WEBVTT

1
00:00:00.000 --> 00:00:01.700
Mikaela Miller: So you're gonna hear a little bit

2
00:00:02.450 --> 00:00:05.600
Mikaela Miller: Little announcement. Perfect. Thank you, Stephanie.

3
00:00:06.760 --> 00:00:11.980
Mikaela Miller: Alright, I'm going to let Shelly read the acknowledgement for today.

4
00:00:13.980 --> 00:00:27.140
Shelly Eagen: So, this is something that we have done over the last several meetings, and will continue to do at each meeting, and we want to thank the families, the caregivers, the committee members, as well as advocates for their contributions to the MT

5
00:00:27.340 --> 00:00:47.700
Shelly Eagen: to the Montana Newborn Screening Program. We recognize that each condition reviewed does affect children and families of Montana. We strive to balance the emotions as well as the vulnerabilities shared with the need for careful and sometimes difficult discussions on logistics as well as finances.

6
00:00:47.700 --> 00:00:55.120
Shelly Eagen: Our goal is to ensure that the process is publicly accessible, transparent, as well as carefully examined.

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00:01:00.530 --> 00:01:01.849
Mikaela Miller: Thank you, Shelly.

8
00:01:04.080 --> 00:01:23.079
Mikaela Miller: Okay, as usual, the goal is to try not to use any form of acronyms if we can so help it. But one acronym you may be hearing slip through today is Metachromatic leukodystrophy, which is also known as MLD, and that is the condition that we will be discussing today.

9
00:01:26.870 --> 00:01:36.170
Mikaela Miller: Quick look at the agenda. So we've completed the welcome roll call. We're gonna do just a few minutes of a lab presentation, so we're gonna have

10
00:01:36.200 --> 00:01:44.900
Mikaela Miller: Jeanne let us know some information she received from Mae Baker over the period of time since our last meeting.

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00:01:44.900 --> 00:01:59.580
Mikaela Miller: And then we're going to give you some more time to have that discussion, about 45 minutes or so, and then we've got a public comment period, as usual. We'll always have that 10-minute period at the end of our meetings.

12
00:01:59.660 --> 00:02:12.970
Mikaela Miller: And then we're going to go through some brief next steps, and then close out the meeting. So we have it ending today at 11.30, but of course it's always flexible, because we're really here just to give you some time to have that discussion, so…

13
00:02:13.270 --> 00:02:16.039
Mikaela Miller: We'll make sure we have enough time for that.

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00:02:18.150 --> 00:02:32.460
Mikaela Miller: Just to review the public comment period rules at the beginning here, this is reserved for members of the public, not the committee members, so we try to reserve that time, if we have anyone hop on by the end of the meeting.

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00:02:32.700 --> 00:02:45.580
Mikaela Miller: We do request that they raise their hands, and then unmute themselves, and then it's a 2-minute maximum per comment, and then any additional commentary can be emailed up to an hour after the meeting ends.

16
00:02:49.370 --> 00:03:08.359
Mikaela Miller: Okay, so as always, just some brief ground rules for the meeting. So, please just keep yourself muted when you're not talking, just helps with some background noise. You can use the chat during the meeting. If you have any questions or run into any technical issues, you can chat or email Stephanie or I.

17
00:03:08.360 --> 00:03:16.679
Mikaela Miller: Any clarifying questions, please feel free to ask. Now is a great time to do so. Since this meeting is

18
00:03:16.690 --> 00:03:25.849
Mikaela Miller: time-bound. I will interject if need be, if we kind of, run into any time constraints, but hopefully we'll have plenty today.

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00:03:25.870 --> 00:03:39.669
Mikaela Miller: Next steps or action items, we'll try to keep track of all of those. We will be taking notes in the background that will get sent out after the meeting in order to ensure equity of voice and engagement.

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00:03:39.760 --> 00:03:49.139
Mikaela Miller: We may call on certain individuals to ask questions. This also, just to note, is a safe meeting space.

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00:03:49.200 --> 00:04:02.790
Mikaela Miller: So if you don't feel comfortable sharing out loud, please just let me or Stephanie, and I know in a separate chat, and we can either communicate with you in another way after the meeting, or we can, share your question for you.

22
00:04:04.740 --> 00:04:18.659

Mikaela Miller: Okay, hopefully those all seemed fairly familiar. We do review those at each meeting, but I'm gonna go ahead and roll into the lab updates next, and I'm going to pass it off to Jeannie.

23
00:04:21.120 --> 00:04:22.640
Jeanne Lee: Thanks, Mikaela.

24
00:04:22.930 --> 00:04:41.100
Jeanne Lee: So I… I just have just a small update for you guys today. I did have a chat with Mae Baker, and she is developing an assay for MLD, and she hopes to have it ready at the end of this year.

25
00:04:41.450 --> 00:04:45.350
Jeanne Lee: And when we talked about…

26
00:04:45.520 --> 00:05:05.370
Jeanne Lee: her assay. She is hoping to, multiplex it with XALD, but I'm not sure if that was, finalized or not yet, but, if she is able to multiplex it,

27
00:05:05.630 --> 00:05:23.040
Jeanne Lee: that, as you recall, makes it pretty easy for testing, because it just takes one punch out of the blood spot, and they can run… would be able to run both the XALD test and the MLD test. So…

28
00:05:23.120 --> 00:05:40.289
Jeanne Lee: It was still up in the air whether it'd be multiplexed or not, but, and then when she, gave us a price, she said probably up to $3, per screen. And so…

29
00:05:40.680 --> 00:05:48.639
Jeanne Lee: You know, it might be a little bit cheaper if she's able to multiplex it, but I… I don't think it'll be…

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00:05:49.000 --> 00:05:52.329
Jeanne Lee: Any more expensive than $3 per screen.

31
00:05:53.770 --> 00:05:58.659
Jeanne Lee: I think that… I think that's all I have.

32
00:06:06.410 --> 00:06:08.150
Mikaela Miller: Great, thank you, Jeannie.

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00:06:08.500 --> 00:06:18.319
Mikaela Miller: I'm gonna go ahead and just go straight into the discussion, so if anyone has any questions, we can go ahead and get started, and I'm gonna let Shelly lead that.

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00:06:19.800 --> 00:06:23.079
Shelly Eagen: Yeah, and I know that, as part of

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00:06:23.200 --> 00:06:32.359
Shelly Eagen: the poll for this, discussion and this meeting as well. There was an opportunity to put questions in ahead of time. I think

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00:06:32.580 --> 00:06:50.250
Shelly Eagen: probably a lot of the questions was what Jeanne just answered for us, is kind of where are things at with Wisconsin, and what kind of… maybe what other states that are already implementing this are doing. But I want to open it up at this point to committee members for further discussion or questions.

37
00:06:55.090 --> 00:06:56.360
Shelly Eagen: Dr. Elias.

38
00:06:57.300 --> 00:07:17.010
Abe Elias: Yeah, yeah, thanks, Ginny, for the update. One question I have there, I did Mae, mention, so, what there… so, whether or not that price and the validation that she's working right now, or development, I guess, at this point that she's working right now, is, includes both the

39
00:07:17.050 --> 00:07:22.260
Abe Elias: Initial, the first tier, you know, which is usually the, the,

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00:07:22.860 --> 00:07:31.949
Abe Elias: the sulfatide, detection, and then second here, if it's positive, enzyme, enzyme testing. Did she mention that?

41
00:07:35.940 --> 00:07:39.639
Jeanne Lee: You know, Dr. Elias, I don't have the answer to that.

42
00:07:40.680 --> 00:07:57.450
Jeanne Lee: I'm… I'm assuming that she is, just working on the first tier right now, but, I… I can ask her, if she has a second tier assay.

43
00:07:57.610 --> 00:08:00.970
Jeanne Lee: for the sulfatides.

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00:08:03.140 --> 00:08:09.800
Abe Elias: Yeah, that might be good. You know, it's interesting, because for MLD, the… Think…

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00:08:10.010 --> 00:08:25.029
Abe Elias: We are kind of in a similar situation in many other states and labs currently. They're, you know, they're thinking about it, there are some of the, it's, you know, it is obviously on the rust, and it's, with the FDA approval of the treatment.

46
00:08:25.030 --> 00:08:47.829
Abe Elias: I think there has been… become a new push, but a lot of labs are still working on that development, because the first tier is really a, is a mass spec analysis that we do a lot of the other metabolic

analytes, of course, but it's a little bit more difficult because it's sulfatide, so developing that is one thing. And then, I think the

47
00:08:47.830 --> 00:08:51.179
Abe Elias: Most labs that, in most…

48
00:08:51.190 --> 00:09:02.509
Abe Elias: places that are developing. There's a second tier to develop to reduce the false positives and to increase the specificity, and I think that is commonly

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00:09:02.690 --> 00:09:08.680
Abe Elias: The… the NDAM activity, so it would be… would be nice to know.

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00:09:08.800 --> 00:09:25.649
Abe Elias: Maybe we know where she is there, or where the lab is, and also from a price perspective. Also, whether or not this can be done on the same… whether or not they would need a second spot there, or this could also be done on the same one.

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00:09:27.630 --> 00:09:35.760
Jeanne Lee: I do know that, she, I… I was hoping that she'd be able to join the call today to…

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00:09:35.880 --> 00:09:51.640
Jeanne Lee: to give us a little bit more information about her assay, but she wasn't able to, join today. She was at a meeting. So, so we can get clarification on that, and,

53
00:09:51.640 --> 00:10:05.390
Jeanne Lee: And if… if possible, perhaps we can even have her, pop in on our October meeting, or whenever our meeting is this fall, before voting, if… if we…

54
00:10:05.390 --> 00:10:07.369
Jeanne Lee: If we need that.

55
00:10:10.840 --> 00:10:12.320
Abe Elias: Thank you, Jeanne.

56
00:10:17.190 --> 00:10:20.079
Shelly Eagen: Other questions or comments?

57
00:10:38.230 --> 00:10:39.110
Shelly Eagen: Bridget?

58
00:10:40.200 --> 00:10:56.320
Bridget Evans: I had a question about the administrative rules of Montana portion. So if the testing, say, is ready at the end of the year, do we still have to wait for babies to start the testing if it's… if we, you know, implement this, until the arms are completely done?

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00:10:57.210 --> 00:11:04.420
Jeanne Lee: Yes. So, unfortunately, that's kind of part of our long process of…

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00:11:04.460 --> 00:11:22.959
Jeanne Lee: getting conditions added to the rule. So since we'll be looking at, a legislative session, in January 2027, unfortunately, that even makes it a little bit longer for us, because they put us on hold

61
00:11:23.100 --> 00:11:27.420
Jeanne Lee: For, you know, starting new,

62
00:11:29.040 --> 00:11:39.210
Jeanne Lee: you know, amending the administrative rules 3 months before the legislative session. So, we will have to wait until

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00:11:39.300 --> 00:11:50.810
Jeanne Lee: after the legislative session, or halfway through, I can't remember which… which way it was, if it's, if we get to start in April, or if we have to wait till May.

64
00:11:51.270 --> 00:11:53.040
Jeanne Lee: But,

65
00:11:53.190 --> 00:12:11.589
Jeanne Lee: when we start the administrative rule, then if MLD is added to the panel, we'll have to wait till then to add it to the rule, and then that even, Bridget takes a little bit of time. I think, you know, probably, you know, up to 9 months.

66
00:12:11.590 --> 00:12:14.109
Jeanne Lee: To get it on the,

67
00:12:14.170 --> 00:12:19.850
Jeanne Lee: Administrative role to go through that process before we'll be able to start screening.

68
00:12:20.050 --> 00:12:29.189
Jeanne Lee: So, I… I would say, if the committee votes to add MLD, we'd probably be looking at the end

69
00:12:29.420 --> 00:12:34.890
Jeanne Lee: Of, 2027, before we can start screening.

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00:12:35.060 --> 00:12:43.019
Jeanne Lee: Or, early… January 2028, so…

71
00:12:43.310 --> 00:12:45.759
Jeanne Lee: And, you know, that's kind of…

72
00:12:46.600 --> 00:12:59.899
Jeanne Lee: what we're in right now with ASMD is that it just takes some time to get through that administrative rule before we can start screening. So, you know, we are looking at January

73
00:13:00.100 --> 00:13:01.860
Jeanne Lee: 2027.

74
00:13:02.100 --> 00:13:04.460
Jeanne Lee: to start screening ASMD.

75
00:13:14.390 --> 00:13:19.410
Shelly Eagen: Thank you for that clarification. Additional questions, comments?

76
00:13:21.180 --> 00:13:37.149
Lynne Wood: I apologize if this was discussed already, since I had to jump off of the call for a second to answer a page. I think I recall from our last meeting that if a child screens positive, the result will go to…

77
00:13:37.330 --> 00:13:41.410
Lynne Wood: Chodair, and then they'll get referred out to get, like, a brain MRI and things like that.

78
00:13:47.900 --> 00:13:52.290
Abe Elias: I can just comment on it. So, you know, I think the, the,

79
00:13:52.720 --> 00:14:09.210
Abe Elias: The screening itself, includes the… typically, so the kind of the recommendations for screening, they have come out, in… in part, to a big part, from a large, European pilot, I think just

80
00:14:09.950 --> 00:14:11.549
Abe Elias: A couple of years ago.

81
00:14:11.660 --> 00:14:28.959
Abe Elias: Or less, and they, they, recommended to, at least as a minimum for screening, to, to, to do, the mass spec for, for the, sulfatides, and then, to confirm that, with,

82
00:14:29.140 --> 00:14:48.409
Abe Elias: with, enzyme testing. Some… some labs, I think they're thinking about doing sequencing right away. That's another issue. Part of it is, pseudodeficiency alleles, which are quite common. But then also, because there's such a well-established genotype, phenotype,

83
00:14:49.480 --> 00:15:07.609
Abe Elias: genotype-phenotype relationship. Now, that can also be done in the confirmatory testing. But in any case, so once you do… you do have the screening, then you have to do confirmatory testing, and that, that usually includes, the, ARSA enzyme activity.

84
00:15:07.610 --> 00:15:12.979
Abe Elias: In, you know, leukocytes, then urine sulfatites, and then

85
00:15:13.080 --> 00:15:35.520
Abe Elias: I think, generally, you… you want to do molecular testing for ARSA as well, to… because that can really predict, pretty… there's so much… I think when, if you remember, Josh Bunkowski at, at our last meeting, he mentioned that there's just a lot of, and it's true, there's just a lot of, we know a lot about the variants, and,

86
00:15:35.520 --> 00:15:36.340
Abe Elias: about…

87
00:15:36.480 --> 00:15:48.540
Abe Elias: what effect they have, and can predict, whether or not this will be a, like, a late infantile to an early juvenile, late juvenile, or adult. So, so that would be the confirmatory part.

88
00:15:48.710 --> 00:16:12.560
Abe Elias: And that would be done, and that's typically done from… through the newborn screening follow-up. And then once it's confirmed, and the recommendations there is that you should have, you know, that confirmation within, ideally within two weeks of initial patient contact, that's kind of the current recommendations. Then you would do the, the referral to a neurology with MRI and, and, and whatnot with that,

89
00:16:13.500 --> 00:16:22.640
Abe Elias: that's kind of the general framework, and I think that would be… that's pretty much in line, so that part would be a relatively…

90
00:16:23.120 --> 00:16:30.019
Abe Elias: I think straightforward. I think that the main thing, the main,

91
00:16:31.430 --> 00:16:46.679
Abe Elias: question is really the Wisconsin lab, where they are, because it is, and I know that some, you know, in, I think New York has a universal pilot right now. I know Minnesota is trying to get it on, but it's, it's not…

92
00:16:46.980 --> 00:16:52.650
Abe Elias: It's not… even though it's… it's a test that can be, can be,

93
00:16:53.220 --> 00:16:58.959
Abe Elias: multiplexed with the XALD, but easily, relatively easily. The…

94
00:17:00.170 --> 00:17:12.490
Abe Elias: developing a test that's both sensitive and specific and robust, because these are… the sulfur tides are a little bit finicky, I think, to detect them with that method. It's,

95

00:17:13.030 --> 00:17:32.740
Abe Elias: you know, it's a little hard to know when this actually will be. You know, one is to have it actually developed, and then they have to validate it on a sufficient number of cases, too. So I would not be surprised if they are still in the process of working that out, that it could potentially be longer than

96
00:17:32.750 --> 00:17:49.500
Abe Elias: you know, then perhaps the end of the year, that might be when they have it ready, and then they have to validate it. So it would be really interesting to hear a little bit from A Baker where they are. But as Jeanne said, there's going to be a legislative session too, so…

97
00:17:49.540 --> 00:18:04.250
Abe Elias: I think, but so from a follow-up perspective, once we have that, once it's, you know, we have a test that's both sensitive, specific, and well-validated, the discrete… the follow-up would be relatively straightforward, I think.

98
00:18:04.250 --> 00:18:12.170
Lynne Wood: Awesome. I'm so sorry if you had to say that twice. I think you were talking about the sulfatides just as I got back on the call, so I'm really sorry if we had to repeat everything because of me.

99
00:18:12.290 --> 00:18:15.829
Lynne Wood: But that helps me know, kind of, where we're at.

100
00:18:20.970 --> 00:18:23.480
Shelly Eagen: Additional questions or comments?

101
00:18:31.600 --> 00:18:38.870
Shelly Eagen: Mikaela and Stephanie, were there any questions or comments that were, submitted ahead of time that we haven't touched on yet?

102
00:18:40.610 --> 00:18:58.440
Mikaela Miller: Yes, I may chime in here. So most of the information was covered, but we did have an additional question that said, what treatment centers are available in Montana for MLD? If there are none, where's the next closest treatment?

103
00:18:59.180 --> 00:19:00.190
Mikaela Miller: center.

104
00:19:00.330 --> 00:19:04.760
Mikaela Miller: And I believe Nikki might have some more information on that.

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00:19:06.540 --> 00:19:19.310
Nikki Goosen: Yeah, so unfortunately, Montana doesn't have a qualified treatment center, but the closest one for us is gonna be the Primary Children's Hospital in Salt Lake.

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00:19:19.620 --> 00:19:29.190
Nikki Goosen: There are 8 total right now, so Salt Lake's the closest. They have one at the University of

Minnesota, as well, in Minneapolis.

107
00:19:29.440 --> 00:19:40.139
Nikki Goosen: And then they have one at San Francisco, too, so I think those three would probably be the closest options for us here in Montana.

108
00:20:03.160 --> 00:20:10.330
Shelly Eagen: Correct me if I'm wrong, but they would be able to get some of the initial workup done in Montana, correct?

109
00:20:14.300 --> 00:20:18.309
Nikki Goosen: Or do you mean, like, the follow-up testing-wise, or…

110
00:20:18.310 --> 00:20:19.120
Shelly Eagen: Yeah, yeah.

111
00:20:19.120 --> 00:20:19.470
Nikki Goosen: Yes, ma'.

112
00:20:19.470 --> 00:20:21.950
Shelly Eagen: The initial workup would be able to be done locally.

113
00:20:22.020 --> 00:20:23.440
Nikki Goosen: Yeah, yup.

114
00:20:24.960 --> 00:20:35.990
Nikki Goosen: Some… some of them might have to, you know, depending if it's a really, like, rural place, they might have to go, you know, to a bigger hospital to get some of the testing drawn, but… but if…

115
00:20:36.180 --> 00:20:38.590
Nikki Goosen: Within Montana, yeah.

116
00:20:40.470 --> 00:20:53.080
Jeanne Lee: I feel like, though we, did not think this was a big barrier, for Montana to… to get the MRI done, do you feel that way, Dr. Wood?

117
00:20:54.890 --> 00:21:07.319
Lynne Wood: I… you know, MRI can be performed everywhere. SPECT scanning may be a little bit more limited, as far as… as far as its interpretation. That was part of why I was wondering, like, hey, what's the follow-up plan?

118
00:21:07.750 --> 00:21:09.660
Lynne Wood: I think it might be worth it.

119

00:21:09.990 --> 00:21:21.399
Lynne Wood: for… I mean, I guess I don't know if it would be appropriate for this committee or somebody on this committee to do a little bit of research about, well, gosh, where are the scanners in the labs that would be able to do these?

120
00:21:21.490 --> 00:21:32.060
Lynne Wood: send-outs, and then the specific scans, because I'm sure they exist, and we'll just want to maybe identify them and somehow have that information out there for people who are following up on these tests.

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00:21:35.930 --> 00:21:37.270
Shelly Eagen: Yeah, Bridget, go ahead.

122
00:21:37.650 --> 00:21:42.549
Bridget Evans: So I know we're talking about MRI scans, but if I'm remembering correctly.

123
00:21:42.800 --> 00:21:57.040
Bridget Evans: the treatment for the early onset with the, bone marrow, with the gene therapy, you can't have MRI involvement to qualify for that one, correct? So how do we know that our providers are going to be educated on that and not give an MRI?

124
00:22:00.090 --> 00:22:05.319
Lynne Wood: Oh, sorry, be educated on that and not give an MRI? What do you mean?

125
00:22:05.950 --> 00:22:15.909
Bridget Evans: I guess, like, if we're doing some early stuff in Montana, or trying to, how do we know that the provider that they're seeing isn't going to refer for an MRI?

126
00:22:16.650 --> 00:22:25.660
Shelly Eagen: I think it… no, the MRI would be done if there's involvement, like, findings from the MRI,

127
00:22:26.180 --> 00:22:30.050
Shelly Eagen: That may make them ineligible for certain treatment options.

128
00:22:30.050 --> 00:22:33.609
Bridget Evans: Oh, I see, the findings from the MRI, not MRI involvement.

129
00:22:34.010 --> 00:22:34.870
Bridget Evans: Okay.

130
00:22:34.870 --> 00:22:41.550
Lynne Wood: Right, yeah, it's not like if you get an MRI, you're disqualified, it's if you see the signs that the white matter is already injured.

131
00:22:42.000 --> 00:22:55.510

Lynne Wood: then the risk of this transplant is probably not worth it, from what I understand, or, like, you wouldn't get as much benefit, and so if they have the, like, actual changes on the MRI, then they don't get some of the therapies.

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00:22:55.510 --> 00:22:57.190
Bridget Evans: Okay, that makes more sense. Thank you.

133
00:22:59.800 --> 00:23:08.969
Shelly Eagen: Stephanie put a note in here. Would it be helpful to review what the treatment process looks like? Do you guys feel like that would be helpful as a group?

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00:23:09.620 --> 00:23:10.910
Shelly Eagen: To review that?

135
00:23:11.050 --> 00:23:12.320
Steven Shapero: Yes, I agree.

136
00:23:12.490 --> 00:23:17.949
Shelly Eagen: Okay, Dr. Elias, I'm gonna let you go first, since you have your hand up, and then maybe we can get into that.

137
00:23:18.220 --> 00:23:42.709
Abe Elias: I think you're a little bit related to that. I… and I apologize, I was distracted, I had to answer a question here, and Nikki, when you mentioned earlier the treatment centers, could you just repeat quickly which they are? And then I wanted to ask, are those centers specifically, with regard to that FDA, the Lenmaldi, the… okay, I'm sorry, I heard you in the background, but then it's… I'm not good at multitasking.

138
00:23:43.220 --> 00:23:59.689
Nikki Goosen: No, that's okay. Yeah, so these are specifically related to that Lynne Meldy. Closest one for us is gonna be Primary Children's in Salt Lake, but there's also University of Minnesota, and then San Francisco in California.

139
00:23:59.820 --> 00:24:04.580
Nikki Goosen: And what I can do is I'm actually gonna share this in the chat.

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00:24:05.400 --> 00:24:09.330
Nikki Goosen: There we go. That way you can look at that link as well.

141
00:24:09.960 --> 00:24:32.819
Abe Elias: Thanks so much. Yeah, no problem. It's funny, you think you can, you know, but then you… it just kind of goes away. You know, I wonder, actually, just related to that review of the treatment, because Dr. Bonkowski, last time he gave that initial… and I wonder if we could ask him, he's the head of that treatment center, basically, in primary, if that's… if that is one of the default centers, or…

142
00:24:32.820 --> 00:24:40.419
Abe Elias: nearest, I guess. I wonder if you could ask him if he was able to join us to our… for our next

meeting.

143
00:24:40.420 --> 00:24:42.909
Abe Elias: Up, that might be something.

144
00:24:42.940 --> 00:24:48.319
Abe Elias: So, it might be nice just to specifically talk about that.

145
00:25:04.950 --> 00:25:12.590
Shelly Eagen: Is there somebody that feels comfortable talking about what the treatment process looks like, or is it best to refer to this link?

146
00:25:21.110 --> 00:25:29.320
Abe Elias: I think it probably would be good to have, in terms of the details, because the principle is really, that you,

147
00:25:29.560 --> 00:25:41.249
Abe Elias: You basically… it's… it's a… it's an ex vivo therapy, so you do, you get, bone marrow, you… you ablate the,

148
00:25:41.630 --> 00:26:04.159
Abe Elias: much of the cells, and just leave a certain subgroup of cells, certain CD… they have certain markers there, and then what you really do use a lentivirus-like vector, a virus, basically, to introduce a functional copy of that enzyme that is not working. So for the early infantile… sorry, late infantile form.

149
00:26:04.160 --> 00:26:07.949
Abe Elias: If you remember, there was a… he, he,

150
00:26:07.950 --> 00:26:23.180
Abe Elias: Burkerkovsky was talking about, there is a genotype-phenotype relationship, so those that are, that require really… the earliest are the ones that have two copies of… both copies of the genes, both parental copies, essentially not working, so loss of function.

151
00:26:23.180 --> 00:26:47.200
Abe Elias: And, and so, what you do is you introduce a functional copy, in that, vect… with that vector that is transcribed, so from that copy, which really provides the recipe to make that protein, that enzyme, not as efficiently as if we have two functional copies, but you don't really need, you know, a full transcription.

152
00:26:47.200 --> 00:26:57.950
Abe Elias: Which we know, because if you're a carrier, you have zero symptoms. So, so you basically, introduce a functional copy, and… and that is sufficient

153
00:26:58.060 --> 00:26:59.849
Abe Elias: To,

154

00:26:59.990 --> 00:27:17.849
Abe Elias: those cells, and then in the… you… you infuse that back, and those cells, they basically know where to go. They hone into the, the, you know, sort of blood-brain barrier, and, and, are able to, at least,

155
00:27:21.630 --> 00:27:28.820
Abe Elias: take some of the functions, basically. And, the outcomes have been, certainly for… for,

156
00:27:29.020 --> 00:27:35.549
Abe Elias: Survival and cognitive functions have been really, quite good if it's done early enough.

157
00:27:35.680 --> 00:27:49.010
Abe Elias: But that's really just the principle, so it would be good, probably, to… or maybe anyone else, maybe Lynne, I don't know if you… Dr. Wood, if you have some more, but it, it's, you know, the devil is always in the details.

158
00:27:51.450 --> 00:28:01.380
Lynne Wood: No, I think that's a great summary, and, you know, certainly as someone who has not treated one of these kiddos at one of those qualified centers, that would probably, summarize up my understanding, too.

159
00:28:08.720 --> 00:28:11.319
Shelly Eagen: Additional questions, comments?

160
00:28:12.060 --> 00:28:13.830
Shelly Eagen: Things we need to address.

161
00:28:21.620 --> 00:28:37.659
Lynne Wood: I… again, I'm so sorry if this has already gone over, and I missed it when I had to jump off the call, but I think the goal in setting this interim meeting was to follow up with Jeannie as far as, hey, what have we learned from May about when this test may be available, and how it might look?

162
00:28:37.800 --> 00:28:49.770
Lynne Wood: To sort of inform, hey, are we gonna move forward with this or not at our next meeting? To me, it sounds like we've probably gathered a lot of the information that we're going to need to make that initial decision.

163
00:28:49.770 --> 00:29:03.880
Lynne Wood: outside of, you know, some of those side projects people brought up, as far as, like, well, you know, where are the labs and where are the scanners that we would need to actually do the follow-up testing and then get a kiddo to a specialized treatment center if they need it? Does that sound right to everybody else?

164
00:29:10.420 --> 00:29:21.180
Jeanne Lee: Yeah, so just the part that you missed was that when I did chat with Mae Baker that she is going to have an assay available.

165

00:29:21.270 --> 00:29:36.519
Jeanne Lee: And… and there was a little bit of question whether she would be able to multiplex it with XALD, or if it would be its own assay. So she… she was working through that.

166
00:29:36.790 --> 00:29:47.889
Jeanne Lee: But I think her hope was to multiplex it so that, you know, like, with one punch, she would be able to test both XALD and MLD.

167
00:29:48.140 --> 00:29:58.959
Jeanne Lee: And so, doctor did have a question, Dr. Elias did have a question about whether second-tier testing, would also be done.

168
00:29:58.960 --> 00:30:10.269
Jeanne Lee: Through the same blood spot, or if it will, need to be recollected. And so, I… I will find that out from her.

169
00:30:10.370 --> 00:30:18.230
Jeanne Lee: For the next meeting. And then… and then the other question was, how much wood screening

170
00:30:18.330 --> 00:30:32.510
Jeanne Lee: raise the newborn, fee by, and so I think we're looking at, you know, up to $3 per test, but it may be a little bit lower if she's able to

171
00:30:32.820 --> 00:30:35.729
Jeanne Lee: multiplex it. Great.

172
00:30:35.890 --> 00:30:40.659
Lynne Wood: Yeah, to me, that sounds like we… we did get everything we needed. Does everybody else agree?

173
00:30:41.220 --> 00:30:52.480
Douglas Harrington: Yeah, hey, I'm not able to raise my hand for some reason, but I just wanted to add, the… most labs that I am familiar with are doing a,

174
00:30:52.810 --> 00:31:10.730
Douglas Harrington: a two- or three-tiered blood spot test that's along the lines of what Dr. Elias said. The first tier is measuring the sulfatides, the second tier is the ARSA enzyme, and the third tier is genetic confirmation through sequencing, and

175
00:31:11.430 --> 00:31:17.979
Douglas Harrington: And it's all done from the one blood spot, so that's kind of the good news. And then…

176
00:31:18.540 --> 00:31:36.529
Douglas Harrington: that has a… that system has about a 99% to 100% sensitivity and specificity, so it's really good. And, the other thing that came out of most of the stuff that I've reviewed in the past is that, you know, the… the,

177
00:31:36.840 --> 00:31:43.900
Douglas Harrington: 50 to 60% of kids that have late infantile onset.

178
00:31:44.250 --> 00:31:52.290
Douglas Harrington: need to be treated before symptoms develop to have a good outcome with the FDA-approved product.

179
00:31:52.530 --> 00:31:57.030
Douglas Harrington: And it's unknown how that will play out in

180
00:31:57.200 --> 00:32:02.980
Douglas Harrington: the other forms which appear later in life, so… I think it's…

181
00:32:03.560 --> 00:32:11.890
Douglas Harrington: probably likely that, as Jeannie said, they'll follow that same protocol that other labs have validated. Okay, sorry about that.

182
00:32:14.050 --> 00:32:16.310
Lynne Wood: Yeah, no need to apologize, super helpful.

183
00:32:20.300 --> 00:32:21.090
Shelly Eagen: Nikki?

184
00:32:22.250 --> 00:32:35.699
Nikki Goosen: Yeah, I can't remember if I said this at our last meeting, but we did actually increase our blood spot collection cards from 5 spots to the 8 spots, so we do have a little more blood to work with, which is nice.

185
00:32:38.190 --> 00:32:40.480
Lynne Wood: I think I do recall you saying that at the last meeting.

186
00:32:48.340 --> 00:32:49.690
Shelly Eagen: Anybody else?

187
00:32:56.980 --> 00:33:05.669
Lynne Wood: I think if we covered all the questions… I can't remember if we have to follow Robert's rules here. Do I have to, like, motion to adjourn the meeting, or no?

188
00:33:09.340 --> 00:33:15.780
Shelly Eagen: I think if there's no more questions, we, actually, Mikaela, is there anybody that has joined from the public?

189
00:33:17.280 --> 00:33:19.899

Mikaela Miller: No, I was just gonna say,

190
00:33:20.670 --> 00:33:39.990
Mikaela Miller: We can, skip this 10-minute public comment period, but this is kind of a good moment to take a final call if there's anything else, that may be helpful for you all to cast your vote in the fall. Any other lingering information you'd like to have before you're able to make a decision on that vote?

191
00:33:51.550 --> 00:33:52.560
Mikaela Miller: Okay.

192
00:33:53.930 --> 00:33:57.949
Mikaela Miller: Always try to take a few breaths here to give everyone a moment.

193
00:33:58.070 --> 00:34:10.689
Mikaela Miller: But I think that covers it. Feel free to email Stephanie or I, or this general NBS committee email if you have anything else. Otherwise, we'll go ahead and…

194
00:34:10.929 --> 00:34:22.110
Mikaela Miller: close out the meeting, so any follow-up that we'll have, we've got some materials that'll be shared online, and I will email them out to all of you individually as well.

195
00:34:22.210 --> 00:34:39.180
Mikaela Miller: That's going to be the recording, the notes, and the slides. And we'll also update the public website with all of that information. And then, as you all know, we've got that meeting this fall. We've sent a poll out at this point, but I believe we're waiting on a few more responses before we can solidify that.

196
00:34:39.179 --> 00:34:54.909
Mikaela Miller: But it's coming very soon, and that's where we'll hold the vote, and then we'll also hear a new presentation for mucopolysaccharidosis, type 2 and PS2. We'll hear that condition next.

197
00:34:55.060 --> 00:34:59.350
Mikaela Miller: Is there anything else, Shelly, that you'd like to say as we close out the meeting?

198
00:34:59.350 --> 00:35:09.009
Shelly Eagen: Well, just again, welcome our new member, Bridget, to the committee. Thank you all for joining, and please get the poll done.

199
00:35:09.010 --> 00:35:20.980
Shelly Eagen: To make sure that we can figure out the timing of our next meeting and block our calendars appropriately. And then, if you haven't all done your conflict of interest, please make sure that that is done as well.

200
00:35:25.620 --> 00:35:27.139
Shelly Eagen: And we'll see you in the fall!

201

00:35:27.800 --> 00:35:32.479
Mikaela Miller: Alright, thanks so much. Please enjoy having some extra time back in your day, everyone.

202
00:35:34.210 --> 00:35:35.690
Abe Elias: Thank you, bye-bye.

203
00:35:36.160 --> 00:35:36.970
Mikaela Miller: Okay.