WEBVTT NOTE duration:"01:20:41.6370000" NOTE language:en-us NOTE Confidence: 0.8107822 00:00:03.780 --> 00:00:06.644 OK, my friends. I think we'll get started. NOTE Confidence: 0.8107822 $00:00:06.650 \rightarrow 00:00:08.648$ Welcome to our evening Ethics seminar NOTE Confidence: 0.8107822 $00:00:08.648 \rightarrow 00:00:11.180$ series hosted by the Yale School of NOTE Confidence: 0.8107822 00:00:11.180 --> 00:00:13.075 Medicine Program for Biomedical Ethics NOTE Confidence: 0.8107822 00:00:13.075 --> 00:00:15.301 to tonight's program is hosted by NOTE Confidence: 0.8107822 00:00:15.301 --> 00:00:17.056 the Yale Pediatric Ethics Program. NOTE Confidence: 0.8107822 $00{:}00{:}17.060 \dashrightarrow 00{:}00{:}18.860$ They are very much interrelated, NOTE Confidence: 0.8107822 $00{:}00{:}18.860 \dashrightarrow 00{:}00{:}22.076$ but to night we have a very special academic NOTE Confidence: 0.8107822 $00:00:22.076 \rightarrow 00:00:24.595$ pediatrician with us before we get to that, NOTE Confidence: 0.8107822 00:00:24.600 --> 00:00:26.868 I just want to comment you notice NOTE Confidence: 0.8107822 $00:00:26.868 \rightarrow 00:00:28.686$ before perhaps that our upcoming NOTE Confidence: 0.8107822 $00:00:28.686 \rightarrow 00:00:30.641$ session is a conversation with NOTE Confidence: 0.8107822 00:00:30.641 --> 00:00:32.310 Doctor Professor Tim Snyder. NOTE Confidence: 0.8107822 $00:00:32.310 \longrightarrow 00:00:33.084$ About his book.

- NOTE Confidence: 0.8107822
- $00:00:33.084 \rightarrow 00:00:34.885$ Our malady, which is a very easy

00:00:34.885 - > 00:00:35.913 read and very inexpensive.

NOTE Confidence: 0.8107822

 $00:00:35.920 \rightarrow 00:00:38.332$ It's it's either a very small book or along

NOTE Confidence: 0.8107822

 $00:00:38.332 \rightarrow 00:00:40.309$ pamphlet depending on how you look at it,

NOTE Confidence: 0.8107822

 $00{:}00{:}40{.}310 \dashrightarrow 00{:}00{:}42{.}109$ but it's a it's an excellent read.

NOTE Confidence: 0.8107822

 $00{:}00{:}42.110 \dashrightarrow 00{:}00{:}43.944$ I mentioned it's now to the medical

NOTE Confidence: 0.8107822

 $00{:}00{:}43.944 \dashrightarrow 00{:}00{:}45.053$ students because some medical

NOTE Confidence: 0.8107822

 $00:00:45.053 \rightarrow 00:00:46.679$ students have copies that are still

NOTE Confidence: 0.8107822

 $00{:}00{:}46.679 \dashrightarrow 00{:}00{:}48.466$ available to be picked up that we're

NOTE Confidence: 0.8107822

00:00:48.466 --> 00:00:50.135 still waiting for you to pick up.

NOTE Confidence: 0.8107822

 $00:00:50.135 \rightarrow 00:00:51.560$ We're looking forward to having

NOTE Confidence: 0.8107822

 $00{:}00{:}51.560 \dashrightarrow 00{:}00{:}52.700$ several medical students on

NOTE Confidence: 0.8107822

00:00:52.755 --> 00:00:53.979 that call coming up in May.

NOTE Confidence: 0.802747

00:00:57.060 --> 00:00:58.495 The Tonight speaker is someone

NOTE Confidence: 0.802747

 $00:00:58.495 \dashrightarrow 00:01:00.606$ I've known for many years in the NOTE Confidence: 0.802747

2

00:01:00.606 --> 00:01:01.838 world of pediatric bioethics,

NOTE Confidence: 0.802747

00:01:01.840 --> 00:01:04.024 and he's been here once before to speak

NOTE Confidence: 0.802747

 $00{:}01{:}04{.}024 \dashrightarrow 00{:}01{:}06{.}417$ and and Doctor Ben will fund is has

NOTE Confidence: 0.802747

 $00{:}01{:}06{.}417 \dashrightarrow 00{:}01{:}08{.}419$ had a remarkable career in bioethics.

NOTE Confidence: 0.802747

 $00:01:08.420 \longrightarrow 00:01:09.560$ Believe it or not,

NOTE Confidence: 0.802747

 $00{:}01{:}09{.}560 \dashrightarrow 00{:}01{:}11{.}710$ the center of gravity in pediatric ethics,

NOTE Confidence: 0.802747

00:01:11.710 --> 00:01:13.510 United States is not New Haven.

NOTE Confidence: 0.802747

00:01:13.510 --> 00:01:15.000 You know, we're working on

NOTE Confidence: 0.802747

 $00{:}01{:}15{.}000 \dashrightarrow 00{:}01{:}16{.}192$ the center of gravity.

NOTE Confidence: 0.802747

 $00:01:16.200 \rightarrow 00:01:18.584$ 1.1 could have argued that it was Wisconsin.

NOTE Confidence: 0.802747

 $00:01:18.590 \dashrightarrow 00:01:20.378$ Maybe some would still say so,

NOTE Confidence: 0.802747

 $00:01:20.380 \longrightarrow 00:01:22.529$ but I think it kind of shifted

NOTE Confidence: 0.802747

 $00{:}01{:}22.529 \dashrightarrow 00{:}01{:}24.483$ West from Wisconsin to a place

NOTE Confidence: 0.802747

 $00{:}01{:}24.483 \dashrightarrow 00{:}01{:}26.093$ called the Treuman Katz Center.

NOTE Confidence: 0.802747

 $00:01:26.100 \rightarrow 00:01:28.025$ I'll talk about that in a moment,

NOTE Confidence: 0.802747

 $00:01:28.030 \longrightarrow 00:01:29.662$ but tonight's speaker to let you

- NOTE Confidence: 0.802747
- $00:01:29.662 \rightarrow 00:01:31.340$ know is Doctor Ben will font.
- NOTE Confidence: 0.802747
- $00{:}01{:}31{.}340 \dashrightarrow 00{:}01{:}32{.}990$ He is professor of the vision
- NOTE Confidence: 0.802747
- $00:01:32.990 \longrightarrow 00:01:34.457$ of bioethics and palliative care
- NOTE Confidence: 0.802747
- $00:01:34.457 \rightarrow 00:01:36.027$ and pulmonary Sleep Medicine in
- NOTE Confidence: 0.802747
- $00{:}01{:}36{.}027 \dashrightarrow 00{:}01{:}37{.}642$ the Department of Pediatrics at
- NOTE Confidence: 0.802747
- $00:01:37.642 \longrightarrow 00:01:38.878$ the University of Washington.
- NOTE Confidence: 0.802747
- $00{:}01{:}38.880 \dashrightarrow 00{:}01{:}41.981$ He's an investigator at the Truman Katz
- NOTE Confidence: 0.802747
- $00:01:41.981 \dashrightarrow 00:01:44.939$ Center for Pediatric Bioethics in Seattle.
- NOTE Confidence: 0.802747
- 00:01:44.940 --> 00:01:46.730 Doctor Wilfong attended Muhlenberg College,
- NOTE Confidence: 0.802747
- 00:01:46.730 --> 00:01:48.535 Rutgers University of New Jersey
- NOTE Confidence: 0.802747
- 00:01:48.535 --> 00:01:49.979 Medical School and completed
- NOTE Confidence: 0.802747
- 00:01:49.979 --> 00:01:51.871 his postgraduate training at the
- NOTE Confidence: 0.802747
- 00:01:51.871 --> 00:01:53.706 University of Wisconsin and he's
- NOTE Confidence: 0.802747
- 00:01:53.706 --> 00:01:55.463 held faculty appointments at the
- NOTE Confidence: 0.802747
- 00:01:55.463 --> 00:01:57.461 University of Arizona at the NIH
- NOTE Confidence: 0.802747

00:01:57.470 --> 00:01:59.850 and at Johns Hopkins University.

NOTE Confidence: 0.802747

00:01:59.850 --> 00:02:00.681 Several years ago,

NOTE Confidence: 0.802747

00:02:00.681 --> 00:02:02.343 Doctor Wilfong moved out to Seattle,

NOTE Confidence: 0.802747

 $00:02:02.350 \dashrightarrow 00:02:04.350$ which is part of the reason why the

NOTE Confidence: 0.802747

 $00:02:04.350 \longrightarrow 00:02:06.238$ center of gravity made that shift,

NOTE Confidence: 0.802747

 $00:02:06.240 \longrightarrow 00:02:07.580$ and he's actually the founding

NOTE Confidence: 0.802747

00:02:07.580 --> 00:02:09.300 director of the Truman Katz Center,

NOTE Confidence: 0.802747

 $00:02:09.300 \dashrightarrow 00:02:11.260$ and I will tell you that I have

NOTE Confidence: 0.802747

00:02:11.260 --> 00:02:13.443 had the pleasure of being out there

NOTE Confidence: 0.802747

00:02:13.443 --> 00:02:15.628 a few times for meetings to speak

NOTE Confidence: 0.802747

 $00{:}02{:}15.628 \dashrightarrow 00{:}02{:}17.086$ and to learn an it is.

NOTE Confidence: 0.802747

 $00:02:17.090 \longrightarrow 00:02:18.889$ It's a real pleasure to have the

NOTE Confidence: 0.802747

00:02:18.889 --> 00:02:20.314 founding director of that very

NOTE Confidence: 0.802747

 $00:02:20.314 \rightarrow 00:02:21.804$ important Institute here with us.

NOTE Confidence: 0.78285277

 $00{:}02{:}23.880 \dashrightarrow 00{:}02{:}25.506$ Doctor Wilfong's got leadership roles in

NOTE Confidence: 0.78285277

 $00:02:25.506 \rightarrow 00:02:27.118$ various national initiatives, and we're

 $00:02:27.118 \rightarrow 00:02:30.310$ really not going to get into that now.

NOTE Confidence: 0.78285277

 $00{:}02{:}30{.}310 \dashrightarrow 00{:}02{:}31{.}828$ But if there's a quite done,

NOTE Confidence: 0.78285277

 $00{:}02{:}31{.}830 \dashrightarrow 00{:}02{:}33{.}990$ I'll throw in one of his titles or one of

NOTE Confidence: 0.78285277

 $00:02:34.043 \rightarrow 00:02:36.150$ his committees or something if you want,

NOTE Confidence: 0.78285277

00:02:36.150 --> 00:02:37.420 but it's really not. Essentially,

NOTE Confidence: 0.78285277

 $00{:}02{:}37{.}420 \dashrightarrow 00{:}02{:}39{.}292$ I want to get to the conversation and

NOTE Confidence: 0.78285277

 $00:02:39.292 \rightarrow 00:02:41.488$ the way the conversation is going to go,

NOTE Confidence: 0.78285277

 $00:02:41.490 \longrightarrow 00:02:43.093$ as many of you are familiar with

NOTE Confidence: 0.78285277

 $00{:}02{:}43.093 \dashrightarrow 00{:}02{:}44.693$ is Doctor will finally speak for

NOTE Confidence: 0.78285277

 $00:02:44.693 \rightarrow 00:02:46.312$ about 45 minutes, plus or minus.

NOTE Confidence: 0.78285277

 $00:02:46.312 \longrightarrow 00:02:47.836$ After that, I will invite you

NOTE Confidence: 0.78285277

 $00:02:47.836 \longrightarrow 00:02:49.106$ to submit questions via Q&A,

NOTE Confidence: 0.78285277

 $00{:}02{:}49{.}110 \dashrightarrow 00{:}02{:}50{.}832$ and then I will be reading them

NOTE Confidence: 0.78285277

 $00{:}02{:}50{.}832 \dashrightarrow 00{:}02{:}51{.}900$ to Doctor will fund.

NOTE Confidence: 0.78285277

 $00:02:51.900 \dashrightarrow 00:02:54.186$ So if you have any questions along the way,

 $00:02:54.190 \longrightarrow 00:02:55.912$ that's how we'll do that and we

NOTE Confidence: 0.78285277

 $00:02:55.912 \longrightarrow 00:02:57.737$ will have a hard stop at 6:30.

NOTE Confidence: 0.78285277

00:02:57.740 --> 00:03:00.026 So if you're if I didn't get to your

NOTE Confidence: 0.78285277

 $00:03:00.026 \dashrightarrow 00:03:01.560$ question, I apologize in advance.

NOTE Confidence: 0.78285277

 $00{:}03{:}01{.}560 \dashrightarrow 00{:}03{:}03{.}820$ But we'll do the best we can.

NOTE Confidence: 0.78285277

 $00{:}03{:}03{.}820 \dashrightarrow 00{:}03{:}06{.}032$ I think it's going to be very

NOTE Confidence: 0.78285277

00:03:06.032 $\operatorname{-->}$ 00:03:08.290 interesting night on a problem that I

NOTE Confidence: 0.78285277

 $00{:}03{:}08{.}290 \dashrightarrow 00{:}03{:}10{.}546$ think is something that's not an easy

NOTE Confidence: 0.78285277

 $00{:}03{:}10.546 \dashrightarrow 00{:}03{:}12.520$ question both in the clinical sphere

NOTE Confidence: 0.78285277

 $00{:}03{:}12.520 \dashrightarrow 00{:}03{:}14.666$ as well as in the research arena,

NOTE Confidence: 0.78285277

 $00:03:14.666 \rightarrow 00:03:17.099$ and Doctor Wilson has a great deal of NOTE Confidence: 0.78285277

 $00:03:17.099 \rightarrow 00:03:19.001$ experience and expertise in bioethics in NOTE Confidence: 0.78285277

 $00:03:19.001 \rightarrow 00:03:21.259$ both the clinical and research arena,

NOTE Confidence: 0.78285277

 $00{:}03{:}21{.}260 \dashrightarrow 00{:}03{:}23{.}012$ and so with that genetic information

NOTE Confidence: 0.78285277

 $00{:}03{:}23.012 \dashrightarrow 00{:}03{:}24.180$ disclosure and whole genome

NOTE Confidence: 0.78285277

 $00:03:24.234 \rightarrow 00:03:25.790$ sequencing welcome Doctor Wilfong.

- NOTE Confidence: 0.8764313
- 00:03:26.690 --> 00:03:29.546 Thank you, I'm gonna go ahead and

 $00:03:29.546 \longrightarrow 00:03:31.688$ share my screen. To start with.

NOTE Confidence: 0.82752955

 $00{:}03{:}35{.}030 \dashrightarrow 00{:}03{:}35{.}760$ And.

NOTE Confidence: 0.8072139

00:03:39.050 --> 00:03:40.849 Oh, I'm kind of in the beginning.

NOTE Confidence: 0.8072139

 $00{:}03{:}40.850 \dashrightarrow 00{:}03{:}44.080$ Sorry, OK, here we are.

NOTE Confidence: 0.8072139

 $00:03:44.080 \longrightarrow 00:03:45.592$ So first of all,

NOTE Confidence: 0.8072139

 $00:03:45.592 \rightarrow 00:03:47.860$ thank you Mark for that introduction.

NOTE Confidence: 0.8072139

00:03:47.860 - 00:03:50.876 It's really a pleasure to be back here,

NOTE Confidence: 0.8072139

 $00{:}03{:}50.880 \dashrightarrow 00{:}03{:}53.904$ at least virtually with you in New Haven.

NOTE Confidence: 0.8072139

 $00:03:53.910 \rightarrow 00:03:56.913$ And I look forward to the opportunity

NOTE Confidence: 0.8072139

 $00:03:56.913 \rightarrow 00:03:59.797$ of being back here in person so.

NOTE Confidence: 0.8072139

 $00{:}03{:}59{.}800 \dashrightarrow 00{:}04{:}01{.}858$ For those of you who are thinking

NOTE Confidence: 0.8072139

 $00{:}04{:}01{.}858 \dashrightarrow 00{:}04{:}03{.}401$ about my title and wondering

NOTE Confidence: 0.8072139

 $00{:}04{:}03{.}401 \dashrightarrow 00{:}04{:}05{.}494$ what am I going to talk about,

NOTE Confidence: 0.8072139

 $00:04:05.500 \longrightarrow 00:04:07.502$ I'm going to ask you since it's

00:04:07.502 --> 00:04:09.697 been a year of Covid Anna year,

NOTE Confidence: 0.8072139

 $00:04:09.700 \longrightarrow 00:04:11.200$ I'll probably watching more Netflix

NOTE Confidence: 0.8072139

 $00:04:11.200 \longrightarrow 00:04:12.700$ then you've watched in a
while.

NOTE Confidence: 0.8072139

 $00:04:12.700 \longrightarrow 00:04:14.863$ I want you to think of what

NOTE Confidence: 0.8072139

00:04:14.863 -> 00:04:16.899 movie is conjured up by my title.

NOTE Confidence: 0.8072139

 $00{:}04{:}16{.}900 \dashrightarrow 00{:}04{:}20{.}418$ I want you to think of what movie is sort of

NOTE Confidence: 0.8072139

 $00:04:20.418 \rightarrow 00:04:23.586$ captures your imagination of what I might be.

NOTE Confidence: 0.8072139

00:04:23.590 --> 00:04:25.935 Talking about today will let

NOTE Confidence: 0.8072139

 $00{:}04{:}25{.}935 \dashrightarrow 00{:}04{:}28{.}810$ you pause and think about that.

NOTE Confidence: 0.8072139

 $00:04:28.810 \longrightarrow 00:04:29.946$ Because of our format,

NOTE Confidence: 0.8072139

 $00:04:29.946 \dashrightarrow 00:04:32.369$ I can't really hear what your answers are,

NOTE Confidence: 0.8072139

 $00:04:32.370 \longrightarrow 00:04:34.818$ but I'm going to assume that.

NOTE Confidence: 0.8072139

00:04:34.820 --> 00:04:36.860 At least some of you have

NOTE Confidence: 0.8072139

 $00:04:36.860 \longrightarrow 00:04:38.220$ thought of this movie.

NOTE Confidence: 0.8072139

 $00{:}04{:}38{.}220 \dashrightarrow 00{:}04{:}40{.}411$ Gattaca was at 25 years old and

NOTE Confidence: 0.8072139

 $00:04:40.411 \longrightarrow 00:04:42.490$ it was really this future where

- NOTE Confidence: 0.8072139
- $00:04:42.490 \rightarrow 00:04:44.975$ the world your place in the world
- NOTE Confidence: 0.8072139
- 00:04:45.048 --> 00:04:46.718 was based upon your DNA.
- NOTE Confidence: 0.8072139
- $00{:}04{:}46.720 \dashrightarrow 00{:}04{:}49.000$ And of course this is sort of a
- NOTE Confidence: 0.8072139
- $00:04:49.000 \rightarrow 00:04:50.815$ futuristic version of the implications
- NOTE Confidence: 0.8072139
- $00:04:50.815 \rightarrow 00:04:53.173$ of sequencing of knowledge about genetics.
- NOTE Confidence: 0.8072139
- $00:04:53.180 \rightarrow 00:04:55.328$ Being able to predict how people
- NOTE Confidence: 0.8072139
- $00:04:55.328 \rightarrow 00:04:57.940$ will behave and how they will do it.
- NOTE Confidence: 0.8072139
- $00:04:57.940 \longrightarrow 00:05:00.430$ Certainly very sort of a
- NOTE Confidence: 0.8072139
- $00{:}05{:}00{.}430 \dashrightarrow 00{:}05{:}02{.}920$ dystopian vision of the world.
- NOTE Confidence: 0.8072139
- $00:05:02.920 \dashrightarrow 00:05:05.360$ I hope for a variety of reasons that
- NOTE Confidence: 0.8072139
- $00:05:05.360 \longrightarrow 00:05:07.649$ you'll leave something more optimistic,
- NOTE Confidence: 0.8072139
- $00:05:07.650 \rightarrow 00:05:10.692$ and I'm actually going to share
- NOTE Confidence: 0.8072139
- $00:05:10.692 \longrightarrow 00:05:13.290$ with you another movie that.
- NOTE Confidence: 0.8072139
- $00{:}05{:}13.290 \dashrightarrow 00{:}05{:}16.644$ I mean partly seriously and partly
- NOTE Confidence: 0.8072139
- $00:05:16.644 \rightarrow 00:05:20.509$ jokingly is a better image for what?
- NOTE Confidence: 0.8072139

 $00:05:20.510 \longrightarrow 00:05:23.457$ Genetics may bring us in the future.

NOTE Confidence: 0.8072139

 $00:05:23.460 \longrightarrow 00:05:25.968$ That's the graduate.

NOTE Confidence: 0.8072139

 $00{:}05{:}25{.}970 \dashrightarrow 00{:}05{:}28{.}700$ Now the reason why I'm using this

NOTE Confidence: 0.8072139

 $00:05:28.700 \longrightarrow 00:05:31.509$ movie is because of for those of you.

NOTE Confidence: 0.8072139

 $00:05:31.510 \longrightarrow 00:05:33.718$ This is about 50 years old.

NOTE Confidence: 0.8072139

 $00:05:33.720 \rightarrow 00:05:37.032$ For those of you who have not seen it,

NOTE Confidence: 0.8072139

 $00:05:37.040 \longrightarrow 00:05:38.880$ there is a very particular

NOTE Confidence: 0.8072139

 $00:05:38.880 \longrightarrow 00:05:40.352$ scene where the protagonist,

NOTE Confidence: 0.8072139

 $00{:}05{:}40{.}360 \dashrightarrow 00{:}05{:}42{.}448$ who's also named Benjamin in this

NOTE Confidence: 0.8072139

 $00:05:42.448 \rightarrow 00:05:44.420$ case played by Dustin Hoffman,

NOTE Confidence: 0.8072139

 $00{:}05{:}44{.}420 \dashrightarrow 00{:}05{:}46{.}712$ is getting advice from a friend

NOTE Confidence: 0.8072139

 $00:05:46.712 \longrightarrow 00:05:48.990$ of his father's about what he

NOTE Confidence: 0.8072139

 $00:05:48.990 \longrightarrow 00:05:50.685$ should do for his career.

NOTE Confidence: 0.8072139

 $00{:}05{:}50{.}690 \dashrightarrow 00{:}05{:}54{.}197$ And he has one word for Benjamin.

NOTE Confidence: 0.8072139

 $00{:}05{:}54{.}200 \dashrightarrow 00{:}05{:}57{.}380$ And that word is plastics.

NOTE Confidence: 0.8072139

 $00:05:57.380 \rightarrow 00:05:59.788$ This is taking place and you know this.

- NOTE Confidence: 0.8072139
- $00:05:59.790 \longrightarrow 00:06:02.198$ This film came out in the early 60s.

 $00{:}06{:}02.200 \dashrightarrow 00{:}06{:}03.705$ It's supposed to take place

NOTE Confidence: 0.8072139

 $00{:}06{:}03.705 \dashrightarrow 00{:}06{:}05.210$ like in the late 50s.

NOTE Confidence: 0.8072139

 $00:06:05.210 \longrightarrow 00:06:06.992$ So clearly plastics at that point

NOTE Confidence: 0.8072139

 $00:06:06.992 \longrightarrow 00:06:08.820$ was the future of the world.

NOTE Confidence: 0.8072139

00:06:08.820 --> 00:06:10.927 So why am I telling you this?

NOTE Confidence: 0.8072139

 $00{:}06{:}10{.}930 \dashrightarrow 00{:}06{:}12{.}771$ Because I want to share with you

NOTE Confidence: 0.8072139

 $00:06:12.771 \longrightarrow 00:06:14.654$ a story of what happened to me

NOTE Confidence: 0.8072139

00:06:14.654 --> 00:06:16.737 when I was a fellow in Wisc
consin

NOTE Confidence: 0.8072139

 $00{:}06{:}16.737 \dashrightarrow 00{:}06{:}19.117$ doing a combined fellowship in

NOTE Confidence: 0.8072139

00:06:19.117 --> 00:06:20.545 pediatric pulmonologist and.

NOTE Confidence: 0.8072139

 $00{:}06{:}20.550 \dashrightarrow 00{:}06{:}22.602$ In bioethics and trying to think

NOTE Confidence: 0.8072139

 $00{:}06{:}22.602 \dashrightarrow 00{:}06{:}24.579$ about what I would work on,

NOTE Confidence: 0.8072139

 $00{:}06{:}24.580 \dashrightarrow 00{:}06{:}26.260$ and I had some ideas,

NOTE Confidence: 0.8072139

 $00:06:26.260 \longrightarrow 00:06:27.640$ but the Phil Farrell,

 $00:06:27.640 \rightarrow 00:06:30.628$ who was the chair of Pediatrics at that time,

NOTE Confidence: 0.8072139

 $00{:}06{:}30{.}630 \dashrightarrow 00{:}06{:}32{.}646$ sort of reminiscent of the graduate,

NOTE Confidence: 0.8072139

 $00:06:32.650 \longrightarrow 00:06:34.660$ came to me with one word.

NOTE Confidence: 0.8072139

00:06:34.660 --> 00:06:36.004 Although it's actually maybe

NOTE Confidence: 0.8072139

 $00:06:36.004 \rightarrow 00:06:37.348$ two in this case,

NOTE Confidence: 0.8072139

 $00:06:37.350 \longrightarrow 00:06:39.366$ maybe 2 words or three words.

NOTE Confidence: 0.8072139

 $00:06:39.370 \longrightarrow 00:06:41.386$ I guess it was began with

NOTE Confidence: 0.8072139

00:06:41.386 --> 00:06:42.730 the P wasn't plastics,

NOTE Confidence: 0.81737673

00:06:42.730 --> 00:06:45.960 it was polymerase chain reaction.

NOTE Confidence: 0.81737673

00:06:45.960 --> 00:06:49.272 This is 1988. He said, you know,

NOTE Confidence: 0.81737673

 $00:06:49.272 \rightarrow 00:06:51.729$ three years ago they developed this new

NOTE Confidence: 0.81737673

 $00:06:51.729 \rightarrow 00:06:54.224$ technique and it's going to change the world.

NOTE Confidence: 0.81737673

 $00{:}06{:}54{.}230 \dashrightarrow 00{:}06{:}56{.}883$ You need to think about the ethical

NOTE Confidence: 0.81737673

 $00{:}06{:}56{.}883 \dashrightarrow 00{:}06{:}58{.}580$ implications of molecular biology.

NOTE Confidence: 0.81737673

00:06:58.580 --> 00:07:00.626 He didn't even use word genetics,

NOTE Confidence: 0.81737673

 $00:07:00.630 \rightarrow 00:07:02.926$ it was microbiology and you should talk

 $00{:}07{:}02{.}926 \dashrightarrow 00{:}07{:}05{.}645$ with norm thoughts to his director of your

NOTE Confidence: 0.81737673

 $00{:}07{:}05.645 \dashrightarrow 00{:}07{:}07.732$ residency program because he thinks about

NOTE Confidence: 0.81737673

 $00:07:07.732 \longrightarrow 00:07:09.868$ the ethical issues of genetic testing.

NOTE Confidence: 0.81737673

 $00{:}07{:}09{.}870 \dashrightarrow 00{:}07{:}11{.}178$ And by the way,

NOTE Confidence: 0.81737673

00:07:11.178 --> 00:07:12.813 your appeared pulmonary fellow and

NOTE Confidence: 0.81737673

 $00{:}07{:}12.813 \dashrightarrow 00{:}07{:}15.095$ you know any year they're going to

NOTE Confidence: 0.81737673

 $00:07:15.095 \dashrightarrow 00:07:17.390$ identify the gene for cystic fibrosis.

NOTE Confidence: 0.81737673

 $00{:}07{:}17.390 \dashrightarrow 00{:}07{:}20.550$ So you should think about what are the

NOTE Confidence: 0.81737673

 $00{:}07{:}20.550 \dashrightarrow 00{:}07{:}22.197$ ethical implications of identifying

NOTE Confidence: 0.81737673

 $00:07:22.197 \longrightarrow 00:07:24.555$ the gene for cystic fibrosis so.

NOTE Confidence: 0.81737673

00:07:24.560 --> 00:07:28.530 I took his advice and as a fellow with Norm,

NOTE Confidence: 0.81737673

00:07:28.530 --> 00:07:31.099 Fost wrote a paper that came out

NOTE Confidence: 0.81737673

00:07:31.099 --> 00:07:34.011 about six months after the gene for

NOTE Confidence: 0.81737673

 $00{:}07{:}34.011 \dashrightarrow 00{:}07{:}36.181$ cystic fibrosis was identified and

NOTE Confidence: 0.81737673

 $00{:}07{:}36.181 \dashrightarrow 00{:}07{:}38.565$ what happened in that six month

00:07:38.565 --> 00:07:41.232 period is that a number of companies

NOTE Confidence: 0.81737673

 $00{:}07{:}41.232 \dashrightarrow 00{:}07{:}42.816$ started immediately market marketing.

NOTE Confidence: 0.85681623

 $00:07:45.160 \dashrightarrow 00:07:47.758$ Cystic fibrosis carrier testing to the NOTE Confidence: 0.85681623

00:07:47.758 --> 00:07:49.903 general population and the question

NOTE Confidence: 0.85681623

 $00{:}07{:}49{.}903 \dashrightarrow 00{:}07{:}52{.}283$ that we addressed in article is how

NOTE Confidence: 0.85681623

 $00:07:52.283 \dashrightarrow 00:07:54.900$ do you think about genetic testing?

NOTE Confidence: 0.85681623

 $00:07:54.900 \rightarrow 00:07:57.336$ But when it becomes technically feasible? NOTE Confidence: 0.85681623

 $00{:}07{:}57{.}340 \dashrightarrow 00{:}07{:}59{.}780$ Ian commercially available what process

NOTE Confidence: 0.85681623

00:07:59.780 --> 00:08:02.628 should be used for deciding whether

NOTE Confidence: 0.85681623

 $00:08:02.628 \dashrightarrow 00:08:05.580$ or when or how to implement this on a NOTE Confidence: 0.85681623

00:08:05.654 --> 00:08:08.356 wide scale basis and what we identified NOTE Confidence: 0.85681623

 $00{:}08{:}08{.}356 \dashrightarrow 00{:}08{:}11{.}460$ in this paper is a theme that really

NOTE Confidence: 0.85681623

 $00{:}08{:}11.460 \dashrightarrow 00{:}08{:}13.990$ has followed through my entire career.

NOTE Confidence: 0.85681623

 $00{:}08{:}13.990 \dashrightarrow 00{:}08{:}16.849$ The notion that.

NOTE Confidence: 0.85681623

 $00{:}08{:}16.850 \dashrightarrow 00{:}08{:}18.578$ Genetic testing requires evidence

NOTE Confidence: 0.85681623

 $00{:}08{:}18{.}578 \dashrightarrow 00{:}08{:}20{.}738$ of benefit and relative evidence

- NOTE Confidence: 0.85681623
- $00{:}08{:}20.738 \dashrightarrow 00{:}08{:}23.469$ of lack of harm to sort of have

 $00{:}08{:}23.469 \dashrightarrow 00{:}08{:}25.496$ an appropriate balance that it

NOTE Confidence: 0.85681623

 $00:08:25.496 \rightarrow 00:08:26.999$ requires understanding thoughts,

NOTE Confidence: 0.85681623

 $00:08:27.000 \rightarrow 00:08:29.436$ issues of informed consent of education,

NOTE Confidence: 0.85681623

 $00:08:29.440 \longrightarrow 00:08:30.246$ quality control,

NOTE Confidence: 0.85681623

 $00{:}08{:}30{.}246 \dashrightarrow 00{:}08{:}33{.}067$ counseling and that we have to think

NOTE Confidence: 0.85681623

 $00:08:33.067 \rightarrow 00:08:35.526$ about the workforce issues as well,

NOTE Confidence: 0.85681623

 $00:08:35.530 \rightarrow 00:08:39.590$ and so this paper that I worked on in 1990,

NOTE Confidence: 0.85681623

 $00:08:39.590 \rightarrow 00:08:42.886$ the themes of this are still present today

NOTE Confidence: 0.85681623

 $00:08:42.886 \dashrightarrow 00:08:46.625$ as we think about this as a relates to.

NOTE Confidence: 0.85681623

00:08:46.630 --> 00:08:46.999 Sequencing,

NOTE Confidence: 0.85681623

 $00{:}08{:}46{.}999 \dashrightarrow 00{:}08{:}50{.}320$ so I want to begin with this sort of

NOTE Confidence: 0.85681623

 $00{:}08{:}50{.}399 \dashrightarrow 00{:}08{:}52{.}889$ historical introduction to the issues that

NOTE Confidence: 0.85681623

 $00{:}08{:}52{.}889 \dashrightarrow 00{:}08{:}56{.}859$ I and how I got interested in this issue.

NOTE Confidence: 0.85681623

00:08:56.860 -> 00:08:57.536 Uhm?

- 00:08:57.536 --> 00:09:00.240 I've gotta Fast forward.
- NOTE Confidence: 0.85681623
- $00:09:00.240 \longrightarrow 00:09:02.720$ 15 years.
- NOTE Confidence: 0.85681623
- $00:09:02.720 \longrightarrow 00:09:03.569$ 14 years, 2004.
- NOTE Confidence: 0.85681623
- $00{:}09{:}03.569 \dashrightarrow 00{:}09{:}05.550$ At this point I was working at
- NOTE Confidence: 0.85681623
- $00{:}09{:}05{.}615 \dashrightarrow 00{:}09{:}07{.}643$ the NIH and the Department of
- NOTE Confidence: 0.85681623
- $00{:}09{:}07.643 \dashrightarrow 00{:}09{:}09.310$ Clinical Bioethics at the NIH.
- NOTE Confidence: 0.85681623
- $00{:}09{:}09{.}310 \dashrightarrow 00{:}09{:}11.648$ I was also working with the Human
- NOTE Confidence: 0.85681623
- 00:09:11.648 --> 00:09:13.157 Genome Research Institute and one
- NOTE Confidence: 0.85681623
- 00:09:13.157 --> 00:09:14.925 of the things I did while I was
- NOTE Confidence: 0.85681623
- 00:09:14.985 --> 00:09:17.603 there is that we did research ethics
- NOTE Confidence: 0.85681623
- $00{:}09{:}17.603 \dashrightarrow 00{:}09{:}19.044$ consultations for researchers who
- NOTE Confidence: 0.85681623
- $00{:}09{:}19{.}044 \dashrightarrow 00{:}09{:}20{.}928$ had questions that they were facing.
- NOTE Confidence: 0.85681623
- $00:09:20.930 \rightarrow 00:09:23.438$ There were challenging for the research.
- NOTE Confidence: 0.85681623
- $00{:}09{:}23{.}440 \dashrightarrow 00{:}09{:}27{.}360$ And so the phone Rang and it was
- NOTE Confidence: 0.85681623
- $00:09:27.360 \longrightarrow 00:09:30.638$ a investigator from the National
- NOTE Confidence: 0.85681623
- $00:09:30.638 \longrightarrow 00:09:32.960$ Cancer Institute. It wasn't.

 $00:09:32.960 \longrightarrow 00:09:35.540$ At the time I thought myself,

NOTE Confidence: 0.85681623

 $00:09:35.540 \longrightarrow 00:09:37.496$ I didn't know that there were

NOTE Confidence: 0.85681623

 $00:09:37.496 \rightarrow 00:09:39.080$ dermatologists working at the NCI.

NOTE Confidence: 0.85681623

 $00:09:39.080 \longrightarrow 00:09:41.357$ He then went on and and told me that

NOTE Confidence: 0.85681623

00:09:41.357 --> 00:09:43.944 he studied the disease called 0 Derma

NOTE Confidence: 0.85681623

 $00:09:43.944 \dashrightarrow 00:09:46.170$ Pigmentosa which I have to recall.

NOTE Confidence: 0.85681623

 $00{:}09{:}46.170 \dashrightarrow 00{:}09{:}48.590$ I had a vague idea of what it was but

NOTE Confidence: 0.85681623

 $00:09:48.658 \rightarrow 00:09:50.553$ couldn't remember although he explained

NOTE Confidence: 0.85681623

 $00:09:50.553 \dashrightarrow 00:09:53.569$ much more to me on that phone call.

NOTE Confidence: 0.85681623

 $00{:}09{:}53{.}570 \dashrightarrow 00{:}09{:}55{.}724$ This is a disease genetic disease

NOTE Confidence: 0.85681623

 $00:09:55.724 \rightarrow 00:09:57.849$ that's very rare 100,000 people and

NOTE Confidence: 0.85681623

 $00:09:57.849 \dashrightarrow 00:10:00.329$ it's caused by defect in the jeans for.

NOTE Confidence: 0.85681623

00:10:00.330 --> 00:10:01.106 DNA repair,

NOTE Confidence: 0.85681623

00:10:01.106 --> 00:10:01.882 in fact,

NOTE Confidence: 0.85681623

 $00{:}10{:}01{.}882 \dashrightarrow 00{:}10{:}04{.}210$ it was through this disease that

 $00{:}10{:}04{.}290 \dashrightarrow 00{:}10{:}06{.}840$ the whole mechanisms of DNA repair

NOTE Confidence: 0.85681623

 $00{:}10{:}06.840 \dashrightarrow 00{:}10{:}09.694$ were elucidated and the NIH had been

NOTE Confidence: 0.85681623

 $00:10:09.694 \rightarrow 00:10:12.207$ studying this for the last 30 years,

NOTE Confidence: 0.85681623

 $00{:}10{:}12{.}210 \dashrightarrow 00{:}10{:}12{.}610$ and.

NOTE Confidence: 0.8571908

 $00{:}10{:}14.730 \dashrightarrow 00{:}10{:}17.404$ The implication of this disease is that

NOTE Confidence: 0.8571908

 $00:10:17.404 \rightarrow 00:10:20.297$ because of this problem with DNA repair,

NOTE Confidence: 0.8571908

 $00{:}10{:}20{.}300 \dashrightarrow 00{:}10{:}23{.}128$ children who have this disease are at

NOTE Confidence: 0.8571908

 $00:10:23.128 \rightarrow 00:10:25.868$ a huge increase risk of sun cancer,

NOTE Confidence: 0.8571908

 $00{:}10{:}25{.}870 \dashrightarrow 00{:}10{:}28{.}150$ so any exposure to UV light

NOTE Confidence: 0.8571908

 $00{:}10{:}28.150 \dashrightarrow 00{:}10{:}30.650$ increases their risk of skin cancer.

NOTE Confidence: 0.8571908

 $00{:}10{:}30.650 \dashrightarrow 00{:}10{:}33.140$ Some of them also have neurologic

NOTE Confidence: 0.8571908

 $00{:}10{:}33.140 \dashrightarrow 00{:}10{:}36.159$ problems as well that develop overtime.

NOTE Confidence: 0.8571908

 $00:10:36.160 \rightarrow 00:10:38.768$ The risk is so great that there's actually

NOTE Confidence: 0.8571908

00:10:38.768 --> 00:10:41.416 a camp in New York called Camp Sundown

NOTE Confidence: 0.8571908

 $00:10:41.416 \rightarrow 00:10:44.098$ that is for kids with this condition,

NOTE Confidence: 0.8571908

 $00:10:44.100 \longrightarrow 00:10:46.578$ so they can actually have a normal

- NOTE Confidence: 0.8571908
- $00:10:46.578 \rightarrow 00:10:49.344$ life by having a week of camp that

 $00:10:49.344 \rightarrow 00:10:51.728$ occurs all night long and they sleep

NOTE Confidence: 0.8571908

 $00:10:51.728 \longrightarrow 00:10:54.448$ all day and this is picture in front

NOTE Confidence: 0.8571908

 $00:10:54.450 \rightarrow 00:10:57.210$ of you is an image from that camp.

NOTE Confidence: 0.8571908

 $00:10:57.210 \longrightarrow 00:10:59.280$ So why did he call me?

NOTE Confidence: 0.8571908

 $00{:}10{:}59{.}280 \dashrightarrow 00{:}11{:}01{.}975$ Well, he the investigator was using at

NOTE Confidence: 0.8571908

 $00{:}11{:}01{.}975 \dashrightarrow 00{:}11{:}04{.}990$ that time the state of the art techniques

NOTE Confidence: 0.8571908

 $00{:}11{:}04{.}990 \dashrightarrow 00{:}11{:}07{.}660$ which had to do with organizing.

NOTE Confidence: 0.8571908

 $00{:}11{:}07.660 \dashrightarrow 00{:}11{:}09.935$ Patients with this condition into

NOTE Confidence: 0.8571908

 $00{:}11{:}09{.}935 \dashrightarrow 00{:}11{:}12{.}703$ seven different groups based upon a

NOTE Confidence: 0.8571908

 $00:11:12.703 \rightarrow 00:11:14.798$ laboratory analysis of their cells.

NOTE Confidence: 0.8571908

 $00{:}11{:}14.800 \dashrightarrow 00{:}11{:}17.030$ That and this was so.

NOTE Confidence: 0.8571908

00:11:17.030 --> 00:11:19.700 This was not typical genetic testing,

NOTE Confidence: 0.8571908

 $00:11:19.700 \rightarrow 00:11:22.794$ but it was genetic based testing that NOTE Confidence: 0.8571908

 $00:11:22.794 \rightarrow 00:11:25.553$ really describe these seven different

- $00:11:25.553 \rightarrow 00:11:30.199$ categories. And these categories.
- NOTE Confidence: 0.8571908
- $00{:}11{:}30{.}200 \dashrightarrow 00{:}11{:}32{.}636$ Might have some ability to predict
- NOTE Confidence: 0.8571908
- 00:11:32.636 --> 00:11:34.779 how someone is going to do,
- NOTE Confidence: 0.8571908
- $00:11:34.780 \longrightarrow 00:11:37.573$ but there's a lot of uncertainty about
- NOTE Confidence: 0.8571908
- $00:11:37.573 \rightarrow 00:11:39.930$ how meaningful these categories were.
- NOTE Confidence: 0.8571908
- $00{:}11{:}39{.}930 \dashrightarrow 00{:}11{:}40{.}785$ At the NIH,
- NOTE Confidence: 0.8571908
- 00:11:40.785 --> 00:11:42.495 they were doing a Natural History
- NOTE Confidence: 0.8571908
- $00:11:42.495 \rightarrow 00:11:44.652$ study where they were doing research
- NOTE Confidence: 0.8571908
- $00{:}11{:}44.652 \dashrightarrow 00{:}11{:}46.467$ with patients with this condition.
- NOTE Confidence: 0.8571908
- $00:11:46.470 \rightarrow 00:11:48.110$ They were doing these studies,
- NOTE Confidence: 0.8571908
- $00:11:48.110 \longrightarrow 00:11:49.982$ but they were not routinely disclosing NOTE Confidence: 0.8571908
- $00{:}11{:}49{.}982 \dashrightarrow 00{:}11{:}52{.}030$ the results because they were uncertain.
- NOTE Confidence: 0.8571908
- 00:11:52.030 --> 00:11:54.400 Their ambiguous, but they're using
- NOTE Confidence: 0.8571908
- $00:11:54.400 \dashrightarrow 00:11:57.369$ it for their research because of the.
- NOTE Confidence: 0.8571908
- 00:11:57.370 --> 00:12:00.210 Community was very small.
- NOTE Confidence: 0.8571908
- $00:12:00.210 \longrightarrow 00:12:02.208$ People were aware that in other

- NOTE Confidence: 0.8571908
- 00:12:02.208 --> 00:12:04.520 countries in the UK in particular,

00:12:04.520 --> 00:12:07.688 it was actually common to find out as part

NOTE Confidence: 0.8571908

 $00:12:07.688 \rightarrow 00:12:10.895$ what what complementary group you were in.

NOTE Confidence: 0.8571908

 $00:12:10.900 \longrightarrow 00:12:12.224$ And so the people.

NOTE Confidence: 0.8571908

 $00:12:12.224 \longrightarrow 00:12:13.548$ Then I hate you,

NOTE Confidence: 0.8571908

 $00{:}12{:}13.550 \dashrightarrow 00{:}12{:}15.518$ and parts that we want to

NOTE Confidence: 0.8571908

 $00:12:15.518 \longrightarrow 00:12:17.190$ find out our results too.

NOTE Confidence: 0.8571908

 $00{:}12{:}17{.}190 \dashrightarrow 00{:}12{:}18{.}850$ And the investigator contacted me

NOTE Confidence: 0.8571908

 $00:12:18.850 \longrightarrow 00:12:20.830$ because he was concerned about this.

NOTE Confidence: 0.8571908

 $00:12:20.830 \longrightarrow 00:12:22.816$ He was not sure whether this

NOTE Confidence: 0.8571908

 $00:12:22.816 \longrightarrow 00:12:24.140$ was appropriate to do,

NOTE Confidence: 0.8571908

 $00{:}12{:}24{.}140 \dashrightarrow 00{:}12{:}26{.}724$ and so he really raised us as a

NOTE Confidence: 0.8571908

 $00:12:26.724 \longrightarrow 00:12:28.767$ question for me to think about.

NOTE Confidence: 0.8571908

 $00{:}12{:}28.770 \dashrightarrow 00{:}12{:}30.890$ And this was the first time I really

NOTE Confidence: 0.8571908

 $00{:}12{:}30{.}890 \dashrightarrow 00{:}12{:}32{.}672$ thought very specifically about the

- $00:12:32.672 \rightarrow 00:12:35.054$ question of results disclosure in research,
- NOTE Confidence: 0.8571908
- $00{:}12{:}35{.}060 \dashrightarrow 00{:}12{:}36{.}715$ where there was no prior
- NOTE Confidence: 0.8571908
- 00:12:36.715 --> 00:12:38.039 expectation of returning results,
- NOTE Confidence: 0.8571908
- $00:12:38.040 \rightarrow 00:12:40.026$ but patients were wanting this information.
- NOTE Confidence: 0.8571908
- $00{:}12{:}40{.}030 \dashrightarrow 00{:}12{:}40{.}806$ This resulted.
- NOTE Confidence: 0.8571908
- 00:12:40.806 --> 00:12:42.746 Overtime and myself working with.
- NOTE Confidence: 0.8571908
- $00:12:42.750 \longrightarrow 00:12:43.374$ With that,
- NOTE Confidence: 0.8571908
- $00{:}12{:}43.374 \dashrightarrow 00{:}12{:}45.870$ a fellow at the NIH for Deep Rovinsky
- NOTE Confidence: 0.8571908
- $00{:}12{:}45{.}941 \dashrightarrow 00{:}12{:}48{.}347$ who's now the University Of Montreal?
- NOTE Confidence: 0.8571908
- $00{:}12{:}48{.}350 \dashrightarrow 00{:}12{:}50{.}702$ We wrote a paper about thinking about
- NOTE Confidence: 0.8571908
- $00{:}12{:}50{.}702 \dashrightarrow 00{:}12{:}53{.}229$ the issues of disclosing individual.
- NOTE Confidence: 0.8571908
- 00:12:53.230 --> 00:12:54.925 Research results to participants and
- NOTE Confidence: 0.8571908
- $00{:}12{:}54{.}925 \dashrightarrow 00{:}12{:}57{.}805$ we try to think through what we are the
- NOTE Confidence: 0.8571908
- $00{:}12{:}57{.}805 \dashrightarrow 00{:}13{:}00{.}010$ criteria for thinking about how to do this.
- NOTE Confidence: 0.8571908
- $00{:}13{:}00{.}010 \dashrightarrow 00{:}13{:}01{.}630$ Because the debate at the time
- NOTE Confidence: 0.8571908
- $00:13:01.630 \rightarrow 00:13:03.390$ was really in two directions.

- NOTE Confidence: 0.8571908
- $00:13:03.390 \longrightarrow 00:13:05.558$ 1 would say look this is for research

 $00{:}13{:}05{.}558 \dashrightarrow 00{:}13{:}07{.}189$ purposes only and therefore there's

NOTE Confidence: 0.8571908

 $00:13:07.189 \rightarrow 00:13:09.247$ no obligation to return any results.

NOTE Confidence: 0.8571908

 $00:13:09.250 \rightarrow 00:13:11.086$ The other view was to say,

NOTE Confidence: 0.8571908

 $00:13:11.090 \rightarrow 00:13:13.554$ you know people if we think about autonomy,

NOTE Confidence: 0.8571908

 $00:13:13.560 \rightarrow 00:13:14.793$ people should have.

NOTE Confidence: 0.8571908

 $00:13:14.793 \rightarrow 00:13:16.848$ Access to information about themselves,

NOTE Confidence: 0.8571908

 $00:13:16.850 \longrightarrow 00:13:17.208$ particularly.

NOTE Confidence: 0.8571908

 $00:13:17.208 \longrightarrow 00:13:18.998$ There were the ones who

NOTE Confidence: 0.8571908

00:13:18.998 --> 00:13:20.072 contribute this information,

NOTE Confidence: 0.8571908

00:13:20.080 --> 00:13:21.504 regardless of its meaning.

NOTE Confidence: 0.8571908

00:13:21.504 --> 00:13:23.640 So the approach that Guardian I

NOTE Confidence: 0.85168897

 $00:13:23.701 \longrightarrow 00:13:26.301$ took was to suggest that in fact rather

NOTE Confidence: 0.85168897

 $00{:}13{:}26{.}301 \dashrightarrow 00{:}13{:}28{.}512$ than thinking of either a research

NOTE Confidence: 0.85168897

 $00:13:28.512 \rightarrow 00:13:30.756$ focused or an autonomy focused approach,

 $00:13:30.760 \rightarrow 00:13:34.054$ we should think of we described as a result

NOTE Confidence: 0.85168897

 $00{:}13{:}34.054 \dashrightarrow 00{:}13{:}36.539$ focused approach thinking about what the NOTE Confidence: 0.85168897

00:13:36.539 --> 00:13:39.879 result itself is to help guide what to do.

NOTE Confidence: 0.85168897

00:13:39.880 --> 00:13:42.358 Now, the way we thought about the

NOTE Confidence: 0.85168897

00:13:42.358 --> 00:13:44.474 result focused approach is to think

NOTE Confidence: 0.85168897

 $00{:}13{:}44.474 \dashrightarrow 00{:}13{:}46.448$ about 2 broad types of considerations NOTE Confidence: 0.85168897

 $00{:}13{:}46{.}448 \dashrightarrow 00{:}13{:}49{.}430$ one had to do with what I will describe NOTE Confidence: 0.85168897

 $00:13:49.430 \longrightarrow 00:13:50.592$ as informational considerations.

NOTE Confidence: 0.85168897

 $00{:}13{:}50{.}592 \dashrightarrow 00{:}13{:}52{.}640$ The other contextual considerations.

NOTE Confidence: 0.85168897

 $00{:}13{:}52{.}640 \dashrightarrow 00{:}13{:}54{.}880$ And but informational considerations

NOTE Confidence: 0.85168897

 $00{:}13{:}54{.}880 \dashrightarrow 00{:}13{:}57{.}680$ include two important concepts one

NOTE Confidence: 0.85168897

 $00:13:57.680 \longrightarrow 00:14:00.657$ has to do with analytic validity

NOTE Confidence: 0.85168897

 $00:14:00.657 \longrightarrow 00:14:02.058$ and clinical utility.

NOTE Confidence: 0.85168897

 $00:14:02.060 \longrightarrow 00:14:04.044$ Analytic validity refers to

NOTE Confidence: 0.85168897

 $00{:}14{:}04{.}044 \dashrightarrow 00{:}14{:}06{.}028$ whether or not the.

NOTE Confidence: 0.85168897

00:14:06.030 --> 00:14:09.789 Study the result you get is actually.

- NOTE Confidence: 0.85168897
- $00{:}14{:}09{.}790 \dashrightarrow 00{:}14{:}10{.}771$ A true result.
- NOTE Confidence: 0.85168897
- $00:14:10.771 \longrightarrow 00:14:12.733$ An analytic validity refers to whether
- NOTE Confidence: 0.85168897
- $00:14:12.733 \longrightarrow 00:14:14.870$ or not the samples were mixed up.
- NOTE Confidence: 0.85168897
- $00:14:14.870 \longrightarrow 00:14:16.823$ It has to do with a wide range of
- NOTE Confidence: 0.85168897
- $00:14:16.823 \longrightarrow 00:14:18.384$ issues that have nothing to do
- NOTE Confidence: 0.85168897
- $00:14:18.384 \rightarrow 00:14:20.100$ with the meaning of the result,
- NOTE Confidence: 0.85168897
- $00:14:20.100 \longrightarrow 00:14:22.038$ but just whether or not the
- NOTE Confidence: 0.85168897
- $00:14:22.038 \longrightarrow 00:14:23.330$ result is reliably true.
- NOTE Confidence: 0.85168897
- $00:14:23.330 \longrightarrow 00:14:25.110$ Clinical utility as another critically
- NOTE Confidence: 0.85168897
- $00:14:25.110 \longrightarrow 00:14:27.211$ important concept is the notion of
- NOTE Confidence: 0.85168897
- $00{:}14{:}27{.}211 \dashrightarrow 00{:}14{:}29{.}038$ what will you do with the information.
- NOTE Confidence: 0.85168897
- $00{:}14{:}29{.}040 \dashrightarrow 00{:}14{:}30{.}625$ How will the information be
- NOTE Confidence: 0.85168897
- $00:14:30.625 \rightarrow 00:14:32.210$ useful to drive clinical care?
- NOTE Confidence: 0.85168897
- 00:14:32.210 --> 00:14:34.338 And you'll see that these notions of
- NOTE Confidence: 0.85168897
- $00:14:34.338 \rightarrow 00:14:35.943$ analytic validity and clinical utility
- NOTE Confidence: 0.85168897

 $00{:}14{:}35{.}943 \dashrightarrow 00{:}14{:}38{.}176$ are still relevant when we talk about

NOTE Confidence: 0.85168897

 $00{:}14{:}38{.}176 \dashrightarrow 00{:}14{:}40{.}126$ issues related to sequencing as well,

NOTE Confidence: 0.85168897

 $00{:}14{:}40{.}130 \dashrightarrow 00{:}14{:}41{.}710$ and another word for often

NOTE Confidence: 0.85168897

 $00{:}14{:}41.710 \dashrightarrow 00{:}14{:}42.974$ used for clinical utilities.

NOTE Confidence: 0.85168897

 $00:14:42.980 \longrightarrow 00:14:44.380$ Notion of action ability.

NOTE Confidence: 0.85168897

 $00:14:44.380 \rightarrow 00:14:47.380$ What will you do with the information?

NOTE Confidence: 0.85168897

 $00{:}14{:}47{.}380 \dashrightarrow 00{:}14{:}49{.}823$ Now I want to distinguish between analytic

NOTE Confidence: 0.85168897

 $00:14:49.823 \rightarrow 00:14:52.390$ validity and what I call clinical validity.

NOTE Confidence: 0.85168897

00:14:52.390 --> 00:14:55.589 I don't let it validity refers to.

NOTE Confidence: 0.85168897

 $00{:}14{:}55{.}590 \dashrightarrow 00{:}14{:}58{.}110$ Whether or not the result is real.

NOTE Confidence: 0.85168897

 $00{:}14{:}58{.}110 \dashrightarrow 00{:}14{:}59{.}860$ Critical validity refers to whether

NOTE Confidence: 0.85168897

 $00{:}14{:}59{.}860 \dashrightarrow 00{:}15{:}01{.}610$ or not it represents something

NOTE Confidence: 0.85168897

 $00:15:01.666 \rightarrow 00:15:02.936$ about the person or not,

NOTE Confidence: 0.85168897

 $00:15:02.940 \longrightarrow 00:15:04.872$ because in fact in many cases

NOTE Confidence: 0.85168897

 $00:15:04.872 \rightarrow 00:15:06.160$ some of the uncertainty,

NOTE Confidence: 0.85168897

00:15:06.160 --> 00:15:07.770 just as was been described

- NOTE Confidence: 0.85168897
- $00:15:07.770 \longrightarrow 00:15:08.736$ with zero dermatosis,
- NOTE Confidence: 0.85168897
- $00:15:08.740 \longrightarrow 00:15:11.568$ that they didn't really know what the.
- NOTE Confidence: 0.85168897
- $00:15:11.570 \rightarrow 00:15:13.460$ Clinical validity of the information is,
- NOTE Confidence: 0.85168897
- $00:15:13.460 \longrightarrow 00:15:14.387$ in other words,
- NOTE Confidence: 0.85168897
- 00:15:14.387 --> 00:15:17.903 what does it mean to have a certain to be
- NOTE Confidence: 0.85168897
- $00:15:17.903 \rightarrow 00:15:20.447$ one of these certain complement groups?
- NOTE Confidence: 0.85168897
- $00{:}15{:}20{.}450 \dashrightarrow 00{:}15{:}22{.}557$ It was unclear now the issue though.
- NOTE Confidence: 0.85168897
- $00:15:22.560 \longrightarrow 00:15:24.560$ For the people in that study is that
- NOTE Confidence: 0.85168897
- $00{:}15{:}24.560 \dashrightarrow 00{:}15{:}26.354$ it had personal meaning for them
- NOTE Confidence: 0.85168897
- $00{:}15{:}26{.}354 \dashrightarrow 00{:}15{:}28{.}625$ that it mattered to them to know
- NOTE Confidence: 0.85168897
- $00:15:28.625 \rightarrow 00:15:30.420$ what their complement group was,
- NOTE Confidence: 0.85168897
- 00:15:30.420 --> 00:15:32.226 even if it wasn't clinically valid.
- NOTE Confidence: 0.85168897
- $00:15:32.230 \rightarrow 00:15:34.138$ So these notions of clinical validity NOTE Confidence: 0.85168897
- $00{:}15{:}34{.}138 \dashrightarrow 00{:}15{:}35{.}757$ and personal and personal meeting NOTE Confidence: 0.85168897
- $00{:}15{:}35{.}757 \dashrightarrow 00{:}15{:}37{.}485$ are two other dimensions that are
- NOTE Confidence: 0.85168897

 $00:15:37.485 \rightarrow 00:15:39.269$ important as we think about genetic

NOTE Confidence: 0.85168897

 $00{:}15{:}39{.}269 \dashrightarrow 00{:}15{:}41{.}285$ results and why people may want them,

NOTE Confidence: 0.85168897

 $00:15:41.290 \longrightarrow 00:15:42.795$ why clinicians may be reluctant

NOTE Confidence: 0.85168897

 $00:15:42.795 \longrightarrow 00:15:43.698$ to return them,

NOTE Confidence: 0.85168897

 $00:15:43.700 \longrightarrow 00:15:45.932$ but these were the issues we

NOTE Confidence: 0.85168897

 $00:15:45.932 \longrightarrow 00:15:47.420$ have to think about.

NOTE Confidence: 0.85168897

 $00:15:47.420 \longrightarrow 00:15:49.758$ The second set of issues are right

NOTE Confidence: 0.85168897

 $00{:}15{:}49.758 \dashrightarrow 00{:}15{:}51.155$ described as contextual considerations

NOTE Confidence: 0.85168897

00:15:51.155 --> 00:15:53.827 and that refers to in a research study

NOTE Confidence: 0.85168897

 $00{:}15{:}53{.}827 \dashrightarrow 00{:}15{:}55{.}947$ with the relationship might be between NOTE Confidence: 0.85168897

 $00{:}15{:}55{.}947 \dashrightarrow 00{:}15{:}58{.}034$ the researcher and the the participants.

NOTE Confidence: 0.85168897

 $00{:}15{:}58{.}034 \dashrightarrow 00{:}16{:}00{.}158$ Is this a longitudinal study which

NOTE Confidence: 0.85168897

00:16:00.158 --> 00:16:02.808 was as it was occurring at the NIH?

NOTE Confidence: 0.85168897

00:16:02.810 --> 00:16:05.141 Is this a biobank study in which

NOTE Confidence: 0.85168897

 $00{:}16{:}05{.}141 \dashrightarrow 00{:}16{:}06{.}776$ people donated samples that are

NOTE Confidence: 0.85168897

 $00:16:06.776 \longrightarrow 00:16:08.486$ being used by people 10 years

- NOTE Confidence: 0.85168897
- 00:16:08.486 --> 00:16:10.329 later at another institution,
- NOTE Confidence: 0.85168897
- $00{:}16{:}10{.}330 \dashrightarrow 00{:}16{:}12{.}724$ or is there something else like this?
- NOTE Confidence: 0.85168897
- 00:16:12.730 --> 00:16:13.054 Also,
- NOTE Confidence: 0.85168897
- $00{:}16{:}13.054 \dashrightarrow 00{:}16{:}15.322$ what is the capacity of the research
- NOTE Confidence: 0.85168897
- $00:16:15.322 \rightarrow 00:16:17.460$ team to provide those results?
- NOTE Confidence: 0.85168897
- $00{:}16{:}17{.}460 \dashrightarrow 00{:}16{:}20{.}099$ If we assume that there are certain
- NOTE Confidence: 0.85168897
- $00:16:20.099 \rightarrow 00:16:21.230$ expectations regarding consent
- NOTE Confidence: 0.84569705
- $00:16:21.291 \rightarrow 00:16:23.072$ counseling, things like that.
- NOTE Confidence: 0.84569705
- $00{:}16{:}23.072 \dashrightarrow 00{:}16{:}25.376$ What if the researchers are being
- NOTE Confidence: 0.84569705
- $00{:}16{:}25{.}376 \dashrightarrow 00{:}16{:}27{.}969$ what the research is being done in
- NOTE Confidence: 0.84569705
- $00:16:27.969 \rightarrow 00:16:30.389$ a laboratory where the folks don't
- NOTE Confidence: 0.84569705
- $00{:}16{:}30{.}389 \dashrightarrow 00{:}16{:}32{.}684$ have that capacity and another
- NOTE Confidence: 0.84569705
- $00{:}16{:}32.684 \dashrightarrow 00{:}16{:}34.878$ consideration is whether this is
- NOTE Confidence: 0.84569705
- $00{:}16{:}34.878 \dashrightarrow 00{:}16{:}36.898$ uniquely available only through research,
- NOTE Confidence: 0.84569705
- $00:16:36.900 \longrightarrow 00:16:38.895$ or whether this is otherwise
- NOTE Confidence: 0.84569705

00:16:38.895 - 00:16:40.491 easily accessible through any

NOTE Confidence: 0.84569705

 $00{:}16{:}40{.}491 \dashrightarrow 00{:}16{:}42{.}168$ variety of other approaches,

NOTE Confidence: 0.84569705

 $00{:}16{:}42{.}170$ --> $00{:}16{:}44{.}905$ including Now these days direct NOTE Confidence: 0.84569705

00:16:44.905 --> 00:16:47.640 to consumer marketing as well.

NOTE Confidence: 0.84569705

 $00{:}16{:}47.640 \dashrightarrow 00{:}16{:}49.845$ So I want to Fast forward now

NOTE Confidence: 0.84569705

 $00:16:49.845 \longrightarrow 00:16:51.920$ briefly to a collaboration.

NOTE Confidence: 0.84569705

00:16:51.920 --> 00:16:53.894 I began a number of years ago

NOTE Confidence: 0.84569705

 $00{:}16{:}53.894 \dashrightarrow 00{:}16{:}56.185$ with a bunch of neurologists who

NOTE Confidence: 0.84569705

 $00{:}16{:}56{.}185 \dashrightarrow 00{:}16{:}58{.}470$ were interested in thinking about

NOTE Confidence: 0.84569705

 $00{:}16{:}58{.}470 \dashrightarrow 00{:}17{:}00{.}727$ a longitudinal study of chronic

NOTE Confidence: 0.84569705

00:17:00.727 --> 00:17:02.419 kidney disease in children,

NOTE Confidence: 0.84569705

 $00:17:02.420 \longrightarrow 00:17:04.760$ and this study began in 2004,

NOTE Confidence: 0.84569705

00:17:04.760 --> 00:17:07.088 and we spoke initially like 2018,

NOTE Confidence: 0.84569705

 $00{:}17{:}07{.}090 \dashrightarrow 00{:}17{:}10{.}690$ and the issue was based upon.

NOTE Confidence: 0.84569705

 $00{:}17{:}10.690 \dashrightarrow 00{:}17{:}12.922$ An evolving consensus that we should

NOTE Confidence: 0.84569705

 $00:17:12.922 \longrightarrow 00:17:15.593$ only we should not be routinely

00:17:15.593 --> 00:17:17.258 disclosing research results,

NOTE Confidence: 0.84569705

00:17:17.260 --> 00:17:20.032 particularly if there was the lack of

NOTE Confidence: 0.84569705

 $00:17:20.032 \rightarrow 00:17:22.520$ analytic ability and clinical validity,

NOTE Confidence: 0.84569705

 $00{:}17{:}22.520$ --> $00{:}17{:}24.710$ and therefore is probably appropriate.

NOTE Confidence: 0.84569705

 $00{:}17{:}24.710$ --> $00{:}17{:}27.446$ Most research to set expectations that NOTE Confidence: 0.84569705

 $00:17:27.446 \longrightarrow 00:17:30.679$ you would not be getting results.

NOTE Confidence: 0.84569705

 $00:17:30.680 \rightarrow 00:17:32.660$ That this group is the dilemma.

NOTE Confidence: 0.84569705

 $00:17:32.660 \rightarrow 00:17:34.285$ Because suddenly as they started

NOTE Confidence: 0.84569705

00:17:34.285 --> 00:17:36.290 doing more analysis over the years,

NOTE Confidence: 0.84569705

 $00{:}17{:}36{.}290 \dashrightarrow 00{:}17{:}38{.}280$ they started realizing through one

NOTE Confidence: 0.84569705

 $00{:}17{:}38{.}280 \dashrightarrow 00{:}17{:}40{.}590$ of the analytics studies they did.

NOTE Confidence: 0.84569705

 $00{:}17{:}40.590 \dashrightarrow 00{:}17{:}42.690$ That the diagnosis of the cause of

NOTE Confidence: 0.84569705

00:17:42.690 --> 00:17:44.241 the contingency that was provided

NOTE Confidence: 0.84569705

00:17:44.241 $\operatorname{-->}$ 00:17:46.306 clinically was not the same as they

NOTE Confidence: 0.84569705

 $00:17:46.306 \rightarrow 00:17:48.567$ found out through their genetic research,

 $00{:}17{:}48.570 \dashrightarrow 00{:}17{:}50.551$ and so it raised question of what

NOTE Confidence: 0.84569705

00:17:50.551 --> 00:17:52.710 they should do in this circumstance,

NOTE Confidence: 0.84569705

 $00{:}17{:}52.710 \dashrightarrow 00{:}17{:}54.290$ particularly when the consent forms

NOTE Confidence: 0.84569705

 $00{:}17{:}54.290 \dashrightarrow 00{:}17{:}56.859$ said we will not disclose any results there.

NOTE Confidence: 0.84569705

 $00:17:56.860 \longrightarrow 00:17:57.428$ Question is,

NOTE Confidence: 0.84569705

 $00{:}17{:}57{.}428 \dashrightarrow 00{:}17{:}59{.}132$ what obligations do we have to

NOTE Confidence: 0.84569705

 $00:17:59.132 \rightarrow 00:18:00.690$ think about returning results,

NOTE Confidence: 0.84569705

 $00:18:00.690 \longrightarrow 00:18:01.962$ disclosing results when we

NOTE Confidence: 0.84569705

 $00:18:01.962 \longrightarrow 00:18:03.234$ told them they wouldn't?

NOTE Confidence: 0.84569705

 $00:18:03.240 \longrightarrow 00:18:04.830$ But this might be actually

NOTE Confidence: 0.84569705

 $00{:}18{:}04{.}830 \dashrightarrow 00{:}18{:}06{.}102$ relevant to their management.

NOTE Confidence: 0.84569705

 $00{:}18{:}06{.}110 \dashrightarrow 00{:}18{:}08{.}891$ Now I won't be spending a lot of time

NOTE Confidence: 0.84569705

 $00{:}18{:}08{.}891 \dashrightarrow 00{:}18{:}10{.}998$ getting into how we address this.

NOTE Confidence: 0.84569705

 $00:18:11.000 \rightarrow 00:18:13.065$ Other than simply say one of the

NOTE Confidence: 0.84569705

 $00:18:13.065 \rightarrow 00:18:15.427$ approaches we use and we recommend it was,

NOTE Confidence: 0.84569705

 $00:18:15.430 \rightarrow 00:18:15.736$ well,

 $00:18:15.736 \longrightarrow 00:18:17.572$ talk to the participants and see

NOTE Confidence: 0.84569705

 $00:18:17.572 \rightarrow 00:18:19.706$ in general what they think of this

NOTE Confidence: 0.84569705

00:18:19.706 --> 00:18:21.440 issue and what advice they would NOTE Confidence: 0.84569705

to 11 connuclee: 0.01000100

00:18:21.500 --> 00:18:23.168 give and was interesting here is

NOTE Confidence: 0.84569705

00:18:23.168 --> 00:18:25.102 that there was a range of different

NOTE Confidence: 0.84569705

 $00{:}18{:}25{.}102 \dashrightarrow 00{:}18{:}26{.}746$ results and some I mentioned to

NOTE Confidence: 0.84569705

 $00{:}18{:}26.746 \dashrightarrow 00{:}18{:}28.635$ you that there were some results

NOTE Confidence: 0.84569705

00:18:28.635 --> 00:18:29.879 related to kidney disease,

NOTE Confidence: 0.84569705

 $00{:}18{:}29{.}880 \dashrightarrow 00{:}18{:}31{.}777$ but there are other results that were

NOTE Confidence: 0.84569705

 $00{:}18{:}31.777 \dashrightarrow 00{:}18{:}33.420$ more ambiguous and more extraneous.

NOTE Confidence: 0.84569705

 $00:18:33.420 \longrightarrow 00:18:36.356$ They had that related to the way in

NOTE Confidence: 0.84569705

 $00{:}18{:}36{.}356 \dashrightarrow 00{:}18{:}39{.}187$ which various results had some impact on.

NOTE Confidence: 0.84569705

 $00:18:39.190 \longrightarrow 00:18:41.248$ On IQ at on learning and there

NOTE Confidence: 0.84569705

 $00{:}18{:}41{.}248 \dashrightarrow 00{:}18{:}43{.}531$ was much more interest among the

NOTE Confidence: 0.84569705

 $00{:}18{:}43.531 \dashrightarrow 00{:}18{:}45.299$ participants in those results

 $00:18:45.299 \rightarrow 00:18:47.830$ related to kidney disease and less

NOTE Confidence: 0.84569705

 $00{:}18{:}47{.}830 \dashrightarrow 00{:}18{:}50{.}104$ interest in the ones regarding IQ.

NOTE Confidence: 0.84569705

00:18:50.110 --> 00:18:53.090 So I want to put this into context so you

NOTE Confidence: 0.84569705

 $00:18:53.169 \rightarrow 00:18:56.299$ can be thinking so you have a sense of how.

NOTE Confidence: 0.84569705

 $00{:}18{:}56{.}300 \dashrightarrow 00{:}18{:}57{.}825$ The research issues have been

NOTE Confidence: 0.84569705

 $00{:}18{:}57.825 \dashrightarrow 00{:}18{:}59.045$ evolving over the decades,

NOTE Confidence: 0.84569705

 $00{:}18{:}59{.}050 \dashrightarrow 00{:}19{:}01{.}948$ and at this point I want to shift gears

NOTE Confidence: 0.84569705

 $00{:}19{:}01{.}948 \dashrightarrow 00{:}19{:}04{.}939$ a little bit and I'm going to share

NOTE Confidence: 0.84569705

 $00{:}19{:}04{.}939 \dashrightarrow 00{:}19{:}08{.}180$ with you over the next 20 minutes or so.

NOTE Confidence: 0.84569705

 $00{:}19{:}08{.}180 \dashrightarrow 00{:}19{:}11{.}366$ Two different projects that I've been

NOTE Confidence: 0.84569705

 $00:19:11.366 \rightarrow 00:19:15.240$ involved with that that involve sequencing.

NOTE Confidence: 0.84569705

 $00{:}19{:}15{.}240 \dashrightarrow 00{:}19{:}16{.}997$ And the first one is sequencing in

NOTE Confidence: 0.84569705

 $00{:}19{:}16{.}997 \dashrightarrow 00{:}19{:}18{.}730$ the context of carrier screening.

NOTE Confidence: 0.84569705

 $00:19:18.730 \longrightarrow 00:19:19.021$ Now,

NOTE Confidence: 0.84569705

 $00{:}19{:}19{.}021 \dashrightarrow 00{:}19{:}21{.}058$ if you recall from my first slide,

NOTE Confidence: 0.84569705

 $00:19:21.060 \rightarrow 00:19:23.868$ I showed you from.

- NOTE Confidence: 0.84569705
- $00:19:23.870 \longrightarrow 00:19:25.904$ From the paper on CF carrier
- NOTE Confidence: 0.84569705
- $00{:}19{:}25{.}904 \dashrightarrow 00{:}19{:}27{.}260$ testing is that one
- NOTE Confidence: 0.784771299999999
- $00:19:27.338 \longrightarrow 00:19:29.462$ of the five that was actually
- NOTE Confidence: 0.784771299999999
- $00:19:29.462 \longrightarrow 00:19:31.757$ the very first paper ever wrote
- NOTE Confidence: 0.784771299999999
- $00{:}19{:}31{.}757 \dashrightarrow 00{:}19{:}33{.}777$ was about CF carrier testing.
- NOTE Confidence: 0.784771299999999
- $00{:}19{:}33.780 \dashrightarrow 00{:}19{:}36.716$ Over the next decade by as CF terror
- NOTE Confidence: 0.784771299999999
- $00:19:36.716 \longrightarrow 00:19:38.761$ testing involved, I became a.
- NOTE Confidence: 0.784771299999999
- 00:19:38.761 --> 00:19:41.383 Mixed critic supporter of carrier testing.
- NOTE Confidence: 0.784771299999999
- $00{:}19{:}41{.}390 \dashrightarrow 00{:}19{:}43{.}910$ I was critical of the enthusiasm to
- NOTE Confidence: 0.784771299999999
- 00:19:43.910 --> 00:19:46.358 do without research, but I was in,
- NOTE Confidence: 0.784771299999999
- 00:19:46.358 --> 00:19:49.226 but I was supportive of research to learn
- NOTE Confidence: 0.784771299999999
- $00{:}19{:}49{.}226 \dashrightarrow 00{:}19{:}51{.}823$ how to do CF carrier testing better.
- NOTE Confidence: 0.784771299999999
- $00{:}19{:}51{.}830 \dashrightarrow 00{:}19{:}55{.}043$ One of the concerns I had about CF carrier
- NOTE Confidence: 0.784771299999999
- $00{:}19{:}55{.}043 \dashrightarrow 00{:}19{:}57{.}398$ testing as quickly as a pulmonologist
- NOTE Confidence: 0.784771299999999
- $00{:}19{:}57{.}398 \dashrightarrow 00{:}20{:}00{.}258$ is that it wasn't really clear to me.
- NOTE Confidence: 0.784771299999999
$00:20:00.260 \rightarrow 00:20:02.794$ What one should do with that information,

NOTE Confidence: 0.784771299999999

 $00{:}20{:}02{.}800 \dashrightarrow 00{:}20{:}04{.}248$ particularly as a disease

NOTE Confidence: 0.784771299999999

00:20:04.248 --> 00:20:05.334 like cystic fibrosis,

NOTE Confidence: 0.784771299999999

 $00:20:05.340 \rightarrow 00:20:07.145$ was evolving clinically and I

NOTE Confidence: 0.784771299999999

00:20:07.145 -> 00:20:09.340 was worried that when this went,

NOTE Confidence: 0.784771299999999

00:20:09.340 --> 00:20:11.385 information about cystic fibrosis would

NOTE Confidence: 0.784771299999999

 $00{:}20{:}11.385 \dashrightarrow 00{:}20{:}13.863$ be presented to individuals who are

NOTE Confidence: 0.784771299999999

 $00:20:13.863 \rightarrow 00:20:16.107$ seeking carrier testing that both the

NOTE Confidence: 0.784771299999999

00:20:16.107 --> 00:20:18.170 obstetricians who might be doing the

NOTE Confidence: 0.784771299999999

 $00{:}20{:}18.170 \dashrightarrow 00{:}20{:}19.928$ testing and the patients might have

NOTE Confidence: 0.784771299999999

 $00{:}20{:}19{.}928 \dashrightarrow 00{:}20{:}22{.}396$ no idea of what cystic fibrosis was.

NOTE Confidence: 0.784771299999999

 $00{:}20{:}22{.}400 \dashrightarrow 00{:}20{:}24{.}326$ I actually reflected back to when

NOTE Confidence: 0.784771299999999

 $00{:}20{:}24.326 \dashrightarrow 00{:}20{:}26.918$ I was a medical student and I

NOTE Confidence: 0.784771299999999

00:20:26.918 --> 00:20:28.566 thought about cystic fibrosis,

NOTE Confidence: 0.784771299999999

00:20:28.570 --> 00:20:30.700 multiple sclerosis and muscular dystrophy.

NOTE Confidence: 0.784771299999999

00:20:30.700 --> 00:20:32.320 I remember thinking to myself

- NOTE Confidence: 0.784771299999999
- $00:20:32.320 \longrightarrow 00:20:33.616$ which one is which.
- NOTE Confidence: 0.784771299999999
- $00:20:33.620 \rightarrow 00:20:35.402$ These are really confusing words and
- NOTE Confidence: 0.784771299999999
- $00{:}20{:}35{.}402 \dashrightarrow 00{:}20{:}37{.}535$ I can't tell which of these diseases
- NOTE Confidence: 0.784771299999999
- $00{:}20{:}37{.}535 \dashrightarrow 00{:}20{:}39{.}614$ means which thing 'cause so I worried
- NOTE Confidence: 0.784771299999999
- $00{:}20{:}39.668 \dashrightarrow 00{:}20{:}41.870$ very much a cystic fibrosis carrier
- NOTE Confidence: 0.784771299999999
- $00:20:41.870 \longrightarrow 00:20:43.663$ testing was actually potentially well.
- NOTE Confidence: 0.784771299999999
- 00:20:43.663 --> 00:20:45.278 It could be done well.
- NOTE Confidence: 0.784771299999999
- $00:20:45.280 \longrightarrow 00:20:47.226$ It could also be done poorly and
- NOTE Confidence: 0.784771299999999
- $00{:}20{:}47{.}226 \dashrightarrow 00{:}20{:}49{.}687$ one of the places in my opinion that
- NOTE Confidence: 0.784771299999999
- $00:20:49.687 \rightarrow 00:20:52.084$ in the 1990s was doing it fairly
- NOTE Confidence: 0.784771299999999
- 00:20:52.084 --> 00:20:54.024 poorly was Kaiser in California,
- NOTE Confidence: 0.784771299999999
- $00:20:54.030 \rightarrow 00:20:55.605$ where they were routinely offering
- NOTE Confidence: 0.784771299999999
- $00{:}20{:}55{.}605 \dashrightarrow 00{:}20{:}57{.}893$ this without a whole lot of discussion
- NOTE Confidence: 0.784771299999999
- $00{:}20{:}57{.}893 \dashrightarrow 00{:}20{:}59{.}855$ without a whole lot of information,
- NOTE Confidence: 0.784771299999999
- $00{:}20{:}59{.}860 \dashrightarrow 00{:}21{:}02{.}086$ and they demonstrated in that setting.
- NOTE Confidence: 0.784771299999999

 $00:21:02.090 \rightarrow 00:21:03.966$ But there are lots of people who

NOTE Confidence: 0.784771299999999

 $00:21:03.966 \longrightarrow 00:21:05.706$ were interested in it and that

NOTE Confidence: 0.784771299999999

00:21:05.706 --> 00:21:06.878 didn't surprise me because.

NOTE Confidence: 0.784771299999999

00:21:06.880 --> 00:21:07.058 Yeah,

NOTE Confidence: 0.784771299999999

 $00:21:07.058 \longrightarrow 00:21:08.482$ if you present it in a way that

NOTE Confidence: 0.784771299999999

00:21:08.482 --> 00:21:09.949 makes it sound very straightforward,

NOTE Confidence: 0.784771299999999

 $00:21:09.950 \longrightarrow 00:21:11.960$ of course people will say yes.

NOTE Confidence: 0.784771299999999

00:21:11.960 --> 00:21:14.320 So why am I telling you that background

NOTE Confidence: 0.784771299999999

 $00:21:14.320 \rightarrow 00:21:16.867$ now and in relationship to this paper?

NOTE Confidence: 0.784771299999999

00:21:16.870 --> 00:21:17.135 Wells,

NOTE Confidence: 0.784771299999999

00:21:17.135 --> 00:21:19.520 because the about in 2013 I got a phone

NOTE Confidence: 0.784771299999999

 $00:21:19.582 \rightarrow 00:21:21.427$ call from a genetic epidemiologist

NOTE Confidence: 0.784771299999999

 $00{:}21{:}21{.}427 \dashrightarrow 00{:}21{:}24{.}001$ named Katrina Goddard whom we end up

NOTE Confidence: 0.784771299999999

 $00:21:24.001 \rightarrow 00:21:26.005$ becoming Kopi eyes with this large

NOTE Confidence: 0.784771299999999

 $00{:}21{:}26.005 \dashrightarrow 00{:}21{:}27.868$ project together that was looking at

NOTE Confidence: 0.784771299999999

 $00:21:27.868 \rightarrow 00:21:29.353$ doing genomic based carrier testing

- NOTE Confidence: 0.784771299999999
- $00:21:29.353 \rightarrow 00:21:31.474$ for people who are already seeking
- NOTE Confidence: 0.784771299999999
- $00{:}21{:}31{.}474 \dashrightarrow 00{:}21{:}32{.}886$ cystic fibrosis carrier testing.
- NOTE Confidence: 0.784771299999999
- $00{:}21{:}32{.}890 \dashrightarrow 00{:}21{:}35{.}506$ The first thing I said to Katrina when
- NOTE Confidence: 0.784771299999999
- $00:21:35.506 \rightarrow 00:21:38.115$ we first spoke on the phone is that,
- NOTE Confidence: 0.784771299999999
- 00:21:38.120 --> 00:21:38.802 you know,
- NOTE Confidence: 0.784771299999999
- 00:21:38.802 --> 00:21:41.530 Are you sure you want me to work
- NOTE Confidence: 0.784771299999999
- $00:21:41.611 \longrightarrow 00:21:42.910$ on this project?
- NOTE Confidence: 0.784771299999999
- $00:21:42.910 \longrightarrow 00:21:46.760$ I've been pretty sceptical of.
- NOTE Confidence: 0.784771299999999
- 00:21:46.760 --> 00:21:47.904 Cystic fibrosis carrier testing,
- NOTE Confidence: 0.784771299999999
- 00:21:47.904 --> 00:21:48.476 particularly Kaiser,
- NOTE Confidence: 0.784771299999999
- $00{:}21{:}48{.}480 \dashrightarrow 00{:}21{:}50{.}489$ and she continue to work at Kaiser
- NOTE Confidence: 0.784771299999999
- $00:21:50.489 \longrightarrow 00:21:52.211$ in an Oregon, which is yeah,
- NOTE Confidence: 0.784771299999999
- 00:21:52.211 --> 00:21:53.646 I know that in fact,
- NOTE Confidence: 0.784771299999999
- 00:21:53.650 --> 00:21:55.282 that's actually why I want you
- NOTE Confidence: 0.784771299999999
- $00:21:55.282 \rightarrow 00:21:57.090$ to help me with this study.
- NOTE Confidence: 0.784771299999999

00:21:57.090 --> 00:21:59.288 I want to make sure we're doing

NOTE Confidence: 0.784771299999999

 $00{:}21{:}59{.}288 \dashrightarrow 00{:}22{:}01{.}298$ it ethically and I want to do

NOTE Confidence: 0.784771299999999

 $00:22:01.298 \longrightarrow 00:22:03.120$ this the very best way we can.

NOTE Confidence: 0.784771299999999

 $00:22:03.120 \longrightarrow 00:22:03.792$ So we were.

NOTE Confidence: 0.784771299999999

 $00{:}22{:}03.792 \dashrightarrow 00{:}22{:}06.140$ We wrote a grant and we were became part

NOTE Confidence: 0.784771299999999

 $00:22:06.140 \longrightarrow 00:22:08.288$ of what's called the Caesar Consortium,

NOTE Confidence: 0.784771299999999

 $00{:}22{:}08{.}290 \dashrightarrow 00{:}22{:}09{.}856$ which stands for the stands for

NOTE Confidence: 0.784771299999999

 $00:22:09.856 \longrightarrow 00:22:10.900$ the clinical sequencing exploratory

NOTE Confidence: 0.784771299999999

00:22:10.948 --> 00:22:11.730 Research Consortium,

NOTE Confidence: 0.784771299999999

 $00:22:11.730 \longrightarrow 00:22:13.380$ which were group of studies that

NOTE Confidence: 0.784771299999999

 $00:22:13.380 \rightarrow 00:22:15.169$ we're trying to address questions of.

NOTE Confidence: 0.8142179

 $00:22:15.170 \longrightarrow 00:22:18.070$ How is it that?

NOTE Confidence: 0.8142179

 $00:22:18.070 \rightarrow 00:22:19.984$ Clinical sequencing can be introduced into

NOTE Confidence: 0.8142179

00:22:19.984 --> 00:22:22.049 clinical practice in a meaningful way.

NOTE Confidence: 0.8142179

 $00:22:22.050 \longrightarrow 00:22:23.398$ What is the evidence?

NOTE Confidence: 0.8142179

 $00:22:23.398 \rightarrow 00:22:26.040$ We need to determine how to do this,

 $00:22:26.040 \rightarrow 00:22:28.929$ and so we wrote a grant regarding how to

NOTE Confidence: 0.8142179

 $00{:}22{:}28{.}929 \dashrightarrow 00{:}22{:}32{.}070$ do this for cystic for carrier testing

NOTE Confidence: 0.8142179

 $00:22:32.070 \longrightarrow 00:22:34.963$ using sequencing and what I want to

NOTE Confidence: 0.8142179

 $00:22:34.963 \longrightarrow 00:22:37.266$ do is share with you briefly some.

NOTE Confidence: 0.8142179

 $00:22:37.270 \longrightarrow 00:22:39.503$ Some of the key things we learn

NOTE Confidence: 0.8142179

 $00{:}22{:}39{.}503 \dashrightarrow 00{:}22{:}41{.}919$ from this study and those key points

NOTE Confidence: 0.8142179

 $00{:}22{:}41{.}919 \dashrightarrow 00{:}22{:}44{.}469$ are based upon this paper that was

NOTE Confidence: 0.8142179

 $00:22:44.469 \longrightarrow 00:22:46.857$ published in 2018 and Health Affairs.

NOTE Confidence: 0.8142179

 $00:22:46.860 \longrightarrow 00:22:48.415$ Really, the lessons that we

NOTE Confidence: 0.8142179

 $00:22:48.415 \rightarrow 00:22:50.478$ learned from doing this study for

NOTE Confidence: 0.8142179

00:22:50.478 --> 00:22:51.828 reproductive decision making.

NOTE Confidence: 0.8142179

00:22:51.830 --> 00:22:54.470 So I briefly describe the study to you

NOTE Confidence: 0.8142179

 $00:22:54.470 \longrightarrow 00:22:57.505$ in a couple of this one slide here.

NOTE Confidence: 0.8142179

 $00{:}22{:}57{.}510 \dashrightarrow 00{:}22{:}59{.}205$ Essentially what happened is that

NOTE Confidence: 0.8142179

 $00:22:59.205 \longrightarrow 00:23:01.306$ for people who on their own's

 $00:23:01.306 \longrightarrow 00:23:03.340$ sought out a clinical carrier test

NOTE Confidence: 0.8142179

 $00:23:03.340 \longrightarrow 00:23:04.960$ often for cystic fibrosis,

NOTE Confidence: 0.8142179

 $00{:}23{:}04{.}960 \dashrightarrow 00{:}23{:}07{.}640$ we approach them an and.

NOTE Confidence: 0.8142179

 $00:23:07.640 \longrightarrow 00:23:09.152$ To roll on the study and the

NOTE Confidence: 0.8142179

 $00{:}23{:}09{.}152 \dashrightarrow 00{:}23{:}10{.}630$ way this would work is that if

NOTE Confidence: 0.8142179

 $00{:}23{:}10.630 \dashrightarrow 00{:}23{:}12.160$ they agree to be in the study.

NOTE Confidence: 0.8142179

00:23:12.160 --> 00:23:14.086 Half of them will get sequencing,

NOTE Confidence: 0.8142179

 $00:23:14.090 \longrightarrow 00:23:16.370$ other half would just go on with their

NOTE Confidence: 0.8142179

00:23:16.370 --> 00:23:18.160 usual life other than completing a

NOTE Confidence: 0.8142179

 $00{:}23{:}18{.}160 \dashrightarrow 00{:}23{:}21{.}101$ bunch of surveys for us as well as us NOTE Confidence: 0.8142179

 $00:23:21.101 \longrightarrow 00:23:22.613$ evaluating their health utilization

NOTE Confidence: 0.8142179

 $00{:}23{:}22{.}613 \dashrightarrow 00{:}23{:}25{.}360$ within Kaiser over the next year or so.

NOTE Confidence: 0.8142179

 $00{:}23{:}25{.}360 \dashrightarrow 00{:}23{:}27{.}970$ And So what I want to show you here is

NOTE Confidence: 0.8142179

 $00{:}23{:}28.043 \dashrightarrow 00{:}23{:}30.395$ briefly the findings and the important

NOTE Confidence: 0.8142179

 $00{:}23{:}30{.}395 \dashrightarrow 00{:}23{:}33{.}409$ thing here is that we were not we were.

NOTE Confidence: 0.8142179

 $00:23:33.410 \longrightarrow 00:23:34.698$ This is actually important

- NOTE Confidence: 0.8142179
- $00:23:34.698 \rightarrow 00:23:35.664$ about about sequencing.

00:23:35.670 --> 00:23:36.633 Sequencing doesn't mean

NOTE Confidence: 0.8142179

00:23:36.633 --> 00:23:37.917 like the movie Gattaca.

NOTE Confidence: 0.8142179

00:23:37.920 --> 00:23:39.530 You see everything you see

NOTE Confidence: 0.8142179

 $00:23:39.530 \longrightarrow 00:23:40.818$ that everything that's there,

NOTE Confidence: 0.8142179

 $00:23:40.820 \longrightarrow 00:23:42.878$ you have to make a decision.

NOTE Confidence: 0.8142179

 $00:23:42.880 \rightarrow 00:23:45.976$ What information do you want to look at?

NOTE Confidence: 0.8142179

 $00{:}23{:}45{.}980 \dashrightarrow 00{:}23{:}48{.}662$ So we had to for this study that was

NOTE Confidence: 0.8142179

 $00{:}23{:}48.662 \dashrightarrow 00{:}23{:}51.396$ done in collaboration with GAIL Jarvik

NOTE Confidence: 0.8142179

 $00{:}23{:}51{.}396 \dashrightarrow 00{:}23{:}54{.}215$ at the University of Washington is

NOTE Confidence: 0.8142179

 $00{:}23{:}54{.}215 \dashrightarrow 00{:}23{:}56{.}483$ decide how many conditions would we

NOTE Confidence: 0.8142179

 $00{:}23{:}56{.}483 \dashrightarrow 00{:}23{:}58{.}838$ be looking at for carrier testing.

NOTE Confidence: 0.8142179

00:23:58.838 --> 00:24:01.274 There were many places that offered

NOTE Confidence: 0.8142179

 $00:24:01.274 \longrightarrow 00:24:03.438$ 567 some place as many as 100,

NOTE Confidence: 0.8142179

 $00:24:03.440 \longrightarrow 00:24:06.051$ and for this study we ended up

 $00:24:06.051 \longrightarrow 00:24:07.650$ identifying 700 conditions to

NOTE Confidence: 0.8142179

 $00:24:07.650 \longrightarrow 00:24:08.877$ offer carrier testing.

NOTE Confidence: 0.8142179

00:24:08.880 --> 00:24:10.023 4 and Additionally,

NOTE Confidence: 0.8142179

 $00{:}24{:}10.023 \dashrightarrow 00{:}24{:}12.309$ we also identified people who had

NOTE Confidence: 0.8142179

00:24:12.309 --> 00:24:14.030 secondary findings in AutoZone.

NOTE Confidence: 0.8142179

00:24:14.030 --> 00:24:15.795 Dominant conditions that by this

NOTE Confidence: 0.8142179

 $00{:}24{:}15.795 \dashrightarrow 00{:}24{:}17.560$ point we're being recommended by

NOTE Confidence: 0.8142179

 $00{:}24{:}17.623 \dashrightarrow 00{:}24{:}19.498$ the American College of Medical

NOTE Confidence: 0.8142179

00:24:19.498 --> 00:24:21.373 Genetics because they were considered

NOTE Confidence: 0.8142179

 $00:24:21.433 \longrightarrow 00:24:22.737$ to be highly actionable,

NOTE Confidence: 0.8142179

 $00{:}24{:}22{.}740 \dashrightarrow 00{:}24{:}25{.}134$ i.e things related to cancer or heart

NOTE Confidence: 0.8142179

 $00:24:25.134 \rightarrow 00:24:27.817$ disease or might be sort of interventions.

NOTE Confidence: 0.8142179

 $00{:}24{:}27.820 \dashrightarrow 00{:}24{:}29.998$ So I want to briefly say,

NOTE Confidence: 0.8142179

00:24:30.000 --> 00:24:30.696 you know,

NOTE Confidence: 0.8142179

 $00{:}24{:}30.696 \dashrightarrow 00{:}24{:}32.784$ described to you and have you

NOTE Confidence: 0.8142179

 $00:24:32.784 \longrightarrow 00:24:35.371$ see how we approach this and to

- NOTE Confidence: 0.8142179
- $00:24:35.371 \rightarrow 00:24:37.630$ show you what our results were.

 $00:24:37.630 \longrightarrow 00:24:39.640$ We actually found that there were

NOTE Confidence: 0.8142179

00:24:39.640 --> 00:24:42.340 roughly out of the 300 individuals who,

NOTE Confidence: 0.8142179

 $00{:}24{:}42{.}340 \dashrightarrow 00{:}24{:}43{.}796$ out of 133 women.

NOTE Confidence: 0.8142179

 $00:24:43.796 \longrightarrow 00:24:45.616$ Who enrolled in the study?

NOTE Confidence: 0.8142179

 $00:24:45.620 \longrightarrow 00:24:47.564$ There were 12 high risk couples

NOTE Confidence: 0.8142179

00:24:47.564 --> 00:24:49.220 identified who are risk of

NOTE Confidence: 0.8142179

 $00{:}24{:}49{.}220 \dashrightarrow 00{:}24{:}50{.}990$ having a child with an AutoZone.

NOTE Confidence: 0.8142179

 $00{:}24{:}50{.}990 \dashrightarrow 00{:}24{:}52{.}714$ Will recessive condition and

NOTE Confidence: 0.8142179

 $00{:}24{:}52{.}714 \dashrightarrow 00{:}24{:}54{.}869$ three who had excellent conditions

NOTE Confidence: 0.8142179

 $00:24:54.869 \longrightarrow 00:24:57.342$ for which they were also risk of

NOTE Confidence: 0.8142179

 $00{:}24{:}57{.}342 \dashrightarrow 00{:}24{:}59{.}500$ having a child because it only one.

NOTE Confidence: 0.8142179

 $00{:}24{:}59{.}500 \dashrightarrow 00{:}25{:}02{.}188$ Up only the one that was necessary to

NOTE Confidence: 0.8142179

 $00{:}25{:}02{.}188 \dashrightarrow 00{:}25{:}05{.}585$ be a carrier for them to have a child.

NOTE Confidence: 0.8142179

 $00:25:05.590 \rightarrow 00:25:07.726$ We also saw several secondary conditions,

00:25:07.730 --> 00:25:09.520 including actually 7-7 people had

NOTE Confidence: 0.8142179

 $00{:}25{:}09{.}520 \dashrightarrow 00{:}25{:}10{.}594$ autosomal dominant conditions.

NOTE Confidence: 0.8142179

00:25:10.600 --> 00:25:13.127 So gives you some idea of the

NOTE Confidence: 0.8142179

 $00:25:13.127 \longrightarrow 00:25:14.210$ scope of what

NOTE Confidence: 0.8073976

 $00:25:14.298 \rightarrow 00:25:18.074$ you can learn from sequencing in the context.

NOTE Confidence: 0.8073976

 $00{:}25{:}18.080 \dashrightarrow 00{:}25{:}20.824$ So we're trying to do in this

NOTE Confidence: 0.8073976

 $00:25:20.824 \longrightarrow 00:25:23.130$ one slide is summarized.

NOTE Confidence: 0.8073976

00:25:23.130 --> 00:25:25.180 15 manuscripts into one slide,

NOTE Confidence: 0.8073976

 $00{:}25{:}25{.}180 \dashrightarrow 00{:}25{:}27{.}460$ so again, we offered 728 condition

NOTE Confidence: 0.8073976

 $00:25:27.460 \longrightarrow 00:25:29.504$ gene condition pairs that we

NOTE Confidence: 0.8073976

 $00{:}25{:}29{.}504 \dashrightarrow 00{:}25{:}31{.}328$ categorized into five categories,

NOTE Confidence: 0.8073976

 $00{:}25{:}31{.}330 \dashrightarrow 00{:}25{:}34{.}018$ and I'll say more about the

NOTE Confidence: 0.8073976

 $00{:}25{:}34.018 \dashrightarrow 00{:}25{:}36.440$ categories on the next slide.

NOTE Confidence: 0.8073976

 $00{:}25{:}36{.}440 \dashrightarrow 00{:}25{:}38{.}281$ We gave people the option of which

NOTE Confidence: 0.8073976

 $00{:}25{:}38{.}281 \dashrightarrow 00{:}25{:}39{.}571$ categories they could choose from

NOTE Confidence: 0.8073976

00:25:39.571 - 00:25:40.992 and 89% selected all the categories,

- NOTE Confidence: 0.8073976
- $00:25:40.992 \longrightarrow 00:25:42.450$ although many of them also value

 $00{:}25{:}42{.}495 \dashrightarrow 00{:}25{:}43{.}825$ the fact that they're giving

NOTE Confidence: 0.8073976

 $00{:}25{:}43.825 \dashrightarrow 00{:}25{:}44.889$ choices between the categories,

NOTE Confidence: 0.8073976

 $00:25:44.890 \rightarrow 00:25:46.938$ and I'll say more about the categories again,

NOTE Confidence: 0.8073976

 $00:25:46.940 \longrightarrow 00:25:49.600$ in a few moments.

NOTE Confidence: 0.8073976

 $00:25:49.600 \longrightarrow 00:25:51.940$ We also learned that if we

NOTE Confidence: 0.8073976

 $00:25:51.940 \longrightarrow 00:25:53.500$ did this that essentially.

NOTE Confidence: 0.8073976

 $00:25:53.500 \rightarrow 00:25:54.940$ Of the individuals who enrolled,

NOTE Confidence: 0.8073976

 $00{:}25{:}54{.}940 \dashrightarrow 00{:}25{:}56{.}656$ 78% had at least one finding,

NOTE Confidence: 0.8073976

 $00:25:56.660 \longrightarrow 00:25:58.090$ which meant that if you

NOTE Confidence: 0.8073976

 $00:25:58.090 \rightarrow 00:25:59.234$ test for enough things,

NOTE Confidence: 0.8073976

 $00{:}25{:}59{.}240 \dashrightarrow 00{:}26{:}00{.}956$ every body will have a positive result,

NOTE Confidence: 0.8073976

 $00:26:00.960 \longrightarrow 00:26:02.380$ and so this clearly has

NOTE Confidence: 0.8073976

 $00{:}26{:}02{.}380 \dashrightarrow 00{:}26{:}03{.}516$ implications for the workforce

NOTE Confidence: 0.8073976

 $00:26:03.516 \rightarrow 00:26:04.988$ capacity of genetic counselors to

 $00:26:04.988 \longrightarrow 00:26:06.413$ explain what these results mean.

NOTE Confidence: 0.8073976

 $00:26:06.420 \rightarrow 00:26:08.415$ And we found that in our study,

NOTE Confidence: 0.8073976

 $00:26:08.420 \longrightarrow 00:26:10.296$ the amount of time it took to

NOTE Confidence: 0.8073976

 $00:26:10.296 \longrightarrow 00:26:12.160$ explain this was about 50 minutes,

NOTE Confidence: 0.8073976

 $00{:}26{:}12.160 \dashrightarrow 00{:}26{:}13.595$ and for more uncommon conditions

NOTE Confidence: 0.8073976

 $00{:}26{:}13.595 \dashrightarrow 00{:}26{:}15.030$ around 8 around 84 minutes.

NOTE Confidence: 0.8073976

 $00:26:15.030 \rightarrow 00:26:17.318$ So if this was to be a nationally,

NOTE Confidence: 0.8073976

 $00:26:17.320 \longrightarrow 00:26:19.588$ this will be a huge amount of

NOTE Confidence: 0.8073976

 $00{:}26{:}19.588 \dashrightarrow 00{:}26{:}21.669$ effort that would require on the.

NOTE Confidence: 0.8073976

00:26:21.670 --> 00:26:24.319 On the workforce.

NOTE Confidence: 0.8073976

 $00{:}26{:}24{.}320 \dashrightarrow 00{:}26{:}26{.}856$ We look carefully at both benefits and harms.

NOTE Confidence: 0.8073976

 $00{:}26{:}26{.}860 \dashrightarrow 00{:}26{:}28{.}996$ I think one of the things I learned

NOTE Confidence: 0.8073976

 $00{:}26{:}28{.}996 \dashrightarrow 00{:}26{:}30{.}969$ along time ago was the importance

NOTE Confidence: 0.8073976

 $00:26:30.969 \rightarrow 00:26:33.027$ of trying to evaluate and understand

NOTE Confidence: 0.8073976

00:26:33.091 -> 00:26:34.776 what the benefits and themes,

NOTE Confidence: 0.8073976

 $00:26:34.780 \longrightarrow 00:26:36.365$ benefits and risks are of

- NOTE Confidence: 0.8073976
- $00:26:36.365 \longrightarrow 00:26:37.316$ a particular invention.

 $00:26:37.320 \longrightarrow 00:26:39.147$ And so we looked to see whether

NOTE Confidence: 0.8073976

 $00:26:39.147 \longrightarrow 00:26:41.154$ or not there's any long term

NOTE Confidence: 0.8073976

 $00:26:41.154 \rightarrow 00:26:42.710$ increase in psychosocial outcomes,

NOTE Confidence: 0.8073976

00:26:42.710 --> 00:26:43.598 such as anxiety,

NOTE Confidence: 0.8073976

 $00{:}26{:}43.598 \dashrightarrow 00{:}26{:}44.782$ whether there's any increased

NOTE Confidence: 0.8073976

 $00:26:44.782 \longrightarrow 00:26:46.510$ use of mental health services,

NOTE Confidence: 0.8073976

 $00:26:46.510 \longrightarrow 00:26:48.729$ and we didn't find any of that.

NOTE Confidence: 0.8073976

 $00{:}26{:}48.730 \dashrightarrow 00{:}26{:}50.641$ We did find in a few specific

NOTE Confidence: 0.8073976

 $00{:}26{:}50.641 \dashrightarrow 00{:}26{:}51.888$ cases some confusion between

NOTE Confidence: 0.8073976

 $00:26:51.888 \longrightarrow 00:26:53.718$ the carrier results and its

NOTE Confidence: 0.8073976

 $00{:}26{:}53.718 \dashrightarrow 00{:}26{:}55.182$ implications for chromosome oh.

NOTE Confidence: 0.8073976

 $00{:}26{:}55{.}190 \dashrightarrow 00{:}26{:}57{.}308$ Conditions which were not included here,

NOTE Confidence: 0.8073976

 $00{:}26{:}57{.}310 \dashrightarrow 00{:}27{:}00{.}496$ so not not, I would say it's not surprising,

NOTE Confidence: 0.8073976

 $00:27:00.500 \rightarrow 00:27:02.675$ but not everybody fully understood

 $00:27:02.675 \rightarrow 00:27:04.850$ exactly what these results meant.

NOTE Confidence: 0.8073976

00:27:04.850 --> 00:27:07.370 Most of the participants did value the

NOTE Confidence: 0.8073976

00:27:07.370 --> 00:27:08.803 information for reproductive planning NOTE Confidence: 0.8073976

 $00{:}27{:}08{.}803 \dashrightarrow 00{:}27{:}10{.}833$ and another benefit is that we did

NOTE Confidence: 0.8073976

 $00{:}27{:}10.833 \dashrightarrow 00{:}27{:}12.926$ identify a small number of people who,

NOTE Confidence: 0.8073976

00:27:12.930 --> 00:27:13.938 unbeknownst to them,

NOTE Confidence: 0.8073976

 $00{:}27{:}13{.}938 \dashrightarrow 00{:}27{:}15{.}618$ had secondary findings at which

NOTE Confidence: 0.8073976

 $00:27:15.618 \longrightarrow 00:27:17.554$ put them at risk of having

NOTE Confidence: 0.8073976

 $00{:}27{:}17.554 \dashrightarrow 00{:}27{:}19.054$ medical problems in the future.

NOTE Confidence: 0.8073976

 $00:27:19.060 \longrightarrow 00:27:21.412$ So this was also seen by us as

NOTE Confidence: 0.8073976

 $00:27:21.412 \longrightarrow 00:27:23.985$ well as by the patients as a

NOTE Confidence: 0.8073976

00:27:23.985 --> 00:27:26.384 beneficial thing for them to know

NOTE Confidence: 0.8073976

 $00{:}27{:}26{.}384 \dashrightarrow 00{:}27{:}28{.}910$ their future risk of a condition.

NOTE Confidence: 0.8073976

 $00{:}27{:}28{.}910 \dashrightarrow 00{:}27{:}30{.}968$ In fact, there was a couple.

NOTE Confidence: 0.8073976

 $00:27:30.970 \rightarrow 00:27:34.466$ There's one instance in particular where a A.

NOTE Confidence: 0.8073976

 $00{:}27{:}34{.}470 \dashrightarrow 00{:}27{:}36{.}510$ Mother was identified as a carrier

 $00{:}27{:}36{.}510 \dashrightarrow 00{:}27{:}38{.}655$ for hemophilia and based upon that

NOTE Confidence: 0.8073976

 $00{:}27{:}38.655 \dashrightarrow 00{:}27{:}40.470$ the perinatal management of her

NOTE Confidence: 0.8073976

 $00{:}27{:}40{.}470 \dashrightarrow 00{:}27{:}42{.}345$ pregnancy was changed so she was NOTE Confidence: 0.8073976

 $00{:}27{:}42{.}345 \dashrightarrow 00{:}27{:}44{.}331$ able to deliver in a place that was

NOTE Confidence: 0.8073976

 $00{:}27{:}44{.}331 \dashrightarrow 00{:}27{:}46{.}077$ prepared for the possibility that she NOTE Confidence: 0.8073976

 $00:27:46.077 \rightarrow 00:27:48.189$ might have a child with hemophilia,

NOTE Confidence: 0.8073976

 $00:27:48.190 \longrightarrow 00:27:50.129$ which in fact when she ended up

NOTE Confidence: 0.8073976

 $00{:}27{:}50{.}129 \dashrightarrow 00{:}27{:}52{.}132$ having and the child actually ended

NOTE Confidence: 0.8073976

 $00{:}27{:}52.132 \dashrightarrow 00{:}27{:}53.972$ up having a subdural hemorrhage

NOTE Confidence: 0.8073976

 $00:27:53.972 \longrightarrow 00:27:56.156$ within a couple of days of life.

NOTE Confidence: 0.8073976

 $00{:}27{:}56{.}160 \dashrightarrow 00{:}27{:}58{.}068$ But because it was known what

NOTE Confidence: 0.8073976

 $00{:}27{:}58.068 \dashrightarrow 00{:}27{:}59.022$ the problem was,

NOTE Confidence: 0.8073976

 $00:27:59.030 \rightarrow 00:28:02.648$ they were able to intervene appropriately.

NOTE Confidence: 0.8073976

 $00{:}28{:}02{.}650 \dashrightarrow 00{:}28{:}05{.}492$ I want to share with you briefly

NOTE Confidence: 0.8073976

 $00{:}28{:}05{.}492 \dashrightarrow 00{:}28{:}06{.}710$ some information regarding

 $00:28:06.779 \rightarrow 00:28:09.621$ the five categories that we used to

NOTE Confidence: 0.869017

 $00{:}28{:}09{.}621 \dashrightarrow 00{:}28{:}11{.}597$ differentiate the 78728 conditions and

NOTE Confidence: 0.869017

 $00{:}28{:}11{.}597 \dashrightarrow 00{:}28{:}14{.}758$ when I also share with you that the process NOTE Confidence: 0.869017

 $00:28:14.758 \rightarrow 00:28:17.614$ of coming up with these categories took

NOTE Confidence: 0.869017

 $00{:}28{:}17.614 \dashrightarrow 00{:}28{:}20.199$ us literally about a year and another,

NOTE Confidence: 0.869017

 $00{:}28{:}20{.}200 \dashrightarrow 00{:}28{:}22{.}552$ we spent a lot of time thinking how

NOTE Confidence: 0.869017

 $00{:}28{:}22.552 \dashrightarrow 00{:}28{:}24.518$ should we categorize conditions for

NOTE Confidence: 0.869017

00:28:24.518 --> 00:28:27.140 CF carrier testing or for carrier

NOTE Confidence: 0.869017

 $00{:}28{:}27{.}140 \dashrightarrow 00{:}28{:}29{.}771$ testing so people could make decisions

NOTE Confidence: 0.869017

00:28:29.771 --> 00:28:31.891 about which conditions they wanted

NOTE Confidence: 0.869017

 $00:28:31.900 \rightarrow 00:28:34.306$ and ultimately came up with was.

NOTE Confidence: 0.869017

00:28:34.310 --> 00:28:36.844 To make a distinction between 3 broad

NOTE Confidence: 0.869017

 $00{:}28{:}36{.}844 \dashrightarrow 00{:}28{:}39{.}020$ categories based upon severity, either

NOTE Confidence: 0.869017

 $00{:}28{:}39{.}020 \dashrightarrow 00{:}28{:}41{.}840$ those with a significant shorten lifespan.

NOTE Confidence: 0.869017

 $00{:}28{:}41.840 \dashrightarrow 00{:}28{:}43.044$ Those that were serious.

NOTE Confidence: 0.869017

 $00:28:43.044 \rightarrow 00:28:46.211$ And those that were mild and you can see here

- NOTE Confidence: 0.869017
- $00:28:46.211 \longrightarrow 00:28:48.155$ the definitions of what those categories

 $00:28:48.218 \rightarrow 00:28:50.610$ were in terms of the level of seriousness.

NOTE Confidence: 0.869017

 $00:28:50.610 \longrightarrow 00:28:51.714$ We spent many iterations

NOTE Confidence: 0.869017

 $00:28:51.714 \rightarrow 00:28:53.094$ trying to think through this.

NOTE Confidence: 0.869017

 $00:28:53.100 \longrightarrow 00:28:54.200$ How to organize this?

NOTE Confidence: 0.869017

 $00{:}28{:}54{.}200 \dashrightarrow 00{:}28{:}56{.}424$ And then of course we had to do

NOTE Confidence: 0.869017

 $00:28:56.424 \rightarrow 00:28:58.223$ is place all of the categories all

NOTE Confidence: 0.869017

 $00:28:58.223 \rightarrow 00:29:00.357$ the diseases into these categories.

NOTE Confidence: 0.869017

 $00{:}29{:}00{.}360 \dashrightarrow 00{:}29{:}02{.}236$ We also had a category that was

NOTE Confidence: 0.869017

00:29:02.236 --> 00:29:04.100 based upon its unpredictability.

NOTE Confidence: 0.869017

 $00:29:04.100 \longrightarrow 00:29:05.120$ So for example,

NOTE Confidence: 0.869017

 $00:29:05.120 \rightarrow 00:29:07.840$ there are some conditions in which you, they.

NOTE Confidence: 0.869017

 $00:29:07.840 \longrightarrow 00:29:09.540$ Some may be very mild,

NOTE Confidence: 0.869017

 $00:29:09.540 \longrightarrow 00:29:11.240$ some may be very serious,

NOTE Confidence: 0.869017

 $00{:}29{:}11{.}240 \dashrightarrow 00{:}29{:}12{.}260$ including life threatening.

 $00:29:12.260 \longrightarrow 00:29:14.640$ So we decided to call that out.

NOTE Confidence: 0.869017

 $00:29:14.640 \rightarrow 00:29:16.680$ We also called out those conditions

NOTE Confidence: 0.869017

 $00:29:16.680 \longrightarrow 00:29:18.040$ that begin as adults.

NOTE Confidence: 0.869017

 $00:29:18.040 \longrightarrow 00:29:19.105$ So for example,

NOTE Confidence: 0.869017

 $00:29:19.105 \rightarrow 00:29:21.235$ children who have no symptoms during

NOTE Confidence: 0.869017

 $00:29:21.235 \rightarrow 00:29:23.478$ childhood but may have problems as adults,

NOTE Confidence: 0.869017

 $00{:}29{:}23.480 \dashrightarrow 00{:}29{:}25.105$ and these could be behavioral

NOTE Confidence: 0.869017

 $00:29:25.105 \rightarrow 00:29:27.143$ that could be related to hearing

NOTE Confidence: 0.869017

 $00{:}29{:}27.143 \dashrightarrow 00{:}29{:}29.255$ they could be related to medical.

NOTE Confidence: 0.869017

 $00:29:29.260 \longrightarrow 00:29:29.992$ So for example.

NOTE Confidence: 0.869017

 $00{:}29{:}29{.}992 \dashrightarrow 00{:}29{:}31{.}700$ We you know I mentioned to you

NOTE Confidence: 0.869017

 $00:29:31.755 \rightarrow 00:29:33.789$ that some of the secondary findings

NOTE Confidence: 0.869017

 $00:29:33.789 \longrightarrow 00:29:35.566$ related to hereditary cancer syndromes

NOTE Confidence: 0.869017

 $00:29:35.566 \longrightarrow 00:29:37.010$ that occur in a dulthood.

NOTE Confidence: 0.869017

 $00{:}29{:}37.010 \dashrightarrow 00{:}29{:}39.034$ So the question was and the idea

NOTE Confidence: 0.869017

 $00:29:39.034 \longrightarrow 00:29:40.891$ here is that these are categories

- NOTE Confidence: 0.869017
- $00:29:40.891 \longrightarrow 00:29:42.823$ that they could be testing for

00:29:42.888 --> 00:29:44.040 as well in children,

NOTE Confidence: 0.869017

 $00:29:44.040 \longrightarrow 00:29:45.798$ and also if they chose it.

NOTE Confidence: 0.9021238

00:29:50.810 - > 00:29:52.914 I was also going to share with you

NOTE Confidence: 0.9021238

 $00:29:52.914 \longrightarrow 00:29:54.843$ the actual results that we actually

NOTE Confidence: 0.9021238

 $00:29:54.843 \rightarrow 00:29:56.847$ found in these in these couples,

NOTE Confidence: 0.9021238

 $00:29:56.850 \rightarrow 00:30:00.260$ and So what you can see here is that this is.

NOTE Confidence: 0.9021238

 $00:30:00.260 \dashrightarrow 00:30:02.540$ On the on the high risk couples

NOTE Confidence: 0.9021238

 $00:30:02.540 \longrightarrow 00:30:04.580$ had the following conditions.

NOTE Confidence: 0.8571314

 $00:30:07.030 \dashrightarrow 00:30:08.836$ In which two people were carers.

NOTE Confidence: 0.8571314

 $00{:}30{:}08{.}840 \dashrightarrow 00{:}30{:}10{.}919$ Now I want to point out that the most

NOTE Confidence: 0.8571314

00:30:10.919 --> 00:30:13.048 common one was hereditary hemochromatosis,

NOTE Confidence: 0.8571314

 $00:30:13.050 \rightarrow 00:30:15.210$ which is a condition that is not routinely

NOTE Confidence: 0.8571314

 $00{:}30{:}15{.}210 \dashrightarrow 00{:}30{:}17{.}109$ tested for in carrier testing because

NOTE Confidence: 0.8571314

 $00:30:17.109 \dashrightarrow 00:30:19.065$ of its treat ability and variability.

 $00:30:19.070 \rightarrow 00:30:21.177$ That was actually the most common one.

NOTE Confidence: 0.8571314

 $00{:}30{:}21.180 \dashrightarrow 00{:}30{:}23.520$ The and then you can see some of the

NOTE Confidence: 0.8571314

 $00{:}30{:}23{.}520 \dashrightarrow 00{:}30{:}25{.}191$ examples of the individual secondary

NOTE Confidence: 0.8571314

 $00:30:25.191 \rightarrow 00:30:27.500$ findings as well that we learn now.

NOTE Confidence: 0.8571314

 $00{:}30{:}27{.}500 \dashrightarrow 00{:}30{:}29{.}939$ The reason why we chose to do all of

NOTE Confidence: 0.8571314

00:30:29.939 --> 00:30:32.177 these 'cause we were trying to learn

NOTE Confidence: 0.8571314

 $00{:}30{:}32{.}177 \dashrightarrow 00{:}30{:}34{.}418$ what the impact of this would be.

NOTE Confidence: 0.8571314

 $00:30:34.420 \longrightarrow 00:30:36.625$ And so the idea was to learn.

NOTE Confidence: 0.8571314

 $00{:}30{:}36{.}630 \dashrightarrow 00{:}30{:}38{.}646$ About what the outcome and impact would be

NOTE Confidence: 0.8571314

 $00{:}30{:}38.646 \dashrightarrow 00{:}30{:}40.710$ of testing for all these things, and so,

NOTE Confidence: 0.8571314

 $00:30:40.710 \rightarrow 00:30:42.495$ therefore we decided in a research setting,

NOTE Confidence: 0.8571314

 $00{:}30{:}42.500 \dashrightarrow 00{:}30{:}44.900$ it was reasonable to be more broad than

NOTE Confidence: 0.8571314

 $00{:}30{:}44{.}900 \dashrightarrow 00{:}30{:}47{.}115$ we otherwise might be doing if we were

NOTE Confidence: 0.8571314

 $00{:}30{:}47.115 \dashrightarrow 00{:}30{:}49.200$ providing this as a clinical service.

NOTE Confidence: 0.8571314

 $00:30:49.200 \longrightarrow 00:30:52.398$ So I want to point out.

NOTE Confidence: 0.8571314

 $00:30:52.400 \dashrightarrow 00:30:55.124$ To make a couple comments about

- NOTE Confidence: 0.8571314
- $00:30:55.124 \longrightarrow 00:30:57.510$ the implications of this study.

 $00{:}30{:}57{.}510 \dashrightarrow 00{:}30{:}59{.}922$ So one of the things is that even though

NOTE Confidence: 0.8571314

00:30:59.922 --> 00:31:02.445 we had 300 people who were randomized,

NOTE Confidence: 0.8571314

 $00:31:02.450 \longrightarrow 00:31:05.066$ we got limited data on the impact of

NOTE Confidence: 0.8571314

 $00{:}31{:}05{.}066 \dashrightarrow 00{:}31{:}07{.}398$ couples who had more than a 25% chance

NOTE Confidence: 0.8571314

 $00:31:07.398 \rightarrow 00:31:09.246$ of having a condition that child.

NOTE Confidence: 0.8571314

 $00:31:09.250 \longrightarrow 00:31:11.570$ We only had 12 couples.

NOTE Confidence: 0.8571314

00:31:11.570 --> 00:31:11.883 Also,

NOTE Confidence: 0.8571314

 $00{:}31{:}11{.}883 \dashrightarrow 00{:}31{:}14.074$ because this was on people who initially

NOTE Confidence: 0.8571314

 $00:31:14.074 \rightarrow 00:31:16.358$ sought out on their own carrier testing,

NOTE Confidence: 0.8571314

 $00:31:16.360 \longrightarrow 00:31:18.100$ it didn't include people who were

NOTE Confidence: 0.8571314

 $00{:}31{:}18.100 \dashrightarrow 00{:}31{:}20.179$ less likely to value carrier testing,

NOTE Confidence: 0.8571314

00:31:20.180 --> 00:31:22.452 so may
be the results that we found this

NOTE Confidence: 0.8571314

 $00:31:22.452 \longrightarrow 00:31:24.457$ was well received really was really

NOTE Confidence: 0.8571314

 $00{:}31{:}24.457 \dashrightarrow 00{:}31{:}26.880$ based upon who was selected for this.

- $00:31:26.880 \rightarrow 00:31:29.424$ So while we learn certain things from it,
- NOTE Confidence: 0.8571314
- 00:31:29.430 --> 00:31:31.134 our approach was to be very
- NOTE Confidence: 0.8571314
- $00{:}31{:}31{.}134 \dashrightarrow 00{:}31{:}32{.}940$ careful with who we including.
- NOTE Confidence: 0.8571314
- $00:31:32.940 \longrightarrow 00:31:35.390$ The study might have limited
- NOTE Confidence: 0.8571314
- $00{:}31{:}35{.}390 \dashrightarrow 00{:}31{:}37{.}840$ what we learn from it.
- NOTE Confidence: 0.8571314
- 00:31:37.840 --> 00:31:39.916 And so I think to me,
- NOTE Confidence: 0.8571314
- $00:31:39.920 \longrightarrow 00:31:41.308$ one of the challenges
- NOTE Confidence: 0.8571314
- $00:31:41.308 \longrightarrow 00:31:42.696$ of doing something like.
- NOTE Confidence: 0.8380644
- 00:31:45.470 --> 00:31:46.945 Critical sequencing are for carrier
- NOTE Confidence: 0.8380644
- $00{:}31{:}46{.}945 \dashrightarrow 00{:}31{:}49{.}273$ testing is how to make the test available
- NOTE Confidence: 0.8380644
- $00{:}31{:}49{.}273 \dashrightarrow 00{:}31{:}51{.}128$ to those who would value it while
- NOTE Confidence: 0.8380644
- $00:31:51.180 \rightarrow 00:31:53.046$ avoiding routinely offering in a way
- NOTE Confidence: 0.8380644
- $00{:}31{:}53.046 \dashrightarrow 00{:}31{:}55.172$ that influences those to get tested who
- NOTE Confidence: 0.8380644
- $00:31:55.172 \rightarrow 00:31:57.230$ might otherwise not value it to meet.
- NOTE Confidence: 0.8380644
- $00:31:57.230 \dashrightarrow 00:31:58.994$ This has always been the biggest
- NOTE Confidence: 0.8380644
- 00:31:58.994 --> 00:32:00.170 challenge of carrier testing,

- NOTE Confidence: 0.8380644
- 00:32:00.170 -> 00:32:01.928 depending on how we explain it,

 $00:32:01.930 \longrightarrow 00:32:03.785$ how we define it, people might choose

NOTE Confidence: 0.8380644

00:32:03.785 --> 00:32:05.759 it not because it's really important,

NOTE Confidence: 0.8380644

00:32:05.760 --> 00:32:07.804 but it seems like it makes sense,

NOTE Confidence: 0.8380644

 $00:32:07.810 \longrightarrow 00:32:09.280$ but they don't really quite

NOTE Confidence: 0.8380644

 $00:32:09.280 \longrightarrow 00:32:10.456$ know what they're getting.

NOTE Confidence: 0.8380644

 $00:32:10.460 \longrightarrow 00:32:11.548$ Into the other hand,

NOTE Confidence: 0.8380644

 $00{:}32{:}11.548 \dashrightarrow 00{:}32{:}13.865$ there are some people for whom this is

NOTE Confidence: 0.8380644

 $00:32:13.865 \rightarrow 00:32:15.743$ really meaningful and really want this,

NOTE Confidence: 0.8380644

 $00:32:15.750 \longrightarrow 00:32:17.610$ but I think this is really.

NOTE Confidence: 0.8380644

 $00{:}32{:}17.610 \dashrightarrow 00{:}32{:}21.756$ Attention. The other thing that this.

NOTE Confidence: 0.8380644

 $00{:}32{:}21.760 \dashrightarrow 00{:}32{:}23.540$ Study brought out to me,

NOTE Confidence: 0.8380644

 $00{:}32{:}23{.}540$ --> $00{:}32{:}25{.}577$ which will come back towards the end

NOTE Confidence: 0.8380644

 $00{:}32{:}25{.}577 \dashrightarrow 00{:}32{:}27{.}893$ is that the value and importance of

NOTE Confidence: 0.8380644

 $00{:}32{:}27.893 \dashrightarrow 00{:}32{:}29.963$ carrier testing is because of its

 $00:32:30.033 \rightarrow 00:32:32.123$ personal value to the individuals

NOTE Confidence: 0.8380644

 $00{:}32{:}32{.}123 \dashrightarrow 00{:}32{:}34{.}213$ about themselves about their lives,

NOTE Confidence: 0.8380644

 $00{:}32{:}34{.}220 \dashrightarrow 00{:}32{:}37{.}484$ about their children and how do we think

NOTE Confidence: 0.8380644

 $00:32:37.484 \longrightarrow 00:32:40.380$ about this type of reason for doing?

NOTE Confidence: 0.8380644

 $00{:}32{:}40{.}380 \dashrightarrow 00{:}32{:}41{.}850$ Genetic testing when we have

NOTE Confidence: 0.8380644

00:32:41.850 --> 00:32:42.438 limited resources,

NOTE Confidence: 0.8380644

 $00:32:42.440 \longrightarrow 00:32:44.351$ we're trying to figure out how we

NOTE Confidence: 0.8380644

 $00{:}32{:}44{.}351 \dashrightarrow 00{:}32{:}45{.}970$ should prioritize our health resource

NOTE Confidence: 0.8380644

 $00{:}32{:}45{.}970 \dashrightarrow 00{:}32{:}48{.}720$ and you'll see this issue will come up

NOTE Confidence: 0.8380644

 $00{:}32{:}48.720 \dashrightarrow 00{:}32{:}52.059$ again as we talk about the next project.

NOTE Confidence: 0.8380644

 $00:32:52.060 \rightarrow 00:32:53.467$ Before we talk about the next project,

NOTE Confidence: 0.8380644

 $00{:}32{:}53{.}470 \dashrightarrow 00{:}32{:}55{.}787$ I want one of the things I

NOTE Confidence: 0.8380644

 $00:32:55.787 \longrightarrow 00:32:57.419$ want to mention to you.

NOTE Confidence: 0.8380644

 $00:32:57.420 \longrightarrow 00:32:59.316$ Is that I mentioned you are study was

NOTE Confidence: 0.8380644

 $00:32:59.316 \longrightarrow 00:33:01.563$ part of a larger consortium of nine

NOTE Confidence: 0.8380644

 $00:33:01.563 \rightarrow 00:33:03.278$ studies called the Caesar Consortium,

 $00:33:03.280 \rightarrow 00:33:05.616$ and so one of the things that happens.

NOTE Confidence: 0.8380644

 $00{:}33{:}05{.}620 \dashrightarrow 00{:}33{:}07{.}388$ Part of this consortium is that we all

NOTE Confidence: 0.8380644

 $00:33:07.388 \longrightarrow 00:33:09.680$ get it would get together and write

NOTE Confidence: 0.8380644

00:33:09.680 --> 00:33:11.475 manuscripts about our collective experience,

NOTE Confidence: 0.8380644

 $00:33:11.480 \longrightarrow 00:33:13.811$ and so one of the projects that I got

NOTE Confidence: 0.8380644

 $00:33:13.811 \rightarrow 00:33:16.170$ involved with with with my collaborator Juno.

NOTE Confidence: 0.8380644

00:33:16.170 - 00:33:18.807 You as well as a bunch of other folks,

NOTE Confidence: 0.8380644

 $00:33:18.810 \dashrightarrow 00:33:20.794$ is part of the consortium was to think

NOTE Confidence: 0.8380644

 $00{:}33{:}20.794 \dashrightarrow 00{:}33{:}22.654$ about the questions of consent for

NOTE Confidence: 0.8380644

 $00:33:22.654 \rightarrow 00:33:24.284$ clinical sequencing even though all

NOTE Confidence: 0.8380644

00:33:24.284 --> 00:33:26.426 of our studies were part of research,

NOTE Confidence: 0.8380644

 $00{:}33{:}26{.}430 \dashrightarrow 00{:}33{:}28{.}000$ the intention was to learn.

NOTE Confidence: 0.8380644

 $00:33:28.000 \rightarrow 00:33:30.527$ How do we do this better clinically?

NOTE Confidence: 0.8380644

 $00{:}33{:}30{.}530 \dashrightarrow 00{:}33{:}32{.}294$ So a number of us got together

NOTE Confidence: 0.8380644

 $00{:}33{:}32{.}294 \dashrightarrow 00{:}33{:}34{.}477$ to write a manuscript about the

 $00:33:34.477 \rightarrow 00:33:36.297$ considerations for clinical sequencing

NOTE Confidence: 0.8380644

 $00{:}33{:}36{.}297 \dashrightarrow 00{:}33{:}38{.}830$ based upon all of our experience,

NOTE Confidence: 0.8380644

 $00{:}33{:}38{.}830 \dashrightarrow 00{:}33{:}41.798$ and I want to share with you briefly

NOTE Confidence: 0.8380644

 $00:33:41.798 \longrightarrow 00:33:44.358$ 3 tables from that manuscript.

NOTE Confidence: 0.8380644

00:33:44.360 --> 00:33:46.663 The first is the table of considerations

NOTE Confidence: 0.8380644

 $00{:}33{:}46.663 \dashrightarrow 00{:}33{:}49.203$ and the idea of these considerations are NOTE Confidence: 0.8380644

 $00{:}33{:}49{.}203 \dashrightarrow 00{:}33{:}51{.}896$ things for people to think about before

NOTE Confidence: 0.8380644

 $00{:}33{:}51{.}896$ --> $00{:}33{:}54{.}236$ they think about the consent process.

NOTE Confidence: 0.8380644

 $00{:}33{:}54{.}240 \dashrightarrow 00{:}33{:}56{.}739$ The first thing to remember is that

NOTE Confidence: 0.8380644

 $00{:}33{:}56{.}739 \dashrightarrow 00{:}33{:}59{.}347$ questions that sequence are in the context

NOTE Confidence: 0.8380644

00:33:59.347 --> 00:34:01.192 of ongoing clinical decision making,

NOTE Confidence: 0.8380644

 $00:34:01.200 \rightarrow 00:34:03.390$ whether it's clinical decision making better,

NOTE Confidence: 0.8380644

 $00:34:03.390 \longrightarrow 00:34:04.088$ better pregnancy,

NOTE Confidence: 0.8380644

 $00{:}34{:}04.088 \dashrightarrow 00{:}34{:}05.833$ whether that's a clinical decision

NOTE Confidence: 0.8380644

 $00:34:05.833 \dashrightarrow 00:34:07.420$ making about cancer treatment.

NOTE Confidence: 0.8380644

 $00:34:07.420 \longrightarrow 00:34:09.250$ It's about some other question

- NOTE Confidence: 0.8380644
- $00:34:09.250 \rightarrow 00:34:10.348$ that's coming up,
- NOTE Confidence: 0.8380644
- $00:34:10.350 \rightarrow 00:34:12.540$ which is why clinical sequence is
- NOTE Confidence: 0.8380644
- $00:34:12.540 \rightarrow 00:34:14.329$ being introduced. So for example.
- NOTE Confidence: 0.8380644
- 00:34:14.329 --> 00:34:16.447 Two of the most common reasons
- NOTE Confidence: 0.8380644
- $00{:}34{:}16{.}447 \dashrightarrow 00{:}34{:}18{.}227$ currently why sequencing is done
- NOTE Confidence: 0.8380644
- $00{:}34{:}18.227 \dashrightarrow 00{:}34{:}20.780$ clinically is in the context of cancer.
- NOTE Confidence: 0.8380644
- $00{:}34{:}20.780 \dashrightarrow 00{:}34{:}22.620$ An evaluation of cancer treatments.
- NOTE Confidence: 0.8380644
- $00:34:22.620 \longrightarrow 00:34:24.455$ Another reason is for children
- NOTE Confidence: 0.8380644
- $00{:}34{:}24{.}455 \dashrightarrow 00{:}34{:}25{.}556$ with developmental delay.
- NOTE Confidence: 0.8380644
- $00:34:25.560 \rightarrow 00:34:27.400$ An evaluation of developmental delay.
- NOTE Confidence: 0.8380644
- $00:34:27.400 \longrightarrow 00:34:29.495$ So thinking about the clinical
- NOTE Confidence: 0.8380644
- 00:34:29.495 --> 00:34:32.190 context may help think about what
- NOTE Confidence: 0.8380644
- $00{:}34{:}32{.}190 \dashrightarrow 00{:}34{:}34{.}795$ information is appropriate to disclose.
- NOTE Confidence: 0.81924385
- $00{:}34{:}34{.}800 \dashrightarrow 00{:}34{:}36{.}495$ The next consideration which will
- NOTE Confidence: 0.81924385
- 00:34:36.495 --> 00:34:38.900 I'll elaborate on in the next slide,
- NOTE Confidence: 0.81924385

 $00:34:38.900 \longrightarrow 00:34:41.812$ is that the process needs to be

NOTE Confidence: 0.81924385

 $00{:}34{:}41.812 \dashrightarrow 00{:}34{:}43.670$ succinct and responsive to the.

NOTE Confidence: 0.81924385

00:34:43.670 --> 00:34:44.982 Person's personal situation context,

NOTE Confidence: 0.81924385

 $00:34:44.982 \rightarrow 00:34:46.622$ 'cause typically and actually three,

NOTE Confidence: 0.81924385

 $00:34:46.630 \rightarrow 00:34:49.267$ is related to that too, and the converse,

NOTE Confidence: 0.81924385

 $00:34:49.267 \rightarrow 00:34:51.570$ because often typically was done for consent,

NOTE Confidence: 0.81924385

00:34:51.570 - 00:34:53.914 is let's make sure we give a lot

NOTE Confidence: 0.81924385

 $00:34:53.914 \dashrightarrow 00:34:55.510$ of information about genetics.

NOTE Confidence: 0.81924385

 $00{:}34{:}55{.}510$ --> $00{:}34{:}56{.}642$ Alot of technical information,

NOTE Confidence: 0.81924385

 $00:34:56.642 \rightarrow 00:34:59.521$ but that may not be as necessary as knowing

NOTE Confidence: 0.81924385

 $00{:}34{:}59{.}521 \dashrightarrow 00{:}35{:}01{.}765$ what's important to the individual person.

NOTE Confidence: 0.81924385

 $00:35:01.770 \longrightarrow 00:35:04.549$ So the analogy I like to give

NOTE Confidence: 0.81924385

 $00:35:04.549 \longrightarrow 00:35:07.068$ is that when we do an MRI.

NOTE Confidence: 0.81924385

 $00:35:07.070 \rightarrow 00:35:09.272$ The consent process does not usually

NOTE Confidence: 0.81924385

 $00:35:09.272 \dashrightarrow 00:35:11.195$ begin with a conversation about

NOTE Confidence: 0.81924385

00:35:11.195 --> 00:35:13.165 quantum mechanics and atomic spin,

 $00:35:13.170 \longrightarrow 00:35:16.599$ which is the basis of how MRI is done.

NOTE Confidence: 0.81924385

 $00{:}35{:}16.600 \dashrightarrow 00{:}35{:}19.260$ But yet when we do genetic testing,

NOTE Confidence: 0.81924385

 $00:35:19.260 \rightarrow 00:35:23.628$ we often think it's important to know.

NOTE Confidence: 0.81924385

 $00:35:23.630 \rightarrow 00:35:24.830$ What base pairs are,

NOTE Confidence: 0.81924385

 $00:35:24.830 \longrightarrow 00:35:26.630$ which is how even the movie

NOTE Confidence: 0.81924385

00:35:26.696 --> 00:35:28.008 Gattaca got its name,

NOTE Confidence: 0.81924385

 $00:35:28.010 \longrightarrow 00:35:31.104$ but are base pairs necessary to think

NOTE Confidence: 0.81924385

 $00:35:31.104 \rightarrow 00:35:33.910$ about the implications of genetic testing?

NOTE Confidence: 0.81924385

 $00{:}35{:}33{.}910 \dashrightarrow 00{:}35{:}35{.}998$ The other broad set of considerations

NOTE Confidence: 0.81924385

 $00:35:35.998 \rightarrow 00:35:38.391$ for consent that we came up with

NOTE Confidence: 0.81924385

 $00:35:38.391 \rightarrow 00:35:40.568$ collectively was that we have to think

NOTE Confidence: 0.81924385

 $00{:}35{:}40{.}638 \dashrightarrow 00{:}35{:}42{.}778$ very explicitly about 3 considerations

NOTE Confidence: 0.81924385

 $00{:}35{:}42.778$ --> $00{:}35{:}45.286$ regarding the results and their implications.

NOTE Confidence: 0.81924385

 $00{:}35{:}45{.}286$ --> $00{:}35{:}48{.}214$ One is to acknowledge the range of results,

NOTE Confidence: 0.81924385

 $00:35:48.220 \rightarrow 00:35:50.060$ including the limitations of interpretations,

 $00:35:50.060 \rightarrow 00:35:52.466$ so that the expectations are reasonable

NOTE Confidence: 0.81924385

 $00:35:52.466 \rightarrow 00:35:55.690$ and we don't have two high expectations.

NOTE Confidence: 0.81924385

 $00{:}35{:}55{.}690 \dashrightarrow 00{:}35{:}57{.}390$ And.

NOTE Confidence: 0.81924385

 $00{:}35{:}57{.}390 \dashrightarrow 00{:}35{:}59{.}112$ One of the other things that

NOTE Confidence: 0.81924385

00:35:59.112 --> 00:35:59.973 clinicians can facilitate.

NOTE Confidence: 0.81924385

 $00:35:59.980 \longrightarrow 00:36:01.385$ The decision making process by

NOTE Confidence: 0.81924385

 $00:36:01.385 \longrightarrow 00:36:02.790$ thinking about the potential clinical

NOTE Confidence: 0.81924385

 $00{:}36{:}02{.}839 \dashrightarrow 00{:}36{:}03{.}999$ and emotional implications for

NOTE Confidence: 0.81924385

 $00{:}36{:}03{.}999 \dashrightarrow 00{:}36{:}05{.}449$ the individual of those results.

NOTE Confidence: 0.81924385

 $00{:}36{:}05{.}450 \dashrightarrow 00{:}36{:}07{.}421$ How will it feel to you if you need

NOTE Confidence: 0.81924385

 $00:36{:}07.421 \dashrightarrow 00{:}36{:}09.489$ to have more doctors appointments?

NOTE Confidence: 0.81924385

00:36:09.490 --> 00:36:11.786 How will it feel to you to know?

NOTE Confidence: 0.81924385

00:36:11.790 --> 00:36:13.476 You might have risk of having

NOTE Confidence: 0.81924385

 $00:36:13.476 \longrightarrow 00:36:14.960$ a problem later in life,

NOTE Confidence: 0.81924385

 $00{:}36{:}14.960 \dashrightarrow 00{:}36{:}16.780$ so at least getting the clinician and

NOTE Confidence: 0.81924385

 $00:36:16.780 \longrightarrow 00:36:18.870$ the patient be thinking about that and

 $00:36:18.870 \longrightarrow 00:36:20.445$ also thinking about the implications

NOTE Confidence: 0.81924385

 $00:36:20.445 \longrightarrow 00:36:22.160$ of the results for the family,

NOTE Confidence: 0.81924385

 $00:36:22.160 \longrightarrow 00:36:24.176$ and being aware that of how they want

NOTE Confidence: 0.81924385

 $00:36:24.176 \longrightarrow 00:36:26.476$ to deal with that piece of information.

NOTE Confidence: 0.81924385

 $00{:}36{:}26{.}480 \dashrightarrow 00{:}36{:}27{.}464$ So the idea.

NOTE Confidence: 0.81924385

 $00{:}36{:}27{.}464 \dashrightarrow 00{:}36{:}29{.}432$ Was to have these six considerations

NOTE Confidence: 0.81924385

 $00:36:29.432 \rightarrow 00:36:31.906$ in mind when thinking about consent.

NOTE Confidence: 0.81924385

 $00:36:31.910 \longrightarrow 00:36:34.325$ I want to really have us focus

NOTE Confidence: 0.81924385

 $00:36:34.325 \longrightarrow 00:36:36.538$ on Table 2 for a moment,

NOTE Confidence: 0.81924385

 $00{:}36{:}36{.}540 \dashrightarrow 00{:}36{:}39{.}403$ which has to do with sample examples

NOTE Confidence: 0.81924385

 $00{:}36{:}39{.}403 \dashrightarrow 00{:}36{:}41{.}541$ of questions that really relate

NOTE Confidence: 0.81924385

 $00{:}36{:}41{.}541 \dashrightarrow 00{:}36{:}43{.}205$ back to the second.

NOTE Confidence: 0.81924385

 $00:36:43.210 \longrightarrow 00:36:45.426$ But can't relate back to number 2 here,

NOTE Confidence: 0.81924385

 $00:36:45.430 \longrightarrow 00:36:46.924$ which is the consent process should

NOTE Confidence: 0.81924385

 $00{:}36{:}46{.}924 \dashrightarrow 00{:}36{:}48{.}613$ be should be responsive to the

 $00:36:48.613 \rightarrow 00:36:49.576$ individuals personal situation.

NOTE Confidence: 0.81924385

 $00:36:49.580 \longrightarrow 00:36:51.519$ Context. Well, how do you do that?

NOTE Confidence: 0.81924385

 $00{:}36{:}51{.}520 \dashrightarrow 00{:}36{:}53{.}165$ The idea here is that consent is

NOTE Confidence: 0.81924385

 $00:36:53.165 \rightarrow 00:36:54.840$ not about giving people information,

NOTE Confidence: 0.81924385

 $00:36:54.840 \dashrightarrow 00:36:56.230$ but actually asking them questions.

NOTE Confidence: 0.81924385

 $00{:}36{:}56{.}230$ --> $00{:}36{:}57{.}620$ Finding out from them information NOTE Confidence: 0.81924385

 $00:36:57.620 \dashrightarrow 00:36:59.325$ about themselves to help you decide

NOTE Confidence: 0.81924385

 $00:36:59.325 \rightarrow 00:37:00.939$ what information they need to know.

NOTE Confidence: 0.81924385

 $00{:}37{:}00{.}940 \dashrightarrow 00{:}37{:}02{.}868$ And so here's a list of what those

NOTE Confidence: 0.81924385

 $00:37:02.868 \rightarrow 00:37:04.817$ questions are fine with their experiences.

NOTE Confidence: 0.81924385

 $00:37:04.820 \longrightarrow 00:37:06.200$ Then finally what they think

NOTE Confidence: 0.81924385

 $00{:}37{:}06{.}200 \dashrightarrow 00{:}37{:}07{.}028$ the benefits are.

NOTE Confidence: 0.81924385

 $00:37:07.030 \rightarrow 00:37:08.969$ Find out questions and concerns they have.

NOTE Confidence: 0.81924385

 $00:37:08.970 \rightarrow 00:37:10.909$ Find out maybe they've already made decision.

NOTE Confidence: 0.81924385

 $00:37:10.910 \dashrightarrow 00:37:13.254$ One of the things we've learned is that.

NOTE Confidence: 0.81924385

 $00:37:13.260 \longrightarrow 00:37:14.432$ Often people when they,

 $00:37:14.432 \rightarrow 00:37:16.190$ when they are engaged with clinician

NOTE Confidence: 0.81924385

 $00:37:16.241 \longrightarrow 00:37:17.641$ or they have an idea of what

NOTE Confidence: 0.81924385

 $00{:}37{:}17.641 \dashrightarrow 00{:}37{:}19.230$ they want to do and so starting

NOTE Confidence: 0.81924385

 $00:37:19.230 \longrightarrow 00:37:20.634$ all over and acting as though

NOTE Confidence: 0.8407035

 $00:37:20.640 \longrightarrow 00:37:22.140$ they have no idea might not

NOTE Confidence: 0.8407035

 $00:37:22.140 \longrightarrow 00:37:23.140$ be really reflecting where

NOTE Confidence: 0.8407035

 $00:37:23.189 \longrightarrow 00:37:24.575$ they are there at that moment.

NOTE Confidence: 0.8407035

 $00{:}37{:}24.580 \dashrightarrow 00{:}37{:}26.292$ But one of the other points and this

NOTE Confidence: 0.8407035

 $00{:}37{:}26.292 \dashrightarrow 00{:}37{:}28.425$ is really brought up by many of our

NOTE Confidence: 0.8407035

 $00{:}37{:}28.425 \dashrightarrow 00{:}37{:}29.525$ genetic counseling colleagues has

NOTE Confidence: 0.8407035

 $00{:}37{:}29{.}578 \dashrightarrow 00{:}37{:}31{.}482$ been the notion of trying to get them

NOTE Confidence: 0.8407035

 $00{:}37{:}31{.}482 \dashrightarrow 00{:}37{:}32{.}968$ to anticipate results and say so.

NOTE Confidence: 0.8407035

 $00:37:32.968 \dashrightarrow 00:37:35.374$ If you get a result like X, how do you? NOTE Confidence: 0.8407035

00:37:35.374 --> 00:37:37.370 What do you think you'll do with that?

NOTE Confidence: 0.8407035

 $00{:}37{:}37{.}370 \dashrightarrow 00{:}37{:}39{.}260$ And as a way of just getting into sort

 $00:37:39.260 \longrightarrow 00:37:41.248$ of walk through weather what this means

NOTE Confidence: 0.8407035

 $00:37:41.248 \longrightarrow 00:37:43.528$ to them and whether they really do order.

NOTE Confidence: 0.8407035

 $00:37:43.530 \longrightarrow 00:37:45.870$ Don't want this and why?

NOTE Confidence: 0.8407035

 $00{:}37{:}45.870 \dashrightarrow 00{:}37{:}46.659$ I want to.

NOTE Confidence: 0.9192076

 $00{:}37{:}50{.}480 \dashrightarrow 00{:}37{:}54{.}332$ Also. Share with you that I'm not

NOTE Confidence: 0.9192076

 $00{:}37{:}54{.}332 \dashrightarrow 00{:}37{:}56{.}064$ gonna go through these in details

NOTE Confidence: 0.9192076

00:37:56.064 --> 00:37:58.379 because I want to move on to the next

NOTE Confidence: 0.9192076

 $00{:}37{:}58{.}379 \dashrightarrow 00{:}38{:}00{.}584$ part of the talk is that we went from

NOTE Confidence: 0.9192076

00:38:00.590 --> 00:38:01.925 those considerations to having twelve

NOTE Confidence: 0.9192076

 $00{:}38{:}01{.}925 \dashrightarrow 00{:}38{:}03{.}260$ key considerations or key points,

NOTE Confidence: 0.9192076

 $00{:}38{:}03{.}260 \dashrightarrow 00{:}38{:}05{.}316$ and the idea is that these are the

NOTE Confidence: 0.9192076

 $00{:}38{:}05{.}316 \dashrightarrow 00{:}38{:}07{.}123$ key points that need to be made

NOTE Confidence: 0.9192076

 $00:38:07.123 \longrightarrow 00:38:08.870$ as part of the consent process,

NOTE Confidence: 0.9192076

 $00{:}38{:}08{.}870 \dashrightarrow 00{:}38{:}11{.}160$ and what I want to do here to night is not

NOTE Confidence: 0.9192076

 $00:38:11.224 \rightarrow 00:38:13.408$ going through each of these key points,

NOTE Confidence: 0.9192076

 $00:38:13.410 \rightarrow 00:38:15.237$ but point out to you the categories

 $00:38:15.237 \rightarrow 00:38:16.791$ and sub categories are the first

NOTE Confidence: 0.9192076

 $00{:}38{:}16.791 \dashrightarrow 00{:}38{:}18.237$ had to do with information about

NOTE Confidence: 0.9192076

 $00:38:18.237 \longrightarrow 00:38:20.078$ the range and limitation of results.

NOTE Confidence: 0.9192076

 $00{:}38{:}20{.}080 \dashrightarrow 00{:}38{:}21{.}865$ So talking about both the types of

NOTE Confidence: 0.9192076

 $00{:}38{:}21.865 \dashrightarrow 00{:}38{:}23.410$ results available and the limitations.

NOTE Confidence: 0.9192076

 $00{:}38{:}23{.}410 \dashrightarrow 00{:}38{:}25{.}108$ And again, the idea is trying

NOTE Confidence: 0.9192076

 $00:38:25.108 \longrightarrow 00:38:26.240$ to make this simple.

NOTE Confidence: 0.9192076

 $00{:}38{:}26{.}240 \dashrightarrow 00{:}38{:}27{.}625$ Trying to make this sufficiently

NOTE Confidence: 0.9192076

 $00{:}38{:}27.625 \dashrightarrow 00{:}38{:}29.010$ straightforward that this can be

NOTE Confidence: 0.9192076

 $00:38:29.061 \rightarrow 00:38:30.765$ communicated by anybody to anybody else.

NOTE Confidence: 0.9192076

 $00{:}38{:}30{.}770 \dashrightarrow 00{:}38{:}32{.}185$ This does not require some

NOTE Confidence: 0.9192076

 $00{:}38{:}32{.}185 \dashrightarrow 00{:}38{:}33{.}600$ of the PhD in genetics,

NOTE Confidence: 0.9192076

 $00:38:33.600 \dashrightarrow 00:38:36.066$ so you could communicate these concepts.

NOTE Confidence: 0.9192076

 $00{:}38{:}36{.}070 \dashrightarrow 00{:}38{:}37{.}924$ The second is to think through

NOTE Confidence: 0.9192076

 $00{:}38{:}37{.}924 \dashrightarrow 00{:}38{:}39{.}680$ the implications of the results.
$00:38:39.680 \longrightarrow 00:38:41.320$ Think about the clinical implications

NOTE Confidence: 0.9192076

 $00:38:41.320 \longrightarrow 00:38:42.960$ for the person being tested.

NOTE Confidence: 0.9192076

00:38:42.960 --> 00:38:44.312 Thinking about the emotional

NOTE Confidence: 0.9192076

 $00:38:44.312 \dashrightarrow 00:38:46.002$ implications and thinking about the

NOTE Confidence: 0.9192076

 $00{:}38{:}46.002 \dashrightarrow 00{:}38{:}47.220$ implications for family members.

NOTE Confidence: 0.9192076

 $00{:}38{:}47{.}220 \dashrightarrow 00{:}38{:}47{.}920$ So again,

NOTE Confidence: 0.9192076

 $00{:}38{:}47{.}920 \dashrightarrow 00{:}38{:}50{.}370$ a way of organizing or thinking about

NOTE Confidence: 0.9192076

 $00{:}38{:}50{.}370 \dashrightarrow 00{:}38{:}52{.}777$ what should be the conversation to

NOTE Confidence: 0.9192076

 $00{:}38{:}52.777 \dashrightarrow 00{:}38{:}55.240$ help with the decision making about.

NOTE Confidence: 0.9192076

 $00:38:55.240 \longrightarrow 00:38:56.004$ About testing,

NOTE Confidence: 0.9192076

00:38:56.004 - 00:38:59.060 so as we were working on our study,

NOTE Confidence: 0.9192076

00:38:59.060 --> 00:39:01.404 the next thing I want to share with

NOTE Confidence: 0.9192076

 $00{:}39{:}01{.}404 \dashrightarrow 00{:}39{:}03{.}555$ you is something that happened

NOTE Confidence: 0.9192076

00:39:03.555 --> 00:39:05.547 happened during our study,

NOTE Confidence: 0.9192076

 $00{:}39{:}05{.}550 \dashrightarrow 00{:}39{:}07{.}955$ we realized that our approach

NOTE Confidence: 0.9192076

00:39:07.955 --> 00:39:09.879 to doing carrier testing.

- NOTE Confidence: 0.9192076
- $00:39:09.880 \longrightarrow 00:39:11.980$ Within Kaiser for people who

00:39:11.980 --> 00:39:13.660 are having cystic fibrosis,

NOTE Confidence: 0.9192076

 $00:39:13.660 \rightarrow 00:39:16.642$ carrier testing resulted in people who

NOTE Confidence: 0.9192076

 $00:39:16.642 \rightarrow 00:39:19.096$ were highly educated value genetic

NOTE Confidence: 0.9192076

 $00:39:19.096 \dashrightarrow 00:39:21.770$ testing a lot and were mostly white

NOTE Confidence: 0.9192076

 $00:39{:}21.770 \dashrightarrow 00{:}39{:}25.070$ and we started realizing that you know.

NOTE Confidence: 0.9192076

00:39:25.070 --> 00:39:26.605 If Genic testing is becoming

NOTE Confidence: 0.9192076

00:39:26.605 --> 00:39:27.526 more widely available,

NOTE Confidence: 0.9192076

 $00:39:27.530 \longrightarrow 00:39:29.486$ we need to be thinking more

NOTE Confidence: 0.9192076

 $00:39:29.486 \longrightarrow 00:39:31.230$ broadly than the example of CF.

NOTE Confidence: 0.9192076

00:39:31.230 --> 00:39:31.858 Carrier testing,

NOTE Confidence: 0.9192076

 $00{:}39{:}31.858 \dashrightarrow 00{:}39{:}34.620$ and in the context that we did that in.

NOTE Confidence: 0.9192076

 $00:39:34.620 \longrightarrow 00:39:36.924$ So we ended up with a number of

NOTE Confidence: 0.9192076

 $00{:}39{:}36{.}924 \dashrightarrow 00{:}39{:}38{.}534$ collaborators are doing a series

NOTE Confidence: 0.9192076

 $00{:}39{:}38{.}534 \dashrightarrow 00{:}39{:}40{.}472$ of interviews and focus groups at

 $00:39:40.472 \rightarrow 00:39:42.627$ safety at hospitals in the in Oregon.

NOTE Confidence: 0.9192076

 $00:39:42.630 \longrightarrow 00:39:43.522$ These were.

NOTE Confidence: 0.9192076

 $00:39:43.522 \rightarrow 00:39:45.306$ Federally qualified health centers

NOTE Confidence: 0.9192076

 $00{:}39{:}45{.}306 \dashrightarrow 00{:}39{:}47{.}510$ where we interviewed both the

NOTE Confidence: 0.9192076

 $00{:}39{:}47{.}510$ --> $00{:}39{:}49{.}275$ clinicians and patients to find

NOTE Confidence: 0.9192076

 $00{:}39{:}49{.}275 \dashrightarrow 00{:}39{:}51{.}777$ out what they thought of a variety

NOTE Confidence: 0.9192076

 $00:39:51.777 \longrightarrow 00:39:52.800$ of genetic services.

NOTE Confidence: 0.9192076

 $00:39:52.800 \dashrightarrow 00:39:54.640$ The first big point of this paper was

NOTE Confidence: 0.9192076

 $00{:}39{:}54.640 \dashrightarrow 00{:}39{:}56.798$ that the conditions were highly sceptical.

NOTE Confidence: 0.9192076

 $00:39:56.800 \dashrightarrow 00:39:58.879$ The idea of genetics in this setting

NOTE Confidence: 0.9192076

 $00:39:58.879 \longrightarrow 00:40:00.319$ and their response was look.

NOTE Confidence: 0.9192076

00:40:00.320 --> 00:40:01.000 You know,

NOTE Confidence: 0.9192076

 $00:40:01.000 \longrightarrow 00:40:03.380$ people have a lot of things to

NOTE Confidence: 0.9192076

 $00{:}40{:}03{.}380 \dashrightarrow 00{:}40{:}05{.}317$ deal with in their lives.

NOTE Confidence: 0.9192076

 $00{:}40{:}05{.}320 \dashrightarrow 00{:}40{:}06{.}409$ They have many,

NOTE Confidence: 0.9192076

 $00:40:06.409 \rightarrow 00:40:08.224$ and again typically now this

- NOTE Confidence: 0.9192076
- $00:40:08.224 \longrightarrow 00:40:10.319$ was done by three years ago.

00:40:10.320 --> 00:40:10.938 But now,

NOTE Confidence: 0.9192076

 $00{:}40{:}10.938 \dashrightarrow 00{:}40{:}12.792$ with all the focus on systemic

NOTE Confidence: 0.9192076

 $00{:}40{:}12.792 \dashrightarrow 00{:}40{:}14.837$ racism and thinking about all the

NOTE Confidence: 0.9192076

 $00{:}40{:}14.837 \dashrightarrow 00{:}40{:}16.877$ various ways that people who are

NOTE Confidence: 0.9192076

 $00:40:16.944 \longrightarrow 00:40:18.984$ under represented in research are

NOTE Confidence: 0.9192076

 $00:40:18.984 \rightarrow 00:40:21.353$ facing huge number of challenges that

NOTE Confidence: 0.9192076

 $00{:}40{:}21.353 \dashrightarrow 00{:}40{:}22.968$ something like genetics seems like

NOTE Confidence: 0.9192076

 $00{:}40{:}22.968 \dashrightarrow 00{:}40{:}24.950$ it is like completely unnecessary.

NOTE Confidence: 0.9192076

 $00:40:24.950 \longrightarrow 00:40:26.334$ Inappropriate when people are

NOTE Confidence: 0.9192076

 $00{:}40{:}26{.}334 \dashrightarrow 00{:}40{:}27{.}718$ facing concerns about housing

NOTE Confidence: 0.9192076

00:40:27.718 --> 00:40:29.240 about addiction about depression,

NOTE Confidence: 0.9192076

 $00:40:29.240 \longrightarrow 00:40:31.598$ why would you even think of

NOTE Confidence: 0.9192076

 $00:40:31.598 \longrightarrow 00:40:32.777$ offering genetic services?

NOTE Confidence: 0.9192076

 $00:40:32.780 \rightarrow 00:40:35.336$ We talked to the patient so and they were

00:40:35.336 --> 00:40:37.726 actually much more open and thought you know,

NOTE Confidence: 0.9192076

00:40:37.730 --> 00:40:38.017 actually.

NOTE Confidence: 0.9192076

 $00{:}40{:}38.017 \dashrightarrow 00{:}40{:}40.600$ In fact we are interested in these types of

NOTE Confidence: 0.84534127

 $00:40:40.660 \longrightarrow 00:40:41.800$ services and in fact,

NOTE Confidence: 0.84534127

 $00{:}40{:}41.800 \dashrightarrow 00{:}40{:}42.964$ particularly because that these

NOTE Confidence: 0.84534127

 $00:40:42.964 \rightarrow 00:40:44.710$ are being offered to other folks.

NOTE Confidence: 0.84534127

 $00:40:44.710 \longrightarrow 00:40:46.170$ We want these two now.

NOTE Confidence: 0.84534127

 $00:40:46.170 \longrightarrow 00:40:48.186$ One thing I want to show you

NOTE Confidence: 0.84534127

 $00{:}40{:}48.186 \dashrightarrow 00{:}40{:}49.977$ and illustrate for you is how

NOTE Confidence: 0.84534127

 $00:40:49.977 \longrightarrow 00:40:51.693$ do we ask people this question?

NOTE Confidence: 0.84534127

 $00:40:51.700 \longrightarrow 00:40:53.428$ 'cause it's not easy to ask

NOTE Confidence: 0.84534127

 $00:40:53.428 \rightarrow 00:40:54.925$ people about genetics when they

NOTE Confidence: 0.84534127

 $00:40:54.925 \longrightarrow 00:40:56.350$ may be less familiar about.

NOTE Confidence: 0.84534127

 $00{:}40{:}56{.}350 \dashrightarrow 00{:}40{:}59{.}002$ So the approach we took without

NOTE Confidence: 0.84534127

 $00{:}40{:}59{.}002 \dashrightarrow 00{:}41{:}00{.}328$ with some collaborators.

NOTE Confidence: 0.84534127

00:41:00.330 --> 00:41:02.370 We created a series of PowerPoint

 $00{:}41{:}02{.}370 \dashrightarrow 00{:}41{:}04{.}524$ that that had animated stories and

NOTE Confidence: 0.84534127

 $00:41:04.524 \rightarrow 00:41:06.344$ we actually had three stories.

NOTE Confidence: 0.84534127

00:41:06.350 - 00:41:08.120 One was about carrier testing,

NOTE Confidence: 0.84534127

 $00:41:08.120 \longrightarrow 00:41:09.885$ one was about genetic testing

NOTE Confidence: 0.84534127

 $00{:}41{:}09{.}885 \dashrightarrow 00{:}41{:}10{.}944$ for developmental disabilities,

NOTE Confidence: 0.84534127

 $00{:}41{:}10.950 \dashrightarrow 00{:}41{:}12.973$ and the third one illustrated here had

NOTE Confidence: 0.84534127

 $00{:}41{:}12.973 \dashrightarrow 00{:}41{:}15.549$ to do with hereditary cancer syndromes,

NOTE Confidence: 0.84534127

 $00:41:15.550 \rightarrow 00:41:19.645$ with the notion that somebody who has.

NOTE Confidence: 0.84534127

 $00{:}41{:}19.650 \dashrightarrow 00{:}41{:}22.146$ A risk of cancer and the fact that

NOTE Confidence: 0.84534127

 $00:41:22.146 \longrightarrow 00:41:24.476$ that risk of cancer in this case.

NOTE Confidence: 0.84023404

00:41:28.090 --> 00:41:30.538 Suggest that, or in this case she had cancer,

NOTE Confidence: 0.84023404

 $00{:}41{:}30{.}540 \dashrightarrow 00{:}41{:}32{.}353$ suggested she might be at risk of

NOTE Confidence: 0.84023404

00:41:32.353 --> 00:41:33.755 having a hereditary cancer syndrome

NOTE Confidence: 0.84023404

 $00{:}41{:}33{.}755 \dashrightarrow 00{:}41{:}35{.}483$ and perhaps testing might be helpful

NOTE Confidence: 0.84023404

00:41:35.483 --> 00:41:37.664 to her to know more about what the

 $00:41:37.664 \rightarrow 00:41:39.438$ implications might be for her daughters.

NOTE Confidence: 0.84023404

 $00:41:39.438 \longrightarrow 00:41:41.652$ When we turned out that of

NOTE Confidence: 0.84023404

 $00:41:41.652 \rightarrow 00:41:43.698$ the three examples we gave,

NOTE Confidence: 0.84023404

 $00{:}41{:}43.700 \dashrightarrow 00{:}41{:}46.580$ the one that the conditions were at least

NOTE Confidence: 0.84023404

 $00:41:46.580 \rightarrow 00:41:48.288$ plausibly interested in was this one.

NOTE Confidence: 0.84023404

 $00{:}41{:}48{.}290 \dashrightarrow 00{:}41{:}51{.}233$ An while the patient stress and all of them.

NOTE Confidence: 0.84023404

 $00:41:51.240 \rightarrow 00:41:53.536$ This also resonated the most with them,

NOTE Confidence: 0.84023404

 $00:41:53.540 \rightarrow 00:41:55.180$ in part because they realize

NOTE Confidence: 0.84023404

 $00:41:55.180 \longrightarrow 00:41:56.820$ that cancer can affect anybody,

NOTE Confidence: 0.84023404

 $00:41:56.820 \longrightarrow 00:41:58.116$ whereas whereas these care,

NOTE Confidence: 0.84023404

00:41:58.116 --> 00:41:59.736 these rare diseases are pretty

NOTE Confidence: 0.84023404

 $00{:}41{:}59{.}736 \dashrightarrow 00{:}42{:}01{.}419$ rare and in fact developmental

NOTE Confidence: 0.84023404

 $00:42:01.419 \longrightarrow 00:42:03.049$ delays don't happen that often.

NOTE Confidence: 0.84023404

 $00:42:03.050 \rightarrow 00:42:04.685$ But everyone everyone had cancer

NOTE Confidence: 0.84023404

00:42:04.685 --> 00:42:05.993 somewhere in their families,

NOTE Confidence: 0.84023404

 $00:42:06.000 \rightarrow 00:42:09.010$ so they all could resonate with that.

- NOTE Confidence: 0.84023404
- $00:42:09.010 \longrightarrow 00:42:13.120$ So. And in fact.
- NOTE Confidence: 0.84023404
- $00:42:13.120 \longrightarrow 00:42:15.365$ There are guidelines for both
- NOTE Confidence: 0.84023404
- $00:42:15.365 \rightarrow 00:42:18.124$ hereditary breast cancer as well As
- NOTE Confidence: 0.84023404
- $00:42:18.124 \rightarrow 00:42:20.299$ for what's called Lynch syndrome,
- NOTE Confidence: 0.84023404
- $00:42:20.300 \rightarrow 00:42:23.450$ which is actually a hereditary colon cancer,
- NOTE Confidence: 0.84023404
- $00{:}42{:}23.450 \dashrightarrow 00{:}42{:}24.794$ an individual cancer.
- NOTE Confidence: 0.84023404
- $00:42:24.794 \longrightarrow 00:42:27.034$ So these are two genetic.
- NOTE Confidence: 0.85791844
- $00:42:29.750 \longrightarrow 00:42:31.014$ Conditions are hereditary cancer
- NOTE Confidence: 0.85791844
- $00:42:31.014 \rightarrow 00:42:32.910$ syndromes for which there are clear
- NOTE Confidence: 0.85791844
- $00:42:32.961 \rightarrow 00:42:34.521$ recommendations that if there's a family
- NOTE Confidence: 0.85791844
- $00{:}42{:}34{.}521 \dashrightarrow 00{:}42{:}36{.}728$ history or of a person of a certain age,
- NOTE Confidence: 0.85791844
- $00{:}42{:}36{.}730 \dashrightarrow 00{:}42{:}38{.}398$ it's recommended that testing be done.
- NOTE Confidence: 0.85791844
- $00:42:38.400 \rightarrow 00:42:40.624$ So the idea here is not something hey,
- NOTE Confidence: 0.85791844
- $00:42:40.630 \longrightarrow 00:42:41.746$ do you want this?
- NOTE Confidence: 0.85791844
- $00:42:41.746 \longrightarrow 00:42:43.420$ Maybe you should think about this,
- NOTE Confidence: 0.85791844

 $00:42:43.420 \rightarrow 00:42:45.652$ but you should have this done in the same

NOTE Confidence: 0.85791844

 $00:42:45.652 \rightarrow 00:42:47.885$ way that somebody is having chest pain.

NOTE Confidence: 0.85791844

 $00:42:47.890 \longrightarrow 00:42:49.280$ We might say, you know,

NOTE Confidence: 0.85791844

 $00{:}42{:}49{.}280 \dashrightarrow 00{:}42{:}51{.}782$ we really we really need to do an EKG.

NOTE Confidence: 0.85791844

00:42:51.790 --> 00:42:53.736 It's not just you want an EKG,

NOTE Confidence: 0.85791844

 $00:42:53.740 \longrightarrow 00:42:55.414$ so no, you should have this

NOTE Confidence: 0.85791844

 $00:42:55.414 \rightarrow 00:42:57.090$ and the same idea here is,

NOTE Confidence: 0.85791844

 $00:42:57.090 \rightarrow 00:43:00.000$ yet you should have this done.

NOTE Confidence: 0.85791844

 $00{:}43{:}00{.}000 \dashrightarrow 00{:}43{:}03{.}448$ So the question then becomes the fact that.

NOTE Confidence: 0.9320227

 $00:43:06.000 \rightarrow 00:43:10.648$ People who are. Low in low income

NOTE Confidence: 0.9320227

 $00{:}43{:}10.648 \dashrightarrow 00{:}43{:}12.176$ or underrepresented in research

NOTE Confidence: 0.9320227

 $00{:}43{:}12.176 \dashrightarrow 00{:}43{:}14.205$ and clinical care are less likely

NOTE Confidence: 0.9320227

 $00:43:14.205 \rightarrow 00:43:16.390$ to have this type of testing done.

NOTE Confidence: 0.9320227

 $00:43:16.390 \longrightarrow 00:43:18.280$ They may. Not have had if they

NOTE Confidence: 0.9320227

 $00:43:18.280 \longrightarrow 00:43:20.228$ may not have a family history,

NOTE Confidence: 0.9320227

 $00:43:20.230 \rightarrow 00:43:22.282$ they may not have had a family history taken.

- NOTE Confidence: 0.9320227
- $00:43:22.290 \rightarrow 00:43:23.886$ They may not have gotten a referral.

 $00{:}43{:}23.890 \dashrightarrow 00{:}43{:}25.690$ They may not have gotten.

NOTE Confidence: 0.9320227

 $00:43:25.690 \longrightarrow 00:43:26.882$ Jenna testing they meant

NOTE Confidence: 0.9320227

00:43:26.882 --> 00:43:27.776 I've gotten diagnosis,

NOTE Confidence: 0.9320227

 $00{:}43{:}27.780 \dashrightarrow 00{:}43{:}29.270$ so we started realizing that

NOTE Confidence: 0.9320227

 $00{:}43{:}29{.}270 \dashrightarrow 00{:}43{:}30{.}760$ if there was a pop,

NOTE Confidence: 0.9320227

 $00:43:30.760 \longrightarrow 00:43:32.524$ if there was a that if we're

NOTE Confidence: 0.9320227

 $00{:}43{:}32{.}524 \dashrightarrow 00{:}43{:}34{.}220$ looking for an opportunity to do

NOTE Confidence: 0.9320227

 $00{:}43{:}34{.}220 \dashrightarrow 00{:}43{:}36{.}271$ testing in a way that could have

NOTE Confidence: 0.9320227

 $00{:}43{:}36{.}333 \dashrightarrow 00{:}43{:}38{.}208$ an impact on health disparities.

NOTE Confidence: 0.9320227

 $00{:}43{:}38{.}210 \dashrightarrow 00{:}43{:}39{.}885$ That thinking about the issues

NOTE Confidence: 0.9320227

00:43:39.885 --> 00:43:41.225 of hereditary cancer syndromes

NOTE Confidence: 0.9320227

00:43:41.225 --> 00:43:42.967 might be very important to do so,

NOTE Confidence: 0.9320227

00:43:42.970 --> 00:43:44.758 even though I was a pediatrician,

NOTE Confidence: 0.9320227

 $00{:}43{:}44.760 \dashrightarrow 00{:}43{:}46.284$ we our country and I decided

 $00:43:46.284 \longrightarrow 00:43:48.398$ to apply for the next round of

NOTE Confidence: 0.9320227

 $00:43:48.398 \rightarrow 00:43:50.128$ funding from the Caesar Consortium,

NOTE Confidence: 0.9320227

 $00:43:50.130 \longrightarrow 00:43:51.018$ and it's interesting.

NOTE Confidence: 0.9320227

00:43:51.018 --> 00:43:51.906 The next round,

NOTE Confidence: 0.9320227

 $00{:}43{:}51{.}910 \dashrightarrow 00{:}43{:}53{.}898$ five of the six studies were about

NOTE Confidence: 0.9320227

 $00{:}43{:}53.898 \dashrightarrow 00{:}43{:}55.190$ Pediatrics and pediatric context,

NOTE Confidence: 0.9320227

 $00:43:55.190 \longrightarrow 00:43:57.098$ and ours was the only one.

NOTE Confidence: 0.9320227

00:43:57.100 --> 00:43:59.050 That was among adults where we

NOTE Confidence: 0.9320227

 $00{:}43{:}59{.}050 \dashrightarrow 00{:}44{:}00{.}897$ really focused on a dults with

NOTE Confidence: 0.9320227

00:44:00.897 - 00:44:02.358 hereditary cancer syndromes.

NOTE Confidence: 0.9320227

 $00:44:02.360 \longrightarrow 00:44:05.060$ In a primary care setting.

NOTE Confidence: 0.9320227

 $00:44:05.060 \rightarrow 00:44:07.036$ Well I want to show you on this

NOTE Confidence: 0.9320227

 $00{:}44{:}07{.}036 \dashrightarrow 00{:}44{:}09{.}050$ slide is to acknowledge that there's

NOTE Confidence: 0.9320227

 $00:44:09.050 \longrightarrow 00:44:11.234$ a series of decisions that need

NOTE Confidence: 0.9320227

 $00:44:11.302 \rightarrow 00:44:13.276$ to be made regarding doing this,

NOTE Confidence: 0.9320227

 $00:44:13.280 \rightarrow 00:44:15.485$ and it's very complicated to do so.

- NOTE Confidence: 0.9320227
- $00:44:15.490 \longrightarrow 00:44:17.576$ Part of what our task was and

00:44:17.576 --> 00:44:19.600 thinking about our study was saying,

NOTE Confidence: 0.9320227

 $00:44:19.600 \longrightarrow 00:44:21.496$ how can we make this process

NOTE Confidence: 0.9320227

 $00:44:21.496 \rightarrow 00:44:22.760$ simpler and less complicated?

NOTE Confidence: 0.9320227

 $00{:}44{:}22.760 \dashrightarrow 00{:}44{:}24.638$ To make it more accessible to

NOTE Confidence: 0.9320227

 $00{:}44{:}24.638 \dashrightarrow 00{:}44{:}26.246$ people who otherwise might not

NOTE Confidence: 0.9320227

 $00:44:26.246 \longrightarrow 00:44:27.701$ have access to these services

NOTE Confidence: 0.9320227

 $00:44:27.701 \longrightarrow 00:44:29.709$ where it's hard to be identified,

NOTE Confidence: 0.9320227

 $00{:}44{:}29{.}710 \dashrightarrow 00{:}44{:}31{.}544$ it's hard to get to come in

NOTE Confidence: 0.9320227

 $00:44:31.544 \longrightarrow 00:44:33.180$ to get an evaluation,

NOTE Confidence: 0.9320227

 $00{:}44{:}33.180 \dashrightarrow 00{:}44{:}35.208$ and that and the agenda counselor.

NOTE Confidence: 0.9320227

 $00{:}44{:}35{.}210 \dashrightarrow 00{:}44{:}36{.}715$ And to come back several

NOTE Confidence: 0.9320227

 $00{:}44{:}36{.}715 \dashrightarrow 00{:}44{:}37{.}919$ times or several visits.

NOTE Confidence: 0.9320227

 $00{:}44{:}37{.}920 \dashrightarrow 00{:}44{:}39{.}904$ So we one of the things that we

NOTE Confidence: 0.9320227

 $00:44:39.904 \longrightarrow 00:44:41.903$ do this in a more simplified

 $00:44:41.903 \rightarrow 00:44:43.335$ way and still effective.

NOTE Confidence: 0.9320227

 $00:44:43.340 \longrightarrow 00:44:45.080$ So the study that we developed

NOTE Confidence: 0.9320227

 $00:44:45.080 \longrightarrow 00:44:46.650$ is called the Charm study,

NOTE Confidence: 0.9320227

 $00{:}44{:}46.650 \dashrightarrow 00{:}44{:}48.155$ which stands for cancer Health

NOTE Confidence: 0.9320227

 $00{:}44{:}48.155 \dashrightarrow 00{:}44{:}49.058$ assessments reaching many.

NOTE Confidence: 0.9320227

 $00{:}44{:}49{.}060 \dashrightarrow 00{:}44{:}51{.}634$ And the idea here is that this was a

NOTE Confidence: 0.9320227

00:44:51.634 --> 00:44:53.988 study was done through Kaiser and it

NOTE Confidence: 0.9320227

 $00{:}44{:}53{.}988 \dashrightarrow 00{:}44{:}56{.}759$ was really done in English and Spanish.

NOTE Confidence: 0.9320227

 $00{:}44{:}56{.}760$ --> $00{:}44{:}59{.}352$ And the team was a very diverse team NOTE Confidence: 0.9320227

00:44:59.352 --> 00:45:01.700 from people from Kaiser in Portland,

NOTE Confidence: 0.9320227

 $00{:}45{:}01.700 \dashrightarrow 00{:}45{:}03.470$ but also a Denver health,

NOTE Confidence: 0.9320227

 $00{:}45{:}03{.}470 \dashrightarrow 00{:}45{:}05{.}220$ which is a federally qualified

NOTE Confidence: 0.9320227

 $00{:}45{:}05{.}220 \dashrightarrow 00{:}45{:}07{.}803$ Health Center in Denver as well as

NOTE Confidence: 0.9320227

 $00{:}45{:}07{.}803 \dashrightarrow 00{:}45{:}09{.}708$ with collaborators at University of

NOTE Confidence: 0.9320227

 $00{:}45{:}09{.}708 \dashrightarrow 00{:}45{:}11{.}548$ Washington who did the sequencing

NOTE Confidence: 0.9320227

 $00{:}45{:}11.548 \dashrightarrow 00{:}45{:}13.746$ for us as well as collaborators at

- NOTE Confidence: 0.9320227
- $00:45:13.746 \longrightarrow 00:45:15.568$ a variety of other institutions,

00:45:15.568 --> 00:45:17.830 including UCSF as well as Emery

NOTE Confidence: 0.9320227

00:45:17.891 --> 00:45:19.346 and Dana Farber and RTI,

NOTE Confidence: 0.9320227

 $00:45:19.350 \longrightarrow 00:45:22.241$ who helped us with both the risk

NOTE Confidence: 0.9320227

 $00:45:22.241 \longrightarrow 00:45:24.308$ assessment evaluation and our study

NOTE Confidence: 0.9320227

 $00:45:24.308 \rightarrow 00:45:27.324$ included a range of both evaluations of of.

NOTE Confidence: 0.9320227

 $00:45:27.330 \longrightarrow 00:45:27.683$ Of.

NOTE Confidence: 0.9320227

 $00{:}45{:}27.683 \dashrightarrow 00{:}45{:}29.448$ Doing lots of interviews with

NOTE Confidence: 0.9320227

 $00{:}45{:}29{.}448 \dashrightarrow 00{:}45{:}31{.}221$ participants along the course of

NOTE Confidence: 0.9320227

 $00:45:31.221 \longrightarrow 00:45:33.003$ the study to learn about their

NOTE Confidence: 0.9320227

 $00{:}45{:}33.003 \dashrightarrow 00{:}45{:}34.731$ experience was like which is part

NOTE Confidence: 0.9320227

00:45:34.731 --> 00:45:36.781 of which is why we were involved in

NOTE Confidence: 0.9320227

00:45:36.781 --> 00:45:38.227 Seattle Children's as well because of

NOTE Confidence: 0.9320227

 $00{:}45{:}38{.}227 \dashrightarrow 00{:}45{:}39{.}889$ our expertise in qualitative research.

NOTE Confidence: 0.9320227

 $00:45:39.890 \longrightarrow 00:45:40.906$ The study.

 $00:45:40.906 \rightarrow 00:45:43.954$ Included individuals aged 18 to 49

NOTE Confidence: 0.9320227

 $00:45:43.954 \rightarrow 00:45:47.337$ who either spoke English or Spanish.

NOTE Confidence: 0.9320227

 $00{:}45{:}47{.}340 \dashrightarrow 00{:}45{:}50{.}420$ This was done at.

NOTE Confidence: 0.9320227

 $00{:}45{:}50{.}420 \dashrightarrow 00{:}45{:}52{.}120$ Kaiser in the Portland area,

NOTE Confidence: 0.9320227

 $00{:}45{:}52{.}120 \dashrightarrow 00{:}45{:}54{.}160$ as well as a Denver health.

NOTE Confidence: 0.81246525

 $00:45:54.160 \longrightarrow 00:45:55.860$ We had our different range

NOTE Confidence: 0.81246525

 $00:45:55.860 \rightarrow 00:45:57.560$ of ways of recruiting people,

NOTE Confidence: 0.81246525

 $00:45:57.560 \longrightarrow 00:45:58.580$ either through email,

NOTE Confidence: 0.81246525

 $00{:}45{:}58{.}580 \dashrightarrow 00{:}46{:}00{.}280$ text messages or in person.

NOTE Confidence: 0.81246525

 $00:46:00.280 \rightarrow 00:46:02.392$ In this and similar approaches were

NOTE Confidence: 0.81246525

00:46:02.392 --> 00:46:04.359 used at Denver Health as well,

NOTE Confidence: 0.81246525

 $00{:}46{:}04{.}360 \dashrightarrow 00{:}46{:}05{.}712$ including in Denver help.

NOTE Confidence: 0.81246525

 $00:46:05.712 \longrightarrow 00:46:07.740$ Also by our direct referrals by

NOTE Confidence: 0.81246525

 $00:46:07.800 \longrightarrow 00:46:09.172$ providers for those individuals

NOTE Confidence: 0.81246525

 $00:46:09.172 \longrightarrow 00:46:11.624$ who they believe may be at risk

NOTE Confidence: 0.81246525

 $00:46:11.624 \rightarrow 00:46:13.580$ for having hereditary cancer in the

 $00:46:13.580 \longrightarrow 00:46:17.025$ family but had not yet been had an

NOTE Confidence: 0.81246525

 $00{:}46{:}17.025 \dashrightarrow 00{:}46{:}18.997$ opportunity for genetic testing.

NOTE Confidence: 0.81246525

 $00{:}46{:}19{.}000 \dashrightarrow 00{:}46{:}20{.}750$ The main outcome of our study was

NOTE Confidence: 0.81246525

 $00:46:20.750 \longrightarrow 00:46:22.716$ to look at the positive findings

NOTE Confidence: 0.81246525

 $00{:}46{:}22.716 \dashrightarrow 00{:}46{:}24.646$ related to hereditary breast and

NOTE Confidence: 0.81246525

00:46:24.646 --> 00:46:26.390 Varian Cancer Orland syndrome,

NOTE Confidence: 0.81246525

00:46:26.390 --> 00:46:28.567 as well as a range of secondary

NOTE Confidence: 0.81246525

 $00{:}46{:}28{.}567 \dashrightarrow 00{:}46{:}30{.}185$ outcomes that included both the

NOTE Confidence: 0.81246525

 $00{:}46{:}30.185 \dashrightarrow 00{:}46{:}32.362$ number of of other findings but also

NOTE Confidence: 0.81246525

 $00{:}46{:}32{.}362 \dashrightarrow 00{:}46{:}34{.}119$ included health care utilization.

NOTE Confidence: 0.81246525

 $00{:}46{:}34.120 \dashrightarrow 00{:}46{:}35.800$ Their understanding of the care,

NOTE Confidence: 0.81246525

 $00{:}46{:}35{.}800 \dashrightarrow 00{:}46{:}37{.}144$ their satisfaction, their communication,

NOTE Confidence: 0.81246525

 $00:46:37.144 \rightarrow 00:46:39.160$ and well say the very end.

NOTE Confidence: 0.81246525

 $00:46:39.160 \longrightarrow 00:46:40.705$ About personal utility.

NOTE Confidence: 0.81246525

 $00{:}46{:}40.705 \dashrightarrow 00{:}46{:}43.795$ The value of this for them.

 $00{:}46{:}43.800 \dashrightarrow 00{:}46{:}46.000$ But I want to share briefly here and NOTE Confidence: 0.81246525

00:46:46.000 --> 00:46:48.318 I want to try to be mindful of our

NOTE Confidence: 0.81246525

 $00{:}46{:}48{.}318$ --> $00{:}46{:}50{.}418$ time so we have time for questions NOTE Confidence: 0.81246525

 $00:46:50.418 \rightarrow 00:46:53.320$ is to suggest that this was a very NOTE Confidence: 0.81246525

 $00:46:53.320 \rightarrow 00:46:58.030$ complex project where we did. Is.

NOTE Confidence: 0.81246525

 $00{:}46{:}58.030 \dashrightarrow 00{:}47{:}00.814$ If you look at this range of services. NOTE Confidence: 0.81246525

 $00:47:00.820 \longrightarrow 00:47:02.630$ We identified 12 different things

NOTE Confidence: 0.81246525

 $00:47:02.630 \longrightarrow 00:47:05.171$ that we tried to do differently to

NOTE Confidence: 0.81246525

 $00{:}47{:}05{.}171 \dashrightarrow 00{:}47{:}07{.}795$ see how well it would work at it,

NOTE Confidence: 0.81246525

 $00{:}47{:}07{.}800 \dashrightarrow 00{:}47{:}10{.}229$ including how we try to reach people NOTE Confidence: 0.81246525

 $00{:}47{:}10.229 \dashrightarrow 00{:}47{:}12.301$ by identifying people by email or

NOTE Confidence: 0.81246525

 $00{:}47{:}12{.}301 \dashrightarrow 00{:}47{:}14{.}239$ text which we collected our family

NOTE Confidence: 0.81246525

00:47:14.239 --> 00:47:16.180 history on a standardized patient,

NOTE Confidence: 0.81246525

 $00:47:16.180 \longrightarrow 00:47:18.349$ patient facing tool.

NOTE Confidence: 0.81246525

 $00{:}47{:}18.350 \dashrightarrow 00{:}47{:}20.744$ We included people who have limited

NOTE Confidence: 0.81246525

 $00{:}47{:}20.744 \dashrightarrow 00{:}47{:}22.340$ information about their families

 $00:47:22.403 \longrightarrow 00:47:23.927$ as a way of being broader.

NOTE Confidence: 0.81246525

 $00{:}47{:}23{.}930 \dashrightarrow 00{:}47{:}25{.}952$ We provide based on this information

NOTE Confidence: 0.81246525

00:47:25.952 --> 00:47:28.037 we provided an online risk assessment

NOTE Confidence: 0.81246525

 $00:47:28.037 \longrightarrow 00:47:30.396$ and then we also did our pretest

NOTE Confidence: 0.81246525

 $00:47:30.396 \rightarrow 00:47:32.309$ education counseling online as well,

NOTE Confidence: 0.81246525

 $00{:}47{:}32{.}310 \dashrightarrow 00{:}47{:}35{.}094$ and so the idea here is that the

NOTE Confidence: 0.81246525

 $00:47:35.094 \rightarrow 00:47:36.625$ individuals who were obtaining

NOTE Confidence: 0.81246525

 $00:47:36.625 \longrightarrow 00:47:39.264$ this did not actually have to go

NOTE Confidence: 0.81246525

 $00:47:39.264 \longrightarrow 00:47:41.030$ into a doctor's office.

NOTE Confidence: 0.81246525

 $00:47:41.030 \longrightarrow 00:47:43.366$ Not only one, not only more than once,

NOTE Confidence: 0.81246525

 $00:47:43.370 \longrightarrow 00:47:45.989$ but even once all this could be done online,

NOTE Confidence: 0.81246525

 $00{:}47{:}45{.}990 \dashrightarrow 00{:}47{:}47{.}425$ we also provided halfway through

NOTE Confidence: 0.81246525

 $00{:}47{:}47{.}425 \dashrightarrow 00{:}47{:}49{.}442$ the study a decision aid to help

NOTE Confidence: 0.81246525

 $00{:}47{:}49{.}442 \dashrightarrow 00{:}47{:}50{.}772$ people decide whether they did

NOTE Confidence: 0.81246525

 $00{:}47{:}50.772 \dashrightarrow 00{:}47{:}52.253$ or didn't want secondary findings

 $00:47:52.253 \rightarrow 00:47:54.125$ because we found we offer people

NOTE Confidence: 0.81246525

 $00{:}47{:}54.125 \dashrightarrow 00{:}47{:}55.626$ the choice of secondary fires.

NOTE Confidence: 0.81246525

00:47:55.626 --> 00:47:56.794 Almost everybody said yes,

NOTE Confidence: 0.81246525

00:47:56.800 - 00:47:59.092 and we worried you know people

NOTE Confidence: 0.81246525

 $00:47:59.092 \longrightarrow 00:48:00.238$ saying yes because.

NOTE Confidence: 0.81246525

 $00:48:00.240 \longrightarrow 00:48:00.908$ Why not?

NOTE Confidence: 0.81246525

 $00{:}48{:}00{.}908 \dashrightarrow 00{:}48{:}02{.}578$ It's free, more is better,

NOTE Confidence: 0.81246525

 $00{:}48{:}02{.}580 \dashrightarrow 00{:}48{:}04{.}578$ so we actually created a decision.

NOTE Confidence: 0.81246525

00:48:04.580 --> 00:48:07.317 We tried to really slow people down

NOTE Confidence: 0.81246525

 $00{:}48{:}07{.}317 \dashrightarrow 00{:}48{:}10{.}370$ to think about this more deliberately.

NOTE Confidence: 0.81246525

 $00{:}48{:}10{.}370 \dashrightarrow 00{:}48{:}12{.}708$ I want to comment about our results.

NOTE Confidence: 0.81246525

 $00:48:12.710 \longrightarrow 00:48:14.046$ Disclosure is that we,

NOTE Confidence: 0.81246525

 $00:48:14.046 \rightarrow 00:48:16.050$ for those people had negative results.

NOTE Confidence: 0.81246525

 $00:48:16.050 \longrightarrow 00:48:17.386$ We provided that information

NOTE Confidence: 0.81246525

 $00{:}48{:}17{.}386 \dashrightarrow 00{:}48{:}18{.}388$ initially by phone,

NOTE Confidence: 0.81246525

 $00:48:18.390 \rightarrow 00:48:21.387$ but all my letter for ease of doing this.

- NOTE Confidence: 0.81246525
- 00:48:21.390 --> 00:48:24.054 The key, if you notice on the side,

 $00:48:24.060 \longrightarrow 00:48:25.660$ there's a couple places where

NOTE Confidence: 0.81246525

 $00:48:25.660 \longrightarrow 00:48:28.013$ there are some dice the dice refer

NOTE Confidence: 0.81246525

 $00:48:28.013 \rightarrow 00:48:29.738$ to things that we randomized,

NOTE Confidence: 0.81246525

 $00:48:29.740 \longrightarrow 00:48:31.016$ so we did randomize.

NOTE Confidence: 0.81246525

 $00{:}48{:}31{.}016 \dashrightarrow 00{:}48{:}32{.}930$ People explain this in more detail

NOTE Confidence: 0.81246525

 $00{:}48{:}32{.}989 \dashrightarrow 00{:}48{:}35{.}138$ in a few moments to two different

NOTE Confidence: 0.81246525

00:48:35.138 --> 00:48:36.760 approaches to Jenna Counseling.

NOTE Confidence: 0.81246525

 $00{:}48{:}36{.}760 \dashrightarrow 00{:}48{:}38{.}525$ We also randomized training for

NOTE Confidence: 0.81246525

 $00{:}48{:}38{.}525 \dashrightarrow 00{:}48{:}40{.}580$ interpreters to see whether or not.

NOTE Confidence: 0.81246525

 $00{:}48{:}40{.}580 \dashrightarrow 00{:}48{:}41{.}904$ Having trained interpreters would

NOTE Confidence: 0.81246525

 $00{:}48{:}41{.}904 \dashrightarrow 00{:}48{:}43{.}559$ be helpful to communicating this

NOTE Confidence: 0.81246525

 $00:48:43.559 \longrightarrow 00:48:44.280$ information as well,

NOTE Confidence: 0.81246525

 $00{:}48{:}44{.}280 \dashrightarrow 00{:}48{:}46{.}116$ because this study is still ongoing,

NOTE Confidence: 0.81246525

 $00:48:46.120 \longrightarrow 00:48:47.920$ I'm not going to present the

- $00:48:47.920 \longrightarrow 00:48:49.120$ results of this in
- NOTE Confidence: 0.84673136
- 00:48:49.185 --> 00:48:51.285 the purpose of me and I will
- NOTE Confidence: 0.84673136
- $00{:}48{:}51{.}285 \dashrightarrow 00{:}48{:}53{.}186$ describe some more of the features
- NOTE Confidence: 0.84673136
- $00:48:53.186 \longrightarrow 00:48:55.058$ of the study in more detail,
- NOTE Confidence: 0.84673136
- 00:48:55.060 --> 00:48:57.520 but the message I want to
- NOTE Confidence: 0.84673136
- $00:48:57.520 \longrightarrow 00:48:59.700$ convey is to suggest that.
- NOTE Confidence: 0.84673136
- $00{:}48{:}59{.}700 \dashrightarrow 00{:}49{:}02{.}244$ Studies like this that try to look at
- NOTE Confidence: 0.84673136
- $00:49:02.244 \rightarrow 00:49:04.002$ the possibility doing sequencing in
- NOTE Confidence: 0.84673136
- 00:49:04.002 --> 00:49:06.126 the context of clinical practice is
- NOTE Confidence: 0.84673136
- 00:49:06.126 --> 00:49:08.508 how we will learn how to do it better
- NOTE Confidence: 0.84673136
- $00{:}49{:}08{.}508 \dashrightarrow 00{:}49{:}10{.}608$ and how we learn how to innovate and NOTE Confidence: 0.84673136
- $00:49:10.608 \rightarrow 00:49:13.029$ see if we can do this effectively.
- NOTE Confidence: 0.84673136
- $00:49:13.030 \longrightarrow 00:49:15.130$ So I want to share with you
- NOTE Confidence: 0.84673136
- $00:49:15.130 \longrightarrow 00:49:17.058$ a few aspects of our study.
- NOTE Confidence: 0.84673136
- $00:49:17.060 \longrightarrow 00:49:18.812$ One is that our approach to
- NOTE Confidence: 0.84673136
- $00:49:18.812 \rightarrow 00:49:20.470$ consent was was done online.

- NOTE Confidence: 0.84673136
- $00{:}49{:}20{.}470 \dashrightarrow 00{:}49{:}22{.}890$ We used a multimedia approach

 $00:49:22.890 \longrightarrow 00:49:24.826$ where we used images.

NOTE Confidence: 0.84673136

00:49:24.830 --> 00:49:25.075 Actually,

NOTE Confidence: 0.84673136

00:49:25.075 - 00:49:26.300 the same colleagues have done

NOTE Confidence: 0.84673136

 $00{:}49{:}26{.}300 \dashrightarrow 00{:}49{:}27{.}820$ our images for the previous study

NOTE Confidence: 0.84673136

 $00:49:27.820 \longrightarrow 00:49:29.055$ of the of the PowerPoints.

NOTE Confidence: 0.84673136

 $00:49:29.060 \longrightarrow 00:49:30.560$ Here we use images with very

NOTE Confidence: 0.84673136

 $00:49:30.560 \longrightarrow 00:49:31.060$ brief information.

NOTE Confidence: 0.84673136

 $00{:}49{:}31.060 \dashrightarrow 00{:}49{:}32.796$ So we really tried to think about.

NOTE Confidence: 0.851953569999999

 $00:49:34.910 \rightarrow 00:49:37.446$ How to be as concise as possible with

NOTE Confidence: 0.851953569999999

 $00{:}49{:}37{.}446 \dashrightarrow 00{:}49{:}39{.}935$ what this test will tell me and one

NOTE Confidence: 0.851953569999999

 $00{:}49{:}39{.}935 \dashrightarrow 00{:}49{:}42{.}473$ of the things that I have been a very

NOTE Confidence: 0.851953569999999

 $00{:}49{:}42{.}473 \dashrightarrow 00{:}49{:}44{.}582$ big fan of with the concept of consent

NOTE Confidence: 0.851953569999999

 $00{:}49{:}44.582 \dashrightarrow 00{:}49{:}46.500$ is trying to be concise by making

NOTE Confidence: 0.851953569999999

 $00{:}49{:}46.561 \dashrightarrow 00{:}49{:}48.780$ suggestions for what the reasons might be,

 $00:49:48.780 \longrightarrow 00:49:50.550$ why some people may want it,

NOTE Confidence: 0.851953569999999

00:49:50.550 - 00:49:52.910 and why some people may not want it.

NOTE Confidence: 0.851953569999999

00:49:52.910 --> 00:49:54.947 And it's a way of both communicating

NOTE Confidence: 0.851953569999999

 $00:49:54.947 \rightarrow 00:49:56.738$ that there's more than one choice,

NOTE Confidence: 0.851953569999999

 $00:49:56.740 \longrightarrow 00:49:58.964$ but to do it in a way that

NOTE Confidence: 0.851953569999999

 $00:49:58.964 \longrightarrow 00:50:00.400$ makes it fairly so.

NOTE Confidence: 0.851953569999999

 $00{:}50{:}00{.}400 \dashrightarrow 00{:}50{:}02{.}068$ Simple to see what those reasons

NOTE Confidence: 0.851953569999999

 $00:50:02.068 \rightarrow 00:50:03.505$ are rather than thinking there's

NOTE Confidence: 0.851953569999999

00:50:03.505 - 00:50:04.850 a whole litany of things,

NOTE Confidence: 0.851953569999999

 $00:50:04.850 \longrightarrow 00:50:07.230$ but really trying to boil down to

NOTE Confidence: 0.851953569999999

 $00{:}50{:}07{.}230 \dashrightarrow 00{:}50{:}09{.}340$ some upsides and some downsides.

NOTE Confidence: 0.851953569999999

 $00:50:09.340 \rightarrow 00:50:11.930$ The other thing that we did is that we just

NOTE Confidence: 0.851953569999999

 $00{:}50{:}11{.}990 \dashrightarrow 00{:}50{:}14{.}398$ about this was done as a research study.

NOTE Confidence: 0.851953569999999

 $00:50:14.400 \rightarrow 00:50:16.200$ We distinguish between consent for testing

NOTE Confidence: 0.851953569999999

 $00:50:16.200 \rightarrow 00:50:18.049$ and consent from being in the study,

NOTE Confidence: 0.851953569999999

 $00:50:18.050 \rightarrow 00:50:19.938$ and so that the first thing we tried

 $00:50:19.938 \longrightarrow 00:50:22.116$ to do is to walk them through whether

NOTE Confidence: 0.851953569999999

 $00:50:22.116 \longrightarrow 00:50:23.950$ or not from their perspective,

NOTE Confidence: 0.851953569999999

 $00:50:23.950 \rightarrow 00:50:25.636$ genetic testing made sense for them,

NOTE Confidence: 0.851953569999999

 $00:50:25.640 \longrightarrow 00:50:27.425$ and then then say the option for

NOTE Confidence: 0.851953569999999

 $00:50:27.425 \longrightarrow 00:50:29.371$ you is to have it done through

NOTE Confidence: 0.851953569999999

 $00:50:29.371 \longrightarrow 00:50:31.540$ the study or talk to your doctor.

NOTE Confidence: 0.851953569999999

 $00:50:31.540 \longrightarrow 00:50:31.780$ Further,

NOTE Confidence: 0.851953569999999

 $00:50:31.780 \longrightarrow 00:50:33.940$ if you don't want to be in the study

NOTE Confidence: 0.851953569999999

 $00{:}50{:}33{.}996 \dashrightarrow 00{:}50{:}35{.}970$ so that the consent was really broken

NOTE Confidence: 0.851953569999999

 $00:50:35.970 \rightarrow 00:50:37.998$ down into these two different pieces,

NOTE Confidence: 0.851953569999999

 $00:50:38.000 \rightarrow 00:50:40.100$ I want to say briefly something about.

NOTE Confidence: 0.851953569999999

00:50:40.100 --> 00:50:42.024 Our risk assessment approach,

NOTE Confidence: 0.851953569999999

 $00{:}50{:}42.024 \dashrightarrow 00{:}50{:}45.560$ 'cause the idea is that people had to.

NOTE Confidence: 0.851953569999999

00:50:45.560 --> 00:50:47.460 Normally family history is completed

NOTE Confidence: 0.851953569999999

00:50:47.460 --> 00:50:50.140 by a clinician and there are validated NOTE Confidence: 0.851953569999999 $00:50:50.140 \longrightarrow 00:50:52.288$ tools for clinicians to use for.

NOTE Confidence: 0.73455286

00:50:54.460 --> 00:50:55.820 For testing for Lynch syndrome,

NOTE Confidence: 0.73455286

 $00:50:55.820 \longrightarrow 00:50:57.955$ for example, so we wanted to do

NOTE Confidence: 0.73455286

 $00:50:57.955 \rightarrow 00:50:59.785$ with working with the collaborative

NOTE Confidence: 0.73455286

 $00{:}50{:}59{.}785 \dashrightarrow 00{:}51{:}02{.}100$ what's called the Prim. We

NOTE Confidence: 0.7347506

 $00:51:04.380 \longrightarrow 00:51:06.348$ modified it so this could be.

NOTE Confidence: 0.8597141

00:51:08.370 --> 00:51:10.407 Adapted so it will be accessible to

NOTE Confidence: 0.8597141

 $00:51:10.407 \rightarrow 00:51:12.349$ patients who are doing this online,

NOTE Confidence: 0.8597141

 $00{:}51{:}12{.}350 \dashrightarrow 00{:}51{:}15{.}410$ so we had to really make some changes in it.

NOTE Confidence: 0.8597141

 $00:51:15.410 \dashrightarrow 00:51:17.240$ We had we simplified the language.

NOTE Confidence: 0.8597141

 $00{:}51{:}17{.}240 \dashrightarrow 00{:}51{:}18{.}770$ We use less medical terms.

NOTE Confidence: 0.8597141

 $00:51:18.770 \longrightarrow 00:51:20.684$ We actually did the calculations for

NOTE Confidence: 0.8597141

 $00{:}51{:}20{.}684 \dashrightarrow 00{:}51{:}22{.}877$ the participants and we had visual aids

NOTE Confidence: 0.8597141

 $00{:}51{:}22.877 \dashrightarrow 00{:}51{:}24.886$ also to help describe what this meant,

NOTE Confidence: 0.8597141

 $00:51:24.890 \rightarrow 00:51:28.986$ and we also have done some of valuations.

NOTE Confidence: 0.8597141

 $00:51:28.990 \longrightarrow 00:51:30.790$ Actually forgot that we also do

- NOTE Confidence: 0.8597141
- $00:51:30.790 \longrightarrow 00:51:32.748$ some evaluations of how well this

00:51:32.748 --> 00:51:34.836 modification work and present that today,

NOTE Confidence: 0.8597141

 $00:51:34.840 \longrightarrow 00:51:36.465$ but this is actually trying

NOTE Confidence: 0.8597141

 $00:51:36.465 \rightarrow 00:51:38.090$ to work really very well,

NOTE Confidence: 0.8597141

 $00{:}51{:}38{.}090 \dashrightarrow 00{:}51{:}40{.}619$ so we have been able to create and develop

NOTE Confidence: 0.8597141

 $00{:}51{:}40{.}619 \dashrightarrow 00{:}51{:}43{.}158$ a tool that people can fill out both

NOTE Confidence: 0.8597141

 $00:51:43.158 \dashrightarrow 00:51:45.568$ in English and Spanish that describe.

NOTE Confidence: 0.8597141

 $00:51:45.570 \rightarrow 00:51:48.314$ It helps to understand what their actual risk

NOTE Confidence: 0.8597141

 $00:51:48.314 \rightarrow 00:51:50.997$ of having hereditary cancer syndrome are.

NOTE Confidence: 0.8597141

 $00:51:51.000 \rightarrow 00:51:53.616$ They were the most important things we done.

NOTE Confidence: 0.8597141

 $00{:}51{:}53{.}620 \dashrightarrow 00{:}51{:}55{.}924$ The study is that we wanted to think

NOTE Confidence: 0.8597141

00:51:55.924 --> 00:51:57.965 through how to do genetic counseling

NOTE Confidence: 0.8597141

 $00{:}51{:}57{.}965 \dashrightarrow 00{:}52{:}00{.}059$ most effectively and we wanted to

NOTE Confidence: 0.8597141

 $00{:}52{:}00{.}120 \dashrightarrow 00{:}52{:}02{.}106$ use the information that you saw

NOTE Confidence: 0.8597141

 $00:52:02.106 \rightarrow 00:52:03.756$ in that previous paper regarding

 $00:52:03.756 \longrightarrow 00:52:05.386$ an approach to informed consent.

NOTE Confidence: 0.8597141

 $00{:}52{:}05{.}390 \dashrightarrow 00{:}52{:}08{.}038$ And we also want to apply that to

NOTE Confidence: 0.8597141

 $00{:}52{:}08.038 \dashrightarrow 00{:}52{:}09.967$ Jenna Counseling an for this study.

NOTE Confidence: 0.8597141

 $00{:}52{:}09{.}970 \dashrightarrow 00{:}52{:}11.600$ We actually collaborated with women

NOTE Confidence: 0.8597141

 $00{:}52{:}11.600 \dashrightarrow 00{:}52{:}13.190$ in effort PA. Logistic gallon.

NOTE Confidence: 0.8597141

00:52:13.190 --> 00:52:15.020 Joseph is at University of California

NOTE Confidence: 0.8597141

00:52:15.020 --> 00:52:16.180 at San Francisco.

NOTE Confidence: 0.8597141

 $00:52:16.180 \longrightarrow 00:52:18.172$ Who's been studying Jenna councils for

NOTE Confidence: 0.8597141

 $00{:}52{:}18.172 \dashrightarrow 00{:}52{:}20.200$ years and together we collaborate on

NOTE Confidence: 0.8597141

 $00:52:20.200 \rightarrow 00:52:22.768$ creating what we have now described as the.

NOTE Confidence: 0.8597141

 $00{:}52{:}22{.}770 \dashrightarrow 00{:}52{:}24{.}945$ Aria model of gender counseling

NOTE Confidence: 0.8597141

 $00{:}52{:}24.945 \dashrightarrow 00{:}52{:}27.120$ that refers to being accessible,

NOTE Confidence: 0.8597141

 $00{:}52{:}27{.}120 \dashrightarrow 00{:}52{:}29{.}240$ relational, inclusive, an actionable,

NOTE Confidence: 0.8597141

 $00:52:29.240 \longrightarrow 00:52:32.420$ and so the idea is to.

NOTE Confidence: 0.8597141

00:52:32.420 --> 00:52:32.832 Again,

NOTE Confidence: 0.8597141

 $00:52:32.832 \rightarrow 00:52:35.304$ make the information easy to understand.

 $00{:}52{:}35{.}310 \dashrightarrow 00{:}52{:}37{.}674$ Really focus on the relationship between

NOTE Confidence: 0.8597141

 $00{:}52{:}37{.}674 \dashrightarrow 00{:}52{:}41{.}032$ the clinician and the provider as a way

NOTE Confidence: 0.8597141

 $00:52:41.032 \rightarrow 00:52:42.748$ of helping facilitate understanding,

NOTE Confidence: 0.8597141

 $00:52:42.750 \longrightarrow 00:52:44.151$ but more importantly,

NOTE Confidence: 0.8597141

 $00{:}52{:}44{.}151 \dashrightarrow 00{:}52{:}46{.}953$ helping to facilitate and ensure that

NOTE Confidence: 0.8597141

 $00{:}52{:}46{.}953 \dashrightarrow 00{:}52{:}48{.}924$ patients understand the actions and

NOTE Confidence: 0.8597141

 $00{:}52{:}48{.}924 \dashrightarrow 00{:}52{:}51{.}830$ know what steps to do the next time.

NOTE Confidence: 0.8597141

 $00:52:51.830 \longrightarrow 00:52:55.134$ So the idea, and So what we did,

NOTE Confidence: 0.8597141

 $00{:}52{:}55{.}140 \dashrightarrow 00{:}52{:}57{.}385$ we developed a fairly detailed

NOTE Confidence: 0.8597141

 $00:52:57.385 \longrightarrow 00:52:59.630$ curriculum to train 2 genetic

NOTE Confidence: 0.8597141

 $00{:}52{:}59{.}712 \dashrightarrow 00{:}53{:}02{.}407$ counselors to use the Aria model an.

NOTE Confidence: 0.8597141

 $00{:}53{:}02{.}410 \dashrightarrow 00{:}53{:}03{.}418$ Other two genetic counselors.

NOTE Confidence: 0.8597141

 $00{:}53{:}03{.}418 \dashrightarrow 00{:}53{:}04{.}930$ Did this get their standard approach

NOTE Confidence: 0.8597141

 $00{:}53{:}04{.}975 \dashrightarrow 00{:}53{:}06{.}427$ to gender counseling that begins with.

NOTE Confidence: 0.8597141

 $00:53:06.430 \longrightarrow 00:53:07.430$ This is the gene.

 $00:53:07.430 \rightarrow 00:53:08.430$ This is a chromosome.

NOTE Confidence: 0.8597141

00:53:08.430 --> 00:53:09.690 Let's take your family history.

NOTE Confidence: 0.8597141

 $00:53:09.690 \rightarrow 00:53:11.447$ Let's tell you what your results are.

NOTE Confidence: 0.8597141

 $00:53:11.450 \longrightarrow 00:53:12.950$ Let's tell you what it means.

NOTE Confidence: 0.8597141

00:53:12.950 --> 00:53:14.714 Let's get into the complexity of

NOTE Confidence: 0.8597141

 $00{:}53{:}14.714 \dashrightarrow 00{:}53{:}16.570$ this because it's always complicated.

NOTE Confidence: 0.8597141

 $00:53:16.570 \longrightarrow 00:53:20.170$ And then will tell you what to do, maybe so.

NOTE Confidence: 0.8597141

 $00:53:20.170 \longrightarrow 00:53:20.530$ Again,

NOTE Confidence: 0.8597141

00:53:20.530 --> 00:53:21.250 here's that.

NOTE Confidence: 0.8597141

 $00:53:21.250 \longrightarrow 00:53:23.536$ Here's a very quick example of

NOTE Confidence: 0.8597141

 $00{:}53{:}23{.}536 \dashrightarrow 00{:}53{:}25{.}060$ the distinction between usual

NOTE Confidence: 0.8597141

 $00:53:25.125 \longrightarrow 00:53:26.650$ care and the Aria model.

NOTE Confidence: 0.8597141

 $00:53:26.650 \rightarrow 00:53:29.170$ I'll put the right hand side first.

NOTE Confidence: 0.8597141

00:53:29.170 --> 00:53:29.466 Again,

NOTE Confidence: 0.8597141

 $00{:}53{:}29{.}466 \dashrightarrow 00{:}53{:}31{.}538$ this notion that the emphasis and usual

NOTE Confidence: 0.8597141

 $00{:}53{:}31{.}538 \dashrightarrow 00{:}53{:}33{.}486$ care is on transferred information

- NOTE Confidence: 0.8597141
- $00:53:33.486 \rightarrow 00:53:35.646$ is usually built technical language.
- NOTE Confidence: 0.8597141
- $00:53:35.650 \rightarrow 00:53:37.810$ It's usually more of a monologue
- NOTE Confidence: 0.8597141
- 00:53:37.810 --> 00:53:38.890 less participant engagement,
- NOTE Confidence: 0.8597141
- $00:53:38.890 \rightarrow 00:53:41.410$ an often it's historically is non directive.
- NOTE Confidence: 0.8597141
- $00:53:41.410 \longrightarrow 00:53:44.290$ It's like you may want to do this.
- NOTE Confidence: 0.8597141
- $00:53:44.290 \longrightarrow 00:53:46.558$ You may want to do this.
- NOTE Confidence: 0.8597141
- $00:53:46.560 \longrightarrow 00:53:48.935$ Whereas the Aria model was
- NOTE Confidence: 0.8597141
- $00:53:48.935 \longrightarrow 00:53:51.310$ meant to really focus on.
- NOTE Confidence: 0.8597141
- 00:53:51.310 --> 00:53:52.954 A model of psychosocial
- NOTE Confidence: 0.8597141
- $00{:}53{:}52{.}954 \dashrightarrow 00{:}53{:}54{.}187$ counseling establishing a
- NOTE Confidence: 0.8597141
- $00{:}53{:}54{.}187 \dashrightarrow 00{:}53{:}56{.}180$ relationship with the participant.
- NOTE Confidence: 0.8597141
- $00{:}53{:}56{.}180 \dashrightarrow 00{:}53{:}58{.}838$ You are using direct and simple
- NOTE Confidence: 0.8597141
- $00:53:58.838 \longrightarrow 00:54:00.610$ language being very clear.
- NOTE Confidence: 0.8597141
- $00{:}54{:}00{.}610 \dashrightarrow 00{:}54{:}03{.}850$ What you should do next.
- NOTE Confidence: 0.8597141
- 00:54:03.850 --> 00:54:06.755 You you should do X&X as well,
- NOTE Confidence: 0.8597141

 $00:54:06.760 \rightarrow 00:54:08.420$ avoiding uncertainty with this.

NOTE Confidence: 0.8762343

00:54:11.910 --> 00:54:13.814 If more if Mark if you invite

NOTE Confidence: 0.8762343

 $00:54:13.814 \longrightarrow 00:54:15.789$ me back in a year from now,

NOTE Confidence: 0.8762343

 $00:54:15.790 \longrightarrow 00:54:17.380$ we will tell you the results

NOTE Confidence: 0.8762343

 $00:54:17.380 \longrightarrow 00:54:19.110$ of this part of the study.

NOTE Confidence: 0.8762343

 $00:54:19.110 \longrightarrow 00:54:20.766$ I don't have those results yet,

NOTE Confidence: 0.8762343

00:54:20.770 --> 00:54:22.758 but I can share with you because

NOTE Confidence: 0.8762343

 $00:54:22.758 \rightarrow 00:54:24.145$ we've now completed the testing

NOTE Confidence: 0.8762343

00:54:24.145 --> 00:54:26.065 is I can at least share with you

NOTE Confidence: 0.8762343

 $00{:}54{:}26{.}123 \dashrightarrow 00{:}54{:}27{.}977$ briefly who enrolled in the study.

NOTE Confidence: 0.8762343

 $00:54:27.980 \longrightarrow 00:54:29.360$ We have roughly 800 people

NOTE Confidence: 0.8762343

 $00:54:29.360 \longrightarrow 00:54:30.464$ enrolled in the study,

NOTE Confidence: 0.8762343

 $00{:}54{:}30{.}470 \dashrightarrow 00{:}54{:}32{.}526$ and what I want to share with you

NOTE Confidence: 0.8762343

 $00{:}54{:}32{.}526 \dashrightarrow 00{:}54{:}34{.}741$ briefly is the fact that we were able to

NOTE Confidence: 0.8762343

 $00{:}54{:}34{.}741 \dashrightarrow 00{:}54{:}36{.}811$ get a more diverse population that we NOTE Confidence: 0.8762343

 $00:54:36.811 \rightarrow 00:54:39.063$ had with our first study that roughly,

- NOTE Confidence: 0.8762343
- 00:54:39.063 --> 00:54:41.044 under under 50% identified as white and

 $00{:}54{:}41{.}044 \dashrightarrow 00{:}54{:}43{.}000$ the rest of the patients identified.

NOTE Confidence: 0.8762343

00:54:43.000 --> 00:54:43.608 Primarily Hispanic,

NOTE Confidence: 0.8762343

 $00:54:43.608 \rightarrow 00:54:46.600$ but in a variety of other groups as well too.

NOTE Confidence: 0.8762343

 $00:54:46.600 \longrightarrow 00:54:48.768$ We are also able to try to have

NOTE Confidence: 0.8762343

 $00:54:48.768 \longrightarrow 00:54:50.559$ a slightly increased amount in

NOTE Confidence: 0.8762343

00:54:50.559 --> 00:54:52.135 terms of educational diversity

NOTE Confidence: 0.8762343

 $00:54:52.135 \rightarrow 00:54:54.480$ as well as income diversity.

NOTE Confidence: 0.8762343

 $00{:}54{:}54{.}480 \dashrightarrow 00{:}54{:}56{.}538$ I without going into the details,

NOTE Confidence: 0.8762343

 $00{:}54{:}56{.}540 \dashrightarrow 00{:}54{:}58{.}388$ I briefly want to acknowledge for

NOTE Confidence: 0.8762343

 $00{:}54{:}58{.}388 \dashrightarrow 00{:}55{:}00{.}819$ those of you in the audience that

NOTE Confidence: 0.8762343

 $00{:}55{:}00{.}819 \dashrightarrow 00{:}55{:}02{.}704$ really are curious about this.

NOTE Confidence: 0.8762343

00:55:02.710 --> 00:55:03.708 In fact,

NOTE Confidence: 0.8762343

 $00{:}55{:}03.708 \dashrightarrow 00{:}55{:}06.702$ we were able to identify through

NOTE Confidence: 0.8762343

 $00:55:06.702 \rightarrow 00:55:08.390$ the charm study.

 $00:55:08.390 \longrightarrow 00:55:10.215$ Quite a number of positive

NOTE Confidence: 0.8762343

 $00:55:10.215 \rightarrow 00:55:12.610$ results of the 800 people we had,

NOTE Confidence: 0.8762343

 $00{:}55{:}12.610 \dashrightarrow 00{:}55{:}14.898$ we had 36 people who had positive results

NOTE Confidence: 0.8762343

 $00:55:14.898 \rightarrow 00:55:17.189$ or other pathogenic likely pathogenic.

NOTE Confidence: 0.8762343

 $00{:}55{:}17{.}190 \dashrightarrow 00{:}55{:}19{.}398$ We also did some additional additional

NOTE Confidence: 0.8762343

 $00:55:19.398 \rightarrow 00:55:21.247$ findings related to other things

NOTE Confidence: 0.8762343

 $00:55:21.247 \rightarrow 00:55:23.167$ that can be found in sequencing.

NOTE Confidence: 0.8762343

00:55:23.170 --> 00:55:23.523 Again,

NOTE Confidence: 0.8762343

 $00{:}55{:}23.523 \dashrightarrow 00{:}55{:}25.288$ want to emphasize that sequencing

NOTE Confidence: 0.8762343

00:55:25.288 --> 00:55:27.100 doesn't mean you find everything

NOTE Confidence: 0.8762343

 $00{:}55{:}27.100 \dashrightarrow 00{:}55{:}28.805$ we had to make decisions.

NOTE Confidence: 0.8762343

 $00:55:28.810 \longrightarrow 00:55:31.232$ About 126 genes that we felt were

NOTE Confidence: 0.8762343

 $00{:}55{:}31{.}232 \dashrightarrow 00{:}55{:}33{.}007$ appropriate to include in our

NOTE Confidence: 0.8762343

00:55:33.007 - > 00:55:34.343 additional findings panel that

NOTE Confidence: 0.8762343

 $00:55:34.343 \rightarrow 00:55:36.549$ we were going to be reporting.

NOTE Confidence: 0.8762343

 $00:55:36.550 \rightarrow 00:55:39.448$ So anytime you hear the word sequencing.

- NOTE Confidence: 0.8762343
- $00:55:39.450 \longrightarrow 00:55:40.734$ It doesn't mean everything.

 $00{:}55{:}40{.}734 \dashrightarrow 00{:}55{:}42{.}660$ It means a deliberate choice to

NOTE Confidence: 0.8762343

 $00:55:42.715 \rightarrow 00:55:44.617$ include some things and not others.

NOTE Confidence: 0.8762343

 $00:55:44.620 \rightarrow 00:55:46.420$ Last point I'll make before I

NOTE Confidence: 0.8762343

 $00{:}55{:}46{.}420 \dashrightarrow 00{:}55{:}48{.}392$ end it 'cause Mark said I have

NOTE Confidence: 0.8762343

 $00:55:48.392 \longrightarrow 00:55:49.808$ to have up to an hour.

NOTE Confidence: 0.8762343

 $00{:}55{:}49{.}810 \dashrightarrow 00{:}55{:}51{.}418$ We have time for conversation is

NOTE Confidence: 0.8762343

 $00:55:51.418 \rightarrow 00:55:53.360$ as much about two points to make,

NOTE Confidence: 0.8762343

 $00{:}55{:}53{.}360 \dashrightarrow 00{:}55{:}56{.}312$ so another 5 minutes I want to at least

NOTE Confidence: 0.8762343

 $00{:}55{:}56{.}312 \dashrightarrow 00{:}55{:}58{.}052$ acknowledge that we were successful

NOTE Confidence: 0.8762343

 $00{:}55{:}58.052 \dashrightarrow 00{:}56{:}00.436$ in reaching people who had a bear

NOTE Confidence: 0.8762343

 $00{:}56{:}00{.}436 \dashrightarrow 00{:}56{:}02{.}900$ who had a range of barriers to access.

NOTE Confidence: 0.8762343

 $00{:}56{:}02{.}900 \dashrightarrow 00{:}56{:}05{.}399$ And have it broken down by these.

NOTE Confidence: 0.8762343

00:56:05.400 --> 00:56:07.220 By these categories I also want to

NOTE Confidence: 0.8762343

 $00{:}56{:}07{.}220 \dashrightarrow 00{:}56{:}09{.}139$ point out and I thought this was

 $00:56:09.139 \dashrightarrow 00:56:11.090$ very important that but we were able.

NOTE Confidence: 0.8762343

 $00{:}56{:}11.090 \dashrightarrow 00{:}56{:}12.987$ We also included and we also asked

NOTE Confidence: 0.8762343

00:56:12.987 --> 00:56:14.328 about ****** or gender minorities,

NOTE Confidence: 0.8762343

00:56:14.328 --> 00:56:16.190 which are often a group that are

NOTE Confidence: 0.8762343

 $00:56:16.239 \longrightarrow 00:56:17.835$ maybe less likely have access to

NOTE Confidence: 0.8762343

 $00{:}56{:}17.835 \dashrightarrow 00{:}56{:}19.490$ testing for a variety of reasons.

NOTE Confidence: 0.8762343

00:56:19.490 --> 00:56:21.093 I also have an complications of what

NOTE Confidence: 0.8762343

 $00{:}56{:}21.093 \dashrightarrow 00{:}56{:}22.539$ testing may be important to them

NOTE Confidence: 0.8762343

 $00{:}56{:}22{.}539 \dashrightarrow 00{:}56{:}23{.}967$ because they may not have access

NOTE Confidence: 0.8762343

 $00{:}56{:}23.967 \dashrightarrow 00{:}56{:}25.663$ to family history information if

NOTE Confidence: 0.8762343

 $00{:}56{:}25{.}663 \dashrightarrow 00{:}56{:}27{.}075$ they have strained relationships.

NOTE Confidence: 0.8762343

 $00{:}56{:}27.080 \dashrightarrow 00{:}56{:}29.636$ And this is one of the things that we

NOTE Confidence: 0.8762343

 $00{:}56{:}29.636 \dashrightarrow 00{:}56{:}31.547$ learned from interviewing people from

NOTE Confidence: 0.8762343

 $00{:}56{:}31{.}547 \dashrightarrow 00{:}56{:}34{.}589$ that population who were part of our study.

NOTE Confidence: 0.8762343

 $00:56:34.590 \rightarrow 00:56:36.936$ This side very quickly just suggested

NOTE Confidence: 0.8762343

 $00:56:36.936 \dashrightarrow 00:56:39.249$ during the course of the study.

- NOTE Confidence: 0.8762343
- $00:56:39.250 \rightarrow 00:56:41.386$ Which include the risk assessment tool,

 $00:56:41.390 \longrightarrow 00:56:42.461$ which term eligibility

NOTE Confidence: 0.8762343

 $00:56:42.461 \longrightarrow 00:56:44.603$ agreement to be in the study,

NOTE Confidence: 0.8762343

 $00{:}56{:}44.610 \dashrightarrow 00{:}56{:}47.403$ returning a saliva kit and then completing

NOTE Confidence: 0.8762343

 $00{:}56{:}47{.}403 \dashrightarrow 00{:}56{:}49{.}600$ results disclosure as as you can see,

NOTE Confidence: 0.8762343

 $00{:}56{:}49{.}600 \dashrightarrow 00{:}56{:}51{.}028$ there was a gradual

NOTE Confidence: 0.8762343

 $00:56:51.028 \rightarrow 00:56:52.456$ degradation along the way,

NOTE Confidence: 0.82956165

 $00:56:52.460 \longrightarrow 00:56:54.902$ but this was not the study

NOTE Confidence: 0.82956165

 $00{:}56{:}54{.}902 \dashrightarrow 00{:}56{:}56{.}530$ successfully retained that population

NOTE Confidence: 0.82956165

 $00{:}56{:}56{.}597 \dashrightarrow 00{:}56{:}58{.}505$ who did have barriers to access

NOTE Confidence: 0.82956165

 $00{:}56{:}58{.}505 \dashrightarrow 00{:}57{:}00{.}668$ a related to being in the study.

NOTE Confidence: 0.85935867

 $00{:}57{:}03{.}400 \dashrightarrow 00{:}57{:}05{.}872$ Because I want to skip this slide simply

NOTE Confidence: 0.85935867

 $00{:}57{:}05{.}872 \dashrightarrow 00{:}57{:}07{.}810$ because we're running short on time.

NOTE Confidence: 0.85935867

00:57:07.810 --> 00:57:10.330 I just want knowledge we're still doing this.

NOTE Confidence: 0.85935867

 $00{:}57{:}10{.}330 \dashrightarrow 00{:}57{:}12{.}409$ We are still trying to evaluate potential
$00:57:12.409 \longrightarrow 00:57:14.684$ risks of this approach in terms of over

NOTE Confidence: 0.85935867

 $00{:}57{:}14.684 \dashrightarrow 00{:}57{:}16.819$ testing in terms of the results themselves

NOTE Confidence: 0.85935867

 $00{:}57{:}16.819 \dashrightarrow 00{:}57{:}19.465$ in terms of the implications for counselors.

NOTE Confidence: 0.85935867

 $00:57:19.470 \rightarrow 00:57:22.296$ So there's a lot of work we're still doing,

NOTE Confidence: 0.85935867

 $00:57:22.300 \dashrightarrow 00:57:24.862$ but I want to end with switching

NOTE Confidence: 0.85935867

 $00{:}57{:}24.862 \dashrightarrow 00{:}57{:}26.963$ gears slightly and talking about

NOTE Confidence: 0.85935867

 $00:57:26.963 \rightarrow 00:57:28.827$ something else that will.

NOTE Confidence: 0.85935867

 $00:57:28.830 \rightarrow 00:57:31.770$ To give you some final thoughts.

NOTE Confidence: 0.85935867

 $00:57:31.770 \dashrightarrow 00:57:35.074$ That's right, so here's my final thought.

NOTE Confidence: 0.85935867

 $00{:}57{:}35{.}080 \dashrightarrow 00{:}57{:}38{.}760$ And it's it's kind of thinking back to.

NOTE Confidence: 0.85935867

 $00{:}57{:}38{.}760 \dashrightarrow 00{:}57{:}40{.}413$ Related back to.

NOTE Confidence: 0.85935867

00:57:40.413 --> 00:57:42.066 Gattaca an graduate,

NOTE Confidence: 0.85935867

 $00{:}57{:}42.070 \dashrightarrow 00{:}57{:}44.114$ and so I want to end with

NOTE Confidence: 0.85935867

 $00{:}57{:}44.114 \dashrightarrow 00{:}57{:}45.619$ something from Winnie the Pooh.

NOTE Confidence: 0.85935867

 $00:57:45.620 \rightarrow 00:57:47.820$ So a number of years ago I wrote an essay

NOTE Confidence: 0.85935867

 $00:57:47.881 \rightarrow 00:57:49.876$ in response to somebody who is looking

- NOTE Confidence: 0.85935867
- $00:57:49.876 \rightarrow 00:57:51.958$ at the ethical implications of genome

00:57:51.958 --> 00:57:53.908 sequencing in the prenatal context,

NOTE Confidence: 0.85935867

 $00:57:53.910 \longrightarrow 00:57:56.046$ and as I thought about that

NOTE Confidence: 0.85935867

 $00:57:56.046 \rightarrow 00:57:57.470$ question of doing prenatal.

NOTE Confidence: 0.85935867

00:57:57.470 --> 00:57:59.194 Genome sequencing audit infant,

NOTE Confidence: 0.85935867

00:57:59.194 --> 00:58:01.780 I thought about Winnie the Pooh,

NOTE Confidence: 0.85935867

 $00:58:01.780 \longrightarrow 00:58:03.940$ and I thought about that.

NOTE Confidence: 0.85935867

 $00:58:03.940 \rightarrow 00:58:06.520$ The phenotypes of Winnie the Pooh,

NOTE Confidence: 0.85935867

 $00:58:06.520 \rightarrow 00:58:09.106$ and I thought about various characters,

NOTE Confidence: 0.85935867

 $00:58:09.110 \longrightarrow 00:58:12.200$ so ER. The person is sceptical.

NOTE Confidence: 0.85935867

 $00:58:12.200 \longrightarrow 00:58:14.447$ Jack testing thinks of low in despair.

NOTE Confidence: 0.85935867

 $00{:}58{:}14.450 \dashrightarrow 00{:}58{:}16.376$ This is a really bad idea,

NOTE Confidence: 0.85935867

 $00:58:16.380 \longrightarrow 00:58:17.660$ and thinking about Tigger

NOTE Confidence: 0.85935867

00:58:17.660 --> 00:58:18.300 always enthusiastic,

NOTE Confidence: 0.85935867

 $00:58:18.300 \rightarrow 00:58:21.810$ always wanting to say well we can do this.

 $00:58:21.810 \rightarrow 00:58:24.393$ And it seems to be a lot of enthusiasm.

NOTE Confidence: 0.85935867

00:58:24.400 --> 00:58:26.376 A lot of the issues in genomics are

NOTE Confidence: 0.85935867

 $00:58:26.376 \longrightarrow 00:58:28.544$ the ERS in the triggers and 1:00

NOTE Confidence: 0.85935867

00:58:28.544 --> 00:58:30.154 remind us of Christopher Robin,

NOTE Confidence: 0.85935867

00:58:30.160 --> 00:58:31.812 who as the protagonist you know how

NOTE Confidence: 0.85935867

 $00{:}58{:}31{.}812 \dashrightarrow 00{:}58{:}34{.}085$ to get a commitment to moving forward NOTE Confidence: 0.85935867

 $00{:}58{:}34.085 \dashrightarrow 00{:}58{:}35.965$ to nurturing those relationships with

NOTE Confidence: 0.85935867

 $00{:}58{:}35{.}965 \dashrightarrow 00{:}58{:}38{.}225$ his friends in the in the wood.

NOTE Confidence: 0.85935867

 $00{:}58{:}38{.}230 \dashrightarrow 00{:}58{:}39{.}940$ But also realizing how to leave

NOTE Confidence: 0.85935867

 $00{:}58{:}39{.}940 \dashrightarrow 00{:}58{:}41{.}995$ there and had to be very careful

NOTE Confidence: 0.85935867

00:58:41.995 --> 00:58:43.990 and thoughtful of how he did it.

NOTE Confidence: 0.85935867

 $00{:}58{:}43{.}990 \dashrightarrow 00{:}58{:}45{.}510$ And of course, poo.

NOTE Confidence: 0.85935867

00:58:45.510 --> 00:58:47.790 Let's let's find some more honey

NOTE Confidence: 0.85935867

 $00:58:47.864 \rightarrow 00:58:50.125$ who thought this is a bunch of.

NOTE Confidence: 0.85935867

 $00{:}58{:}50{.}130 \dashrightarrow 00{:}58{:}53{.}138$ Crap bunch of honey.

NOTE Confidence: 0.85935867

 $00:58:53.140 \rightarrow 00:58:55.020$ There's more important things than

 $00:58:55.020 \rightarrow 00:58:56.524$ worrying about genetic testing.

NOTE Confidence: 0.85935867

 $00{:}58{:}56{.}530 \dashrightarrow 00{:}58{:}58{.}186$ One of the things that people

NOTE Confidence: 0.85935867

 $00:58:58.186 \longrightarrow 00:58:59.780$ worry about with Jenn testing,

NOTE Confidence: 0.85935867

00:58:59.780 --> 00:59:00.629 particularly for children,

NOTE Confidence: 0.85935867

 $00:59:00.629 \rightarrow 00:59:03.020$ and if we're looking for adult onset disease.

NOTE Confidence: 0.85935867

 $00:59:03.020 \longrightarrow 00:59:04.790$ Even for things like BRCA one

NOTE Confidence: 0.85935867

00:59:04.790 --> 00:59:05.970 or for Lynch syndrome,

NOTE Confidence: 0.85935867

 $00:59:05.970 \dashrightarrow 00:59:08.330$ is what does that mean for the child?

NOTE Confidence: 0.85935867

 $00{:}59{:}08{.}330 \dashrightarrow 00{:}59{:}09{.}805$ Whatever child finds out when

NOTE Confidence: 0.85935867

 $00:59:09.805 \longrightarrow 00:59:11.280$ they're there for the born,

NOTE Confidence: 0.85935867

 $00:59:11.280 \longrightarrow 00:59:13.080$ the parents right before the

NOTE Confidence: 0.85935867

 $00{:}59{:}13.080 \dashrightarrow 00{:}59{:}15.419$ board they carry the BRCA 1 gene.

NOTE Confidence: 0.85935867

 $00:59:15.420 \longrightarrow 00:59:17.643$ Is that appropriate to do that for a child?

NOTE Confidence: 0.85935867

 $00{:}59{:}17.650 \dashrightarrow 00{:}59{:}19.522$ Don't they have a right to an open

NOTE Confidence: 0.85935867

 $00{:}59{:}19{.}522 \dashrightarrow 00{:}59{:}21{.}092$ future which is a term that's been

 $00:59:21.092 \longrightarrow 00:59:22.772$ used for the last 20 years as

NOTE Confidence: 0.85935867

 $00{:}59{:}22.772 \dashrightarrow 00{:}59{:}24.217$ we were thinking about questions

NOTE Confidence: 0.85935867

 $00:59:24.217 \longrightarrow 00:59:25.337$ about testing in children?

NOTE Confidence: 0.85935867

 $00:59:25.337 \rightarrow 00:59:27.313$ Since this is a pediatric audience in part,

NOTE Confidence: 0.85935867

 $00{:}59{:}27{.}320 \dashrightarrow 00{:}59{:}30{.}414$ I want to end on this note.

NOTE Confidence: 0.85935867

 $00{:}59{:}30{.}420 \dashrightarrow 00{:}59{:}32{.}380$ To suggest is that the right and open

NOTE Confidence: 0.85935867

 $00{:}59{:}32{.}380 \dashrightarrow 00{:}59{:}34{.}282$ future is a considerations that we

NOTE Confidence: 0.85935867

 $00:59:34.282 \rightarrow 00:59:36.316$ thought about by parents and clinicians,

NOTE Confidence: 0.85935867

 $00{:}59{:}36{.}320 \dashrightarrow 00{:}59{:}38{.}385$ but perhaps not is a final answer.

NOTE Confidence: 0.85935867

 $00{:}59{:}38{.}390 \dashrightarrow 00{:}59{:}40{.}614$ But when the parents may want to think

NOTE Confidence: 0.85935867

 $00{:}59{:}40{.}614 \dashrightarrow 00{:}59{:}42{.}722$ about into account all decisions that

NOTE Confidence: 0.85935867

 $00:59:42.722 \dashrightarrow 00:59:44.582$ parents make affect their child.

NOTE Confidence: 0.85935867

 $00{:}59{:}44{.}590 \dashrightarrow 00{:}59{:}46{.}634$ And it really depends on the context

NOTE Confidence: 0.85935867

 $00{:}59{:}46{.}634 \dashrightarrow 00{:}59{:}48{.}829$ of the request about whether or not

NOTE Confidence: 0.85935867

 $00{:}59{:}48.829 \dashrightarrow 00{:}59{:}50.719$ this really will have a meaningful

NOTE Confidence: 0.8734655

 $00:59:50.777 \rightarrow 00:59:51.977$ impact on the future,

- NOTE Confidence: 0.8734655
- $00:59:51.980 \dashrightarrow 00:59:55.346$ in a way that's even concerning.

 $00{:}59{:}55{.}350 \dashrightarrow 00{:}59{:}57{.}835$ And even if we cannot predict problems,

NOTE Confidence: 0.8734655

 $00:59:57.840 \rightarrow 01:00:00.458$ it doesn't mean that we should preclude.

NOTE Confidence: 0.8734655

 $01:00:00.460 \rightarrow 01:00:02.568$ Decisions by parents based

NOTE Confidence: 0.8734655

 $01:00:02.568 \longrightarrow 01:00:04.149$ upon their interests.

NOTE Confidence: 0.8734655

 $01:00:04.150 \longrightarrow 01:00:05.760$ And so, in other words,

NOTE Confidence: 0.8734655

 $01:00:05.760 \longrightarrow 01:00:07.692$ even if we know that there might

NOTE Confidence: 0.8734655

 $01:00:07.692 \rightarrow 01:00:09.555$ be some possible concerns for some

NOTE Confidence: 0.8734655

 $01{:}00{:}09{.}555 \dashrightarrow 01{:}00{:}11{.}185$ parents about doing testing early

NOTE Confidence: 0.8734655

01:00:11.185 --> 01:00:13.140 on for double onset diseases,

NOTE Confidence: 0.8734655

 $01{:}00{:}13.140 \dashrightarrow 01{:}00{:}16.140$ parents might still make that decision.

NOTE Confidence: 0.8734655

 $01:00:16.140 \longrightarrow 01:00:18.760$ Now. Why is that so?

NOTE Confidence: 0.8734655

 $01:00:18.760 \longrightarrow 01:00:19.860$ That maybe is a question

NOTE Confidence: 0.8734655

 $01:00:19.860 \longrightarrow 01:00:21.049$ maybe for the discussion,

NOTE Confidence: 0.8734655

 $01{:}00{:}21.050 \dashrightarrow 01{:}00{:}23.900$ but what I want to end on to have you

 $01:00:23.983 \rightarrow 01:00:27.070$ think about that is the following image.

NOTE Confidence: 0.8734655

 $01{:}00{:}27.070 \dashrightarrow 01{:}00{:}29.126$ For those of you who might know this,

NOTE Confidence: 0.8734655

01:00:29.130 --> 01:00:31.452 I'm not normally would ask this in a talk,

NOTE Confidence: 0.8734655

 $01:00:31.460 \rightarrow 01:00:34.358$ but this is actually Christopher Robin.

NOTE Confidence: 0.8734655

01:00:34.360 --> 01:00:34.760 Again,

NOTE Confidence: 0.8734655

 $01{:}00{:}34.760 \dashrightarrow 01{:}00{:}37.560$ the the instigator for Winnie the Pooh.

NOTE Confidence: 0.8734655

 $01{:}00{:}37{.}560 \dashrightarrow 01{:}00{:}39{.}464$ What you may some of you who are

NOTE Confidence: 0.8734655

 $01{:}00{:}39{.}464 \dashrightarrow 01{:}00{:}40{.}947$ familiar with Christopher Robin in the

NOTE Confidence: 0.8734655

 $01{:}00{:}40{.}947 \dashrightarrow 01{:}00{:}43{.}299$ story of a A Milne is that while this NOTE Confidence: 0.8734655

01:00:43.299 --> 01:00:45.039 while we're all familiar with Winnie NOTE Confidence: 0.8734655

 $01{:}00{:}45{.}039 \dashrightarrow 01{:}00{:}46{.}980$ the Pooh stories because we tell it

NOTE Confidence: 0.8734655

 $01{:}00{:}46.980 \dashrightarrow 01{:}00{:}48.900$ to our children and grandchildren.

NOTE Confidence: 0.8734655

01:00:48.900 --> 01:00:52.620 Christopher Robin was very unhappy

NOTE Confidence: 0.8734655

 $01{:}00{:}52.620 \dashrightarrow 01{:}00{:}54.540$ with his father because of those

NOTE Confidence: 0.8734655

 $01:00:54.540 \longrightarrow 01:00:56.220$ stories he felt he was bullied.

NOTE Confidence: 0.8734655

 $01{:}00{:}56{.}220 \dashrightarrow 01{:}00{:}57{.}740$ He felt he was ost racized.

- NOTE Confidence: 0.8734655
- $01{:}00{:}57.740 \dashrightarrow 01{:}00{:}59.940$ He became a strange for him from his

 $01{:}00{:}59{.}940 \dashrightarrow 01{:}01{:}01{.}834$ father for decades and only really

NOTE Confidence: 0.8734655

 $01:01:01.834 \longrightarrow 01:01:04.120$ towards the end of his father's life.

NOTE Confidence: 0.8734655

 $01:01:04.120 \rightarrow 01:01:06.250$ Did they reconcile?

NOTE Confidence: 0.8734655

 $01:01:06.250 \longrightarrow 01:01:08.800$ The point of this is that.

NOTE Confidence: 0.8734655

 $01:01:08.800 \longrightarrow 01:01:10.350$ Amil's decision to tell these

NOTE Confidence: 0.8734655

 $01:01:10.350 \longrightarrow 01:01:12.420$ stories had an impact on his son.

NOTE Confidence: 0.8734655

 $01:01:12.420 \rightarrow 01:01:15.138$ We don't know whether he was aware of that,

NOTE Confidence: 0.8734655

 $01:01:15.140 \longrightarrow 01:01:16.650$ whether he thought about that,

NOTE Confidence: 0.8734655

 $01:01:16.650 \rightarrow 01:01:18.160$ whether they should or shouldn't

NOTE Confidence: 0.8734655

 $01{:}01{:}18{.}160 \dashrightarrow 01{:}01{:}19{.}368$ have influenced his decision.

NOTE Confidence: 0.8734655

01:01:19.370 --> 01:01:19.978 He made,

NOTE Confidence: 0.8734655

 $01{:}01{:}19{.}978 \dashrightarrow 01{:}01{:}22{.}410$ and so I really want to point out

NOTE Confidence: 0.8734655

 $01:01:22.487 \rightarrow 01:01:24.827$ in a in a in a very sincere way

NOTE Confidence: 0.8734655

 $01{:}01{:}24.827 \dashrightarrow 01{:}01{:}27.124$ that while we do think about the

 $01:01:27.124 \rightarrow 01:01:29.238$ the impact on the open future,

NOTE Confidence: 0.8734655

 $01:01:29.238 \rightarrow 01:01:32.066$ we don't know what anyone's future will

NOTE Confidence: 0.8734655

 $01:01:32.066 \rightarrow 01:01:34.899$ be and what the impact will be and.

NOTE Confidence: 0.8734655

 $01:01:34.900 \longrightarrow 01:01:37.150$ Just like a million Christopher Robin.

NOTE Confidence: 0.8683241

 $01:01:39.210 \longrightarrow 01:01:41.016$ It's a journey, so with that we

NOTE Confidence: 0.8683241

 $01:01:41.016 \rightarrow 01:01:43.118$ have about 25 minutes for questions,

NOTE Confidence: 0.8683241

01:01:43.120 --> 01:01:45.145 so hopefully I've given you

NOTE Confidence: 0.8683241

 $01:01:45.145 \longrightarrow 01:01:47.170$ enough things to think about.

NOTE Confidence: 0.8683241

01:01:47.170 --> 01:01:48.930 Emma, stop sharing my screen.

NOTE Confidence: 0.83240086

 $01:01:50.960 \longrightarrow 01:01:52.406$ Given its last to think about,

NOTE Confidence: 0.83240086

 $01:01:52.410 \longrightarrow 01:01:55.560$ then thank you very much.

NOTE Confidence: 0.83240086

 $01:01:55.560 \longrightarrow 01:01:56.715$ Now we'll just take a

NOTE Confidence: 0.83240086

 $01:01:56.715 \longrightarrow 01:01:57.870$ minute to get your breath.

NOTE Confidence: 0.83240086

01:01:57.870 --> 01:01:59.622 Take a sip of water an I'm gonna

NOTE Confidence: 0.83240086

 $01{:}01{:}59.622 \dashrightarrow 01{:}02{:}01.128$ invite you folks please to send

NOTE Confidence: 0.83240086

 $01:02:01.128 \longrightarrow 01:02:02.652$ your questions in via the Q&A

- NOTE Confidence: 0.83240086
- $01:02:02.710 \longrightarrow 01:02:04.334$ and then I will read him to bed.
- NOTE Confidence: 0.82474625
- $01{:}02{:}09.860 \dashrightarrow 01{:}02{:}12.240$ So then the first question that one
- NOTE Confidence: 0.82474625
- $01:02:12.240 \longrightarrow 01:02:14.727$ of the things that occurred to me,
- NOTE Confidence: 0.82474625
- $01:02:14.730 \longrightarrow 01:02:17.170$ kind of in broad strokes, was the.
- NOTE Confidence: 0.9138662
- $01:02:19.200 \longrightarrow 01:02:21.868$ Is the question about.
- NOTE Confidence: 0.9138662
- $01:02:21.870 \longrightarrow 01:02:22.356$ About paternalism,
- NOTE Confidence: 0.9138662
- $01:02:22.356 \rightarrow 01:02:24.057$ you know you talked about clinical utility.
- NOTE Confidence: 0.9138662
- $01:02:24.060 \longrightarrow 01:02:25.986$ Of course, that for those of us in the
- NOTE Confidence: 0.9138662
- $01:02:25.986 \rightarrow 01:02:27.946$ room who are who are mainly clinicians,
- NOTE Confidence: 0.9138662
- $01:02:27.950 \longrightarrow 01:02:28.922$ this really hits home.
- NOTE Confidence: 0.9138662
- 01:02:28.922 --> 01:02:30.380 You're going to do this test.
- NOTE Confidence: 0.9138662
- $01:02:30.380 \longrightarrow 01:02:31.832$ What are you going to do
- NOTE Confidence: 0.9138662
- $01:02:31.832 \longrightarrow 01:02:32.558$ with the information?
- NOTE Confidence: 0.9138662
- $01{:}02{:}32{.}560 \dashrightarrow 01{:}02{:}34{.}261$ And if parents were to say or
- NOTE Confidence: 0.9138662
- 01:02:34.261 --> 01:02:35.440 a physician says, you know,
- NOTE Confidence: 0.9138662

 $01:02:35.440 \longrightarrow 01:02:37.015$ I just want to know that we

NOTE Confidence: 0.9138662

01:02:37.015 --> 01:02:38.390 greatly in 'cause I'm curious,

NOTE Confidence: 0.9138662

 $01{:}02{:}38{.}390 \dashrightarrow 01{:}02{:}40{.}098$ we greatly discourage him from doing it.

NOTE Confidence: 0.9138662

 $01{:}02{:}40{.}100 \dashrightarrow 01{:}02{:}42{.}179$ Seems like a poor use of resources and some

NOTE Confidence: 0.9138662

 $01{:}02{:}42.179 \dashrightarrow 01{:}02{:}44.469$ of these tests carry risks for the kid too.

NOTE Confidence: 0.9138662

 $01:02:44.470 \rightarrow 01:02:45.826$ But now with the genetic testing

NOTE Confidence: 0.9138662

 $01:02:45.826 \longrightarrow 01:02:47.140$ and of course the parents.

NOTE Confidence: 0.9138662

01:02:47.140 --> 01:02:49.212 So if a parent said, you know what,

NOTE Confidence: 0.9138662

01:02:49.212 --> 01:02:51.200 I'd really like you to do an

NOTE Confidence: 0.9138662

 $01{:}02{:}51{.}267 \dashrightarrow 01{:}02{:}52{.}387$ X Ray of my kids.

NOTE Confidence: 0.9138662

 $01{:}02{:}52{.}390 \dashrightarrow 01{:}02{:}53{.}131$ Kids need 'cause.

NOTE Confidence: 0.9138662

 $01:02:53.131 \rightarrow 01:02:54.860$ I'm curious to see what he looks

NOTE Confidence: 0.9138662

 $01:02:54.909 \rightarrow 01:02:56.439$ like when we're not doing that.

NOTE Confidence: 0.9138662

 $01:02:56.440 \rightarrow 01:02:57.700$ If there's not clinically indicated,

NOTE Confidence: 0.9138662

 $01:02:57.700 \longrightarrow 01:02:59.218$ at least we should, right so,

NOTE Confidence: 0.9138662

 $01:02:59.220 \longrightarrow 01:03:00.606$ so we limit what's available to

- NOTE Confidence: 0.9138662
- $01:03:00.606 \rightarrow 01:03:02.409$ them and with some of the sequencing

 $01:03:02.409 \longrightarrow 01:03:03.517$ that you've referred to.

NOTE Confidence: 0.9138662

 $01{:}03{:}03{.}520 \dashrightarrow 01{:}03{:}05{.}158$ But an awful lot of this information

NOTE Confidence: 0.9138662

 $01:03:05.158 \rightarrow 01:03:07.060$ that is kind of optional for parents,

NOTE Confidence: 0.9138662

 $01{:}03{:}07{.}060 \dashrightarrow 01{:}03{:}07{.}566$ this is.

NOTE Confidence: 0.9138662

01:03:07.566 --> 01:03:08.072 There's no.

NOTE Confidence: 0.9138662

 $01:03:08.072 \rightarrow 01:03:09.337$ Not necessarily an increased expense,

NOTE Confidence: 0.9138662

 $01{:}03{:}09{.}340 \dashrightarrow 01{:}03{:}10{.}792$ and certainly not an increase risk

NOTE Confidence: 0.9138662

 $01:03:10.792 \longrightarrow 01:03:12.851$ of doing the test that the risk of

NOTE Confidence: 0.9138662

 $01:03:12.851 \rightarrow 01:03:14.146$ the information is certainly one.

NOTE Confidence: 0.9138662

 $01{:}03{:}14.150 \dashrightarrow 01{:}03{:}16.190$ But I guess what I'm trying to get at is

NOTE Confidence: 0.9138662

 $01{:}03{:}16.249 \dashrightarrow 01{:}03{:}18.447$ if parents say 'cause you're talking about.

NOTE Confidence: 0.9138662

 $01{:}03{:}18.450 \dashrightarrow 01{:}03{:}20.214$ Some people just think more is better.

NOTE Confidence: 0.9138662

 $01:03:20.220 \longrightarrow 01:03:21.236$ So if parents say,

NOTE Confidence: 0.9138662

01:03:21.236 --> 01:03:22.026 yeah, you know,

- $01:03:22.026 \longrightarrow 01:03:23.634$ I'd like to know in particular.
- NOTE Confidence: 0.9138662
- $01{:}03{:}23.640 \dashrightarrow 01{:}03{:}27.114$ If some body in the lab knows I wanna know.
- NOTE Confidence: 0.9138662
- 01:03:27.120 --> 01:03:28.530 Then is it overly paternalistic
- NOTE Confidence: 0.9138662
- $01:03:28.530 \longrightarrow 01:03:29.940$ of us to say now?
- NOTE Confidence: 0.9138662
- $01{:}03{:}29{.}940 \dashrightarrow 01{:}03{:}32{.}760$ I know it's really not good for you to know.
- NOTE Confidence: 0.8251437
- $01{:}03{:}34{.}190 \dashrightarrow 01{:}03{:}35{.}690$ So that's a great question.
- NOTE Confidence: 0.8251437
- 01:03:35.690 --> 01:03:37.120 Actually, it means that I
- NOTE Confidence: 0.8251437
- $01:03:37.120 \longrightarrow 01:03:39.269$ have to go back to the slide,
- NOTE Confidence: 0.8251437
- $01{:}03{:}39{.}270 \dashrightarrow 01{:}03{:}40{.}770$ skipped over towards the end,
- NOTE Confidence: 0.8251437
- $01:03:40.770 \rightarrow 01:03:42.444$ because that's actually the slides I
- NOTE Confidence: 0.8251437
- $01{:}03{:}42{.}444 \dashrightarrow 01{:}03{:}44{.}660$ slipped over that I skipped over briefly.
- NOTE Confidence: 0.8251437
- 01:03:44.660 --> 01:03:45.852 You know, I think.
- NOTE Confidence: 0.8251437
- $01{:}03{:}45{.}852 \dashrightarrow 01{:}03{:}47{.}938$ Well, I think there's been a shift.
- NOTE Confidence: 0.8251437
- $01:03:47.940 \longrightarrow 01:03:50.929$ So in other words, I think that.
- NOTE Confidence: 0.8251437
- $01:03:50.930 \rightarrow 01:03:53.546$ There has been this shift in several ways.
- NOTE Confidence: 0.8251437
- $01:03:53.550 \dashrightarrow 01:03:56.097$ One is, I do think just like the example

- NOTE Confidence: 0.8251437
- $01:03:56.097 \rightarrow 01:03:58.777$ they gave you from zero during pigmentosa,
- NOTE Confidence: 0.8251437
- $01{:}03{:}58.780 \dashrightarrow 01{:}04{:}00.742$ even though from the clinician from
- NOTE Confidence: 0.8251437
- 01:04:00.742 --> 01:04:02.380 the researchers point of view,
- NOTE Confidence: 0.8251437
- $01:04:02.380 \longrightarrow 01:04:04.662$ we don't know what to do with
- NOTE Confidence: 0.8251437
- $01:04:04.662 \longrightarrow 01:04:05.314$ this information.
- NOTE Confidence: 0.8251437
- $01:04:05.320 \longrightarrow 01:04:07.350$ These families who are part
- NOTE Confidence: 0.8251437
- $01:04:07.350 \longrightarrow 01:04:09.380$ of a close knit group.
- NOTE Confidence: 0.8251437
- $01{:}04{:}09{.}380 \dashrightarrow 01{:}04{:}10.755$ Felt this information was important
- NOTE Confidence: 0.8251437
- $01{:}04{:}10.755 \dashrightarrow 01{:}04{:}12.130$ and they knew its limitations.
- NOTE Confidence: 0.8251437
- $01:04:12.130 \longrightarrow 01:04:14.330$ They knew that it was uncertain what it
- NOTE Confidence: 0.8251437
- $01:04:14.330 \longrightarrow 01:04:16.528$ meant, but they knew other folks had it.
- NOTE Confidence: 0.8251437
- $01:04:16.530 \longrightarrow 01:04:17.630$ They wanted it to,
- NOTE Confidence: 0.8251437
- $01:04:17.630 \longrightarrow 01:04:18.455$ and in fact,
- NOTE Confidence: 0.8251437
- 01:04:18.460 --> 01:04:19.735 part of our conversation with
- NOTE Confidence: 0.8251437
- $01:04:19.735 \longrightarrow 01:04:21.329$ that group resulted in them over
- NOTE Confidence: 0.8251437

 $01:04:21.329 \rightarrow 01:04:22.919$ the next number of years revising

NOTE Confidence: 0.8251437

 $01{:}04{:}22{.}919 \dashrightarrow 01{:}04{:}24{.}072$ their approaches and getting

NOTE Confidence: 0.8251437

 $01:04:24.072 \rightarrow 01:04:25.328$ input from their community.

NOTE Confidence: 0.8251437

 $01:04:25.330 \longrightarrow 01:04:25.884$ And actually,

NOTE Confidence: 0.8251437

 $01:04:25.884 \longrightarrow 01:04:27.546$ they made the decision to make

NOTE Confidence: 0.8251437

 $01{:}04{:}27.546 \dashrightarrow 01{:}04{:}28.737$ that information available with

NOTE Confidence: 0.8251437

 $01{:}04{:}28.737 \dashrightarrow 01{:}04{:}30.192$ the limitations and with the

NOTE Confidence: 0.8251437

 $01:04:30.192 \longrightarrow 01:04:31.594$ explanations of what this may

NOTE Confidence: 0.8251437

 $01{:}04{:}31{.}594 \dashrightarrow 01{:}04{:}33{.}028$ mean with the limitations are so.

NOTE Confidence: 0.8251437

 $01:04:33.030 \rightarrow 01:04:34.126$ That's an example where,

NOTE Confidence: 0.8251437

 $01{:}04{:}34{.}126 \dashrightarrow 01{:}04{:}36{.}878$ but I think the one of the issues I want.

NOTE Confidence: 0.8251437

 $01:04:36.880 \longrightarrow 01:04:38.614$ That's a very specific group of

NOTE Confidence: 0.8251437

 $01{:}04{:}38.614 \dashrightarrow 01{:}04{:}40.849$ people who are high risk of a disease.

NOTE Confidence: 0.8251437

 $01{:}04{:}40.850 \dashrightarrow 01{:}04{:}43.550$ Where it was meaningful to them to do that,

NOTE Confidence: 0.8251437

 $01:04:43.550 \rightarrow 01:04:45.384$ I think other questions come up when

NOTE Confidence: 0.8251437

 $01:04:45.384 \rightarrow 01:04:47.373$ we get the thing with the context

- NOTE Confidence: 0.8251437
- $01:04:47.373 \rightarrow 01:04:49.481$ of when is people are people asking

 $01{:}04{:}49{.}481 \dashrightarrow 01{:}04{:}51{.}868$ for this information and I want to

NOTE Confidence: 0.8251437

 $01:04:51.868 \rightarrow 01:04:53.094$ distinguish between the random.

NOTE Confidence: 0.8251437

 $01:04:53.094 \rightarrow 01:04:55.270$ And So what I want to clarify is

NOTE Confidence: 0.8251437

 $01:04:55.339 \rightarrow 01:04:57.563$ that right now we're not at a place,

NOTE Confidence: 0.8251437

01:04:57.570 --> 01:04:59.613 nor do I think we are going to place,

NOTE Confidence: 0.8251437

01:04:59.620 --> 01:05:00.106 like Gattaca,

NOTE Confidence: 0.8251437

01:05:00.106 --> 01:05:01.321 where everyone we tested for

NOTE Confidence: 0.8251437

 $01:05:01.321 \rightarrow 01:05:02.502$ everything for everything that we've

NOTE Confidence: 0.8251437

 $01{:}05{:}02{.}502 \dashrightarrow 01{:}05{:}03{.}522$ done for reasons for particular

NOTE Confidence: 0.8251437

 $01:05:03.522 \rightarrow 01:05:04.640$ reasons may be good reasons,

NOTE Confidence: 0.8251437

 $01{:}05{:}04.640 \dashrightarrow 01{:}05{:}05.321$ maybe bad reasons,

NOTE Confidence: 0.8251437

 $01{:}05{:}05{.}321 \dashrightarrow 01{:}05{:}06{.}683$ but there are currently based reasons.

NOTE Confidence: 0.8251437

 $01{:}05{:}06.690 \dashrightarrow 01{:}05{:}07.890$ But the question is,

NOTE Confidence: 0.8251437

 $01:05:07.890 \rightarrow 01:05:09.390$ what should the scope be?

 $01:05:09.390 \longrightarrow 01:05:10.815$ Once testing is done because

NOTE Confidence: 0.8251437

01:05:10.815 --> 01:05:12.240 with sequencing we have the

NOTE Confidence: 0.8251437

 $01:05:12.295 \rightarrow 01:05:14.130$ opportunity to add additional tests,

NOTE Confidence: 0.8251437

 $01:05:14.130 \longrightarrow 01:05:15.390$ something you know these

NOTE Confidence: 0.8251437

01:05:15.390 --> 01:05:16.335 actionable tests are.

NOTE Confidence: 0.8251437

 $01{:}05{:}16.340 \dashrightarrow 01{:}05{:}18.206$ These actionable tests include tests that

NOTE Confidence: 0.8251437

 $01:05:18.206 \rightarrow 01:05:20.130$ ensure the only develop in adulthood,

NOTE Confidence: 0.8251437

 $01:05:20.130 \longrightarrow 01:05:21.394$ and that's actually the

NOTE Confidence: 0.8251437

 $01{:}05{:}21{.}394 \dashrightarrow 01{:}05{:}22{.}658$ question you're getting at.

NOTE Confidence: 0.8251437

 $01{:}05{:}22.660 \dashrightarrow 01{:}05{:}26.548$ Mark and I want to share my screen again.

NOTE Confidence: 0.8251437

 $01{:}05{:}26.550 \dashrightarrow 01{:}05{:}27.270$ And.

NOTE Confidence: 0.897526

 $01:05:29.400 \longrightarrow 01:05:32.184$ See if I can get to this here.

NOTE Confidence: 0.897526

 $01:05:32.190 \rightarrow 01:05:35.590$ I want to go back to one slide but when I.

NOTE Confidence: 0.897526

 $01:05:35.590 \longrightarrow 01:05:38.298$ Skipped over right here.

NOTE Confidence: 0.897526

 $01:05:38.300 \longrightarrow 01:05:41.036$ So one of the things that.

NOTE Confidence: 0.897526

 $01:05:41.040 \longrightarrow 01:05:42.408$ That are the consortium

- NOTE Confidence: 0.897526
- $01:05:42.408 \rightarrow 01:05:44.118$ is doing this time around,
- NOTE Confidence: 0.897526
- $01:05:44.120 \rightarrow 01:05:46.166$ just like show the consent paper.
- NOTE Confidence: 0.897526
- $01:05:46.170 \longrightarrow 01:05:47.398$ This is a project.
- NOTE Confidence: 0.897526
- $01:05:47.398 \longrightarrow 01:05:50.077$ This is the paper that is being led
- NOTE Confidence: 0.897526
- 01:05:50.077 --> 01:05:52.375 by Amy McGuire and Stephanie Marine
- NOTE Confidence: 0.897526
- $01{:}05{:}52{.}375 \dashrightarrow 01{:}05{:}55{.}145$ at Baylor and a bunch of other Anna
- NOTE Confidence: 0.897526
- $01{:}05{:}55{.}145 \dashrightarrow 01{:}05{:}57{.}696$ Bunch of other folks in which there
- NOTE Confidence: 0.897526
- $01{:}05{:}57{.}696 \dashrightarrow 01{:}05{:}59{.}806$ we are trying to collectively.
- NOTE Confidence: 0.897526
- $01{:}05{:}59{.}810 \dashrightarrow 01{:}06{:}01{.}760$ Articulate the range of perceived
- NOTE Confidence: 0.897526
- 01:06:01.760 --> 01:06:04.451 utility from the point of view of
- NOTE Confidence: 0.897526
- $01{:}06{:}04{.}451 \dashrightarrow 01{:}06{:}06{.}473$ patients for genetics and the point
- NOTE Confidence: 0.897526
- $01{:}06{:}06{.}473 \dashrightarrow 01{:}06{:}08{.}700$ of this model is to suggest that
- NOTE Confidence: 0.897526
- $01:06:08.700 \longrightarrow 01:06:10.536$ there are a range of utilities
- NOTE Confidence: 0.897526
- $01{:}06{:}10.536 \dashrightarrow 01{:}06{:}12.266$ and that we can distinguish,
- NOTE Confidence: 0.897526
- $01{:}06{:}12.270 \dashrightarrow 01{:}06{:}14.853$ and we spent a lot of time just like
- NOTE Confidence: 0.897526

 $01:06:14.853 \rightarrow 01:06:17.148$ the issue of categories that describe

NOTE Confidence: 0.897526

 $01{:}06{:}17.148 \dashrightarrow 01{:}06{:}19.880$ for the for the next Gen paper,

NOTE Confidence: 0.897526

 $01{:}06{:}19.880 \dashrightarrow 01{:}06{:}21.610$ the categories of diseases here,

NOTE Confidence: 0.897526

01:06:21.610 --> 01:06:23.158 which categories of utility,

NOTE Confidence: 0.897526

 $01{:}06{:}23.158 \dashrightarrow 01{:}06{:}25.480$ and we did interviews with participants

NOTE Confidence: 0.897526

 $01{:}06{:}25{.}541 \dashrightarrow 01{:}06{:}27{.}761$ in the various Caesar studies about

NOTE Confidence: 0.897526

 $01:06:27.761 \longrightarrow 01:06:29.599$ their experience with genetic testing

NOTE Confidence: 0.897526

 $01{:}06{:}29{.}599 \dashrightarrow 01{:}06{:}31{.}975$ in order to come up with these domains.

NOTE Confidence: 0.897526

 $01{:}06{:}31{.}980 \dashrightarrow 01{:}06{:}33{.}312$ Of perceived utility.

NOTE Confidence: 0.897526

 $01{:}06{:}33{.}312 \dashrightarrow 01{:}06{:}35{.}976$ One of those domains is clinical.

NOTE Confidence: 0.897526

 $01:06:35.980 \rightarrow 01:06:38.092$ The way in which the information will you NOTE Confidence: 0.897526

 $01:06:38.092 \rightarrow 01:06:39.969$ know either established new diagnosis,

NOTE Confidence: 0.897526

 $01{:}06{:}39{.}970 \dashrightarrow 01{:}06{:}41{.}460$ informed clinical management may result

NOTE Confidence: 0.897526

 $01:06:41.460 \rightarrow 01:06:43.959$ in referral to a trial may allow screening.

NOTE Confidence: 0.897526

 $01:06:43.960 \longrightarrow 01:06:45.188$ These are clinical outcomes

NOTE Confidence: 0.897526

01:06:45.188 --> 01:06:46.109 and clinical values,

- NOTE Confidence: 0.897526
- $01:06:46.110 \longrightarrow 01:06:47.820$ but we also learned was that

 $01:06:47.820 \longrightarrow 01:06:49.800$ there were range of other values,

NOTE Confidence: 0.897526

 $01:06:49.800 \longrightarrow 01:06:51.636$ and some of these are positive.

NOTE Confidence: 0.897526

 $01:06:51.640 \longrightarrow 01:06:53.170$ Some of these are negative.

NOTE Confidence: 0.897526

 $01{:}06{:}53{.}170 \dashrightarrow 01{:}06{:}55{.}933$ We look at the emotional one at the bottom.

NOTE Confidence: 0.897526

01:06:55.940 --> 01:06:57.390 You can identify the potential

NOTE Confidence: 0.897526

 $01{:}06{:}57{.}390 \dashrightarrow 01{:}06{:}59{.}240$ for adverse feelings as well as

NOTE Confidence: 0.897526

01:06:59.240 --> 01:06:59.928 positive feelings,

NOTE Confidence: 0.897526

 $01{:}06{:}59{.}930 \dashrightarrow 01{:}07{:}02{.}354$ and I think the point here is simply

NOTE Confidence: 0.897526

 $01{:}07{:}02.354 \dashrightarrow 01{:}07{:}04.623$ acknowledge that one of the impacts of

NOTE Confidence: 0.897526

 $01{:}07{:}04.623 \dashrightarrow 01{:}07{:}06.460$ testing with positive thinking can be.

NOTE Confidence: 0.897526

 $01{:}07{:}06.460 \dashrightarrow 01{:}07{:}09.076$ An emotional impact and.

NOTE Confidence: 0.897526

 $01:07:09.076 \longrightarrow 01:07:09.730$ Additionally,

NOTE Confidence: 0.897526

 $01{:}07{:}09.730 \dashrightarrow 01{:}07{:}12.140$ there can be behavioral impacts, and again,

NOTE Confidence: 0.897526

 $01{:}07{:}12{.}140 \dashrightarrow 01{:}07{:}14{.}210$ here we're talking about behavioral impacts.

 $01:07:14.210 \rightarrow 01:07:16.268$ They're not the same as clinical,

NOTE Confidence: 0.897526

 $01:07:16.270 \longrightarrow 01:07:17.514$ but might have meaning,

NOTE Confidence: 0.897526

 $01{:}07{:}17.514 \dashrightarrow 01{:}07{:}19.380$ and here we include things like

NOTE Confidence: 0.897526

 $01:07:19.439 \longrightarrow 01:07:20.738$ changing health habits,

NOTE Confidence: 0.897526

 $01:07:20.740 \longrightarrow 01:07:21.428$ future planning,

NOTE Confidence: 0.897526

 $01{:}07{:}21.428 \dashrightarrow 01{:}07{:}23.836$ whether it's for financial or career choices,

NOTE Confidence: 0.897526

01:07:23.840 --> 01:07:24.869 parental decision making,

NOTE Confidence: 0.897526

01:07:24.869 --> 01:07:25.898 reproductive decision making,

NOTE Confidence: 0.897526

01:07:25.900 --> 01:07:26.590 influence coverage.

NOTE Confidence: 0.897526

 $01:07:26.590 \longrightarrow 01:07:28.315$ These are things that people

NOTE Confidence: 0.897526

 $01{:}07{:}28.315 \dashrightarrow 01{:}07{:}30.412$ might do with the information on

NOTE Confidence: 0.897526

 $01:07:30.412 \longrightarrow 01:07:31.748$ the issue of curiosity.

NOTE Confidence: 0.897526

 $01{:}07{:}31.750 \dashrightarrow 01{:}07{:}34.494$ I think we would describe as cognitive is.

NOTE Confidence: 0.897526

 $01:07:34.500 \longrightarrow 01:07:35.529$ In other words,

NOTE Confidence: 0.897526

 $01{:}07{:}35{.}529 \dashrightarrow 01{:}07{:}37{.}244$ it might impact their understanding.

NOTE Confidence: 0.897526

 $01:07:37.250 \rightarrow 01:07:40.616$ It might be satisfying their curiosity.

- NOTE Confidence: 0.897526
- $01:07:40.620 \rightarrow 01:07:43.140$ And it may be relevant to the how they view

 $01{:}07{:}43.199 \dashrightarrow 01{:}07{:}45.659$ the information related to overall health.

NOTE Confidence: 0.897526

01:07:45.660 --> 01:07:47.250 And finally, there's a social

NOTE Confidence: 0.897526

 $01:07:47.250 \longrightarrow 01:07:49.130$ impact of testing that may.

NOTE Confidence: 0.897526

 $01{:}07{:}49{.}130 \dashrightarrow 01{:}07{:}50{.}120$ So, for example,

NOTE Confidence: 0.897526

 $01{:}07{:}50{.}120 \dashrightarrow 01{:}07{:}52{.}430$ one of the things that we learned

NOTE Confidence: 0.897526

 $01:07:52.499 \longrightarrow 01:07:54.480$ and it not only in this state,

NOTE Confidence: 0.897526

 $01:07:54.480 \longrightarrow 01:07:55.383$ but previous studies,

NOTE Confidence: 0.897526

 $01{:}07{:}55{.}383 \dashrightarrow 01{:}07{:}57{.}490$ is that often having a genic diagnosis

NOTE Confidence: 0.897526

01:07:57.549 --> 01:07:59.209 ocular for children for children,

NOTE Confidence: 0.897526

 $01:07:59.210 \longrightarrow 01:08:00.402$ parents with rare diseases,

NOTE Confidence: 0.897526

 $01{:}08{:}00{.}402 \dashrightarrow 01{:}08{:}02{.}591$ is a way of being part of

NOTE Confidence: 0.897526

01:08:02.591 --> 01:08:03.927 a social support network.

NOTE Confidence: 0.897526

 $01{:}08{:}03{.}930 \dashrightarrow 01{:}08{:}06{.}450$ Its way of having access to social services.

NOTE Confidence: 0.897526

 $01:08:06.450 \longrightarrow 01:08:08.382$ When I was at the NIH working

 $01:08:08.382 \rightarrow 01:08:09.920$ in the Genome Institute,

NOTE Confidence: 0.897526

 $01:08:09.920 \longrightarrow 01:08:11.196$ they would see patients.

NOTE Confidence: 0.897526

 $01{:}08{:}11{.}196 \dashrightarrow 01{:}08{:}13{.}110$ With rare diseases and it turned

NOTE Confidence: 0.897526

 $01:08:13.166 \longrightarrow 01:08:14.840$ out one of the most stressful

NOTE Confidence: 0.897526

 $01{:}08{:}14.840 \dashrightarrow 01{:}08{:}16.602$ things when a family would come

NOTE Confidence: 0.897526

 $01:08:16.602 \longrightarrow 01:08:18.348$ in with a perceived rare disease

NOTE Confidence: 0.897526

 $01:08:18.348 \rightarrow 01:08:20.090$ in which the Jess would say,

NOTE Confidence: 0.897526

 $01:08:20.090 \rightarrow 01:08:20.380$ hey,

NOTE Confidence: 0.862932

 $01{:}08{:}20{.}380 \dashrightarrow 01{:}08{:}22{.}410$ we've got some good news for you.

NOTE Confidence: 0.862932

01:08:22.410 --> 01:08:24.120 You don't have this terrible disease

NOTE Confidence: 0.862932

 $01{:}08{:}24{.}120 \dashrightarrow 01{:}08{:}26{.}178$ you thought you had is actually this,

NOTE Confidence: 0.862932

 $01:08:26.180 \longrightarrow 01:08:28.210$ or we don't know what it is,

NOTE Confidence: 0.862932

 $01{:}08{:}28{.}210 \dashrightarrow 01{:}08{:}30{.}303$ but it's not that and people were

NOTE Confidence: 0.862932

 $01{:}08{:}30{.}303 \dashrightarrow 01{:}08{:}32{.}084$ often unhappy to be told they

NOTE Confidence: 0.862932

 $01:08:32.084 \rightarrow 01:08:34.009$ were no longer part of the group,

NOTE Confidence: 0.862932

 $01:08:34.010 \longrightarrow 01:08:36.188$ and so the point here Mark

- NOTE Confidence: 0.862932
- $01:08:36.188 \longrightarrow 01:08:38.920$ is that is to suggest that.

 $01{:}08{:}38{.}920 \dashrightarrow 01{:}08{:}41{.}216$ There is a range of value of this.

NOTE Confidence: 0.862932

 $01:08:41.220 \longrightarrow 01:08:43.074$ One of the other things that

NOTE Confidence: 0.862932

 $01:08:43.074 \rightarrow 01:08:44.758$ the Caesar consortium is doing

NOTE Confidence: 0.862932

 $01:08:44.758 \longrightarrow 01:08:46.298$ is interacting with payers.

NOTE Confidence: 0.862932

 $01{:}08{:}46{.}300 \dashrightarrow 01{:}08{:}48{.}005$ People who provide the coverage

NOTE Confidence: 0.862932

 $01{:}08{:}48.005 \dashrightarrow 01{:}08{:}50.580$ for testing and the try to at least

NOTE Confidence: 0.862932

 $01:08:50.580 \longrightarrow 01:08:52.854$ have on the agenda that some of the

NOTE Confidence: 0.862932

 $01{:}08{:}52{.}854 \dashrightarrow 01{:}08{:}54{.}559$ value of testing for individuals

NOTE Confidence: 0.862932

 $01:08:54.559 \rightarrow 01:08:56.251$ is not simply only clinical,

NOTE Confidence: 0.862932

 $01{:}08{:}56{.}251 \dashrightarrow 01{:}08{:}58{.}177$ but it's these other ranges too.

NOTE Confidence: 0.862932

 $01{:}08{:}58{.}180 \dashrightarrow 01{:}09{:}00{.}100$ And the question is, you know,

NOTE Confidence: 0.862932

 $01{:}09{:}00{.}100 \dashrightarrow 01{:}09{:}02{.}252$ where do we fit this in two questions

NOTE Confidence: 0.862932

 $01:09:02.252 \rightarrow 01:09:03.630$ about resource prioritization?

NOTE Confidence: 0.862932

 $01{:}09{:}03.630 \dashrightarrow 01{:}09{:}05.240$ And I don't want to.

 $01:09:05.240 \longrightarrow 01:09:07.166$ I can't answer that for sure,

NOTE Confidence: 0.862932

 $01{:}09{:}07{.}170 \dashrightarrow 01{:}09{:}10{.}095$ but I think the idea is to at least

NOTE Confidence: 0.862932

 $01:09:10.095 \rightarrow 01:09:12.615$ suggest it ought to be on the table,

NOTE Confidence: 0.862932

 $01:09:12.620 \longrightarrow 01:09:14.685$ and I wouldn't suggest that

NOTE Confidence: 0.862932

 $01:09:14.685 \rightarrow 01:09:16.337$ testing simply for that.

NOTE Confidence: 0.862932

 $01:09:16.340 \longrightarrow 01:09:17.804$ That cognitive utilities have

NOTE Confidence: 0.862932

01:09:17.804 --> 01:09:19.634 no meaning for an individual,

NOTE Confidence: 0.862932

 $01:09:19.640 \longrightarrow 01:09:22.576$ and I think the best example of that,

NOTE Confidence: 0.862932

 $01{:}09{:}22.580 \dashrightarrow 01{:}09{:}25.966$ frankly, is. Yeah, I do have a diagnosis.

NOTE Confidence: 0.862932

 $01:09:25.970 \longrightarrow 01:09:26.672$ Is that really?

NOTE Confidence: 0.862932

 $01{:}09{:}26.672 \dashrightarrow 01{:}09{:}28.076$ If it's a diagnosis that we

NOTE Confidence: 0.862932

01:09:28.076 --> 01:09:29.179 can't do anything back,

NOTE Confidence: 0.862932

 $01:09:29.180 \longrightarrow 01:09:30.960$ we can simply say you've

NOTE Confidence: 0.862932

 $01{:}09{:}30{.}960 \dashrightarrow 01{:}09{:}32{.}384$ got a rare condition.

NOTE Confidence: 0.862932

 $01:09:32.390 \longrightarrow 01:09:35.060$ Would it be appropriate for a

NOTE Confidence: 0.862932

01:09:35.060 - 01:09:36.395 insurance insurance company?

- NOTE Confidence: 0.862932
- $01:09:36.400 \longrightarrow 01:09:37.729$ Say we don't?
- NOTE Confidence: 0.7978275
- $01:09:39.780 \longrightarrow 01:09:41.100$ We shouldn't pay for that.
- NOTE Confidence: 0.7978275
- $01:09:41.100 \longrightarrow 01:09:42.195$ Do anything with.
- NOTE Confidence: 0.7978275
- $01:09:42.195 \longrightarrow 01:09:43.655$ We usually will say.
- NOTE Confidence: 0.7978275
- $01{:}09{:}43.660 \dashrightarrow 01{:}09{:}45.196$ You know that that may be
- NOTE Confidence: 0.7978275
- $01:09:45.196 \longrightarrow 01:09:46.220$ valuable to the person,
- NOTE Confidence: 0.7978275
- $01:09:46.220 \longrightarrow 01:09:47.500$ and therefore it's worth doing.
- NOTE Confidence: 0.7978275
- $01:09:47.500 \longrightarrow 01:09:48.776$ So I think the biggest,
- NOTE Confidence: 0.7978275
- 01:09:48.776 --> 01:09:49.544 biggest reasons for
- NOTE Confidence: 0.7978275
- $01:09:49.544 \longrightarrow 01:09:50.320$ clinical sequencing, but
- NOTE Confidence: 0.81839126
- $01:09:50.320 \rightarrow 01:09:51.565$ maybe not sometimes these tests
- NOTE Confidence: 0.81839126
- $01:09:51.565 \rightarrow 01:09:53.122$ are not to satisfy the curiosity
- NOTE Confidence: 0.81839126
- $01:09:53.122 \longrightarrow 01:09:54.664$ of the patient or the parent,
- NOTE Confidence: 0.81839126
- 01:09:54.670 --> 01:09:56.332 but the curiosity of the physician
- NOTE Confidence: 0.81839126
- $01{:}09{:}56{.}332 \dashrightarrow 01{:}09{:}57{.}945$ where this isn't actually going to
- NOTE Confidence: 0.81839126

- $01:09:57.945 \longrightarrow 01:09:59.527$ help us manage this patient at all,
- NOTE Confidence: 0.81839126
- $01:09:59.530 \longrightarrow 01:10:00.343$ but it'll it'll.
- NOTE Confidence: 0.81839126
- $01:10:00.343 \rightarrow 01:10:01.969$ Best Buy sometimes our curiosity and
- NOTE Confidence: 0.81839126
- $01:10:01.969 \longrightarrow 01:10:03.688$ that that's a whole separate issue,
- NOTE Confidence: 0.81839126
- $01{:}10{:}03.690 \dashrightarrow 01{:}10{:}05.786$ and as far as the the the the,
- NOTE Confidence: 0.81839126
- $01{:}10{:}05{.}790 \dashrightarrow 01{:}10{:}07{.}624$ the emotional outcomes or benefits of this.
- NOTE Confidence: 0.81839126
- $01:10:07.630 \longrightarrow 01:10:09.438$ I mean, I'd say I remember that when
- NOTE Confidence: 0.81839126
- $01:10:09.438 \longrightarrow 01:10:11.415$ the first time with my second child
- NOTE Confidence: 0.81839126
- $01{:}10{:}11{.}415 \dashrightarrow 01{:}12{.}890$ was our first prenatal ultrasound,
- NOTE Confidence: 0.81839126
- $01{:}10{:}12.890 \dashrightarrow 01{:}10{:}14.498$ and when the afterwards they said NOTE Confidence: 0.81839126
- 01:10:14.498 --> 01:10:16.310 would you like to know the ***?
- NOTE Confidence: 0.81839126
- 01:10:16.310 --> 01:10:17.894 And I recall exactly I knew
- NOTE Confidence: 0.81839126
- 01:10:17.894 --> 01:10:19.521 exactly what I was thinking and
- NOTE Confidence: 0.81839126
- $01:10:19.521 \longrightarrow 01:10:21.306$ I said to my wife and said,
- NOTE Confidence: 0.81839126
- 01:10:21.310 --> 01:10:22.934 you know, I said I didn't really
- NOTE Confidence: 0.81839126
- 01:10:22.934 --> 01:10:24.459 want to know it necessarily,

- NOTE Confidence: 0.81839126
- $01:10:24.460 \longrightarrow 01:10:26.038$ but I'm looking at this tech
- NOTE Confidence: 0.81839126
- 01:10:26.038 --> 01:10:27.093 and I'm thinking, well,
- NOTE Confidence: 0.81839126
- 01:10:27.093 --> 01:10:29.197 if you know what I want to know,
- NOTE Confidence: 0.81839126
- $01:10:29.200 \longrightarrow 01:10:30.880$ if you know what I mean.
- NOTE Confidence: 0.81839126
- 01:10:30.880 --> 01:10:32.105 If some
body else is so
- NOTE Confidence: 0.81839126
- $01:10:32.105 \longrightarrow 01:10:33.085$ you about other people,
- NOTE Confidence: 0.81839126
- $01{:}10{:}33{.}090 \dashrightarrow 01{:}10{:}34{.}738$ but it could be the same with the
- NOTE Confidence: 0.81839126
- $01:10:34.738 \longrightarrow 01:10:36.192$ IT could potentially be the same
- NOTE Confidence: 0.81839126
- $01{:}10{:}36{.}192 \dashrightarrow 01{:}10{:}38{.}296$ with some people to say to their to
- NOTE Confidence: 0.81839126
- $01{:}10{:}38.296 \dashrightarrow 01{:}10{:}39.736$ the researcher or the clinician.
- NOTE Confidence: 0.81839126
- 01:10:39.740 --> 01:10:41.700 Well, if you're aware of something like this,
- NOTE Confidence: 0.81839126
- $01{:}10{:}41.700 \dashrightarrow 01{:}10{:}43.815$ I want to be aware of if nobody knows
- NOTE Confidence: 0.81839126
- $01:10:43.815 \rightarrow 01:10:46.125$ if it's, you know if it's in the dark.
- NOTE Confidence: 0.81839126
- $01{:}10{:}46.130 \dashrightarrow 01{:}10{:}47.360$ We've never looked at that.
- NOTE Confidence: 0.81839126
- $01:10:47.360 \longrightarrow 01:10:48.344$ We've never examined that.
- NOTE Confidence: 0.81839126

01:10:48.344 --> 01:10:49.328 That's a different feeling,

NOTE Confidence: 0.81839126

 $01:10:49.330 \longrightarrow 01:10:51.018$ potentially for the patient

NOTE Confidence: 0.81839126

 $01:10:51.018 \rightarrow 01:10:53.128$ or the research subject then.

NOTE Confidence: 0.81839126

 $01:10:53.130 \longrightarrow 01:10:55.629$ Then then you know somebody else knows

NOTE Confidence: 0.81839126

 $01:10:55.629 \rightarrow 01:10:58.030$ it and excuse me, sorry bout that.

NOTE Confidence: 0.8288017

 $01:11:03.570 \longrightarrow 01:11:04.810$ Sorry about what happened.

NOTE Confidence: 0.8288017

 $01:11:04.810 \longrightarrow 01:11:06.670$ I keep hitting this pilot is

NOTE Confidence: 0.8288017

01:11:06.670 --> 01:11:09.163 not working, I know but you know so let

NOTE Confidence: 0.8288017

 $01{:}11{:}09{.}163 \dashrightarrow 01{:}11{:}11{.}320$ me answer your second question first.

NOTE Confidence: 0.8288017

 $01:11:11.320 \longrightarrow 01:11:13.180$ I gotta get to the next

NOTE Confidence: 0.8288017

01:11:13.180 --> 01:11:14.516 question here Ben. OK,

NOTE Confidence: 0.8288017

 $01{:}11{:}14.516 \dashrightarrow 01{:}11{:}17.208$ we've got a couple of questions to get to.

NOTE Confidence: 0.8288017

 $01:11:17.210 \longrightarrow 01:11:18.800$ What have you learned from the

NOTE Confidence: 0.8288017

01:11:18.800 --> 01:11:20.518 studies like charm that would inform

NOTE Confidence: 0.8288017

 $01:11:20.518 \rightarrow 01:11:22.018$ the argument for doing genetic

NOTE Confidence: 0.8288017

 $01{:}11{:}22.018 \dashrightarrow 01{:}11{:}24.087$ screening as part of the newborn

- NOTE Confidence: 0.8288017
- 01:11:24.087 --> 01:11:25.575 congenital disease screening programs?

01:11:26.850 --> 01:11:28.005 Oh that's oh,

NOTE Confidence: 0.81407756

01:11:28.005 --> 01:11:30.315 thank you for asking that question.

NOTE Confidence: 0.81407756

01:11:30.320 --> 01:11:32.636 I'm, you know. First of all,

NOTE Confidence: 0.81407756

01:11:32.640 --> 01:11:34.968 I think there's different ways of

NOTE Confidence: 0.81407756

 $01{:}11{:}34{.}968 \dashrightarrow 01{:}11{:}36{.}938$ thinking about new born screening and

NOTE Confidence: 0.81407756

 $01:11:36.938 \rightarrow 01:11:39.086$ thinking about sequencing and the role

NOTE Confidence: 0.81407756

 $01{:}11{:}39.086 \dashrightarrow 01{:}11{:}41.519$ of sequencing with the new born screening.

NOTE Confidence: 0.81407756

01:11:41.520 --> 01:11:42.426 And So what?

NOTE Confidence: 0.81407756

 $01{:}11{:}42{.}426 \dashrightarrow 01{:}11{:}45{.}468$ I want to comment is that is that there

NOTE Confidence: 0.81407756

 $01{:}11{:}45{.}468 \dashrightarrow 01{:}11{:}48{.}457$ is sometimes sequencing done as part of

NOTE Confidence: 0.81407756

 $01{:}11{:}48{.}457 \dashrightarrow 01{:}11{:}51{.}170$ new born screening as a secondary test.

NOTE Confidence: 0.81407756

 $01:11:51.170 \longrightarrow 01:11:52.954$ So in other words,

NOTE Confidence: 0.81407756

 $01{:}11{:}52{.}954 \dashrightarrow 01{:}11{:}55{.}184$ by using the various biochemical

NOTE Confidence: 0.81407756

 $01:11:55.184 \rightarrow 01:11:58.215$ tests or even genetic tests that once.

- $01:11:58.220 \longrightarrow 01:12:01.380$ That some.
- NOTE Confidence: 0.81407756
- $01:12:01.380 \longrightarrow 01:12:03.100$ Currently in fact some some

 $01:12:03.100 \longrightarrow 01:12:04.476$ sequencing is being done.

NOTE Confidence: 0.81407756

01:12:04.480 --> 01:12:06.538 I will acknowledge I'm I think.

NOTE Confidence: 0.81407756

01:12:06.540 --> 01:12:07.536 I think we have.

NOTE Confidence: 0.81407756

 $01{:}12{:}07{.}536 \dashrightarrow 01{:}12{:}09{.}422$ This has to be done very thoughtfully

NOTE Confidence: 0.81407756

 $01:12:09.422 \rightarrow 01:12:12.046$ and I'm going to give you a very

NOTE Confidence: 0.81407756

 $01:12:12.046 \longrightarrow 01:12:13.957$ specific example related cystic

NOTE Confidence: 0.81407756

01:12:13.957 --> 01:12:15.475 fibrosis newborn screening,

NOTE Confidence: 0.81407756

01:12:15.480 --> 01:12:17.920 which is done in many places by a

NOTE Confidence: 0.81407756

01:12:17.920 --> 01:12:19.658 combination of biochemical as well

NOTE Confidence: 0.81407756

01:12:19.658 --> 01:12:21.433 as by either reflex sequencing

NOTE Confidence: 0.81407756

 $01{:}12{:}21{.}433 \dashrightarrow 01{:}12{:}23{.}739$ or in the state of California.

NOTE Confidence: 0.81407756

 $01:12:23.740 \longrightarrow 01:12:26.398$ Signal sequencing is done on every.

NOTE Confidence: 0.81407756

 $01{:}12{:}26{.}400 \dashrightarrow 01{:}12{:}29{.}627$ Patient who is a positive IRTA test.

NOTE Confidence: 0.81407756

01:12:29.630 --> 01:12:31.570 And what happens in California?

- NOTE Confidence: 0.81407756
- 01:12:31.570 --> 01:12:33.862 Because they do so much sequencing

 $01{:}12{:}33.862 \dashrightarrow 01{:}12{:}36.431$ is that they identify many more

NOTE Confidence: 0.81407756

 $01:12:36.431 \longrightarrow 01:12:38.371$ individuals who have variants

NOTE Confidence: 0.81407756

 $01:12:38.371 \longrightarrow 01:12:39.826$ of unknown significance.

NOTE Confidence: 0.81407756

 $01{:}12{:}39{.}830 \dashrightarrow 01{:}12{:}42.672$ And in cystic fibrosis they have a

NOTE Confidence: 0.81407756

 $01{:}12{:}42.672 \dashrightarrow 01{:}12{:}45.796$ term that they use when you have one.

NOTE Confidence: 0.81407756

 $01:12:45.800 \longrightarrow 01:12:47.870$ Of the genes for cystic fibrosis.

NOTE Confidence: 0.81407756

 $01{:}12{:}47.870 \dashrightarrow 01{:}12{:}50.306$ One of the variants that can cause

NOTE Confidence: 0.81407756

 $01{:}12{:}50{.}306 \dashrightarrow 01{:}12{:}52{.}026$ cystic fibrosis and a variant

NOTE Confidence: 0.81407756

 $01:12:52.026 \rightarrow 01:12:54.147$ that is not yet known to cause

NOTE Confidence: 0.81407756

01:12:54.147 --> 01:12:55.809 cystic fibrosis is unclear.

NOTE Confidence: 0.81407756

01:12:55.810 --> 01:12:56.604 It's unknown.

NOTE Confidence: 0.81407756

 $01:12:56.604 \rightarrow 01:12:59.383$ And you have a normal sweat test.

NOTE Confidence: 0.81407756

 $01{:}12{:}59{.}390 \dashrightarrow 01{:}13{:}01{.}757$ They then go ahead and say that you have

NOTE Confidence: 0.81407756

 $01:13:01.757 \longrightarrow 01:13:04.287$ a cystic fibrosis metabolic syndrome,

 $01:13:04.290 \longrightarrow 01:13:06.040$ which means that you don't

NOTE Confidence: 0.81407756

 $01:13:06.040 \longrightarrow 01:13:07.090$ have cystic fibrosis.

NOTE Confidence: 0.81407756

01:13:07.090 --> 01:13:08.994 You normal sweat test,

NOTE Confidence: 0.81407756

 $01:13:08.994 \rightarrow 01:13:09.470$ but.

NOTE Confidence: 0.81407756

01:13:09.470 --> 01:13:11.335 We don't know whether other

NOTE Confidence: 0.81407756

01:13:11.335 --> 01:13:13.615 variant means and you might get NOTE Confidence: 0.81407756

 $01:13:13.615 \longrightarrow 01:13:15.691$ cystic fibrosis and we need to

NOTE Confidence: 0.81407756

01:13:15.691 --> 01:13:18.017 keep testing you an one of my

NOTE Confidence: 0.81407756

01:13:18.017 --> 01:13:19.847 former fellows who I don't think NOTE Confidence: 0.81407756

01:13:19.850 --> 01:13:21.926 she'll mind me sharing this who,

NOTE Confidence: 0.81407756

 $01:13:21.930 \longrightarrow 01:13:24.345$ like many of us in the world,

NOTE Confidence: 0.81407756

01:13:24.350 --> 01:13:26.891 worries a lot ahead of child who

NOTE Confidence: 0.81407756

 $01:13:26.891 \longrightarrow 01:13:29.188$ had this happen to Ann for her.

NOTE Confidence: 0.81407756

 $01{:}13{:}29{.}190 \dashrightarrow 01{:}13{:}31{.}560$ This was a big worry.

NOTE Confidence: 0.81407756

 $01{:}13{:}31{.}560 \dashrightarrow 01{:}13{:}34{.}506$ That that the fact that she.

NOTE Confidence: 0.81407756

01:13:34.510 --> 01:13:36.418 Was now said your child doesn't

- NOTE Confidence: 0.81407756
- $01:13:36.418 \longrightarrow 01:13:37.372$ have cystic fibrosis,
- NOTE Confidence: 0.81407756
- $01:13:37.380 \longrightarrow 01:13:39.417$ but maybe he could get it at
- NOTE Confidence: 0.81407756
- $01:13:39.417 \longrightarrow 01:13:41.317$ some point for her was weighed
- NOTE Confidence: 0.81407756
- $01:13:41.317 \rightarrow 01:13:43.956$ very heavily on her and for her I
- NOTE Confidence: 0.81407756
- $01{:}13{:}43.956 \dashrightarrow 01{:}13{:}45.990$ think was very distressful to me.
- NOTE Confidence: 0.81407756
- $01:13:45.990 \longrightarrow 01:13:47.067$ With this means,
- NOTE Confidence: 0.81407756
- $01:13:47.067 \longrightarrow 01:13:48.862$ I actually think that newborn
- NOTE Confidence: 0.81407756
- $01:13:48.862 \rightarrow 01:13:50.717$ screening is an area where we
- NOTE Confidence: 0.81407756
- $01:13:50.717 \rightarrow 01:13:52.690$ have to be very careful for that,
- NOTE Confidence: 0.81407756
- $01:13:52.690 \rightarrow 01:13:54.290$ not only for that reason,
- NOTE Confidence: 0.81407756
- $01:13:54.290 \rightarrow 01:13:56.198$ but for a variety of reasons.
- NOTE Confidence: 0.81407756
- 01:13:56.200 --> 01:13:58.769 So I do not see new born screening
- NOTE Confidence: 0.81407756
- $01:13:58.769 \rightarrow 01:14:01.749$ shifting to a sequencing based model.
- NOTE Confidence: 0.81407756
- $01:14:01.750 \longrightarrow 01:14:03.958$ As the first place this goes,
- NOTE Confidence: 0.81407756
- $01:14:03.960 \rightarrow 01:14:06.168$ will it go there eventually possibly?
- NOTE Confidence: 0.81407756

01:14:06.170 --> 01:14:08.396 I think it will require a very

NOTE Confidence: 0.81407756

 $01{:}14{:}08.396 \dashrightarrow 01{:}14{:}10.274$ careful thought about the curation

NOTE Confidence: 0.81407756

 $01{:}14{:}10.274 \dashrightarrow 01{:}14{:}12.414$ of the information making decisions,

NOTE Confidence: 0.81407756

 $01:14:12.420 \rightarrow 01:14:13.892$ which variants to report,

NOTE Confidence: 0.81407756

 $01:14:13.892 \rightarrow 01:14:17.250$ which diseases to consider, how to do this.

NOTE Confidence: 0.81407756

 $01:14:17.250 \longrightarrow 01:14:19.130$ So I actually think.

NOTE Confidence: 0.81407756

 $01:14:19.130 \longrightarrow 01:14:21.674$ In 20 years we will be doing that,

NOTE Confidence: 0.81407756

 $01:14:21.680 \longrightarrow 01:14:23.815$ but I hope that the process to

NOTE Confidence: 0.81407756

 $01{:}14{:}23.815 \dashrightarrow 01{:}14{:}26.002$ get there will have allowed us to

NOTE Confidence: 0.81407756

 $01:14:26.002 \rightarrow 01:14:29.020$ develop how to do this in a thoughtful way.

NOTE Confidence: 0.81407756

 $01{:}14{:}29{.}020 \dashrightarrow 01{:}14{:}30{.}928$ I also think that it raises,

NOTE Confidence: 0.81407756

 $01:14:30.930 \longrightarrow 01:14:32.454$ as has been discussed,

NOTE Confidence: 0.81407756

 $01{:}14{:}32{.}454 \dashrightarrow 01{:}14{:}34{.}740$ for new born screening in general raises

NOTE Confidence: 0.87807447

 $01:14:34.803 \rightarrow 01:14:36.643$ new questions about informed consent

NOTE Confidence: 0.87807447

 $01:14:36.643 \rightarrow 01:14:38.860$ in the following way is that.

NOTE Confidence: 0.87807447

 $01:14:38.860 \rightarrow 01:14:40.610$ Historically, newborn screening is often

 $01:14:40.610 \rightarrow 01:14:42.790$ done without much discussion with parents,

NOTE Confidence: 0.87807447

 $01{:}14{:}42.790 \dashrightarrow 01{:}14{:}44.662$ and there's been a long gradual

NOTE Confidence: 0.87807447

01:14:44.662 --> 01:14:46.751 thrust in trying to do more

NOTE Confidence: 0.87807447

 $01:14:46.751 \rightarrow 01:14:48.295$ education about newborn screening

NOTE Confidence: 0.87807447

01:14:48.295 --> 01:14:50.640 prior to delivery and at delivery,

NOTE Confidence: 0.87807447

 $01{:}14{:}50{.}640 \dashrightarrow 01{:}14{:}53{.}040$ and I suspect if sequencing becomes

NOTE Confidence: 0.87807447

 $01:14:53.040 \longrightarrow 01:14:56.320$ part of it will be one more reason

NOTE Confidence: 0.87807447

 $01{:}14{:}56{.}320 \dashrightarrow 01{:}14{:}59{.}232$ why there will be more education and

NOTE Confidence: 0.87807447

 $01:14:59.232 \longrightarrow 01:15:01.980$ more engagement at that time. So.

NOTE Confidence: 0.8257083

 $01:15:03.110 \longrightarrow 01:15:05.336$ Thank you the next question please,

NOTE Confidence: 0.8257083

 $01{:}15{:}05{.}340 \dashrightarrow 01{:}15{:}07{.}643$ how do you counsel parents when you

NOTE Confidence: 0.8257083

 $01:15:07.643 \rightarrow 01:15:09.809$ identify mutations with variable penetrance,

NOTE Confidence: 0.8257083

 $01:15:09.810 \longrightarrow 01:15:12.036$ even if predicted to be pathogenic?

NOTE Confidence: 0.9043739

01:15:14.080 $\operatorname{-->}$ 01:15:17.608 You know, I actually that
's I think

NOTE Confidence: 0.9043739

01:15:17.608 --> 01:15:20.864 that's easier. I want to easier,
$01:15:20.864 \rightarrow 01:15:23.430$ it's more common. Because.

NOTE Confidence: 0.92042774

 $01:15:25.640 \rightarrow 01:15:30.938$ There's many circumstances where the outcome.

NOTE Confidence: 0.92042774

 $01:15:30.940 \rightarrow 01:15:33.516$ It's not clear what that's going to be,

NOTE Confidence: 0.92042774

 $01:15:33.520 \rightarrow 01:15:35.837$ so let's take cystic fibrosis and diabetes

NOTE Confidence: 0.92042774

 $01{:}15{:}35{.}837 \dashrightarrow 01{:}15{:}37{.}733$ and thinking about typically about two

NOTE Confidence: 0.92042774

 $01:15:37.733 \rightarrow 01:15:40.115$ or three year olds for four year olds

NOTE Confidence: 0.92042774

 $01:15:40.115 \rightarrow 01:15:42.209$ who are diagnosed with these conditions.

NOTE Confidence: 0.8497331

 $01:15:45.460 \longrightarrow 01:15:47.320$ What is variable? I mean,

NOTE Confidence: 0.8497331

 $01{:}15{:}47{.}320 \dashrightarrow 01{:}15{:}50{.}296$ use patterns as an example as a metaphor.

NOTE Confidence: 0.8497331

 $01:15:50.300 \rightarrow 01:15:52.160$ Which variable is the outcome?

NOTE Confidence: 0.8497331

01:15:52.160 --> 01:15:53.985 There are some children who

NOTE Confidence: 0.8497331

 $01:15:53.985 \longrightarrow 01:15:56.250$ have who will do really well.

NOTE Confidence: 0.8497331

 $01{:}15{:}56{.}250 \dashrightarrow 01{:}15{:}58{.}476$ Other children who will do who

NOTE Confidence: 0.8497331

 $01:15:58.476 \longrightarrow 01:16:00.604$ have challenges, and so it's so.

NOTE Confidence: 0.8497331

 $01:16:00.604 \rightarrow 01:16:02.334$ This notion of uncertainty that

NOTE Confidence: 0.8497331

01:16:02.334 --> 01:16:04.797 a copy is a clinical diagnosis,

- NOTE Confidence: 0.8497331
- $01{:}16{:}04.800 \dashrightarrow 01{:}16{:}06.840$ I think, is part and parcel
- NOTE Confidence: 0.8497331
- $01:16:06.840 \longrightarrow 01:16:08.900$ with much of clinical practice.
- NOTE Confidence: 0.8497331
- 01:16:08.900 --> 01:16:10.755 I think for some individuals
- NOTE Confidence: 0.8497331
- $01:16:10.755 \rightarrow 01:16:12.239$ that uncertainty is debilitating,
- NOTE Confidence: 0.8497331
- $01:16:12.240 \rightarrow 01:16:14.550$ but for many people they really.
- NOTE Confidence: 0.8497331
- $01:16:14.550 \longrightarrow 01:16:15.405$ Adapt to that,
- NOTE Confidence: 0.8497331
- $01:16:15.405 \rightarrow 01:16:17.400$ and so I think there's many circumstances.
- NOTE Confidence: 0.8497331
- 01:16:17.400 --> 01:16:19.110 I guess my own, as pulmonologist,
- NOTE Confidence: 0.8497331
- 01:16:19.110 $\operatorname{-->}$ 01:16:20.867 I take care of children with chronic
- NOTE Confidence: 0.8497331
- 01:16:20.867 --> 01:16:22.250 lung disease of prematurity.
- NOTE Confidence: 0.8497331
- 01:16:22.250 --> 01:16:24.238 Putting kids were born at 2425 weeks.
- NOTE Confidence: 0.8497331
- 01:16:24.240 --> 01:16:25.080 Have lung disease,
- NOTE Confidence: 0.8497331
- $01{:}16{:}25.080 \dashrightarrow 01{:}16{:}26.480$ that time that they're leaving
- NOTE Confidence: 0.8497331
- 01:16:26.480 --> 01:16:27.663 the NICU there, oxygen,
- NOTE Confidence: 0.8497331
- $01{:}16{:}27.663 \dashrightarrow 01{:}16{:}29.644$ the feeding tube and you know what
- NOTE Confidence: 0.8497331

01:16:29.644 $\operatorname{-->}$ 01:16:32.168 I can tell them is that I don't know

NOTE Confidence: 0.8497331

 $01:16:32.168 \longrightarrow 01:16:33.930$ where you'll be in three years.

NOTE Confidence: 0.8497331

01:16:33.930 --> 01:16:35.215 There's a there's a reasonable

NOTE Confidence: 0.8497331

 $01:16:35.215 \longrightarrow 01:16:37.157$ chance that you will be off all

NOTE Confidence: 0.8497331

 $01:16:37.157 \rightarrow 01:16:38.357$ these supports and developmentally

NOTE Confidence: 0.8497331

01:16:38.357 --> 01:16:40.186 you're doing fine or you'll be

NOTE Confidence: 0.8497331

 $01:16:40.186 \longrightarrow 01:16:41.902$ off the support that you have

NOTE Confidence: 0.8497331

 $01:16:41.902 \longrightarrow 01:16:43.410$ developmental issues or vice versa.

NOTE Confidence: 0.8497331

01:16:43.410 --> 01:16:44.960 And I say you know.

NOTE Confidence: 0.8497331

 $01:16:44.960 \rightarrow 01:16:46.490$ The the We don't know,

NOTE Confidence: 0.8497331

 $01{:}16{:}46{.}490 \dashrightarrow 01{:}16{:}49{.}235$ but part of my answer back to your question.

NOTE Confidence: 0.8497331

01:16:49.240 --> 01:16:51.776 Patrick is one thing I say to my

NOTE Confidence: 0.8497331

01:16:51.776 --> 01:16:53.625 families myself is why I don't

NOTE Confidence: 0.8497331

 $01:16:53.625 \rightarrow 01:16:55.055$ know where this will go.

NOTE Confidence: 0.8497331

01:16:55.060 --> 01:16:57.062 I'll be with you for that journey

NOTE Confidence: 0.8497331

 $01{:}16{:}57.062 \dashrightarrow 01{:}16{:}59.317$ and we will continue to work together

- NOTE Confidence: 0.8497331
- 01:16:59.317 --> 01:17:01.943 to sort that out over time and I
- NOTE Confidence: 0.8497331
- $01{:}17{:}01{.}943 \dashrightarrow 01{:}17{:}03{.}945$ think for me that's a critical issue
- NOTE Confidence: 0.8497331
- $01:17:03.945 \rightarrow 01:17:05.432$ as relates to genetic information.
- NOTE Confidence: 0.8497331
- $01:17:05.432 \longrightarrow 01:17:07.220$ Is the notion of follow up
- NOTE Confidence: 0.8497331
- $01{:}17{:}07{.}274 \dashrightarrow 01{:}17{:}09{.}122$ follow up with their primary care
- NOTE Confidence: 0.8497331
- 01:17:09.122 --> 01:17:10.354 provider or somebody else?
- NOTE Confidence: 0.8497331
- $01:17:10.360 \longrightarrow 01:17:12.208$ The fact that the family feels
- NOTE Confidence: 0.8497331
- $01:17:12.208 \longrightarrow 01:17:13.812$ like they're connected to somebody
- NOTE Confidence: 0.8497331
- $01{:}17{:}13.812 \dashrightarrow 01{:}17{:}15.696$ whom they can ask questions of.
- NOTE Confidence: 0.8497331
- $01:17:15.700 \longrightarrow 01:17:16.808$ As time goes on.
- NOTE Confidence: 0.8476957
- 01:17:18.600 --> 01:17:20.704 Thanks Ben I we've got time for one
- NOTE Confidence: 0.8476957
- 01:17:20.704 --> 01:17:22.308 more question which I'm going to.
- NOTE Confidence: 0.8476957
- $01{:}17{:}22.310 \dashrightarrow 01{:}17{:}24.430$ I'm going to take myself if I can.
- NOTE Confidence: 0.8476957
- 01:17:24.430 --> 01:17:26.334 You know you were talking about trying to
- NOTE Confidence: 0.8476957
- $01{:}17{:}26.334 \dashrightarrow 01{:}17{:}28.396$ get a sense from patients, for example,
- NOTE Confidence: 0.8476957

01:17:28.396 --> 01:17:30.790 or subjects in a study for what you know,

NOTE Confidence: 0.8476957

01:17:30.790 --> 01:17:32.710 what if what whether they would or would

NOTE Confidence: 0.8476957

 $01{:}17{:}32{.}710$ --> $01{:}17{:}34{.}371$ not value this certain information and

NOTE Confidence: 0.8476957

 $01:17:34.371 \longrightarrow 01:17:36.360$ trying to trying to figure out that.

NOTE Confidence: 0.8476957

 $01{:}17{:}36{.}360 \dashrightarrow 01{:}17{:}37{.}680$ Is this something people want.

NOTE Confidence: 0.8476957

 $01{:}17{:}37{.}680 \dashrightarrow 01{:}17{:}39{.}507$ I'd be curious to know if there are data NOTE Confidence: 0.8476957

 $01:17:39.507 \rightarrow 01:17:41.584$ or if you've got personal experience with

NOTE Confidence: 0.8476957

 $01:17:41.584 \rightarrow 01:17:43.510$ this about people looking in retrospect,

NOTE Confidence: 0.8476957

01:17:43.510 --> 01:17:44.293 not, you know,

NOTE Confidence: 0.8476957

 $01:17:44.293 \rightarrow 01:17:46.430$ trying to figure out what they would want,

NOTE Confidence: 0.8476957

 $01:17:46.430 \longrightarrow 01:17:48.100$ but people who found out.

NOTE Confidence: 0.8476957

01:17:48.100 --> 01:17:50.074 Good news or bad news or

NOTE Confidence: 0.8476957

01:17:50.074 --> 01:17:51.390 equivocal news to people?

NOTE Confidence: 0.8476957

01:17:51.390 --> 01:17:53.184 Look after the fact in in

NOTE Confidence: 0.8476957

 $01{:}17{:}53{.}184 \dashrightarrow 01{:}17{:}55{.}010$ parents or in patients directly.

NOTE Confidence: 0.8476957

01:17:55.010 --> 01:17:57.306 You know, I wish I hadn't known.

- NOTE Confidence: 0.8476957
- 01:17:57.310 --> 01:17:59.620 I'm glad I knew someone studied that.

NOTE Confidence: 0.8476957

01:17:59.620 --> 01:17:59.940 Yeah,

NOTE Confidence: 0.8225839

 $01:17:59.940 \rightarrow 01:18:00.930$ actually, we certainly

NOTE Confidence: 0.8225839

 $01:18:00.930 \longrightarrow 01:18:02.910$ have done a lot of that.

NOTE Confidence: 0.8225839

 $01:18:02.910 \longrightarrow 01:18:05.206$ We, you know, one of the things

NOTE Confidence: 0.8225839

 $01:18:05.206 \rightarrow 01:18:07.840$ that we do as part of our vote,

NOTE Confidence: 0.8225839

 $01:18:07.840 \longrightarrow 01:18:09.490$ both the next Gen study,

NOTE Confidence: 0.8225839

 $01:18:09.490 \rightarrow 01:18:11.842$ which was a carrier study as well

NOTE Confidence: 0.8225839

 $01{:}18{:}11{.}842 \dashrightarrow 01{:}18{:}14{.}634$ as the charm is we do a range of

NOTE Confidence: 0.8225839

 $01:18:14.634 \longrightarrow 01:18:16.297$ of the side surveys interviews

NOTE Confidence: 0.8225839

 $01{:}18{:}16.297 \dashrightarrow 01{:}18{:}18.547$ where we talked to people.

NOTE Confidence: 0.8225839

01:18:18.550 --> 01:18:20.657 That a month or so after the

NOTE Confidence: 0.8225839

01:18:20.657 --> 01:18:22.234 results disclosure to find out

NOTE Confidence: 0.8225839

 $01{:}18{:}22{.}234 \dashrightarrow 01{:}18{:}23{.}769$ how that experience went with

NOTE Confidence: 0.8225839

 $01{:}18{:}23.769 \dashrightarrow 01{:}18{:}25.689$ a follow the information and we

NOTE Confidence: 0.8225839

 $01:18:25.689 \rightarrow 01:18:27.880$ also have done that where we take

NOTE Confidence: 0.8225839

 $01{:}18{:}27.880 \dashrightarrow 01{:}18{:}29.440$ people who got negative results.

NOTE Confidence: 0.8225839

 $01:18:29.440 \longrightarrow 01:18:30.990$ Positive results were uncertain results.

NOTE Confidence: 0.8225839

 $01{:}18{:}30{.}990 \dashrightarrow 01{:}18{:}33{.}286$ We also have a separate set of interviews

NOTE Confidence: 0.8225839

 $01{:}18{:}33.286 \dashrightarrow 01{:}18{:}35.965$ that we did regarding this idea of utility.

NOTE Confidence: 0.8225839

01:18:35.970 --> 01:18:38.450 You know what's important to them as well,

NOTE Confidence: 0.8225839

 $01{:}18{:}38{.}450 \dashrightarrow 01{:}18{:}41{.}195$ and so it's a way of trying to understand

NOTE Confidence: 0.8225839

 $01:18:41.195 \rightarrow 01:18:43.736$ better what people think of this as well.

NOTE Confidence: 0.8225839

01:18:43.740 --> 01:18:45.032 Well, say is that.

NOTE Confidence: 0.8225839

 $01:18:45.032 \rightarrow 01:18:47.847$ And this may be again even in this

NOTE Confidence: 0.8225839

 $01:18:47.847 \longrightarrow 01:18:49.847$ context of this selection bias,

NOTE Confidence: 0.8225839

 $01{:}18{:}49{.}850 \dashrightarrow 01{:}18{:}52{.}130$ because people had to sign up for this

NOTE Confidence: 0.8225839

 $01{:}18{:}52{.}130 \dashrightarrow 01{:}18{:}54{.}457$ and that most people don't regret it.

NOTE Confidence: 0.8225839

 $01:18:54.460 \longrightarrow 01:18:55.990$ They don't feel like God.

NOTE Confidence: 0.8225839

01:18:55.990 --> 01:18:57.826 I wish I didn't know this.

NOTE Confidence: 0.8225839

 $01:18:57.830 \longrightarrow 01:18:59.672$ They figure out how to sort

- NOTE Confidence: 0.8225839
- $01:18:59.672 \longrightarrow 01:19:00.593$ of incorporate it.
- NOTE Confidence: 0.8225839
- 01:19:00.600 --> 01:19:01.860 And so you know,
- NOTE Confidence: 0.8225839
- $01{:}19{:}01{.}860 \dashrightarrow 01{:}19{:}04{.}196$ while I still am sceptical and wanted
- NOTE Confidence: 0.8225839
- $01:19:04.196 \rightarrow 01:19:06.440$ and really want to urge caution.
- NOTE Confidence: 0.8225839
- $01:19:06.440 \rightarrow 01:19:09.079$ With how we roll this out carefully,
- NOTE Confidence: 0.8225839
- $01:19:09.080 \longrightarrow 01:19:10.912$ we do it well.
- NOTE Confidence: 0.8225839
- $01:19:10.912 \rightarrow 01:19:14.240$ I'm less worried that most people have.
- NOTE Confidence: 0.8225839
- $01:19:14.240 \longrightarrow 01:19:15.544$ Adverse consequences of the
- NOTE Confidence: 0.8225839
- 01:19:15.544 --> 01:19:16.196 information itself,
- NOTE Confidence: 0.8225839
- $01:19:16.200 \rightarrow 01:19:18.237$ but I believe that the way we
- NOTE Confidence: 0.8225839
- $01:19:18.237 \rightarrow 01:19:20.450$ minimize that is by being thoughtful,
- NOTE Confidence: 0.8225839
- 01:19:20.450 --> 01:19:22.316 but making sure resources are available
- NOTE Confidence: 0.8225839
- $01{:}19{:}22{.}316 \dashrightarrow 01{:}19{:}24{.}380$ and also by tempering our enthusiasm.
- NOTE Confidence: 0.8225839
- 01:19:24.380 $\operatorname{-->}$ 01:19:26.660 And I think the example of back to
- NOTE Confidence: 0.8225839
- $01{:}19{:}26.660 \dashrightarrow 01{:}19{:}28.808$ the issue of new born screening is
- NOTE Confidence: 0.8225839

 $01:19:28.808 \rightarrow 01:19:31.064$ really a good example where that's

NOTE Confidence: 0.8225839

 $01{:}19{:}31{.}137 \dashrightarrow 01{:}19{:}33{.}272$ probably one of the last places I

NOTE Confidence: 0.8225839

 $01:19:33.272 \longrightarrow 01:19:35.266$ would want to go for doing this,

NOTE Confidence: 0.8225839

 $01{:}19{:}35{.}266 \dashrightarrow 01{:}19{:}37{.}716$ but in the context of of some body who

NOTE Confidence: 0.8225839

 $01:19:37.716 \longrightarrow 01:19:40.397$ is at risk for hereditary cancer syndrome,

NOTE Confidence: 0.8225839

 $01:19:40.400 \longrightarrow 01:19:42.352$ in the context of a family of a

NOTE Confidence: 0.8225839

01:19:42.352 --> 01:19:44.203 child who has parent development

NOTE Confidence: 0.8225839

 $01:19:44.203 \rightarrow 01:19:46.363$ disabilities looking for the cause.

NOTE Confidence: 0.8225839

01:19:46.370 --> 01:19:48.452 I think it's rare that some
body

NOTE Confidence: 0.8225839

01:19:48.452 --> 01:19:49.493 has testing done,

NOTE Confidence: 0.8225839

 $01{:}19{:}49{.}500 \dashrightarrow 01{:}19{:}51{.}936$ even if the result is an unexpected

NOTE Confidence: 0.8225839

 $01{:}19{:}51{.}936 \dashrightarrow 01{:}19{:}53{.}330$ actual finding somewhere else.

NOTE Confidence: 0.8225839

 $01:19:53.330 \longrightarrow 01:19:55.070$ They're usually open to that

NOTE Confidence: 0.8225839

 $01:19:55.070 \longrightarrow 01:19:56.810$ idea that usually accept that.

NOTE Confidence: 0.9123243

01:19:58.590 --> 01:20:00.938 Thank you very much.

NOTE Confidence: 0.9123243

 $01{:}20{:}00{.}940 \dashrightarrow 01{:}20{:}02{.}548$ Doctor Ben will fund this advance

- NOTE Confidence: 0.9123243
- $01:20:02.548 \longrightarrow 01:20:03.352$ tremendous session to night.

NOTE Confidence: 0.9123243

 $01:20:03.360 \longrightarrow 01:20:04.440$ We really appreciate it.

NOTE Confidence: 0.9123243

 $01:20:04.440 \rightarrow 01:20:06.590$ We're sorry we couldn't do this in person,

NOTE Confidence: 0.9123243

 $01{:}20{:}06{.}590 \dashrightarrow 01{:}20{:}09{.}292$ but sometime down the road we will

NOTE Confidence: 0.9123243

 $01:20:09.292 \longrightarrow 01:20:11.688$ absolutely get you to come join us.

NOTE Confidence: 0.9123243

 $01{:}20{:}11.690 \dashrightarrow 01{:}20{:}13.322$ And thank you very much and I thank

NOTE Confidence: 0.9123243

 $01:20:13.322 \rightarrow 01:20:15.157$ you all for attending on the website.

NOTE Confidence: 0.9123243

 $01:20:15.160 \rightarrow 01:20:16.546$ You can see when the upcoming

NOTE Confidence: 0.9123243

 $01:20:16.546 \longrightarrow 01:20:17.470$ seminars are a bending.

NOTE Confidence: 0.9123243

01:20:17.470 --> 01:20:19.279 Like if you come here we take you out

NOTE Confidence: 0.9123243

 $01{:}20{:}19.279 \dashrightarrow 01{:}20{:}21.257$ to dinner and but then you'd have a

NOTE Confidence: 0.9123243

 $01{:}20{:}21{.}257 \dashrightarrow 01{:}20{:}23{.}009$ long flight home and all this stuff.

NOTE Confidence: 0.9123243

01:20:23.010 --> 01:20:24.706 So now you know no dinner but you

NOTE Confidence: 0.9123243

01:20:24.706 --> 01:20:26.621 get to just to kind of turn off

NOTE Confidence: 0.9123243

 $01{:}20{:}26.621 \dashrightarrow 01{:}20{:}28.320$ the computer and put your feet up.

NOTE Confidence: 0.9123243

- 01:20:28.320 --> 01:20:29.248 No airplane right either.
- NOTE Confidence: 0.8262469
- $01{:}20{:}30{.}550 \dashrightarrow 01{:}20{:}32{.}468$ This is true. It's a pleasure to
- NOTE Confidence: 0.8262469
- $01{:}20{:}32{.}468 \dashrightarrow 01{:}20{:}34{.}586$ see all of you as especially you
- NOTE Confidence: 0.8262469
- 01:20:34.586 --> 01:20:36.750 Mark and Karen and see you next
- NOTE Confidence: 0.8262469
- $01:20:36.750 \longrightarrow 01:20:38.220$ time. See you next time.
- NOTE Confidence: 0.8262469
- 01:20:38.220 --> 01:20:41.637 Thanks folks. Goodnight