

WEBVTT

NOTE duration:"00:19:53.7610000"

NOTE language:en-us

NOTE Confidence: 0.78761697

00:00:00.000 --> 00:00:02.562 It's my great pleasure to now

NOTE Confidence: 0.78761697

00:00:02.562 --> 00:00:04.270 introduce our next speaker,

NOTE Confidence: 0.78761697

00:00:04.270 --> 00:00:05.461 Doctor Yunk Lijiang.

NOTE Confidence: 0.78761697

00:00:05.461 --> 00:00:08.240 Doctor Zhang is a professor of genetics,

NOTE Confidence: 0.78761697

00:00:08.240 --> 00:00:09.359 neuroscience, and Pediatrics,

NOTE Confidence: 0.78761697

00:00:09.359 --> 00:00:12.610 as well as a chief of medical genetics.

NOTE Confidence: 0.78761697

00:00:12.610 --> 00:00:16.174 He joined Yale in 2019 from Duke at Duke.

NOTE Confidence: 0.78761697

00:00:16.180 --> 00:00:18.884 He was a key member of the NIH

NOTE Confidence: 0.78761697

00:00:18.884 --> 00:00:20.497 Undiagnosed Disease Network and

NOTE Confidence: 0.78761697

00:00:20.497 --> 00:00:22.662 his research interests are genomics

NOTE Confidence: 0.78761697

00:00:22.662 --> 00:00:24.910 of rare diseases and modeling.

NOTE Confidence: 0.78761697

00:00:24.910 --> 00:00:26.104 The neurodevelopmental disorders

NOTE Confidence: 0.78761697

00:00:26.104 --> 00:00:27.298 using functional approaches.

NOTE Confidence: 0.78761697

00:00:27.300 --> 00:00:30.079 The floor is now yours Doctor Young.

NOTE Confidence: 0.63141394

00:00:36.050 --> 00:00:39.116 Thank you Nicole for kind introduction on.

NOTE Confidence: 0.63141394

00:00:39.120 --> 00:00:41.320 I'm taking about 15 minutes.

NOTE Confidence: 0.63141394

00:00:41.320 --> 00:00:43.956 You can introduce you kind of

NOTE Confidence: 0.63141394

00:00:43.956 --> 00:00:46.590 class backed off the genomic spoj.

NOTE Confidence: 0.7496509

00:00:50.350 --> 00:00:54.370 You got everyone's same page.

NOTE Confidence: 0.7496509

00:00:54.370 --> 00:00:57.340 Then I have no conflict, and indeed,

NOTE Confidence: 0.7496509

00:00:57.340 --> 00:01:00.308 and the undiagnosed disease, a rare disease.

NOTE Confidence: 0.7496509

00:01:00.310 --> 00:01:02.848 Bilateral mean is easy to understand,

NOTE Confidence: 0.7496509

00:01:02.850 --> 00:01:05.094 but in I actually indeed have

NOTE Confidence: 0.7496509

00:01:05.094 --> 00:01:07.174 us very specific and terminology

NOTE Confidence: 0.7496509

00:01:07.174 --> 00:01:09.206 to describe this term,

NOTE Confidence: 0.7496509

00:01:09.210 --> 00:01:11.330 which is basically sweet term.

NOTE Confidence: 0.7496509

00:01:11.330 --> 00:01:14.298 It's a real previous can lung disease,

NOTE Confidence: 0.7496509

00:01:14.300 --> 00:01:15.828 has won the SEC.

NOTE Confidence: 0.7496509

00:01:15.828 --> 00:01:18.960 It's knew yet to be discribed disorder,

NOTE Confidence: 0.7496509

00:01:18.960 --> 00:01:21.080 but presumably Israel to the
NOTE Confidence: 0.7496509

00:01:21.080 --> 00:01:23.200 server could be common disease,
NOTE Confidence: 0.7496509

00:01:23.200 --> 00:01:25.510 but it's rare atypical presentation.
NOTE Confidence: 0.7496509

00:01:25.510 --> 00:01:28.114 Which of these is EU DNS and
NOTE Confidence: 0.7496509

00:01:28.114 --> 00:01:30.309 Nicosia loaded 2 hours apart?
NOTE Confidence: 0.7496509

00:01:30.310 --> 00:01:32.718 Up with the the team before I
NOTE Confidence: 0.7496509

00:01:32.718 --> 00:01:35.110 left the Duke forward disease.
NOTE Confidence: 0.7496509

00:01:35.110 --> 00:01:37.576 Of course this is oftentimes the
NOTE Confidence: 0.7496509

00:01:37.576 --> 00:01:39.510 policy definition in the US,
NOTE Confidence: 0.7496509

00:01:39.510 --> 00:01:42.782 which is often drug act in 1983 as
NOTE Confidence: 0.7496509

00:01:42.782 --> 00:01:45.260 it described as less than 200,000
NOTE Confidence: 0.7496509

00:01:45.260 --> 00:01:48.310 people or one in 1500 in the US,
NOTE Confidence: 0.7496509

00:01:48.310 --> 00:01:50.710 will qualify as a real disease.
NOTE Confidence: 0.7496509

00:01:50.710 --> 00:01:51.084 Roughly,
NOTE Confidence: 0.7496509

00:01:51.084 --> 00:01:53.328 we know about 7000 rare disease
NOTE Confidence: 0.7496509

00:01:53.328 --> 00:01:55.629 are known and destination you.

NOTE Confidence: 0.7496509
00:01:55.630 --> 00:01:59.110 One discovered over the time.
NOTE Confidence: 0.7496509
00:01:59.110 --> 00:02:00.870 Individual rare disease is real,
NOTE Confidence: 0.7496509
00:02:00.870 --> 00:02:03.320 however, as a group is now well,
NOTE Confidence: 0.7496509
00:02:03.320 --> 00:02:05.518 one in a 10 or estimated 25
NOTE Confidence: 0.7496509
00:02:05.518 --> 00:02:08.075 million US are living in the US
NOTE Confidence: 0.7496509
00:02:08.075 --> 00:02:09.990 are living with real disease.
NOTE Confidence: 0.7496509
00:02:09.990 --> 00:02:12.582 You can see that number is
NOTE Confidence: 0.7496509
00:02:12.582 --> 00:02:13.878 pretty pretty challenge.
NOTE Confidence: 0.7496509
00:02:13.880 --> 00:02:16.432 Summit register maybe diagnosis
NOTE Confidence: 0.7496509
00:02:16.432 --> 00:02:18.984 or undiagnosed for both.
NOTE Confidence: 0.7496509
00:02:18.990 --> 00:02:21.060 The reason they got the janome
NOTE Confidence: 0.7496509
00:02:21.060 --> 00:02:23.015 assists almost an estimated 80% of
NOTE Confidence: 0.7496509
00:02:23.015 --> 00:02:24.690 undiagnosed disease or rare disease.
NOTE Confidence: 0.7496509
00:02:24.690 --> 00:02:27.448 Have a genetic etiology so I'll think
NOTE Confidence: 0.7496509
00:02:27.448 --> 00:02:30.638 about so called the same page on that one.
NOTE Confidence: 0.7496509

00:02:30.640 --> 00:02:31.016 So,
NOTE Confidence: 0.7496509

00:02:31.016 --> 00:02:32.896 and I indeed have many,
NOTE Confidence: 0.7496509

00:02:32.900 --> 00:02:34.400 many program intramural extramural
NOTE Confidence: 0.7496509

00:02:34.400 --> 00:02:36.275 program dedicated for rare disease,
NOTE Confidence: 0.7496509

00:02:36.280 --> 00:02:38.012 just list some here.
NOTE Confidence: 0.7496509

00:02:38.012 --> 00:02:41.445 I'm Luckily I was Luckily as part of
NOTE Confidence: 0.7496509

00:02:41.445 --> 00:02:44.860 the UDN before left a year left of you.
NOTE Confidence: 0.7496509

00:02:44.860 --> 00:02:47.499 As you probably also know that the
NOTE Confidence: 0.7496509

00:02:47.499 --> 00:02:50.148 sellers and the parents of private,
NOTE Confidence: 0.7496509

00:02:50.150 --> 00:02:51.766 industrial or philanthropy sponsor
NOTE Confidence: 0.7496509

00:02:51.766 --> 00:02:53.786 efforts and our foundation is
NOTE Confidence: 0.7496509

00:02:53.786 --> 00:02:55.440 oftentimes real disease specific.
NOTE Confidence: 0.7496509

00:02:55.440 --> 00:02:57.876 In the US in the world,
NOTE Confidence: 0.7496509

00:02:57.880 --> 00:02:59.915 the partner between NIH program
NOTE Confidence: 0.7496509

00:02:59.915 --> 00:03:01.136 and Parents Group.
NOTE Confidence: 0.7496509

00:03:01.140 --> 00:03:01.656 Indeed,

NOTE Confidence: 0.7496509

00:03:01.656 --> 00:03:04.752 it's become very powerful force to

NOTE Confidence: 0.7496509

00:03:04.752 --> 00:03:07.738 promoting the research for rare disease.

NOTE Confidence: 0.7496509

00:03:07.740 --> 00:03:08.348 Of course,

NOTE Confidence: 0.7496509

00:03:08.348 --> 00:03:10.780 in the history in the year that for

NOTE Confidence: 0.7496509

00:03:10.846 --> 00:03:13.816 real disease or undiagnosed disease research,

NOTE Confidence: 0.7496509

00:03:13.820 --> 00:03:14.200 definition,

NOTE Confidence: 0.7496509

00:03:14.200 --> 00:03:16.860 outstanding and just highlight a few summer.

NOTE Confidence: 0.7496509

00:03:16.860 --> 00:03:19.032 Probably you know that Lee Rosenberg

NOTE Confidence: 0.7496509

00:03:19.032 --> 00:03:21.736 and was the former chair of the

NOTE Confidence: 0.7496509

00:03:21.736 --> 00:03:23.316 Department of Human Genetics.

NOTE Confidence: 0.7496509

00:03:23.320 --> 00:03:25.980 Also Dean of the School of Medicine.

NOTE Confidence: 0.7496509

00:03:25.980 --> 00:03:27.388 He highlighted one lecture,

NOTE Confidence: 0.7496509

00:03:27.388 --> 00:03:29.148 said for children and yield

NOTE Confidence: 0.7496509

00:03:29.148 --> 00:03:30.919 making awful human geneticists.

NOTE Confidence: 0.7496509

00:03:30.920 --> 00:03:33.895 I would argue probably making the grade

NOTE Confidence: 0.7496509

00:03:33.895 --> 00:03:37.189 of Human Genetics in our country on history.

NOTE Confidence: 0.7496509

00:03:37.190 --> 00:03:39.260 An awesome notice have very many

NOTE Confidence: 0.7496509

00:03:39.260 --> 00:03:41.073 many other notable fellow from

NOTE Confidence: 0.7496509

00:03:41.073 --> 00:03:42.549 the year Training program.

NOTE Confidence: 0.7496509

00:03:42.550 --> 00:03:45.241 Just to list a few on here and I

NOTE Confidence: 0.7496509

00:03:45.241 --> 00:03:47.898 don't have to say anything further.

NOTE Confidence: 0.7496509

00:03:47.900 --> 00:03:49.148 For most recent one,

NOTE Confidence: 0.7496509

00:03:49.148 --> 00:03:51.020 like Clifton also make a huge

NOTE Confidence: 0.7496509

00:03:51.087 --> 00:03:54.840 contribution for real disease for sure.

NOTE Confidence: 0.7496509

00:03:54.840 --> 00:03:56.675 So it's kind of problematic

NOTE Confidence: 0.7496509

00:03:56.675 --> 00:03:57.776 at medical education.

NOTE Confidence: 0.7496509

00:03:57.780 --> 00:04:00.349 Often time we're saying making a medical

NOTE Confidence: 0.7496509

00:04:00.349 --> 00:04:02.548 diagnosis is the art of medicine.

NOTE Confidence: 0.7496509

00:04:02.550 --> 00:04:04.380 Sometimes can be quite easy,

NOTE Confidence: 0.7496509

00:04:04.380 --> 00:04:06.900 such as like this if you encounter a

NOTE Confidence: 0.7496509

00:04:06.900 --> 00:04:09.890 patient in the clinic like this presentation,

NOTE Confidence: 0.7496509
00:04:09.890 --> 00:04:10.988 you will say,
NOTE Confidence: 0.7496509
00:04:10.988 --> 00:04:11.354 OK,
NOTE Confidence: 0.7496509
00:04:11.354 --> 00:04:13.184 I know what it is.
NOTE Confidence: 0.8053164
00:04:13.190 --> 00:04:15.030 If you got some training.
NOTE Confidence: 0.8053164
00:04:15.030 --> 00:04:17.585 However, they often that's not very easy.
NOTE Confidence: 0.8053164
00:04:17.590 --> 00:04:19.425 We often time car diagnostic
NOTE Confidence: 0.8053164
00:04:19.425 --> 00:04:20.893 Odyssey for real disease.
NOTE Confidence: 0.8053164
00:04:20.900 --> 00:04:23.828 This is a slide I took from alumina.
NOTE Confidence: 0.8053164
00:04:23.830 --> 00:04:26.110 Kind of modified a little bit.
NOTE Confidence: 0.8053164
00:04:26.110 --> 00:04:28.666 And that shows you the challenge
NOTE Confidence: 0.8053164
00:04:28.666 --> 00:04:31.065 before the next generation sequencing
NOTE Confidence: 0.8053164
00:04:31.065 --> 00:04:33.549 technology available on average,
NOTE Confidence: 0.8053164
00:04:33.550 --> 00:04:35.990 the real disease diagnosed taking
NOTE Confidence: 0.8053164
00:04:35.990 --> 00:04:39.009 about five years and kind of
NOTE Confidence: 0.8053164
00:04:39.009 --> 00:04:41.489 looking for a different position,
NOTE Confidence: 0.8053164

00:04:41.490 --> 00:04:43.970 an average final which diagnosis,
NOTE Confidence: 0.8053164

00:04:43.970 --> 00:04:45.256 diagnosis oftentimes
NOTE Confidence: 0.8053164

00:04:45.256 --> 00:04:47.828 also misdiagnosis at end.
NOTE Confidence: 0.8053164

00:04:47.830 --> 00:04:48.744 So therefore,
NOTE Confidence: 0.8053164

00:04:48.744 --> 00:04:51.029 which the new generation sequencing
NOTE Confidence: 0.8053164

00:04:51.029 --> 00:04:53.822 now really look at the possibility
NOTE Confidence: 0.8053164

00:04:53.822 --> 00:04:56.072 comma ending the diagnostic Odyssey
NOTE Confidence: 0.8053164

00:04:56.072 --> 00:04:58.764 by global efforts for this one or
NOTE Confidence: 0.8053164

00:04:58.764 --> 00:05:00.942 in the US government also pushed
NOTE Confidence: 0.8053164

00:05:00.942 --> 00:05:04.106 for the policy or or two supporting
NOTE Confidence: 0.8053164

00:05:04.106 --> 00:05:06.777 this effort study in 2019 that
NOTE Confidence: 0.8053164

00:05:06.777 --> 00:05:09.291 HR4144 on passing the House but
NOTE Confidence: 0.8053164

00:05:09.377 --> 00:05:12.016 not yet in the Senate and renamed
NOTE Confidence: 0.8053164

00:05:12.016 --> 00:05:14.413 in the 2020 called ending the
NOTE Confidence: 0.8053164

00:05:14.413 --> 00:05:16.097 Diagnostic obviously act hopeful.
NOTE Confidence: 0.8053164

00:05:16.100 --> 00:05:18.310 We got this one pass.

NOTE Confidence: 0.8053164

00:05:18.310 --> 00:05:21.075 We really would benefit a four million

NOTE Confidence: 0.8053164

00:05:21.075 --> 00:05:23.360 offer where disease patient family.

NOTE Confidence: 0.8053164

00:05:23.360 --> 00:05:24.138 So really,

NOTE Confidence: 0.8053164

00:05:24.138 --> 00:05:26.083 not on the next generation

NOTE Confidence: 0.8053164

00:05:26.083 --> 00:05:28.020 sequencing and Internet diagnosing,

NOTE Confidence: 0.8053164

00:05:28.020 --> 00:05:30.044 the clinics are restarting

NOTE Confidence: 0.8053164

00:05:30.044 --> 00:05:32.068 roughly earlier for 2000.

NOTE Confidence: 0.8053164

00:05:32.070 --> 00:05:34.626 Can a 2011 yell definitely Distaff

NOTE Confidence: 0.8053164

00:05:34.626 --> 00:05:36.847 Latinist efforts which shows this

NOTE Confidence: 0.8053164

00:05:36.847 --> 00:05:39.047 publication lot about Rick Lifton?

NOTE Confidence: 0.8053164

00:05:39.050 --> 00:05:42.346 Here he was the former chair in the

NOTE Confidence: 0.8053164

00:05:42.346 --> 00:05:45.354 Department and shows at the same time

NOTE Confidence: 0.8053164

00:05:45.354 --> 00:05:48.200 is asking another group from Seattle,

NOTE Confidence: 0.8053164

00:05:48.200 --> 00:05:51.200 WA and publish almost the same

NOTE Confidence: 0.8053164

00:05:51.200 --> 00:05:53.200 time just to demonstrate.

NOTE Confidence: 0.8053164

00:05:53.200 --> 00:05:55.979 The whole exon capture and sequencing has
NOTE Confidence: 0.8053164

00:05:55.979 --> 00:05:58.519 significant value or effectively can use.
NOTE Confidence: 0.8053164

00:05:58.520 --> 00:06:01.824 It can be used in the real
NOTE Confidence: 0.8053164

00:06:01.824 --> 00:06:04.450 disease diagnosis in the clinic.
NOTE Confidence: 0.8053164

00:06:04.450 --> 00:06:04.848 So,
NOTE Confidence: 0.8053164

00:06:04.848 --> 00:06:05.246 uh,
NOTE Confidence: 0.8053164

00:06:05.246 --> 00:06:07.634 some of you probably know and
NOTE Confidence: 0.8053164

00:06:07.634 --> 00:06:09.762 Baylor Genetics consider powerhouse
NOTE Confidence: 0.8053164

00:06:09.762 --> 00:06:12.266 for medical genetic diagnosis.
NOTE Confidence: 0.8053164

00:06:12.270 --> 00:06:14.937 Last for decades and then in the
NOTE Confidence: 0.8053164

00:06:14.937 --> 00:06:18.133 2011 and two years after initial
NOTE Confidence: 0.8053164

00:06:18.133 --> 00:06:20.749 researchers are the demonstration.
NOTE Confidence: 0.8053164

00:06:20.750 --> 00:06:23.750 Yeah, Baylor considered the first offer.
NOTE Confidence: 0.8053164

00:06:23.750 --> 00:06:26.805 The whole exome testing in
NOTE Confidence: 0.8053164

00:06:26.805 --> 00:06:29.860 the clinic and about 2019.
NOTE Confidence: 0.8053164

00:06:29.860 --> 00:06:32.056 Baylor also introduced the whole exon.

NOTE Confidence: 0.8053164

00:06:32.060 --> 00:06:35.345 Of course, that's the place I was trained as.

NOTE Confidence: 0.8053164

00:06:35.350 --> 00:06:37.546 Happy for that kind of advancement.

NOTE Confidence: 0.8053164

00:06:37.550 --> 00:06:39.740 And then, on the other hand,

NOTE Confidence: 0.8053164

00:06:39.740 --> 00:06:42.668 I should also try to make an argument.

NOTE Confidence: 0.8053164

00:06:42.670 --> 00:06:44.866 Yield does deserve a huge credit

NOTE Confidence: 0.8053164

00:06:44.866 --> 00:06:46.330 for this particular area,

NOTE Confidence: 0.8053164

00:06:46.330 --> 00:06:48.418 or even consider the same level

NOTE Confidence: 0.8053164

00:06:48.418 --> 00:06:50.644 which you can shows here under

NOTE Confidence: 0.8053164

00:06:50.644 --> 00:06:52.544 leadership of Alan Bell Net,

NOTE Confidence: 0.8053164

00:06:52.550 --> 00:06:54.380 which you can see here,

NOTE Confidence: 0.8053164

00:06:54.380 --> 00:06:56.576 offer the first whole exon in

NOTE Confidence: 0.8053164

00:06:56.576 --> 00:06:58.040 the December of 2011.

NOTE Confidence: 0.8053164

00:06:58.040 --> 00:07:00.368 Literally, it's one month after Baylor.

NOTE Confidence: 0.8053164

00:07:00.370 --> 00:07:03.366 Introduce the whole exon in a clinic,

NOTE Confidence: 0.8053164

00:07:03.370 --> 00:07:05.956 which is your second in the

NOTE Confidence: 0.8053164

00:07:05.956 --> 00:07:08.520 country and similarly in the 2019.

NOTE Confidence: 0.8053164

00:07:08.520 --> 00:07:10.292 A couple months later,

NOTE Confidence: 0.8053164

00:07:10.292 --> 00:07:12.507 year offered rapid whole genome

NOTE Confidence: 0.8053164

00:07:12.507 --> 00:07:14.528 sequencing in New Haven Hospital.

NOTE Confidence: 0.8053164

00:07:14.530 --> 00:07:16.670 Patient any liver flavor to

NOTE Confidence: 0.8053164

00:07:16.670 --> 00:07:18.382 that which is Apple.

NOTE Confidence: 0.8053164

00:07:18.390 --> 00:07:21.757 There was the chair of the genetics

NOTE Confidence: 0.8053164

00:07:21.757 --> 00:07:24.988 at Baylor when I took it off for

NOTE Confidence: 0.8053164

00:07:24.988 --> 00:07:27.830 a year and he was telling me,

NOTE Confidence: 0.8053164

00:07:27.830 --> 00:07:30.500 well when I moved to the.

NOTE Confidence: 0.8053164

00:07:30.500 --> 00:07:33.076 A Mailer stunning genetics the year is

NOTE Confidence: 0.8053164

00:07:33.076 --> 00:07:35.787 the model I've tried to replicate it.

NOTE Confidence: 0.8053164

00:07:35.790 --> 00:07:38.058 That's worse than the siding here.

NOTE Confidence: 0.8053164

00:07:38.060 --> 00:07:38.820 Code here.

NOTE Confidence: 0.8053164

00:07:38.820 --> 00:07:40.720 When he received the victim

NOTE Confidence: 0.8053164

00:07:40.720 --> 00:07:42.220 Acustica Leadership Award 2017,

NOTE Confidence: 0.8053164
00:07:42.220 --> 00:07:44.470 which is kind of interesting
NOTE Confidence: 0.8053164
00:07:44.470 --> 00:07:46.270 flavor for this story.
NOTE Confidence: 0.8053164
00:07:46.270 --> 00:07:49.460 I just got a 2 slide to show what our
NOTE Confidence: 0.84979755
00:07:49.552 --> 00:07:52.860 medical journals. Glad you can offer
NOTE Confidence: 0.84979755
00:07:52.860 --> 00:07:55.160 for patient care or diagnostic.
NOTE Confidence: 0.84979755
00:07:55.160 --> 00:07:57.686 Purpose so this is a a
NOTE Confidence: 0.84979755
00:07:57.686 --> 00:07:58.949 leadership without laughter.
NOTE Confidence: 0.84979755
00:07:58.950 --> 00:08:02.739 It directed by Alan Bell and way Jonah here.
NOTE Confidence: 0.84979755
00:08:02.740 --> 00:08:05.687 Just highlight a few or we offer.
NOTE Confidence: 0.84979755
00:08:05.690 --> 00:08:07.790 Here is a critical access,
NOTE Confidence: 0.84979755
00:08:07.790 --> 00:08:09.900 you know, start in 2011.
NOTE Confidence: 0.84979755
00:08:09.900 --> 00:08:12.000 We offer both inpatient outpatient
NOTE Confidence: 0.84979755
00:08:12.000 --> 00:08:13.260 impatient regardless insurance.
NOTE Confidence: 0.84979755
00:08:13.260 --> 00:08:15.732 Your health will cover it's very
NOTE Confidence: 0.84979755
00:08:15.732 --> 00:08:18.319 generous asanka keys and many others.
NOTE Confidence: 0.84979755

00:08:18.320 --> 00:08:20.420 General support for this effort.
NOTE Confidence: 0.84979755

00:08:20.420 --> 00:08:23.815 The Russia case and the whole exon
NOTE Confidence: 0.84979755

00:08:23.815 --> 00:08:26.507 lab reporting within 7 to 12 days.
NOTE Confidence: 0.84979755

00:08:26.510 --> 00:08:29.191 The regular is 2 months time to
NOTE Confidence: 0.84979755

00:08:29.191 --> 00:08:31.599 reporting out the result totally.
NOTE Confidence: 0.84979755

00:08:31.600 --> 00:08:34.198 Lab has performed about more than
NOTE Confidence: 0.84979755

00:08:34.198 --> 00:08:37.032 10,000 whole exon case and the
NOTE Confidence: 0.84979755

00:08:37.032 --> 00:08:39.072 lab also contributed generation
NOTE Confidence: 0.84979755

00:08:39.072 --> 00:08:41.112 project for the reporting.
NOTE Confidence: 0.84979755

00:08:41.120 --> 00:08:42.960 On 2019, we introduce rap.
NOTE Confidence: 0.84979755

00:08:42.960 --> 00:08:46.160 The whole genome sequencing is a few in
NOTE Confidence: 0.84979755

00:08:46.160 --> 00:08:49.080 the country and we offer the critical.
NOTE Confidence: 0.84979755

00:08:49.080 --> 00:08:51.648 Your patient in the NICU and PICU in
NOTE Confidence: 0.84979755

00:08:51.648 --> 00:08:53.920 the New Haven Children Hospital and
NOTE Confidence: 0.84979755

00:08:53.920 --> 00:08:57.036 the turn around time is 3 and five
NOTE Confidence: 0.84979755

00:08:57.036 --> 00:08:59.535 days with our support from why SGA.

NOTE Confidence: 0.84979755

00:08:59.540 --> 00:09:03.572 The total number so far we performed is 19.

NOTE Confidence: 0.84979755

00:09:03.580 --> 00:09:05.914 I was touching out later for

NOTE Confidence: 0.84979755

00:09:05.914 --> 00:09:09.076 the detail and one of the also

NOTE Confidence: 0.84979755

00:09:09.076 --> 00:09:11.601 significant strengths and the lab

NOTE Confidence: 0.84979755

00:09:11.601 --> 00:09:14.434 drackett inform me it's in our

NOTE Confidence: 0.84979755

00:09:14.434 --> 00:09:16.599 lab DNA diagrams diagnostic lab.

NOTE Confidence: 0.84979755

00:09:16.600 --> 00:09:18.850 We are often customer testing,

NOTE Confidence: 0.84979755

00:09:18.850 --> 00:09:19.746 customized testing,

NOTE Confidence: 0.84979755

00:09:19.746 --> 00:09:21.090 including origin interesting.

NOTE Confidence: 0.84979755

00:09:21.090 --> 00:09:23.364 The team is awaiting to discuss

NOTE Confidence: 0.84979755

00:09:23.364 --> 00:09:25.902 a walking with the clinical team

NOTE Confidence: 0.84979755

00:09:25.902 --> 00:09:28.252 for any specialty or subspecialty

NOTE Confidence: 0.84979755

00:09:28.252 --> 00:09:30.580 for design and validation.

NOTE Confidence: 0.84979755

00:09:30.580 --> 00:09:32.820 Which is very significant for this type

NOTE Confidence: 0.84979755

00:09:32.820 --> 00:09:35.549 are often a diagnostic testing purpose.

NOTE Confidence: 0.84979755

00:09:35.550 --> 00:09:38.028 There's a lot of strengths are listed
NOTE Confidence: 0.84979755

00:09:38.028 --> 00:09:40.129 here for experienced laboratory team.
NOTE Confidence: 0.84979755

00:09:40.130 --> 00:09:42.538 Easy ordering from epic and we have
NOTE Confidence: 0.84979755

00:09:42.538 --> 00:09:44.495 a team clinical team assisting
NOTE Confidence: 0.84979755

00:09:44.495 --> 00:09:46.620 you for the complicated case.
NOTE Confidence: 0.84979755

00:09:46.620 --> 00:09:48.575 The Russian service available 7227
NOTE Confidence: 0.84979755

00:09:48.575 --> 00:09:51.441 to 12 days for the whole exon
NOTE Confidence: 0.84979755

00:09:51.441 --> 00:09:53.925 is impatient and we offer the
NOTE Confidence: 0.84979755

00:09:53.925 --> 00:09:56.452 printer testing and I sort of the
NOTE Confidence: 0.84979755

00:09:56.452 --> 00:09:58.498 child OneNote four and ask for
NOTE Confidence: 0.84979755

00:09:58.498 --> 00:10:01.006 generation project for the raw data.
NOTE Confidence: 0.84979755

00:10:01.010 --> 00:10:03.677 Here are we offer if the order
NOTE Confidence: 0.84979755

00:10:03.677 --> 00:10:05.250 and provide interesting data.
NOTE Confidence: 0.84979755

00:10:05.250 --> 00:10:06.686 Raw data hold Gina,
NOTE Confidence: 0.84979755

00:10:06.686 --> 00:10:08.840 who acts on laptop with able
NOTE Confidence: 0.84979755

00:10:08.918 --> 00:10:10.638 to accommodate to that.

NOTE Confidence: 0.84979755

00:10:10.640 --> 00:10:13.280 We also got a huge support for ICJ

NOTE Confidence: 0.84979755

00:10:13.280 --> 00:10:15.759 II laboratory is it's a side of

NOTE Confidence: 0.84979755

00:10:15.759 --> 00:10:17.509 genetic laboratory is directed by

NOTE Confidence: 0.84979755

00:10:17.584 --> 00:10:20.344 pending Lee and very quickly they

NOTE Confidence: 0.84979755

00:10:20.344 --> 00:10:22.184 offer the traditional karyotyping

NOTE Confidence: 0.84979755

00:10:22.190 --> 00:10:24.878 and fish and in 2006 and they often

NOTE Confidence: 0.84979755

00:10:24.878 --> 00:10:26.920 genome wide copy number analysis

NOTE Confidence: 0.84979755

00:10:26.920 --> 00:10:29.500 by Michael Away which is regular.

NOTE Confidence: 0.84979755

00:10:29.500 --> 00:10:31.660 Taking one 2-3 weeks time.

NOTE Confidence: 0.84979755

00:10:31.660 --> 00:10:33.604 Performing more than 7000 cases for

NOTE Confidence: 0.84979755

00:10:33.604 --> 00:10:36.110 both prenatal and adult pediatric population.

NOTE Confidence: 0.84979755

00:10:36.110 --> 00:10:36.482 Again,

NOTE Confidence: 0.84979755

00:10:36.482 --> 00:10:37.970 they're very experienced team.

NOTE Confidence: 0.84979755

00:10:37.970 --> 00:10:39.820 It's very easy to ordering.

NOTE Confidence: 0.84979755

00:10:39.820 --> 00:10:42.076 The team is assistant for you

NOTE Confidence: 0.84979755

00:10:42.076 --> 00:10:44.000 for interpretation and then you
NOTE Confidence: 0.84979755

00:10:44.000 --> 00:10:45.760 can order Rush rush service.
NOTE Confidence: 0.84979755

00:10:45.760 --> 00:10:47.041 If you need,
NOTE Confidence: 0.84979755

00:10:47.041 --> 00:10:50.030 just contact the lab and they will
NOTE Confidence: 0.84979755

00:10:50.121 --> 00:10:53.208 help you out for the patient care.
NOTE Confidence: 0.84979755

00:10:53.210 --> 00:10:54.890 Stop for rest of the time.
NOTE Confidence: 0.84979755

00:10:54.890 --> 00:10:56.450 I'm gonna just as you probably
NOTE Confidence: 0.84979755

00:10:56.450 --> 00:10:57.970 heard from the tax partner.
NOTE Confidence: 0.84979755

00:10:57.970 --> 00:10:59.342 Kind of interesting case.
NOTE Confidence: 0.84979755

00:10:59.342 --> 00:11:01.820 I'm going to show you a few
NOTE Confidence: 0.84979755

00:11:01.820 --> 00:11:03.570 more case how the genetic.
NOTE Confidence: 0.84979755

00:11:03.570 --> 00:11:06.218 May lead into the different type of the,
NOTE Confidence: 0.84979755

00:11:06.220 --> 00:11:08.104 so the revolution from the clinical
NOTE Confidence: 0.84979755

00:11:08.104 --> 00:11:10.450 care and then the details on the
NOTE Confidence: 0.84979755

00:11:10.450 --> 00:11:12.502 clinical Side Story is not important.
NOTE Confidence: 0.8291808

00:11:12.510 --> 00:11:14.090 You pretty much you can't

NOTE Confidence: 0.8291808

00:11:14.090 --> 00:11:16.150 just come look at the title.

NOTE Confidence: 0.8291808

00:11:16.150 --> 00:11:18.530 So the point I want to make it to show

NOTE Confidence: 0.8291808

00:11:18.593 --> 00:11:20.957 you the success opportunity and also

NOTE Confidence: 0.8291808

00:11:20.957 --> 00:11:23.305 challenge was still facing for using

NOTE Confidence: 0.8291808

00:11:23.305 --> 00:11:25.417 the application for the whole exon.

NOTE Confidence: 0.8291808

00:11:25.420 --> 00:11:27.388 Hoje no, in the clinical 7

NOTE Confidence: 0.8291808

00:11:27.388 --> 00:11:29.060 so this case number one.

NOTE Confidence: 0.8291808

00:11:29.060 --> 00:11:30.026 It's very quickly.

NOTE Confidence: 0.8291808

00:11:30.026 --> 00:11:32.280 It's a 30 week and SJ in

NOTE Confidence: 0.8291808

00:11:32.357 --> 00:11:34.377 the Premier baby with this.

NOTE Confidence: 0.8291808

00:11:34.380 --> 00:11:36.232 With their short Lambda

NOTE Confidence: 0.8291808

00:11:36.232 --> 00:11:38.084 and abnormal rib cage.

NOTE Confidence: 0.8291808

00:11:38.090 --> 00:11:40.582 And this sort of quiet kind of

NOTE Confidence: 0.8291808

00:11:40.582 --> 00:11:42.457 significant recipe support and also

NOTE Confidence: 0.8291808

00:11:42.457 --> 00:11:44.302 when incubated ventilators are and

NOTE Confidence: 0.8291808

00:11:44.302 --> 00:11:46.599 had a difficult to win ventilator.
NOTE Confidence: 0.8291808

00:11:46.600 --> 00:11:49.190 So here's the initial actually at birth,
NOTE Confidence: 0.8291808

00:11:49.190 --> 00:11:51.040 which you can see here.
NOTE Confidence: 0.8291808

00:11:51.040 --> 00:11:54.000 Let me see if I can get my.
NOTE Confidence: 0.7772621

00:11:56.390 --> 00:11:59.747 Pointer so this is a normal X Ray and
NOTE Confidence: 0.7772621

00:11:59.747 --> 00:12:02.878 this is the patients actually on the
NOTE Confidence: 0.7772621

00:12:02.878 --> 00:12:06.840 left side and this is the one in the
NOTE Confidence: 0.7772621

00:12:06.840 --> 00:12:09.366 middle which is tactical from textbook
NOTE Confidence: 0.7772621

00:12:09.366 --> 00:12:11.546 which initially actually report about
NOTE Confidence: 0.7772621

00:12:11.546 --> 00:12:14.360 radiologist say OK this is very similar.
NOTE Confidence: 0.7772621

00:12:14.360 --> 00:12:16.032 The patient based similar
NOTE Confidence: 0.7772621

00:12:16.032 --> 00:12:18.122 to a one known condition,
NOTE Confidence: 0.7772621

00:12:18.130 --> 00:12:19.798 comma center, foreca displeasure.
NOTE Confidence: 0.7772621

00:12:19.798 --> 00:12:21.466 It's a skeletal dysplasias,
NOTE Confidence: 0.7772621

00:12:21.470 --> 00:12:24.284 oftentimes caused by FGFR receptor mutation
NOTE Confidence: 0.7772621

00:12:24.284 --> 00:12:26.410 disease often time considerably so.

NOTE Confidence: 0.7772621

00:12:26.410 --> 00:12:28.954 Post Natal reappearing natively so the

NOTE Confidence: 0.7772621

00:12:28.954 --> 00:12:32.513 whole exon was done and the quickly getting

NOTE Confidence: 0.7772621

00:12:32.513 --> 00:12:35.189 report initial report and no mutation

NOTE Confidence: 0.7772621

00:12:35.268 --> 00:12:38.047 FGFR 3 that pretty much accounted for,

NOTE Confidence: 0.7772621

00:12:38.050 --> 00:12:40.205 now confirmed for the clinical

NOTE Confidence: 0.7772621

00:12:40.205 --> 00:12:42.360 impression by an X Ray.

NOTE Confidence: 0.7772621

00:12:42.360 --> 00:12:44.992 And indeed we find a homozygous mutation

NOTE Confidence: 0.7772621

00:12:44.992 --> 00:12:48.387 for a splice inside mutation foreign Eugene.

NOTE Confidence: 0.7772621

00:12:48.390 --> 00:12:50.114 Can IMPP L1 gene,

NOTE Confidence: 0.7772621

00:12:50.114 --> 00:12:51.838 it's extremely rare condition.

NOTE Confidence: 0.7772621

00:12:51.840 --> 00:12:54.426 You can see the earlier frequency.

NOTE Confidence: 0.7772621

00:12:54.430 --> 00:12:56.190 It's very very low.

NOTE Confidence: 0.7772621

00:12:56.190 --> 00:12:58.830 But this gene mutation homozygous terroir.

NOTE Confidence: 0.7772621

00:12:58.830 --> 00:13:01.342 Stage cost another different

NOTE Confidence: 0.7772621

00:13:01.342 --> 00:13:02.598 scatter displeasure,

NOTE Confidence: 0.7772621

00:13:02.600 --> 00:13:07.100 and this one indeed clinically.
NOTE Confidence: 0.7772621

00:13:07.100 --> 00:13:09.760 The symbol for the silverback
NOTE Confidence: 0.7772621

00:13:09.760 --> 00:13:10.824 Assire Friedrich.
NOTE Confidence: 0.7772621

00:13:10.830 --> 00:13:12.450 Centerfire before your displeasure,
NOTE Confidence: 0.7772621

00:13:12.450 --> 00:13:14.880 but the patient often times survive,
NOTE Confidence: 0.7772621

00:13:14.880 --> 00:13:17.300 and the prognosis is very
NOTE Confidence: 0.7772621

00:13:17.300 --> 00:13:19.236 different for this one.
NOTE Confidence: 0.7772621

00:13:19.240 --> 00:13:21.080 Fun are MPPL optimal
NOTE Confidence: 0.7772621

00:13:21.080 --> 00:13:22.460 displeasure on presentation.
NOTE Confidence: 0.7772621

00:13:22.460 --> 00:13:25.680 They often time after past and you
NOTE Confidence: 0.7772621

00:13:25.680 --> 00:13:27.060 need to stage.
NOTE Confidence: 0.7772621

00:13:27.060 --> 00:13:28.335 This is viable.
NOTE Confidence: 0.7772621

00:13:28.335 --> 00:13:28.760 Well,
NOTE Confidence: 0.7772621

00:13:28.760 --> 00:13:30.885 we wish the treatment particular
NOTE Confidence: 0.7772621

00:13:30.885 --> 00:13:33.816 as a treatment available by by
NOTE Confidence: 0.7772621

00:13:33.816 --> 00:13:35.756 phosphonate infusion can increase

NOTE Confidence: 0.7772621

00:13:35.756 --> 00:13:38.560 the bone health and good mobility.

NOTE Confidence: 0.7772621

00:13:38.560 --> 00:13:41.780 So indeed this initial diagnosis by genetics,

NOTE Confidence: 0.7772621

00:13:41.780 --> 00:13:44.545 changing management and also potentially

NOTE Confidence: 0.7772621

00:13:44.545 --> 00:13:47.990 for the prognosis for the future.

NOTE Confidence: 0.7772621

00:13:47.990 --> 00:13:50.174 The second it's a I want to

NOTE Confidence: 0.7772621

00:13:50.174 --> 00:13:52.109 show you it's my own case.

NOTE Confidence: 0.7772621

00:13:52.110 --> 00:13:54.276 Other cases are shared by Doctor

NOTE Confidence: 0.7772621

00:13:54.276 --> 00:13:56.799 Phalen other my other colleague here.

NOTE Confidence: 0.7772621

00:13:56.800 --> 00:13:59.020 It's the final ideological

NOTE Confidence: 0.7772621

00:13:59.020 --> 00:14:01.795 diagnosis that's waiting 20 years.

NOTE Confidence: 0.7772621

00:14:01.800 --> 00:14:04.432 So this is a 26 year old female

NOTE Confidence: 0.7772621

00:14:04.432 --> 00:14:06.894 present to my clinic last summer

NOTE Confidence: 0.7772621

00:14:06.894 --> 00:14:09.480 which continue onset of a seizure

NOTE Confidence: 0.7772621

00:14:09.558 --> 00:14:12.190 and cognition deley and she had a

NOTE Confidence: 0.7772621

00:14:12.190 --> 00:14:14.709 history of our Department delay or

NOTE Confidence: 0.7772621

00:14:14.709 --> 00:14:16.561 intellectual disability was evaluated
NOTE Confidence: 0.7772621

00:14:16.561 --> 00:14:19.238 in the genetics Connect 20 years ago
NOTE Confidence: 0.7772621

00:14:19.238 --> 00:14:21.769 by my colleague and yell at that time,
NOTE Confidence: 0.7772621

00:14:21.770 --> 00:14:22.961 as you know,
NOTE Confidence: 0.7772621

00:14:22.961 --> 00:14:24.946 is very limited genetic testing
NOTE Confidence: 0.7772621

00:14:24.946 --> 00:14:27.426 available and no 'cause it was fun
NOTE Confidence: 0.7772621

00:14:27.426 --> 00:14:29.943 because it's a new problem then the
NOTE Confidence: 0.7772621

00:14:29.943 --> 00:14:32.033 physician appreciate physician not refer
NOTE Confidence: 0.7772621

00:14:32.033 --> 00:14:35.768 back to the generic say hey help it again.
NOTE Confidence: 0.7772621

00:14:35.770 --> 00:14:38.356 See if you understand why these
NOTE Confidence: 0.7772621

00:14:38.356 --> 00:14:40.596 patients suddenly have sort of
NOTE Confidence: 0.7772621

00:14:40.596 --> 00:14:43.224 deterioration or other on you symptoms.
NOTE Confidence: 0.7772621

00:14:43.230 --> 00:14:46.902 So we indeed who acts on the outpatient
NOTE Confidence: 0.7772621

00:14:46.902 --> 00:14:48.727 setting identify exonic dilution
NOTE Confidence: 0.7772621

00:14:48.727 --> 00:14:52.007 for 27 or 30 in than Eugene Comma,
NOTE Confidence: 0.7772621

00:14:52.010 --> 00:14:54.650 USP 96 is X linked gene,

NOTE Confidence: 0.7772621

00:14:54.650 --> 00:14:57.278 and usually it's a show dominant.

NOTE Confidence: 0.7772621

00:14:57.280 --> 00:15:00.668 Therefore you will see the female has

NOTE Confidence: 0.7772621

00:15:00.668 --> 00:15:03.416 a significant presentation and the gene

NOTE Confidence: 0.7772621

00:15:03.416 --> 00:15:05.840 is encoding our Du broken eighting.

NOTE Confidence: 0.7772621

00:15:05.840 --> 00:15:08.498 Enzyme in surpass way shows here.

NOTE Confidence: 0.7772621

00:15:08.500 --> 00:15:11.412 This is you population are passed away

NOTE Confidence: 0.7772621

00:15:11.412 --> 00:15:14.123 from E1E2E3 for putting degradation but

NOTE Confidence: 0.7772621

00:15:14.123 --> 00:15:17.357 USPX is on the Du population answer.

NOTE Confidence: 0.7772621

00:15:17.360 --> 00:15:20.440 Of course this is a relatively new

NOTE Confidence: 0.7772621

00:15:20.440 --> 00:15:22.674 condition was first reported 2014

NOTE Confidence: 0.7772621

00:15:22.674 --> 00:15:25.579 and by the Spider 2021 is only

NOTE Confidence: 0.7772621

00:15:25.579 --> 00:15:28.820 totals 38 case report but we are

NOTE Confidence: 0.7772621

00:15:28.820 --> 00:15:31.092 very convincing convinced that their

NOTE Confidence: 0.7772621

00:15:31.092 --> 00:15:33.744 leases are cause for her condition.

NOTE Confidence: 0.7772621

00:15:33.750 --> 00:15:36.408 But that took almost 20 years.

NOTE Confidence: 0.81014601

00:15:36.410 --> 00:15:38.510 We finally. Check it out.
NOTE Confidence: 0.81014601

00:15:38.510 --> 00:15:41.390 The next one is, as you probably know,
NOTE Confidence: 0.81014601

00:15:41.390 --> 00:15:43.550 and we are primary practice in
NOTE Confidence: 0.81014601

00:15:43.550 --> 00:15:45.710 pediatric hospital, but we indeed see
NOTE Confidence: 0.81014601

00:15:45.710 --> 00:15:47.870 that all the patient different age.
NOTE Confidence: 0.81014601

00:15:47.870 --> 00:15:51.470 This is the one case and the case number 3.
NOTE Confidence: 0.81014601

00:15:51.470 --> 00:15:53.990 The whole Axon allowed to the funding.
NOTE Confidence: 0.81014601

00:15:53.990 --> 00:15:55.950 Of course for adults.
NOTE Confidence: 0.81014601

00:15:55.950 --> 00:15:58.778 Which cancer, which is 70 years
NOTE Confidence: 0.81014601

00:15:58.778 --> 00:16:02.172 71 years old man and which showed
NOTE Confidence: 0.81014601

00:16:02.172 --> 00:16:05.756 the cancer to 64 and then which?
NOTE Confidence: 0.81014601

00:16:05.760 --> 00:16:08.742 History of the uterine fibroids and then
NOTE Confidence: 0.81014601

00:16:08.742 --> 00:16:12.323 also has a skin layer myomeres and also
NOTE Confidence: 0.81014601

00:16:12.323 --> 00:16:15.709 found a colon polyps but unknown type.
NOTE Confidence: 0.81014601

00:16:15.710 --> 00:16:18.176 Lots of the scheme funding for
NOTE Confidence: 0.81014601

00:16:18.176 --> 00:16:19.820 Leiomyoma in an interesting

NOTE Confidence: 0.81014601
00:16:19.892 --> 00:16:22.160 family history on as shows here,
NOTE Confidence: 0.81014601
00:16:22.160 --> 00:16:23.772 which is clearly suggesting
NOTE Confidence: 0.81014601
00:16:23.772 --> 00:16:24.578 genetic contributed.
NOTE Confidence: 0.81014601
00:16:24.580 --> 00:16:26.986 Further genetic cancer risk for sure.
NOTE Confidence: 0.81014601
00:16:26.990 --> 00:16:30.617 This is the proband here and 71 years old,
NOTE Confidence: 0.81014601
00:16:30.620 --> 00:16:32.640 crazy and to the clinic.
NOTE Confidence: 0.81014601
00:16:32.640 --> 00:16:35.844 I'm glad and then the physician
NOTE Confidence: 0.81014601
00:16:35.844 --> 00:16:37.446 ordered how excellent.
NOTE Confidence: 0.81014601
00:16:37.450 --> 00:16:39.916 But initial, who acts on testing,
NOTE Confidence: 0.81014601
00:16:39.920 --> 00:16:41.564 looking small mutation point
NOTE Confidence: 0.81014601
00:16:41.564 --> 00:16:43.619 mutation in India was negative.
NOTE Confidence: 0.81014601
00:16:43.620 --> 00:16:46.080 Then the further looking for detail.
NOTE Confidence: 0.81014601
00:16:46.080 --> 00:16:48.792 Indeed who accent identify a 2.3
NOTE Confidence: 0.81014601
00:16:48.792 --> 00:16:51.439 mega base confirmed by Michael Ray
NOTE Confidence: 0.81014601
00:16:51.439 --> 00:16:54.085 later about seeing at 9 Gene was
NOTE Confidence: 0.81014601

00:16:54.085 --> 00:16:56.863 a diluted in the comments on one
NOTE Confidence: 0.81014601

00:16:56.863 --> 00:16:59.775 Q 4 sweet region among all nine
NOTE Confidence: 0.81014601

00:16:59.775 --> 00:17:02.505 gene at one particular gene in
NOTE Confidence: 0.81014601

00:17:02.505 --> 00:17:05.472 the film erator Hardrick how tasty
NOTE Confidence: 0.81014601

00:17:05.472 --> 00:17:08.819 Lincoln FHT in is the Omen Gin?
NOTE Confidence: 0.81014601

00:17:08.820 --> 00:17:11.250 In the literature we know Homosassa
NOTE Confidence: 0.81014601

00:17:11.250 --> 00:17:13.284 mutation interesting 'cause they Sevier
NOTE Confidence: 0.81014601

00:17:13.284 --> 00:17:15.378 in your Natal and stuff allopathy.
NOTE Confidence: 0.81014601

00:17:15.380 --> 00:17:19.258 But ahead was expectation is to contribute
NOTE Confidence: 0.81014601

00:17:19.258 --> 00:17:22.540 leiomyoma and renal cell cancer risk.
NOTE Confidence: 0.81014601

00:17:22.540 --> 00:17:23.420 Interesting Lee,
NOTE Confidence: 0.81014601

00:17:23.420 --> 00:17:25.180 for this contiguous dilution.
NOTE Confidence: 0.81014601

00:17:25.180 --> 00:17:26.264 It's very,
NOTE Confidence: 0.81014601

00:17:26.264 --> 00:17:28.974 very rare in the literature.
NOTE Confidence: 0.81014601

00:17:28.980 --> 00:17:30.500 So this is a.
NOTE Confidence: 0.81014601

00:17:30.500 --> 00:17:32.400 It's also interesting to notice

NOTE Confidence: 0.81014601

00:17:32.400 --> 00:17:35.149 this gene is in the TCA cycle and

NOTE Confidence: 0.81014601

00:17:35.149 --> 00:17:37.499 the key enzyme in the TCA cycle,

NOTE Confidence: 0.81014601

00:17:37.500 --> 00:17:40.699 which is a contributor to condition in

NOTE Confidence: 0.81014601

00:17:40.699 --> 00:17:43.989 the heavens eggs versus the homozygous.

NOTE Confidence: 0.81014601

00:17:43.990 --> 00:17:46.528 The next one is very quickly,

NOTE Confidence: 0.81014601

00:17:46.530 --> 00:17:48.038 so see you often.

NOTE Confidence: 0.81014601

00:17:48.038 --> 00:17:49.923 Time is clearly challenge and

NOTE Confidence: 0.81014601

00:17:49.923 --> 00:17:51.180 your physicians,

NOTE Confidence: 0.81014601

00:17:51.180 --> 00:17:52.872 persistent efforts or technical

NOTE Confidence: 0.81014601

00:17:52.872 --> 00:17:54.987 improvement can lead into diagnosis,

NOTE Confidence: 0.81014601

00:17:54.990 --> 00:17:57.944 which shows you know re analysis here,

NOTE Confidence: 0.81014601

00:17:57.950 --> 00:18:00.060 which is 9 years older,

NOTE Confidence: 0.81014601

00:18:00.060 --> 00:18:02.598 2018 with the Autism no diagnosis

NOTE Confidence: 0.81014601

00:18:02.598 --> 00:18:04.290 industry will come back.

NOTE Confidence: 0.81014601

00:18:04.290 --> 00:18:07.350 Return with the whole exome reanalysis

NOTE Confidence: 0.81014601

00:18:07.350 --> 00:18:10.448 will indeed find the cause for
NOTE Confidence: 0.81014601

00:18:10.448 --> 00:18:12.858 her for his autism diagnosis.
NOTE Confidence: 0.81014601

00:18:12.860 --> 00:18:15.590 So very quickly it's gonna last the
NOTE Confidence: 0.81014601

00:18:15.590 --> 00:18:18.706 case which I want to share with you,
NOTE Confidence: 0.81014601

00:18:18.710 --> 00:18:19.853 which is interesting.
NOTE Confidence: 0.81014601

00:18:19.853 --> 00:18:23.015 Which is us that's the case I learn
NOTE Confidence: 0.81014601

00:18:23.015 --> 00:18:25.892 from Duke as part of a genomic
NOTE Confidence: 0.81014601

00:18:25.892 --> 00:18:28.069 sequencing program is 15 months old.
NOTE Confidence: 0.81014601

00:18:28.070 --> 00:18:30.405 Girl shows you progressive neurological
NOTE Confidence: 0.81014601

00:18:30.405 --> 00:18:33.106 deterioration can occur on set was
NOTE Confidence: 0.81014601

00:18:33.106 --> 00:18:35.474 to try out of many men in treatment.
NOTE Confidence: 0.81014601

00:18:35.480 --> 00:18:38.210 And this including this or cortico steroid,
NOTE Confidence: 0.81014601

00:18:38.210 --> 00:18:40.625 Ivy IG and no significant
NOTE Confidence: 0.81014601

00:18:40.625 --> 00:18:42.074 improvement was considered.
NOTE Confidence: 0.81014601

00:18:42.080 --> 00:18:43.728 Chemotherapy by neurology and
NOTE Confidence: 0.81014601

00:18:43.728 --> 00:18:45.791 the families insisted say, OK,

NOTE Confidence: 0.81014601
00:18:45.791 --> 00:18:48.257 you need to figure out what's
NOTE Confidence: 0.81014601
00:18:48.257 --> 00:18:49.079 the radiologist.
NOTE Confidence: 0.81014601
00:18:49.080 --> 00:18:51.621 Let me present to us asking help
NOTE Confidence: 0.81014601
00:18:51.621 --> 00:18:53.620 for the genetic evaluation.
NOTE Confidence: 0.81014601
00:18:53.620 --> 00:18:57.319 Indeed, this is a very quick a whole accent.
NOTE Confidence: 0.81014601
00:18:57.320 --> 00:19:00.001 11 days and we come from the
NOTE Confidence: 0.81014601
00:19:00.001 --> 00:19:03.009 mutation in the one off the vitamin
NOTE Confidence: 0.81014601
00:19:03.009 --> 00:19:05.667 B2 transporter and in the weeks
NOTE Confidence: 0.81014601
00:19:05.763 --> 00:19:08.109 three we offer that high dose
NOTE Confidence: 0.81014601
00:19:08.109 --> 00:19:09.673 to offer up for
NOTE Confidence: 0.82039505
00:19:09.680 --> 00:19:12.520 even treatment. Here's the outcome.
NOTE Confidence: 0.82039505
00:19:12.520 --> 00:19:13.820 Quickly I can see it.
NOTE Confidence: 0.8001305
00:19:16.010 --> 00:19:17.390 Show the video.
NOTE Confidence: 0.8451408
00:19:20.280 --> 00:19:24.296 Oh, I cannot see the videos now working.
NOTE Confidence: 0.7038002
00:19:29.400 --> 00:19:33.120 Oh, I'm unfortunate videos not working.
NOTE Confidence: 0.7038002

00:19:33.120 --> 00:19:35.220 Erica, can you help it? Look at video.

NOTE Confidence: 0.8398877

00:19:38.790 --> 00:19:41.076 So basically, for week after treatment,

NOTE Confidence: 0.8398877

00:19:41.080 --> 00:19:43.348 I wish you can see the video

NOTE Confidence: 0.8398877

00:19:43.348 --> 00:19:45.280 is a strikingly improvement.

NOTE Confidence: 0.8398877

00:19:45.280 --> 00:19:47.260 Even only four weeks with high

NOTE Confidence: 0.8398877

00:19:47.260 --> 00:19:49.122 dose of vitamin B2 treatment

NOTE Confidence: 0.8398877

00:19:49.122 --> 00:19:51.006 for this particular case,

NOTE Confidence: 0.8398877

00:19:51.010 --> 00:19:53.761 let's move on to the next slide.