

WEBVTT

NOTE duration:"01:08:21"

NOTE recognizability:0.857

NOTE language:en-us

NOTE Confidence: 0.840858526666667

00:00:00.000 --> 00:00:02.460 Well, I think we'll get started

NOTE Confidence: 0.840858526666667

00:00:02.460 --> 00:00:04.588 up so my name is Claire Healy.

NOTE Confidence: 0.840858526666667

00:00:04.590 --> 00:00:06.977 I'm one of the managers in this

NOTE Confidence: 0.840858526666667

00:00:06.977 --> 00:00:08.813 Milo cancer genetics and prevention

NOTE Confidence: 0.840858526666667

00:00:08.813 --> 00:00:11.051 program and we're very excited to

NOTE Confidence: 0.840858526666667

00:00:11.051 --> 00:00:13.376 have all of you join us tonight.

NOTE Confidence: 0.840858526666667

00:00:13.380 --> 00:00:15.809 This is a two night seminar series

NOTE Confidence: 0.840858526666667

00:00:15.809 --> 00:00:18.142 and we've titled it Identifying and

NOTE Confidence: 0.840858526666667

00:00:18.142 --> 00:00:20.182 caring for individuals with high

NOTE Confidence: 0.840858526666667

00:00:20.182 --> 00:00:22.770 risk and inherited cancer syndrome

NOTE Confidence: 0.840858526666667

00:00:22.770 --> 00:00:24.569 and the goal of this seminar series

NOTE Confidence: 0.840858526666667

00:00:24.569 --> 00:00:26.421 is really to help provide some

NOTE Confidence: 0.840858526666667

00:00:26.421 --> 00:00:28.473 information to our colleagues in the

NOTE Confidence: 0.840858526666667

00:00:28.473 --> 00:00:30.427 community on how to recognize patients.
NOTE Confidence: 0.840858526666667

00:00:30.430 --> 00:00:33.286 At elevated risk to develop cancer
NOTE Confidence: 0.840858526666667

00:00:33.290 --> 00:00:36.503 and that those who may have an
NOTE Confidence: 0.840858526666667

00:00:36.503 --> 00:00:38.350 underlying hereditary cancer risk.
NOTE Confidence: 0.840858526666667

00:00:38.350 --> 00:00:40.660 There will be utilizing their
NOTE Confidence: 0.840858526666667

00:00:40.660 --> 00:00:41.984 Q&A feature for questions,
NOTE Confidence: 0.840858526666667

00:00:41.984 --> 00:00:44.418 so feel free to type your questions
NOTE Confidence: 0.840858526666667

00:00:44.418 --> 00:00:47.811 in as the talks goes on and we will
NOTE Confidence: 0.840858526666667

00:00:47.811 --> 00:00:50.030 plan to have a question and answer
NOTE Confidence: 0.840858526666667

00:00:50.105 --> 00:00:52.590 session at the end with our speaker,
NOTE Confidence: 0.840858526666667

00:00:52.590 --> 00:00:54.165 so I'll introduce our speaker
NOTE Confidence: 0.840858526666667

00:00:54.165 --> 00:00:56.199 and then turn things over to her.
NOTE Confidence: 0.840858526666667

00:00:56.200 --> 00:00:58.324 So tonight we're pleased to welcome
NOTE Confidence: 0.840858526666667

00:00:58.324 --> 00:01:01.098 Amy Kelly and Amy received her Masters
NOTE Confidence: 0.840858526666667

00:01:01.098 --> 00:01:03.666 degree in genetic counseling from the
NOTE Confidence: 0.840858526666667

00:01:03.666 --> 00:01:06.438 ICANN School of Medicine at Mount Sinai.

NOTE Confidence: 0.840858526666667
00:01:06.440 --> 00:01:07.380 This band with a smile.
NOTE Confidence: 0.840858526666667
00:01:07.380 --> 00:01:09.230 Oh cancer genetics and prevention
NOTE Confidence: 0.840858526666667
00:01:09.230 --> 00:01:11.919 program for four years and she's one
NOTE Confidence: 0.840858526666667
00:01:11.919 --> 00:01:13.854 of our more experienced counselors
NOTE Confidence: 0.840858526666667
00:01:13.860 --> 00:01:16.164 at a genetic counselor to level
NOTE Confidence: 0.840858526666667
00:01:16.164 --> 00:01:18.381 and her research interests include
NOTE Confidence: 0.840858526666667
00:01:18.381 --> 00:01:20.217 hereditary pancreatic cancer.
NOTE Confidence: 0.840858526666667
00:01:20.220 --> 00:01:21.921 So we're thrilled to have Amy and
NOTE Confidence: 0.840858526666667
00:01:21.921 --> 00:01:23.478 Amy thank you for joining us.
NOTE Confidence: 0.840858526666667
00:01:23.480 --> 00:01:24.746 I'll turn it over to you.
NOTE Confidence: 0.87619315375
00:01:27.280 --> 00:01:29.245 Great, thanks so much Claire
NOTE Confidence: 0.87619315375
00:01:29.245 --> 00:01:30.424 for the introduction.
NOTE Confidence: 0.87619315375
00:01:30.430 --> 00:01:31.510 Good evening everyone.
NOTE Confidence: 0.87619315375
00:01:31.510 --> 00:01:33.670 I'm very excited to be talking
NOTE Confidence: 0.87619315375
00:01:33.670 --> 00:01:35.488 to you about hereditary cancer
NOTE Confidence: 0.87619315375

00:01:35.488 --> 00:01:38.020 and things to look for and things
NOTE Confidence: 0.87619315375

00:01:38.020 --> 00:01:39.915 to consider when thinking about
NOTE Confidence: 0.87619315375

00:01:39.915 --> 00:01:42.354 a referral to harass her cancer.
NOTE Confidence: 0.87619315375

00:01:42.354 --> 00:01:46.909 So I am just going to share my screen.
NOTE Confidence: 0.45747572

00:01:58.980 --> 00:02:02.201 Amy, I'm not seeing your slides. Yep,
NOTE Confidence: 0.45747572

00:02:02.201 --> 00:02:06.120 I just put it on pause just for one. OK.
NOTE Confidence: 0.59584844

00:02:08.260 --> 00:02:09.050 And.
NOTE Confidence: 0.73375588

00:02:13.310 --> 00:02:15.214 These things always take a second day.
NOTE Confidence: 0.73375588

00:02:15.220 --> 00:02:19.700 OK, there we go. Great OK perfect.
NOTE Confidence: 0.853983905

00:02:22.040 --> 00:02:25.076 So just for the agenda today,
NOTE Confidence: 0.853983905

00:02:25.080 --> 00:02:27.527 what I want to go over as I want to review
NOTE Confidence: 0.853983905

00:02:27.527 --> 00:02:29.787 referral guidelines for hereditary cancer,
NOTE Confidence: 0.853983905

00:02:29.790 --> 00:02:32.527 I want to review the more common
NOTE Confidence: 0.853983905

00:02:32.527 --> 00:02:33.700 hereditary cancer indications.
NOTE Confidence: 0.853983905

00:02:33.700 --> 00:02:36.157 Then I want to review a couple of more
NOTE Confidence: 0.853983905

00:02:36.157 --> 00:02:38.409 rare hereditary cancer indications,

NOTE Confidence: 0.853983905

00:02:38.410 --> 00:02:39.640 then talk about self pay,

NOTE Confidence: 0.853983905

00:02:39.640 --> 00:02:41.435 genetic testing and also direct

NOTE Confidence: 0.853983905

00:02:41.435 --> 00:02:43.230 to consumer genetic testing and

NOTE Confidence: 0.853983905

00:02:43.289 --> 00:02:45.371 talk about some of the practical

NOTE Confidence: 0.853983905

00:02:45.371 --> 00:02:47.280 practical aspects of genetic casting.

NOTE Confidence: 0.853983905

00:02:47.280 --> 00:02:49.758 Why someone may want to have genetic

NOTE Confidence: 0.853983905

00:02:49.758 --> 00:02:52.570 testing and talk about genetic counseling.

NOTE Confidence: 0.853983905

00:02:52.570 --> 00:02:54.940 As a whole.

NOTE Confidence: 0.853983905

00:02:54.940 --> 00:02:56.965 So to start talking about

NOTE Confidence: 0.853983905

00:02:56.965 --> 00:02:58.180 hereditary breast cancer,

NOTE Confidence: 0.853983905

00:02:58.180 --> 00:03:00.988 so we cannot talk about hereditary

NOTE Confidence: 0.853983905

00:03:00.988 --> 00:03:03.323 cancer without talking about BRCA

NOTE Confidence: 0.853983905

00:03:03.323 --> 00:03:06.025 one and BRCA 2 related to hereditary

NOTE Confidence: 0.853983905

00:03:06.025 --> 00:03:08.658 breast and ovarian cancer syndrome.

NOTE Confidence: 0.853983905

00:03:08.660 --> 00:03:11.528 These genes are probably the poster

NOTE Confidence: 0.853983905

00:03:11.528 --> 00:03:13.440 children of hereditary cancer
NOTE Confidence: 0.853983905

00:03:13.520 --> 00:03:15.860 testing and I know that everyone
NOTE Confidence: 0.853983905

00:03:15.860 --> 00:03:18.380 here has heard about them before.
NOTE Confidence: 0.853983905

00:03:18.380 --> 00:03:21.019 As we know they make up mutations
NOTE Confidence: 0.853983905

00:03:21.019 --> 00:03:22.150 in these genes.
NOTE Confidence: 0.853983905

00:03:22.150 --> 00:03:23.770 Germline mutations in these genes
NOTE Confidence: 0.853983905

00:03:23.770 --> 00:03:26.134 are associated with a high risk of
NOTE Confidence: 0.853983905

00:03:26.134 --> 00:03:28.180 breast cancer in women which depends
NOTE Confidence: 0.853983905

00:03:28.180 --> 00:03:29.717 slightly dependent on BRCA one or
NOTE Confidence: 0.853983905

00:03:29.717 --> 00:03:31.710 BRCA 2 but the risk is between 50
NOTE Confidence: 0.853983905

00:03:31.710 --> 00:03:34.475 to 75% lifetime risk and also a
NOTE Confidence: 0.853983905

00:03:34.475 --> 00:03:36.242 significantly increased risk of
NOTE Confidence: 0.853983905

00:03:36.242 --> 00:03:38.828 ovarian cancer in women with again
NOTE Confidence: 0.853983905

00:03:38.828 --> 00:03:40.976 arranged depending on the gene but
NOTE Confidence: 0.853983905

00:03:40.976 --> 00:03:44.328 between 15 up to 60% lifetime risk.
NOTE Confidence: 0.853983905

00:03:44.328 --> 00:03:48.104 So definitely significant lifetime risks and.

NOTE Confidence: 0.853983905

00:03:48.104 --> 00:03:49.680 This this I mentioned.

NOTE Confidence: 0.853983905

00:03:49.680 --> 00:03:51.108 These jeans are the poster children

NOTE Confidence: 0.853983905

00:03:51.108 --> 00:03:52.909 of why we do genetic testing.

NOTE Confidence: 0.853983905

00:03:52.910 --> 00:03:54.644 Is that even though these are

NOTE Confidence: 0.853983905

00:03:54.644 --> 00:03:56.489 considered to be high risk genes,

NOTE Confidence: 0.853983905

00:03:56.490 --> 00:03:58.158 there are management guidelines.

NOTE Confidence: 0.853983905

00:03:58.158 --> 00:04:00.243 There's screening options for women

NOTE Confidence: 0.853983905

00:04:00.243 --> 00:04:02.407 who test positive for a mutation,

NOTE Confidence: 0.853983905

00:04:02.410 --> 00:04:05.650 BRCA one and BRCA 2 in terms of beginning

NOTE Confidence: 0.853983905

00:04:05.650 --> 00:04:07.960 breast imaging at a younger age.

NOTE Confidence: 0.853983905

00:04:07.960 --> 00:04:10.720 Adding on breast MRI for more

NOTE Confidence: 0.853983905

00:04:10.720 --> 00:04:11.640 sensitive imaging.

NOTE Confidence: 0.853983905

00:04:11.640 --> 00:04:12.768 Prophylactic bilateral mastectomy's.

NOTE Confidence: 0.853983905

00:04:12.768 --> 00:04:15.024 I feel like they're often talked

NOTE Confidence: 0.853983905

00:04:15.024 --> 00:04:16.506 about in the media at once.

NOTE Confidence: 0.853983905

00:04:16.510 --> 00:04:17.770 Part of Angelina Jolie.
NOTE Confidence: 0.853983905

00:04:17.770 --> 00:04:20.503 Is an option women decided to pursue and
NOTE Confidence: 0.853983905

00:04:20.503 --> 00:04:22.855 of course and of course prophylactic
NOTE Confidence: 0.853983905

00:04:22.855 --> 00:04:25.305 bilateral salpingo over ectomy to be
NOTE Confidence: 0.853983905

00:04:25.305 --> 00:04:26.909 preventative against ovarian cancer.
NOTE Confidence: 0.853983905

00:04:26.910 --> 00:04:29.590 So in the advent of genetic testing for
NOTE Confidence: 0.853983905

00:04:29.590 --> 00:04:31.574 these genes and establishing guidelines
NOTE Confidence: 0.853983905

00:04:31.574 --> 00:04:34.058 you know women and other people
NOTE Confidence: 0.853983905

00:04:34.058 --> 00:04:35.960 who are at higher risk of pressure,
NOTE Confidence: 0.853983905

00:04:35.960 --> 00:04:38.486 right cancer have really been able
NOTE Confidence: 0.853983905

00:04:38.486 --> 00:04:40.973 to take this information and use
NOTE Confidence: 0.853983905

00:04:40.973 --> 00:04:43.157 it to actually lengthen their life
NOTE Confidence: 0.853983905

00:04:43.157 --> 00:04:45.175 and have a better quality of life.
NOTE Confidence: 0.853983905

00:04:45.175 --> 00:04:46.035 So really we can't.
NOTE Confidence: 0.853983905

00:04:46.040 --> 00:04:47.365 We can't talk about hereditary
NOTE Confidence: 0.853983905

00:04:47.365 --> 00:04:48.690 cancer without talking about these.

NOTE Confidence: 0.853983905

00:04:48.690 --> 00:04:50.990 2 genes.

NOTE Confidence: 0.853983905

00:04:50.990 --> 00:04:53.270 However.

NOTE Confidence: 0.853983905

00:04:53.270 --> 00:04:56.470 We are now past the era of only

NOTE Confidence: 0.853983905

00:04:56.470 --> 00:04:58.729 testing BRCA one and BRCA 2.

NOTE Confidence: 0.853983905

00:04:58.730 --> 00:05:01.850 It's not uncommon for me to see a

NOTE Confidence: 0.853983905

00:05:01.850 --> 00:05:04.901 patient and say oh I'm here for the

NOTE Confidence: 0.853983905

00:05:04.901 --> 00:05:07.568 the Braca test but now you know,

NOTE Confidence: 0.853983905

00:05:07.570 --> 00:05:11.650 especially since about 2015 now.

NOTE Confidence: 0.853983905

00:05:11.650 --> 00:05:14.674 Our testing includes many other genes

NOTE Confidence: 0.853983905

00:05:14.674 --> 00:05:18.427 that we now know of relating to

NOTE Confidence: 0.853983905

00:05:18.427 --> 00:05:21.431 hereditary risk of breast cancer and

NOTE Confidence: 0.853983905

00:05:21.431 --> 00:05:23.258 we know that these genes they they

NOTE Confidence: 0.853983905

00:05:23.258 --> 00:05:25.346 can be arranged so some of these.

NOTE Confidence: 0.853983905

00:05:25.350 --> 00:05:27.120 And broke up a little categories

NOTE Confidence: 0.853983905

00:05:27.120 --> 00:05:29.615 but BRCA one and two are related to

NOTE Confidence: 0.853983905

00:05:29.615 --> 00:05:31.493 are considered a high risk breast
NOTE Confidence: 0.853983905

00:05:31.556 --> 00:05:33.211 cancer gene and there's other
NOTE Confidence: 0.853983905

00:05:33.211 --> 00:05:34.866 genes that fall into that
NOTE Confidence: 0.930804264285714

00:05:34.870 --> 00:05:38.335 category as well that also have guidelines.
NOTE Confidence: 0.930804264285714

00:05:38.340 --> 00:05:40.338 Now some of these higher risk
NOTE Confidence: 0.930804264285714

00:05:40.338 --> 00:05:42.103 genes often have other features
NOTE Confidence: 0.930804264285714

00:05:42.103 --> 00:05:44.299 that are uncommon or more rare.
NOTE Confidence: 0.930804264285714

00:05:44.300 --> 00:05:46.550 For example, the gene TP 53.
NOTE Confidence: 0.930804264285714

00:05:46.550 --> 00:05:48.686 Many people have heard of related
NOTE Confidence: 0.930804264285714

00:05:48.686 --> 00:05:51.497 to leave from many syndrome which is
NOTE Confidence: 0.930804264285714

00:05:51.497 --> 00:05:53.687 most characteristic of early onset
NOTE Confidence: 0.930804264285714

00:05:53.687 --> 00:05:56.420 sarcomas or early onset osteosarcomas.
NOTE Confidence: 0.930804264285714

00:05:56.420 --> 00:05:58.212 Very young breast cancer
NOTE Confidence: 0.930804264285714

00:05:58.212 --> 00:05:59.556 in childhood cancers.
NOTE Confidence: 0.930804264285714

00:05:59.560 --> 00:06:03.112 So not every single high risk gene is
NOTE Confidence: 0.930804264285714

00:06:03.112 --> 00:06:06.171 necessarily a gene of interest for a

NOTE Confidence: 0.930804264285714

00:06:06.171 --> 00:06:08.488 woman who has early onset breast cancer.

NOTE Confidence: 0.930804264285714

00:06:08.490 --> 00:06:10.626 But with the advent of larger

NOTE Confidence: 0.930804264285714

00:06:10.626 --> 00:06:13.218 testing we can now test for these

NOTE Confidence: 0.930804264285714

00:06:13.218 --> 00:06:15.028 genes all the same time.

NOTE Confidence: 0.930804264285714

00:06:15.030 --> 00:06:17.358 And what we now know is there are

NOTE Confidence: 0.930804264285714

00:06:17.358 --> 00:06:20.022 other genes that fall into more of the

NOTE Confidence: 0.930804264285714

00:06:20.022 --> 00:06:22.600 moderate risk which would be ATM and CHEK 2.

NOTE Confidence: 0.930804264285714

00:06:22.600 --> 00:06:24.904 You can see in the middle and the

NOTE Confidence: 0.930804264285714

00:06:24.904 --> 00:06:27.102 blue column there you know the risk.

NOTE Confidence: 0.930804264285714

00:06:27.102 --> 00:06:29.360 Ranges between 20 to 40% lifetime risk.

NOTE Confidence: 0.930804264285714

00:06:29.360 --> 00:06:32.750 You can see in the high risk category PAL B2,

NOTE Confidence: 0.930804264285714

00:06:32.750 --> 00:06:34.478 which has been in the news.

NOTE Confidence: 0.930804264285714

00:06:34.480 --> 00:06:36.250 More recently we're getting a

NOTE Confidence: 0.930804264285714

00:06:36.250 --> 00:06:38.280 lot of messages about PAL B2,

NOTE Confidence: 0.930804264285714

00:06:38.280 --> 00:06:40.198 but we consider to be a moderate

NOTE Confidence: 0.930804264285714
00:06:40.198 --> 00:06:42.248 to high risk breast cancer gene,
NOTE Confidence: 0.930804264285714
00:06:42.250 --> 00:06:44.861 which is why it's in the high
NOTE Confidence: 0.930804264285714
00:06:44.861 --> 00:06:47.056 risk category where the risk can
NOTE Confidence: 0.930804264285714
00:06:47.056 --> 00:06:49.184 be upwards of 58% lifetime risk.
NOTE Confidence: 0.930804264285714
00:06:49.184 --> 00:06:51.452 So there are other genes that
NOTE Confidence: 0.930804264285714
00:06:51.452 --> 00:06:54.061 we definitely want to be testing
NOTE Confidence: 0.930804264285714
00:06:54.061 --> 00:06:56.960 individuals for if they have a family.
NOTE Confidence: 0.930804264285714
00:06:56.960 --> 00:06:58.406 History that's concerning.
NOTE Confidence: 0.930804264285714
00:06:58.406 --> 00:07:00.334 For hereditary breast cancer.
NOTE Confidence: 0.930804264285714
00:07:00.340 --> 00:07:02.460 And now there's you see in the green
NOTE Confidence: 0.930804264285714
00:07:02.460 --> 00:07:04.225 the green column there are genes
NOTE Confidence: 0.930804264285714
00:07:04.225 --> 00:07:06.416 that have an unknown risk of breast
NOTE Confidence: 0.930804264285714
00:07:06.416 --> 00:07:08.636 cancer because as we've studied genes,
NOTE Confidence: 0.930804264285714
00:07:08.640 --> 00:07:10.719 and as we've learned more about genes,
NOTE Confidence: 0.930804264285714
00:07:10.720 --> 00:07:13.051 the risk is sometimes unclear where the
NOTE Confidence: 0.930804264285714

00:07:13.051 --> 00:07:14.991 there might be literature suggesting
NOTE Confidence: 0.930804264285714

00:07:14.991 --> 00:07:17.625 an increased risk of breast cancer,
NOTE Confidence: 0.930804264285714

00:07:17.630 --> 00:07:19.982 but maybe not enough to be
NOTE Confidence: 0.930804264285714

00:07:19.982 --> 00:07:22.399 significant or more data is needed.
NOTE Confidence: 0.930804264285714

00:07:22.400 --> 00:07:24.570 So some of these genes we off
NOTE Confidence: 0.930804264285714

00:07:24.570 --> 00:07:27.130 we can are included on genetic.
NOTE Confidence: 0.930804264285714

00:07:27.130 --> 00:07:30.845 Testing and this again just just
NOTE Confidence: 0.930804264285714

00:07:30.845 --> 00:07:31.736 shows the range.
NOTE Confidence: 0.930804264285714

00:07:31.740 --> 00:07:32.069 Specifically,
NOTE Confidence: 0.930804264285714

00:07:32.069 --> 00:07:34.701 I would say the the major ones that
NOTE Confidence: 0.930804264285714

00:07:34.701 --> 00:07:37.057 we would test for like ATM Chek 2,
NOTE Confidence: 0.930804264285714

00:07:37.060 --> 00:07:37.810 PALB 2,
NOTE Confidence: 0.930804264285714

00:07:37.810 --> 00:07:40.435 where the the lighter blue is the
NOTE Confidence: 0.930804264285714

00:07:40.435 --> 00:07:42.909 higher end end of the range and
NOTE Confidence: 0.930804264285714

00:07:42.909 --> 00:07:45.319 the darker blue is the lower end
NOTE Confidence: 0.930804264285714

00:07:45.319 --> 00:07:47.615 of the range so you can see.

NOTE Confidence: 0.930804264285714
00:07:47.620 --> 00:07:50.600 Regina is different and that's
NOTE Confidence: 0.930804264285714
00:07:50.600 --> 00:07:52.322 that's why we're now past this
NOTE Confidence: 0.930804264285714
00:07:52.322 --> 00:07:54.449 era of only doing testing for BRCA
NOTE Confidence: 0.930804264285714
00:07:54.449 --> 00:07:56.584 one and BRCA 2 because these genes
NOTE Confidence: 0.930804264285714
00:07:56.647 --> 00:07:58.765 some of them do have significant
NOTE Confidence: 0.930804264285714
00:07:58.765 --> 00:08:00.856 risks and most importantly we have
NOTE Confidence: 0.930804264285714
00:08:00.856 --> 00:08:01.870 guidelines for screening.
NOTE Confidence: 0.915834705333333
00:08:04.220 --> 00:08:06.796 Now this is sometimes a very common thing
NOTE Confidence: 0.915834705333333
00:08:06.796 --> 00:08:09.360 that we will see with with patients.
NOTE Confidence: 0.915834705333333
00:08:09.360 --> 00:08:11.815 Usually 1-2 testing has been
NOTE Confidence: 0.915834705333333
00:08:11.815 --> 00:08:14.144 around for at least 20 years,
NOTE Confidence: 0.915834705333333
00:08:14.144 --> 00:08:17.378 so it's not uncommon for for patients to say,
NOTE Confidence: 0.915834705333333
00:08:17.380 --> 00:08:18.718 oh, I've already had this test.
NOTE Confidence: 0.915834705333333
00:08:18.720 --> 00:08:21.030 I had this test in 2012.
NOTE Confidence: 0.915834705333333
00:08:21.030 --> 00:08:22.780 It was it was negative,
NOTE Confidence: 0.915834705333333

00:08:22.780 --> 00:08:25.650 so I was often common to see
NOTE Confidence: 0.915834705333333
00:08:25.650 --> 00:08:27.959 a test report like this.
NOTE Confidence: 0.915834705333333
00:08:27.960 --> 00:08:30.582 But back before I would say
NOTE Confidence: 0.915834705333333
00:08:30.582 --> 00:08:32.870 definitely before 2014 and probably
NOTE Confidence: 0.915834705333333
00:08:32.870 --> 00:08:35.576 a little bit even before then.
NOTE Confidence: 0.915834705333333
00:08:35.580 --> 00:08:38.370 Specially BRCA one and two testing
NOTE Confidence: 0.915834705333333
00:08:38.370 --> 00:08:40.940 was really limited to sequencing
NOTE Confidence: 0.915834705333333
00:08:40.940 --> 00:08:44.268 only and sequencing for BRCA one
NOTE Confidence: 0.915834705333333
00:08:44.268 --> 00:08:49.326 and BRCA 2 can detect up to 80% of
NOTE Confidence: 0.915834705333333
00:08:49.326 --> 00:08:52.169 detectable pathogenic mutations and
NOTE Confidence: 0.915834705333333
00:08:52.169 --> 00:08:53.605 sequencing covers the clinically
NOTE Confidence: 0.915834705333333
00:08:53.605 --> 00:08:55.400 important regions of each gene,
NOTE Confidence: 0.915834705333333
00:08:55.400 --> 00:08:56.820 including the coding exons,
NOTE Confidence: 0.915834705333333
00:08:56.820 --> 00:09:00.071 so that is that is a very informative
NOTE Confidence: 0.915834705333333
00:09:00.071 --> 00:09:03.130 test but sequencing cannot detect
NOTE Confidence: 0.915834705333333
00:09:03.130 --> 00:09:05.670 large deletions or duplications.

NOTE Confidence: 0.915834705333333

00:09:05.670 --> 00:09:08.658 Of your C1 and BRCA 2 so it is

NOTE Confidence: 0.915834705333333

00:09:08.658 --> 00:09:11.193 important when a patient has had

NOTE Confidence: 0.915834705333333

00:09:11.193 --> 00:09:13.872 genetic testing to know what year

NOTE Confidence: 0.915834705333333

00:09:13.872 --> 00:09:16.698 was it and also confirmed with

NOTE Confidence: 0.915834705333333

00:09:16.698 --> 00:09:19.056 records because for example in this

NOTE Confidence: 0.915834705333333

00:09:19.056 --> 00:09:21.730 case it says BRCA one and BRCA

NOTE Confidence: 0.915834705333333

00:09:21.730 --> 00:09:24.090 1 sequencing BRCA 2 sequencing.

NOTE Confidence: 0.915834705333333

00:09:24.090 --> 00:09:27.198 No mutation detected but this test

NOTE Confidence: 0.915834705333333

00:09:27.198 --> 00:09:30.861 would not include what we consider

NOTE Confidence: 0.915834705333333

00:09:30.861 --> 00:09:33.099 large rearrangement analysis.

NOTE Confidence: 0.915834705333333

00:09:33.100 --> 00:09:33.982 Which is on,

NOTE Confidence: 0.915834705333333

00:09:33.982 --> 00:09:36.968 but you can say he see here BRCA one

NOTE Confidence: 0.915834705333333

00:09:36.968 --> 00:09:39.950 and BRCA 2 full gene rearrangement also

NOTE Confidence: 0.915834705333333

00:09:39.950 --> 00:09:42.266 sometimes called deletion duplication

NOTE Confidence: 0.915834705333333

00:09:42.266 --> 00:09:44.678 which detects intragenic deletions

NOTE Confidence: 0.915834705333333

00:09:44.678 --> 00:09:47.891 and duplications at at single exons.
NOTE Confidence: 0.9158347053333333

00:09:47.891 --> 00:09:51.160 Some people also call this a the
NOTE Confidence: 0.9158347053333333

00:09:51.264 --> 00:09:54.510 Bart test because back until 2013,
NOTE Confidence: 0.9158347053333333

00:09:54.510 --> 00:09:56.195 Myriad Genetics had the patent
NOTE Confidence: 0.9158347053333333

00:09:56.195 --> 00:09:57.880 on BRCA one and two,
NOTE Confidence: 0.9158347053333333

00:09:57.880 --> 00:10:00.912 so their actual test name for BRCA one
NOTE Confidence: 0.9158347053333333

00:10:00.912 --> 00:10:04.060 and two large rearrangement testing.
NOTE Confidence: 0.9158347053333333

00:10:04.060 --> 00:10:06.760 Was Bart so some people might.
NOTE Confidence: 0.9158347053333333

00:10:06.760 --> 00:10:08.170 You might hear patients talking
NOTE Confidence: 0.9158347053333333

00:10:08.170 --> 00:10:09.016 about bar testing.
NOTE Confidence: 0.9158347053333333

00:10:09.020 --> 00:10:10.796 You might hear about bar testing.
NOTE Confidence: 0.9158347053333333

00:10:10.800 --> 00:10:15.388 Bart is just a name that Myriad gave
NOTE Confidence: 0.9158347053333333

00:10:15.388 --> 00:10:17.920 their own tasks for large rearrangement.
NOTE Confidence: 0.9158347053333333

00:10:17.920 --> 00:10:19.805 But now that Myriad Genetics
NOTE Confidence: 0.9158347053333333

00:10:19.805 --> 00:10:21.313 don't have the patent,
NOTE Confidence: 0.9158347053333333

00:10:21.320 --> 00:10:22.046 there's plenty.

NOTE Confidence: 0.915834705333333
00:10:22.046 --> 00:10:23.498 There's many other labs
NOTE Confidence: 0.915834705333333
00:10:23.498 --> 00:10:24.950 doing doing this test,
NOTE Confidence: 0.915834705333333
00:10:24.950 --> 00:10:26.098 and nowadays it's standard
NOTE Confidence: 0.915834705333333
00:10:26.098 --> 00:10:27.533 of care when we do.
NOTE Confidence: 0.915834705333333
00:10:27.540 --> 00:10:31.140 BRCA one and two testing to do full gene
NOTE Confidence: 0.915834705333333
00:10:31.140 --> 00:10:33.578 sequencing and deletion duplication.
NOTE Confidence: 0.915834705333333
00:10:33.580 --> 00:10:35.925 So what's also important is
NOTE Confidence: 0.915834705333333
00:10:35.925 --> 00:10:38.853 that if a patient reports BRC
NOTE Confidence: 0.915834705333333
00:10:38.853 --> 00:10:41.288 wanted to testing prior 2013,
NOTE Confidence: 0.915834705333333
00:10:41.290 --> 00:10:43.544 one is possible they may have only
NOTE Confidence: 0.915834705333333
00:10:43.544 --> 00:10:45.995 had BRCA one and two sequencing
NOTE Confidence: 0.915834705333333
00:10:45.995 --> 00:10:47.875 and not deletion duplication.
NOTE Confidence: 0.915834705333333
00:10:47.880 --> 00:10:50.771 But even if they had full gene
NOTE Confidence: 0.915834705333333
00:10:50.771 --> 00:10:53.010 sequencing and deletion duplication,
NOTE Confidence: 0.915834705333333
00:10:53.010 --> 00:10:55.185 updated genetic testing is likely
NOTE Confidence: 0.915834705333333

00:10:55.185 --> 00:10:57.360 to be indicated because prior
NOTE Confidence: 0.915834705333333
00:10:57.438 --> 00:10:59.592 2013 those other genes I had
NOTE Confidence: 0.915834705333333
00:10:59.592 --> 00:11:01.600 talked about related to hereditary
NOTE Confidence: 0.915834705333333
00:11:01.600 --> 00:11:04.075 breast cancer were not included.
NOTE Confidence: 0.915834705333333
00:11:04.080 --> 00:11:05.235 On genetic testing,
NOTE Confidence: 0.915834705333333
00:11:05.235 --> 00:11:07.545 or at least very rarely included.
NOTE Confidence: 0.915834705333333
00:11:07.550 --> 00:11:10.664 So if patients before 2013 say
NOTE Confidence: 0.915834705333333
00:11:10.664 --> 00:11:13.920 that they had negative testing in,
NOTE Confidence: 0.915834705333333
00:11:13.920 --> 00:11:16.069 likely is worth or a new referral
NOTE Confidence: 0.915834705333333
00:11:16.069 --> 00:11:18.099 to talk about updated testing,
NOTE Confidence: 0.915834705333333
00:11:18.100 --> 00:11:19.840 and they say 2013.
NOTE Confidence: 0.915834705333333
00:11:19.840 --> 00:11:22.051 But even patients that 2014,
NOTE Confidence: 0.915834705333333
00:11:22.051 --> 00:11:24.877 2015 some of these bigger tests,
NOTE Confidence: 0.915834705333333
00:11:24.880 --> 00:11:26.670 these bigger panels were still
NOTE Confidence: 0.915834705333333
00:11:26.670 --> 00:11:27.744 coming into play,
NOTE Confidence: 0.915834705333333
00:11:27.750 --> 00:11:31.047 but I would say anything before 2013

NOTE Confidence: 0.915834705333333
00:11:31.047 --> 00:11:33.980 is definitely worth a new evaluation.
NOTE Confidence: 0.915834705333333
00:11:33.980 --> 00:11:35.350 So I'm talking about hereditary
NOTE Confidence: 0.915834705333333
00:11:35.350 --> 00:11:36.724 breast cancer. So what?
NOTE Confidence: 0.915834705333333
00:11:36.724 --> 00:11:37.960 So what should?
NOTE Confidence: 0.915834705333333
00:11:37.960 --> 00:11:39.395 What should you be thinking
NOTE Confidence: 0.915834705333333
00:11:39.395 --> 00:11:40.830 about what I would indicate
NOTE Confidence: 0.904235814666667
00:11:40.890 --> 00:11:42.460 a referral to cancer genetics?
NOTE Confidence: 0.904235814666667
00:11:42.460 --> 00:11:43.956 If we're thinking about
NOTE Confidence: 0.904235814666667
00:11:43.956 --> 00:11:45.078 hereditary breast cancer?
NOTE Confidence: 0.904235814666667
00:11:45.080 --> 00:11:47.984 So our bread and butter for
NOTE Confidence: 0.904235814666667
00:11:47.984 --> 00:11:49.920 guidelines is NCCN guidelines.
NOTE Confidence: 0.904235814666667
00:11:49.920 --> 00:11:51.628 Based from the National
NOTE Confidence: 0.904235814666667
00:11:51.628 --> 00:11:52.909 Comprehensive Cancer Network,
NOTE Confidence: 0.904235814666667
00:11:52.910 --> 00:11:55.472 and these guidelines change every 6
NOTE Confidence: 0.904235814666667
00:11:55.472 --> 00:11:58.242 to 12 months before it was focusing
NOTE Confidence: 0.904235814666667

00:11:58.242 --> 00:12:01.366 on just pure C1 and BRCA 2 but now
NOTE Confidence: 0.904235814666667

00:12:01.366 --> 00:12:03.640 in the advent of panel testing,
NOTE Confidence: 0.904235814666667

00:12:03.640 --> 00:12:06.368 moving past the era of just BRCA 1.
NOTE Confidence: 0.904235814666667

00:12:06.370 --> 00:12:08.596 And BRCA two we know there are
NOTE Confidence: 0.904235814666667

00:12:08.596 --> 00:12:10.617 other genes if someone would
NOTE Confidence: 0.904235814666667

00:12:10.617 --> 00:12:12.980 meet clinical guidelines for so.
NOTE Confidence: 0.904235814666667

00:12:12.980 --> 00:12:15.640 NCCN used to have a nice package
NOTE Confidence: 0.904235814666667

00:12:15.640 --> 00:12:19.090 of just BRCA one and two relating
NOTE Confidence: 0.904235814666667

00:12:19.090 --> 00:12:21.166 to hereditary breast cancer.
NOTE Confidence: 0.904235814666667

00:12:21.170 --> 00:12:24.026 But now they've broken it up a little
NOTE Confidence: 0.904235814666667

00:12:24.026 --> 00:12:27.230 bit into hereditary breast cancer genes,
NOTE Confidence: 0.904235814666667

00:12:27.230 --> 00:12:30.160 hereditary ovarian cancer genes correct,
NOTE Confidence: 0.904235814666667

00:12:30.160 --> 00:12:30.627 right?
NOTE Confidence: 0.904235814666667

00:12:30.627 --> 00:12:32.495 Pancreatic cancer genes and
NOTE Confidence: 0.904235814666667

00:12:32.495 --> 00:12:34.363 hereditary prostate cancer genes.
NOTE Confidence: 0.904235814666667

00:12:34.370 --> 00:12:36.380 The reason for this is because.

NOTE Confidence: 0.904235814666667

00:12:36.380 --> 00:12:38.823 Mutations in BRCA one and BRCA two

NOTE Confidence: 0.904235814666667

00:12:38.823 --> 00:12:40.800 are related to hereditary breast

NOTE Confidence: 0.904235814666667

00:12:40.800 --> 00:12:43.362 cancer and ovarian cancer in women

NOTE Confidence: 0.904235814666667

00:12:43.362 --> 00:12:46.041 and and breast cancer in men but

NOTE Confidence: 0.904235814666667

00:12:46.041 --> 00:12:48.190 also they are related to prostate.

NOTE Confidence: 0.904235814666667

00:12:48.190 --> 00:12:50.590 Cancer risk in men and pancreatic

NOTE Confidence: 0.904235814666667

00:12:50.590 --> 00:12:52.829 cancer risk in men and women.

NOTE Confidence: 0.904235814666667

00:12:52.830 --> 00:12:56.030 So that's why NCCM has so many guidelines.

NOTE Confidence: 0.904235814666667

00:12:56.030 --> 00:12:57.618 And definitely it's not.

NOTE Confidence: 0.904235814666667

00:12:57.618 --> 00:12:58.809 These are in.

NOTE Confidence: 0.904235814666667

00:12:58.810 --> 00:13:00.426 Again, they change every 6 to 12 months,

NOTE Confidence: 0.904235814666667

00:13:00.430 --> 00:13:02.369 so you are not expected to be

NOTE Confidence: 0.904235814666667

00:13:02.369 --> 00:13:04.359 the expert on these guidelines.

NOTE Confidence: 0.904235814666667

00:13:04.360 --> 00:13:04.706 Again,

NOTE Confidence: 0.904235814666667

00:13:04.706 --> 00:13:05.744 as cancer genetics,

NOTE Confidence: 0.904235814666667

00:13:05.744 --> 00:13:08.570 we want to be a resource to you
NOTE Confidence: 0.904235814666667

00:13:08.570 --> 00:13:09.587 for your patience.
NOTE Confidence: 0.904235814666667

00:13:09.590 --> 00:13:12.348 So just to really distill some of
NOTE Confidence: 0.904235814666667

00:13:12.348 --> 00:13:14.895 these guidelines down and just some
NOTE Confidence: 0.904235814666667

00:13:14.895 --> 00:13:17.045 suggesting subjects things to think
NOTE Confidence: 0.904235814666667

00:13:17.045 --> 00:13:19.340 about when placing a referral.
NOTE Confidence: 0.904235814666667

00:13:19.340 --> 00:13:22.798 Is that if someone with a diagnosis
NOTE Confidence: 0.904235814666667

00:13:22.798 --> 00:13:25.229 of breast cancer under 50,
NOTE Confidence: 0.904235814666667

00:13:25.230 --> 00:13:26.264 anything premenopausal?
NOTE Confidence: 0.904235814666667

00:13:26.264 --> 00:13:29.883 I will say you know there there
NOTE Confidence: 0.904235814666667

00:13:29.883 --> 00:13:32.318 isn't blue for for testing if
NOTE Confidence: 0.904235814666667

00:13:32.318 --> 00:13:35.501 someone is at 50 or at 49 with
NOTE Confidence: 0.904235814666667

00:13:35.501 --> 00:13:37.197 no other family history.
NOTE Confidence: 0.904235814666667

00:13:37.200 --> 00:13:39.264 It's possible this possible they may
NOTE Confidence: 0.904235814666667

00:13:39.264 --> 00:13:41.051 not meet their insurance guidelines
NOTE Confidence: 0.904235814666667

00:13:41.051 --> 00:13:43.181 to be protesting to be clinically

NOTE Confidence: 0.904235814666667

00:13:43.181 --> 00:13:45.490 indicated and meet insurance guidelines.

NOTE Confidence: 0.904235814666667

00:13:45.490 --> 00:13:47.580 Someone would need to be

NOTE Confidence: 0.904235814666667

00:13:47.580 --> 00:13:49.670 diagnosed at 45 or younger.

NOTE Confidence: 0.904235814666667

00:13:49.670 --> 00:13:52.022 But the reason why I posted before

NOTE Confidence: 0.904235814666667

00:13:52.022 --> 00:13:54.467 50s because it is worth a referral,

NOTE Confidence: 0.904235814666667

00:13:54.470 --> 00:13:56.105 because sometimes there may be

NOTE Confidence: 0.904235814666667

00:13:56.105 --> 00:13:57.740 other factors that we would

NOTE Confidence: 0.904235814666667

00:13:57.803 --> 00:13:59.609 want to look into in the family,

NOTE Confidence: 0.904235814666667

00:13:59.610 --> 00:14:01.735 including small family size or

NOTE Confidence: 0.904235814666667

00:14:01.735 --> 00:14:04.330 limited structure or and also also

NOTE Confidence: 0.904235814666667

00:14:04.330 --> 00:14:06.670 just to go through more distant

NOTE Confidence: 0.904235814666667

00:14:06.670 --> 00:14:09.363 relatives and take the time to go

NOTE Confidence: 0.904235814666667

00:14:09.363 --> 00:14:11.173 through a whole family history.

NOTE Confidence: 0.904235814666667

00:14:11.180 --> 00:14:13.048 Now NCCN guidelines recommending

NOTE Confidence: 0.904235814666667

00:14:13.048 --> 00:14:15.383 genetic testing for anyone with

NOTE Confidence: 0.904235814666667

00:14:15.383 --> 00:14:17.500 triple negative breast cancer at
NOTE Confidence: 0.904235814666667

00:14:17.500 --> 00:14:19.870 any age and triple negative breast
NOTE Confidence: 0.904235814666667

00:14:19.936 --> 00:14:22.264 cancer is breast cancer that test
NOTE Confidence: 0.904235814666667

00:14:22.264 --> 00:14:24.340 negative for the estrogen receptor.
NOTE Confidence: 0.904235814666667

00:14:24.340 --> 00:14:26.420 Progesterone receptor and access
NOTE Confidence: 0.904235814666667

00:14:26.420 --> 00:14:29.020 of the her two protein.
NOTE Confidence: 0.904235814666667

00:14:29.020 --> 00:14:30.820 So this means essentially that the
NOTE Confidence: 0.904235814666667

00:14:30.820 --> 00:14:33.440 growth of the cancer is not fueled by
NOTE Confidence: 0.904235814666667

00:14:33.440 --> 00:14:35.140 the hormones estrogen and progesterone,
NOTE Confidence: 0.904235814666667

00:14:35.140 --> 00:14:37.247 and is also not fueled by the
NOTE Confidence: 0.904235814666667

00:14:37.247 --> 00:14:38.150 her two protein.
NOTE Confidence: 0.904235814666667

00:14:38.150 --> 00:14:40.678 So triple negative breast cancer tends to be.
NOTE Confidence: 0.904235814666667

00:14:40.680 --> 00:14:42.876 Much harder to treat and more
NOTE Confidence: 0.904235814666667

00:14:42.876 --> 00:14:44.833 aggressive because it does not
NOTE Confidence: 0.904235814666667

00:14:44.833 --> 00:14:47.023 respond to hormonal therapy, so.
NOTE Confidence: 0.904235814666667

00:14:47.023 --> 00:14:49.588 NCCN guidelines is now recommending

NOTE Confidence: 0.904235814666667

00:14:49.588 --> 00:14:52.540 triple negative breast cancer at any age.

NOTE Confidence: 0.904235814666667

00:14:52.540 --> 00:14:54.068 Because triple negative breast

NOTE Confidence: 0.904235814666667

00:14:54.068 --> 00:14:56.867 cancer is more likely to have a

NOTE Confidence: 0.904235814666667

00:14:56.867 --> 00:14:58.623 hereditary component than non

NOTE Confidence: 0.904235814666667

00:14:58.623 --> 00:15:00.379 triple negative breast cancer.

NOTE Confidence: 0.869235684

00:15:00.380 --> 00:15:03.230 For for most insurance purposes,

NOTE Confidence: 0.869235684

00:15:03.230 --> 00:15:05.138 since insurance tends to be slower

NOTE Confidence: 0.869235684

00:15:05.138 --> 00:15:06.800 on the uptake than guidelines

NOTE Confidence: 0.869235684

00:15:06.800 --> 00:15:09.110 that change every 6 to 12 months,

NOTE Confidence: 0.869235684

00:15:09.110 --> 00:15:11.280 insurance won't cover testing and

NOTE Confidence: 0.869235684

00:15:11.280 --> 00:15:13.895 without any other family history unless

NOTE Confidence: 0.869235684

00:15:13.895 --> 00:15:16.485 someone is diagnosed at 60 or younger.

NOTE Confidence: 0.869235684

00:15:16.490 --> 00:15:18.026 So something to keep in mind,

NOTE Confidence: 0.869235684

00:15:18.030 --> 00:15:21.444 but for in terms of a referral, it is def.

NOTE Confidence: 0.869235684

00:15:21.444 --> 00:15:22.756 I would say indicated,

NOTE Confidence: 0.869235684

00:15:22.760 --> 00:15:24.580 so something that we can
NOTE Confidence: 0.869235684

00:15:24.580 --> 00:15:26.400 talk about with the patient.
NOTE Confidence: 0.869235684

00:15:26.400 --> 00:15:29.634 Anyone with ovarian flopping to her primary
NOTE Confidence: 0.869235684

00:15:29.634 --> 00:15:32.497 parotta Neil Cancer at any age is genetic.
NOTE Confidence: 0.869235684

00:15:32.497 --> 00:15:33.838 Testing is indicated.
NOTE Confidence: 0.869235684

00:15:33.838 --> 00:15:37.139 Men diagnosed with breast cancer at any age.
NOTE Confidence: 0.869235684

00:15:37.140 --> 00:15:38.936 Metastatic intraductal prostate cancer
NOTE Confidence: 0.869235684

00:15:38.936 --> 00:15:42.336 diagnosis at any age and this is a
NOTE Confidence: 0.869235684

00:15:42.336 --> 00:15:44.555 more I would say a newer development
NOTE Confidence: 0.869235684

00:15:44.555 --> 00:15:47.395 within the last two years where there's
NOTE Confidence: 0.869235684

00:15:47.395 --> 00:15:49.441 been literature has suggested that
NOTE Confidence: 0.869235684

00:15:49.441 --> 00:15:52.248 men who have meta static or prostate
NOTE Confidence: 0.869235684

00:15:52.248 --> 00:15:54.730 cancer that's more locally advanced,
NOTE Confidence: 0.869235684

00:15:54.730 --> 00:15:56.984 particularly if it's a higher Gleason score.
NOTE Confidence: 0.869235684

00:15:56.990 --> 00:15:59.882 Which is used to rate aggressiveness
NOTE Confidence: 0.869235684

00:15:59.882 --> 00:16:01.810 of the prostate cancer.

NOTE Confidence: 0.869235684

00:16:01.810 --> 00:16:03.586 There is a higher prevalence of

NOTE Confidence: 0.869235684

00:16:03.586 --> 00:16:05.130 germline mutations in that group,

NOTE Confidence: 0.869235684

00:16:05.130 --> 00:16:07.741 so that's why for men with metastatic

NOTE Confidence: 0.869235684

00:16:07.741 --> 00:16:09.350 or intraductal prostate cancer,

NOTE Confidence: 0.869235684

00:16:09.350 --> 00:16:11.074 regardless of their AIDS,

NOTE Confidence: 0.869235684

00:16:11.074 --> 00:16:12.798 genetic testing is indicated,

NOTE Confidence: 0.869235684

00:16:12.800 --> 00:16:15.326 particularly since it can be helpful

NOTE Confidence: 0.869235684

00:16:15.326 --> 00:16:17.660 in their own treatment options.

NOTE Confidence: 0.869235684

00:16:17.660 --> 00:16:19.280 Same thing with pancreatic cancer.

NOTE Confidence: 0.869235684

00:16:19.280 --> 00:16:21.710 This is a relatively new development

NOTE Confidence: 0.869235684

00:16:21.710 --> 00:16:22.925 since about 2019.

NOTE Confidence: 0.869235684

00:16:22.930 --> 00:16:25.731 I guess it's not so recent, but all things.

NOTE Confidence: 0.869235684

00:16:25.731 --> 00:16:27.873 But when I started in order

NOTE Confidence: 0.869235684

00:16:27.873 --> 00:16:30.137 for pancreatic cancer to be,

NOTE Confidence: 0.869235684

00:16:30.140 --> 00:16:31.265 someone meet criteria,

NOTE Confidence: 0.869235684

00:16:31.265 --> 00:16:33.515 they will need additional family history,
NOTE Confidence: 0.869235684

00:16:33.520 --> 00:16:35.566 which is now not the case.
NOTE Confidence: 0.869235684

00:16:35.570 --> 00:16:38.366 Pancreatic cancer based on newer literature,
NOTE Confidence: 0.869235684

00:16:38.370 --> 00:16:40.520 kind of similar to metastatic
NOTE Confidence: 0.869235684

00:16:40.520 --> 00:16:41.380 prostate cancer,
NOTE Confidence: 0.869235684

00:16:41.380 --> 00:16:43.739 is more likely to have a germline
NOTE Confidence: 0.869235684

00:16:43.739 --> 00:16:45.789 mutation than more common cancers,
NOTE Confidence: 0.869235684

00:16:45.790 --> 00:16:47.092 so given that.
NOTE Confidence: 0.869235684

00:16:47.092 --> 00:16:47.960 If so,
NOTE Confidence: 0.869235684

00:16:47.960 --> 00:16:50.210 given that that's why pancreatic
NOTE Confidence: 0.869235684

00:16:50.210 --> 00:16:53.042 cancer at any age is clinically
NOTE Confidence: 0.869235684

00:16:53.042 --> 00:16:55.106 indicated for genetic testing.
NOTE Confidence: 0.869235684

00:16:55.110 --> 00:16:59.526 Also, because there is treatment options.
NOTE Confidence: 0.869235684

00:16:59.530 --> 00:17:00.282 And also,
NOTE Confidence: 0.869235684

00:17:00.282 --> 00:17:02.914 if there's three cases of the same
NOTE Confidence: 0.869235684

00:17:02.914 --> 00:17:04.399 cancer or or associated cancers

NOTE Confidence: 0.869235684

00:17:04.399 --> 00:17:06.450 on the same side of the family,

NOTE Confidence: 0.869235684

00:17:06.450 --> 00:17:09.047 so someone who has breast cancer and

NOTE Confidence: 0.869235684

00:17:09.047 --> 00:17:11.519 two relatives with breast cancer or

NOTE Confidence: 0.869235684

00:17:11.519 --> 00:17:13.669 someone with breast breast cancer,

NOTE Confidence: 0.869235684

00:17:13.670 --> 00:17:16.190 some cousin with ovarian cancer,

NOTE Confidence: 0.869235684

00:17:16.190 --> 00:17:16.520 someone,

NOTE Confidence: 0.869235684

00:17:16.520 --> 00:17:18.500 a man with prostate cancer and

NOTE Confidence: 0.869235684

00:17:18.500 --> 00:17:20.390 his mother with breast cancer.

NOTE Confidence: 0.869235684

00:17:20.390 --> 00:17:23.374 So those cancers all in the family is.

NOTE Confidence: 0.869235684

00:17:23.380 --> 00:17:25.276 If you were seeing that especially

NOTE Confidence: 0.869235684

00:17:25.276 --> 00:17:25.908 multiple generations,

NOTE Confidence: 0.869235684

00:17:25.910 --> 00:17:30.140 multiple relatives is definitely worth over.

NOTE Confidence: 0.869235684

00:17:30.140 --> 00:17:32.020 Referral.

NOTE Confidence: 0.869235684

00:17:32.020 --> 00:17:32.842 And finally,

NOTE Confidence: 0.869235684

00:17:32.842 --> 00:17:35.308 someone who has multiple primary cancers,

NOTE Confidence: 0.869235684

00:17:35.310 --> 00:17:37.921 such as someone who has breast and
NOTE Confidence: 0.869235684

00:17:37.921 --> 00:17:40.800 ovarian cancer and bilateral breast cancer,
NOTE Confidence: 0.869235684

00:17:40.800 --> 00:17:43.010 may be worth the referral.
NOTE Confidence: 0.869235684

00:17:43.010 --> 00:17:44.975 Bilateral breast cancer if there's
NOTE Confidence: 0.869235684

00:17:44.975 --> 00:17:47.342 no other family history and with
NOTE Confidence: 0.869235684

00:17:47.342 --> 00:17:49.982 depending on the age and may not meet
NOTE Confidence: 0.869235684

00:17:49.982 --> 00:17:52.218 insurance criteria for genetic testing,
NOTE Confidence: 0.869235684

00:17:52.220 --> 00:17:54.650 but it is worth a referral just for us
NOTE Confidence: 0.869235684

00:17:54.650 --> 00:17:57.117 to fully flush out the family history
NOTE Confidence: 0.869235684

00:17:57.117 --> 00:17:59.729 and talk about that with the patient.
NOTE Confidence: 0.9104423475

00:18:01.820 --> 00:18:04.836 So this is similar to the previous slide,
NOTE Confidence: 0.9104423475

00:18:04.840 --> 00:18:06.712 but for just those patients with
NOTE Confidence: 0.9104423475

00:18:06.712 --> 00:18:08.600 only a family history of cancer.
NOTE Confidence: 0.9104423475

00:18:08.600 --> 00:18:10.785 Something to consider is someone
NOTE Confidence: 0.9104423475

00:18:10.785 --> 00:18:12.970 who is Ashkenazi Jewish ancestry,
NOTE Confidence: 0.9104423475

00:18:12.970 --> 00:18:15.892 one in 40 individuals of Ashkenazi

NOTE Confidence: 0.9104423475
00:18:15.892 --> 00:18:18.290 Jewish ancestry will have a
NOTE Confidence: 0.9104423475
00:18:18.290 --> 00:18:20.450 BRCA one and BRCA 2 mutation.
NOTE Confidence: 0.9104423475
00:18:20.450 --> 00:18:22.910 There are three founder mutations
NOTE Confidence: 0.9104423475
00:18:22.910 --> 00:18:25.370 that are seen very commonly
NOTE Confidence: 0.9104423475
00:18:25.451 --> 00:18:27.707 in the Ashkenazi population.
NOTE Confidence: 0.9104423475
00:18:27.710 --> 00:18:29.362 95% of Ashkenazi Jewish
NOTE Confidence: 0.9104423475
00:18:29.362 --> 00:18:31.840 individuals who have a BRCA 1.
NOTE Confidence: 0.9104423475
00:18:31.840 --> 00:18:33.826 Or BRCA 2 mutation will have
NOTE Confidence: 0.9104423475
00:18:33.826 --> 00:18:35.930 one of these three mutations.
NOTE Confidence: 0.9104423475
00:18:35.930 --> 00:18:38.583 So it is more common in the
NOTE Confidence: 0.9104423475
00:18:38.583 --> 00:18:40.490 Ashkenazi population while in the
NOTE Confidence: 0.9104423475
00:18:40.490 --> 00:18:42.240 non Ashkenazy population the risk
NOTE Confidence: 0.9104423475
00:18:42.240 --> 00:18:45.146 of a BRCA mutations about one in
NOTE Confidence: 0.9104423475
00:18:45.146 --> 00:18:47.366 400 so is significantly increased.
NOTE Confidence: 0.9104423475
00:18:47.370 --> 00:18:49.874 What I will say is that without a
NOTE Confidence: 0.9104423475

00:18:49.874 --> 00:18:51.680 family history of breast cancer,
NOTE Confidence: 0.9104423475

00:18:51.680 --> 00:18:53.504 ovarian, prostate cancer,
NOTE Confidence: 0.9104423475

00:18:53.504 --> 00:18:54.720 pancreatic cancer,
NOTE Confidence: 0.9104423475

00:18:54.720 --> 00:18:56.685 the likelihood someone would have
NOTE Confidence: 0.9104423475

00:18:56.685 --> 00:18:58.650 a mutation is still relatively
NOTE Confidence: 0.9104423475

00:18:58.715 --> 00:19:00.479 low and at this point in time
NOTE Confidence: 0.9104423475

00:19:00.479 --> 00:19:02.280 there is no recommendations.
NOTE Confidence: 0.9104423475

00:19:02.280 --> 00:19:04.992 For general screening of the Ashkenazi
NOTE Confidence: 0.9104423475

00:19:04.992 --> 00:19:07.900 Jewish population without a family history,
NOTE Confidence: 0.9104423475

00:19:07.900 --> 00:19:10.112 so it is something I think worth
NOTE Confidence: 0.9104423475

00:19:10.112 --> 00:19:11.860 to consider for a referral,
NOTE Confidence: 0.9104423475

00:19:11.860 --> 00:19:13.804 because at least then we can talk about
NOTE Confidence: 0.9104423475

00:19:13.804 --> 00:19:15.865 with the patient and we can go through,
NOTE Confidence: 0.9104423475

00:19:15.870 --> 00:19:17.220 go through the family history.
NOTE Confidence: 0.9104423475

00:19:17.220 --> 00:19:20.916 But I do want to make that caviar.
NOTE Confidence: 0.9104423475

00:19:20.920 --> 00:19:22.376 Kind of similar to the before slide,

NOTE Confidence: 0.9104423475

00:19:22.380 --> 00:19:25.117 if someone has at least three breast

NOTE Confidence: 0.9104423475

00:19:25.117 --> 00:19:27.679 cancers in the family and their,

NOTE Confidence: 0.9104423475

00:19:27.680 --> 00:19:29.129 you know in their mother and their

NOTE Confidence: 0.9104423475

00:19:29.129 --> 00:19:30.552 aunt and a cousin again clustered

NOTE Confidence: 0.9104423475

00:19:30.552 --> 00:19:32.295 on the same side of the family,

NOTE Confidence: 0.9104423475

00:19:32.300 --> 00:19:34.510 that was worth the referral.

NOTE Confidence: 0.9104423475

00:19:34.510 --> 00:19:37.078 Someone who has a close relative

NOTE Confidence: 0.9104423475

00:19:37.078 --> 00:19:39.511 with breast cancer diagnosed at 45

NOTE Confidence: 0.9104423475

00:19:39.511 --> 00:19:41.455 or younger is worth a referral.

NOTE Confidence: 0.9104423475

00:19:41.460 --> 00:19:44.028 And someone who has a relative

NOTE Confidence: 0.9104423475

00:19:44.028 --> 00:19:45.312 with ovarian cancer.

NOTE Confidence: 0.9104423475

00:19:45.320 --> 00:19:47.784 Someone if someone reports out there on

NOTE Confidence: 0.9104423475

00:19:47.784 --> 00:19:50.196 how to ovarian cancer that individual

NOTE Confidence: 0.9104423475

00:19:50.196 --> 00:19:53.206 does meet criteria based on family history.

NOTE Confidence: 0.9104423475

00:19:53.210 --> 00:19:54.968 Same thing with male breast cancer.

NOTE Confidence: 0.9104423475

00:19:54.970 --> 00:19:57.644 Having an uncle or father with male
NOTE Confidence: 0.9104423475

00:19:57.644 --> 00:20:00.630 breast cancer is a referral is indicated.
NOTE Confidence: 0.9104423475

00:20:00.630 --> 00:20:03.726 Pancreatic cancer in a close relative,
NOTE Confidence: 0.9104423475

00:20:03.730 --> 00:20:05.554 particularly if specifically
NOTE Confidence: 0.9104423475

00:20:05.554 --> 00:20:07.378 if it's apparent.
NOTE Confidence: 0.9104423475

00:20:07.380 --> 00:20:09.492 For a child or a sibling that is
NOTE Confidence: 0.9104423475

00:20:09.492 --> 00:20:11.946 worth a referral and same thing
NOTE Confidence: 0.9104423475

00:20:11.946 --> 00:20:13.798 with metastatic prostate cancer.
NOTE Confidence: 0.9104423475

00:20:13.800 --> 00:20:17.010 This in a close male relative.
NOTE Confidence: 0.9104423475

00:20:17.010 --> 00:20:19.618 And finally, as before.
NOTE Confidence: 0.9104423475

00:20:19.620 --> 00:20:21.312 Leave any close relatives with a
NOTE Confidence: 0.9104423475

00:20:21.312 --> 00:20:23.139 combination of the above cancer types.
NOTE Confidence: 0.9104423475

00:20:23.140 --> 00:20:25.796 Again, if you're seeing in the same family,
NOTE Confidence: 0.9104423475

00:20:25.800 --> 00:20:27.354 prostate, ovarian, breast,
NOTE Confidence: 0.9104423475

00:20:27.354 --> 00:20:29.944 pancreatic, any combination of that,
NOTE Confidence: 0.9104423475

00:20:29.950 --> 00:20:31.498 that is definitely worth a referral.

NOTE Confidence: 0.9104423475

00:20:31.500 --> 00:20:34.176 'cause cancers all can be associated

NOTE Confidence: 0.9104423475

00:20:34.176 --> 00:20:36.730 through a single gene mutation.

NOTE Confidence: 0.9104423475

00:20:36.730 --> 00:20:38.190 Now switching gears and going

NOTE Confidence: 0.9104423475

00:20:38.190 --> 00:20:40.110 to her age right colon cancer.

NOTE Confidence: 0.9104423475

00:20:40.110 --> 00:20:42.399 Kind of like with hereditary breast cancer.

NOTE Confidence: 0.9104423475

00:20:42.400 --> 00:20:44.824 We cannot talk about her age rate colon

NOTE Confidence: 0.9104423475

00:20:44.824 --> 00:20:47.118 cancer without talking about Lynch syndrome.

NOTE Confidence: 0.9104423475

00:20:47.120 --> 00:20:49.316 Lynch syndrome is caused by germline

NOTE Confidence: 0.9104423475

00:20:49.316 --> 00:20:51.439 mutations in one of the five,

NOTE Confidence: 0.9104423475

00:20:51.440 --> 00:20:54.304 so we one of the four mismatch repair

NOTE Confidence: 0.9104423475

00:20:54.304 --> 00:20:57.150 proteins or deletions in the Epcam gene.

NOTE Confidence: 0.9104423475

00:20:57.150 --> 00:21:00.046 The risk is very dependent upon the gene,

NOTE Confidence: 0.9104423475

00:21:00.050 --> 00:21:01.868 especially in the last few years.

NOTE Confidence: 0.9104423475

00:21:01.870 --> 00:21:04.420 We've actually realized that the lifetime

NOTE Confidence: 0.9104423475

00:21:04.420 --> 00:21:06.790 risks with specifically with mutations.

NOTE Confidence: 0.9104423475

00:21:06.790 --> 00:21:07.422 And TM,
NOTE Confidence: 0.9104423475

00:21:07.422 --> 00:21:09.634 S2 and MSH six are quite different
NOTE Confidence: 0.9104423475

00:21:09.634 --> 00:21:12.107 than the risks associated with them.
NOTE Confidence: 0.9104423475

00:21:12.110 --> 00:21:13.246 Which one and MSH.
NOTE Confidence: 0.9104423475

00:21:13.246 --> 00:21:14.950 2 and that could actually be
NOTE Confidence: 0.749313972941176

00:21:15.013 --> 00:21:17.127 a whole whole talking in of itself.
NOTE Confidence: 0.749313972941176

00:21:17.130 --> 00:21:19.065 So I'm not going to go into all the
NOTE Confidence: 0.749313972941176

00:21:19.065 --> 00:21:20.578 details of how they're different,
NOTE Confidence: 0.749313972941176

00:21:20.580 --> 00:21:24.492 but essentially the main cancer risks
NOTE Confidence: 0.749313972941176

00:21:24.492 --> 00:21:27.560 with Lynch syndrome is colorectal cancer
NOTE Confidence: 0.749313972941176

00:21:27.560 --> 00:21:29.970 and uterine or endometrial cancer.
NOTE Confidence: 0.749313972941176

00:21:29.970 --> 00:21:31.496 As you can see in this chart,
NOTE Confidence: 0.749313972941176

00:21:31.500 --> 00:21:33.628 which is mainly based on the high,
NOTE Confidence: 0.749313972941176

00:21:33.630 --> 00:21:35.688 I would say the more high
NOTE Confidence: 0.749313972941176

00:21:35.688 --> 00:21:37.519 penetrance genes Emily Quan and H.
NOTE Confidence: 0.749313972941176

00:21:37.520 --> 00:21:39.686 Two, but there are risk of

NOTE Confidence: 0.749313972941176
00:21:39.686 --> 00:21:41.130 other types of cancers,
NOTE Confidence: 0.749313972941176
00:21:41.130 --> 00:21:43.125 including other GI cancers like
NOTE Confidence: 0.749313972941176
00:21:43.125 --> 00:21:45.120 cancer of the stomach file,
NOTE Confidence: 0.749313972941176
00:21:45.120 --> 00:21:47.520 duct cancer of the pancreas.
NOTE Confidence: 0.749313972941176
00:21:47.520 --> 00:21:49.410 You can also see ovarian cancer
NOTE Confidence: 0.749313972941176
00:21:49.410 --> 00:21:51.619 and women who have Lynch syndrome
NOTE Confidence: 0.749313972941176
00:21:51.620 --> 00:21:53.540 and cancer of the urinary tract,
NOTE Confidence: 0.749313972941176
00:21:53.540 --> 00:21:55.910 like the bladder and also individuals
NOTE Confidence: 0.749313972941176
00:21:55.910 --> 00:21:58.343 with Lynch syndrome can have sebaceous
NOTE Confidence: 0.749313972941176
00:21:58.343 --> 00:22:00.313 adenomas which are very specific
NOTE Confidence: 0.749313972941176
00:22:00.313 --> 00:22:02.670 types of skin of skin findings.
NOTE Confidence: 0.79931447
00:22:05.060 --> 00:22:06.860 So when we think about hereditary
NOTE Confidence: 0.79931447
00:22:06.860 --> 00:22:09.195 colon cancer, I often think about in
NOTE Confidence: 0.79931447
00:22:09.195 --> 00:22:11.964 these two buckets where one bucket is
NOTE Confidence: 0.79931447
00:22:11.964 --> 00:22:14.119 what we call nonpolyposis syndromes,
NOTE Confidence: 0.79931447

00:22:14.120 --> 00:22:16.856 which essentially is mainly Lynch syndrome,

NOTE Confidence: 0.79931447

00:22:16.860 --> 00:22:18.468 maybe other hereditary

NOTE Confidence: 0.79931447

00:22:18.468 --> 00:22:20.076 colon cancer syndromes.

NOTE Confidence: 0.79931447

00:22:20.080 --> 00:22:23.168 This just means that someone is at a

NOTE Confidence: 0.79931447

00:22:23.168 --> 00:22:25.924 high risk genetically of colon cancer,

NOTE Confidence: 0.79931447

00:22:25.924 --> 00:22:28.900 but it's not necessarily because they're

NOTE Confidence: 0.79931447

00:22:28.972 --> 00:22:31.558 developing a large number of polyps.

NOTE Confidence: 0.79931447

00:22:31.560 --> 00:22:33.275 The polyps and Lynch syndrome

NOTE Confidence: 0.79931447

00:22:33.275 --> 00:22:34.990 are not numerous like with.

NOTE Confidence: 0.79931447

00:22:34.990 --> 00:22:35.786 Polyp process,

NOTE Confidence: 0.79931447

00:22:35.786 --> 00:22:38.174 but the polyps in Lynch syndrome

NOTE Confidence: 0.79931447

00:22:38.174 --> 00:22:40.903 do advance much faster than non

NOTE Confidence: 0.79931447

00:22:40.903 --> 00:22:42.759 lynch syndrome associated polyps.

NOTE Confidence: 0.79931447

00:22:42.760 --> 00:22:44.805 So nonpolyposis syndromes you would

NOTE Confidence: 0.79931447

00:22:44.805 --> 00:22:47.983 see a high risk of colon cancer but

NOTE Confidence: 0.79931447

00:22:47.983 --> 00:22:50.834 not the high risk of numerous polyps.

NOTE Confidence: 0.79931447

00:22:50.834 --> 00:22:53.569 Poly Post syndromes are different

NOTE Confidence: 0.79931447

00:22:53.569 --> 00:22:56.335 because an individual would be

NOTE Confidence: 0.79931447

00:22:56.335 --> 00:22:58.431 genetically predisposed to developing

NOTE Confidence: 0.79931447

00:22:58.431 --> 00:23:01.290 large number of column of colon.

NOTE Confidence: 0.79931447

00:23:01.290 --> 00:23:01.996 Polyps depend.

NOTE Confidence: 0.79931447

00:23:01.996 --> 00:23:04.467 The type is dependent on the the

NOTE Confidence: 0.79931447

00:23:04.467 --> 00:23:06.129 polyposis syndrome and I will.

NOTE Confidence: 0.79931447

00:23:06.130 --> 00:23:08.447 Review those briefly on the next slide,

NOTE Confidence: 0.79931447

00:23:08.450 --> 00:23:10.410 but typically they polyps tend

NOTE Confidence: 0.79931447

00:23:10.410 --> 00:23:12.766 to be numerous, usually over 10,

NOTE Confidence: 0.79931447

00:23:12.766 --> 00:23:14.726 and that numerous those numerous

NOTE Confidence: 0.79931447

00:23:14.726 --> 00:23:16.822 polyps or what's causing the

NOTE Confidence: 0.79931447

00:23:16.822 --> 00:23:18.897 increased risk of colon cancer.

NOTE Confidence: 0.581294212

00:23:21.170 --> 00:23:23.000 So hurry, Polly pulses syndrome.

NOTE Confidence: 0.581294212

00:23:23.000 --> 00:23:25.712 Most people have heard about familial

NOTE Confidence: 0.581294212

00:23:25.712 --> 00:23:28.039 adenomatous polyp ossis or FAP
NOTE Confidence: 0.581294212

00:23:28.039 --> 00:23:30.264 which is characterized by someone
NOTE Confidence: 0.581294212

00:23:30.264 --> 00:23:32.470 developing in the classic form,
NOTE Confidence: 0.581294212

00:23:32.470 --> 00:23:33.610 developing hundreds,
NOTE Confidence: 0.581294212

00:23:33.610 --> 00:23:37.030 possibly up to 1000 adenomatous colon
NOTE Confidence: 0.581294212

00:23:37.030 --> 00:23:39.769 polyps beginning at a very young age,
NOTE Confidence: 0.581294212

00:23:39.770 --> 00:23:42.326 usually in the 20s and 20s.
NOTE Confidence: 0.581294212

00:23:42.330 --> 00:23:44.844 Eat or even younger and for
NOTE Confidence: 0.581294212

00:23:44.844 --> 00:23:46.520 the majority of individuals.
NOTE Confidence: 0.581294212

00:23:46.520 --> 00:23:48.970 If this was left untreated,
NOTE Confidence: 0.581294212

00:23:48.970 --> 00:23:51.580 it's with with a total colectomy.
NOTE Confidence: 0.581294212

00:23:51.580 --> 00:23:52.633 There would, uh,
NOTE Confidence: 0.581294212

00:23:52.633 --> 00:23:54.388 the risk of colon cancer
NOTE Confidence: 0.581294212

00:23:54.388 --> 00:23:56.740 approaches 100% even by age 40,
NOTE Confidence: 0.581294212

00:23:56.740 --> 00:23:58.765 so this is a significant
NOTE Confidence: 0.581294212

00:23:58.765 --> 00:23:59.980 Poly pulses syndrome,

NOTE Confidence: 0.581294212

00:23:59.980 --> 00:24:02.578 attenuated as as the name implies,

NOTE Confidence: 0.581294212

00:24:02.580 --> 00:24:03.298 is attenuated,

NOTE Confidence: 0.581294212

00:24:03.298 --> 00:24:05.093 meaning that the polyps develop

NOTE Confidence: 0.581294212

00:24:05.093 --> 00:24:07.443 at a later age and there can

NOTE Confidence: 0.581294212

00:24:07.443 --> 00:24:09.255 be a smaller number of polyps,

NOTE Confidence: 0.581294212

00:24:09.260 --> 00:24:12.344 such usually over usually over 12

NOTE Confidence: 0.581294212

00:24:12.344 --> 00:24:15.770 / 20 up to 100 adenomatous polyps,

NOTE Confidence: 0.581294212

00:24:15.770 --> 00:24:18.871 usually beginning in the 30s or 40s

NOTE Confidence: 0.581294212

00:24:18.871 --> 00:24:21.868 fifties and we have seen a variety of.

NOTE Confidence: 0.581294212

00:24:21.870 --> 00:24:23.610 Even very attenuated forms

NOTE Confidence: 0.581294212

00:24:23.610 --> 00:24:25.785 even in our own clinic.

NOTE Confidence: 0.581294212

00:24:25.790 --> 00:24:28.492 So there does appear to be some

NOTE Confidence: 0.581294212

00:24:28.492 --> 00:24:31.020 variability we're discovering map can

NOTE Confidence: 0.581294212

00:24:31.020 --> 00:24:34.445 sometimes mimic attenuated faps UM,

NOTE Confidence: 0.581294212

00:24:34.450 --> 00:24:36.375 but the only difference with that is

NOTE Confidence: 0.581294212

00:24:36.375 --> 00:24:38.785 that the all the cancer syndromes I've
NOTE Confidence: 0.581294212

00:24:38.785 --> 00:24:40.690 talked about or autosomal dominant,
NOTE Confidence: 0.581294212

00:24:40.690 --> 00:24:43.854 meaning someone would need to inherit a
NOTE Confidence: 0.581294212

00:24:43.854 --> 00:24:46.344 single pathogenic variant in one copy
NOTE Confidence: 0.581294212

00:24:46.344 --> 00:24:48.822 of the gene map is autosomal recessive.
NOTE Confidence: 0.581294212

00:24:48.830 --> 00:24:50.618 So for someone to have map,
NOTE Confidence: 0.581294212

00:24:50.620 --> 00:24:52.020 they would need their parents,
NOTE Confidence: 0.581294212

00:24:52.020 --> 00:24:53.670 we need to be carriers.
NOTE Confidence: 0.581294212

00:24:53.670 --> 00:24:54.248 In math,
NOTE Confidence: 0.581294212

00:24:54.248 --> 00:24:56.560 is can present in a similar way too,
NOTE Confidence: 0.581294212

00:24:56.560 --> 00:25:00.050 attenuated faps with again over,
NOTE Confidence: 0.581294212

00:25:00.050 --> 00:25:02.969 usually over a dozen polyps up to
NOTE Confidence: 0.581294212

00:25:02.969 --> 00:25:05.262 100 polyps beginning in someones 40s
NOTE Confidence: 0.581294212

00:25:05.262 --> 00:25:07.766 or or 50s Paula Mac and sometimes
NOTE Confidence: 0.581294212

00:25:07.766 --> 00:25:10.070 present with a more mixed Poly
NOTE Confidence: 0.581294212

00:25:10.150 --> 00:25:12.500 poesis type where there's adenomas

NOTE Confidence: 0.581294212

00:25:12.500 --> 00:25:14.850 but also non adenomatous polyps

NOTE Confidence: 0.581294212

00:25:14.924 --> 00:25:17.489 like hyperplastic polyps and others.

NOTE Confidence: 0.581294212

00:25:17.490 --> 00:25:20.040 Play Seeger syndrome is a is

NOTE Confidence: 0.581294212

00:25:20.040 --> 00:25:22.810 A is a rare syndrome.

NOTE Confidence: 0.581294212

00:25:22.810 --> 00:25:24.386 It presents particularly with

NOTE Confidence: 0.581294212

00:25:24.386 --> 00:25:26.750 Poly pulses of the small bowel.

NOTE Confidence: 0.581294212

00:25:26.750 --> 00:25:28.798 You could also see it in the colon,

NOTE Confidence: 0.581294212

00:25:28.800 --> 00:25:30.426 but the small bowel is the

NOTE Confidence: 0.581294212

00:25:30.426 --> 00:25:31.510 part that's mostly affected.

NOTE Confidence: 0.581294212

00:25:31.510 --> 00:25:34.240 They develop specific pretty agurs polyps.

NOTE Confidence: 0.581294212

00:25:34.240 --> 00:25:36.710 There's other features such as

NOTE Confidence: 0.581294212

00:25:36.710 --> 00:25:38.192 Miko cutaneous pigmentation

NOTE Confidence: 0.581294212

00:25:38.192 --> 00:25:40.440 that's present in childhood around

NOTE Confidence: 0.581294212

00:25:40.440 --> 00:25:42.425 the lips on the fingertips,

NOTE Confidence: 0.581294212

00:25:42.430 --> 00:25:43.982 even on the nostrils,

NOTE Confidence: 0.581294212

00:25:43.982 --> 00:25:46.310 and there isn't also associated risk
NOTE Confidence: 0.581294212

00:25:46.385 --> 00:25:48.995 of breast cancer and pancreatic cancer.
NOTE Confidence: 0.581294212

00:25:49.000 --> 00:25:51.205 Juvenile polyp Ossis syndrome is
NOTE Confidence: 0.581294212

00:25:51.205 --> 00:25:54.852 not related to the age of onset of
NOTE Confidence: 0.581294212

00:25:54.852 --> 00:25:57.576 polyps because is not necessarily mean.
NOTE Confidence: 0.581294212

00:25:57.580 --> 00:25:59.152 Someone develops polyps as
NOTE Confidence: 0.581294212

00:25:59.152 --> 00:26:01.117 a juvenile or a child,
NOTE Confidence: 0.581294212

00:26:01.120 --> 00:26:03.385 but the these individuals develop
NOTE Confidence: 0.581294212

00:26:03.385 --> 00:26:05.650 specific types of juvenile polyps
NOTE Confidence: 0.581294212

00:26:05.724 --> 00:26:07.619 and juvenile polyps are not.
NOTE Confidence: 0.581294212

00:26:07.620 --> 00:26:10.180 They're not common types of polyps to have,
NOTE Confidence: 0.581294212

00:26:10.180 --> 00:26:13.726 so someone who presents with multiple
NOTE Confidence: 0.581294212

00:26:13.726 --> 00:26:16.090 juvenile polyps would warrant
NOTE Confidence: 0.581294212

00:26:16.182 --> 00:26:18.726 investigation or juvenile polipo SIS.
NOTE Confidence: 0.581294212

00:26:18.726 --> 00:26:20.820 And then there's other more newly
NOTE Confidence: 0.581294212

00:26:20.886 --> 00:26:22.910 described Poly closest Jeanswear.

NOTE Confidence: 0.581294212

00:26:22.910 --> 00:26:24.404 The risk there does appear to

NOTE Confidence: 0.581294212

00:26:24.404 --> 00:26:26.290 be some risk of of Poly poesis,

NOTE Confidence: 0.581294212

00:26:26.290 --> 00:26:31.514 but how that presents exactly is less clear.

NOTE Confidence: 0.581294212

00:26:31.520 --> 00:26:32.970 So since there are those,

NOTE Confidence: 0.581294212

00:26:32.970 --> 00:26:33.861 those two buckets,

NOTE Confidence: 0.581294212

00:26:33.861 --> 00:26:36.363 so let's talk about some of the the

NOTE Confidence: 0.581294212

00:26:36.363 --> 00:26:38.148 guidelines for the referral guidelines

NOTE Confidence: 0.581294212

00:26:38.148 --> 00:26:40.410 for her age right colon cancer.

NOTE Confidence: 0.581294212

00:26:40.410 --> 00:26:42.972 So Lynch syndrome and the guidelines

NOTE Confidence: 0.581294212

00:26:42.972 --> 00:26:46.558 on NCCN are a little bit more clear

NOTE Confidence: 0.581294212

00:26:46.560 --> 00:26:50.496 in terms of when to when to refer.

NOTE Confidence: 0.922782083181818

00:26:50.500 --> 00:26:52.556 I'm not going to go through all of

NOTE Confidence: 0.922782083181818

00:26:52.556 --> 00:26:54.602 this because I know it's a lot and

NOTE Confidence: 0.922782083181818

00:26:54.602 --> 00:26:56.080 I'm gonna again distill it down.

NOTE Confidence: 0.922782083181818

00:26:56.080 --> 00:26:59.384 But essentially it is based in a similar

NOTE Confidence: 0.922782083181818

00:26:59.384 --> 00:27:03.609 way to BRC wanted to based on the age of
NOTE Confidence: 0.922782083181818

00:27:03.609 --> 00:27:07.170 diagnosis and the number of people affected.
NOTE Confidence: 0.922782083181818

00:27:07.170 --> 00:27:08.136 Poly post syndromes.
NOTE Confidence: 0.922782083181818

00:27:08.136 --> 00:27:10.919 The testing strategy can be as you can see,
NOTE Confidence: 0.922782083181818

00:27:10.920 --> 00:27:13.132 branched off a lot so as to
NOTE Confidence: 0.922782083181818

00:27:13.132 --> 00:27:14.929 focus on this part here.
NOTE Confidence: 0.922782083181818

00:27:14.930 --> 00:27:17.145 Probably most important thing to
NOTE Confidence: 0.922782083181818

00:27:17.145 --> 00:27:20.200 think about is the number of polyps.
NOTE Confidence: 0.922782083181818

00:27:20.200 --> 00:27:22.790 It says here over 10 helps me.
NOTE Confidence: 0.922782083181818

00:27:22.790 --> 00:27:24.311 Over 20 adenomas.
NOTE Confidence: 0.922782083181818

00:27:24.311 --> 00:27:26.846 We sometimes are more conservative
NOTE Confidence: 0.922782083181818

00:27:26.846 --> 00:27:30.450 and if we see more than 10 adenomas,
NOTE Confidence: 0.922782083181818

00:27:30.450 --> 00:27:32.334 it's it's worth a referral just
NOTE Confidence: 0.922782083181818

00:27:32.334 --> 00:27:34.690 to look at the family history.
NOTE Confidence: 0.922782083181818

00:27:34.690 --> 00:27:37.960 If someone has exactly 10, you know,
NOTE Confidence: 0.922782083181818

00:27:37.960 --> 00:27:39.985 depending on their their age,

NOTE Confidence: 0.922782083181818

00:27:39.990 --> 00:27:42.150 how many colonoscopies they've had.

NOTE Confidence: 0.922782083181818

00:27:42.150 --> 00:27:43.542 Again, adenomas are common,

NOTE Confidence: 0.922782083181818

00:27:43.542 --> 00:27:45.282 especially if people get older.

NOTE Confidence: 0.922782083181818

00:27:45.290 --> 00:27:47.873 So having a few or a couple adenomas is

NOTE Confidence: 0.922782083181818

00:27:47.873 --> 00:27:50.836 not does not mean someone has Polly Pocest.

NOTE Confidence: 0.922782083181818

00:27:50.840 --> 00:27:52.620 But something to think about.

NOTE Confidence: 0.922782083181818

00:27:52.620 --> 00:27:54.116 Also, as someone's age.

NOTE Confidence: 0.922782083181818

00:27:54.116 --> 00:27:54.490 Also,

NOTE Confidence: 0.922782083181818

00:27:54.490 --> 00:27:57.186 the size of the polop a young person

NOTE Confidence: 0.922782083181818

00:27:57.186 --> 00:27:59.977 with a very large adenomas polyp,

NOTE Confidence: 0.922782083181818

00:27:59.980 --> 00:28:02.339 even with a single a single polyp,

NOTE Confidence: 0.922782083181818

00:28:02.340 --> 00:28:05.735 may still be worth worth the referral.

NOTE Confidence: 0.922782083181818

00:28:05.740 --> 00:28:08.260 But these are these are suggestions,

NOTE Confidence: 0.922782083181818

00:28:08.260 --> 00:28:10.521 so anyone with a colon or uterine

NOTE Confidence: 0.922782083181818

00:28:10.521 --> 00:28:11.892 and demetral cancer diagnosis

NOTE Confidence: 0.922782083181818

00:28:11.892 --> 00:28:14.620 under the age of 50 that would be
NOTE Confidence: 0.922782083181818

00:28:14.620 --> 00:28:16.101 specifically suspicious of Lynch
NOTE Confidence: 0.922782083181818

00:28:16.101 --> 00:28:18.255 syndrome because we expect to see
NOTE Confidence: 0.922782083181818

00:28:18.255 --> 00:28:20.692 those cancers at younger ages.
NOTE Confidence: 0.922782083181818

00:28:20.692 --> 00:28:23.282 Any Lynch syndrome associated cancer,
NOTE Confidence: 0.922782083181818

00:28:23.290 --> 00:28:25.570 that is microsatellite unstable
NOTE Confidence: 0.922782083181818

00:28:25.570 --> 00:28:28.342 or has abnormal MMR staining
NOTE Confidence: 0.922782083181818

00:28:28.342 --> 00:28:29.326 by immunohistochemistry.
NOTE Confidence: 0.922782083181818

00:28:29.326 --> 00:28:34.072 So during it once now here at at a young
NOTE Confidence: 0.922782083181818

00:28:34.072 --> 00:28:36.914 and haven and offered other other places,
NOTE Confidence: 0.922782083181818

00:28:36.920 --> 00:28:40.380 they're now doing universal screening
NOTE Confidence: 0.922782083181818

00:28:40.380 --> 00:28:43.840 of colon and endometrial tumors,
NOTE Confidence: 0.922782083181818

00:28:43.840 --> 00:28:46.090 which is done through immunohistochemistry.
NOTE Confidence: 0.922782083181818

00:28:46.090 --> 00:28:49.393 So staining of of the tumor to look for
NOTE Confidence: 0.922782083181818

00:28:49.393 --> 00:28:52.930 the presence or absence of MMR proteins.
NOTE Confidence: 0.922782083181818

00:28:52.930 --> 00:28:55.348 For individuals who have Lynch syndrome,

NOTE Confidence: 0.922782083181818

00:28:55.350 --> 00:28:58.465 if we would expect that those MMR

NOTE Confidence: 0.922782083181818

00:28:58.465 --> 00:29:00.623 proteins would actually be absent

NOTE Confidence: 0.922782083181818

00:29:00.623 --> 00:29:02.849 on IHC staining and if those

NOTE Confidence: 0.922782083181818

00:29:02.850 --> 00:29:04.806 MMR proteins are absent,

NOTE Confidence: 0.922782083181818

00:29:04.806 --> 00:29:06.762 it actually results in

NOTE Confidence: 0.922782083181818

00:29:06.762 --> 00:29:07.740 microsatellite instability.

NOTE Confidence: 0.922782083181818

00:29:07.740 --> 00:29:09.786 So this is something that is

NOTE Confidence: 0.922782083181818

00:29:09.786 --> 00:29:11.150 on is on pathology.

NOTE Confidence: 0.922782083181818

00:29:11.150 --> 00:29:13.467 It would comment if there was loss

NOTE Confidence: 0.922782083181818

00:29:13.467 --> 00:29:16.132 or presence of these proteins if

NOTE Confidence: 0.922782083181818

00:29:16.132 --> 00:29:18.260 there was microsatellite instability.

NOTE Confidence: 0.922782083181818

00:29:18.260 --> 00:29:20.983 And so the presence of MMR proteins

NOTE Confidence: 0.922782083181818

00:29:20.983 --> 00:29:23.040 would actually be reassuring.

NOTE Confidence: 0.922782083181818

00:29:23.040 --> 00:29:24.140 And it actually would reduce

NOTE Confidence: 0.922782083181818

00:29:24.140 --> 00:29:25.240 the risk of Lynch syndrome,

NOTE Confidence: 0.922782083181818

00:29:25.240 --> 00:29:28.546 but that's why any cancer with
NOTE Confidence: 0.922782083181818

00:29:28.546 --> 00:29:31.760 abnormal IHC missing MMR proteins,
NOTE Confidence: 0.922782083181818

00:29:31.760 --> 00:29:36.440 regardless of age, is is warrants a referral.
NOTE Confidence: 0.922782083181818

00:29:36.440 --> 00:29:38.180 As I mentioned before,
NOTE Confidence: 0.922782083181818

00:29:38.180 --> 00:29:41.272 multiple or you know over over 10
NOTE Confidence: 0.922782083181818

00:29:41.272 --> 00:29:43.760 or early onset gastro GI polyps,
NOTE Confidence: 0.922782083181818

00:29:43.760 --> 00:29:46.490 particularly in the in the colon,
NOTE Confidence: 0.922782083181818

00:29:46.490 --> 00:29:48.320 but also again I mentioned
NOTE Confidence: 0.922782083181818

00:29:48.320 --> 00:29:49.784 with puts Jaeger syndrome.
NOTE Confidence: 0.922782083181818

00:29:49.790 --> 00:29:52.823 If there is or there are polyps of the
NOTE Confidence: 0.922782083181818

00:29:52.823 --> 00:29:57.940 small bowel that can also warrant a referral.
NOTE Confidence: 0.922782083181818

00:29:57.940 --> 00:29:58.780 And specifically,
NOTE Confidence: 0.922782083181818

00:29:58.780 --> 00:30:01.300 I would say over 10 adenoma
NOTE Confidence: 0.922782083181818

00:30:01.300 --> 00:30:04.079 dis or mixed Histology polyps.
NOTE Confidence: 0.922782083181818

00:30:04.080 --> 00:30:06.976 Meaning if someone had a mix of adenomas
NOTE Confidence: 0.922782083181818

00:30:06.976 --> 00:30:09.916 or other polyps like hyperplastic polyps

NOTE Confidence: 0.922782083181818
00:30:09.916 --> 00:30:12.586 that weren't warrant or referral.
NOTE Confidence: 0.922782083181818
00:30:12.590 --> 00:30:14.850 With since Hammertoe medicine juvenile
NOTE Confidence: 0.922782083181818
00:30:14.850 --> 00:30:17.600 polyps are more rare polyp types,
NOTE Confidence: 0.922782083181818
00:30:17.600 --> 00:30:19.812 if we if we see there's essentially
NOTE Confidence: 0.922782083181818
00:30:19.812 --> 00:30:20.760 a lower threshold
NOTE Confidence: 0.85586542875
00:30:20.827 --> 00:30:22.675 for the number of those polyps.
NOTE Confidence: 0.85586542875
00:30:22.680 --> 00:30:25.240 So seeing at least five of those polyps
NOTE Confidence: 0.85586542875
00:30:25.240 --> 00:30:27.696 could warrant a referral and ***** aggers
NOTE Confidence: 0.85586542875
00:30:27.696 --> 00:30:30.370 polyps are a particular type of colon,
NOTE Confidence: 0.85586542875
00:30:30.370 --> 00:30:32.810 polyp or small bowel polyp,
NOTE Confidence: 0.85586542875
00:30:32.810 --> 00:30:35.169 so if that is confirmed on pathology,
NOTE Confidence: 0.85586542875
00:30:35.170 --> 00:30:37.605 at least two it definitely
NOTE Confidence: 0.85586542875
00:30:37.605 --> 00:30:39.553 is warrants or referral.
NOTE Confidence: 0.85586542875
00:30:39.560 --> 00:30:41.310 I'm adding this here for
NOTE Confidence: 0.85586542875
00:30:41.310 --> 00:30:42.360 diffuse gastric cancer.
NOTE Confidence: 0.85586542875

00:30:42.360 --> 00:30:44.376 It's not necessarily related to her edge,
NOTE Confidence: 0.85586542875

00:30:44.380 --> 00:30:45.853 right colon cancer,
NOTE Confidence: 0.85586542875

00:30:45.853 --> 00:30:48.308 but diffuse gastric cancer is
NOTE Confidence: 0.85586542875

00:30:48.308 --> 00:30:51.398 a rare type of gastric cancer,
NOTE Confidence: 0.85586542875

00:30:51.400 --> 00:30:53.056 and it actually can be related.
NOTE Confidence: 0.85586542875

00:30:53.060 --> 00:30:55.280 It is related to win this
NOTE Confidence: 0.85586542875

00:30:55.280 --> 00:30:57.291 hereditary related also to lobular
NOTE Confidence: 0.85586542875

00:30:57.291 --> 00:30:59.546 risk of lobular breast cancer.
NOTE Confidence: 0.85586542875

00:30:59.550 --> 00:31:02.959 So diffuse gastric cancer again is rare.
NOTE Confidence: 0.85586542875

00:31:02.960 --> 00:31:04.718 A specific pathology,
NOTE Confidence: 0.85586542875

00:31:04.718 --> 00:31:07.062 but definitely diagnosis under
NOTE Confidence: 0.85586542875

00:31:07.062 --> 00:31:09.930 50 would warrant or referral.
NOTE Confidence: 0.85586542875

00:31:09.930 --> 00:31:12.114 And similar to BRCA one and two,
NOTE Confidence: 0.85586542875

00:31:12.120 --> 00:31:15.544 if there's three cases of the same cancer
NOTE Confidence: 0.85586542875

00:31:15.544 --> 00:31:19.796 like 3 colon cancers or colon uterine colon,
NOTE Confidence: 0.85586542875

00:31:19.800 --> 00:31:20.316 stomach,

NOTE Confidence: 0.85586542875

00:31:20.316 --> 00:31:21.348 bladder, stomach,

NOTE Confidence: 0.85586542875

00:31:21.348 --> 00:31:23.928 colon all in close relatives

NOTE Confidence: 0.85586542875

00:31:23.928 --> 00:31:25.950 and multiple generations.

NOTE Confidence: 0.85586542875

00:31:25.950 --> 00:31:28.380 You know looking for those associated

NOTE Confidence: 0.85586542875

00:31:28.380 --> 00:31:31.340 cancers that would warrant a referral.

NOTE Confidence: 0.85586542875

00:31:31.340 --> 00:31:34.076 And similar for another for BRCA one and

NOTE Confidence: 0.85586542875

00:31:34.076 --> 00:31:36.510 two multiple cancers in one individual,

NOTE Confidence: 0.85586542875

00:31:36.510 --> 00:31:39.186 such as someone who's had two

NOTE Confidence: 0.85586542875

00:31:39.186 --> 00:31:41.533 colon cancers or someone who's

NOTE Confidence: 0.85586542875

00:31:41.533 --> 00:31:44.365 had colon and any material cancer.

NOTE Confidence: 0.85586542875

00:31:44.370 --> 00:31:46.284 For his patients with with just

NOTE Confidence: 0.85586542875

00:31:46.284 --> 00:31:48.070 a family history of cancer.

NOTE Confidence: 0.85586542875

00:31:48.070 --> 00:31:49.930 Again, as I mentioned before,

NOTE Confidence: 0.85586542875

00:31:49.930 --> 00:31:52.264 someone with at least three colon

NOTE Confidence: 0.85586542875

00:31:52.264 --> 00:31:54.307 cancers or Lynch syndrome associated

NOTE Confidence: 0.85586542875

00:31:54.307 --> 00:31:56.833 cancers in first and second degree
NOTE Confidence: 0.85586542875

00:31:56.833 --> 00:31:58.830 relatives so close relatives.
NOTE Confidence: 0.85586542875

00:31:58.830 --> 00:32:00.797 At least one first of your relative
NOTE Confidence: 0.85586542875

00:32:00.797 --> 00:32:02.346 with colon or endometrial cancer
NOTE Confidence: 0.85586542875

00:32:02.346 --> 00:32:03.946 under the age of 50.
NOTE Confidence: 0.85586542875

00:32:03.950 --> 00:32:06.188 So if someone's parent had color,
NOTE Confidence: 0.85586542875

00:32:06.190 --> 00:32:08.690 sibling had colon cancer under
NOTE Confidence: 0.85586542875

00:32:08.690 --> 00:32:11.550 50 or uterine cancer under 50.
NOTE Confidence: 0.85586542875

00:32:11.550 --> 00:32:13.279 And for second or third of your
NOTE Confidence: 0.85586542875

00:32:13.279 --> 00:32:14.399 relatives with a combination
NOTE Confidence: 0.85586542875

00:32:14.399 --> 00:32:16.029 of the above cancer types,
NOTE Confidence: 0.85586542875

00:32:16.030 --> 00:32:19.300 again looking for those patterns of cancer.
NOTE Confidence: 0.820837004347826

00:32:21.380 --> 00:32:23.636 So I'm I'm. I'm sure everyone here again
NOTE Confidence: 0.820837004347826

00:32:23.636 --> 00:32:26.226 as I talked about with knows about BRCA
NOTE Confidence: 0.820837004347826

00:32:26.226 --> 00:32:28.918 one and two Lynch syndrome Poly Pulsus.
NOTE Confidence: 0.820837004347826

00:32:28.920 --> 00:32:31.035 I want to talk about a few of more

NOTE Confidence: 0.820837004347826
00:32:31.035 --> 00:32:33.473 rare indications that you may not come
NOTE Confidence: 0.820837004347826
00:32:33.473 --> 00:32:35.526 across but just something I wanted
NOTE Confidence: 0.820837004347826
00:32:35.526 --> 00:32:37.500 to bring up as a rare indication.
NOTE Confidence: 0.820837004347826
00:32:37.500 --> 00:32:40.124 So hereditary paraganglioma and
NOTE Confidence: 0.820837004347826
00:32:40.124 --> 00:32:43.404 Pheochromocytoma syndrome is is very
NOTE Confidence: 0.820837004347826
00:32:43.404 --> 00:32:46.577 rare and as as the name implies,
NOTE Confidence: 0.820837004347826
00:32:46.580 --> 00:32:49.080 individuals or increased risk
NOTE Confidence: 0.820837004347826
00:32:49.080 --> 00:32:51.580 to develop rare tumor.
NOTE Confidence: 0.820837004347826
00:32:51.580 --> 00:32:54.244 Here is called Paraganglioma
NOTE Confidence: 0.820837004347826
00:32:54.244 --> 00:32:55.576 and pheochromocytomas,
NOTE Confidence: 0.820837004347826
00:32:55.580 --> 00:32:57.355 so they develop from specific
NOTE Confidence: 0.820837004347826
00:32:57.355 --> 00:32:58.420 nerve nerve cells.
NOTE Confidence: 0.820837004347826
00:32:58.420 --> 00:33:00.365 The the chromaffin cells and
NOTE Confidence: 0.820837004347826
00:33:00.365 --> 00:33:01.921 paragangliomas developed along the
NOTE Confidence: 0.820837004347826
00:33:01.921 --> 00:33:03.740 Para vertebral axis from the base
NOTE Confidence: 0.820837004347826

00:33:03.740 --> 00:33:05.896 of the skull to the pelvis so they
NOTE Confidence: 0.820837004347826

00:33:05.896 --> 00:33:07.842 can show up in the head and neck.
NOTE Confidence: 0.820837004347826

00:33:07.842 --> 00:33:09.676 They can show up in the abdomen.
NOTE Confidence: 0.820837004347826

00:33:09.680 --> 00:33:12.112 Pheochromocytomas are a specific
NOTE Confidence: 0.820837004347826

00:33:12.112 --> 00:33:15.152 type of paraganglioma that is
NOTE Confidence: 0.820837004347826

00:33:15.152 --> 00:33:17.730 confined to the adrenal medula.
NOTE Confidence: 0.820837004347826

00:33:17.730 --> 00:33:20.315 Both of these tumors can
NOTE Confidence: 0.820837004347826

00:33:20.315 --> 00:33:21.866 secrete calcaneus specifically.
NOTE Confidence: 0.820837004347826

00:33:21.870 --> 00:33:24.222 Feos almost always secrete
NOTE Confidence: 0.820837004347826

00:33:24.222 --> 00:33:26.574 these excess of hormones,
NOTE Confidence: 0.820837004347826

00:33:26.580 --> 00:33:29.408 so someone who has a pheochromocytoma can
NOTE Confidence: 0.820837004347826

00:33:29.408 --> 00:33:31.969 actually present with severe hypertension,
NOTE Confidence: 0.820837004347826

00:33:31.970 --> 00:33:33.276 flushing, sweating,
NOTE Confidence: 0.820837004347826

00:33:33.276 --> 00:33:34.582 heart palpitations.
NOTE Confidence: 0.820837004347826

00:33:34.582 --> 00:33:39.230 The majority of these tumors are benign,
NOTE Confidence: 0.820837004347826

00:33:39.230 --> 00:33:42.310 but they some of them can be malignant,

NOTE Confidence: 0.820837004347826
00:33:42.310 --> 00:33:45.750 particularly for some of these
NOTE Confidence: 0.820837004347826
00:33:45.750 --> 00:33:47.126 hereditary predispositions.
NOTE Confidence: 0.820837004347826
00:33:47.130 --> 00:33:48.670 So it's something that that's
NOTE Confidence: 0.820837004347826
00:33:48.670 --> 00:33:50.934 why if some you know a patient
NOTE Confidence: 0.820837004347826
00:33:50.934 --> 00:33:53.046 presents with a sudden neck, neck,
NOTE Confidence: 0.820837004347826
00:33:53.046 --> 00:33:56.230 mass or a mass mass in the abdomen,
NOTE Confidence: 0.820837004347826
00:33:56.230 --> 00:34:00.290 or is complaining of episodes of sweating,
NOTE Confidence: 0.820837004347826
00:34:00.290 --> 00:34:02.677 fatigue, anxiety, that makes me not fatigue,
NOTE Confidence: 0.820837004347826
00:34:02.680 --> 00:34:04.318 anxiety, heart palpitations,
NOTE Confidence: 0.820837004347826
00:34:04.318 --> 00:34:07.048 it's you know it might.
NOTE Confidence: 0.820837004347826
00:34:07.050 --> 00:34:08.170 It's it's, it's something.
NOTE Confidence: 0.820837004347826
00:34:08.170 --> 00:34:10.014 Something to think about, you know,
NOTE Confidence: 0.820837004347826
00:34:10.014 --> 00:34:11.824 maybe a referral to endocrinology.
NOTE Confidence: 0.820837004347826
00:34:11.830 --> 00:34:13.462 Not saying that everyone that presents
NOTE Confidence: 0.820837004347826
00:34:13.462 --> 00:34:15.210 with that will have these tumors,
NOTE Confidence: 0.820837004347826

00:34:15.210 --> 00:34:17.420 'cause they're very rare, but.
NOTE Confidence: 0.820837004347826

00:34:17.420 --> 00:34:19.598 They are something that we do.
NOTE Confidence: 0.820837004347826

00:34:19.600 --> 00:34:20.708 We do screen form.
NOTE Confidence: 0.820837004347826

00:34:20.708 --> 00:34:22.810 We do see patients with these tumors
NOTE Confidence: 0.820837004347826

00:34:22.810 --> 00:34:24.690 because there is also screening.
NOTE Confidence: 0.820837004347826

00:34:24.690 --> 00:34:27.010 For individuals who have a
NOTE Confidence: 0.820837004347826

00:34:27.010 --> 00:34:27.938 hereditary predisposition.
NOTE Confidence: 0.78671463125

00:34:30.200 --> 00:34:34.100 Another, more rare hereditary predisposition
NOTE Confidence: 0.78671463125

00:34:34.100 --> 00:34:36.440 is hereditary hyperparathyroidism,
NOTE Confidence: 0.78671463125

00:34:36.440 --> 00:34:38.659 and I think when people think of
NOTE Confidence: 0.78671463125

00:34:38.659 --> 00:34:40.225 hyperparathyroidism, they may not
NOTE Confidence: 0.78671463125

00:34:40.225 --> 00:34:42.300 initially think of hereditary cancer.
NOTE Confidence: 0.78671463125

00:34:42.300 --> 00:34:44.288 'cause hyperparathyroidism is not
NOTE Confidence: 0.78671463125

00:34:44.288 --> 00:34:46.784 a cancer diagnosis, of course,
NOTE Confidence: 0.78671463125

00:34:46.784 --> 00:34:49.796 is just an overactive parathyroid gland,
NOTE Confidence: 0.78671463125

00:34:49.800 --> 00:34:52.976 but hereditary hyperparathyroidism can

NOTE Confidence: 0.78671463125

00:34:52.976 --> 00:34:56.708 be related to other risks of humors,

NOTE Confidence: 0.78671463125

00:34:56.708 --> 00:34:58.730 so the most common form of

NOTE Confidence: 0.78671463125

00:34:58.798 --> 00:35:00.930 hereditary hyperparathyroidism.

NOTE Confidence: 0.78671463125

00:35:00.930 --> 00:35:03.570 Is multiple endocrine neoplasia type

NOTE Confidence: 0.78671463125

00:35:03.570 --> 00:35:06.865 one or M1 where the individuals with

NOTE Confidence: 0.78671463125

00:35:06.865 --> 00:35:10.658 this with me and one will develop

NOTE Confidence: 0.78671463125

00:35:10.658 --> 00:35:13.165 hyperparathyroidism over 90% of

NOTE Confidence: 0.78671463125

00:35:13.165 --> 00:35:15.605 the time by the time they are 50,

NOTE Confidence: 0.78671463125

00:35:15.610 --> 00:35:18.322 usually when they're in their 20s or 30s

NOTE Confidence: 0.78671463125

00:35:18.322 --> 00:35:21.272 and they are at risk of other endocrine

NOTE Confidence: 0.78671463125

00:35:21.272 --> 00:35:23.630 tumors of of the endocrine system,

NOTE Confidence: 0.78671463125

00:35:23.630 --> 00:35:26.456 such as neuroendocrine tumors of the

NOTE Confidence: 0.78671463125

00:35:26.456 --> 00:35:29.450 pancreas of the stomach of carcinoid.

NOTE Confidence: 0.78671463125

00:35:29.450 --> 00:35:31.306 So it is important.

NOTE Confidence: 0.78671463125

00:35:31.306 --> 00:35:34.090 When someone has particularly early onset

NOTE Confidence: 0.78671463125

00:35:34.175 --> 00:35:37.299 hyperparathyroidism to actually evaluate,
NOTE Confidence: 0.78671463125

00:35:37.300 --> 00:35:40.042 to evaluate if they have possibly
NOTE Confidence: 0.78671463125

00:35:40.042 --> 00:35:43.258 have me in one and possibly some of
NOTE Confidence: 0.78671463125

00:35:43.258 --> 00:35:45.895 these less common forms of fresh
NOTE Confidence: 0.78671463125

00:35:45.895 --> 00:35:47.835 or hyperparathyroidism because it
NOTE Confidence: 0.78671463125

00:35:47.835 --> 00:35:50.513 possibly could mean they might be
NOTE Confidence: 0.78671463125

00:35:50.513 --> 00:35:52.799 at risk of other endocrine tumors.
NOTE Confidence: 0.78671463125

00:35:52.800 --> 00:35:54.738 So those are just two examples,
NOTE Confidence: 0.78671463125

00:35:54.740 --> 00:35:56.546 but just some of the other
NOTE Confidence: 0.78671463125

00:35:56.546 --> 00:35:58.416 indications that you know maybe may
NOTE Confidence: 0.78671463125

00:35:58.416 --> 00:36:00.222 not immediately come to mind when
NOTE Confidence: 0.78671463125

00:36:00.222 --> 00:36:02.280 we think about hereditary cancer.
NOTE Confidence: 0.78671463125

00:36:02.280 --> 00:36:05.500 Some of the more rare indications anyone
NOTE Confidence: 0.78671463125

00:36:05.500 --> 00:36:08.109 with with medullary thyroid cancer,
NOTE Confidence: 0.78671463125

00:36:08.110 --> 00:36:09.474 adrenocortical carcinoma,
NOTE Confidence: 0.78671463125

00:36:09.474 --> 00:36:10.838 paraganglioma, pheochromocytoma,

NOTE Confidence: 0.78671463125

00:36:10.838 --> 00:36:15.831 or red no blastoma at any age warrants

NOTE Confidence: 0.78671463125

00:36:15.831 --> 00:36:19.079 a referral or anyone that has a close

NOTE Confidence: 0.78671463125

00:36:19.165 --> 00:36:22.009 relative with any of these cancers.

NOTE Confidence: 0.78671463125

00:36:22.010 --> 00:36:24.410 Anyone with any again rare or

NOTE Confidence: 0.78671463125

00:36:24.410 --> 00:36:26.495 unusual tumors or physical findings

NOTE Confidence: 0.78671463125

00:36:26.495 --> 00:36:28.937 so there are specific skin findings

NOTE Confidence: 0.78671463125

00:36:28.937 --> 00:36:31.309 that even if they're benign,

NOTE Confidence: 0.78671463125

00:36:31.310 --> 00:36:33.055 they actually can be indicative

NOTE Confidence: 0.78671463125

00:36:33.055 --> 00:36:34.800 of a hereditary cancer syndrome,

NOTE Confidence: 0.78671463125

00:36:34.800 --> 00:36:36.660 such as sebaceous carcinoma or

NOTE Confidence: 0.78671463125

00:36:36.660 --> 00:36:38.932 sebaceous adenomas which we can see

NOTE Confidence: 0.78671463125

00:36:38.932 --> 00:36:40.854 with Lynch syndrome or tricholoma

NOTE Confidence: 0.78671463125

00:36:40.854 --> 00:36:43.948 which we can see with Cowden syndrome,

NOTE Confidence: 0.78671463125

00:36:43.950 --> 00:36:46.250 which is related to the.

NOTE Confidence: 0.78671463125

00:36:46.250 --> 00:36:50.678 The P10 gene. Melanoma is is common.

NOTE Confidence: 0.78671463125

00:36:50.678 --> 00:36:52.838 The biggest risk factor for
NOTE Confidence: 0.78671463125

00:36:52.838 --> 00:36:55.321 Melanoma or any skin cancer is of
NOTE Confidence: 0.78671463125

00:36:55.321 --> 00:36:57.669 course fair skin and sun exposure.
NOTE Confidence: 0.78671463125

00:36:57.670 --> 00:37:00.385 But if someone's had multiple
NOTE Confidence: 0.78671463125

00:37:00.385 --> 00:37:03.100 melanomas usually over over 3,
NOTE Confidence: 0.78671463125

00:37:03.100 --> 00:37:05.200 specially if they're at a younger age,
NOTE Confidence: 0.78671463125

00:37:05.200 --> 00:37:07.630 that would warrant a referral
NOTE Confidence: 0.78671463125

00:37:07.630 --> 00:37:10.758 to cancer genetics to rule out
NOTE Confidence: 0.78671463125

00:37:10.758 --> 00:37:13.190 a hereditary Melanoma syndrome.
NOTE Confidence: 0.78671463125

00:37:13.190 --> 00:37:16.046 Leukemia in children is unfortunately common,
NOTE Confidence: 0.78671463125

00:37:16.050 --> 00:37:18.192 but if there are other rare
NOTE Confidence: 0.78671463125

00:37:18.192 --> 00:37:19.620 childhood tumors that might,
NOTE Confidence: 0.78671463125

00:37:19.620 --> 00:37:21.396 that would be an indication for
NOTE Confidence: 0.78671463125

00:37:21.396 --> 00:37:22.580 a cancer genetics referral,
NOTE Confidence: 0.78671463125

00:37:22.580 --> 00:37:26.846 such as again, a Renault blastoma.
NOTE Confidence: 0.78671463125

00:37:26.850 --> 00:37:28.440 Sarcoma and osteosarcoma.

NOTE Confidence: 0.78671463125

00:37:28.440 --> 00:37:31.620 Some tumors that are diagnosed in

NOTE Confidence: 0.78671463125

00:37:31.620 --> 00:37:35.410 children can be indicative of a

NOTE Confidence: 0.78671463125

00:37:35.410 --> 00:37:38.018 rare hereditary cancer syndrome.

NOTE Confidence: 0.78671463125

00:37:38.020 --> 00:37:40.125 And anyone diagnosed with primary

NOTE Confidence: 0.78671463125

00:37:40.125 --> 00:37:41.809 hyperparathyroidism before the age

NOTE Confidence: 0.78671463125

00:37:41.809 --> 00:37:44.017 of 50 warrants a referral again,

NOTE Confidence: 0.78671463125

00:37:44.020 --> 00:37:47.890 since the most common type of

NOTE Confidence: 0.78671463125

00:37:47.890 --> 00:37:50.982 hyperparathyroidism, me and one.

NOTE Confidence: 0.78671463125

00:37:50.982 --> 00:37:52.610 I'm hypercalcemia,

NOTE Confidence: 0.78671463125

00:37:52.610 --> 00:37:55.270 which often is the first presenting sign

NOTE Confidence: 0.78671463125

00:37:55.270 --> 00:37:57.046 that someone has hyperparathyroidism

NOTE Confidence: 0.78671463125

00:37:57.046 --> 00:37:59.314 because elevated parathyroid hormone

NOTE Confidence: 0.78671463125

00:37:59.314 --> 00:38:02.750 causes an elevation of serum calcium.

NOTE Confidence: 0.78671463125

00:38:02.750 --> 00:38:04.808 The almost all people with me and

NOTE Confidence: 0.78671463125

00:38:04.808 --> 00:38:06.532 one will have hypercalcemia by

NOTE Confidence: 0.78671463125

00:38:06.532 --> 00:38:08.076 the time they're 50,
NOTE Confidence: 0.78671463125

00:38:08.080 --> 00:38:10.105 so if someone has hyperparathyroidism
NOTE Confidence: 0.78671463125

00:38:10.105 --> 00:38:11.320 before they're 50,
NOTE Confidence: 0.78671463125

00:38:11.320 --> 00:38:13.785 that could indicate that you
NOTE Confidence: 0.78671463125

00:38:13.785 --> 00:38:16.640 know it is important to rule
NOTE Confidence: 0.78671463125

00:38:16.640 --> 00:38:18.528 out another previous position.
NOTE Confidence: 0.78671463125

00:38:18.530 --> 00:38:20.540 And I say this for the end because it is
NOTE Confidence: 0.868109920714286

00:38:20.600 --> 00:38:21.708 not a rare indication,
NOTE Confidence: 0.868109920714286

00:38:21.710 --> 00:38:23.966 but anyone that has a fame
NOTE Confidence: 0.868109920714286

00:38:23.966 --> 00:38:25.470 reports a family history,
NOTE Confidence: 0.868109920714286

00:38:25.470 --> 00:38:29.607 but no mutation in any cancer gene
NOTE Confidence: 0.868109920714286

00:38:29.607 --> 00:38:33.187 Jersey one MLH one pal B2 ATM.
NOTE Confidence: 0.868109920714286

00:38:33.187 --> 00:38:37.120 Any cancer gene you know in a cousin,
NOTE Confidence: 0.868109920714286

00:38:37.120 --> 00:38:39.430 even in a first cousin once removed,
NOTE Confidence: 0.868109920714286

00:38:39.430 --> 00:38:41.964 if someone is reporting that that is
NOTE Confidence: 0.868109920714286

00:38:41.964 --> 00:38:43.727 a definite indication for genetic

NOTE Confidence: 0.868109920714286
00:38:43.727 --> 00:38:45.869 testing and it is very important if
NOTE Confidence: 0.868109920714286
00:38:45.869 --> 00:38:48.297 a patient is reporting that to you.
NOTE Confidence: 0.868109920714286
00:38:48.300 --> 00:38:50.268 That you do encourage them to get a
NOTE Confidence: 0.868109920714286
00:38:50.268 --> 00:38:52.479 copy of their relatives test results,
NOTE Confidence: 0.868109920714286
00:38:52.480 --> 00:38:54.586 because that can actually be really
NOTE Confidence: 0.868109920714286
00:38:54.586 --> 00:38:56.933 helpful in their own testing and
NOTE Confidence: 0.868109920714286
00:38:56.933 --> 00:38:58.689 actually ensuring their tests.
NOTE Confidence: 0.868109920714286
00:38:58.690 --> 00:39:02.910 Their own testing is accurate.
NOTE Confidence: 0.868109920714286
00:39:02.910 --> 00:39:05.514 So let's talk about out of pocket
NOTE Confidence: 0.868109920714286
00:39:05.514 --> 00:39:07.100 genetic casting options and
NOTE Confidence: 0.868109920714286
00:39:07.100 --> 00:39:08.716 direct to consumer testing.
NOTE Confidence: 0.868109920714286
00:39:08.720 --> 00:39:11.317 So out of pocket genetic testing options.
NOTE Confidence: 0.868109920714286
00:39:11.320 --> 00:39:13.336 Luckily in the last few years the
NOTE Confidence: 0.868109920714286
00:39:13.336 --> 00:39:15.726 cost of genetic testing has gone down
NOTE Confidence: 0.868109920714286
00:39:15.726 --> 00:39:17.916 significantly in the last several years,
NOTE Confidence: 0.868109920714286

00:39:17.920 --> 00:39:19.906 so years, years and years ago,
NOTE Confidence: 0.868109920714286

00:39:19.910 --> 00:39:21.250 over ten years ago,
NOTE Confidence: 0.868109920714286

00:39:21.250 --> 00:39:23.602 testing may have been a few \$1000
NOTE Confidence: 0.868109920714286

00:39:23.602 --> 00:39:25.996 even just for BRCA one and two.
NOTE Confidence: 0.868109920714286

00:39:26.000 --> 00:39:30.480 Testing now because the technology is faster,
NOTE Confidence: 0.868109920714286

00:39:30.480 --> 00:39:32.580 it's better. It's cheaper now.
NOTE Confidence: 0.868109920714286

00:39:32.580 --> 00:39:34.840 Genetic testing can be performed
NOTE Confidence: 0.868109920714286

00:39:34.840 --> 00:39:37.660 through a large panel of genes.
NOTE Confidence: 0.868109920714286

00:39:37.660 --> 00:39:39.820 Again, all the genes that we've talked about.
NOTE Confidence: 0.868109920714286

00:39:39.820 --> 00:39:42.054 For an out of pocket costs of \$250,
NOTE Confidence: 0.868109920714286

00:39:42.054 --> 00:39:43.874 there are a few commercial
NOTE Confidence: 0.868109920714286

00:39:43.874 --> 00:39:45.730 labs that are offering this,
NOTE Confidence: 0.868109920714286

00:39:45.730 --> 00:39:48.322 which has been really excellent because
NOTE Confidence: 0.868109920714286

00:39:48.322 --> 00:39:51.640 it doesn't prove access to genetic testing
NOTE Confidence: 0.868109920714286

00:39:51.640 --> 00:39:54.170 for patients without insurance coverage.
NOTE Confidence: 0.868109920714286

00:39:54.170 --> 00:39:56.070 For those patients who are

NOTE Confidence: 0.868109920714286
00:39:56.070 --> 00:39:57.970 motivated to pursue genetic testing,
NOTE Confidence: 0.868109920714286
00:39:57.970 --> 00:39:59.944 even if we don't seal are risk
NOTE Confidence: 0.868109920714286
00:39:59.944 --> 00:40:01.280 factors in their family.
NOTE Confidence: 0.868109920714286
00:40:01.280 --> 00:40:03.730 For a hereditary cancer syndrome
NOTE Confidence: 0.868109920714286
00:40:03.730 --> 00:40:06.320 and just one example here in vitae,
NOTE Confidence: 0.868109920714286
00:40:06.320 --> 00:40:08.258 they do actually have their own
NOTE Confidence: 0.868109920714286
00:40:08.258 --> 00:40:09.491 self pay option.
NOTE Confidence: 0.868109920714286
00:40:09.491 --> 00:40:12.977 Where to be \$250 for diagnostic
NOTE Confidence: 0.868109920714286
00:40:12.977 --> 00:40:16.727 testing and this testing is different
NOTE Confidence: 0.868109920714286
00:40:16.727 --> 00:40:20.525 than the direct to consumer testing
NOTE Confidence: 0.868109920714286
00:40:20.525 --> 00:40:22.984 like 23 and me or ancestry.com
NOTE Confidence: 0.868109920714286
00:40:22.984 --> 00:40:25.354 which this test again detained
NOTE Confidence: 0.868109920714286
00:40:25.354 --> 00:40:28.615 other other labs is testing is a
NOTE Confidence: 0.868109920714286
00:40:28.615 --> 00:40:30.760 clinical diagnostic test but other
NOTE Confidence: 0.868109920714286
00:40:30.760 --> 00:40:33.199 direct to consumer testing like 23
NOTE Confidence: 0.868109920714286

00:40:33.199 --> 00:40:35.974 and me and ancestry.com is well how
NOTE Confidence: 0.868109920714286

00:40:35.974 --> 00:40:37.618 I've heard it described.
NOTE Confidence: 0.868109920714286

00:40:37.620 --> 00:40:38.990 It is like cocktail genetics.
NOTE Confidence: 0.868109920714286

00:40:38.990 --> 00:40:40.958 It's it's something fun.
NOTE Confidence: 0.868109920714286

00:40:40.958 --> 00:40:42.994 It's, you know, an ancestry.
NOTE Confidence: 0.868109920714286

00:40:42.994 --> 00:40:44.924 There's nothing indicating that the
NOTE Confidence: 0.868109920714286

00:40:44.924 --> 00:40:46.867 ancestry is actually is inaccurate.
NOTE Confidence: 0.868109920714286

00:40:46.870 --> 00:40:48.446 If anything actually appears
NOTE Confidence: 0.868109920714286

00:40:48.446 --> 00:40:50.416 to be quite quite accurate,
NOTE Confidence: 0.868109920714286

00:40:50.420 --> 00:40:52.820 but it's not something that should
NOTE Confidence: 0.868109920714286

00:40:52.820 --> 00:40:55.660 be used for clinical decision making.
NOTE Confidence: 0.868109920714286

00:40:55.660 --> 00:40:58.308 So just something to.
NOTE Confidence: 0.868109920714286

00:40:58.308 --> 00:41:00.875 To think about so.
NOTE Confidence: 0.868109920714286

00:41:00.875 --> 00:41:03.750 As a as an example,
NOTE Confidence: 0.868109920714286

00:41:03.750 --> 00:41:04.244 you know,
NOTE Confidence: 0.868109920714286

00:41:04.244 --> 00:41:06.690 let's say Diana is a 36 year old female.

NOTE Confidence: 0.868109920714286
00:41:06.690 --> 00:41:07.778 She's in good health.
NOTE Confidence: 0.868109920714286
00:41:07.778 --> 00:41:09.760 She comes in for an annual exam.
NOTE Confidence: 0.868109920714286
00:41:09.760 --> 00:41:12.052 She has Spanish ancestry.
NOTE Confidence: 0.868109920714286
00:41:12.052 --> 00:41:15.280 She's reports her mom had breast
NOTE Confidence: 0.868109920714286
00:41:15.280 --> 00:41:17.080 cancer 44 and passed away,
NOTE Confidence: 0.868109920714286
00:41:17.080 --> 00:41:19.390 but no other family history of cancer.
NOTE Confidence: 0.868109920714286
00:41:19.390 --> 00:41:21.763 And then Diana reports that she sent
NOTE Confidence: 0.868109920714286
00:41:21.763 --> 00:41:24.119 in a sample for genetic testing at
NOTE Confidence: 0.868109920714286
00:41:24.119 --> 00:41:26.823 23 and me and that her results were
NOTE Confidence: 0.868109920714286
00:41:26.823 --> 00:41:29.350 negative for BRCA one and BRCA 2.
NOTE Confidence: 0.868109920714286
00:41:29.350 --> 00:41:32.430 So the question I have and I was going to do.
NOTE Confidence: 0.868109920714286
00:41:32.430 --> 00:41:32.834 Cool,
NOTE Confidence: 0.868109920714286
00:41:32.834 --> 00:41:35.258 but it wasn't working out so
NOTE Confidence: 0.868109920714286
00:41:35.258 --> 00:41:36.800 I'm just going to.
NOTE Confidence: 0.868109920714286
00:41:36.800 --> 00:41:38.116 Close the question for
NOTE Confidence: 0.868109920714286

00:41:38.116 --> 00:41:40.090 you to think about and you
NOTE Confidence: 0.905090555238095

00:41:40.165 --> 00:41:41.645 can answer out loud where
NOTE Confidence: 0.905090555238095

00:41:41.645 --> 00:41:43.530 you are or in your head.
NOTE Confidence: 0.905090555238095

00:41:43.530 --> 00:41:47.758 Are Diane is genetic tests isn't informative.
NOTE Confidence: 0.905090555238095

00:41:47.760 --> 00:41:49.762 And does she need to be retested
NOTE Confidence: 0.905090555238095

00:41:49.762 --> 00:41:51.892 for BRCA one and BRCA 2 if
NOTE Confidence: 0.905090555238095

00:41:51.892 --> 00:41:53.100 her testing is negative?
NOTE Confidence: 0.905090555238095

00:41:53.100 --> 00:41:55.484 And let's say we actually have the report.
NOTE Confidence: 0.905090555238095

00:41:55.490 --> 00:41:58.250 Is confirmed does she need to be retested?
NOTE Confidence: 0.905090555238095

00:41:58.250 --> 00:41:59.279 Is this informative?
NOTE Confidence: 0.85469599

00:42:01.660 --> 00:42:05.750 And the answer is no,
NOTE Confidence: 0.85469599

00:42:05.750 --> 00:42:10.220 and and no so. So so 23andMe?
NOTE Confidence: 0.85469599

00:42:10.220 --> 00:42:12.620 Uhm, it's not to say so.
NOTE Confidence: 0.85469599

00:42:12.620 --> 00:42:15.405 Essentially what is important to
NOTE Confidence: 0.85469599

00:42:15.405 --> 00:42:19.817 know is that 23 NI they do offer
NOTE Confidence: 0.85469599

00:42:19.817 --> 00:42:23.338 testing for BRCA one and BRCA 2.

NOTE Confidence: 0.85469599

00:42:23.340 --> 00:42:26.168 But only selected variants

NOTE Confidence: 0.85469599

00:42:26.168 --> 00:42:28.289 you might remember.

NOTE Confidence: 0.85469599

00:42:28.290 --> 00:42:30.674 I said that earlier in the earlier in

NOTE Confidence: 0.85469599

00:42:30.674 --> 00:42:33.473 the talk I said that there are three

NOTE Confidence: 0.85469599

00:42:33.473 --> 00:42:35.778 specific mutations that are seen commonly

NOTE Confidence: 0.85469599

00:42:35.778 --> 00:42:38.208 in people that are Ashkenazi Jewish.

NOTE Confidence: 0.85469599

00:42:38.210 --> 00:42:40.910 Those are the three variants that.

NOTE Confidence: 0.85469599

00:42:40.910 --> 00:42:43.780 Are being tested for, so for Diana

NOTE Confidence: 0.85469599

00:42:43.780 --> 00:42:47.240 she may not have those variants and.

NOTE Confidence: 0.85469599

00:42:47.240 --> 00:42:51.000 A manned, but she also has Spanish ancestry,

NOTE Confidence: 0.85469599

00:42:51.000 --> 00:42:53.135 so she would not have been expected

NOTE Confidence: 0.85469599

00:42:53.135 --> 00:42:55.110 to have those variants at all,

NOTE Confidence: 0.85469599

00:42:55.110 --> 00:42:57.198 and 23 me does not do full gene

NOTE Confidence: 0.85469599

00:42:57.198 --> 00:42:58.997 sequencing of beers they wanted to.

NOTE Confidence: 0.85469599

00:42:59.000 --> 00:43:01.700 Does not do deletion duplication,

NOTE Confidence: 0.85469599

00:43:01.700 --> 00:43:04.352 and those three mutations are seen
NOTE Confidence: 0.85469599

00:43:04.352 --> 00:43:06.559 almost exclusively in people or
NOTE Confidence: 0.85469599

00:43:06.559 --> 00:43:08.749 Ashkenazi Jewish make up only three
NOTE Confidence: 0.85469599

00:43:08.749 --> 00:43:11.389 of 1000 known pathogenic mutations.
NOTE Confidence: 0.85469599

00:43:11.390 --> 00:43:14.222 So for Diana this is a very uninformative
NOTE Confidence: 0.85469599

00:43:14.222 --> 00:43:17.349 test and she would be recommended to have.
NOTE Confidence: 0.85469599

00:43:17.350 --> 00:43:21.130 Formal BRC wanted two testing gene
NOTE Confidence: 0.85469599

00:43:21.130 --> 00:43:23.250 sequencing and deletion duplication.
NOTE Confidence: 0.85469599

00:43:23.250 --> 00:43:24.298 And that's essentially what
NOTE Confidence: 0.85469599

00:43:24.298 --> 00:43:25.608 direct to consumer testing does.
NOTE Confidence: 0.85469599

00:43:25.610 --> 00:43:27.840 It looks at specific variants,
NOTE Confidence: 0.85469599

00:43:27.840 --> 00:43:30.056 and some of these variants may have a
NOTE Confidence: 0.85469599

00:43:30.056 --> 00:43:32.618 very small effect on health and may have
NOTE Confidence: 0.85469599

00:43:32.618 --> 00:43:34.550 a very questionable effect on health,
NOTE Confidence: 0.85469599

00:43:34.550 --> 00:43:35.417 and it's not.
NOTE Confidence: 0.85469599

00:43:35.417 --> 00:43:37.151 It's not very clear some for

NOTE Confidence: 0.85469599
00:43:37.151 --> 00:43:38.610 some of these variants,
NOTE Confidence: 0.85469599
00:43:38.610 --> 00:43:40.610 for even other diseases,
NOTE Confidence: 0.85469599
00:43:40.610 --> 00:43:43.610 how much risk is actually giving,
NOTE Confidence: 0.85469599
00:43:43.610 --> 00:43:46.321 and if something is not clear exactly how
NOTE Confidence: 0.85469599
00:43:46.321 --> 00:43:48.218 little of the gene that actually looking
NOTE Confidence: 0.85469599
00:43:48.218 --> 00:43:50.439 at so direct to consumer testing is,
NOTE Confidence: 0.85469599
00:43:50.440 --> 00:43:53.347 you know it is something that the the people.
NOTE Confidence: 0.85469599
00:43:53.350 --> 00:43:53.688 Lou,
NOTE Confidence: 0.85469599
00:43:53.688 --> 00:43:56.054 but it's something that should not be
NOTE Confidence: 0.85469599
00:43:56.054 --> 00:43:58.600 used for any clinical determination.
NOTE Confidence: 0.85469599
00:43:58.600 --> 00:44:00.748 What's what some people have been
NOTE Confidence: 0.85469599
00:44:00.748 --> 00:44:03.356 doing is that they can take the raw
NOTE Confidence: 0.85469599
00:44:03.356 --> 00:44:05.518 data from 23 and me and actually
NOTE Confidence: 0.85469599
00:44:05.518 --> 00:44:08.556 upload it to programs like one called
NOTE Confidence: 0.85469599
00:44:08.556 --> 00:44:11.365 promethease that actually builds a
NOTE Confidence: 0.85469599

00:44:11.365 --> 00:44:15.184 personal DNA report by looking at the
NOTE Confidence: 0.85469599

00:44:15.184 --> 00:44:18.010 genotype so they actually they actually
NOTE Confidence: 0.85469599

00:44:18.095 --> 00:44:20.735 can do that at not a large cost,
NOTE Confidence: 0.85469599

00:44:20.740 --> 00:44:23.288 but the problem with that is actually
NOTE Confidence: 0.85469599

00:44:23.288 --> 00:44:25.149 uploading these raw data files.
NOTE Confidence: 0.85469599

00:44:25.150 --> 00:44:28.594 There's actually a high false positive rate.
NOTE Confidence: 0.85469599

00:44:28.600 --> 00:44:31.096 Because just in terms of of how the
NOTE Confidence: 0.85469599

00:44:31.096 --> 00:44:33.571 data is uploaded and how the programs
NOTE Confidence: 0.85469599

00:44:33.571 --> 00:44:36.269 are there is it can actually report
NOTE Confidence: 0.85469599

00:44:36.269 --> 00:44:38.795 a pathogenic mutation that if we
NOTE Confidence: 0.85469599

00:44:38.795 --> 00:44:41.473 look test the patient clinically
NOTE Confidence: 0.85469599

00:44:41.473 --> 00:44:43.917 limitation is not there.
NOTE Confidence: 0.85469599

00:44:43.920 --> 00:44:47.764 So this is so this is is 11
NOTE Confidence: 0.85469599

00:44:47.764 --> 00:44:50.608 figure from from this test which
NOTE Confidence: 0.85469599

00:44:50.608 --> 00:44:52.709 actually showed that out of.
NOTE Confidence: 0.85469599

00:44:52.710 --> 00:44:55.494 Out of the the number of patients that

NOTE Confidence: 0.85469599

00:44:55.494 --> 00:44:57.536 report that had pathogenic variant

NOTE Confidence: 0.85469599

00:44:57.536 --> 00:45:00.734 detected by uploading the raw data,

NOTE Confidence: 0.85469599

00:45:00.740 --> 00:45:03.132 40% were a false positive and you can

NOTE Confidence: 0.85469599

00:45:03.132 --> 00:45:05.625 see most of those were in jeans that

NOTE Confidence: 0.85469599

00:45:05.625 --> 00:45:07.758 we often know about PRC two BRCA,

NOTE Confidence: 0.85469599

00:45:07.760 --> 00:45:11.656 one ATM MLH one related to Lynch syndrome.

NOTE Confidence: 0.85469599

00:45:11.660 --> 00:45:16.470 Check two TB 53 so genes that if

NOTE Confidence: 0.85469599

00:45:16.470 --> 00:45:18.990 someone had a mutation those genes

NOTE Confidence: 0.85469599

00:45:18.990 --> 00:45:21.327 that has clinical significance so

NOTE Confidence: 0.85469599

00:45:21.327 --> 00:45:24.309 individuals if they aren't aware of.

NOTE Confidence: 0.85469599

00:45:24.310 --> 00:45:26.280 The risk of false positives.

NOTE Confidence: 0.85469599

00:45:26.280 --> 00:45:28.734 They may be doing screening or

NOTE Confidence: 0.85469599

00:45:28.734 --> 00:45:30.370 preventative surgeries that may

NOTE Confidence: 0.922767203571429

00:45:30.440 --> 00:45:31.760 not be indicated so,

NOTE Confidence: 0.922767203571429

00:45:31.760 --> 00:45:33.550 particularly with things like raw

NOTE Confidence: 0.922767203571429

00:45:33.550 --> 00:45:35.870 data and direct to consumer testing.
NOTE Confidence: 0.922767203571429

00:45:35.870 --> 00:45:37.310 If there is a family history
NOTE Confidence: 0.922767203571429

00:45:37.310 --> 00:45:38.860 and a patient is concerned,
NOTE Confidence: 0.922767203571429

00:45:38.860 --> 00:45:40.940 the best thing they can do is meet
NOTE Confidence: 0.922767203571429

00:45:40.940 --> 00:45:43.585 with a genetic counselor and have a
NOTE Confidence: 0.922767203571429

00:45:43.585 --> 00:45:45.645 clinical test for hereditary cancer.
NOTE Confidence: 0.922767203571429

00:45:45.650 --> 00:45:47.645 And there are position statements
NOTE Confidence: 0.922767203571429

00:45:47.645 --> 00:45:50.117 from the National Society of Genetic
NOTE Confidence: 0.922767203571429

00:45:50.117 --> 00:45:52.553 Counselors about at home testing and
NOTE Confidence: 0.922767203571429

00:45:52.553 --> 00:45:55.016 things to consider and the American
NOTE Confidence: 0.922767203571429

00:45:55.016 --> 00:45:56.981 College of Obese and Gynecologists
NOTE Confidence: 0.922767203571429

00:45:56.981 --> 00:46:00.260 also have a position statement.
NOTE Confidence: 0.922767203571429

00:46:00.260 --> 00:46:01.940 Actually say quite nicely in their
NOTE Confidence: 0.922767203571429

00:46:01.940 --> 00:46:03.610 title that it creates confusion,
NOTE Confidence: 0.922767203571429

00:46:03.610 --> 00:46:06.120 so direct to consumer testing.
NOTE Confidence: 0.922767203571429

00:46:06.120 --> 00:46:07.338 Again for ancestry.

NOTE Confidence: 0.922767203571429
00:46:07.338 --> 00:46:09.368 For small things like that
NOTE Confidence: 0.922767203571429
00:46:09.368 --> 00:46:11.010 is something that again,
NOTE Confidence: 0.922767203571429
00:46:11.010 --> 00:46:13.110 cocktail genetic something that's fun.
NOTE Confidence: 0.922767203571429
00:46:13.110 --> 00:46:14.874 But for things like BRCA one and
NOTE Confidence: 0.922767203571429
00:46:14.874 --> 00:46:16.440 two testing or anything else,
NOTE Confidence: 0.922767203571429
00:46:16.440 --> 00:46:18.246 it's clinically indicated.
NOTE Confidence: 0.922767203571429
00:46:18.246 --> 00:46:20.654 Definitely a clinical diagnostic
NOTE Confidence: 0.922767203571429
00:46:20.654 --> 00:46:22.460 test is warranted,
NOTE Confidence: 0.922767203571429
00:46:22.460 --> 00:46:26.378 particularly if there's a family history.
NOTE Confidence: 0.922767203571429
00:46:26.380 --> 00:46:27.764 So let's talk about,
NOTE Confidence: 0.922767203571429
00:46:27.764 --> 00:46:29.494 you know genetic testing and
NOTE Confidence: 0.922767203571429
00:46:29.494 --> 00:46:31.836 kind of what would we talk about
NOTE Confidence: 0.922767203571429
00:46:31.836 --> 00:46:33.120 with with patients so.
NOTE Confidence: 0.922767203571429
00:46:33.120 --> 00:46:34.440 When we do genetic testing,
NOTE Confidence: 0.922767203571429
00:46:34.440 --> 00:46:36.330 there are three possible results we
NOTE Confidence: 0.922767203571429

00:46:36.330 --> 00:46:38.991 can get from a task which will one
NOTE Confidence: 0.922767203571429

00:46:38.991 --> 00:46:40.995 will stay positive means there is
NOTE Confidence: 0.922767203571429

00:46:41.060 --> 00:46:42.920 a pathogenic mutation detected in
NOTE Confidence: 0.922767203571429

00:46:42.920 --> 00:46:45.584 one of the genes analyzed and that
NOTE Confidence: 0.922767203571429

00:46:45.584 --> 00:46:47.594 there are associated cancer risks.
NOTE Confidence: 0.922767203571429

00:46:47.600 --> 00:46:48.899 Negative straightforward means
NOTE Confidence: 0.922767203571429

00:46:48.899 --> 00:46:51.497 that when the genes are analyzed,
NOTE Confidence: 0.922767203571429

00:46:51.500 --> 00:46:53.690 there's no known pathogenic mutations.
NOTE Confidence: 0.922767203571429

00:46:53.690 --> 00:46:55.542 The jeans look normal,
NOTE Confidence: 0.922767203571429

00:46:55.542 --> 00:46:58.320 there's no detectable hereditary cancer risk.
NOTE Confidence: 0.922767203571429

00:46:58.320 --> 00:47:00.650 And then there's the variant
NOTE Confidence: 0.922767203571429

00:47:00.650 --> 00:47:02.048 of uncertain significance,
NOTE Confidence: 0.922767203571429

00:47:02.050 --> 00:47:05.249 which is a variation in a gene.
NOTE Confidence: 0.922767203571429

00:47:05.250 --> 00:47:08.310 That the laboratory has not yet
NOTE Confidence: 0.922767203571429

00:47:08.310 --> 00:47:10.913 classified to pathogenic meaning it's
NOTE Confidence: 0.922767203571429

00:47:10.913 --> 00:47:14.153 causing cancer risk or benign variation,

NOTE Confidence: 0.922767203571429
00:47:14.153 --> 00:47:17.158 meaning it's just a polymorphism
NOTE Confidence: 0.922767203571429
00:47:17.158 --> 00:47:20.447 that's actually not impacting the gene.
NOTE Confidence: 0.922767203571429
00:47:20.450 --> 00:47:22.550 And variants of uncertain significance.
NOTE Confidence: 0.922767203571429
00:47:22.550 --> 00:47:23.828 They are very.
NOTE Confidence: 0.922767203571429
00:47:23.828 --> 00:47:25.106 They're quite common,
NOTE Confidence: 0.922767203571429
00:47:25.110 --> 00:47:27.576 especially when we do larger testing,
NOTE Confidence: 0.922767203571429
00:47:27.580 --> 00:47:29.052 which we do nowadays.
NOTE Confidence: 0.922767203571429
00:47:29.052 --> 00:47:30.892 We actually find variants of
NOTE Confidence: 0.922767203571429
00:47:30.892 --> 00:47:32.130 uncertain significance.
NOTE Confidence: 0.922767203571429
00:47:32.130 --> 00:47:34.308 20 to 30% of the time.
NOTE Confidence: 0.922767203571429
00:47:34.310 --> 00:47:36.650 This is an example report,
NOTE Confidence: 0.922767203571429
00:47:36.650 --> 00:47:38.510 and particularly it's always hard
NOTE Confidence: 0.922767203571429
00:47:38.510 --> 00:47:41.147 when you get a variant of uncertain
NOTE Confidence: 0.922767203571429
00:47:41.147 --> 00:47:43.625 significance in a gene like BRCA 2,
NOTE Confidence: 0.922767203571429
00:47:43.630 --> 00:47:47.140 but most of these variants are
NOTE Confidence: 0.922767203571429

00:47:47.140 --> 00:47:50.439 reclassified by the laboratory 2 benign.

NOTE Confidence: 0.922767203571429

00:47:50.440 --> 00:47:51.892 Two normal changes.

NOTE Confidence: 0.922767203571429

00:47:51.892 --> 00:47:54.796 That's why on the previous slide,

NOTE Confidence: 0.922767203571429

00:47:54.800 --> 00:47:56.950 if a varying divine stern

NOTE Confidence: 0.922767203571429

00:47:56.950 --> 00:47:58.240 significance is detected,

NOTE Confidence: 0.922767203571429

00:47:58.240 --> 00:47:59.948 medical management is not

NOTE Confidence: 0.922767203571429

00:47:59.948 --> 00:48:01.656 based on that variant.

NOTE Confidence: 0.922767203571429

00:48:01.660 --> 00:48:04.010 It's based only on personal

NOTE Confidence: 0.922767203571429

00:48:04.010 --> 00:48:05.420 family history factors.

NOTE Confidence: 0.922767203571429

00:48:05.420 --> 00:48:07.946 We have seen cases where someone

NOTE Confidence: 0.922767203571429

00:48:07.946 --> 00:48:10.860 is followed as if they have a

NOTE Confidence: 0.922767203571429

00:48:10.860 --> 00:48:13.552 real mutation in BRCA 1, BRCA 2,

NOTE Confidence: 0.922767203571429

00:48:13.552 --> 00:48:15.532 another gene that's actually a

NOTE Confidence: 0.922767203571429

00:48:15.532 --> 00:48:17.600 variant of uncertain significance

NOTE Confidence: 0.922767203571429

00:48:17.600 --> 00:48:20.576 and they're screened. They they may.

NOTE Confidence: 0.922767203571429

00:48:20.576 --> 00:48:22.148 They may have preventive surgery and

NOTE Confidence: 0.922767203571429
00:48:22.148 --> 00:48:23.780 then they find out in the future.
NOTE Confidence: 0.922767203571429
00:48:23.780 --> 00:48:25.817 It is actually a benign variation so
NOTE Confidence: 0.922767203571429
00:48:25.817 --> 00:48:28.139 we always go back to family history
NOTE Confidence: 0.922767203571429
00:48:28.139 --> 00:48:29.864 to guide management is something
NOTE Confidence: 0.922767203571429
00:48:29.864 --> 00:48:32.235 we kind of say innocent till proven
NOTE Confidence: 0.922767203571429
00:48:32.235 --> 00:48:34.585 guilty because most of the time the
NOTE Confidence: 0.922767203571429
00:48:34.585 --> 00:48:36.655 laboratory will update us with the
NOTE Confidence: 0.922767203571429
00:48:36.655 --> 00:48:39.235 report when they can get enough
NOTE Confidence: 0.922767203571429
00:48:39.235 --> 00:48:41.405 information to change their classification.
NOTE Confidence: 0.922767203571429
00:48:41.410 --> 00:48:41.703 Uhm,
NOTE Confidence: 0.922767203571429
00:48:41.703 --> 00:48:41.996 no,
NOTE Confidence: 0.922767203571429
00:48:41.996 --> 00:48:44.047 and actually I already talked about this,
NOTE Confidence: 0.922767203571429
00:48:44.050 --> 00:48:45.698 so I I've already.
NOTE Confidence: 0.922767203571429
00:48:45.698 --> 00:48:48.170 I'm I'm when someone step ahead.
NOTE Confidence: 0.922767203571429
00:48:48.170 --> 00:48:50.826 So really we also I want to mention
NOTE Confidence: 0.922767203571429

00:48:50.826 --> 00:48:54.152 point out we don't test relatives for
NOTE Confidence: 0.922767203571429

00:48:54.152 --> 00:48:56.220 variants of uncertain significance,
NOTE Confidence: 0.922136335

00:48:56.220 --> 00:48:59.258 only if it might be helpful in
NOTE Confidence: 0.922136335

00:48:59.258 --> 00:49:00.560 reclassifying that variant,
NOTE Confidence: 0.922136335

00:49:00.560 --> 00:49:02.480 meaning that sometimes it can be
NOTE Confidence: 0.922136335

00:49:02.480 --> 00:49:04.830 helpful to know did the variant come
NOTE Confidence: 0.922136335

00:49:04.830 --> 00:49:07.161 from your momma from your dad because
NOTE Confidence: 0.922136335

00:49:07.226 --> 00:49:09.038 your dad had pancreatic cancer and
NOTE Confidence: 0.922136335

00:49:09.038 --> 00:49:11.518 your mom is 85 and still living so.
NOTE Confidence: 0.922136335

00:49:11.518 --> 00:49:13.420 If the speakers say two variant
NOTE Confidence: 0.922136335

00:49:13.482 --> 00:49:14.638 came from your mom,
NOTE Confidence: 0.922136335

00:49:14.640 --> 00:49:16.116 I we might be less suspicious,
NOTE Confidence: 0.922136335

00:49:16.120 --> 00:49:18.576 but it came from your dad that maybe
NOTE Confidence: 0.922136335

00:49:18.576 --> 00:49:20.697 could maybe could add evidence or at
NOTE Confidence: 0.922136335

00:49:20.697 --> 00:49:23.290 least keep it at the level that it is.
NOTE Confidence: 0.922136335

00:49:23.290 --> 00:49:25.498 So that's why we only really test relatives.

NOTE Confidence: 0.922136335
00:49:25.500 --> 00:49:26.500 If it might be helpful,
NOTE Confidence: 0.922136335
00:49:26.500 --> 00:49:30.250 and reclassifying the the variant.
NOTE Confidence: 0.922136335
00:49:30.250 --> 00:49:33.013 And and this this can be hard for patients.
NOTE Confidence: 0.922136335
00:49:33.020 --> 00:49:36.346 It can be hard to have that uncertainty of.
NOTE Confidence: 0.922136335
00:49:36.346 --> 00:49:37.816 Is this something I'm gonna
NOTE Confidence: 0.922136335
00:49:37.816 --> 00:49:38.930 have to think about?
NOTE Confidence: 0.922136335
00:49:38.930 --> 00:49:40.792 It is something that is actually going
NOTE Confidence: 0.922136335
00:49:40.792 --> 00:49:43.194 to we find out that I'm as positive in
NOTE Confidence: 0.922136335
00:49:43.194 --> 00:49:45.824 the future and it can be hard to how
NOTE Confidence: 0.922136335
00:49:45.824 --> 00:49:48.010 to communicate this to family members.
NOTE Confidence: 0.922136335
00:49:48.010 --> 00:49:49.024 It's you know,
NOTE Confidence: 0.922136335
00:49:49.024 --> 00:49:50.376 it's something members can
NOTE Confidence: 0.922136335
00:49:50.376 --> 00:49:51.390 won't understand it.
NOTE Confidence: 0.922136335
00:49:51.390 --> 00:49:53.014 They might want to be tested for it.
NOTE Confidence: 0.922136335
00:49:53.020 --> 00:49:55.491 So this can be particularly tricky in
NOTE Confidence: 0.922136335

00:49:55.491 --> 00:49:57.419 communicating this with with patients,
NOTE Confidence: 0.922136335

00:49:57.420 --> 00:49:59.695 which is why we do take time
NOTE Confidence: 0.922136335

00:49:59.695 --> 00:50:00.670 before and after.
NOTE Confidence: 0.922136335

00:50:00.670 --> 00:50:02.350 Even before someone has testing to
NOTE Confidence: 0.922136335

00:50:02.350 --> 00:50:04.378 prep them and let them know this
NOTE Confidence: 0.922136335

00:50:04.378 --> 00:50:06.070 isn't this as a possible result,
NOTE Confidence: 0.922136335

00:50:06.070 --> 00:50:09.619 we can find that is quite common.
NOTE Confidence: 0.922136335

00:50:09.620 --> 00:50:12.620 And testing options are, they're all.
NOTE Confidence: 0.922136335

00:50:12.620 --> 00:50:14.168 They're all a little bit different.
NOTE Confidence: 0.922136335

00:50:14.170 --> 00:50:16.120 We do single site testing.
NOTE Confidence: 0.922136335

00:50:16.120 --> 00:50:18.376 If there's a known mutation in the family
NOTE Confidence: 0.922136335

00:50:18.376 --> 00:50:20.880 and no other suspicious cancer history,
NOTE Confidence: 0.922136335

00:50:20.880 --> 00:50:23.790 meaning that there's a mutation.
NOTE Confidence: 0.922136335

00:50:23.790 --> 00:50:25.420 We've confirmed with their report,
NOTE Confidence: 0.922136335

00:50:25.420 --> 00:50:26.458 there's nothing else.
NOTE Confidence: 0.922136335

00:50:26.458 --> 00:50:28.534 In those cases we would test

NOTE Confidence: 0.922136335
00:50:28.534 --> 00:50:30.619 for just that single mutation.
NOTE Confidence: 0.922136335
00:50:30.620 --> 00:50:32.430 Other options for those families
NOTE Confidence: 0.922136335
00:50:32.430 --> 00:50:34.683 where there's not a no mutation
NOTE Confidence: 0.922136335
00:50:34.683 --> 00:50:36.939 is a cancer site specific panel
NOTE Confidence: 0.922136335
00:50:36.940 --> 00:50:39.108 meaning related to genes.
NOTE Confidence: 0.922136335
00:50:39.108 --> 00:50:39.650 Only.
NOTE Confidence: 0.922136335
00:50:39.650 --> 00:50:41.228 Genes that are only related to
NOTE Confidence: 0.922136335
00:50:41.228 --> 00:50:43.088 the cancer is seen in the family,
NOTE Confidence: 0.922136335
00:50:43.090 --> 00:50:44.944 meaning that maybe if there's just
NOTE Confidence: 0.922136335
00:50:44.944 --> 00:50:46.968 breast or varying cancer in the family,
NOTE Confidence: 0.922136335
00:50:46.970 --> 00:50:49.588 limiting it to jeans that have a
NOTE Confidence: 0.922136335
00:50:49.588 --> 00:50:51.277 known association with hereditary
NOTE Confidence: 0.922136335
00:50:51.277 --> 00:50:53.237 breast or ovarian cancer.
NOTE Confidence: 0.922136335
00:50:53.240 --> 00:50:55.347 Well, this colon cancer in the family,
NOTE Confidence: 0.922136335
00:50:55.350 --> 00:50:58.115 keeping it only to hereditary colon cancer,
NOTE Confidence: 0.922136335

00:50:58.120 --> 00:50:58.780 expanded panels.
NOTE Confidence: 0.922136335

00:50:58.780 --> 00:51:01.110 We've been doing more often, meaning that.
NOTE Confidence: 0.922136335

00:51:01.110 --> 00:51:03.150 You look at multiple genes related
NOTE Confidence: 0.922136335

00:51:03.150 --> 00:51:04.809 to several types of cancer,
NOTE Confidence: 0.922136335

00:51:04.810 --> 00:51:05.546 including cancers.
NOTE Confidence: 0.922136335

00:51:05.546 --> 00:51:08.122 We may not even be seeing in
NOTE Confidence: 0.922136335

00:51:08.122 --> 00:51:09.010 someone's family.
NOTE Confidence: 0.922136335

00:51:09.010 --> 00:51:11.058 Such as colon cancer,
NOTE Confidence: 0.922136335

00:51:11.058 --> 00:51:12.430 Melanoma, uterine cancer,
NOTE Confidence: 0.922136335

00:51:12.430 --> 00:51:13.290 stomach cancer.
NOTE Confidence: 0.922136335

00:51:13.290 --> 00:51:16.270 So essentially all the more common horizon
NOTE Confidence: 0.922136335

00:51:16.270 --> 00:51:19.258 right cancer types we often can do that and
NOTE Confidence: 0.922136335

00:51:19.327 --> 00:51:21.994 will be called an expanded panel testing.
NOTE Confidence: 0.922136335

00:51:22.000 --> 00:51:24.118 But panel testing there are considerations
NOTE Confidence: 0.922136335

00:51:24.118 --> 00:51:26.188 because the more genes we look at,
NOTE Confidence: 0.922136335

00:51:26.190 --> 00:51:28.074 the higher chance we'll find something

NOTE Confidence: 0.922136335
00:51:28.074 --> 00:51:29.640 in a hereditary cancer gene,
NOTE Confidence: 0.922136335
00:51:29.640 --> 00:51:31.564 which could be expected
NOTE Confidence: 0.922136335
00:51:31.564 --> 00:51:33.007 or possibly unexpected.
NOTE Confidence: 0.922136335
00:51:33.010 --> 00:51:35.080 Some patients don't want to
NOTE Confidence: 0.922136335
00:51:35.080 --> 00:51:36.736 find an unexpected result,
NOTE Confidence: 0.922136335
00:51:36.740 --> 00:51:37.908 as I mentioned earlier,
NOTE Confidence: 0.922136335
00:51:37.908 --> 00:51:39.660 there are some genes that are
NOTE Confidence: 0.922136335
00:51:39.720 --> 00:51:41.320 included in testing nowadays,
NOTE Confidence: 0.922136335
00:51:41.320 --> 00:51:43.016 particularly panel testing that
NOTE Confidence: 0.922136335
00:51:43.016 --> 00:51:46.023 are have not been studied as long
NOTE Confidence: 0.922136335
00:51:46.023 --> 00:51:48.327 they have an unknown cancer risk,
NOTE Confidence: 0.922136335
00:51:48.330 --> 00:51:50.730 so there may not actually be
NOTE Confidence: 0.922136335
00:51:50.730 --> 00:51:52.330 screening recommendations 'cause for
NOTE Confidence: 0.93351909
00:51:52.396 --> 00:51:54.261 syndrome. Like Lynch syndrome
NOTE Confidence: 0.93351909
00:51:54.261 --> 00:51:56.009 and mutations in BRCA,
NOTE Confidence: 0.93351909

00:51:56.010 --> 00:51:58.607 one of your C2 there are very
NOTE Confidence: 0.93351909

00:51:58.607 --> 00:51:59.720 clear screen recommendations
NOTE Confidence: 0.93351909

00:51:59.786 --> 00:52:01.766 with some of these newer genes.
NOTE Confidence: 0.93351909

00:52:01.770 --> 00:52:04.274 We can find a mutation and still just
NOTE Confidence: 0.93351909

00:52:04.274 --> 00:52:06.312 follow someone based on personal or
NOTE Confidence: 0.93351909

00:52:06.312 --> 00:52:08.334 family history until we learn about
NOTE Confidence: 0.93351909

00:52:08.400 --> 00:52:10.626 what that gene is doing specifically.
NOTE Confidence: 0.93351909

00:52:10.630 --> 00:52:12.784 And as I mentioned earlier that
NOTE Confidence: 0.93351909

00:52:12.784 --> 00:52:15.572 when we do a larger panel probably
NOTE Confidence: 0.93351909

00:52:15.572 --> 00:52:17.264 more standard panel that we do,
NOTE Confidence: 0.93351909

00:52:17.270 --> 00:52:20.934 we find a VUS 20 to 30% of the
NOTE Confidence: 0.93351909

00:52:20.934 --> 00:52:23.256 time so that's that's quite a.
NOTE Confidence: 0.93351909

00:52:23.260 --> 00:52:26.490 A big chunk of the time we find at least one,
NOTE Confidence: 0.93351909

00:52:26.490 --> 00:52:28.634 and so some patients may not want the
NOTE Confidence: 0.93351909

00:52:28.634 --> 00:52:30.568 possibility of finding uncertain results,
NOTE Confidence: 0.93351909

00:52:30.570 --> 00:52:32.700 so something else to also think

NOTE Confidence: 0.93351909

00:52:32.700 --> 00:52:35.309 about when we offer genetic testing.

NOTE Confidence: 0.93351909

00:52:35.310 --> 00:52:37.305 Some of the benefits of genetic testing,

NOTE Confidence: 0.93351909

00:52:37.310 --> 00:52:39.140 you know it can end uncertainty.

NOTE Confidence: 0.93351909

00:52:39.140 --> 00:52:40.769 You know if there is cancer in the family,

NOTE Confidence: 0.93351909

00:52:40.770 --> 00:52:42.822 it can be helpful to know do is there

NOTE Confidence: 0.93351909

00:52:42.822 --> 00:52:44.825 a reason for why there's cancer in

NOTE Confidence: 0.93351909

00:52:44.825 --> 00:52:47.128 my family and do I have the reason

NOTE Confidence: 0.93351909

00:52:47.128 --> 00:52:48.772 for the cancer in the family?

NOTE Confidence: 0.93351909

00:52:48.780 --> 00:52:51.230 If I cancer risks for an individual,

NOTE Confidence: 0.93351909

00:52:51.230 --> 00:52:53.110 so then we know what screening we can.

NOTE Confidence: 0.93351909

00:52:53.110 --> 00:52:54.370 We need to be doing.

NOTE Confidence: 0.93351909

00:52:54.370 --> 00:52:56.666 It can also clarify cancer risks for

NOTE Confidence: 0.93351909

00:52:56.666 --> 00:52:58.626 relatives. If there is a positive result.

NOTE Confidence: 0.93351909

00:52:58.630 --> 00:53:00.490 We then know that other relatives

NOTE Confidence: 0.93351909

00:53:00.490 --> 00:53:02.478 would be at risk of inheriting

NOTE Confidence: 0.93351909

00:53:02.478 --> 00:53:04.524 that same mutation and then they
NOTE Confidence: 0.93351909

00:53:04.524 --> 00:53:06.700 can also have their own testing.
NOTE Confidence: 0.93351909

00:53:06.700 --> 00:53:09.060 And it can aid in medical decision making.
NOTE Confidence: 0.93351909

00:53:09.060 --> 00:53:10.672 Knowing again what screening
NOTE Confidence: 0.93351909

00:53:10.672 --> 00:53:12.284 they should be doing.
NOTE Confidence: 0.93351909

00:53:12.290 --> 00:53:14.468 Is it worth thinking about a
NOTE Confidence: 0.93351909

00:53:14.468 --> 00:53:15.920 prophylactic bilateral mastectomy and
NOTE Confidence: 0.93351909

00:53:15.978 --> 00:53:17.958 that can actually relieving anxiety?
NOTE Confidence: 0.93351909

00:53:17.960 --> 00:53:20.036 There are studies showing that even
NOTE Confidence: 0.93351909

00:53:20.036 --> 00:53:21.920 a positive result could actually
NOTE Confidence: 0.93351909

00:53:21.920 --> 00:53:23.240 be anxiety relieving,
NOTE Confidence: 0.93351909

00:53:23.240 --> 00:53:26.018 because some actually has the answer.
NOTE Confidence: 0.93351909

00:53:26.020 --> 00:53:27.526 They actually know why there's cancer
NOTE Confidence: 0.93351909

00:53:27.526 --> 00:53:29.576 in the family, what's causing it,
NOTE Confidence: 0.93351909

00:53:29.576 --> 00:53:31.736 and they know the specific
NOTE Confidence: 0.93351909

00:53:31.736 --> 00:53:33.709 risks instead of an unknown.

NOTE Confidence: 0.93351909

00:53:33.710 --> 00:53:35.485 There are some risks and

NOTE Confidence: 0.93351909

00:53:35.485 --> 00:53:36.905 limitations to genetic testing,

NOTE Confidence: 0.93351909

00:53:36.910 --> 00:53:38.915 such as a negative result

NOTE Confidence: 0.93351909

00:53:38.915 --> 00:53:40.118 may be uninformative,

NOTE Confidence: 0.93351909

00:53:40.120 --> 00:53:42.628 or it may be falsely reassuring.

NOTE Confidence: 0.93351909

00:53:42.630 --> 00:53:44.774 There are concerns about

NOTE Confidence: 0.93351909

00:53:44.774 --> 00:53:45.846 genetic discrimination,

NOTE Confidence: 0.93351909

00:53:45.850 --> 00:53:48.026 so there are laws in place that do

NOTE Confidence: 0.93351909

00:53:48.026 --> 00:53:50.374 make it illegal for most health

NOTE Confidence: 0.93351909

00:53:50.374 --> 00:53:52.544 insurance companies and most employers

NOTE Confidence: 0.93351909

00:53:52.544 --> 00:53:54.492 from discriminating against someone

NOTE Confidence: 0.93351909

00:53:54.492 --> 00:53:57.384 with a positive genetic test result,

NOTE Confidence: 0.93351909

00:53:57.390 --> 00:53:59.973 but these do not apply to things

NOTE Confidence: 0.93351909

00:53:59.973 --> 00:54:01.080 like life insurance,

NOTE Confidence: 0.93351909

00:54:01.080 --> 00:54:03.824 long term care or disability and it actually.

NOTE Confidence: 0.93351909

00:54:03.830 --> 00:54:05.598 Possibly could impact fitness
NOTE Confidence: 0.93351909

00:54:05.598 --> 00:54:07.808 for duty in the military,
NOTE Confidence: 0.93351909

00:54:07.810 --> 00:54:10.358 so it is something to think about
NOTE Confidence: 0.93351909

00:54:10.358 --> 00:54:12.689 when someone has genetic testing.
NOTE Confidence: 0.93351909

00:54:12.690 --> 00:54:14.906 You know what policies they have in place?
NOTE Confidence: 0.93351909

00:54:14.910 --> 00:54:16.956 Are they actively serving in the
NOTE Confidence: 0.93351909

00:54:16.956 --> 00:54:19.339 military just to just to think about?
NOTE Confidence: 0.93351909

00:54:19.340 --> 00:54:20.624 In timing of testing,
NOTE Confidence: 0.93351909

00:54:20.624 --> 00:54:22.229 might not be optimal depending
NOTE Confidence: 0.93351909

00:54:22.229 --> 00:54:23.659 on someone's current.
NOTE Confidence: 0.93351909

00:54:23.660 --> 00:54:26.175 Current lifestyle or current current
NOTE Confidence: 0.93351909

00:54:26.175 --> 00:54:28.858 plans and Shank testing is an option.
NOTE Confidence: 0.93351909

00:54:28.860 --> 00:54:30.064 Even though there are
NOTE Confidence: 0.93351909

00:54:30.064 --> 00:54:30.666 clinical recommendations,
NOTE Confidence: 0.93351909

00:54:30.670 --> 00:54:32.602 it is an option and for some
NOTE Confidence: 0.93351909

00:54:32.602 --> 00:54:34.033 patients they actually prefer not

NOTE Confidence: 0.93351909

00:54:34.033 --> 00:54:35.659 to know if they have hereditary

NOTE Confidence: 0.93351909

00:54:35.659 --> 00:54:37.613 risk of cancer and they may not

NOTE Confidence: 0.93351909

00:54:37.613 --> 00:54:39.461 wish to know future cancer risks.

NOTE Confidence: 0.93351909

00:54:39.461 --> 00:54:42.407 There are limitations in the testing,

NOTE Confidence: 0.93351909

00:54:42.410 --> 00:54:45.330 so it is possible even though are the

NOTE Confidence: 0.93351909

00:54:45.330 --> 00:54:47.179 testing technology has grown a lot,

NOTE Confidence: 0.849544856

00:54:47.180 --> 00:54:49.505 though there are sometimes undetectable

NOTE Confidence: 0.849544856

00:54:49.505 --> 00:54:51.830 mutations and current technology that

NOTE Confidence: 0.849544856

00:54:51.896 --> 00:54:54.304 could be missed in a known hereditary

NOTE Confidence: 0.849544856

00:54:54.304 --> 00:54:56.126 cancer gene testing technology will

NOTE Confidence: 0.849544856

00:54:56.126 --> 00:54:57.936 change and improve overtime just

NOTE Confidence: 0.849544856

00:54:57.936 --> 00:55:00.230 like it has in the last ten years.

NOTE Confidence: 0.849544856

00:55:00.230 --> 00:55:03.313 But that is a limitation and not all

NOTE Confidence: 0.849544856

00:55:03.313 --> 00:55:05.318 inherited cancer syndromes are known.

NOTE Confidence: 0.849544856

00:55:05.320 --> 00:55:08.096 So in the future kind of like I

NOTE Confidence: 0.849544856

00:55:08.096 --> 00:55:10.380 mentioned with BRCA one and BRCA 2.
NOTE Confidence: 0.849544856

00:55:10.380 --> 00:55:13.800 Four 2013 is out is outdated.
NOTE Confidence: 0.849544856

00:55:13.800 --> 00:55:15.930 Is possible I could be saying
NOTE Confidence: 0.849544856

00:55:15.930 --> 00:55:18.804 in you know 2030 if any test
NOTE Confidence: 0.849544856

00:55:18.804 --> 00:55:21.111 after 20 before 2033 is outdated.
NOTE Confidence: 0.849544856

00:55:21.111 --> 00:55:23.819 You know we are likely learn more about
NOTE Confidence: 0.849544856

00:55:23.819 --> 00:55:26.327 inherited cancer syndromes and new genes.
NOTE Confidence: 0.849544856

00:55:26.330 --> 00:55:29.066 So more testing may be needed in the future
NOTE Confidence: 0.849544856

00:55:29.066 --> 00:55:31.815 to help clarify risk for some families.
NOTE Confidence: 0.849544856

00:55:31.820 --> 00:55:34.376 And this is an example of.
NOTE Confidence: 0.849544856

00:55:34.380 --> 00:55:35.772 Uninformative genetic testing,
NOTE Confidence: 0.849544856

00:55:35.772 --> 00:55:39.020 where the probe and two were there.
NOTE Confidence: 0.849544856

00:55:39.020 --> 00:55:40.988 The circle there is pointing to
NOTE Confidence: 0.849544856

00:55:40.990 --> 00:55:42.580 she's had negative genetic testing,
NOTE Confidence: 0.849544856

00:55:42.580 --> 00:55:44.736 but has a very striking family history,
NOTE Confidence: 0.849544856

00:55:44.740 --> 00:55:46.888 so her testing is really not

NOTE Confidence: 0.849544856

00:55:46.888 --> 00:55:48.755 informative and it's not someone

NOTE Confidence: 0.849544856

00:55:48.755 --> 00:55:50.945 that you feel comfortable saying oh

NOTE Confidence: 0.849544856

00:55:50.945 --> 00:55:53.728 you are at average risk for cancer.

NOTE Confidence: 0.849544856

00:55:53.730 --> 00:55:55.627 We always go back to family history

NOTE Confidence: 0.849544856

00:55:55.627 --> 00:55:58.276 to This is why we go back to family

NOTE Confidence: 0.849544856

00:55:58.276 --> 00:56:00.238 history to guys screening until we

NOTE Confidence: 0.849544856

00:56:00.238 --> 00:56:02.038 can get informative relative tested

NOTE Confidence: 0.849544856

00:56:02.038 --> 00:56:05.096 and determine is is there a familial.

NOTE Confidence: 0.849544856

00:56:05.096 --> 00:56:05.590 Mutation,

NOTE Confidence: 0.849544856

00:56:05.590 --> 00:56:08.894 but maybe this probe and did not inherit.

NOTE Confidence: 0.849544856

00:56:08.900 --> 00:56:10.104 Which is why, again,

NOTE Confidence: 0.849544856

00:56:10.104 --> 00:56:11.910 it is very important for sometimes

NOTE Confidence: 0.849544856

00:56:11.968 --> 00:56:13.938 testing the affected relative first,

NOTE Confidence: 0.849544856

00:56:13.940 --> 00:56:15.823 and if that relative has had genetic

NOTE Confidence: 0.849544856

00:56:15.823 --> 00:56:17.581 testing it is really important for

NOTE Confidence: 0.849544856

00:56:17.581 --> 00:56:19.653 you to be talking to your patients
NOTE Confidence: 0.849544856

00:56:19.715 --> 00:56:21.623 and explaining it is important to
NOTE Confidence: 0.849544856

00:56:21.623 --> 00:56:25.560 get those medical records for us.
NOTE Confidence: 0.849544856

00:56:25.560 --> 00:56:27.400 Engineer counseling in the session,
NOTE Confidence: 0.849544856

00:56:27.400 --> 00:56:29.974 what would I do and will my colleagues do?
NOTE Confidence: 0.849544856

00:56:29.980 --> 00:56:32.710 It's really a combination of education,
NOTE Confidence: 0.849544856

00:56:32.710 --> 00:56:34.750 talking about the diagnosis
NOTE Confidence: 0.849544856

00:56:34.750 --> 00:56:36.280 of hereditary cancers,
NOTE Confidence: 0.849544856

00:56:36.280 --> 00:56:38.720 syndromes offering psychosocial support,
NOTE Confidence: 0.849544856

00:56:38.720 --> 00:56:41.160 advocating for our patients,
NOTE Confidence: 0.849544856

00:56:41.160 --> 00:56:42.830 talking about talking about risks.
NOTE Confidence: 0.849544856

00:56:42.830 --> 00:56:45.140 So it is really this.
NOTE Confidence: 0.849544856

00:56:45.140 --> 00:56:47.438 We're we're here for resource for,
NOTE Confidence: 0.849544856

00:56:47.440 --> 00:56:49.045 for patients were also here
NOTE Confidence: 0.849544856

00:56:49.045 --> 00:56:50.329 for resource for you.
NOTE Confidence: 0.849544856

00:56:50.330 --> 00:56:51.094 You know,

NOTE Confidence: 0.849544856

00:56:51.094 --> 00:56:53.768 in terms of in terms of risk

NOTE Confidence: 0.849544856

00:56:53.768 --> 00:56:56.297 assessments and patients going forward.

NOTE Confidence: 0.849544856

00:56:56.300 --> 00:56:58.463 The main I'll go through some of

NOTE Confidence: 0.849544856

00:56:58.463 --> 00:57:00.420 this this briefly 'cause I don't

NOTE Confidence: 0.849544856

00:57:00.420 --> 00:57:02.352 wanna go over too much overtime,

NOTE Confidence: 0.849544856

00:57:02.360 --> 00:57:05.140 but there is a lot.

NOTE Confidence: 0.849544856

00:57:05.140 --> 00:57:07.688 There's a lot in a genetic counseling

NOTE Confidence: 0.849544856

00:57:07.688 --> 00:57:10.669 session where we go through medical history.

NOTE Confidence: 0.849544856

00:57:10.670 --> 00:57:12.956 We take a detailed family history.

NOTE Confidence: 0.849544856

00:57:12.960 --> 00:57:14.340 We talked about the risk factors.

NOTE Confidence: 0.849544856

00:57:14.340 --> 00:57:15.164 We've seen, the family,

NOTE Confidence: 0.849544856

00:57:15.164 --> 00:57:16.708 we do, a risk assessment.

NOTE Confidence: 0.849544856

00:57:16.708 --> 00:57:19.340 We talk about specific genes or specific

NOTE Confidence: 0.849544856

00:57:19.406 --> 00:57:22.014 syndromes the patient may be at risk for.

NOTE Confidence: 0.849544856

00:57:22.020 --> 00:57:23.250 We talk about,

NOTE Confidence: 0.849544856

00:57:23.250 --> 00:57:26.120 you know the what hereditary cancer is,

NOTE Confidence: 0.849544856

00:57:26.120 --> 00:57:27.600 why we do testing.

NOTE Confidence: 0.849544856

00:57:27.600 --> 00:57:29.080 What screening would change

NOTE Confidence: 0.849544856

00:57:29.080 --> 00:57:30.730 if someone was positive?

NOTE Confidence: 0.849544856

00:57:30.730 --> 00:57:32.332 You know why are we doing

NOTE Confidence: 0.849544856

00:57:32.332 --> 00:57:33.400 this testing at all?

NOTE Confidence: 0.849544856

00:57:33.400 --> 00:57:35.210 We talk about different testing

NOTE Confidence: 0.849544856

00:57:35.210 --> 00:57:37.020 options in terms of limited

NOTE Confidence: 0.849544856

00:57:37.091 --> 00:57:38.687 panels or expanded panels.

NOTE Confidence: 0.849544856

00:57:38.690 --> 00:57:39.340 There's research.

NOTE Confidence: 0.849544856

00:57:39.340 --> 00:57:41.615 Of course we coordinate the testing we

NOTE Confidence: 0.849544856

00:57:41.615 --> 00:57:43.706 call the patients with their results,

NOTE Confidence: 0.849544856

00:57:43.710 --> 00:57:44.676 explain their results,

NOTE Confidence: 0.849544856

00:57:44.676 --> 00:57:47.662 and then of do any follow up in terms

NOTE Confidence: 0.849544856

00:57:47.662 --> 00:57:49.666 of referrals to specialists and also

NOTE Confidence: 0.849544856

00:57:49.666 --> 00:57:52.110 just be a resource for the patient.

NOTE Confidence: 0.939351119

00:57:52.110 --> 00:57:52.844 Transfer information,

NOTE Confidence: 0.939351119

00:57:52.844 --> 00:57:55.780 so for what you can do to help,

NOTE Confidence: 0.939351119

00:57:55.780 --> 00:57:57.100 have your patients prepare

NOTE Confidence: 0.939351119

00:57:57.100 --> 00:57:58.750 probably the most important thing

NOTE Confidence: 0.939351119

00:57:58.750 --> 00:58:00.531 is to encourage their patients

NOTE Confidence: 0.939351119

00:58:00.531 --> 00:58:02.276 to collect their family history.

NOTE Confidence: 0.939351119

00:58:02.280 --> 00:58:04.098 So if you place a referral

NOTE Confidence: 0.939351119

00:58:04.098 --> 00:58:05.007 for cancer genetics,

NOTE Confidence: 0.939351119

00:58:05.010 --> 00:58:06.684 the most helpful thing is for

NOTE Confidence: 0.939351119

00:58:06.684 --> 00:58:08.754 that patient to be talking to

NOTE Confidence: 0.939351119

00:58:08.754 --> 00:58:10.538 relatives getting that information,

NOTE Confidence: 0.939351119

00:58:10.540 --> 00:58:12.790 particularly if there's no mutation,

NOTE Confidence: 0.939351119

00:58:12.790 --> 00:58:14.854 it's very important for to have

NOTE Confidence: 0.939351119

00:58:14.854 --> 00:58:16.230 that confirmed with records.

NOTE Confidence: 0.939351119

00:58:16.230 --> 00:58:18.631 It actually can be impact the accuracy

NOTE Confidence: 0.939351119

00:58:18.631 --> 00:58:20.480 of the patient's own testing,
NOTE Confidence: 0.939351119

00:58:20.480 --> 00:58:22.718 which some patients may not understand.
NOTE Confidence: 0.939351119

00:58:22.720 --> 00:58:25.372 There can be differences in laboratories
NOTE Confidence: 0.939351119

00:58:25.372 --> 00:58:27.140 and detecting specific variants,
NOTE Confidence: 0.939351119

00:58:27.140 --> 00:58:29.534 so we cannot confirm what lab or
NOTE Confidence: 0.939351119

00:58:29.534 --> 00:58:31.314 what specific variant or relative
NOTE Confidence: 0.939351119

00:58:31.314 --> 00:58:33.024 had it's possible the patient
NOTE Confidence: 0.939351119

00:58:33.024 --> 00:58:34.955 could get an uninformative or
NOTE Confidence: 0.939351119

00:58:34.955 --> 00:58:37.040 even a false negative result,
NOTE Confidence: 0.939351119

00:58:37.040 --> 00:58:38.684 so anything any medical
NOTE Confidence: 0.939351119

00:58:38.684 --> 00:58:39.917 records from relatives,
NOTE Confidence: 0.939351119

00:58:39.920 --> 00:58:42.030 any information is very helpful.
NOTE Confidence: 0.852921282

00:58:44.540 --> 00:58:45.960 Which I just you know,
NOTE Confidence: 0.852921282

00:58:45.960 --> 00:58:49.216 bring those milk records visit to the visits.
NOTE Confidence: 0.852921282

00:58:49.220 --> 00:58:52.783 And. And again, encourage them to get
NOTE Confidence: 0.852921282

00:58:52.783 --> 00:58:56.909 copies of any any genetic test results.

NOTE Confidence: 0.852921282

00:58:56.910 --> 00:59:00.417 So after the referral, it takes about,

NOTE Confidence: 0.852921282

00:59:00.420 --> 00:59:03.052 UM, the initial jet counseling visit is

NOTE Confidence: 0.852921282

00:59:03.052 --> 00:59:06.300 60 minutes. They are sent a questionnaire.

NOTE Confidence: 0.852921282

00:59:06.300 --> 00:59:08.196 And the patient schedules an appointment.

NOTE Confidence: 0.852921282

00:59:08.200 --> 00:59:09.820 They do a very brief intake.

NOTE Confidence: 0.852921282

00:59:09.820 --> 00:59:11.910 Again, the initial genetic counseling

NOTE Confidence: 0.852921282

00:59:11.910 --> 00:59:14.000 visit takes about 60 minutes.

NOTE Confidence: 0.852921282

00:59:14.000 --> 00:59:15.932 We again go through that that whole

NOTE Confidence: 0.852921282

00:59:15.932 --> 00:59:17.849 session and the cases are reviewed.

NOTE Confidence: 0.852921282

00:59:17.850 --> 00:59:21.300 Our weekly cancer genetics and prevention

NOTE Confidence: 0.852921282

00:59:21.300 --> 00:59:24.246 program the just out of a meeting where

NOTE Confidence: 0.852921282

00:59:24.246 --> 00:59:26.918 we review all the cases altogether.

NOTE Confidence: 0.852921282

00:59:26.920 --> 00:59:27.754 Patients are disclosed.

NOTE Confidence: 0.852921282

00:59:27.754 --> 00:59:30.100 The results over the phone or in person.

NOTE Confidence: 0.852921282

00:59:30.100 --> 00:59:32.176 If the patient prefers and the

NOTE Confidence: 0.852921282

00:59:32.176 --> 00:59:34.858 patient does get a copy of their
NOTE Confidence: 0.852921282

00:59:34.858 --> 00:59:37.306 results and a detailed summary letter.
NOTE Confidence: 0.852921282

00:59:37.310 --> 00:59:40.370 So I guess I'll just end this by saying,
NOTE Confidence: 0.852921282

00:59:40.370 --> 00:59:42.680 uhm, you don't have to be the
NOTE Confidence: 0.852921282

00:59:42.680 --> 00:59:44.649 experts on her Ezra Cancer.
NOTE Confidence: 0.852921282

00:59:44.650 --> 00:59:46.526 We care for a resource for you.
NOTE Confidence: 0.852921282

00:59:46.530 --> 00:59:47.498 You're really, you know,
NOTE Confidence: 0.852921282

00:59:47.498 --> 00:59:48.224 the the front,
NOTE Confidence: 0.852921282

00:59:48.230 --> 00:59:50.350 the front line for these patients to really
NOTE Confidence: 0.852921282

00:59:50.350 --> 00:59:52.289 gauge their personal and family history.
NOTE Confidence: 0.852921282

00:59:52.290 --> 00:59:55.827 So I really just less last thing I'll say,
NOTE Confidence: 0.852921282

00:59:55.830 --> 00:59:59.127 and I think everyone for your attention
NOTE Confidence: 0.852921282

00:59:59.130 --> 01:00:00.630 and is a pleasure talking everyone.
NOTE Confidence: 0.852921282

01:00:00.630 --> 01:00:02.280 And I will take any questions.
NOTE Confidence: 0.77375266

01:00:07.420 --> 01:00:09.166 Hi Amy, thanks for a great
NOTE Confidence: 0.77375266

01:00:09.166 --> 01:00:10.650 talk that was really you.

NOTE Confidence: 0.77375266

01:00:10.650 --> 01:00:13.512 You managed to pack in a lot there so

NOTE Confidence: 0.77375266

01:00:13.512 --> 01:00:16.120 thank you so much I do just want to

NOTE Confidence: 0.77375266

01:00:16.120 --> 01:00:18.597 let all of the attendees know that the

NOTE Confidence: 0.77375266

01:00:18.597 --> 01:00:21.339 session is being recorded and a link to

NOTE Confidence: 0.77375266

01:00:21.339 --> 01:00:23.677 the recording will be on our website.

NOTE Confidence: 0.77375266

01:00:23.680 --> 01:00:25.888 So if you wanna go back and listen

NOTE Confidence: 0.77375266

01:00:25.888 --> 01:00:27.952 to parts of Amy's talk you will

NOTE Confidence: 0.77375266

01:00:27.952 --> 01:00:30.169 have access to that in the future.

NOTE Confidence: 0.77375266

01:00:30.170 --> 01:00:32.570 We also have a number of resources on

NOTE Confidence: 0.77375266

01:00:32.570 --> 01:00:34.633 our website including fact sheets that

NOTE Confidence: 0.77375266

01:00:34.633 --> 01:00:37.469 you and your patients may find helpful so.

NOTE Confidence: 0.77375266

01:00:37.470 --> 01:00:38.810 And encourage everyone to

NOTE Confidence: 0.77375266

01:00:38.810 --> 01:00:40.150 look at their website.

NOTE Confidence: 0.77375266

01:00:40.150 --> 01:00:41.670 And I believe that, UM,

NOTE Confidence: 0.77375266

01:00:41.670 --> 01:00:44.916 the website will be included and

NOTE Confidence: 0.77375266

01:00:44.916 --> 01:00:47.016 post conference communications soum.
NOTE Confidence: 0.77375266

01:00:47.016 --> 01:00:50.572 We'd like to encourage people to enter
NOTE Confidence: 0.77375266

01:00:50.572 --> 01:00:54.072 some questions for Amy in the Q&A and Amy,
NOTE Confidence: 0.77375266

01:00:54.072 --> 01:00:56.244 we do have a couple here,
NOTE Confidence: 0.77375266

01:00:56.250 --> 01:00:58.170 and so the first one,
NOTE Confidence: 0.77375266

01:00:58.170 --> 01:00:59.510 the first question we have,
NOTE Confidence: 0.77375266

01:00:59.510 --> 01:01:01.962 is whether Gardner's syndrome
NOTE Confidence: 0.77375266

01:01:01.962 --> 01:01:05.027 is something separate or whether
NOTE Confidence: 0.77375266

01:01:05.027 --> 01:01:07.838 that actually is attenuated.
NOTE Confidence: 0.77375266

01:01:07.840 --> 01:01:09.268 Uhm Polly process.
NOTE Confidence: 0.828105996428571

01:01:10.330 --> 01:01:11.584 Great, great question.
NOTE Confidence: 0.828105996428571

01:01:11.584 --> 01:01:14.092 So Gardner syndrome is I would
NOTE Confidence: 0.828105996428571

01:01:14.092 --> 01:01:16.498 say it's an outdated syndrome.
NOTE Confidence: 0.828105996428571

01:01:16.500 --> 01:01:20.388 It was mainly used to describe
NOTE Confidence: 0.828105996428571

01:01:20.388 --> 01:01:22.980 if someone had specific.
NOTE Confidence: 0.828105996428571

01:01:22.980 --> 01:01:25.560 Less common features that we

NOTE Confidence: 0.828105996428571
01:01:25.560 --> 01:01:28.140 would see with classic faps,
NOTE Confidence: 0.828105996428571
01:01:28.140 --> 01:01:30.432 but we know now that people
NOTE Confidence: 0.828105996428571
01:01:30.432 --> 01:01:32.455 with Gardner syndrome do have
NOTE Confidence: 0.828105996428571
01:01:32.455 --> 01:01:34.535 pathogenic mutations in a PC,
NOTE Confidence: 0.828105996428571
01:01:34.540 --> 01:01:37.516 so we really just prefer to
NOTE Confidence: 0.828105996428571
01:01:37.516 --> 01:01:41.470 call it faps or attenuated faps.
NOTE Confidence: 0.828105996428571
01:01:41.470 --> 01:01:43.374 So it's not so I would say
NOTE Confidence: 0.828105996428571
01:01:43.374 --> 01:01:45.210 the answer to your question.
NOTE Confidence: 0.828105996428571
01:01:45.210 --> 01:01:46.860 They they are the same.
NOTE Confidence: 0.828105996428571
01:01:46.860 --> 01:01:49.667 They just said Gardner syndrome was house,
NOTE Confidence: 0.828105996428571
01:01:49.670 --> 01:01:52.780 certain phenotypes were were described,
NOTE Confidence: 0.828105996428571
01:01:52.780 --> 01:01:54.935 particularly some of the less
NOTE Confidence: 0.828105996428571
01:01:54.935 --> 01:01:57.647 common features of of faps like
NOTE Confidence: 0.828105996428571
01:01:57.647 --> 01:02:00.032 extra teeth and things like
NOTE Confidence: 0.828105996428571
01:02:00.032 --> 01:02:01.940 that oftentimes was described
NOTE Confidence: 0.828105996428571

01:02:02.022 --> 01:02:04.407 with the four Gardner syndrome,
NOTE Confidence: 0.828105996428571

01:02:04.410 --> 01:02:07.416 but nowadays it is packaged together.
NOTE Confidence: 0.828105996428571

01:02:07.420 --> 01:02:09.160 But that is an important point
NOTE Confidence: 0.828105996428571

01:02:09.160 --> 01:02:11.006 that some people still use some
NOTE Confidence: 0.828105996428571

01:02:11.006 --> 01:02:12.250 of the older terminology.
NOTE Confidence: 0.828105996428571

01:02:12.250 --> 01:02:13.246 For Gardner syndrome.
NOTE Confidence: 0.828105996428571

01:02:13.246 --> 01:02:15.570 So if someone does report a family
NOTE Confidence: 0.828105996428571

01:02:15.636 --> 01:02:17.300 history of Gardner syndrome,
NOTE Confidence: 0.828105996428571

01:02:17.300 --> 01:02:18.560 even though that's may not be
NOTE Confidence: 0.828105996428571

01:02:18.560 --> 01:02:19.940 a term we use currently,
NOTE Confidence: 0.828105996428571

01:02:19.940 --> 01:02:21.476 that would also warrant a referral.
NOTE Confidence: 0.933836863

01:02:25.980 --> 01:02:27.860 And the next question we
NOTE Confidence: 0.933836863

01:02:27.860 --> 01:02:29.740 have is an interesting one.
NOTE Confidence: 0.933836863

01:02:29.740 --> 01:02:32.255 There's an attendee who's wondering
NOTE Confidence: 0.933836863

01:02:32.255 --> 01:02:35.690 if we think the ages at the time
NOTE Confidence: 0.933836863

01:02:35.690 --> 01:02:37.610 of diagnosis are getting younger

NOTE Confidence: 0.933836863
01:02:37.685 --> 01:02:39.569 in subsequent generations.
NOTE Confidence: 0.933836863
01:02:39.570 --> 01:02:40.923 So, for example,
NOTE Confidence: 0.933836863
01:02:40.923 --> 01:02:43.178 if someone's mother had breast
NOTE Confidence: 0.933836863
01:02:43.178 --> 01:02:46.710 cancer at 55 with a BRCA mutation,
NOTE Confidence: 0.933836863
01:02:46.710 --> 01:02:48.640 are we seeing her daughter
NOTE Confidence: 0.933836863
01:02:48.640 --> 01:02:51.065 who has a mutation having her
NOTE Confidence: 0.933836863
01:02:51.065 --> 01:02:53.165 breast cancer earlier in life?
NOTE Confidence: 0.889521676666667
01:02:54.470 --> 01:02:58.268 That is, that's a great question.
NOTE Confidence: 0.889521676666667
01:02:58.270 --> 01:03:01.663 As with with BRCA one and BRCA 2 this
NOTE Confidence: 0.889521676666667
01:03:01.663 --> 01:03:05.753 and we we often call this in genetics
NOTE Confidence: 0.889521676666667
01:03:05.753 --> 01:03:08.462 anticipation where the phenotype appears
NOTE Confidence: 0.889521676666667
01:03:08.462 --> 01:03:12.734 to present at a younger and younger age.
NOTE Confidence: 0.889521676666667
01:03:12.740 --> 01:03:15.710 There's no known evidence of
NOTE Confidence: 0.889521676666667
01:03:15.710 --> 01:03:19.520 anticipation in BRCA one and BRCA 2.
NOTE Confidence: 0.889521676666667
01:03:19.520 --> 01:03:22.439 It's it's it's. It's hard to say,
NOTE Confidence: 0.889521676666667

01:03:22.440 --> 01:03:24.456 but there can be what we
NOTE Confidence: 0.889521676666667

01:03:24.456 --> 01:03:25.464 call variable expressivity,
NOTE Confidence: 0.889521676666667

01:03:25.470 --> 01:03:26.875 meaning that someone could have
NOTE Confidence: 0.889521676666667

01:03:26.875 --> 01:03:28.280 the same mutation and beers
NOTE Confidence: 0.889521676666667

01:03:28.332 --> 01:03:29.616 they want to be receipt to,
NOTE Confidence: 0.889521676666667

01:03:29.620 --> 01:03:31.792 but they will not always develop
NOTE Confidence: 0.889521676666667

01:03:31.792 --> 01:03:33.676 breast cancer at the same
NOTE Confidence: 0.889521676666667

01:03:33.676 --> 01:03:35.496 age or even develop breast.
NOTE Confidence: 0.889521676666667

01:03:35.500 --> 01:03:37.225 You know someone made available
NOTE Confidence: 0.889521676666667

01:03:37.225 --> 01:03:39.420 varying cancer or or breast cancer,
NOTE Confidence: 0.889521676666667

01:03:39.420 --> 01:03:41.200 which we call variable expressivity.
NOTE Confidence: 0.889521676666667

01:03:41.200 --> 01:03:43.328 So that might explain why in some
NOTE Confidence: 0.889521676666667

01:03:43.328 --> 01:03:45.340 families it does appear that Amma,
NOTE Confidence: 0.889521676666667

01:03:45.340 --> 01:03:47.545 a mother may have breast cancer at a later
NOTE Confidence: 0.889521676666667

01:03:47.545 --> 01:03:49.915 age and her daughter has breast cancer and.
NOTE Confidence: 0.889521676666667

01:03:49.920 --> 01:03:50.824 Earlier age,

NOTE Confidence: 0.889521676666667
01:03:50.824 --> 01:03:53.536 not because of something like anticipation,
NOTE Confidence: 0.889521676666667
01:03:53.540 --> 01:03:56.996 but possibly do that variable expressivity.
NOTE Confidence: 0.889521676666667
01:03:57.000 --> 01:03:57.777 Although I do,
NOTE Confidence: 0.889521676666667
01:03:57.777 --> 01:04:00.300 I do know that there was some older data.
NOTE Confidence: 0.889521676666667
01:04:00.300 --> 01:04:01.884 I think Claire you mentioned this
NOTE Confidence: 0.889521676666667
01:04:01.884 --> 01:04:04.511 to me a few years ago where there
NOTE Confidence: 0.889521676666667
01:04:04.511 --> 01:04:06.391 is some data suggesting maybe
NOTE Confidence: 0.889521676666667
01:04:06.391 --> 01:04:08.337 anticipation with Lee from Mini
NOTE Confidence: 0.889521676666667
01:04:08.337 --> 01:04:09.841 syndrome where individuals actually
NOTE Confidence: 0.889521676666667
01:04:09.841 --> 01:04:12.440 appear to develop Lee from many
NOTE Confidence: 0.889521676666667
01:04:12.440 --> 01:04:13.820 syndrome associated cancers,
NOTE Confidence: 0.889521676666667
01:04:13.820 --> 01:04:16.830 particularly breast cancer at younger
NOTE Confidence: 0.889521676666667
01:04:16.830 --> 01:04:19.583 age and subsequent generations I.
NOTE Confidence: 0.889521676666667
01:04:19.583 --> 01:04:21.984 In terms of additional research on that,
NOTE Confidence: 0.889521676666667
01:04:21.990 --> 01:04:25.246 I have not seen anything recent about that,
NOTE Confidence: 0.889521676666667

01:04:25.250 --> 01:04:27.679 but I know there was some hypothesis
NOTE Confidence: 0.889521676666667

01:04:27.679 --> 01:04:29.613 that leaf armenie syndrome may
NOTE Confidence: 0.889521676666667

01:04:29.613 --> 01:04:31.678 have some level of anticipation.
NOTE Confidence: 0.889521676666667

01:04:31.680 --> 01:04:33.528 But in terms of other hereditary
NOTE Confidence: 0.889521676666667

01:04:33.528 --> 01:04:34.452 breast cancer syndromes,
NOTE Confidence: 0.889521676666667

01:04:34.460 --> 01:04:36.665 there's none that we know of yet.
NOTE Confidence: 0.890462226923077

01:04:39.910 --> 01:04:42.158 Yeah, and I would just add that even
NOTE Confidence: 0.890462226923077

01:04:42.158 --> 01:04:44.458 with these hereditary cancer syndromes,
NOTE Confidence: 0.890462226923077

01:04:44.460 --> 01:04:46.494 the mutation is not the only
NOTE Confidence: 0.890462226923077

01:04:46.494 --> 01:04:48.210 contributing factor to cancer risk.
NOTE Confidence: 0.890462226923077

01:04:48.210 --> 01:04:48.976 Lifestyle environment.
NOTE Confidence: 0.890462226923077

01:04:48.976 --> 01:04:51.274 All of those things still play
NOTE Confidence: 0.890462226923077

01:04:51.274 --> 01:04:53.520 a role in cancer development,
NOTE Confidence: 0.890462226923077

01:04:53.520 --> 01:04:55.680 and so it's possible that some
NOTE Confidence: 0.890462226923077

01:04:55.680 --> 01:04:57.648 of those earlier cancer diagnosis
NOTE Confidence: 0.890462226923077

01:04:57.648 --> 01:04:59.464 that you're you're mentioning

NOTE Confidence: 0.890462226923077

01:04:59.464 --> 01:05:01.734 are related to other factors

NOTE Confidence: 0.890462226923077

01:05:01.803 --> 01:05:03.938 outside of the genetic mutation,

NOTE Confidence: 0.890462226923077

01:05:03.940 --> 01:05:06.768 or in addition to the genetic mutation.

NOTE Confidence: 0.890462226923077

01:05:06.770 --> 01:05:08.513 And I think we still have a

NOTE Confidence: 0.890462226923077

01:05:08.513 --> 01:05:10.380 lot to learn about hereditary.

NOTE Confidence: 0.890462226923077

01:05:10.380 --> 01:05:12.158 Cancer genetics we've come a long way.

NOTE Confidence: 0.890462226923077

01:05:12.160 --> 01:05:14.128 We know a lot and they will learn

NOTE Confidence: 0.890462226923077

01:05:14.128 --> 01:05:16.242 more and perhaps some of these

NOTE Confidence: 0.890462226923077

01:05:16.242 --> 01:05:17.798 hereditary cancer syndromes do,

NOTE Confidence: 0.890462226923077

01:05:17.800 --> 01:05:19.052 in fact have anticipation.

NOTE Confidence: 0.890462226923077

01:05:19.052 --> 01:05:20.930 And we just don't know yet.

NOTE Confidence: 0.859760127

01:05:25.000 --> 01:05:26.430 Those are all the questions

NOTE Confidence: 0.859760127

01:05:26.430 --> 01:05:28.152 that were entered into the Q&A,

NOTE Confidence: 0.859760127

01:05:28.152 --> 01:05:30.488 so I don't know if any of our

NOTE Confidence: 0.859760127

01:05:30.488 --> 01:05:32.416 panelists have any questions or if

NOTE Confidence: 0.859760127

01:05:32.416 --> 01:05:35.047 we want to give people just a minute

NOTE Confidence: 0.859760127

01:05:35.047 --> 01:05:37.371 in case any other questions come up.

NOTE Confidence: 0.717009476153846

01:05:39.890 --> 01:05:42.410 Maybe while waiting I'll post a

NOTE Confidence: 0.717009476153846

01:05:42.410 --> 01:05:45.199 question Amy that was a great talk.

NOTE Confidence: 0.717009476153846

01:05:45.200 --> 01:05:46.022 So quick question.

NOTE Confidence: 0.717009476153846

01:05:46.022 --> 01:05:47.940 You just you went over very nicely

NOTE Confidence: 0.717009476153846

01:05:47.996 --> 01:05:49.546 about what happened with Brad.

NOTE Confidence: 0.717009476153846

01:05:49.550 --> 01:05:51.850 Can white people got retested

NOTE Confidence: 0.717009476153846

01:05:51.850 --> 01:05:53.807 then and then towards the end?

NOTE Confidence: 0.717009476153846

01:05:53.807 --> 01:05:55.121 You also discuss a little bit

NOTE Confidence: 0.717009476153846

01:05:55.121 --> 01:05:56.368 more that there may be some.

NOTE Confidence: 0.717009476153846

01:05:56.370 --> 01:05:58.944 Uh, in the future,

NOTE Confidence: 0.717009476153846

01:05:58.944 --> 01:06:00.452 opportunities for bettering the

NOTE Confidence: 0.717009476153846

01:06:00.452 --> 01:06:02.469 testing and actually being able to

NOTE Confidence: 0.717009476153846

01:06:02.469 --> 01:06:04.782 detect a few more number 4 so when

NOTE Confidence: 0.717009476153846

01:06:04.782 --> 01:06:07.423 do you think people will will have

NOTE Confidence: 0.717009476153846
01:06:07.423 --> 01:06:10.057 to consider re sending the patient?
NOTE Confidence: 0.717009476153846
01:06:10.060 --> 01:06:11.880 Who previously had tested negative.
NOTE Confidence: 0.717009476153846
01:06:11.880 --> 01:06:13.280 I'm talking in general terms.
NOTE Confidence: 0.717009476153846
01:06:13.280 --> 01:06:16.336 What do you think would be a general
NOTE Confidence: 0.717009476153846
01:06:16.336 --> 01:06:18.090 recommendation that we could follow
NOTE Confidence: 0.717009476153846
01:06:18.090 --> 01:06:20.239 in terms of in terms of that?
NOTE Confidence: 0.717009476153846
01:06:20.240 --> 01:06:21.500 That's a, that's a.
NOTE Confidence: 0.717009476153846
01:06:21.500 --> 01:06:22.760 That's a great question,
NOTE Confidence: 0.717009476153846
01:06:22.760 --> 01:06:24.936 and it's something that patients also ask me.
NOTE Confidence: 0.717009476153846
01:06:24.940 --> 01:06:26.625 Even now they they say, well,
NOTE Confidence: 0.717009476153846
01:06:26.625 --> 01:06:28.250 when should I call you?
NOTE Confidence: 0.717009476153846
01:06:28.250 --> 01:06:31.208 You know, for more testing, I think it's.
NOTE Confidence: 0.717009476153846
01:06:31.208 --> 01:06:34.484 It's hard now that testing has gone so
NOTE Confidence: 0.717009476153846
01:06:34.484 --> 01:06:36.510 comprehensive, I would say so quickly.
NOTE Confidence: 0.717009476153846
01:06:36.510 --> 01:06:38.310 Even the last five years.
NOTE Confidence: 0.717009476153846

01:06:38.310 --> 01:06:39.470 What I've been saying.
NOTE Confidence: 0.717009476153846

01:06:39.470 --> 01:06:41.210 What I was saying to patients,
NOTE Confidence: 0.717009476153846

01:06:41.210 --> 01:06:42.470 and this is,
NOTE Confidence: 0.717009476153846

01:06:42.470 --> 01:06:43.310 but again,
NOTE Confidence: 0.717009476153846

01:06:43.310 --> 01:06:44.480 this is not something that's
NOTE Confidence: 0.717009476153846

01:06:44.480 --> 01:06:45.650 really hard and fast rule.
NOTE Confidence: 0.717009476153846

01:06:45.650 --> 01:06:48.434 I would say at least five
NOTE Confidence: 0.717009476153846

01:06:48.434 --> 01:06:49.971 years to check back,
NOTE Confidence: 0.717009476153846

01:06:49.971 --> 01:06:52.470 unless there was a change in someone's
NOTE Confidence: 0.717009476153846

01:06:52.541 --> 01:06:55.439 family history like a new cancer diagnosis,
NOTE Confidence: 0.717009476153846

01:06:55.440 --> 01:06:57.444 that possibly could mean that other
NOTE Confidence: 0.717009476153846

01:06:57.444 --> 01:06:59.205 genes related to that diagnosis
NOTE Confidence: 0.717009476153846

01:06:59.205 --> 01:07:01.365 were not included in the testing.
NOTE Confidence: 0.717009476153846

01:07:01.370 --> 01:07:03.030 But if there's no changes.
NOTE Confidence: 0.717009476153846

01:07:03.030 --> 01:07:05.340 Someone's personal or family history.
NOTE Confidence: 0.717009476153846

01:07:05.340 --> 01:07:07.716 I would say every five years

NOTE Confidence: 0.717009476153846
01:07:07.720 --> 01:07:09.460 is a good place to check,
NOTE Confidence: 0.717009476153846
01:07:09.460 --> 01:07:11.000 and it's possible even in
NOTE Confidence: 0.717009476153846
01:07:11.000 --> 01:07:12.232 five years from now.
NOTE Confidence: 0.717009476153846
01:07:12.240 --> 01:07:15.760 Maybe it's it might not change as much,
NOTE Confidence: 0.717009476153846
01:07:15.760 --> 01:07:19.002 but I can't imagine the testing jumping
NOTE Confidence: 0.717009476153846
01:07:19.002 --> 01:07:22.376 to such a huge amount of clinical
NOTE Confidence: 0.717009476153846
01:07:22.376 --> 01:07:25.200 significance in less than five years.
NOTE Confidence: 0.717009476153846
01:07:25.200 --> 01:07:26.820 I don't know Clearview any other
NOTE Confidence: 0.717009476153846
01:07:26.820 --> 01:07:28.530 thoughts on that in particular?
NOTE Confidence: 0.879846068
01:07:29.970 --> 01:07:31.020 No, I agree. I mean,
NOTE Confidence: 0.879846068
01:07:31.020 --> 01:07:33.216 I think the testing technology doesn't.
NOTE Confidence: 0.879846068
01:07:33.220 --> 01:07:34.324 It changes quickly,
NOTE Confidence: 0.879846068
01:07:34.324 --> 01:07:35.796 but not that quickly.
NOTE Confidence: 0.879846068
01:07:35.800 --> 01:07:38.726 So five years is probably a good.
NOTE Confidence: 0.879846068
01:07:38.730 --> 01:07:41.114 An estimate, but I would also just reiterate
NOTE Confidence: 0.879846068

01:07:41.114 --> 01:07:43.200 one other thing that you said there.
NOTE Confidence: 0.879846068

01:07:43.200 --> 01:07:45.664 If the personal or family history changes,
NOTE Confidence: 0.879846068

01:07:45.670 --> 01:07:47.266 that's another really important
NOTE Confidence: 0.879846068

01:07:47.266 --> 01:07:49.261 touch point for patients to
NOTE Confidence: 0.879846068

01:07:49.261 --> 01:07:51.248 get back in contact with us,
NOTE Confidence: 0.879846068

01:07:51.250 --> 01:07:54.598 because that could certainly change things,
NOTE Confidence: 0.879846068

01:07:54.600 --> 01:07:57.000 so those are good time point
NOTE Confidence: 0.879846068

01:07:57.000 --> 01:07:58.600 to consider re evaluation.
NOTE Confidence: 0.911991621428571

01:08:02.180 --> 01:08:04.560 Right, I think that's it for question.
NOTE Confidence: 0.911991621428571

01:08:04.560 --> 01:08:06.975 So Amy, thanks again for your talk
NOTE Confidence: 0.911991621428571

01:08:06.975 --> 01:08:09.233 and we hope that everyone will
NOTE Confidence: 0.911991621428571

01:08:09.233 --> 01:08:11.907 join us next week for the second
NOTE Confidence: 0.911991621428571

01:08:11.989 --> 01:08:14.089 half of this seminar series.
NOTE Confidence: 0.949164791428571

01:08:15.530 --> 01:08:17.378 Thank you everyone. Have a good night.