

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 --> 00:00:08.648 Welcome to our evening Ethics seminar

NOTE Confidence: 0.8107822

00:00:08.648 --> 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 --> 00:00:18.860 They are very much interrelated,

NOTE Confidence: 0.8107822

00:00:18.860 --> 00:00:22.076 but tonight we have a very special academic

NOTE Confidence: 0.8107822

00:00:22.076 --> 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 --> 00:00:28.686 before perhaps that our upcoming

NOTE Confidence: 0.8107822

00:00:28.686 --> 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 --> 00:00:33.084 About his book.

NOTE Confidence: 0.8107822

00:00:33.084 --> 00:00:34.885 Our malady, which is a very easy

NOTE Confidence: 0.8107822

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

NOTE Confidence: 0.8107822

00:00:35.920 --> 00:00:38.332 It's it's either a very small book or along

NOTE Confidence: 0.8107822

00:00:38.332 --> 00:00:40.309 pamphlet depending on how you look at it,

NOTE Confidence: 0.8107822

00:00:40.310 --> 00:00:42.109 but it's a it's an excellent read.

NOTE Confidence: 0.8107822

00:00:42.110 --> 00:00:43.944 I mentioned it's now to the medical

NOTE Confidence: 0.8107822

00:00:43.944 --> 00:00:45.053 students because some medical

NOTE Confidence: 0.8107822

00:00:45.053 --> 00:00:46.679 students have copies that are still

NOTE Confidence: 0.8107822

00:00:46.679 --> 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 --> 00:00:51.560 We're looking forward to having

NOTE Confidence: 0.8107822

00:00:51.560 --> 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 --> 00:01:00.606 I've known for many years in the

NOTE Confidence: 0.802747

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 --> 00:01:06.417 and and Doctor Ben will fund is has
NOTE Confidence: 0.802747

00:01:06.417 --> 00:01:08.419 had a remarkable career in bioethics.
NOTE Confidence: 0.802747

00:01:08.420 --> 00:01:09.560 Believe it or not,
NOTE Confidence: 0.802747

00:01:09.560 --> 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 --> 00:01:16.192 the center of gravity.
NOTE Confidence: 0.802747

00:01:16.200 --> 00:01:18.584 1.1 could have argued that it was Wisconsin.
NOTE Confidence: 0.802747

00:01:18.590 --> 00:01:20.378 Maybe some would still say so,
NOTE Confidence: 0.802747

00:01:20.380 --> 00:01:22.529 but I think it kind of shifted
NOTE Confidence: 0.802747

00:01:22.529 --> 00:01:24.483 West from Wisconsin to a place
NOTE Confidence: 0.802747

00:01:24.483 --> 00:01:26.093 called the Treuman Katz Center.
NOTE Confidence: 0.802747

00:01:26.100 --> 00:01:28.025 I'll talk about that in a moment,
NOTE Confidence: 0.802747

00:01:28.030 --> 00:01:29.662 but tonight's speaker to let you

NOTE Confidence: 0.802747
00:01:29.662 --> 00:01:31.340 know is Doctor Ben will font.
NOTE Confidence: 0.802747
00:01:31.340 --> 00:01:32.990 He is professor of the vision
NOTE Confidence: 0.802747
00:01:32.990 --> 00:01:34.457 of bioethics and palliative care
NOTE Confidence: 0.802747
00:01:34.457 --> 00:01:36.027 and pulmonary Sleep Medicine in
NOTE Confidence: 0.802747
00:01:36.027 --> 00:01:37.642 the Department of Pediatrics at
NOTE Confidence: 0.802747
00:01:37.642 --> 00:01:38.878 the University of Washington.
NOTE Confidence: 0.802747
00:01:38.880 --> 00:01:41.981 He's an investigator at the Truman Katz
NOTE Confidence: 0.802747
00:01:41.981 --> 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 --> 00:02:04.350 which is part of the reason why the

NOTE Confidence: 0.802747

00:02:04.350 --> 00:02:06.238 center of gravity made that shift,

NOTE Confidence: 0.802747

00:02:06.240 --> 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 --> 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 --> 00:02:17.086 and to learn an it is.

NOTE Confidence: 0.802747

00:02:17.090 --> 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 --> 00:02:21.804 important Institute here with us.

NOTE Confidence: 0.78285277

00:02:23.880 --> 00:02:25.506 Doctor Wilfong's got leadership roles in

NOTE Confidence: 0.78285277

00:02:25.506 --> 00:02:27.118 various national initiatives, and we're

NOTE Confidence: 0.78285277

00:02:27.118 --> 00:02:30.310 really not going to get into that now.

NOTE Confidence: 0.78285277

00:02:30.310 --> 00:02:31.828 But if there's a quite done,

NOTE Confidence: 0.78285277

00:02:31.830 --> 00:02:33.990 I'll throw in one of his titles or one of

NOTE Confidence: 0.78285277

00:02:34.043 --> 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 --> 00:02:39.292 I want to get to the conversation and

NOTE Confidence: 0.78285277

00:02:39.292 --> 00:02:41.488 the way the conversation is going to go,

NOTE Confidence: 0.78285277

00:02:41.490 --> 00:02:43.093 as many of you are familiar with

NOTE Confidence: 0.78285277

00:02:43.093 --> 00:02:44.693 is Doctor will finally speak for

NOTE Confidence: 0.78285277

00:02:44.693 --> 00:02:46.312 about 45 minutes, plus or minus.

NOTE Confidence: 0.78285277

00:02:46.312 --> 00:02:47.836 After that, I will invite you

NOTE Confidence: 0.78285277

00:02:47.836 --> 00:02:49.106 to submit questions via Q&A,

NOTE Confidence: 0.78285277

00:02:49.110 --> 00:02:50.832 and then I will be reading them

NOTE Confidence: 0.78285277

00:02:50.832 --> 00:02:51.900 to Doctor will fund.

NOTE Confidence: 0.78285277

00:02:51.900 --> 00:02:54.186 So if you have any questions along the way,

NOTE Confidence: 0.78285277

00:02:54.190 --> 00:02:55.912 that's how we'll do that and we
NOTE Confidence: 0.78285277

00:02:55.912 --> 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 --> 00:03:01.560 question, I apologize in advance.
NOTE Confidence: 0.78285277

00:03:01.560 --> 00:03:03.820 But we'll do the best we can.
NOTE Confidence: 0.78285277

00:03:03.820 --> 00:03:06.032 I think it's going to be very
NOTE Confidence: 0.78285277

00:03:06.032 --> 00:03:08.290 interesting night on a problem that I
NOTE Confidence: 0.78285277

00:03:08.290 --> 00:03:10.546 think is something that's not an easy
NOTE Confidence: 0.78285277

00:03:10.546 --> 00:03:12.520 question both in the clinical sphere
NOTE Confidence: 0.78285277

00:03:12.520 --> 00:03:14.666 as well as in the research arena,
NOTE Confidence: 0.78285277

00:03:14.666 --> 00:03:17.099 and Doctor Wilson has a great deal of
NOTE Confidence: 0.78285277

00:03:17.099 --> 00:03:19.001 experience and expertise in bioethics in
NOTE Confidence: 0.78285277

00:03:19.001 --> 00:03:21.259 both the clinical and research arena,
NOTE Confidence: 0.78285277

00:03:21.260 --> 00:03:23.012 and so with that genetic information
NOTE Confidence: 0.78285277

00:03:23.012 --> 00:03:24.180 disclosure and whole genome
NOTE Confidence: 0.78285277

00:03:24.234 --> 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

NOTE Confidence: 0.8764313

00:03:29.546 --> 00:03:31.688 share my screen. To start with.

NOTE Confidence: 0.82752955

00:03:35.030 --> 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 --> 00:03:44.080 Sorry, OK, here we are.

NOTE Confidence: 0.8072139

00:03:44.080 --> 00:03:45.592 So first of all,

NOTE Confidence: 0.8072139

00:03:45.592 --> 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 --> 00:03:53.904 at least virtually with you in New Haven.

NOTE Confidence: 0.8072139

00:03:53.910 --> 00:03:56.913 And I look forward to the opportunity

NOTE Confidence: 0.8072139

00:03:56.913 --> 00:03:59.797 of being back here in person so.

NOTE Confidence: 0.8072139

00:03:59.800 --> 00:04:01.858 For those of you who are thinking

NOTE Confidence: 0.8072139

00:04:01.858 --> 00:04:03.401 about my title and wondering

NOTE Confidence: 0.8072139

00:04:03.401 --> 00:04:05.494 what am I going to talk about,

NOTE Confidence: 0.8072139

00:04:05.500 --> 00:04:07.502 I'm going to ask you since it's

NOTE Confidence: 0.8072139

00:04:07.502 --> 00:04:09.697 been a year of Covid Anna year,
NOTE Confidence: 0.8072139

00:04:09.700 --> 00:04:11.200 I'll probably watching more Netflix
NOTE Confidence: 0.8072139

00:04:11.200 --> 00:04:12.700 then you've watched in awhile.
NOTE Confidence: 0.8072139

00:04:12.700 --> 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 --> 00:04:20.418 I want you to think of what movie is sort of
NOTE Confidence: 0.8072139

00:04:20.418 --> 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 --> 00:04:28.810 you pause and think about that.
NOTE Confidence: 0.8072139

00:04:28.810 --> 00:04:29.946 Because of our format,
NOTE Confidence: 0.8072139

00:04:29.946 --> 00:04:32.369 I can't really hear what your answers are,
NOTE Confidence: 0.8072139

00:04:32.370 --> 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 --> 00:04:38.220 thought of this movie.
NOTE Confidence: 0.8072139

00:04:38.220 --> 00:04:40.411 Gattaca was at 25 years old and
NOTE Confidence: 0.8072139

00:04:40.411 --> 00:04:42.490 it was really this future where

NOTE Confidence: 0.8072139

00:04:42.490 --> 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 --> 00:04:49.000 And of course this is sort of a

NOTE Confidence: 0.8072139

00:04:49.000 --> 00:04:50.815 futuristic version of the implications

NOTE Confidence: 0.8072139

00:04:50.815 --> 00:04:53.173 of sequencing of knowledge about genetics.

NOTE Confidence: 0.8072139

00:04:53.180 --> 00:04:55.328 Being able to predict how people

NOTE Confidence: 0.8072139

00:04:55.328 --> 00:04:57.940 will behave and how they will do it.

NOTE Confidence: 0.8072139

00:04:57.940 --> 00:05:00.430 Certainly very sort of a

NOTE Confidence: 0.8072139

00:05:00.430 --> 00:05:02.920 dystopian vision of the world.

NOTE Confidence: 0.8072139

00:05:02.920 --> 00:05:05.360 I hope for a variety of reasons that

NOTE Confidence: 0.8072139

00:05:05.360 --> 00:05:07.649 you'll leave something more optimistic,

NOTE Confidence: 0.8072139

00:05:07.650 --> 00:05:10.692 and I'm actually going to share

NOTE Confidence: 0.8072139

00:05:10.692 --> 00:05:13.290 with you another movie that.

NOTE Confidence: 0.8072139

00:05:13.290 --> 00:05:16.644 I mean partly seriously and partly

NOTE Confidence: 0.8072139

00:05:16.644 --> 00:05:20.509 jokingly is a better image for what?

NOTE Confidence: 0.8072139

00:05:20.510 --> 00:05:23.457 Genetics may bring us in the future.
NOTE Confidence: 0.8072139

00:05:23.460 --> 00:05:25.968 That's the graduate.
NOTE Confidence: 0.8072139

00:05:25.970 --> 00:05:28.700 Now the reason why I'm using this
NOTE Confidence: 0.8072139

00:05:28.700 --> 00:05:31.509 movie is because of for those of you.
NOTE Confidence: 0.8072139

00:05:31.510 --> 00:05:33.718 This is about 50 years old.
NOTE Confidence: 0.8072139

00:05:33.720 --> 00:05:37.032 For those of you who have not seen it,
NOTE Confidence: 0.8072139

00:05:37.040 --> 00:05:38.880 there is a very particular
NOTE Confidence: 0.8072139

00:05:38.880 --> 00:05:40.352 scene where the protagonist,
NOTE Confidence: 0.8072139

00:05:40.360 --> 00:05:42.448 who's also named Benjamin in this
NOTE Confidence: 0.8072139

00:05:42.448 --> 00:05:44.420 case played by Dustin Hoffman,
NOTE Confidence: 0.8072139

00:05:44.420 --> 00:05:46.712 is getting advice from a friend
NOTE Confidence: 0.8072139

00:05:46.712 --> 00:05:48.990 of his father's about what he
NOTE Confidence: 0.8072139

00:05:48.990 --> 00:05:50.685 should do for his career.
NOTE Confidence: 0.8072139

00:05:50.690 --> 00:05:54.197 And he has one word for Benjamin.
NOTE Confidence: 0.8072139

00:05:54.200 --> 00:05:57.380 And that word is plastics.
NOTE Confidence: 0.8072139

00:05:57.380 --> 00:05:59.788 This is taking place and you know this.

NOTE Confidence: 0.8072139

00:05:59.790 --> 00:06:02.198 This film came out in the early 60s.

NOTE Confidence: 0.8072139

00:06:02.200 --> 00:06:03.705 It's supposed to take place

NOTE Confidence: 0.8072139

00:06:03.705 --> 00:06:05.210 like in the late 50s.

NOTE Confidence: 0.8072139

00:06:05.210 --> 00:06:06.992 So clearly plastics at that point

NOTE Confidence: 0.8072139

00:06:06.992 --> 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 --> 00:06:12.771 Because I want to share with you

NOTE Confidence: 0.8072139

00:06:12.771 --> 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 Wisconsin

NOTE Confidence: 0.8072139

00:06:16.737 --> 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 --> 00:06:22.602 In bioethics and trying to think

NOTE Confidence: 0.8072139

00:06:22.602 --> 00:06:24.579 about what I would work on,

NOTE Confidence: 0.8072139

00:06:24.580 --> 00:06:26.260 and I had some ideas,

NOTE Confidence: 0.8072139

00:06:26.260 --> 00:06:27.640 but the Phil Farrell,

NOTE Confidence: 0.8072139

00:06:27.640 --> 00:06:30.628 who was the chair of Pediatrics at that time,
NOTE Confidence: 0.8072139

00:06:30.630 --> 00:06:32.646 sort of reminiscent of the graduate,
NOTE Confidence: 0.8072139

00:06:32.650 --> 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 --> 00:06:37.348 two in this case,
NOTE Confidence: 0.8072139

00:06:37.350 --> 00:06:39.366 maybe 2 words or three words.
NOTE Confidence: 0.8072139

00:06:39.370 --> 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 --> 00:06:51.729 three years ago they developed this new
NOTE Confidence: 0.81737673

00:06:51.729 --> 00:06:54.224 technique and it's going to change the world.
NOTE Confidence: 0.81737673

00:06:54.230 --> 00:06:56.883 You need to think about the ethical
NOTE Confidence: 0.81737673

00:06:56.883 --> 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 --> 00:07:02.926 it was microbiology and you should talk

NOTE Confidence: 0.81737673

00:07:02.926 --> 00:07:05.645 with norm thoughts to his director of your

NOTE Confidence: 0.81737673

00:07:05.645 --> 00:07:07.732 residency program because he thinks about

NOTE Confidence: 0.81737673

00:07:07.732 --> 00:07:09.868 the ethical issues of genetic testing.

NOTE Confidence: 0.81737673

00:07:09.870 --> 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 --> 00:07:15.095 you know any year they're going to

NOTE Confidence: 0.81737673

00:07:15.095 --> 00:07:17.390 identify the gene for cystic fibrosis.

NOTE Confidence: 0.81737673

00:07:17.390 --> 00:07:20.550 So you should think about what are the

NOTE Confidence: 0.81737673

00:07:20.550 --> 00:07:22.197 ethical implications of identifying

NOTE Confidence: 0.81737673

00:07:22.197 --> 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 --> 00:07:36.181 cystic fibrosis was identified and

NOTE Confidence: 0.81737673

00:07:36.181 --> 00:07:38.565 what happened in that six month

NOTE Confidence: 0.81737673

00:07:38.565 --> 00:07:41.232 period is that a number of companies
NOTE Confidence: 0.81737673

00:07:41.232 --> 00:07:42.816 started immediately market marketing.
NOTE Confidence: 0.85681623

00:07:45.160 --> 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 --> 00:07:52.283 that we addressed in article is how
NOTE Confidence: 0.85681623

00:07:52.283 --> 00:07:54.900 do you think about genetic testing?
NOTE Confidence: 0.85681623

00:07:54.900 --> 00:07:57.336 But when it becomes technically feasible?
NOTE Confidence: 0.85681623

00:07:57.340 --> 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 --> 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 --> 00:08:11.460 in this paper is a theme that really
NOTE Confidence: 0.85681623

00:08:11.460 --> 00:08:13.990 has followed through my entire career.
NOTE Confidence: 0.85681623

00:08:13.990 --> 00:08:16.849 The notion that.
NOTE Confidence: 0.85681623

00:08:16.850 --> 00:08:18.578 Genetic testing requires evidence
NOTE Confidence: 0.85681623

00:08:18.578 --> 00:08:20.738 of benefit and relative evidence

NOTE Confidence: 0.85681623

00:08:20.738 --> 00:08:23.469 of lack of harm to sort of have

NOTE Confidence: 0.85681623

00:08:23.469 --> 00:08:25.496 an appropriate balance that it

NOTE Confidence: 0.85681623

00:08:25.496 --> 00:08:26.999 requires understanding thoughts,

NOTE Confidence: 0.85681623

00:08:27.000 --> 00:08:29.436 issues of informed consent of education,

NOTE Confidence: 0.85681623

00:08:29.440 --> 00:08:30.246 quality control,

NOTE Confidence: 0.85681623

00:08:30.246 --> 00:08:33.067 counseling and that we have to think

NOTE Confidence: 0.85681623

00:08:33.067 --> 00:08:35.526 about the workforce issues as well,

NOTE Confidence: 0.85681623

00:08:35.530 --> 00:08:39.590 and so this paper that I worked on in 1990,

NOTE Confidence: 0.85681623

00:08:39.590 --> 00:08:42.886 the themes of this are still present today

NOTE Confidence: 0.85681623

00:08:42.886 --> 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 --> 00:08:50.320 so I want to begin with this sort of

NOTE Confidence: 0.85681623

00:08:50.399 --> 00:08:52.889 historical introduction to the issues that

NOTE Confidence: 0.85681623

00:08:52.889 --> 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?

NOTE Confidence: 0.85681623

00:08:57.536 --> 00:09:00.240 I've gotta Fast forward.
NOTE Confidence: 0.85681623

00:09:00.240 --> 00:09:02.720 15 years.
NOTE Confidence: 0.85681623

00:09:02.720 --> 00:09:03.569 14 years, 2004.
NOTE Confidence: 0.85681623

00:09:03.569 --> 00:09:05.550 At this point I was working at
NOTE Confidence: 0.85681623

00:09:05.615 --> 00:09:07.643 the NIH and the Department of
NOTE Confidence: 0.85681623

00:09:07.643 --> 00:09:09.310 Clinical Bioethics at the NIH.
NOTE Confidence: 0.85681623

00:09:09.310 --> 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 --> 00:09:19.044 consultations for researchers who
NOTE Confidence: 0.85681623

00:09:19.044 --> 00:09:20.928 had questions that they were facing.
NOTE Confidence: 0.85681623

00:09:20.930 --> 00:09:23.438 There were challenging for the research.
NOTE Confidence: 0.85681623

00:09:23.440 --> 00:09:27.360 And so the phone Rang and it was
NOTE Confidence: 0.85681623

00:09:27.360 --> 00:09:30.638 a investigator from the National
NOTE Confidence: 0.85681623

00:09:30.638 --> 00:09:32.960 Cancer Institute. It wasn't.

NOTE Confidence: 0.85681623

00:09:32.960 --> 00:09:35.540 At the time I thought myself,

NOTE Confidence: 0.85681623

00:09:35.540 --> 00:09:37.496 I didn't know that there were

NOTE Confidence: 0.85681623

00:09:37.496 --> 00:09:39.080 dermatologists working at the NCI.

NOTE Confidence: 0.85681623

00:09:39.080 --> 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 --> 00:09:46.170 Pigmentosa which I have to recall.

NOTE Confidence: 0.85681623

00:09:46.170 --> 00:09:48.590 I had a vague idea of what it was but

NOTE Confidence: 0.85681623

00:09:48.658 --> 00:09:50.553 couldn't remember although he explained

NOTE Confidence: 0.85681623

00:09:50.553 --> 00:09:53.569 much more to me on that phone call.

NOTE Confidence: 0.85681623

00:09:53.570 --> 00:09:55.724 This is a disease genetic disease

NOTE Confidence: 0.85681623

00:09:55.724 --> 00:09:57.849 that's very rare 100,000 people and

NOTE Confidence: 0.85681623

00:09:57.849 --> 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 --> 00:10:04.210 it was through this disease that

NOTE Confidence: 0.85681623

00:10:04.290 --> 00:10:06.840 the whole mechanisms of DNA repair
NOTE Confidence: 0.85681623

00:10:06.840 --> 00:10:09.694 were elucidated and the NIH had been
NOTE Confidence: 0.85681623

00:10:09.694 --> 00:10:12.207 studying this for the last 30 years,
NOTE Confidence: 0.85681623

00:10:12.210 --> 00:10:12.610 and.
NOTE Confidence: 0.8571908

00:10:14.730 --> 00:10:17.404 The implication of this disease is that
NOTE Confidence: 0.8571908

00:10:17.404 --> 00:10:20.297 because of this problem with DNA repair,
NOTE Confidence: 0.8571908

00:10:20.300 --> 00:10:23.128 children who have this disease are at
NOTE Confidence: 0.8571908

00:10:23.128 --> 00:10:25.868 a huge increase risk of sun cancer,
NOTE Confidence: 0.8571908

00:10:25.870 --> 00:10:28.150 so any exposure to UV light
NOTE Confidence: 0.8571908

00:10:28.150 --> 00:10:30.650 increases their risk of skin cancer.
NOTE Confidence: 0.8571908

00:10:30.650 --> 00:10:33.140 Some of them also have neurologic
NOTE Confidence: 0.8571908

00:10:33.140 --> 00:10:36.159 problems as well that develop overtime.
NOTE Confidence: 0.8571908

00:10:36.160 --> 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 --> 00:10:44.098 that is for kids with this condition,
NOTE Confidence: 0.8571908

00:10:44.100 --> 00:10:46.578 so they can actually have a normal

NOTE Confidence: 0.8571908

00:10:46.578 --> 00:10:49.344 life by having a week of camp that

NOTE Confidence: 0.8571908

00:10:49.344 --> 00:10:51.728 occurs all night long and they sleep

NOTE Confidence: 0.8571908

00:10:51.728 --> 00:10:54.448 all day and this is picture in front

NOTE Confidence: 0.8571908

00:10:54.450 --> 00:10:57.210 of you is an image from that camp.

NOTE Confidence: 0.8571908

00:10:57.210 --> 00:10:59.280 So why did he call me?

NOTE Confidence: 0.8571908

00:10:59.280 --> 00:11:01.975 Well, he the investigator was using at

NOTE Confidence: 0.8571908

00:11:01.975 --> 00:11:04.990 that time the state of the art techniques

NOTE Confidence: 0.8571908

00:11:04.990 --> 00:11:07.660 which had to do with organizing.

NOTE Confidence: 0.8571908

00:11:07.660 --> 00:11:09.935 Patients with this condition into

NOTE Confidence: 0.8571908

00:11:09.935 --> 00:11:12.703 seven different groups based upon a

NOTE Confidence: 0.8571908

00:11:12.703 --> 00:11:14.798 laboratory analysis of their cells.

NOTE Confidence: 0.8571908

00:11:14.800 --> 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 --> 00:11:22.794 but it was genetic based testing that

NOTE Confidence: 0.8571908

00:11:22.794 --> 00:11:25.553 really describe these seven different

NOTE Confidence: 0.8571908

00:11:25.553 --> 00:11:30.199 categories. And these categories.
NOTE Confidence: 0.8571908

00:11:30.200 --> 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 --> 00:11:37.573 but there's a lot of uncertainty about
NOTE Confidence: 0.8571908

00:11:37.573 --> 00:11:39.930 how meaningful these categories were.
NOTE Confidence: 0.8571908

00:11:39.930 --> 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 --> 00:11:44.652 study where they were doing research
NOTE Confidence: 0.8571908

00:11:44.652 --> 00:11:46.467 with patients with this condition.
NOTE Confidence: 0.8571908

00:11:46.470 --> 00:11:48.110 They were doing these studies,
NOTE Confidence: 0.8571908

00:11:48.110 --> 00:11:49.982 but they were not routinely disclosing
NOTE Confidence: 0.8571908

00:11:49.982 --> 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 --> 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 --> 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,

NOTE Confidence: 0.8571908

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 --> 00:12:10.895 what what complementary group you were in.

NOTE Confidence: 0.8571908

00:12:10.900 --> 00:12:12.224 And so the people.

NOTE Confidence: 0.8571908

00:12:12.224 --> 00:12:13.548 Then I hate you,

NOTE Confidence: 0.8571908

00:12:13.550 --> 00:12:15.518 and parts that we want to

NOTE Confidence: 0.8571908

00:12:15.518 --> 00:12:17.190 find out our results too.

NOTE Confidence: 0.8571908

00:12:17.190 --> 00:12:18.850 And the investigator contacted me

NOTE Confidence: 0.8571908

00:12:18.850 --> 00:12:20.830 because he was concerned about this.

NOTE Confidence: 0.8571908

00:12:20.830 --> 00:12:22.816 He was not sure whether this

NOTE Confidence: 0.8571908

00:12:22.816 --> 00:12:24.140 was appropriate to do,

NOTE Confidence: 0.8571908

00:12:24.140 --> 00:12:26.724 and so he really raised us as a

NOTE Confidence: 0.8571908

00:12:26.724 --> 00:12:28.767 question for me to think about.

NOTE Confidence: 0.8571908

00:12:28.770 --> 00:12:30.890 And this was the first time I really

NOTE Confidence: 0.8571908

00:12:30.890 --> 00:12:32.672 thought very specifically about the

NOTE Confidence: 0.8571908

00:12:32.672 --> 00:12:35.054 question of results disclosure in research,
NOTE Confidence: 0.8571908

00:12:35.060 --> 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 --> 00:12:40.026 but patients were wanting this information.
NOTE Confidence: 0.8571908

00:12:40.030 --> 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 --> 00:12:43.374 With that,
NOTE Confidence: 0.8571908

00:12:43.374 --> 00:12:45.870 a fellow at the NIH for Deep Rovinsky
NOTE Confidence: 0.8571908

00:12:45.941 --> 00:12:48.347 who's now the University Of Montreal?
NOTE Confidence: 0.8571908

00:12:48.350 --> 00:12:50.702 We wrote a paper about thinking about
NOTE Confidence: 0.8571908

00:12:50.702 --> 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 --> 00:12:57.805 we try to think through what we are the
NOTE Confidence: 0.8571908

00:12:57.805 --> 00:13:00.010 criteria for thinking about how to do this.
NOTE Confidence: 0.8571908

00:13:00.010 --> 00:13:01.630 Because the debate at the time
NOTE Confidence: 0.8571908

00:13:01.630 --> 00:13:03.390 was really in two directions.

NOTE Confidence: 0.8571908

00:13:03.390 --> 00:13:05.558 1 would say look this is for research

NOTE Confidence: 0.8571908

00:13:05.558 --> 00:13:07.189 purposes only and therefore there's

NOTE Confidence: 0.8571908

00:13:07.189 --> 00:13:09.247 no obligation to return any results.

NOTE Confidence: 0.8571908

00:13:09.250 --> 00:13:11.086 The other view was to say,

NOTE Confidence: 0.8571908

00:13:11.090 --> 00:13:13.554 you know people if we think about autonomy,

NOTE Confidence: 0.8571908

00:13:13.560 --> 00:13:14.793 people should have.

NOTE Confidence: 0.8571908

00:13:14.793 --> 00:13:16.848 Access to information about themselves,

NOTE Confidence: 0.8571908

00:13:16.850 --> 00:13:17.208 particularly.

NOTE Confidence: 0.8571908

00:13:17.208 --> 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 --> 00:13:26.301 took was to suggest that in fact rather

NOTE Confidence: 0.85168897

00:13:26.301 --> 00:13:28.512 than thinking of either a research

NOTE Confidence: 0.85168897

00:13:28.512 --> 00:13:30.756 focused or an autonomy focused approach,

NOTE Confidence: 0.85168897

00:13:30.760 --> 00:13:34.054 we should think of we described as a result
NOTE Confidence: 0.85168897

00:13:34.054 --> 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 --> 00:13:46.448 about 2 broad types of considerations
NOTE Confidence: 0.85168897

00:13:46.448 --> 00:13:49.430 one had to do with what I will describe
NOTE Confidence: 0.85168897

00:13:49.430 --> 00:13:50.592 as informational considerations.
NOTE Confidence: 0.85168897

00:13:50.592 --> 00:13:52.640 The other contextual considerations.
NOTE Confidence: 0.85168897

00:13:52.640 --> 00:13:54.880 And but informational considerations
NOTE Confidence: 0.85168897

00:13:54.880 --> 00:13:57.680 include two important concepts one
NOTE Confidence: 0.85168897

00:13:57.680 --> 00:14:00.657 has to do with analytic validity
NOTE Confidence: 0.85168897

00:14:00.657 --> 00:14:02.058 and clinical utility.
NOTE Confidence: 0.85168897

00:14:02.060 --> 00:14:04.044 Analytic validity refers to
NOTE Confidence: 0.85168897

00:14:04.044 --> 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 --> 00:14:10.771 A true result.

NOTE Confidence: 0.85168897

00:14:10.771 --> 00:14:12.733 An analytic validity refers to whether

NOTE Confidence: 0.85168897

00:14:12.733 --> 00:14:14.870 or not the samples were mixed up.

NOTE Confidence: 0.85168897

00:14:14.870 --> 00:14:16.823 It has to do with a wide range of

NOTE Confidence: 0.85168897

00:14:16.823 --> 00:14:18.384 issues that have nothing to do

NOTE Confidence: 0.85168897

00:14:18.384 --> 00:14:20.100 with the meaning of the result,

NOTE Confidence: 0.85168897

00:14:20.100 --> 00:14:22.038 but just whether or not the

NOTE Confidence: 0.85168897

00:14:22.038 --> 00:14:23.330 result is reliably true.

NOTE Confidence: 0.85168897

00:14:23.330 --> 00:14:25.110 Clinical utility as another critically

NOTE Confidence: 0.85168897

00:14:25.110 --> 00:14:27.211 important concept is the notion of

NOTE Confidence: 0.85168897

00:14:27.211 --> 00:14:29.038 what will you do with the information.

NOTE Confidence: 0.85168897

00:14:29.040 --> 00:14:30.625 How will the information be

NOTE Confidence: 0.85168897

00:14:30.625 --> 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 --> 00:14:35.943 analytic validity and clinical utility

NOTE Confidence: 0.85168897

00:14:35.943 --> 00:14:38.176 are still relevant when we talk about
NOTE Confidence: 0.85168897

00:14:38.176 --> 00:14:40.126 issues related to sequencing as well,
NOTE Confidence: 0.85168897

00:14:40.130 --> 00:14:41.710 and another word for often
NOTE Confidence: 0.85168897

00:14:41.710 --> 00:14:42.974 used for clinical utilities.
NOTE Confidence: 0.85168897

00:14:42.980 --> 00:14:44.380 Notion of action ability.
NOTE Confidence: 0.85168897

00:14:44.380 --> 00:14:47.380 What will you do with the information?
NOTE Confidence: 0.85168897

00:14:47.380 --> 00:14:49.823 Now I want to distinguish between analytic
NOTE Confidence: 0.85168897

00:14:49.823 --> 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 --> 00:14:58.110 Whether or not the result is real.
NOTE Confidence: 0.85168897

00:14:58.110 --> 00:14:59.860 Critical validity refers to whether
NOTE Confidence: 0.85168897

00:14:59.860 --> 00:15:01.610 or not it represents something
NOTE Confidence: 0.85168897

00:15:01.666 --> 00:15:02.936 about the person or not,
NOTE Confidence: 0.85168897

00:15:02.940 --> 00:15:04.872 because in fact in many cases
NOTE Confidence: 0.85168897

00:15:04.872 --> 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 --> 00:15:08.736 with zero dermatosis,

NOTE Confidence: 0.85168897

00:15:08.740 --> 00:15:11.568 that they didn't really know what the.

NOTE Confidence: 0.85168897

00:15:11.570 --> 00:15:13.460 Clinical validity of the information is,

NOTE Confidence: 0.85168897

00:15:13.460 --> 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 --> 00:15:20.447 one of these certain complement groups?

NOTE Confidence: 0.85168897

00:15:20.450 --> 00:15:22.557 It was unclear now the issue though.

NOTE Confidence: 0.85168897

00:15:22.560 --> 00:15:24.560 For the people in that study is that

NOTE Confidence: 0.85168897

00:15:24.560 --> 00:15:26.354 it had personal meaning for them

NOTE Confidence: 0.85168897

00:15:26.354 --> 00:15:28.625 that it mattered to them to know

NOTE Confidence: 0.85168897

00:15:28.625 --> 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 --> 00:15:34.138 So these notions of clinical validity

NOTE Confidence: 0.85168897

00:15:34.138 --> 00:15:35.757 and personal and personal meeting

NOTE Confidence: 0.85168897

00:15:35.757 --> 00:15:37.485 are two other dimensions that are

NOTE Confidence: 0.85168897

00:15:37.485 --> 00:15:39.269 important as we think about genetic
NOTE Confidence: 0.85168897

00:15:39.269 --> 00:15:41.285 results and why people may want them,
NOTE Confidence: 0.85168897

00:15:41.290 --> 00:15:42.795 why clinicians may be reluctant
NOTE Confidence: 0.85168897

00:15:42.795 --> 00:15:43.698 to return them,
NOTE Confidence: 0.85168897

00:15:43.700 --> 00:15:45.932 but these were the issues we
NOTE Confidence: 0.85168897

00:15:45.932 --> 00:15:47.420 have to think about.
NOTE Confidence: 0.85168897

00:15:47.420 --> 00:15:49.758 The second set of issues are right
NOTE Confidence: 0.85168897

00:15:49.758 --> 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 --> 00:15:55.947 with the relationship might be between
NOTE Confidence: 0.85168897

00:15:55.947 --> 00:15:58.034 the researcher and the the participants.
NOTE Confidence: 0.85168897

00:15:58.034 --> 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 --> 00:16:06.776 people donated samples that are
NOTE Confidence: 0.85168897

00:16:06.776 --> 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 --> 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 --> 00:16:15.322 what is the capacity of the research

NOTE Confidence: 0.85168897

00:16:15.322 --> 00:16:17.460 team to provide those results?

NOTE Confidence: 0.85168897

00:16:17.460 --> 00:16:20.099 If we assume that there are certain

NOTE Confidence: 0.85168897

00:16:20.099 --> 00:16:21.230 expectations regarding consent

NOTE Confidence: 0.84569705

00:16:21.291 --> 00:16:23.072 counseling, things like that.

NOTE Confidence: 0.84569705

00:16:23.072 --> 00:16:25.376 What if the researchers are being

NOTE Confidence: 0.84569705

00:16:25.376 --> 00:16:27.969 what the research is being done in

NOTE Confidence: 0.84569705

00:16:27.969 --> 00:16:30.389 a laboratory where the folks don't

NOTE Confidence: 0.84569705

00:16:30.389 --> 00:16:32.684 have that capacity and another

NOTE Confidence: 0.84569705

00:16:32.684 --> 00:16:34.878 consideration is whether this is

NOTE Confidence: 0.84569705

00:16:34.878 --> 00:16:36.898 uniquely available only through research,

NOTE Confidence: 0.84569705

00:16:36.900 --> 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 --> 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 --> 00:16:49.845 So I want to Fast forward now
NOTE Confidence: 0.84569705

00:16:49.845 --> 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 --> 00:16:56.185 with a bunch of neurologists who
NOTE Confidence: 0.84569705

00:16:56.185 --> 00:16:58.470 were interested in thinking about
NOTE Confidence: 0.84569705

00:16:58.470 --> 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 --> 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 --> 00:17:10.690 and the issue was based upon.
NOTE Confidence: 0.84569705

00:17:10.690 --> 00:17:12.922 An evolving consensus that we should
NOTE Confidence: 0.84569705

00:17:12.922 --> 00:17:15.593 only we should not be routinely

NOTE Confidence: 0.84569705

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 --> 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 --> 00:17:30.679 you would not be getting results.

NOTE Confidence: 0.84569705

00:17:30.680 --> 00:17:32.660 That this group is the dilemma.

NOTE Confidence: 0.84569705

00:17:32.660 --> 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 --> 00:17:38.280 they started realizing through one

NOTE Confidence: 0.84569705

00:17:38.280 --> 00:17:40.590 of the analytics studies they did.

NOTE Confidence: 0.84569705

00:17:40.590 --> 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 --> 00:17:46.306 clinically was not the same as they

NOTE Confidence: 0.84569705

00:17:46.306 --> 00:17:48.567 found out through their genetic research,

NOTE Confidence: 0.84569705

00:17:48.570 --> 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 --> 00:17:54.290 particularly when the consent forms
NOTE Confidence: 0.84569705

00:17:54.290 --> 00:17:56.859 said we will not disclose any results there.
NOTE Confidence: 0.84569705

00:17:56.860 --> 00:17:57.428 Question is,
NOTE Confidence: 0.84569705

00:17:57.428 --> 00:17:59.132 what obligations do we have to
NOTE Confidence: 0.84569705

00:17:59.132 --> 00:18:00.690 think about returning results,
NOTE Confidence: 0.84569705

00:18:00.690 --> 00:18:01.962 disclosing results when we
NOTE Confidence: 0.84569705

00:18:01.962 --> 00:18:03.234 told them they wouldn't?
NOTE Confidence: 0.84569705

00:18:03.240 --> 00:18:04.830 But this might be actually
NOTE Confidence: 0.84569705

00:18:04.830 --> 00:18:06.102 relevant to their management.
NOTE Confidence: 0.84569705

00:18:06.110 --> 00:18:08.891 Now I won't be spending a lot of time
NOTE Confidence: 0.84569705

00:18:08.891 --> 00:18:10.998 getting into how we address this.
NOTE Confidence: 0.84569705

00:18:11.000 --> 00:18:13.065 Other than simply say one of the
NOTE Confidence: 0.84569705

00:18:13.065 --> 00:18:15.427 approaches we use and we recommend it was,
NOTE Confidence: 0.84569705

00:18:15.430 --> 00:18:15.736 well,

NOTE Confidence: 0.84569705

00:18:15.736 --> 00:18:17.572 talk to the participants and see

NOTE Confidence: 0.84569705

00:18:17.572 --> 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

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 --> 00:18:26.746 results and some I mentioned to

NOTE Confidence: 0.84569705

00:18:26.746 --> 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 --> 00:18:31.777 but there are other results that were

NOTE Confidence: 0.84569705

00:18:31.777 --> 00:18:33.420 more ambiguous and more extraneous.

NOTE Confidence: 0.84569705

00:18:33.420 --> 00:18:36.356 They had that related to the way in

NOTE Confidence: 0.84569705

00:18:36.356 --> 00:18:39.187 which various results had some impact on.

NOTE Confidence: 0.84569705

00:18:39.190 --> 00:18:41.248 On IQ at on learning and there

NOTE Confidence: 0.84569705

00:18:41.248 --> 00:18:43.531 was much more interest among the

NOTE Confidence: 0.84569705

00:18:43.531 --> 00:18:45.299 participants in those results

NOTE Confidence: 0.84569705

00:18:45.299 --> 00:18:47.830 related to kidney disease and less
NOTE Confidence: 0.84569705

00:18:47.830 --> 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 --> 00:18:56.299 can be thinking so you have a sense of how.
NOTE Confidence: 0.84569705

00:18:56.300 --> 00:18:57.825 The research issues have been
NOTE Confidence: 0.84569705

00:18:57.825 --> 00:18:59.045 evolving over the decades,
NOTE Confidence: 0.84569705

00:18:59.050 --> 00:19:01.948 and at this point I want to shift gears
NOTE Confidence: 0.84569705

00:19:01.948 --> 00:19:04.939 a little bit and I'm going to share
NOTE Confidence: 0.84569705

00:19:04.939 --> 00:19:08.180 with you over the next 20 minutes or so.
NOTE Confidence: 0.84569705

00:19:08.180 --> 00:19:11.366 Two different projects that I've been
NOTE Confidence: 0.84569705

00:19:11.366 --> 00:19:15.240 involved with that that involve sequencing.
NOTE Confidence: 0.84569705

00:19:15.240 --> 00:19:16.997 And the first one is sequencing in
NOTE Confidence: 0.84569705

00:19:16.997 --> 00:19:18.730 the context of carrier screening.
NOTE Confidence: 0.84569705

00:19:18.730 --> 00:19:19.021 Now,
NOTE Confidence: 0.84569705

00:19:19.021 --> 00:19:21.058 if you recall from my first slide,
NOTE Confidence: 0.84569705

00:19:21.060 --> 00:19:23.868 I showed you from.

NOTE Confidence: 0.84569705

00:19:23.870 --> 00:19:25.904 From the paper on CF carrier

NOTE Confidence: 0.84569705

00:19:25.904 --> 00:19:27.260 testing is that one

NOTE Confidence: 0.784771299999999

00:19:27.338 --> 00:19:29.462 of the five that was actually

NOTE Confidence: 0.784771299999999

00:19:29.462 --> 00:19:31.757 the very first paper ever wrote

NOTE Confidence: 0.784771299999999

00:19:31.757 --> 00:19:33.777 was about CF carrier testing.

NOTE Confidence: 0.784771299999999

00:19:33.780 --> 00:19:36.716 Over the next decade by as CF terror

NOTE Confidence: 0.784771299999999

00:19:36.716 --> 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 --> 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 --> 00:19:51.823 how to do CF carrier testing better.

NOTE Confidence: 0.784771299999999

00:19:51.830 --> 00:19:55.043 One of the concerns I had about CF carrier

NOTE Confidence: 0.784771299999999

00:19:55.043 --> 00:19:57.398 testing as quickly as a pulmonologist

NOTE Confidence: 0.784771299999999

00:19:57.398 --> 00:20:00.258 is that it wasn't really clear to me.

NOTE Confidence: 0.784771299999999

00:20:00.260 --> 00:20:02.794 What one should do with that information,
NOTE Confidence: 0.7847712999999999

00:20:02.800 --> 00:20:04.248 particularly as a disease
NOTE Confidence: 0.7847712999999999

00:20:04.248 --> 00:20:05.334 like cystic fibrosis,
NOTE Confidence: 0.7847712999999999

00:20:05.340 --> 00:20:07.145 was evolving clinically and I
NOTE Confidence: 0.7847712999999999

00:20:07.145 --> 00:20:09.340 was worried that when this went,
NOTE Confidence: 0.7847712999999999

00:20:09.340 --> 00:20:11.385 information about cystic fibrosis would
NOTE Confidence: 0.7847712999999999

00:20:11.385 --> 00:20:13.863 be presented to individuals who are
NOTE Confidence: 0.7847712999999999

00:20:13.863 --> 00:20:16.107 seeking carrier testing that both the
NOTE Confidence: 0.7847712999999999

00:20:16.107 --> 00:20:18.170 obstetricians who might be doing the
NOTE Confidence: 0.7847712999999999

00:20:18.170 --> 00:20:19.928 testing and the patients might have
NOTE Confidence: 0.7847712999999999

00:20:19.928 --> 00:20:22.396 no idea of what cystic fibrosis was.
NOTE Confidence: 0.7847712999999999

00:20:22.400 --> 00:20:24.326 I actually reflected back to when
NOTE Confidence: 0.7847712999999999

00:20:24.326 --> 00:20:26.918 I was a medical student and I
NOTE Confidence: 0.7847712999999999

00:20:26.918 --> 00:20:28.566 thought about cystic fibrosis,
NOTE Confidence: 0.7847712999999999

00:20:28.570 --> 00:20:30.700 multiple sclerosis and muscular dystrophy.
NOTE Confidence: 0.7847712999999999

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

NOTE Confidence: 0.784771299999999

00:20:32.320 --> 00:20:33.616 which one is which.

NOTE Confidence: 0.784771299999999

00:20:33.620 --> 00:20:35.402 These are really confusing words and

NOTE Confidence: 0.784771299999999

00:20:35.402 --> 00:20:37.535 I can't tell which of these diseases

NOTE Confidence: 0.784771299999999

00:20:37.535 --> 00:20:39.614 means which thing 'cause so I worried

NOTE Confidence: 0.784771299999999

00:20:39.668 --> 00:20:41.870 very much a cystic fibrosis carrier

NOTE Confidence: 0.784771299999999

00:20:41.870 --> 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 --> 00:20:47.226 It could also be done poorly and

NOTE Confidence: 0.784771299999999

00:20:47.226 --> 00:20:49.687 one of the places in my opinion that

NOTE Confidence: 0.784771299999999

00:20:49.687 --> 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 --> 00:20:55.605 where they were routinely offering

NOTE Confidence: 0.784771299999999

00:20:55.605 --> 00:20:57.893 this without a whole lot of discussion

NOTE Confidence: 0.784771299999999

00:20:57.893 --> 00:20:59.855 without a whole lot of information,

NOTE Confidence: 0.784771299999999

00:20:59.860 --> 00:21:02.086 and they demonstrated in that setting.

NOTE Confidence: 0.784771299999999

00:21:02.090 --> 00:21:03.966 But there are lots of people who
NOTE Confidence: 0.7847712999999999

00:21:03.966 --> 00:21:05.706 were interested in it and that
NOTE Confidence: 0.7847712999999999

00:21:05.706 --> 00:21:06.878 didn't surprise me because.
NOTE Confidence: 0.7847712999999999

00:21:06.880 --> 00:21:07.058 Yeah,
NOTE Confidence: 0.7847712999999999

00:21:07.058 --> 00:21:08.482 if you present it in a way that
NOTE Confidence: 0.7847712999999999

00:21:08.482 --> 00:21:09.949 makes it sound very straightforward,
NOTE Confidence: 0.7847712999999999

00:21:09.950 --> 00:21:11.960 of course people will say yes.
NOTE Confidence: 0.7847712999999999

00:21:11.960 --> 00:21:14.320 So why am I telling you that background
NOTE Confidence: 0.7847712999999999

00:21:14.320 --> 00:21:16.867 now and in relationship to this paper?
NOTE Confidence: 0.7847712999999999

00:21:16.870 --> 00:21:17.135 Wells,
NOTE Confidence: 0.7847712999999999

00:21:17.135 --> 00:21:19.520 because the about in 2013 I got a phone
NOTE Confidence: 0.7847712999999999

00:21:19.582 --> 00:21:21.427 call from a genetic epidemiologist
NOTE Confidence: 0.7847712999999999

00:21:21.427 --> 00:21:24.001 named Katrina Goddard whom we end up
NOTE Confidence: 0.7847712999999999

00:21:24.001 --> 00:21:26.005 becoming Kopi eyes with this large
NOTE Confidence: 0.7847712999999999

00:21:26.005 --> 00:21:27.868 project together that was looking at
NOTE Confidence: 0.7847712999999999

00:21:27.868 --> 00:21:29.353 doing genomic based carrier testing

NOTE Confidence: 0.7847712999999999
00:21:29.353 --> 00:21:31.474 for people who are already seeking
NOTE Confidence: 0.7847712999999999
00:21:31.474 --> 00:21:32.886 cystic fibrosis carrier testing.
NOTE Confidence: 0.7847712999999999
00:21:32.890 --> 00:21:35.506 The first thing I said to Katrina when
NOTE Confidence: 0.7847712999999999
00:21:35.506 --> 00:21:38.115 we first spoke on the phone is that,
NOTE Confidence: 0.7847712999999999
00:21:38.120 --> 00:21:38.802 you know,
NOTE Confidence: 0.7847712999999999
00:21:38.802 --> 00:21:41.530 Are you sure you want me to work
NOTE Confidence: 0.7847712999999999
00:21:41.611 --> 00:21:42.910 on this project?
NOTE Confidence: 0.7847712999999999
00:21:42.910 --> 00:21:46.760 I've been pretty sceptical of.
NOTE Confidence: 0.7847712999999999
00:21:46.760 --> 00:21:47.904 Cystic fibrosis carrier testing,
NOTE Confidence: 0.7847712999999999
00:21:47.904 --> 00:21:48.476 particularly Kaiser,
NOTE Confidence: 0.7847712999999999
00:21:48.480 --> 00:21:50.489 and she continue to work at Kaiser
NOTE Confidence: 0.7847712999999999
00:21:50.489 --> 00:21:52.211 in an Oregon, which is yeah,
NOTE Confidence: 0.7847712999999999
00:21:52.211 --> 00:21:53.646 I know that in fact,
NOTE Confidence: 0.7847712999999999
00:21:53.650 --> 00:21:55.282 that's actually why I want you
NOTE Confidence: 0.7847712999999999
00:21:55.282 --> 00:21:57.090 to help me with this study.
NOTE Confidence: 0.7847712999999999

00:21:57.090 --> 00:21:59.288 I want to make sure we're doing
NOTE Confidence: 0.7847712999999999

00:21:59.288 --> 00:22:01.298 it ethically and I want to do
NOTE Confidence: 0.7847712999999999

00:22:01.298 --> 00:22:03.120 this the very best way we can.
NOTE Confidence: 0.7847712999999999

00:22:03.120 --> 00:22:03.792 So we were.
NOTE Confidence: 0.7847712999999999

00:22:03.792 --> 00:22:06.140 We wrote a grant and we were became part
NOTE Confidence: 0.7847712999999999

00:22:06.140 --> 00:22:08.288 of what's called the Caesar Consortium,
NOTE Confidence: 0.7847712999999999

00:22:08.290 --> 00:22:09.856 which stands for the stands for
NOTE Confidence: 0.7847712999999999

00:22:09.856 --> 00:22:10.900 the clinical sequencing exploratory
NOTE Confidence: 0.7847712999999999

00:22:10.948 --> 00:22:11.730 Research Consortium,
NOTE Confidence: 0.7847712999999999

00:22:11.730 --> 00:22:13.380 which were group of studies that
NOTE Confidence: 0.7847712999999999

00:22:13.380 --> 00:22:15.169 we're trying to address questions of.
NOTE Confidence: 0.8142179

00:22:15.170 --> 00:22:18.070 How is it that?
NOTE Confidence: 0.8142179

00:22:18.070 --> 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 --> 00:22:23.398 What is the evidence?
NOTE Confidence: 0.8142179

00:22:23.398 --> 00:22:26.040 We need to determine how to do this,

NOTE Confidence: 0.8142179

00:22:26.040 --> 00:22:28.929 and so we wrote a grant regarding how to

NOTE Confidence: 0.8142179

00:22:28.929 --> 00:22:32.070 do this for cystic for carrier testing

NOTE Confidence: 0.8142179

00:22:32.070 --> 00:22:34.963 using sequencing and what I want to

NOTE Confidence: 0.8142179

00:22:34.963 --> 00:22:37.266 do is share with you briefly some.

NOTE Confidence: 0.8142179

00:22:37.270 --> 00:22:39.503 Some of the key things we learn

NOTE Confidence: 0.8142179

00:22:39.503 --> 00:22:41.919 from this study and those key points

NOTE Confidence: 0.8142179

00:22:41.919 --> 00:22:44.469 are based upon this paper that was

NOTE Confidence: 0.8142179

00:22:44.469 --> 00:22:46.857 published in 2018 and Health Affairs.

NOTE Confidence: 0.8142179

00:22:46.860 --> 00:22:48.415 Really, the lessons that we

NOTE Confidence: 0.8142179

00:22:48.415 --> 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 --> 00:22:57.505 in a couple of this one slide here.

NOTE Confidence: 0.8142179

00:22:57.510 --> 00:22:59.205 Essentially what happened is that

NOTE Confidence: 0.8142179

00:22:59.205 --> 00:23:01.306 for people who on their own's

NOTE Confidence: 0.8142179

00:23:01.306 --> 00:23:03.340 sought out a clinical carrier test
NOTE Confidence: 0.8142179

00:23:03.340 --> 00:23:04.960 often for cystic fibrosis,
NOTE Confidence: 0.8142179

00:23:04.960 --> 00:23:07.640 we approach them an and.
NOTE Confidence: 0.8142179

00:23:07.640 --> 00:23:09.152 To roll on the study and the
NOTE Confidence: 0.8142179

00:23:09.152 --> 00:23:10.630 way this would work is that if
NOTE Confidence: 0.8142179

00:23:10.630 --> 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 --> 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 --> 00:23:21.101 bunch of surveys for us as well as us
NOTE Confidence: 0.8142179

00:23:21.101 --> 00:23:22.613 evaluating their health utilization
NOTE Confidence: 0.8142179

00:23:22.613 --> 00:23:25.360 within Kaiser over the next year or so.
NOTE Confidence: 0.8142179

00:23:25.360 --> 00:23:27.970 And So what I want to show you here is
NOTE Confidence: 0.8142179

00:23:28.043 --> 00:23:30.395 briefly the findings and the important
NOTE Confidence: 0.8142179

00:23:30.395 --> 00:23:33.409 thing here is that we were not we were.
NOTE Confidence: 0.8142179

00:23:33.410 --> 00:23:34.698 This is actually important

NOTE Confidence: 0.8142179
00:23:34.698 --> 00:23:35.664 about about sequencing.
NOTE Confidence: 0.8142179
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 --> 00:23:40.818 that everything that's there,
NOTE Confidence: 0.8142179
00:23:40.820 --> 00:23:42.878 you have to make a decision.
NOTE Confidence: 0.8142179
00:23:42.880 --> 00:23:45.976 What information do you want to look at?
NOTE Confidence: 0.8142179
00:23:45.980 --> 00:23:48.662 So we had to for this study that was
NOTE Confidence: 0.8142179
00:23:48.662 --> 00:23:51.396 done in collaboration with GAIL Jarvik
NOTE Confidence: 0.8142179
00:23:51.396 --> 00:23:54.215 at the University of Washington is
NOTE Confidence: 0.8142179
00:23:54.215 --> 00:23:56.483 decide how many conditions would we
NOTE Confidence: 0.8142179
00:23:56.483 --> 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 --> 00:24:03.438 567 someplace as many as 100,
NOTE Confidence: 0.8142179
00:24:03.440 --> 00:24:06.051 and for this study we ended up
NOTE Confidence: 0.8142179

00:24:06.051 --> 00:24:07.650 identifying 700 conditions to
NOTE Confidence: 0.8142179

00:24:07.650 --> 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 --> 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 --> 00:24:17.560 point we're being recommended by
NOTE Confidence: 0.8142179

00:24:17.623 --> 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 --> 00:24:22.737 to be highly actionable,
NOTE Confidence: 0.8142179

00:24:22.740 --> 00:24:25.134 i.e things related to cancer or heart
NOTE Confidence: 0.8142179

00:24:25.134 --> 00:24:27.817 disease or might be sort of interventions.
NOTE Confidence: 0.8142179

00:24:27.820 --> 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 --> 00:24:32.784 described to you and have you
NOTE Confidence: 0.8142179

00:24:32.784 --> 00:24:35.371 see how we approach this and to

NOTE Confidence: 0.8142179

00:24:35.371 --> 00:24:37.630 show you what our results were.

NOTE Confidence: 0.8142179

00:24:37.630 --> 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 --> 00:24:43.796 out of 133 women.

NOTE Confidence: 0.8142179

00:24:43.796 --> 00:24:45.616 Who enrolled in the study?

NOTE Confidence: 0.8142179

00:24:45.620 --> 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 --> 00:24:50.990 having a child with an AutoZone.

NOTE Confidence: 0.8142179

00:24:50.990 --> 00:24:52.714 Will recessive condition and

NOTE Confidence: 0.8142179

00:24:52.714 --> 00:24:54.869 three who had excellent conditions

NOTE Confidence: 0.8142179

00:24:54.869 --> 00:24:57.342 for which they were also risk of

NOTE Confidence: 0.8142179

00:24:57.342 --> 00:24:59.500 having a child because it only one.

NOTE Confidence: 0.8142179

00:24:59.500 --> 00:25:02.188 Up only the one that was necessary to

NOTE Confidence: 0.8142179

00:25:02.188 --> 00:25:05.585 be a carrier for them to have a child.

NOTE Confidence: 0.8142179

00:25:05.590 --> 00:25:07.726 We also saw several secondary conditions,

NOTE Confidence: 0.8142179

00:25:07.730 --> 00:25:09.520 including actually 7-7 people had
NOTE Confidence: 0.8142179

00:25:09.520 --> 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 --> 00:25:14.210 scope of what
NOTE Confidence: 0.8073976

00:25:14.298 --> 00:25:18.074 you can learn from sequencing in the context.
NOTE Confidence: 0.8073976

00:25:18.080 --> 00:25:20.824 So we're trying to do in this
NOTE Confidence: 0.8073976

00:25:20.824 --> 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 --> 00:25:27.460 so again, we offered 728 condition
NOTE Confidence: 0.8073976

00:25:27.460 --> 00:25:29.504 gene condition pairs that we
NOTE Confidence: 0.8073976

00:25:29.504 --> 00:25:31.328 categorized into five categories,
NOTE Confidence: 0.8073976

00:25:31.330 --> 00:25:34.018 and I'll say more about the
NOTE Confidence: 0.8073976

00:25:34.018 --> 00:25:36.440 categories on the next slide.
NOTE Confidence: 0.8073976

00:25:36.440 --> 00:25:38.281 We gave people the option of which
NOTE Confidence: 0.8073976

00:25:38.281 --> 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 --> 00:25:42.450 although many of them also value

NOTE Confidence: 0.8073976

00:25:42.495 --> 00:25:43.825 the fact that they're giving

NOTE Confidence: 0.8073976

00:25:43.825 --> 00:25:44.889 choices between the categories,

NOTE Confidence: 0.8073976

00:25:44.890 --> 00:25:46.938 and I'll say more about the categories again,

NOTE Confidence: 0.8073976

00:25:46.940 --> 00:25:49.600 in a few moments.

NOTE Confidence: 0.8073976

00:25:49.600 --> 00:25:51.940 We also learned that if we

NOTE Confidence: 0.8073976

00:25:51.940 --> 00:25:53.500 did this that essentially.

NOTE Confidence: 0.8073976

00:25:53.500 --> 00:25:54.940 Of the individuals who enrolled,

NOTE Confidence: 0.8073976

00:25:54.940 --> 00:25:56.656 78% had at least one finding,

NOTE Confidence: 0.8073976

00:25:56.660 --> 00:25:58.090 which meant that if you

NOTE Confidence: 0.8073976

00:25:58.090 --> 00:25:59.234 test for enough things,

NOTE Confidence: 0.8073976

00:25:59.240 --> 00:26:00.956 everybody will have a positive result,

NOTE Confidence: 0.8073976

00:26:00.960 --> 00:26:02.380 and so this clearly has

NOTE Confidence: 0.8073976

00:26:02.380 --> 00:26:03.516 implications for the workforce

NOTE Confidence: 0.8073976

00:26:03.516 --> 00:26:04.988 capacity of genetic counselors to

NOTE Confidence: 0.8073976

00:26:04.988 --> 00:26:06.413 explain what these results mean.

NOTE Confidence: 0.8073976

00:26:06.420 --> 00:26:08.415 And we found that in our study,

NOTE Confidence: 0.8073976

00:26:08.420 --> 00:26:10.296 the amount of time it took to

NOTE Confidence: 0.8073976

00:26:10.296 --> 00:26:12.160 explain this was about 50 minutes,

NOTE Confidence: 0.8073976

00:26:12.160 --> 00:26:13.595 and for more uncommon conditions

NOTE Confidence: 0.8073976

00:26:13.595 --> 00:26:15.030 around 8 around 84 minutes.

NOTE Confidence: 0.8073976

00:26:15.030 --> 00:26:17.318 So if this was to be a nationally,

NOTE Confidence: 0.8073976

00:26:17.320 --> 00:26:19.588 this will be a huge amount of

NOTE Confidence: 0.8073976

00:26:19.588 --> 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 --> 00:26:26.856 We look carefully at both benefits and harms.

NOTE Confidence: 0.8073976

00:26:26.860 --> 00:26:28.996 I think one of the things I learned

NOTE Confidence: 0.8073976

00:26:28.996 --> 00:26:30.969 along time ago was the importance

NOTE Confidence: 0.8073976

00:26:30.969 --> 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 --> 00:26:36.365 benefits and risks are of

NOTE Confidence: 0.8073976

00:26:36.365 --> 00:26:37.316 a particular invention.

NOTE Confidence: 0.8073976

00:26:37.320 --> 00:26:39.147 And so we looked to see whether

NOTE Confidence: 0.8073976

00:26:39.147 --> 00:26:41.154 or not there's any long term

NOTE Confidence: 0.8073976

00:26:41.154 --> 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 --> 00:26:44.782 whether there's any increased

NOTE Confidence: 0.8073976

00:26:44.782 --> 00:26:46.510 use of mental health services,

NOTE Confidence: 0.8073976

00:26:46.510 --> 00:26:48.729 and we didn't find any of that.

NOTE Confidence: 0.8073976

00:26:48.730 --> 00:26:50.641 We did find in a few specific

NOTE Confidence: 0.8073976

00:26:50.641 --> 00:26:51.888 cases some confusion between

NOTE Confidence: 0.8073976

00:26:51.888 --> 00:26:53.718 the carrier results and its

NOTE Confidence: 0.8073976

00:26:53.718 --> 00:26:55.182 implications for chromosome oh.

NOTE Confidence: 0.8073976

00:26:55.190 --> 00:26:57.308 Conditions which were not included here,

NOTE Confidence: 0.8073976

00:26:57.310 --> 00:27:00.496 so not not, I would say it's not surprising,

NOTE Confidence: 0.8073976

00:27:00.500 --> 00:27:02.675 but not everybody fully understood

NOTE Confidence: 0.8073976

00:27:02.675 --> 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 --> 00:27:10.833 and another benefit is that we did
NOTE Confidence: 0.8073976

00:27:10.833 --> 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 --> 00:27:15.618 had secondary findings at which
NOTE Confidence: 0.8073976

00:27:15.618 --> 00:27:17.554 put them at risk of having
NOTE Confidence: 0.8073976

00:27:17.554 --> 00:27:19.054 medical problems in the future.
NOTE Confidence: 0.8073976

00:27:19.060 --> 00:27:21.412 So this was also seen by us as
NOTE Confidence: 0.8073976

00:27:21.412 --> 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 --> 00:27:28.910 their future risk of a condition.
NOTE Confidence: 0.8073976

00:27:28.910 --> 00:27:30.968 In fact, there was a couple.
NOTE Confidence: 0.8073976

00:27:30.970 --> 00:27:34.466 There's one instance in particular where a A.
NOTE Confidence: 0.8073976

00:27:34.470 --> 00:27:36.510 Mother was identified as a carrier

NOTE Confidence: 0.8073976

00:27:36.510 --> 00:27:38.655 for hemophilia and based upon that

NOTE Confidence: 0.8073976

00:27:38.655 --> 00:27:40.470 the perinatal management of her

NOTE Confidence: 0.8073976

00:27:40.470 --> 00:27:42.345 pregnancy was changed so she was

NOTE Confidence: 0.8073976

00:27:42.345 --> 00:27:44.331 able to deliver in a place that was

NOTE Confidence: 0.8073976

00:27:44.331 --> 00:27:46.077 prepared for the possibility that she

NOTE Confidence: 0.8073976

00:27:46.077 --> 00:27:48.189 might have a child with hemophilia,

NOTE Confidence: 0.8073976

00:27:48.190 --> 00:27:50.129 which in fact when she ended up

NOTE Confidence: 0.8073976

00:27:50.129 --> 00:27:52.132 having and the child actually ended

NOTE Confidence: 0.8073976

00:27:52.132 --> 00:27:53.972 up having a subdural hemorrhage

NOTE Confidence: 0.8073976

00:27:53.972 --> 00:27:56.156 within a couple of days of life.

NOTE Confidence: 0.8073976

00:27:56.160 --> 00:27:58.068 But because it was known what

NOTE Confidence: 0.8073976

00:27:58.068 --> 00:27:59.022 the problem was,

NOTE Confidence: 0.8073976

00:27:59.030 --> 00:28:02.648 they were able to intervene appropriately.

NOTE Confidence: 0.8073976

00:28:02.650 --> 00:28:05.492 I want to share with you briefly

NOTE Confidence: 0.8073976

00:28:05.492 --> 00:28:06.710 some information regarding

NOTE Confidence: 0.869017

00:28:06.779 --> 00:28:09.621 the five categories that we used to
NOTE Confidence: 0.869017

00:28:09.621 --> 00:28:11.597 differentiate the 78728 conditions and
NOTE Confidence: 0.869017

00:28:11.597 --> 00:28:14.758 when I also share with you that the process
NOTE Confidence: 0.869017

00:28:14.758 --> 00:28:17.614 of coming up with these categories took
NOTE Confidence: 0.869017

00:28:17.614 --> 00:28:20.199 us literally about a year and another,
NOTE Confidence: 0.869017

00:28:20.200 --> 00:28:22.552 we spent a lot of time thinking how
NOTE Confidence: 0.869017

00:28:22.552 --> 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 --> 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 --> 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 --> 00:28:39.020 categories based upon severity, either
NOTE Confidence: 0.869017

00:28:39.020 --> 00:28:41.840 those with a significant shorten lifespan.
NOTE Confidence: 0.869017

00:28:41.840 --> 00:28:43.044 Those that were serious.
NOTE Confidence: 0.869017

00:28:43.044 --> 00:28:46.211 And those that were mild and you can see here

NOTE Confidence: 0.869017
00:28:46.211 --> 00:28:48.155 the definitions of what those categories
NOTE Confidence: 0.869017
00:28:48.218 --> 00:28:50.610 were in terms of the level of seriousness.
NOTE Confidence: 0.869017
00:28:50.610 --> 00:28:51.714 We spent many iterations
NOTE Confidence: 0.869017
00:28:51.714 --> 00:28:53.094 trying to think through this.
NOTE Confidence: 0.869017
00:28:53.100 --> 00:28:54.200 How to organize this?
NOTE Confidence: 0.869017
00:28:54.200 --> 00:28:56.424 And then of course we had to do
NOTE Confidence: 0.869017
00:28:56.424 --> 00:28:58.223 is place all of the categories all
NOTE Confidence: 0.869017
00:28:58.223 --> 00:29:00.357 the diseases into these categories.
NOTE Confidence: 0.869017
00:29:00.360 --> 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 --> 00:29:05.120 So for example,
NOTE Confidence: 0.869017
00:29:05.120 --> 00:29:07.840 there are some conditions in which you, they.
NOTE Confidence: 0.869017
00:29:07.840 --> 00:29:09.540 Some may be very mild,
NOTE Confidence: 0.869017
00:29:09.540 --> 00:29:11.240 some may be very serious,
NOTE Confidence: 0.869017
00:29:11.240 --> 00:29:12.260 including life threatening.
NOTE Confidence: 0.869017

00:29:12.260 --> 00:29:14.640 So we decided to call that out.
NOTE Confidence: 0.869017

00:29:14.640 --> 00:29:16.680 We also called out those conditions
NOTE Confidence: 0.869017

00:29:16.680 --> 00:29:18.040 that begin as adults.
NOTE Confidence: 0.869017

00:29:18.040 --> 00:29:19.105 So for example,
NOTE Confidence: 0.869017

00:29:19.105 --> 00:29:21.235 children who have no symptoms during
NOTE Confidence: 0.869017

00:29:21.235 --> 00:29:23.478 childhood but may have problems as adults,
NOTE Confidence: 0.869017

00:29:23.480 --> 00:29:25.105 and these could be behavioral
NOTE Confidence: 0.869017

00:29:25.105 --> 00:29:27.143 that could be related to hearing
NOTE Confidence: 0.869017

00:29:27.143 --> 00:29:29.255 they could be related to medical.
NOTE Confidence: 0.869017

00:29:29.260 --> 00:29:29.992 So for example.
NOTE Confidence: 0.869017

00:29:29.992 --> 00:29:31.700 We you know I mentioned to you
NOTE Confidence: 0.869017

00:29:31.755 --> 00:29:33.789 that some of the secondary findings
NOTE Confidence: 0.869017

00:29:33.789 --> 00:29:35.566 related to hereditary cancer syndromes
NOTE Confidence: 0.869017

00:29:35.566 --> 00:29:37.010 that occur in adulthood.
NOTE Confidence: 0.869017

00:29:37.010 --> 00:29:39.034 So the question was and and the idea
NOTE Confidence: 0.869017

00:29:39.034 --> 00:29:40.891 here is that these are categories

NOTE Confidence: 0.869017
00:29:40.891 --> 00:29:42.823 that they could be testing for
NOTE Confidence: 0.869017
00:29:42.888 --> 00:29:44.040 as well in children,
NOTE Confidence: 0.869017
00:29:44.040 --> 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 --> 00:29:54.843 the actual results that we actually
NOTE Confidence: 0.9021238
00:29:54.843 --> 00:29:56.847 found in these in these couples,
NOTE Confidence: 0.9021238
00:29:56.850 --> 00:30:00.260 and So what you can see here is that this is.
NOTE Confidence: 0.9021238
00:30:00.260 --> 00:30:02.540 On the on the the high risk couples
NOTE Confidence: 0.9021238
00:30:02.540 --> 00:30:04.580 had the following conditions.
NOTE Confidence: 0.8571314
00:30:07.030 --> 00:30:08.836 In which two people were carers.
NOTE Confidence: 0.8571314
00:30:08.840 --> 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 --> 00:30:15.210 which is a condition that is not routinely
NOTE Confidence: 0.8571314
00:30:15.210 --> 00:30:17.109 tested for in carrier testing because
NOTE Confidence: 0.8571314
00:30:17.109 --> 00:30:19.065 of its treat ability and variability.
NOTE Confidence: 0.8571314

00:30:19.070 --> 00:30:21.177 That was actually the most common one.

NOTE Confidence: 0.8571314

00:30:21.180 --> 00:30:23.520 The and then you can see some of the

NOTE Confidence: 0.8571314

00:30:23.520 --> 00:30:25.191 examples of the individual secondary

NOTE Confidence: 0.8571314

00:30:25.191 --> 00:30:27.500 findings as well that we learn now.

NOTE Confidence: 0.8571314

00:30:27.500 --> 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 --> 00:30:34.418 what the impact of this would be.

NOTE Confidence: 0.8571314

00:30:34.420 --> 00:30:36.625 And so the idea was to learn.

NOTE Confidence: 0.8571314

00:30:36.630 --> 00:30:38.646 About what the outcome and impact would be

NOTE Confidence: 0.8571314

00:30:38.646 --> 00:30:40.710 of testing for all these things, and so,

NOTE Confidence: 0.8571314

00:30:40.710 --> 00:30:42.495 therefore we decided in a research setting,

NOTE Confidence: 0.8571314

00:30:42.500 --> 00:30:44.900 it was reasonable to be more broad than

NOTE Confidence: 0.8571314

00:30:44.900 --> 00:30:47.115 we otherwise might be doing if we were

NOTE Confidence: 0.8571314

00:30:47.115 --> 00:30:49.200 providing this as a clinical service.

NOTE Confidence: 0.8571314

00:30:49.200 --> 00:30:52.398 So I want to point out.

NOTE Confidence: 0.8571314

00:30:52.400 --> 00:30:55.124 To make a couple comments about

NOTE Confidence: 0.8571314

00:30:55.124 --> 00:30:57.510 the implications of this study.

NOTE Confidence: 0.8571314

00:30:57.510 --> 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 --> 00:31:05.066 we got limited data on the impact of

NOTE Confidence: 0.8571314

00:31:05.066 --> 00:31:07.398 couples who had more than a 25% chance

NOTE Confidence: 0.8571314

00:31:07.398 --> 00:31:09.246 of having a condition that child.

NOTE Confidence: 0.8571314

00:31:09.250 --> 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 --> 00:31:14.074 because this was on people who initially

NOTE Confidence: 0.8571314

00:31:14.074 --> 00:31:16.358 sought out on their own carrier testing,

NOTE Confidence: 0.8571314

00:31:16.360 --> 00:31:18.100 it didn't include people who were

NOTE Confidence: 0.8571314

00:31:18.100 --> 00:31:20.179 less likely to value carrier testing,

NOTE Confidence: 0.8571314

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

NOTE Confidence: 0.8571314

00:31:22.452 --> 00:31:24.457 was well received really was really

NOTE Confidence: 0.8571314

00:31:24.457 --> 00:31:26.880 based upon who was selected for this.

NOTE Confidence: 0.8571314

00:31:26.880 --> 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 --> 00:31:32.940 careful with who we including.
NOTE Confidence: 0.8571314

00:31:32.940 --> 00:31:35.390 The study might have limited
NOTE Confidence: 0.8571314

00:31:35.390 --> 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 --> 00:31:41.308 one of the challenges
NOTE Confidence: 0.8571314

00:31:41.308 --> 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 --> 00:31:49.273 testing is how to make the test available
NOTE Confidence: 0.8380644

00:31:49.273 --> 00:31:51.128 to those who would value it while
NOTE Confidence: 0.8380644

00:31:51.180 --> 00:31:53.046 avoiding routinely offering in a way
NOTE Confidence: 0.8380644

00:31:53.046 --> 00:31:55.172 that influences those to get tested who
NOTE Confidence: 0.8380644

00:31:55.172 --> 00:31:57.230 might otherwise not value it to meet.
NOTE Confidence: 0.8380644

00:31:57.230 --> 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,

NOTE Confidence: 0.8380644

00:32:01.930 --> 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 --> 00:32:09.280 but they don't really quite

NOTE Confidence: 0.8380644

00:32:09.280 --> 00:32:10.456 know what they're getting.

NOTE Confidence: 0.8380644

00:32:10.460 --> 00:32:11.548 Into the other hand,

NOTE Confidence: 0.8380644

00:32:11.548 --> 00:32:13.865 there are some people for whom this is

NOTE Confidence: 0.8380644

00:32:13.865 --> 00:32:15.743 really meaningful and really want this,

NOTE Confidence: 0.8380644

00:32:15.750 --> 00:32:17.610 but I think this is really.

NOTE Confidence: 0.8380644

00:32:17.610 --> 00:32:21.756 Attention. The other thing that this.

NOTE Confidence: 0.8380644

00:32:21.760 --> 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 --> 00:32:27.893 is that the value and importance of

NOTE Confidence: 0.8380644

00:32:27.893 --> 00:32:29.963 carrier testing is because of its

NOTE Confidence: 0.8380644

00:32:30.033 --> 00:32:32.123 personal value to the individuals
NOTE Confidence: 0.8380644

00:32:32.123 --> 00:32:34.213 about themselves about their lives,
NOTE Confidence: 0.8380644

00:32:34.220 --> 00:32:37.484 about their children and how do we think
NOTE Confidence: 0.8380644

00:32:37.484 --> 00:32:40.380 about this type of reason for doing?
NOTE Confidence: 0.8380644

00:32:40.380 --> 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 --> 00:32:44.351 we're trying to figure out how we
NOTE Confidence: 0.8380644

00:32:44.351 --> 00:32:45.970 should prioritize our health resource
NOTE Confidence: 0.8380644

00:32:45.970 --> 00:32:48.720 and you'll see this issue will come up
NOTE Confidence: 0.8380644

00:32:48.720 --> 00:32:52.059 again as we talk about the next project.
NOTE Confidence: 0.8380644

00:32:52.060 --> 00:32:53.467 Before we talk about the next project,
NOTE Confidence: 0.8380644

00:32:53.470 --> 00:32:55.787 I want one of the things I
NOTE Confidence: 0.8380644

00:32:55.787 --> 00:32:57.419 want to mention to you.
NOTE Confidence: 0.8380644

00:32:57.420 --> 00:32:59.316 Is that I mentioned you are study was
NOTE Confidence: 0.8380644

00:32:59.316 --> 00:33:01.563 part of a larger consortium of nine
NOTE Confidence: 0.8380644

00:33:01.563 --> 00:33:03.278 studies called the Caesar Consortium,

NOTE Confidence: 0.8380644

00:33:03.280 --> 00:33:05.616 and so one of the things that happens.

NOTE Confidence: 0.8380644

00:33:05.620 --> 00:33:07.388 Part of this consortium is that we all

NOTE Confidence: 0.8380644

00:33:07.388 --> 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 --> 00:33:13.811 and so one of the projects that I got

NOTE Confidence: 0.8380644

00:33:13.811 --> 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 --> 00:33:20.794 is part of the consortium was to think

NOTE Confidence: 0.8380644

00:33:20.794 --> 00:33:22.654 about the questions of consent for

NOTE Confidence: 0.8380644

00:33:22.654 --> 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 --> 00:33:28.000 the intention was to learn.

NOTE Confidence: 0.8380644

00:33:28.000 --> 00:33:30.527 How do we do this better clinically?

NOTE Confidence: 0.8380644

00:33:30.530 --> 00:33:32.294 So a number of us got together

NOTE Confidence: 0.8380644

00:33:32.294 --> 00:33:34.477 to write a manuscript about the

NOTE Confidence: 0.8380644

00:33:34.477 --> 00:33:36.297 considerations for clinical sequencing
NOTE Confidence: 0.8380644

00:33:36.297 --> 00:33:38.830 based upon all of our experience,
NOTE Confidence: 0.8380644

00:33:38.830 --> 00:33:41.798 and I want to share with you briefly
NOTE Confidence: 0.8380644

00:33:41.798 --> 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 --> 00:33:49.203 and the idea of these considerations are
NOTE Confidence: 0.8380644

00:33:49.203 --> 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 --> 00:33:56.739 The first thing to remember is that
NOTE Confidence: 0.8380644

00:33:56.739 --> 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 --> 00:34:03.390 whether it's clinical decision making better,
NOTE Confidence: 0.8380644

00:34:03.390 --> 00:34:04.088 better pregnancy,
NOTE Confidence: 0.8380644

00:34:04.088 --> 00:34:05.833 whether that's a clinical decision
NOTE Confidence: 0.8380644

00:34:05.833 --> 00:34:07.420 making about cancer treatment.
NOTE Confidence: 0.8380644

00:34:07.420 --> 00:34:09.250 It's about some other question

NOTE Confidence: 0.8380644

00:34:09.250 --> 00:34:10.348 that's coming up,

NOTE Confidence: 0.8380644

00:34:10.350 --> 00:34:12.540 which is why clinical sequence is

NOTE Confidence: 0.8380644

00:34:12.540 --> 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 --> 00:34:18.227 currently why sequencing is done

NOTE Confidence: 0.8380644

00:34:18.227 --> 00:34:20.780 clinically is in the context of cancer.

NOTE Confidence: 0.8380644

00:34:20.780 --> 00:34:22.620 An evaluation of cancer treatments.

NOTE Confidence: 0.8380644

00:34:22.620 --> 00:34:24.455 Another reason is for children

NOTE Confidence: 0.8380644

00:34:24.455 --> 00:34:25.556 with developmental delay.

NOTE Confidence: 0.8380644

00:34:25.560 --> 00:34:27.400 An evaluation of developmental delay.

NOTE Confidence: 0.8380644

00:34:27.400 --> 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 --> 00:34:34.795 information is appropriate to disclose.

NOTE Confidence: 0.81924385

00:34:34.800 --> 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 --> 00:34:41.812 is that the process needs to be
NOTE Confidence: 0.81924385

00:34:41.812 --> 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 --> 00:34:46.622 'cause typically and actually three,
NOTE Confidence: 0.81924385

00:34:46.630 --> 00:34:49.267 is related to that too, and the converse,
NOTE Confidence: 0.81924385

00:34:49.267 --> 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 --> 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 --> 00:34:59.521 but that may not be as necessary as knowing
NOTE Confidence: 0.81924385

00:34:59.521 --> 00:35:01.765 what's important to the individual person.
NOTE Confidence: 0.81924385

00:35:01.770 --> 00:35:04.549 So the analogy I like to give
NOTE Confidence: 0.81924385

00:35:04.549 --> 00:35:07.068 is that when we do an MRI.
NOTE Confidence: 0.81924385

00:35:07.070 --> 00:35:09.272 The consent process does not usually
NOTE Confidence: 0.81924385

00:35:09.272 --> 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,

NOTE Confidence: 0.81924385

00:35:13.170 --> 00:35:16.599 which is the basis of how MRI is done.

NOTE Confidence: 0.81924385

00:35:16.600 --> 00:35:19.260 But yet when we do genetic testing,

NOTE Confidence: 0.81924385

00:35:19.260 --> 00:35:23.628 we often think it's important to know.

NOTE Confidence: 0.81924385

00:35:23.630 --> 00:35:24.830 What base pairs are,

NOTE Confidence: 0.81924385

00:35:24.830 --> 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 --> 00:35:31.104 but are base pairs necessary to think

NOTE Confidence: 0.81924385

00:35:31.104 --> 00:35:33.910 about the implications of genetic testing?

NOTE Confidence: 0.81924385

00:35:33.910 --> 00:35:35.998 The other broad set of considerations

NOTE Confidence: 0.81924385

00:35:35.998 --> 00:35:38.391 for consent that we came up with

NOTE Confidence: 0.81924385

00:35:38.391 --> 00:35:40.568 collectively was that we have to think

NOTE Confidence: 0.81924385

00:35:40.638 --> 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 --> 00:35:50.060 including the limitations of interpretations,

NOTE Confidence: 0.81924385

00:35:50.060 --> 00:35:52.466 so that the expectations are reasonable

NOTE Confidence: 0.81924385

00:35:52.466 --> 00:35:55.690 and we don't have two high expectations.

NOTE Confidence: 0.81924385

00:35:55.690 --> 00:35:57.390 And.

NOTE Confidence: 0.81924385

00:35:57.390 --> 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 --> 00:36:01.385 The decision making process by

NOTE Confidence: 0.81924385

00:36:01.385 --> 00:36:02.790 thinking about the potential clinical

NOTE Confidence: 0.81924385

00:36:02.839 --> 00:36:03.999 and emotional implications for

NOTE Confidence: 0.81924385

00:36:03.999 --> 00:36:05.449 the individual of those results.

NOTE Confidence: 0.81924385

00:36:05.450 --> 00:36:07.421 How will it feel to you if you need

NOTE Confidence: 0.81924385

00:36:07.421 --> 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 --> 00:36:14.960 a problem later in life,

NOTE Confidence: 0.81924385

00:36:14.960 --> 00:36:16.780 so at least getting the clinician and

NOTE Confidence: 0.81924385

00:36:16.780 --> 00:36:18.870 the patient be thinking about that and

NOTE Confidence: 0.81924385

00:36:18.870 --> 00:36:20.445 also thinking about the implications

NOTE Confidence: 0.81924385

00:36:20.445 --> 00:36:22.160 of the results for the family,

NOTE Confidence: 0.81924385

00:36:22.160 --> 00:36:24.176 and being aware that of how they want

NOTE Confidence: 0.81924385

00:36:24.176 --> 00:36:26.476 to deal with that piece of information.

NOTE Confidence: 0.81924385

00:36:26.480 --> 00:36:27.464 So the idea.

NOTE Confidence: 0.81924385

00:36:27.464 --> 00:36:29.432 Was to have these six considerations

NOTE Confidence: 0.81924385

00:36:29.432 --> 00:36:31.906 in mind when thinking about consent.

NOTE Confidence: 0.81924385

00:36:31.910 --> 00:36:34.325 I want to really have us focus

NOTE Confidence: 0.81924385

00:36:34.325 --> 00:36:36.538 on Table 2 for a moment,

NOTE Confidence: 0.81924385

00:36:36.540 --> 00:36:39.403 which has to do with sample examples

NOTE Confidence: 0.81924385

00:36:39.403 --> 00:36:41.541 of questions that really relate

NOTE Confidence: 0.81924385

00:36:41.541 --> 00:36:43.205 back to the second.

NOTE Confidence: 0.81924385

00:36:43.210 --> 00:36:45.426 But can't relate back to number 2 here,

NOTE Confidence: 0.81924385

00:36:45.430 --> 00:36:46.924 which is the consent process should

NOTE Confidence: 0.81924385

00:36:46.924 --> 00:36:48.613 be should be responsive to the

NOTE Confidence: 0.81924385

00:36:48.613 --> 00:36:49.576 individuals personal situation.
NOTE Confidence: 0.81924385

00:36:49.580 --> 00:36:51.519 Context. Well, how do you do that?
NOTE Confidence: 0.81924385

00:36:51.520 --> 00:36:53.165 The idea here is that consent is
NOTE Confidence: 0.81924385

00:36:53.165 --> 00:36:54.840 not about giving people information,
NOTE Confidence: 0.81924385

00:36:54.840 --> 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 --> 00:36:59.325 about themselves to help you decide
NOTE Confidence: 0.81924385

00:36:59.325 --> 00:37:00.939 what information they need to know.
NOTE Confidence: 0.81924385

00:37:00.940 --> 00:37:02.868 And so here's a list of what those
NOTE Confidence: 0.81924385

00:37:02.868 --> 00:37:04.817 questions are fine with their experiences.
NOTE Confidence: 0.81924385

00:37:04.820 --> 00:37:06.200 Then finally what they think
NOTE Confidence: 0.81924385

00:37:06.200 --> 00:37:07.028 the benefits are.
NOTE Confidence: 0.81924385

00:37:07.030 --> 00:37:08.969 Find out questions and concerns they have.
NOTE Confidence: 0.81924385

00:37:08.970 --> 00:37:10.909 Find out maybe they've already made decision.
NOTE Confidence: 0.81924385

00:37:10.910 --> 00:37:13.254 One of the things we've learned is that.
NOTE Confidence: 0.81924385

00:37:13.260 --> 00:37:14.432 Often people when they,

NOTE Confidence: 0.81924385

00:37:14.432 --> 00:37:16.190 when they are engaged with clinician

NOTE Confidence: 0.81924385

00:37:16.241 --> 00:37:17.641 or they have an idea of what

NOTE Confidence: 0.81924385

00:37:17.641 --> 00:37:19.230 they want to do and so starting

NOTE Confidence: 0.81924385

00:37:19.230 --> 00:37:20.634 all over and acting as though

NOTE Confidence: 0.8407035

00:37:20.640 --> 00:37:22.140 they have no idea might not

NOTE Confidence: 0.8407035

00:37:22.140 --> 00:37:23.140 be really reflecting where

NOTE Confidence: 0.8407035

00:37:23.189 --> 00:37:24.575 they are there at that moment.

NOTE Confidence: 0.8407035

00:37:24.580 --> 00:37:26.292 But one of the other points and this

NOTE Confidence: 0.8407035

00:37:26.292 --> 00:37:28.425 is really brought up by many of our

NOTE Confidence: 0.8407035

00:37:28.425 --> 00:37:29.525 genetic counseling colleagues has

NOTE Confidence: 0.8407035

00:37:29.578 --> 00:37:31.482 been the notion of trying to get them

NOTE Confidence: 0.8407035

00:37:31.482 --> 00:37:32.968 to anticipate results and say so.

NOTE Confidence: 0.8407035

00:37:32.968 --> 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 --> 00:37:39.260 And as a way of just getting into sort

NOTE Confidence: 0.8407035

00:37:39.260 --> 00:37:41.248 of walk through weather what this means
NOTE Confidence: 0.8407035

00:37:41.248 --> 00:37:43.528 to them and whether they really do order.
NOTE Confidence: 0.8407035

00:37:43.530 --> 00:37:45.870 Don't want this and why?
NOTE Confidence: 0.8407035

00:37:45.870 --> 00:37:46.659 I want to.
NOTE Confidence: 0.9192076

00:37:50.480 --> 00:37:54.332 Also. Share with you that I'm not
NOTE Confidence: 0.9192076

00:37:54.332 --> 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 --> 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 --> 00:38:03.260 key considerations or key points,
NOTE Confidence: 0.9192076

00:38:03.260 --> 00:38:05.316 and the idea is that these are the
NOTE Confidence: 0.9192076

00:38:05.316 --> 00:38:07.123 key points that need to be made
NOTE Confidence: 0.9192076

00:38:07.123 --> 00:38:08.870 as part of the consent process,
NOTE Confidence: 0.9192076

00:38:08.870 --> 00:38:11.160 and what I want to do here tonight is not
NOTE Confidence: 0.9192076

00:38:11.224 --> 00:38:13.408 going through each of these key points,
NOTE Confidence: 0.9192076

00:38:13.410 --> 00:38:15.237 but point out to you the categories

NOTE Confidence: 0.9192076

00:38:15.237 --> 00:38:16.791 and sub categories are the first

NOTE Confidence: 0.9192076

00:38:16.791 --> 00:38:18.237 had to do with information about

NOTE Confidence: 0.9192076

00:38:18.237 --> 00:38:20.078 the range and limitation of results.

NOTE Confidence: 0.9192076

00:38:20.080 --> 00:38:21.865 So talking about both the types of

NOTE Confidence: 0.9192076

00:38:21.865 --> 00:38:23.410 results available and the limitations.

NOTE Confidence: 0.9192076

00:38:23.410 --> 00:38:25.108 And again, the idea is trying

NOTE Confidence: 0.9192076

00:38:25.108 --> 00:38:26.240 to make this simple.

NOTE Confidence: 0.9192076

00:38:26.240 --> 00:38:27.625 Trying to make this sufficiently

NOTE Confidence: 0.9192076

00:38:27.625 --> 00:38:29.010 straightforward that this can be

NOTE Confidence: 0.9192076

00:38:29.061 --> 00:38:30.765 communicated by anybody to anybody else.

NOTE Confidence: 0.9192076

00:38:30.770 --> 00:38:32.185 This does not require some

NOTE Confidence: 0.9192076

00:38:32.185 --> 00:38:33.600 of the PhD in genetics,

NOTE Confidence: 0.9192076

00:38:33.600 --> 00:38:36.066 so you could communicate these concepts.

NOTE Confidence: 0.9192076

00:38:36.070 --> 00:38:37.924 The second is to think through

NOTE Confidence: 0.9192076

00:38:37.924 --> 00:38:39.680 the implications of the results.

NOTE Confidence: 0.9192076

00:38:39.680 --> 00:38:41.320 Think about the clinical implications
NOTE Confidence: 0.9192076

00:38:41.320 --> 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 --> 00:38:46.002 implications and thinking about the
NOTE Confidence: 0.9192076

00:38:46.002 --> 00:38:47.220 implications for family members.
NOTE Confidence: 0.9192076

00:38:47.220 --> 00:38:47.920 So again,
NOTE Confidence: 0.9192076

00:38:47.920 --> 00:38:50.370 a way of organizing or thinking about
NOTE Confidence: 0.9192076

00:38:50.370 --> 00:38:52.777 what should be the conversation to
NOTE Confidence: 0.9192076

00:38:52.777 --> 00:38:55.240 help with the decision making about.
NOTE Confidence: 0.9192076

00:38:55.240 --> 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 --> 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 --> 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 --> 00:39:11.980 Within Kaiser for people who

NOTE Confidence: 0.9192076

00:39:11.980 --> 00:39:13.660 are having cystic fibrosis,

NOTE Confidence: 0.9192076

00:39:13.660 --> 00:39:16.642 carrier testing resulted in people who

NOTE Confidence: 0.9192076

00:39:16.642 --> 00:39:19.096 were highly educated value genetic

NOTE Confidence: 0.9192076

00:39:19.096 --> 00:39:21.770 testing a lot and were mostly white

NOTE Confidence: 0.9192076

00:39:21.770 --> 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 --> 00:39:29.486 we need to be thinking more

NOTE Confidence: 0.9192076

00:39:29.486 --> 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 --> 00:39:34.620 and in the context that we did that in.

NOTE Confidence: 0.9192076

00:39:34.620 --> 00:39:36.924 So we ended up with a number of

NOTE Confidence: 0.9192076

00:39:36.924 --> 00:39:38.534 collaborators are doing a series

NOTE Confidence: 0.9192076

00:39:38.534 --> 00:39:40.472 of interviews and focus groups at

NOTE Confidence: 0.9192076

00:39:40.472 --> 00:39:42.627 safety at hospitals in the in Oregon.

NOTE Confidence: 0.9192076

00:39:42.630 --> 00:39:43.522 These were.

NOTE Confidence: 0.9192076

00:39:43.522 --> 00:39:45.306 Federally qualified health centers

NOTE Confidence: 0.9192076

00:39:45.306 --> 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 --> 00:39:51.777 out what they thought of a variety

NOTE Confidence: 0.9192076

00:39:51.777 --> 00:39:52.800 of genetic services.

NOTE Confidence: 0.9192076

00:39:52.800 --> 00:39:54.640 The first big point of this paper was

NOTE Confidence: 0.9192076

00:39:54.640 --> 00:39:56.798 that the conditions were highly sceptical.

NOTE Confidence: 0.9192076

00:39:56.800 --> 00:39:58.879 The idea of genetics in this setting

NOTE Confidence: 0.9192076

00:39:58.879 --> 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 --> 00:40:03.380 people have a lot of things to

NOTE Confidence: 0.9192076

00:40:03.380 --> 00:40:05.317 deal with in their lives.

NOTE Confidence: 0.9192076

00:40:05.320 --> 00:40:06.409 They have many,

NOTE Confidence: 0.9192076

00:40:06.409 --> 00:40:08.224 and again typically now this

NOTE Confidence: 0.9192076

00:40:08.224 --> 00:40:10.319 was done by three years ago.

NOTE Confidence: 0.9192076

00:40:10.320 --> 00:40:10.938 But now,

NOTE Confidence: 0.9192076

00:40:10.938 --> 00:40:12.792 with all the focus on systemic

NOTE Confidence: 0.9192076

00:40:12.792 --> 00:40:14.837 racism and thinking about all the

NOTE Confidence: 0.9192076

00:40:14.837 --> 00:40:16.877 various ways that people who are

NOTE Confidence: 0.9192076

00:40:16.944 --> 00:40:18.984 under represented in research are

NOTE Confidence: 0.9192076

00:40:18.984 --> 00:40:21.353 facing huge number of challenges that

NOTE Confidence: 0.9192076

00:40:21.353 --> 00:40:22.968 something like genetics seems like

NOTE Confidence: 0.9192076

00:40:22.968 --> 00:40:24.950 it is like completely unnecessary.

NOTE Confidence: 0.9192076

00:40:24.950 --> 00:40:26.334 Inappropriate when people are

NOTE Confidence: 0.9192076

00:40:26.334 --> 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 --> 00:40:31.598 why would you even think of

NOTE Confidence: 0.9192076

00:40:31.598 --> 00:40:32.777 offering genetic services?

NOTE Confidence: 0.9192076

00:40:32.780 --> 00:40:35.336 We talked to the patient so and they were

NOTE Confidence: 0.9192076

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 --> 00:40:40.600 In fact we are interested in these types of

NOTE Confidence: 0.84534127

00:40:40.660 --> 00:40:41.800 services and in fact,

NOTE Confidence: 0.84534127

00:40:41.800 --> 00:40:42.964 particularly because that these

NOTE Confidence: 0.84534127

00:40:42.964 --> 00:40:44.710 are being offered to other folks.

NOTE Confidence: 0.84534127

00:40:44.710 --> 00:40:46.170 We want these two now.

NOTE Confidence: 0.84534127

00:40:46.170 --> 00:40:48.186 One thing I want to show you

NOTE Confidence: 0.84534127

00:40:48.186 --> 00:40:49.977 and illustrate for you is how

NOTE Confidence: 0.84534127

00:40:49.977 --> 00:40:51.693 do we ask people this question?

NOTE Confidence: 0.84534127

00:40:51.700 --> 00:40:53.428 'cause it's not easy to ask

NOTE Confidence: 0.84534127

00:40:53.428 --> 00:40:54.925 people about genetics when they

NOTE Confidence: 0.84534127

00:40:54.925 --> 00:40:56.350 may be less familiar about.

NOTE Confidence: 0.84534127

00:40:56.350 --> 00:40:59.002 So the approach we took without

NOTE Confidence: 0.84534127

00:40:59.002 --> 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

NOTE Confidence: 0.84534127

00:41:02.370 --> 00:41:04.524 that that had animated stories and

NOTE Confidence: 0.84534127

00:41:04.524 --> 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 --> 00:41:09.885 one was about genetic testing

NOTE Confidence: 0.84534127

00:41:09.885 --> 00:41:10.944 for developmental disabilities,

NOTE Confidence: 0.84534127

00:41:10.950 --> 00:41:12.973 and the third one illustrated here had

NOTE Confidence: 0.84534127

00:41:12.973 --> 00:41:15.549 to do with hereditary cancer syndromes,

NOTE Confidence: 0.84534127

00:41:15.550 --> 00:41:19.645 with the notion that somebody who has.

NOTE Confidence: 0.84534127

00:41:19.650 --> 00:41:22.146 A risk of cancer and the fact that

NOTE Confidence: 0.84534127

00:41:22.146 --> 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 --> 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 --> 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

NOTE Confidence: 0.84023404

00:41:37.664 --> 00:41:39.438 implications might be for her daughters.
NOTE Confidence: 0.84023404

00:41:39.438 --> 00:41:41.652 When we turned out that of
NOTE Confidence: 0.84023404

00:41:41.652 --> 00:41:43.698 the three examples we gave,
NOTE Confidence: 0.84023404

00:41:43.700 --> 00:41:46.580 the one that the conditions were at least
NOTE Confidence: 0.84023404

00:41:46.580 --> 00:41:48.288 plausibly interested in was this one.
NOTE Confidence: 0.84023404

00:41:48.290 --> 00:41:51.233 An while the patient stress and all of them.
NOTE Confidence: 0.84023404

00:41:51.240 --> 00:41:53.536 This also resonated the most with them,
NOTE Confidence: 0.84023404

00:41:53.540 --> 00:41:55.180 in part because they realize
NOTE Confidence: 0.84023404

00:41:55.180 --> 00:41:56.820 that cancer can affect anybody,
NOTE Confidence: 0.84023404

00:41:56.820 --> 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 --> 00:42:01.419 rare and in fact developmental
NOTE Confidence: 0.84023404

00:42:01.419 --> 00:42:03.049 delays don't happen that often.
NOTE Confidence: 0.84023404

00:42:03.050 --> 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 --> 00:42:09.010 so they all could resonate with that.

NOTE Confidence: 0.84023404

00:42:09.010 --> 00:42:13.120 So. And in fact.

NOTE Confidence: 0.84023404

00:42:13.120 --> 00:42:15.365 There are guidelines for both

NOTE Confidence: 0.84023404

00:42:15.365 --> 00:42:18.124 hereditary breast cancer as well As

NOTE Confidence: 0.84023404

00:42:18.124 --> 00:42:20.299 for what's called Lynch syndrome,

NOTE Confidence: 0.84023404

00:42:20.300 --> 00:42:23.450 which is actually a hereditary colon cancer,

NOTE Confidence: 0.84023404

00:42:23.450 --> 00:42:24.794 an individual cancer.

NOTE Confidence: 0.84023404

00:42:24.794 --> 00:42:27.034 So these are two genetic.

NOTE Confidence: 0.85791844

00:42:29.750 --> 00:42:31.014 Conditions are hereditary cancer

NOTE Confidence: 0.85791844

00:42:31.014 --> 00:42:32.910 syndromes for which there are clear

NOTE Confidence: 0.85791844

00:42:32.961 --> 00:42:34.521 recommendations that if there's a family

NOTE Confidence: 0.85791844

00:42:34.521 --> 00:42:36.728 history or of a person of a certain age,

NOTE Confidence: 0.85791844

00:42:36.730 --> 00:42:38.398 it's recommended that testing be done.

NOTE Confidence: 0.85791844

00:42:38.400 --> 00:42:40.624 So the idea here is not something hey,

NOTE Confidence: 0.85791844

00:42:40.630 --> 00:42:41.746 do you want this?

NOTE Confidence: 0.85791844

00:42:41.746 --> 00:42:43.420 Maybe you should think about this,

NOTE Confidence: 0.85791844

00:42:43.420 --> 00:42:45.652 but you should have this done in the same
NOTE Confidence: 0.85791844

00:42:45.652 --> 00:42:47.885 way that somebody is having chest pain.
NOTE Confidence: 0.85791844

00:42:47.890 --> 00:42:49.280 We might say, you know,
NOTE Confidence: 0.85791844

00:42:49.280 --> 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 --> 00:42:55.414 so no, you should have this
NOTE Confidence: 0.85791844

00:42:55.414 --> 00:42:57.090 and the same idea here is,
NOTE Confidence: 0.85791844

00:42:57.090 --> 00:43:00.000 yet you should have this done.
NOTE Confidence: 0.85791844

00:43:00.000 --> 00:43:03.448 So the question then becomes the fact that.
NOTE Confidence: 0.9320227

00:43:06.000 --> 00:43:10.648 People who are. Low in low income
NOTE Confidence: 0.9320227

00:43:10.648 --> 00:43:12.176 or underrepresented in research
NOTE Confidence: 0.9320227

00:43:12.176 --> 00:43:14.205 and clinical care are less likely
NOTE Confidence: 0.9320227

00:43:14.205 --> 00:43:16.390 to have this type of testing done.
NOTE Confidence: 0.9320227

00:43:16.390 --> 00:43:18.280 They may. Not have had if they
NOTE Confidence: 0.9320227

00:43:18.280 --> 00:43:20.228 may not have a family history,
NOTE Confidence: 0.9320227

00:43:20.230 --> 00:43:22.282 they may not have had a family history taken.

NOTE Confidence: 0.9320227
00:43:22.290 --> 00:43:23.886 They may not have gotten a referral.
NOTE Confidence: 0.9320227
00:43:23.890 --> 00:43:25.690 They may not have gotten.
NOTE Confidence: 0.9320227
00:43:25.690 --> 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 --> 00:43:29.270 so we started realizing that
NOTE Confidence: 0.9320227
00:43:29.270 --> 00:43:30.760 if there was a pop,
NOTE Confidence: 0.9320227
00:43:30.760 --> 00:43:32.524 if there was a that if we're
NOTE Confidence: 0.9320227
00:43:32.524 --> 00:43:34.220 looking for an opportunity to do
NOTE Confidence: 0.9320227
00:43:34.220 --> 00:43:36.271 testing in a way that could have
NOTE Confidence: 0.9320227
00:43:36.333 --> 00:43:38.208 an impact on health disparities.
NOTE Confidence: 0.9320227
00:43:38.210 --> 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 --> 00:43:46.284 we our country and I decided
NOTE Confidence: 0.9320227

00:43:46.284 --> 00:43:48.398 to apply for the next round of

NOTE Confidence: 0.9320227

00:43:48.398 --> 00:43:50.128 funding from the Caesar Consortium,

NOTE Confidence: 0.9320227

00:43:50.130 --> 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 --> 00:43:53.898 five of the six studies were about

NOTE Confidence: 0.9320227

00:43:53.898 --> 00:43:55.190 Pediatrics and pediatric context,

NOTE Confidence: 0.9320227

00:43:55.190 --> 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 --> 00:44:00.897 really focused on adults with

NOTE Confidence: 0.9320227

00:44:00.897 --> 00:44:02.358 hereditary cancer syndromes.

NOTE Confidence: 0.9320227

00:44:02.360 --> 00:44:05.060 In a primary care setting.

NOTE Confidence: 0.9320227

00:44:05.060 --> 00:44:07.036 Well I want to show you on this

NOTE Confidence: 0.9320227

00:44:07.036 --> 00:44:09.050 slide is to acknowledge that there's

NOTE Confidence: 0.9320227

00:44:09.050 --> 00:44:11.234 a series of decisions that need

NOTE Confidence: 0.9320227

00:44:11.302 --> 00:44:13.276 to be made regarding doing this,

NOTE Confidence: 0.9320227

00:44:13.280 --> 00:44:15.485 and it's very complicated to do so.

NOTE Confidence: 0.9320227

00:44:15.490 --> 00:44:17.576 Part of what our task was and

NOTE Confidence: 0.9320227

00:44:17.576 --> 00:44:19.600 thinking about our study was saying,

NOTE Confidence: 0.9320227

00:44:19.600 --> 00:44:21.496 how can we make this process

NOTE Confidence: 0.9320227

00:44:21.496 --> 00:44:22.760 simpler and less complicated?

NOTE Confidence: 0.9320227

00:44:22.760 --> 00:44:24.638 To make it more accessible to

NOTE Confidence: 0.9320227

00:44:24.638 --> 00:44:26.246 people who otherwise might not

NOTE Confidence: 0.9320227

00:44:26.246 --> 00:44:27.701 have access to these services

NOTE Confidence: 0.9320227

00:44:27.701 --> 00:44:29.709 where it's hard to be identified,

NOTE Confidence: 0.9320227

00:44:29.710 --> 00:44:31.544 it's hard to get to come in

NOTE Confidence: 0.9320227

00:44:31.544 --> 00:44:33.180 to get an evaluation,

NOTE Confidence: 0.9320227

00:44:33.180 --> 00:44:35.208 and that and the agenda counselor.

NOTE Confidence: 0.9320227

00:44:35.210 --> 00:44:36.715 And to come back several

NOTE Confidence: 0.9320227

00:44:36.715 --> 00:44:37.919 times or several visits.

NOTE Confidence: 0.9320227

00:44:37.920 --> 00:44:39.904 So we one of the things that we

NOTE Confidence: 0.9320227

00:44:39.904 --> 00:44:41.903 do this in a more simplified

NOTE Confidence: 0.9320227

00:44:41.903 --> 00:44:43.335 way and still effective.
NOTE Confidence: 0.9320227

00:44:43.340 --> 00:44:45.080 So the study that we developed
NOTE Confidence: 0.9320227

00:44:45.080 --> 00:44:46.650 is called the Charm study,
NOTE Confidence: 0.9320227

00:44:46.650 --> 00:44:48.155 which stands for cancer Health
NOTE Confidence: 0.9320227

00:44:48.155 --> 00:44:49.058 assessments reaching many.
NOTE Confidence: 0.9320227

00:44:49.060 --> 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 --> 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 --> 00:45:03.470 but also a Denver health,
NOTE Confidence: 0.9320227

00:45:03.470 --> 00:45:05.220 which is a federally qualified
NOTE Confidence: 0.9320227

00:45:05.220 --> 00:45:07.803 Health Center in Denver as well as
NOTE Confidence: 0.9320227

00:45:07.803 --> 00:45:09.708 with collaborators at University of
NOTE Confidence: 0.9320227

00:45:09.708 --> 00:45:11.548 Washington who did the sequencing
NOTE Confidence: 0.9320227

00:45:11.548 --> 00:45:13.746 for us as well as collaborators at

NOTE Confidence: 0.9320227

00:45:13.746 --> 00:45:15.568 a variety of other institutions,

NOTE Confidence: 0.9320227

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 --> 00:45:22.241 who helped us with both the risk

NOTE Confidence: 0.9320227

00:45:22.241 --> 00:45:24.308 assessment evaluation and our study

NOTE Confidence: 0.9320227

00:45:24.308 --> 00:45:27.324 included a range of both evaluations of of.

NOTE Confidence: 0.9320227

00:45:27.330 --> 00:45:27.683 Of.

NOTE Confidence: 0.9320227

00:45:27.683 --> 00:45:29.448 Doing lots of interviews with

NOTE Confidence: 0.9320227

00:45:29.448 --> 00:45:31.221 participants along the course of

NOTE Confidence: 0.9320227

00:45:31.221 --> 00:45:33.003 the study to learn about their

NOTE Confidence: 0.9320227

00:45:33.003 --> 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 --> 00:45:39.889 our expertise in qualitative research.

NOTE Confidence: 0.9320227

00:45:39.890 --> 00:45:40.906 The study.

NOTE Confidence: 0.9320227

00:45:40.906 --> 00:45:43.954 Included individuals aged 18 to 49
NOTE Confidence: 0.9320227

00:45:43.954 --> 00:45:47.337 who either spoke English or Spanish.
NOTE Confidence: 0.9320227

00:45:47.340 --> 00:45:50.420 This was done at.
NOTE Confidence: 0.9320227

00:45:50.420 --> 00:45:52.120 Kaiser in the Portland area,
NOTE Confidence: 0.9320227

00:45:52.120 --> 00:45:54.160 as well as a Denver health.
NOTE Confidence: 0.81246525

00:45:54.160 --> 00:45:55.860 We had our different range
NOTE Confidence: 0.81246525

00:45:55.860 --> 00:45:57.560 of ways of recruiting people,
NOTE Confidence: 0.81246525

00:45:57.560 --> 00:45:58.580 either through email,
NOTE Confidence: 0.81246525

00:45:58.580 --> 00:46:00.280 text messages or in person.
NOTE Confidence: 0.81246525

00:46:00.280 --> 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 --> 00:46:05.712 including in Denver help.
NOTE Confidence: 0.81246525

00:46:05.712 --> 00:46:07.740 Also by our direct referrals by
NOTE Confidence: 0.81246525

00:46:07.800 --> 00:46:09.172 providers for those individuals
NOTE Confidence: 0.81246525

00:46:09.172 --> 00:46:11.624 who they believe may be at risk
NOTE Confidence: 0.81246525

00:46:11.624 --> 00:46:13.580 for having hereditary cancer in the

NOTE Confidence: 0.81246525

00:46:13.580 --> 00:46:17.025 family but had not yet been had an

NOTE Confidence: 0.81246525

00:46:17.025 --> 00:46:18.997 opportunity for genetic testing.

NOTE Confidence: 0.81246525

00:46:19.000 --> 00:46:20.750 The main outcome of our study was

NOTE Confidence: 0.81246525

00:46:20.750 --> 00:46:22.716 to look at the positive findings

NOTE Confidence: 0.81246525

00:46:22.716 --> 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 --> 00:46:30.185 outcomes that included both the

NOTE Confidence: 0.81246525

00:46:30.185 --> 00:46:32.362 number of of other findings but also

NOTE Confidence: 0.81246525

00:46:32.362 --> 00:46:34.119 included health care utilization.

NOTE Confidence: 0.81246525

00:46:34.120 --> 00:46:35.800 Their understanding of the care,

NOTE Confidence: 0.81246525

00:46:35.800 --> 00:46:37.144 their satisfaction, their communication,

NOTE Confidence: 0.81246525

00:46:37.144 --> 00:46:39.160 and well say the very end.

NOTE Confidence: 0.81246525

00:46:39.160 --> 00:46:40.705 About personal utility.

NOTE Confidence: 0.81246525

00:46:40.705 --> 00:46:43.795 The value of this for them.

NOTE Confidence: 0.81246525

00:46:43.800 --> 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 --> 00:46:53.320 is to suggest that this was a very

NOTE Confidence: 0.81246525

00:46:53.320 --> 00:46:58.030 complex project where we did. Is.

NOTE Confidence: 0.81246525

00:46:58.030 --> 00:47:00.814 If you look at this range of services.

NOTE Confidence: 0.81246525

00:47:00.820 --> 00:47:02.630 We identified 12 different things

NOTE Confidence: 0.81246525

00:47:02.630 --> 00:47:05.171 that we tried to do differently to

NOTE Confidence: 0.81246525

00:47:05.171 --> 00:47:07.795 see how well it would work at it,

NOTE Confidence: 0.81246525

00:47:07.800 --> 00:47:10.229 including how we try to reach people

NOTE Confidence: 0.81246525

00:47:10.229 --> 00:47:12.301 by identifying people by email or

NOTE Confidence: 0.81246525

00:47:12.301 --> 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 --> 00:47:18.349 patient facing tool.

NOTE Confidence: 0.81246525

00:47:18.350 --> 00:47:20.744 We included people who have limited

NOTE Confidence: 0.81246525

00:47:20.744 --> 00:47:22.340 information about their families

NOTE Confidence: 0.81246525

00:47:22.403 --> 00:47:23.927 as a way of being broader.

NOTE Confidence: 0.81246525

00:47:23.930 --> 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 --> 00:47:30.396 and then we also did our pretest

NOTE Confidence: 0.81246525

00:47:30.396 --> 00:47:32.309 education counseling online as well,

NOTE Confidence: 0.81246525

00:47:32.310 --> 00:47:35.094 and so the idea here is that the

NOTE Confidence: 0.81246525

00:47:35.094 --> 00:47:36.625 individuals who were obtaining

NOTE Confidence: 0.81246525

00:47:36.625 --> 00:47:39.264 this did not actually have to go

NOTE Confidence: 0.81246525

00:47:39.264 --> 00:47:41.030 into a doctor's office.

NOTE Confidence: 0.81246525

00:47:41.030 --> 00:47:43.366 Not only one, not only more than once,

NOTE Confidence: 0.81246525

00:47:43.370 --> 00:47:45.989 but even once all this could be done online,

NOTE Confidence: 0.81246525

00:47:45.990 --> 00:47:47.425 we also provided halfway through

NOTE Confidence: 0.81246525

00:47:47.425 --> 00:47:49.442 the study a decision aid to help

NOTE Confidence: 0.81246525

00:47:49.442 --> 00:47:50.772 people decide whether they did

NOTE Confidence: 0.81246525

00:47:50.772 --> 00:47:52.253 or didn't want secondary findings

NOTE Confidence: 0.81246525

00:47:52.253 --> 00:47:54.125 because we found we offer people
NOTE Confidence: 0.81246525

00:47:54.125 --> 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 --> 00:48:00.238 saying yes because.
NOTE Confidence: 0.81246525

00:48:00.240 --> 00:48:00.908 Why not?
NOTE Confidence: 0.81246525

00:48:00.908 --> 00:48:02.578 It's free, more is better,
NOTE Confidence: 0.81246525

00:48:02.580 --> 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 --> 00:48:10.370 to think about this more deliberately.
NOTE Confidence: 0.81246525

00:48:10.370 --> 00:48:12.708 I want to comment about our results.
NOTE Confidence: 0.81246525

00:48:12.710 --> 00:48:14.046 Disclosure is that we,
NOTE Confidence: 0.81246525

00:48:14.046 --> 00:48:16.050 for those people had negative results.
NOTE Confidence: 0.81246525

00:48:16.050 --> 00:48:17.386 We provided that information
NOTE Confidence: 0.81246525

00:48:17.386 --> 00:48:18.388 initially by phone,
NOTE Confidence: 0.81246525

00:48:18.390 --> 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,
NOTE Confidence: 0.81246525
00:48:24.060 --> 00:48:25.660 there's a couple places where
NOTE Confidence: 0.81246525
00:48:25.660 --> 00:48:28.013 there are some dice the dice refer
NOTE Confidence: 0.81246525
00:48:28.013 --> 00:48:29.738 to things that we randomized,
NOTE Confidence: 0.81246525
00:48:29.740 --> 00:48:31.016 so we did randomize.
NOTE Confidence: 0.81246525
00:48:31.016 --> 00:48:32.930 People explain this in more detail
NOTE Confidence: 0.81246525
00:48:32.989 --> 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 --> 00:48:38.525 We also randomized training for
NOTE Confidence: 0.81246525
00:48:38.525 --> 00:48:40.580 interpreters to see whether or not.
NOTE Confidence: 0.81246525
00:48:40.580 --> 00:48:41.904 Having trained interpreters would
NOTE Confidence: 0.81246525
00:48:41.904 --> 00:48:43.559 be helpful to communicating this
NOTE Confidence: 0.81246525
00:48:43.559 --> 00:48:44.280 information as well,
NOTE Confidence: 0.81246525
00:48:44.280 --> 00:48:46.116 because this study is still ongoing,
NOTE Confidence: 0.81246525
00:48:46.120 --> 00:48:47.920 I'm not going to present the
NOTE Confidence: 0.81246525

00:48:47.920 --> 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 --> 00:48:53.186 describe some more of the features
NOTE Confidence: 0.84673136

00:48:53.186 --> 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 --> 00:48:59.700 convey is to suggest that.
NOTE Confidence: 0.84673136

00:48:59.700 --> 00:49:02.244 Studies like this that try to look at
NOTE Confidence: 0.84673136

00:49:02.244 --> 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 --> 00:49:10.608 and how we learn how to innovate and
NOTE Confidence: 0.84673136

00:49:10.608 --> 00:49:13.029 see if we can do this effectively.
NOTE Confidence: 0.84673136

00:49:13.030 --> 00:49:15.130 So I want to share with you
NOTE Confidence: 0.84673136

00:49:15.130 --> 00:49:17.058 a few aspects of our study.
NOTE Confidence: 0.84673136

00:49:17.060 --> 00:49:18.812 One is that our approach to
NOTE Confidence: 0.84673136

00:49:18.812 --> 00:49:20.470 consent was was done online.

NOTE Confidence: 0.84673136

00:49:20.470 --> 00:49:22.890 We used a multimedia approach

NOTE Confidence: 0.84673136

00:49:22.890 --> 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 --> 00:49:27.820 our images for the previous study

NOTE Confidence: 0.84673136

00:49:27.820 --> 00:49:29.055 of the of the PowerPoints.

NOTE Confidence: 0.84673136

00:49:29.060 --> 00:49:30.560 Here we use images with very

NOTE Confidence: 0.84673136

00:49:30.560 --> 00:49:31.060 brief information.

NOTE Confidence: 0.84673136

00:49:31.060 --> 00:49:32.796 So we really tried to think about.

NOTE Confidence: 0.851953569999999

00:49:34.910 --> 00:49:37.446 How to be as concise as possible with

NOTE Confidence: 0.851953569999999

00:49:37.446 --> 00:49:39.935 what this test will tell me and one

NOTE Confidence: 0.851953569999999

00:49:39.935 --> 00:49:42.473 of the things that I have been a very

NOTE Confidence: 0.851953569999999

00:49:42.473 --> 00:49:44.582 big fan of with the concept of consent

NOTE Confidence: 0.851953569999999

00:49:44.582 --> 00:49:46.500 is trying to be concise by making

NOTE Confidence: 0.851953569999999

00:49:46.561 --> 00:49:48.780 suggestions for what the reasons might be,

NOTE Confidence: 0.851953569999999

00:49:48.780 --> 00:49:50.550 why some people may want it,
NOTE Confidence: 0.8519535699999999

00:49:50.550 --> 00:49:52.910 and why some people may not want it.
NOTE Confidence: 0.8519535699999999

00:49:52.910 --> 00:49:54.947 And it's a way of both communicating
NOTE Confidence: 0.8519535699999999

00:49:54.947 --> 00:49:56.738 that there's more than one choice,
NOTE Confidence: 0.8519535699999999

00:49:56.740 --> 00:49:58.964 but to do it in a way that
NOTE Confidence: 0.8519535699999999

00:49:58.964 --> 00:50:00.400 makes it fairly so.
NOTE Confidence: 0.8519535699999999

00:50:00.400 --> 00:50:02.068 Simple to see what those reasons
NOTE Confidence: 0.8519535699999999

00:50:02.068 --> 00:50:03.505 are rather than thinking there's
NOTE Confidence: 0.8519535699999999

00:50:03.505 --> 00:50:04.850 a whole litany of things,
NOTE Confidence: 0.8519535699999999

00:50:04.850 --> 00:50:07.230 but really trying to boil down to
NOTE Confidence: 0.8519535699999999

00:50:07.230 --> 00:50:09.340 some upsides and some downsides.
NOTE Confidence: 0.8519535699999999

00:50:09.340 --> 00:50:11.930 The other thing that we did is that we just
NOTE Confidence: 0.8519535699999999

00:50:11.990 --> 00:50:14.398 about this was done as a research study.
NOTE Confidence: 0.8519535699999999

00:50:14.400 --> 00:50:16.200 We distinguish between consent for testing
NOTE Confidence: 0.8519535699999999

00:50:16.200 --> 00:50:18.049 and consent from being in the study,
NOTE Confidence: 0.8519535699999999

00:50:18.050 --> 00:50:19.938 and so that the first thing we tried

NOTE Confidence: 0.851953569999999

00:50:19.938 --> 00:50:22.116 to do is to walk them through whether

NOTE Confidence: 0.851953569999999

00:50:22.116 --> 00:50:23.950 or not from their perspective,

NOTE Confidence: 0.851953569999999

00:50:23.950 --> 00:50:25.636 genetic testing made sense for them,

NOTE Confidence: 0.851953569999999

00:50:25.640 --> 00:50:27.425 and then then say the option for

NOTE Confidence: 0.851953569999999

00:50:27.425 --> 00:50:29.371 you is to have it done through

NOTE Confidence: 0.851953569999999

00:50:29.371 --> 00:50:31.540 the study or talk to your doctor.

NOTE Confidence: 0.851953569999999

00:50:31.540 --> 00:50:31.780 Further,

NOTE Confidence: 0.851953569999999

00:50:31.780 --> 00:50:33.940 if you don't want to be in the study

NOTE Confidence: 0.851953569999999

00:50:33.996 --> 00:50:35.970 so that the consent was really broken

NOTE Confidence: 0.851953569999999

00:50:35.970 --> 00:50:37.998 down into these two different pieces,

NOTE Confidence: 0.851953569999999

00:50:38.000 --> 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 --> 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 --> 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 --> 00:50:57.955 for example, so we wanted to do
NOTE Confidence: 0.73455286

00:50:57.955 --> 00:50:59.785 with working with the collaborative
NOTE Confidence: 0.73455286

00:50:59.785 --> 00:51:02.100 what's called the Prim. We
NOTE Confidence: 0.7347506

00:51:04.380 --> 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 --> 00:51:12.349 patients who are doing this online,
NOTE Confidence: 0.8597141

00:51:12.350 --> 00:51:15.410 so we had to really make some changes in it.
NOTE Confidence: 0.8597141

00:51:15.410 --> 00:51:17.240 We had we simplified the language.
NOTE Confidence: 0.8597141

00:51:17.240 --> 00:51:18.770 We use less medical terms.
NOTE Confidence: 0.8597141

00:51:18.770 --> 00:51:20.684 We actually did the calculations for
NOTE Confidence: 0.8597141

00:51:20.684 --> 00:51:22.877 the participants and we had visual aids
NOTE Confidence: 0.8597141

00:51:22.877 --> 00:51:24.886 also to help describe what this meant,
NOTE Confidence: 0.8597141

00:51:24.890 --> 00:51:28.986 and we also have done some of valuations.
NOTE Confidence: 0.8597141

00:51:28.990 --> 00:51:30.790 Actually forgot that we also do

NOTE Confidence: 0.8597141

00:51:30.790 --> 00:51:32.748 some evaluations of how well this

NOTE Confidence: 0.8597141

00:51:32.748 --> 00:51:34.836 modification work and present that today,

NOTE Confidence: 0.8597141

00:51:34.840 --> 00:51:36.465 but this is actually trying

NOTE Confidence: 0.8597141

00:51:36.465 --> 00:51:38.090 to work really very well,

NOTE Confidence: 0.8597141

00:51:38.090 --> 00:51:40.619 so we have been able to create and develop

NOTE Confidence: 0.8597141

00:51:40.619 --> 00:51:43.158 a tool that people can fill out both

NOTE Confidence: 0.8597141

00:51:43.158 --> 00:51:45.568 in English and Spanish that describe.

NOTE Confidence: 0.8597141

00:51:45.570 --> 00:51:48.314 It helps to understand what their actual risk

NOTE Confidence: 0.8597141

00:51:48.314 --> 00:51:50.997 of having hereditary cancer syndrome are.

NOTE Confidence: 0.8597141

00:51:51.000 --> 00:51:53.616 They were the most important things we done.

NOTE Confidence: 0.8597141

00:51:53.620 --> 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 --> 00:52:00.059 most effectively and we wanted to

NOTE Confidence: 0.8597141

00:52:00.120 --> 00:52:02.106 use the information that you saw

NOTE Confidence: 0.8597141

00:52:02.106 --> 00:52:03.756 in that previous paper regarding

NOTE Confidence: 0.8597141

00:52:03.756 --> 00:52:05.386 an approach to informed consent.
NOTE Confidence: 0.8597141

00:52:05.390 --> 00:52:08.038 And we also want to apply that to
NOTE Confidence: 0.8597141

00:52:08.038 --> 00:52:09.967 Jenna Counseling an for this study.
NOTE Confidence: 0.8597141

00:52:09.970 --> 00:52:11.600 We actually collaborated with women
NOTE Confidence: 0.8597141

00:52:11.600 --> 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 --> 00:52:18.172 Who's been studying Jenna councils for
NOTE Confidence: 0.8597141

00:52:18.172 --> 00:52:20.200 years and together we collaborate on
NOTE Confidence: 0.8597141

00:52:20.200 --> 00:52:22.768 creating what we have now described as the.
NOTE Confidence: 0.8597141

00:52:22.770 --> 00:52:24.945 Aria model of gender counseling
NOTE Confidence: 0.8597141

00:52:24.945 --> 00:52:27.120 that refers to being accessible,
NOTE Confidence: 0.8597141

00:52:27.120 --> 00:52:29.240 relational, inclusive, an actionable,
NOTE Confidence: 0.8597141

00:52:29.240 --> 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 --> 00:52:35.304 make the information easy to understand.

NOTE Confidence: 0.8597141

00:52:35.310 --> 00:52:37.674 Really focus on the relationship between

NOTE Confidence: 0.8597141

00:52:37.674 --> 00:52:41.032 the clinician and the provider as a way

NOTE Confidence: 0.8597141

00:52:41.032 --> 00:52:42.748 of helping facilitate understanding,

NOTE Confidence: 0.8597141

00:52:42.750 --> 00:52:44.151 but more importantly,

NOTE Confidence: 0.8597141

00:52:44.151 --> 00:52:46.953 helping to facilitate and ensure that

NOTE Confidence: 0.8597141

00:52:46.953 --> 00:52:48.924 patients understand the actions and

NOTE Confidence: 0.8597141

00:52:48.924 --> 00:52:51.830 know what steps to do the next time.

NOTE Confidence: 0.8597141

00:52:51.830 --> 00:52:55.134 So the idea, and So what we did,

NOTE Confidence: 0.8597141

00:52:55.140 --> 00:52:57.385 we developed a fairly detailed

NOTE Confidence: 0.8597141

00:52:57.385 --> 00:52:59.630 curriculum to train 2 genetic

NOTE Confidence: 0.8597141

00:52:59.712 --> 00:53:02.407 counselors to use the Aria model an.

NOTE Confidence: 0.8597141

00:53:02.410 --> 00:53:03.418 Other two genetic counselors.

NOTE Confidence: 0.8597141

00:53:03.418 --> 00:53:04.930 Did this get their standard approach

NOTE Confidence: 0.8597141

00:53:04.975 --> 00:53:06.427 to gender counseling that begins with.

NOTE Confidence: 0.8597141

00:53:06.430 --> 00:53:07.430 This is the gene.

NOTE Confidence: 0.8597141

00:53:07.430 --> 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 --> 00:53:11.447 Let's tell you what your results are.
NOTE Confidence: 0.8597141

00:53:11.450 --> 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 --> 00:53:16.570 this because it's always complicated.
NOTE Confidence: 0.8597141

00:53:16.570 --> 00:53:20.170 And then will tell you what to do, maybe so.
NOTE Confidence: 0.8597141

00:53:20.170 --> 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 --> 00:53:23.536 Here's a very quick example of
NOTE Confidence: 0.8597141

00:53:23.536 --> 00:53:25.060 the distinction between usual
NOTE Confidence: 0.8597141

00:53:25.125 --> 00:53:26.650 care and the Aria model.
NOTE Confidence: 0.8597141

00:53:26.650 --> 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 --> 00:53:31.538 this notion that the emphasis and usual
NOTE Confidence: 0.8597141

00:53:31.538 --> 00:53:33.486 care is on transferred information

NOTE Confidence: 0.8597141

00:53:33.486 --> 00:53:35.646 is usually built technical language.

NOTE Confidence: 0.8597141

00:53:35.650 --> 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 --> 00:53:41.410 an often it's historically is non directive.

NOTE Confidence: 0.8597141

00:53:41.410 --> 00:53:44.290 It's like you may want to do this.

NOTE Confidence: 0.8597141

00:53:44.290 --> 00:53:46.558 You may want to do this.

NOTE Confidence: 0.8597141

00:53:46.560 --> 00:53:48.935 Whereas the Aria model was

NOTE Confidence: 0.8597141

00:53:48.935 --> 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 --> 00:53:54.187 counseling establishing a

NOTE Confidence: 0.8597141

00:53:54.187 --> 00:53:56.180 relationship with the participant.

NOTE Confidence: 0.8597141

00:53:56.180 --> 00:53:58.838 You are using direct and simple

NOTE Confidence: 0.8597141

00:53:58.838 --> 00:54:00.610 language being very clear.

NOTE Confidence: 0.8597141

00:54:00.610 --> 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 --> 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 --> 00:54:15.789 me back in a year from now,

NOTE Confidence: 0.8762343

00:54:15.790 --> 00:54:17.380 we will tell you the results

NOTE Confidence: 0.8762343

00:54:17.380 --> 00:54:19.110 of this part of the study.

NOTE Confidence: 0.8762343

00:54:19.110 --> 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 --> 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 --> 00:54:27.977 briefly who enrolled in the study.

NOTE Confidence: 0.8762343

00:54:27.980 --> 00:54:29.360 We have roughly 800 people

NOTE Confidence: 0.8762343

00:54:29.360 --> 00:54:30.464 enrolled in the study,

NOTE Confidence: 0.8762343

00:54:30.470 --> 00:54:32.526 and what I want to share with you

NOTE Confidence: 0.8762343

00:54:32.526 --> 00:54:34.741 briefly is the fact that we were able to

NOTE Confidence: 0.8762343

00:54:34.741 --> 00:54:36.811 get a more diverse population that we

NOTE Confidence: 0.8762343

00:54:36.811 --> 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

NOTE Confidence: 0.8762343

00:54:41.044 --> 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 --> 00:54:46.600 but in a variety of other groups as well too.

NOTE Confidence: 0.8762343

00:54:46.600 --> 00:54:48.768 We are also able to try to have

NOTE Confidence: 0.8762343

00:54:48.768 --> 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 --> 00:54:54.480 as well as income diversity.

NOTE Confidence: 0.8762343

00:54:54.480 --> 00:54:56.538 I without going into the details,

NOTE Confidence: 0.8762343

00:54:56.540 --> 00:54:58.388 I briefly want to acknowledge for

NOTE Confidence: 0.8762343

00:54:58.388 --> 00:55:00.819 those of you in the audience that

NOTE Confidence: 0.8762343

00:55:00.819 --> 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 --> 00:55:06.702 we were able to identify through

NOTE Confidence: 0.8762343

00:55:06.702 --> 00:55:08.390 the charm study.

NOTE Confidence: 0.8762343

00:55:08.390 --> 00:55:10.215 Quite a number of positive
NOTE Confidence: 0.8762343

00:55:10.215 --> 00:55:12.610 results of the 800 people we had,
NOTE Confidence: 0.8762343

00:55:12.610 --> 00:55:14.898 we had 36 people who had positive results
NOTE Confidence: 0.8762343

00:55:14.898 --> 00:55:17.189 or other pathogenic likely pathogenic.
NOTE Confidence: 0.8762343

00:55:17.190 --> 00:55:19.398 We also did some additional additional
NOTE Confidence: 0.8762343

00:55:19.398 --> 00:55:21.247 findings related to other things
NOTE Confidence: 0.8762343

00:55:21.247 --> 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 --> 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 --> 00:55:28.805 we had to make decisions.
NOTE Confidence: 0.8762343

00:55:28.810 --> 00:55:31.232 About 126 genes that we felt were
NOTE Confidence: 0.8762343

00:55:31.232 --> 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 --> 00:55:36.549 we were going to be reporting.
NOTE Confidence: 0.8762343

00:55:36.550 --> 00:55:39.448 So anytime you hear the word sequencing.

NOTE Confidence: 0.8762343

00:55:39.450 --> 00:55:40.734 It doesn't mean everything.

NOTE Confidence: 0.8762343

00:55:40.734 --> 00:55:42.660 It means a deliberate choice to

NOTE Confidence: 0.8762343

00:55:42.715 --> 00:55:44.617 include some things and not others.

NOTE Confidence: 0.8762343

00:55:44.620 --> 00:55:46.420 Last point I'll make before I

NOTE Confidence: 0.8762343

00:55:46.420 --> 00:55:48.392 end it 'cause Mark said I have

NOTE Confidence: 0.8762343

00:55:48.392 --> 00:55:49.808 to have up to an hour.

NOTE Confidence: 0.8762343

00:55:49.810 --> 00:55:51.418 We have time for conversation is

NOTE Confidence: 0.8762343

00:55:51.418 --> 00:55:53.360 as much about two points to make,

NOTE Confidence: 0.8762343

00:55:53.360 --> 00:55:56.312 so another 5 minutes I want to at least

NOTE Confidence: 0.8762343

00:55:56.312 --> 00:55:58.052 acknowledge that we were successful

NOTE Confidence: 0.8762343

00:55:58.052 --> 00:56:00.436 in reaching people who had a bear

NOTE Confidence: 0.8762343

00:56:00.436 --> 00:56:02.900 who had a range of barriers to access.

NOTE Confidence: 0.8762343

00:56:02.900 --> 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 --> 00:56:09.139 point out and I thought this was

NOTE Confidence: 0.8762343

00:56:09.139 --> 00:56:11.090 very important that but we were able.

NOTE Confidence: 0.8762343

00:56:11.090 --> 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 --> 00:56:17.835 maybe less likely have access to

NOTE Confidence: 0.8762343

00:56:17.835 --> 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 --> 00:56:22.539 testing may be important to them

NOTE Confidence: 0.8762343

00:56:22.539 --> 00:56:23.967 because they may not have access

NOTE Confidence: 0.8762343

00:56:23.967 --> 00:56:25.663 to family history information if

NOTE Confidence: 0.8762343

00:56:25.663 --> 00:56:27.075 they have strained relationships.

NOTE Confidence: 0.8762343

00:56:27.080 --> 00:56:29.636 And this is one of the things that we

NOTE Confidence: 0.8762343

00:56:29.636 --> 00:56:31.547 learned from interviewing people from

NOTE Confidence: 0.8762343

00:56:31.547 --> 00:56:34.589 that population who were part of our study.

NOTE Confidence: 0.8762343

00:56:34.590 --> 00:56:36.936 This side very quickly just suggested

NOTE Confidence: 0.8762343

00:56:36.936 --> 00:56:39.249 during the course of the study.

NOTE Confidence: 0.8762343

00:56:39.250 --> 00:56:41.386 Which include the risk assessment tool,

NOTE Confidence: 0.8762343

00:56:41.390 --> 00:56:42.461 which term eligibility

NOTE Confidence: 0.8762343

00:56:42.461 --> 00:56:44.603 agreement to be in the study,

NOTE Confidence: 0.8762343

00:56:44.610 --> 00:56:47.403 returning a saliva kit and then completing

NOTE Confidence: 0.8762343

00:56:47.403 --> 00:56:49.600 results disclosure as as you can see,

NOTE Confidence: 0.8762343

00:56:49.600 --> 00:56:51.028 there was a gradual

NOTE Confidence: 0.8762343

00:56:51.028 --> 00:56:52.456 degradation along the way,

NOTE Confidence: 0.82956165

00:56:52.460 --> 00:56:54.902 but this was not the study

NOTE Confidence: 0.82956165

00:56:54.902 --> 00:56:56.530 successfully retained that population

NOTE Confidence: 0.82956165

00:56:56.597 --> 00:56:58.505 who did have barriers to access

NOTE Confidence: 0.82956165

00:56:58.505 --> 00:57:00.668 a related to being in the study.

NOTE Confidence: 0.85935867

00:57:03.400 --> 00:57:05.872 Because I want to skip this slide simply

NOTE Confidence: 0.85935867

00:57:05.872 --> 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 --> 00:57:12.409 We are still trying to evaluate potential

NOTE Confidence: 0.85935867

00:57:12.409 --> 00:57:14.684 risks of this approach in terms of over
NOTE Confidence: 0.85935867

00:57:14.684 --> 00:57:16.819 testing in terms of the results themselves
NOTE Confidence: 0.85935867

00:57:16.819 --> 00:57:19.465 in terms of the implications for counselors.
NOTE Confidence: 0.85935867

00:57:19.470 --> 00:57:22.296 So there's a lot of work we're still doing,
NOTE Confidence: 0.85935867

00:57:22.300 --> 00:57:24.862 but I want to end with switching
NOTE Confidence: 0.85935867

00:57:24.862 --> 00:57:26.963 gears slightly and talking about
NOTE Confidence: 0.85935867

00:57:26.963 --> 00:57:28.827 something else that will.
NOTE Confidence: 0.85935867

00:57:28.830 --> 00:57:31.770 To give you some final thoughts.
NOTE Confidence: 0.85935867

00:57:31.770 --> 00:57:35.074 That's right, so here's my final thought.
NOTE Confidence: 0.85935867

00:57:35.080 --> 00:57:38.760 And it's it's kind of thinking back to.
NOTE Confidence: 0.85935867

00:57:38.760 --> 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 --> 00:57:44.114 and so I want to end with
NOTE Confidence: 0.85935867

00:57:44.114 --> 00:57:45.619 something from Winnie the Pooh.
NOTE Confidence: 0.85935867

00:57:45.620 --> 00:57:47.820 So a number of years ago I wrote an essay
NOTE Confidence: 0.85935867

00:57:47.881 --> 00:57:49.876 in response to somebody who is looking

NOTE Confidence: 0.85935867
00:57:49.876 --> 00:57:51.958 at the ethical implications of genome
NOTE Confidence: 0.85935867
00:57:51.958 --> 00:57:53.908 sequencing in the prenatal context,
NOTE Confidence: 0.85935867
00:57:53.910 --> 00:57:56.046 and as I thought about that
NOTE Confidence: 0.85935867
00:57:56.046 --> 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 --> 00:58:03.940 and I thought about that.
NOTE Confidence: 0.85935867
00:58:03.940 --> 00:58:06.520 The phenotypes of Winnie the Pooh,
NOTE Confidence: 0.85935867
00:58:06.520 --> 00:58:09.106 and I thought about various characters,
NOTE Confidence: 0.85935867
00:58:09.110 --> 00:58:12.200 so ER. The person is sceptical.
NOTE Confidence: 0.85935867
00:58:12.200 --> 00:58:14.447 Jack testing thinks of low in despair.
NOTE Confidence: 0.85935867
00:58:14.450 --> 00:58:16.376 This is a really bad idea,
NOTE Confidence: 0.85935867
00:58:16.380 --> 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 --> 00:58:21.810 always wanting to say well we can do this.
NOTE Confidence: 0.85935867

00:58:21.810 --> 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 --> 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 --> 00:58:34.085 to get a commitment to moving forward

NOTE Confidence: 0.85935867

00:58:34.085 --> 00:58:35.965 to nurturing those relationships with

NOTE Confidence: 0.85935867

00:58:35.965 --> 00:58:38.225 his friends in the in the in the wood.

NOTE Confidence: 0.85935867

00:58:38.230 --> 00:58:39.940 But also realizing how to leave

NOTE Confidence: 0.85935867

00:58:39.940 --> 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 --> 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 --> 00:58:50.125 who thought this is a bunch of.

NOTE Confidence: 0.85935867

00:58:50.130 --> 00:58:53.138 Crap bunch of honey.

NOTE Confidence: 0.85935867

00:58:53.140 --> 00:58:55.020 There's more important things than

NOTE Confidence: 0.85935867

00:58:55.020 --> 00:58:56.524 worrying about genetic testing.

NOTE Confidence: 0.85935867

00:58:56.530 --> 00:58:58.186 One of the things that people

NOTE Confidence: 0.85935867

00:58:58.186 --> 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 --> 00:59:03.020 and if we're looking for adult onset disease.

NOTE Confidence: 0.85935867

00:59:03.020 --> 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 --> 00:59:08.330 is what does that mean for the child?

NOTE Confidence: 0.85935867

00:59:08.330 --> 00:59:09.805 Whatever child finds out when

NOTE Confidence: 0.85935867

00:59:09.805 --> 00:59:11.280 they're there for the born,

NOTE Confidence: 0.85935867

00:59:11.280 --> 00:59:13.080 the parents right before the

NOTE Confidence: 0.85935867

00:59:13.080 --> 00:59:15.419 board they carry the BRCA 1 gene.

NOTE Confidence: 0.85935867

00:59:15.420 --> 00:59:17.643 Is that appropriate to do that for a child?

NOTE Confidence: 0.85935867

00:59:17.650 --> 00:59:19.522 Don't they have a right to an open

NOTE Confidence: 0.85935867

00:59:19.522 --> 00:59:21.092 future which is a term that's been

NOTE Confidence: 0.85935867

00:59:21.092 --> 00:59:22.772 used for the last 20 years as
NOTE Confidence: 0.85935867

00:59:22.772 --> 00:59:24.217 we were thinking about questions
NOTE Confidence: 0.85935867

00:59:24.217 --> 00:59:25.337 about testing in children?
NOTE Confidence: 0.85935867

00:59:25.337 --> 00:59:27.313 Since this is a pediatric audience in part,
NOTE Confidence: 0.85935867

00:59:27.320 --> 00:59:30.414 I want to end on this note.
NOTE Confidence: 0.85935867

00:59:30.420 --> 00:59:32.380 To suggest is that the right and open
NOTE Confidence: 0.85935867

00:59:32.380 --> 00:59:34.282 future is a considerations that we
NOTE Confidence: 0.85935867

00:59:34.282 --> 00:59:36.316 thought about by parents and clinicians,
NOTE Confidence: 0.85935867

00:59:36.320 --> 00:59:38.385 but perhaps not is a final answer.
NOTE Confidence: 0.85935867

00:59:38.390 --> 00:59:40.614 But when the parents may want to think
NOTE Confidence: 0.85935867

00:59:40.614 --> 00:59:42.722 about into account all decisions that
NOTE Confidence: 0.85935867

00:59:42.722 --> 00:59:44.582 parents make affect their child.
NOTE Confidence: 0.85935867

00:59:44.590 --> 00:59:46.634 And it really depends on the context
NOTE Confidence: 0.85935867

00:59:46.634 --> 00:59:48.829 of the request about whether or not
NOTE Confidence: 0.85935867

00:59:48.829 --> 00:59:50.719 this really will have a meaningful
NOTE Confidence: 0.8734655

00:59:50.777 --> 00:59:51.977 impact on the future,

NOTE Confidence: 0.8734655

00:59:51.980 --> 00:59:55.346 in a way that's even concerning.

NOTE Confidence: 0.8734655

00:59:55.350 --> 00:59:57.835 And even if we cannot predict problems,

NOTE Confidence: 0.8734655

00:59:57.840 --> 01:00:00.458 it doesn't mean that we should preclude.

NOTE Confidence: 0.8734655

01:00:00.460 --> 01:00:02.568 Decisions by parents based

NOTE Confidence: 0.8734655

01:00:02.568 --> 01:00:04.149 upon their interests.

NOTE Confidence: 0.8734655

01:00:04.150 --> 01:00:05.760 And so, in other words,

NOTE Confidence: 0.8734655

01:00:05.760 --> 01:00:07.692 even if we know that there might

NOTE Confidence: 0.8734655

01:00:07.692 --> 01:00:09.555 be some possible concerns for some

NOTE Confidence: 0.8734655

01:00:09.555 --> 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 --> 01:00:16.140 parents might still make that decision.

NOTE Confidence: 0.8734655

01:00:16.140 --> 01:00:18.760 Now. Why is that so?

NOTE Confidence: 0.8734655

01:00:18.760 --> 01:00:19.860 That maybe is a question

NOTE Confidence: 0.8734655

01:00:19.860 --> 01:00:21.049 maybe for the discussion,

NOTE Confidence: 0.8734655

01:00:21.050 --> 01:00:23.900 but what I want to end on to have you

NOTE Confidence: 0.8734655

01:00:23.983 --> 01:00:27.070 think about that is the following image.
NOTE Confidence: 0.8734655

01:00:27.070 --> 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 --> 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 --> 01:00:37.560 the the instigator for Winnie the Pooh.
NOTE Confidence: 0.8734655

01:00:37.560 --> 01:00:39.464 What you may some of you who are
NOTE Confidence: 0.8734655

01:00:39.464 --> 01:00:40.947 familiar with Christopher Robin in the
NOTE Confidence: 0.8734655

01:00:40.947 --> 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 --> 01:00:46.980 the Pooh stories because we tell it
NOTE Confidence: 0.8734655

01:00:46.980 --> 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 --> 01:00:54.540 with his father because of those
NOTE Confidence: 0.8734655

01:00:54.540 --> 01:00:56.220 stories he felt he was bullied.
NOTE Confidence: 0.8734655

01:00:56.220 --> 01:00:57.740 He felt he was ostracized.

NOTE Confidence: 0.8734655

01:00:57.740 --> 01:00:59.940 He became a strange for him from his

NOTE Confidence: 0.8734655

01:00:59.940 --> 01:01:01.834 father for decades and only really

NOTE Confidence: 0.8734655

01:01:01.834 --> 01:01:04.120 towards the end of his father's life.

NOTE Confidence: 0.8734655

01:01:04.120 --> 01:01:06.250 Did they reconcile?

NOTE Confidence: 0.8734655

01:01:06.250 --> 01:01:08.800 The point of this is that.

NOTE Confidence: 0.8734655

01:01:08.800 --> 01:01:10.350 Amil's decision to tell these

NOTE Confidence: 0.8734655

01:01:10.350 --> 01:01:12.420 stories had an impact on his son.

NOTE Confidence: 0.8734655

01:01:12.420 --> 01:01:15.138 We don't know whether he was aware of that,

NOTE Confidence: 0.8734655

01:01:15.140 --> 01:01:16.650 whether he thought about that,

NOTE Confidence: 0.8734655

01:01:16.650 --> 01:01:18.160 whether they should or shouldn't

NOTE Confidence: 0.8734655

01:01:18.160 --> 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 --> 01:01:22.410 and so I really want to point out

NOTE Confidence: 0.8734655

01:01:22.487 --> 01:01:24.827 in a in a in a very sincere way

NOTE Confidence: 0.8734655

01:01:24.827 --> 01:01:27.124 that while we do think about the

NOTE Confidence: 0.8734655

01:01:27.124 --> 01:01:29.238 the impact on the open future,
NOTE Confidence: 0.8734655

01:01:29.238 --> 01:01:32.066 we don't know what anyone's future will
NOTE Confidence: 0.8734655

01:01:32.066 --> 01:01:34.899 be and what the impact will be and.
NOTE Confidence: 0.8734655

01:01:34.900 --> 01:01:37.150 Just like a million Christopher Robin.
NOTE Confidence: 0.8683241

01:01:39.210 --> 01:01:41.016 It's a journey, so with that we
NOTE Confidence: 0.8683241

01:01:41.016 --> 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 --> 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 --> 01:01:52.406 Given its last to think about,
NOTE Confidence: 0.83240086

01:01:52.410 --> 01:01:55.560 then thank you very much.
NOTE Confidence: 0.83240086

01:01:55.560 --> 01:01:56.715 Now we'll just take a
NOTE Confidence: 0.83240086

01:01:56.715 --> 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 --> 01:02:01.128 invite you folks please to send
NOTE Confidence: 0.83240086

01:02:01.128 --> 01:02:02.652 your questions in via the Q&A

NOTE Confidence: 0.83240086

01:02:02.710 --> 01:02:04.334 and then I will read him to bed.

NOTE Confidence: 0.82474625

01:02:09.860 --> 01:02:12.240 So then the first question that one

NOTE Confidence: 0.82474625

01:02:12.240 --> 01:02:14.727 of the things that occurred to me,

NOTE Confidence: 0.82474625

01:02:14.730 --> 01:02:17.170 kind of in broad strokes, was the.

NOTE Confidence: 0.9138662

01:02:19.200 --> 01:02:21.868 Is the question about.

NOTE Confidence: 0.9138662

01:02:21.870 --> 01:02:22.356 About paternalism,

NOTE Confidence: 0.9138662

01:02:22.356 --> 01:02:24.057 you know you talked about clinical utility.

NOTE Confidence: 0.9138662

01:02:24.060 --> 01:02:25.986 Of course, that for those of us in the

NOTE Confidence: 0.9138662

01:02:25.986 --> 01:02:27.946 room who are who are mainly clinicians,

NOTE Confidence: 0.9138662

01:02:27.950 --> 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 --> 01:02:31.832 What are you going to do

NOTE Confidence: 0.9138662

01:02:31.832 --> 01:02:32.558 with the information?

NOTE Confidence: 0.9138662

01:02:32.560 --> 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 --> 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 --> 01:02:40.098 we greatly discourage him from doing it.
NOTE Confidence: 0.9138662

01:02:40.100 --> 01:02:42.179 Seems like a poor use of resources and some
NOTE Confidence: 0.9138662

01:02:42.179 --> 01:02:44.469 of these tests carry risks for the kid too.
NOTE Confidence: 0.9138662

01:02:44.470 --> 01:02:45.826 But now with the genetic testing
NOTE Confidence: 0.9138662

01:02:45.826 --> 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 --> 01:02:52.387 X Ray of my kids.
NOTE Confidence: 0.9138662

01:02:52.390 --> 01:02:53.131 Kids need 'cause.
NOTE Confidence: 0.9138662

01:02:53.131 --> 01:02:54.860 I'm curious to see what he looks
NOTE Confidence: 0.9138662

01:02:54.909 --> 01:02:56.439 like when we're not doing that.
NOTE Confidence: 0.9138662

01:02:56.440 --> 01:02:57.700 If there's not clinically indicated,
NOTE Confidence: 0.9138662

01:02:57.700 --> 01:02:59.218 at least we should, right so,
NOTE Confidence: 0.9138662

01:02:59.220 --> 01:03:00.606 so we limit what's available to

NOTE Confidence: 0.9138662

01:03:00.606 --> 01:03:02.409 them and with some of the sequencing

NOTE Confidence: 0.9138662

01:03:02.409 --> 01:03:03.517 that you've referred to.

NOTE Confidence: 0.9138662

01:03:03.520 --> 01:03:05.158 But an awful lot of this information

NOTE Confidence: 0.9138662

01:03:05.158 --> 01:03:07.060 that is kind of optional for parents,

NOTE Confidence: 0.9138662

01:03:07.060 --> 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 --> 01:03:09.337 Not necessarily an increased expense,

NOTE Confidence: 0.9138662

01:03:09.340 --> 01:03:10.792 and certainly not an increase risk

NOTE Confidence: 0.9138662

01:03:10.792 --> 01:03:12.851 of doing the test that the risk of

NOTE Confidence: 0.9138662

01:03:12.851 --> 01:03:14.146 the information is certainly one.

NOTE Confidence: 0.9138662

01:03:14.150 --> 01:03:16.190 But I guess what I'm trying to get at is

NOTE Confidence: 0.9138662

01:03:16.249 --> 01:03:18.447 if parents say 'cause you're talking about.

NOTE Confidence: 0.9138662

01:03:18.450 --> 01:03:20.214 Some people just think more is better.

NOTE Confidence: 0.9138662

01:03:20.220 --> 01:03:21.236 So if parents say,

NOTE Confidence: 0.9138662

01:03:21.236 --> 01:03:22.026 yeah, you know,

NOTE Confidence: 0.9138662

01:03:22.026 --> 01:03:23.634 I'd like to know in particular.
NOTE Confidence: 0.9138662

01:03:23.640 --> 01:03:27.114 If somebody 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 --> 01:03:29.940 of us to say now?
NOTE Confidence: 0.9138662

01:03:29.940 --> 01:03:32.760 I know it's really not good for you to know.
NOTE Confidence: 0.8251437

01:03:34.190 --> 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 --> 01:03:39.269 have to go back to the slide,
NOTE Confidence: 0.8251437

01:03:39.270 --> 01:03:40.770 skipped over towards the end,
NOTE Confidence: 0.8251437

01:03:40.770 --> 01:03:42.444 because that's actually the slides I
NOTE Confidence: 0.8251437

01:03:42.444 --> 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 --> 01:03:47.938 Well, I think there's been a shift.
NOTE Confidence: 0.8251437

01:03:47.940 --> 01:03:50.929 So in other words, I think that.
NOTE Confidence: 0.8251437

01:03:50.930 --> 01:03:53.546 There has been this shift in several ways.
NOTE Confidence: 0.8251437

01:03:53.550 --> 01:03:56.097 One is, I do think just like the example

NOTE Confidence: 0.8251437

01:03:56.097 --> 01:03:58.777 they gave you from zero during pigmentosa,

NOTE Confidence: 0.8251437

01:03:58.780 --> 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 --> 01:04:04.662 we don't know what to do with

NOTE Confidence: 0.8251437

01:04:04.662 --> 01:04:05.314 this information.

NOTE Confidence: 0.8251437

01:04:05.320 --> 01:04:07.350 These families who are part

NOTE Confidence: 0.8251437

01:04:07.350 --> 01:04:09.380 of a close knit group.

NOTE Confidence: 0.8251437

01:04:09.380 --> 01:04:10.755 Felt this information was important

NOTE Confidence: 0.8251437

01:04:10.755 --> 01:04:12.130 and they knew its limitations.

NOTE Confidence: 0.8251437

01:04:12.130 --> 01:04:14.330 They knew that it was uncertain what it

NOTE Confidence: 0.8251437

01:04:14.330 --> 01:04:16.528 meant, but they knew other folks had it.

NOTE Confidence: 0.8251437

01:04:16.530 --> 01:04:17.630 They wanted it to,

NOTE Confidence: 0.8251437

01:04:17.630 --> 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 --> 01:04:21.329 that group resulted in them over

NOTE Confidence: 0.8251437

01:04:21.329 --> 01:04:22.919 the next number of years revising
NOTE Confidence: 0.8251437

01:04:22.919 --> 01:04:24.072 their approaches and getting
NOTE Confidence: 0.8251437

01:04:24.072 --> 01:04:25.328 input from their community.
NOTE Confidence: 0.8251437

01:04:25.330 --> 01:04:25.884 And actually,
NOTE Confidence: 0.8251437

01:04:25.884 --> 01:04:27.546 they made the decision to make
NOTE Confidence: 0.8251437

01:04:27.546 --> 01:04:28.737 that information available with
NOTE Confidence: 0.8251437

01:04:28.737 --> 01:04:30.192 the limitations and with the
NOTE Confidence: 0.8251437

01:04:30.192 --> 01:04:31.594 explanations of what this may
NOTE Confidence: 0.8251437

01:04:31.594 --> 01:04:33.028 mean with the limitations are so.
NOTE Confidence: 0.8251437

01:04:33.030 --> 01:04:34.126 That's an example where,
NOTE Confidence: 0.8251437

01:04:34.126 --> 01:04:36.878 but I think the one of the issues I want.
NOTE Confidence: 0.8251437

01:04:36.880 --> 01:04:38.614 That's a very specific group of
NOTE Confidence: 0.8251437

01:04:38.614 --> 01:04:40.849 people who are high risk of a disease.
NOTE Confidence: 0.8251437

01:04:40.850 --> 01:04:43.550 Where it was meaningful to them to do that,
NOTE Confidence: 0.8251437

01:04:43.550 --> 01:04:45.384 I think other questions come up when
NOTE Confidence: 0.8251437

01:04:45.384 --> 01:04:47.373 we get the thing with the context

NOTE Confidence: 0.8251437

01:04:47.373 --> 01:04:49.481 of when is people are people asking

NOTE Confidence: 0.8251437

01:04:49.481 --> 01:04:51.868 for this information and I want to

NOTE Confidence: 0.8251437

01:04:51.868 --> 01:04:53.094 distinguish between the random.

NOTE Confidence: 0.8251437

01:04:53.094 --> 01:04:55.270 And So what I want to clarify is

NOTE Confidence: 0.8251437

01:04:55.339 --> 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 --> 01:05:02.502 everything for everything that we've

NOTE Confidence: 0.8251437

01:05:02.502 --> 01:05:03.522 done for reasons for particular

NOTE Confidence: 0.8251437

01:05:03.522 --> 01:05:04.640 reasons may be good reasons,

NOTE Confidence: 0.8251437

01:05:04.640 --> 01:05:05.321 maybe bad reasons,

NOTE Confidence: 0.8251437

01:05:05.321 --> 01:05:06.683 but there are currently based reasons.

NOTE Confidence: 0.8251437

01:05:06.690 --> 01:05:07.890 But the question is,

NOTE Confidence: 0.8251437

01:05:07.890 --> 01:05:09.390 what should the scope be?

NOTE Confidence: 0.8251437

01:05:09.390 --> 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 --> 01:05:14.130 opportunity to add additional tests,
NOTE Confidence: 0.8251437

01:05:14.130 --> 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 --> 01:05:18.206 These actionable tests include tests that
NOTE Confidence: 0.8251437

01:05:18.206 --> 01:05:20.130 ensure the only develop in adulthood,
NOTE Confidence: 0.8251437

01:05:20.130 --> 01:05:21.394 and that's actually the
NOTE Confidence: 0.8251437

01:05:21.394 --> 01:05:22.658 question you're getting at.
NOTE Confidence: 0.8251437

01:05:22.660 --> 01:05:26.548 Mark and I want to share my screen again.
NOTE Confidence: 0.8251437

01:05:26.550 --> 01:05:27.270 And.
NOTE Confidence: 0.897526

01:05:29.400 --> 01:05:32.184 See if I can get to this here.
NOTE Confidence: 0.897526

01:05:32.190 --> 01:05:35.590 I want to go back to one slide but when I.
NOTE Confidence: 0.897526

01:05:35.590 --> 01:05:38.298 Skipped over right here.
NOTE Confidence: 0.897526

01:05:38.300 --> 01:05:41.036 So one of the things that.
NOTE Confidence: 0.897526

01:05:41.040 --> 01:05:42.408 That are the consortium

NOTE Confidence: 0.897526
01:05:42.408 --> 01:05:44.118 is doing this time around,
NOTE Confidence: 0.897526
01:05:44.120 --> 01:05:46.166 just like show the consent paper.
NOTE Confidence: 0.897526
01:05:46.170 --> 01:05:47.398 This is a project.
NOTE Confidence: 0.897526
01:05:47.398 --> 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 --> 01:05:55.145 at Baylor and a bunch of other Anna
NOTE Confidence: 0.897526
01:05:55.145 --> 01:05:57.696 Bunch of other folks in which there
NOTE Confidence: 0.897526
01:05:57.696 --> 01:05:59.806 we are trying to collectively.
NOTE Confidence: 0.897526
01:05:59.810 --> 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 --> 01:06:06.473 patients for genetics and the point
NOTE Confidence: 0.897526
01:06:06.473 --> 01:06:08.700 of this model is to suggest that
NOTE Confidence: 0.897526
01:06:08.700 --> 01:06:10.536 there are a range of utilities
NOTE Confidence: 0.897526
01:06:10.536 --> 01:06:12.266 and that we can distinguish,
NOTE Confidence: 0.897526
01:06:12.270 --> 01:06:14.853 and we spent a lot of time just like
NOTE Confidence: 0.897526

01:06:14.853 --> 01:06:17.148 the issue of categories that describe
NOTE Confidence: 0.897526

01:06:17.148 --> 01:06:19.880 for the for the next Gen paper,
NOTE Confidence: 0.897526

01:06:19.880 --> 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 --> 01:06:25.480 and we did interviews with participants
NOTE Confidence: 0.897526

01:06:25.541 --> 01:06:27.761 in the various Caesar studies about
NOTE Confidence: 0.897526

01:06:27.761 --> 01:06:29.599 their experience with genetic testing
NOTE Confidence: 0.897526

01:06:29.599 --> 01:06:31.975 in order to come up with these domains.
NOTE Confidence: 0.897526

01:06:31.980 --> 01:06:33.312 Of perceived utility.
NOTE Confidence: 0.897526

01:06:33.312 --> 01:06:35.976 One of those domains is clinical.
NOTE Confidence: 0.897526

01:06:35.980 --> 01:06:38.092 The way in which the information will you
NOTE Confidence: 0.897526

01:06:38.092 --> 01:06:39.969 know either established new diagnosis,
NOTE Confidence: 0.897526

01:06:39.970 --> 01:06:41.460 informed clinical management may result
NOTE Confidence: 0.897526

01:06:41.460 --> 01:06:43.959 in referral to a trial may allow screening.
NOTE Confidence: 0.897526

01:06:43.960 --> 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 --> 01:06:47.820 but we also learned was that
NOTE Confidence: 0.897526
01:06:47.820 --> 01:06:49.800 there were range of other values,
NOTE Confidence: 0.897526
01:06:49.800 --> 01:06:51.636 and some of these are positive.
NOTE Confidence: 0.897526
01:06:51.640 --> 01:06:53.170 Some of these are negative.
NOTE Confidence: 0.897526
01:06:53.170 --> 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 --> 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 --> 01:07:02.354 and I think the point here is simply
NOTE Confidence: 0.897526
01:07:02.354 --> 01:07:04.623 acknowledge that one of the impacts of
NOTE Confidence: 0.897526
01:07:04.623 --> 01:07:06.460 testing with positive thinking can be.
NOTE Confidence: 0.897526
01:07:06.460 --> 01:07:09.076 An emotional impact and.
NOTE Confidence: 0.897526
01:07:09.076 --> 01:07:09.730 Additionally,
NOTE Confidence: 0.897526
01:07:09.730 --> 01:07:12.140 there can be behavioral impacts, and again,
NOTE Confidence: 0.897526
01:07:12.140 --> 01:07:14.210 here we're talking about behavioral impacts.
NOTE Confidence: 0.897526

01:07:14.210 --> 01:07:16.268 They're not the same as clinical,
NOTE Confidence: 0.897526

01:07:16.270 --> 01:07:17.514 but might have meaning,
NOTE Confidence: 0.897526

01:07:17.514 --> 01:07:19.380 and here we include things like
NOTE Confidence: 0.897526

01:07:19.439 --> 01:07:20.738 changing health habits,
NOTE Confidence: 0.897526

01:07:20.740 --> 01:07:21.428 future planning,
NOTE Confidence: 0.897526

01:07:21.428 --> 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 --> 01:07:28.315 These are things that people
NOTE Confidence: 0.897526

01:07:28.315 --> 01:07:30.412 might do with the information on
NOTE Confidence: 0.897526

01:07:30.412 --> 01:07:31.748 the issue of curiosity.
NOTE Confidence: 0.897526

01:07:31.750 --> 01:07:34.494 I think we would describe as cognitive is.
NOTE Confidence: 0.897526

01:07:34.500 --> 01:07:35.529 In other words,
NOTE Confidence: 0.897526

01:07:35.529 --> 01:07:37.244 it might impact their understanding.
NOTE Confidence: 0.897526

01:07:37.250 --> 01:07:40.616 It might be satisfying their curiosity.

NOTE Confidence: 0.897526

01:07:40.620 --> 01:07:43.140 And it may be relevant to the how they view

NOTE Confidence: 0.897526

01:07:43.199 --> 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 --> 01:07:49.130 impact of of testing that may.

NOTE Confidence: 0.897526

01:07:49.130 --> 01:07:50.120 So, for example,

NOTE Confidence: 0.897526

01:07:50.120 --> 01:07:52.430 one of the things that we learned

NOTE Confidence: 0.897526

01:07:52.499 --> 01:07:54.480 and it not only in this state,

NOTE Confidence: 0.897526

01:07:54.480 --> 01:07:55.383 but previous studies,

NOTE Confidence: 0.897526

01:07:55.383 --> 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 --> 01:08:00.402 parents with rare diseases,

NOTE Confidence: 0.897526

01:08:00.402 --> 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 --> 01:08:06.450 Its way of having access to social services.

NOTE Confidence: 0.897526

01:08:06.450 --> 01:08:08.382 When I was at the NIH working

NOTE Confidence: 0.897526

01:08:08.382 --> 01:08:09.920 in the Genome Institute,
NOTE Confidence: 0.897526

01:08:09.920 --> 01:08:11.196 they would see patients.
NOTE Confidence: 0.897526

01:08:11.196 --> 01:08:13.110 With rare diseases and it turned
NOTE Confidence: 0.897526

01:08:13.166 --> 01:08:14.840 out one of the most stressful
NOTE Confidence: 0.897526

01:08:14.840 --> 01:08:16.602 things when a family would come
NOTE Confidence: 0.897526

01:08:16.602 --> 01:08:18.348 in with a perceived rare disease
NOTE Confidence: 0.897526

01:08:18.348 --> 01:08:20.090 in which the Jess would say,
NOTE Confidence: 0.897526

01:08:20.090 --> 01:08:20.380 hey,
NOTE Confidence: 0.862932

01:08:20.380 --> 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 --> 01:08:26.178 you thought you had is actually this,
NOTE Confidence: 0.862932

01:08:26.180 --> 01:08:28.210 or we don't know what it is,
NOTE Confidence: 0.862932

01:08:28.210 --> 01:08:30.303 but it's not that and people were
NOTE Confidence: 0.862932

01:08:30.303 --> 01:08:32.084 often unhappy to be told they
NOTE Confidence: 0.862932

01:08:32.084 --> 01:08:34.009 were no longer part of the group,
NOTE Confidence: 0.862932

01:08:34.010 --> 01:08:36.188 and so the point here Mark

NOTE Confidence: 0.862932

01:08:36.188 --> 01:08:38.920 is that is to suggest that.

NOTE Confidence: 0.862932

01:08:38.920 --> 01:08:41.216 There is a range of value of this.

NOTE Confidence: 0.862932

01:08:41.220 --> 01:08:43.074 One of the other things that

NOTE Confidence: 0.862932

01:08:43.074 --> 01:08:44.758 the Caesar consortium is doing

NOTE Confidence: 0.862932

01:08:44.758 --> 01:08:46.298 is interacting with payers.

NOTE Confidence: 0.862932

01:08:46.300 --> 01:08:48.005 People who provide the coverage

NOTE Confidence: 0.862932

01:08:48.005 --> 01:08:50.580 for testing and the try to at least

NOTE Confidence: 0.862932

01:08:50.580 --> 01:08:52.854 have on the agenda that some of the

NOTE Confidence: 0.862932

01:08:52.854 --> 01:08:54.559 value of testing for individuals

NOTE Confidence: 0.862932

01:08:54.559 --> 01:08:56.251 is not simply only clinical,

NOTE Confidence: 0.862932

01:08:56.251 --> 01:08:58.177 but it's these other ranges too.

NOTE Confidence: 0.862932

01:08:58.180 --> 01:09:00.100 And the question is, you know,

NOTE Confidence: 0.862932

01:09:00.100 --> 01:09:02.252 where do we fit this in two questions

NOTE Confidence: 0.862932

01:09:02.252 --> 01:09:03.630 about resource prioritization?

NOTE Confidence: 0.862932

01:09:03.630 --> 01:09:05.240 And I don't want to.

NOTE Confidence: 0.862932

01:09:05.240 --> 01:09:07.166 I can't answer that for sure,
NOTE Confidence: 0.862932

01:09:07.170 --> 01:09:10.095 but I think the idea is to at least
NOTE Confidence: 0.862932

01:09:10.095 --> 01:09:12.615 suggest it ought to be on the table,
NOTE Confidence: 0.862932

01:09:12.620 --> 01:09:14.685 and I wouldn't suggest that
NOTE Confidence: 0.862932

01:09:14.685 --> 01:09:16.337 testing simply for that.
NOTE Confidence: 0.862932

01:09:16.340 --> 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 --> 01:09:22.576 and I think the best example of that,
NOTE Confidence: 0.862932

01:09:22.580 --> 01:09:25.966 frankly, is. Yeah, I do have a diagnosis.
NOTE Confidence: 0.862932

01:09:25.970 --> 01:09:26.672 Is that really?
NOTE Confidence: 0.862932

01:09:26.672 --> 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 --> 01:09:30.960 we can simply say you've
NOTE Confidence: 0.862932

01:09:30.960 --> 01:09:32.384 got a rare condition.
NOTE Confidence: 0.862932

01:09:32.390 --> 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 --> 01:09:37.729 Say we don't?
NOTE Confidence: 0.7978275
01:09:39.780 --> 01:09:41.100 We shouldn't pay for that.
NOTE Confidence: 0.7978275
01:09:41.100 --> 01:09:42.195 Do anything with.
NOTE Confidence: 0.7978275
01:09:42.195 --> 01:09:43.655 We usually will say.
NOTE Confidence: 0.7978275
01:09:43.660 --> 01:09:45.196 You know that that may be
NOTE Confidence: 0.7978275
01:09:45.196 --> 01:09:46.220 valuable to the person,
NOTE Confidence: 0.7978275
01:09:46.220 --> 01:09:47.500 and therefore it's worth doing.
NOTE Confidence: 0.7978275
01:09:47.500 --> 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 --> 01:09:50.320 clinical sequencing, but
NOTE Confidence: 0.81839126
01:09:50.320 --> 01:09:51.565 maybe not sometimes these tests
NOTE Confidence: 0.81839126
01:09:51.565 --> 01:09:53.122 are not to satisfy the curiosity
NOTE Confidence: 0.81839126
01:09:53.122 --> 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 --> 01:09:57.945 where this isn't actually going to
NOTE Confidence: 0.81839126

01:09:57.945 --> 01:09:59.527 help us manage this patient at all,
NOTE Confidence: 0.81839126

01:09:59.530 --> 01:10:00.343 but it'll it'll.
NOTE Confidence: 0.81839126

01:10:00.343 --> 01:10:01.969 Best Buy sometimes our curiosity and
NOTE Confidence: 0.81839126

01:10:01.969 --> 01:10:03.688 that that's a whole separate issue,
NOTE Confidence: 0.81839126

01:10:03.690 --> 01:10:05.786 and as far as the the the the,
NOTE Confidence: 0.81839126

01:10:05.790 --> 01:10:07.624 the emotional outcomes or benefits of this.
NOTE Confidence: 0.81839126

01:10:07.630 --> 01:10:09.438 I mean, I'd say I remember that when
NOTE Confidence: 0.81839126

01:10:09.438 --> 01:10:11.415 the first time with my second child
NOTE Confidence: 0.81839126

01:10:11.415 --> 01:10:12.890 was our first prenatal ultrasound,
NOTE Confidence: 0.81839126

01:10:12.890 --> 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 --> 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 --> 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 --> 01:10:30.880 if you know what I mean.
NOTE Confidence: 0.81839126
01:10:30.880 --> 01:10:32.105 If somebody else is so
NOTE Confidence: 0.81839126
01:10:32.105 --> 01:10:33.085 you about other people,
NOTE Confidence: 0.81839126
01:10:33.090 --> 01:10:34.738 but it could be the same with the
NOTE Confidence: 0.81839126
01:10:34.738 --> 01:10:36.192 IT could potentially be the same
NOTE Confidence: 0.81839126
01:10:36.192 --> 01:10:38.296 with some people to say to their to
NOTE Confidence: 0.81839126
01:10:38.296 --> 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 --> 01:10:43.815 I want to be aware of if nobody knows
NOTE Confidence: 0.81839126
01:10:43.815 --> 01:10:46.125 if it's, you know if it's in the dark.
NOTE Confidence: 0.81839126
01:10:46.130 --> 01:10:47.360 We've never looked at that.
NOTE Confidence: 0.81839126
01:10:47.360 --> 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 --> 01:10:51.018 potentially for the patient
NOTE Confidence: 0.81839126

01:10:51.018 --> 01:10:53.128 or the research subject then.
NOTE Confidence: 0.81839126

01:10:53.130 --> 01:10:55.629 Then then you know somebody else knows
NOTE Confidence: 0.81839126

01:10:55.629 --> 01:10:58.030 it and excuse me, sorry bout that.
NOTE Confidence: 0.8288017

01:11:03.570 --> 01:11:04.810 Sorry about what happened.
NOTE Confidence: 0.8288017

01:11:04.810 --> 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 --> 01:11:11.320 me answer your second question first.
NOTE Confidence: 0.8288017

01:11:11.320 --> 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 --> 01:11:17.208 we've got a couple of questions to get to.
NOTE Confidence: 0.8288017

01:11:17.210 --> 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 --> 01:11:22.018 the argument for doing genetic
NOTE Confidence: 0.8288017

01:11:22.018 --> 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?

NOTE Confidence: 0.81407756

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 --> 01:11:36.938 thinking about newborn screening and

NOTE Confidence: 0.81407756

01:11:36.938 --> 01:11:39.086 thinking about sequencing and the role

NOTE Confidence: 0.81407756

01:11:39.086 --> 01:11:41.519 of sequencing with the newborn screening.

NOTE Confidence: 0.81407756

01:11:41.520 --> 01:11:42.426 And So what?

NOTE Confidence: 0.81407756

01:11:42.426 --> 01:11:45.468 I want to comment is that is that there

NOTE Confidence: 0.81407756

01:11:45.468 --> 01:11:48.457 is sometimes sequencing done as part of

NOTE Confidence: 0.81407756

01:11:48.457 --> 01:11:51.170 newborn screening as a secondary test.

NOTE Confidence: 0.81407756

01:11:51.170 --> 01:11:52.954 So in other words,

NOTE Confidence: 0.81407756

01:11:52.954 --> 01:11:55.184 by using the various biochemical

NOTE Confidence: 0.81407756

01:11:55.184 --> 01:11:58.215 tests or even genetic tests that once.

NOTE Confidence: 0.81407756

01:11:58.220 --> 01:12:01.380 That some.
NOTE Confidence: 0.81407756

01:12:01.380 --> 01:12:03.100 Currently in fact some some
NOTE Confidence: 0.81407756

01:12:03.100 --> 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 --> 01:12:09.422 This has to be done very thoughtfully
NOTE Confidence: 0.81407756

01:12:09.422 --> 01:12:12.046 and I'm going to give you a very
NOTE Confidence: 0.81407756

01:12:12.046 --> 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 --> 01:12:23.739 or in the state of California.
NOTE Confidence: 0.81407756

01:12:23.740 --> 01:12:26.398 Signal sequencing is done on every.
NOTE Confidence: 0.81407756

01:12:26.400 --> 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
NOTE Confidence: 0.81407756
01:12:33.862 --> 01:12:36.431 is that they identify many more
NOTE Confidence: 0.81407756
01:12:36.431 --> 01:12:38.371 individuals who have variants
NOTE Confidence: 0.81407756
01:12:38.371 --> 01:12:39.826 of unknown significance.
NOTE Confidence: 0.81407756
01:12:39.830 --> 01:12:42.672 And in cystic fibrosis they have a
NOTE Confidence: 0.81407756
01:12:42.672 --> 01:12:45.796 term that they use when you have one.
NOTE Confidence: 0.81407756
01:12:45.800 --> 01:12:47.870 Of the genes for cystic fibrosis.
NOTE Confidence: 0.81407756
01:12:47.870 --> 01:12:50.306 One of the variants that can cause
NOTE Confidence: 0.81407756
01:12:50.306 --> 01:12:52.026 cystic fibrosis and a variant
NOTE Confidence: 0.81407756
01:12:52.026 --> 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 --> 01:12:59.383 And you have a normal sweat test.
NOTE Confidence: 0.81407756
01:12:59.390 --> 01:13:01.757 They then go ahead and say that you have
NOTE Confidence: 0.81407756
01:13:01.757 --> 01:13:04.287 a cystic fibrosis metabolic syndrome,
NOTE Confidence: 0.81407756

01:13:04.290 --> 01:13:06.040 which means that you don't
NOTE Confidence: 0.81407756

01:13:06.040 --> 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 --> 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 --> 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 --> 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 --> 01:13:29.188 had this happen to Ann for her.
NOTE Confidence: 0.81407756

01:13:29.190 --> 01:13:31.560 This was a big worry.
NOTE Confidence: 0.81407756

01:13:31.560 --> 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 --> 01:13:37.372 have cystic fibrosis,

NOTE Confidence: 0.81407756

01:13:37.380 --> 01:13:39.417 but maybe he could get it at

NOTE Confidence: 0.81407756

01:13:39.417 --> 01:13:41.317 some point for her was weighed

NOTE Confidence: 0.81407756

01:13:41.317 --> 01:13:43.956 very heavily on her and for her I

NOTE Confidence: 0.81407756

01:13:43.956 --> 01:13:45.990 think was very distressful to me.

NOTE Confidence: 0.81407756

01:13:45.990 --> 01:13:47.067 With this means,

NOTE Confidence: 0.81407756

01:13:47.067 --> 01:13:48.862 I actually think that newborn

NOTE Confidence: 0.81407756

01:13:48.862 --> 01:13:50.717 screening is an area where we

NOTE Confidence: 0.81407756

01:13:50.717 --> 01:13:52.690 have to be very careful for that,

NOTE Confidence: 0.81407756

01:13:52.690 --> 01:13:54.290 not only for that reason,

NOTE Confidence: 0.81407756

01:13:54.290 --> 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 newborn screening

NOTE Confidence: 0.81407756

01:13:58.769 --> 01:14:01.749 shifting to a sequencing based model.

NOTE Confidence: 0.81407756

01:14:01.750 --> 01:14:03.958 As the first place this goes,

NOTE Confidence: 0.81407756

01:14:03.960 --> 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 --> 01:14:10.274 careful thought about the curation
NOTE Confidence: 0.81407756

01:14:10.274 --> 01:14:12.414 of the information making decisions,
NOTE Confidence: 0.81407756

01:14:12.420 --> 01:14:13.892 which variants to report,
NOTE Confidence: 0.81407756

01:14:13.892 --> 01:14:17.250 which diseases to consider, how to do this.
NOTE Confidence: 0.81407756

01:14:17.250 --> 01:14:19.130 So I actually think.
NOTE Confidence: 0.81407756

01:14:19.130 --> 01:14:21.674 In 20 years we will be doing that,
NOTE Confidence: 0.81407756

01:14:21.680 --> 01:14:23.815 but I hope that the process to
NOTE Confidence: 0.81407756

01:14:23.815 --> 01:14:26.002 get there will have allowed us to
NOTE Confidence: 0.81407756

01:14:26.002 --> 01:14:29.020 develop how to do this in a thoughtful way.
NOTE Confidence: 0.81407756

01:14:29.020 --> 01:14:30.928 I also think that it raises,
NOTE Confidence: 0.81407756

01:14:30.930 --> 01:14:32.454 as has been discussed,
NOTE Confidence: 0.81407756

01:14:32.454 --> 01:14:34.740 for newborn screening in general raises
NOTE Confidence: 0.87807447

01:14:34.803 --> 01:14:36.643 new questions about informed consent
NOTE Confidence: 0.87807447

01:14:36.643 --> 01:14:38.860 in the following way is that.
NOTE Confidence: 0.87807447

01:14:38.860 --> 01:14:40.610 Historically, newborn screening is often

NOTE Confidence: 0.87807447

01:14:40.610 --> 01:14:42.790 done without much discussion with parents,

NOTE Confidence: 0.87807447

01:14:42.790 --> 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 --> 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 --> 01:14:53.040 and I suspect if sequencing becomes

NOTE Confidence: 0.87807447

01:14:53.040 --> 01:14:56.320 part of it will be one more reason

NOTE Confidence: 0.87807447

01:14:56.320 --> 01:14:59.232 why there will be more education and

NOTE Confidence: 0.87807447

01:14:59.232 --> 01:15:01.980 more engagement at that time. So.

NOTE Confidence: 0.8257083

01:15:03.110 --> 01:15:05.336 Thank you the next question please,

NOTE Confidence: 0.8257083

01:15:05.340 --> 01:15:07.643 how do you counsel parents when you

NOTE Confidence: 0.8257083

01:15:07.643 --> 01:15:09.809 identify mutations with variable penetrance,

NOTE Confidence: 0.8257083

01:15:09.810 --> 01:15:12.036 even if predicted to be pathogenic?

NOTE Confidence: 0.9043739

01:15:14.080 --> 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,

NOTE Confidence: 0.9043739

01:15:20.864 --> 01:15:23.430 it's more common. Because.
NOTE Confidence: 0.92042774

01:15:25.640 --> 01:15:30.938 There's many circumstances where the outcome.
NOTE Confidence: 0.92042774

01:15:30.940 --> 01:15:33.516 It's not clear what that's going to be,
NOTE Confidence: 0.92042774

01:15:33.520 --> 01:15:35.837 so let's take cystic fibrosis and diabetes
NOTE Confidence: 0.92042774

01:15:35.837 --> 01:15:37.733 and thinking about typically about two
NOTE Confidence: 0.92042774

01:15:37.733 --> 01:15:40.115 or three year olds for four year olds
NOTE Confidence: 0.92042774

01:15:40.115 --> 01:15:42.209 who are diagnosed with these conditions.
NOTE Confidence: 0.8497331

01:15:45.460 --> 01:15:47.320 What is variable? I mean,
NOTE Confidence: 0.8497331

01:15:47.320 --> 01:15:50.296 use patterns as an example as a metaphor.
NOTE Confidence: 0.8497331

01:15:50.300 --> 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 --> 01:15:56.250 have who will do really well.
NOTE Confidence: 0.8497331

01:15:56.250 --> 01:15:58.476 Other children who will do who
NOTE Confidence: 0.8497331

01:15:58.476 --> 01:16:00.604 have challenges, and so it's so.
NOTE Confidence: 0.8497331

01:16:00.604 --> 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 --> 01:16:06.840 I think, is part and parcel

NOTE Confidence: 0.8497331

01:16:06.840 --> 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 --> 01:16:12.239 that uncertainty is debilitating,

NOTE Confidence: 0.8497331

01:16:12.240 --> 01:16:14.550 but for many people they really.

NOTE Confidence: 0.8497331

01:16:14.550 --> 01:16:15.405 Adapt to that,

NOTE Confidence: 0.8497331

01:16:15.405 --> 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 --> 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 --> 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 --> 01:16:29.644 the feeding tube and you know what

NOTE Confidence: 0.8497331

01:16:29.644 --> 01:16:32.168 I can tell them is that I don't know
NOTE Confidence: 0.8497331

01:16:32.168 --> 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 --> 01:16:37.157 chance that you will be off all
NOTE Confidence: 0.8497331

01:16:37.157 --> 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 --> 01:16:41.902 off the support that you have
NOTE Confidence: 0.8497331

01:16:41.902 --> 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 --> 01:16:46.490 The the We don't know,
NOTE Confidence: 0.8497331

01:16:46.490 --> 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 --> 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 --> 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 --> 01:17:03.945 think for me that's a critical issue

NOTE Confidence: 0.8497331

01:17:03.945 --> 01:17:05.432 as relates to genetic information.

NOTE Confidence: 0.8497331

01:17:05.432 --> 01:17:07.220 Is the notion of follow up

NOTE Confidence: 0.8497331

01:17:07.274 --> 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 --> 01:17:12.208 The fact that the family feels

NOTE Confidence: 0.8497331

01:17:12.208 --> 01:17:13.812 like they're connected to somebody

NOTE Confidence: 0.8497331

01:17:13.812 --> 01:17:15.696 whom they can ask questions of.

NOTE Confidence: 0.8497331

01:17:15.700 --> 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 --> 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 --> 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 --> 01:17:36.360 trying to trying to figure out that.
NOTE Confidence: 0.8476957

01:17:36.360 --> 01:17:37.680 Is this something people want.
NOTE Confidence: 0.8476957

01:17:37.680 --> 01:17:39.507 I'd be curious to know if there are data
NOTE Confidence: 0.8476957

01:17:39.507 --> 01:17:41.584 or if you've got personal experience with
NOTE Confidence: 0.8476957

01:17:41.584 --> 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 --> 01:17:46.430 trying to figure out what they would want,
NOTE Confidence: 0.8476957

01:17:46.430 --> 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 --> 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 --> 01:18:00.930 actually, we certainly

NOTE Confidence: 0.8225839

01:18:00.930 --> 01:18:02.910 have done a lot of that.

NOTE Confidence: 0.8225839

01:18:02.910 --> 01:18:05.206 We, you know, one of the things

NOTE Confidence: 0.8225839

01:18:05.206 --> 01:18:07.840 that we do as part of our vote,

NOTE Confidence: 0.8225839

01:18:07.840 --> 01:18:09.490 both the next Gen study,

NOTE Confidence: 0.8225839

01:18:09.490 --> 01:18:11.842 which was a carrier study as well

NOTE Confidence: 0.8225839

01:18:11.842 --> 01:18:14.634 as the charm is we do a range of

NOTE Confidence: 0.8225839

01:18:14.634 --> 01:18:16.297 of the side surveys interviews

NOTE Confidence: 0.8225839

01:18:16.297 --> 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 --> 01:18:23.769 how that experience went with

NOTE Confidence: 0.8225839

01:18:23.769 --> 01:18:25.689 a follow the information and we

NOTE Confidence: 0.8225839

01:18:25.689 --> 01:18:27.880 also have done that where we take
NOTE Confidence: 0.8225839

01:18:27.880 --> 01:18:29.440 people who got negative results.
NOTE Confidence: 0.8225839

01:18:29.440 --> 01:18:30.990 Positive results were uncertain results.
NOTE Confidence: 0.8225839

01:18:30.990 --> 01:18:33.286 We also have a separate set of interviews
NOTE Confidence: 0.8225839

01:18:33.286 --> 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 --> 01:18:41.195 and so it's a way of trying to understand
NOTE Confidence: 0.8225839

01:18:41.195 --> 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 --> 01:18:47.847 And this may be again even in this
NOTE Confidence: 0.8225839

01:18:47.847 --> 01:18:49.847 context of this selection bias,
NOTE Confidence: 0.8225839

01:18:49.850 --> 01:18:52.130 because people had to sign up for this
NOTE Confidence: 0.8225839

01:18:52.130 --> 01:18:54.457 and that most people don't regret it.
NOTE Confidence: 0.8225839

01:18:54.460 --> 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 --> 01:18:59.672 They figure out how to sort

NOTE Confidence: 0.8225839

01:18:59.672 --> 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 --> 01:19:04.196 while I still am sceptical and wanted

NOTE Confidence: 0.8225839

01:19:04.196 --> 01:19:06.440 and really want to urge caution.

NOTE Confidence: 0.8225839

01:19:06.440 --> 01:19:09.079 With how we roll this out carefully,

NOTE Confidence: 0.8225839

01:19:09.080 --> 01:19:10.912 we do it well.

NOTE Confidence: 0.8225839

01:19:10.912 --> 01:19:14.240 I'm less worried that most people have.

NOTE Confidence: 0.8225839

01:19:14.240 --> 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 --> 01:19:18.237 but I believe that the way we

NOTE Confidence: 0.8225839

01:19:18.237 --> 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 --> 01:19:24.380 and also by tempering our enthusiasm.

NOTE Confidence: 0.8225839

01:19:24.380 --> 01:19:26.660 And I think the example of back to

NOTE Confidence: 0.8225839

01:19:26.660 --> 01:19:28.808 the issue of newborn screening is

NOTE Confidence: 0.8225839

01:19:28.808 --> 01:19:31.064 really a good example where that's
NOTE Confidence: 0.8225839

01:19:31.137 --> 01:19:33.272 probably one of the last places I
NOTE Confidence: 0.8225839

01:19:33.272 --> 01:19:35.266 would want to go for doing this,
NOTE Confidence: 0.8225839

01:19:35.266 --> 01:19:37.716 but in the context of of somebody who
NOTE Confidence: 0.8225839

01:19:37.716 --> 01:19:40.397 is at risk for hereditary cancer syndrome,
NOTE Confidence: 0.8225839

01:19:40.400 --> 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 --> 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 somebody
NOTE Confidence: 0.8225839

01:19:48.452 --> 01:19:49.493 has testing done,
NOTE Confidence: 0.8225839

01:19:49.500 --> 01:19:51.936 even if the result is an unexpected
NOTE Confidence: 0.8225839

01:19:51.936 --> 01:19:53.330 actual finding somewhere else.
NOTE Confidence: 0.8225839

01:19:53.330 --> 01:19:55.070 They're usually open to that
NOTE Confidence: 0.8225839

01:19:55.070 --> 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 --> 01:20:02.548 Doctor Ben will fund this advance

NOTE Confidence: 0.9123243

01:20:02.548 --> 01:20:03.352 tremendous session tonight.

NOTE Confidence: 0.9123243

01:20:03.360 --> 01:20:04.440 We really appreciate it.

NOTE Confidence: 0.9123243

01:20:04.440 --> 01:20:06.590 We're sorry we couldn't do this in person,

NOTE Confidence: 0.9123243

01:20:06.590 --> 01:20:09.292 but sometime down the road we will

NOTE Confidence: 0.9123243

01:20:09.292 --> 01:20:11.688 absolutely get you to come join us.

NOTE Confidence: 0.9123243

01:20:11.690 --> 01:20:13.322 And thank you very much and I thank

NOTE Confidence: 0.9123243

01:20:13.322 --> 01:20:15.157 you all for attending on the website.

NOTE Confidence: 0.9123243

01:20:15.160 --> 01:20:16.546 You can see when the upcoming

NOTE Confidence: 0.9123243

01:20:16.546 --> 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 --> 01:20:21.257 to dinner and but then you'd have a

NOTE Confidence: 0.9123243

01:20:21.257 --> 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 --> 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 --> 01:20:32.468 This is true. It's a pleasure to

NOTE Confidence: 0.8262469

01:20:32.468 --> 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 --> 01:20:38.220 time. See you next time.

NOTE Confidence: 0.8262469

01:20:38.220 --> 01:20:41.637 Thanks folks. Goodnight