1
00:00:02.150 --> 00:00:26.149
Mikaela Miller: Oh, thank you, Stephanie. We're gonna start the recording and then we're gonna go ahead and do a quick roll call. If you could. Please just say your name, the organization you represent, what your role is, and your physical description. Jen, do you wanna go ahead and popcorn. All the introductions. Feel free to introduce yourself first, st and then pass it on.

2
00:00:26.670 --> 00:00:36.060
Jenn Banna MTF2FHIC: Okay. Yes, thanks. My name is Jen Banna. I'm with Montana's Family Family Health Information Center. I'm here as the family organization

3
00:00:36.720 --> 00:00:37.120
Jenn Banna MTF2FHIC: and

4
00:00:37.420 --> 00:00:50.880
Jenn Banna MTF2FHIC: I have gray and blue hair that's up in a ponytail, and I'm wearing a gray sweater and glasses today, and I'm gonna go ahead and send it over to Steven to introduce himself.

5
00:00:51.680 --> 00:00:57.880
Steven Shapero: Yes, Hi! My name is Steven Shapiro, and I represent families with rare diseases.

6
00:00:58.010 --> 00:01:09.909
Steven Shapero: I also have emergencies myself, and I am a man. And I'm wearing a a checkered shirt, and I'm wearing glasses and a hat.

7
00:01:11.790 --> 00:01:14.430
Jenn Banna MTF2FHIC: Thank you. Let's go ahead and go to Shannalia.

8
00:01:17.790 --> 00:01:31.890
Shawnalea Chief Goes Out: I'm with Dphhs's Health Resources Division, and my role is representing Medicaid. My physical description is brown skin, brown hair, brown eyes, hair in a bun and

9
00:01:32.230 --> 00:01:33.960

Shawnalea Chief Goes Out: pink cardigan.

10
00:01:35.280 --> 00:01:46.289
Jenn Banna MTF2FHIC: Okay, thank you. I think that is everybody that's here from the voting members shelly. Egan is excused today because she is gone.

11
00:01:47.146 --> 00:01:50.629
Jenn Banna MTF2FHIC: Let's see. Oh, we'll go ahead and let Ava introduce himself.

12
00:01:50.790 --> 00:01:52.800
Stephanie Burkholder: And we have Dr. Wood here as well.

13
00:01:53.340 --> 00:01:55.999
Jenn Banna MTF2FHIC: Okay, I can't come. I can't see Dr. Wood on my screen.

14
00:01:56.420 --> 00:01:57.919
Jenn Banna MTF2FHIC: Sorry, Dr. Wood, go ahead.

15
00:02:00.220 --> 00:02:01.150
Stephanie Burkholder: Oh! Did we lose our.

16
00:02:01.150 --> 00:02:05.530
Mikaela Miller: I don't see her either. She might have fallen off. She was here for a moment, though.

17
00:02:05.980 --> 00:02:07.040
Jenn Banna MTF2FHIC: That's what I thought.

18
00:02:07.448 --> 00:02:11.200
Jenn Banna MTF2FHIC: Hey, Abe, we're just introducing ourselves. Will you go ahead and go next?

19
00:02:11.470 --> 00:02:17.587

Abe Elias: Yeah, yeah, I'm Eve, Elias, Chief Medical Officer show there and sorry for joining late. I

20
00:02:18.350 --> 00:02:22.210
Abe Elias: I've had difficulties and I still have difficulties a little bit with the connection.

21
00:02:23.155 --> 00:02:27.129
Abe Elias: But I think, can you hear me? Okay.

22
00:02:28.920 --> 00:02:32.350
Abe Elias: I. It's huh?

23
00:02:37.180 --> 00:02:38.929
Abe Elias: Okay, I hope I might.

24
00:02:38.930 --> 00:02:41.789
Jenn Banna MTF2FHIC: Are you doing? Okay? Do you want to give a short physical description? Abe?

25
00:02:42.175 --> 00:02:42.945
Abe Elias: Yeah, I'm

26
00:02:45.590 --> 00:02:50.912
Abe Elias: So let's see here, Shaudea, our genetics geneticist. I'm a middle aged,

27
00:02:51.670 --> 00:03:00.325
Abe Elias: black gray haired and wearing a white shirt and glasses.

28
00:03:01.480 --> 00:03:07.170
Jenn Banna MTF2FHIC: You got it, you got it? We'll go to Douglas Harrington next. Who's a non-voting member of the committee.

29
00:03:07.240 --> 00:03:08.029
Douglas Harrington: Have to change.

30
00:03:08.030 --> 00:03:14.829
Douglas Harrington: Yeah. Douglas Harrington. I represent Dphhs. I'm the

31
00:03:15.230 --> 00:03:25.950
Douglas Harrington: State Medical Officer and the Executive Director of the Health Facilities Division. I am an older white male, with gray hair and a beard and glasses.

32
00:03:27.120 --> 00:03:27.990
Jenn Banna MTF2FHIC: Thank you.

33
00:03:28.130 --> 00:03:45.839
Jenn Banna MTF2FHIC: We also have members of the internal committee that are here today, just for everybody's awareness. Amber Bell, Jacqueline Islay and Miranda Redig won't be able to join us today. So let's go ahead and let's see, I saw Chelsea. You go ahead and introduce yourself. Chelsea.

34
00:03:46.550 --> 00:03:55.110
Chelsea Pugh: Yeah. Hello. My name is Chelsea Pugh, and I am with Dphhs as a nurse consultant for children's special health services.

35
00:03:55.300 --> 00:03:59.219
Chelsea Pugh: and today I am wearing a black, long sleeve shirt.

36
00:03:59.330 --> 00:04:04.980
Chelsea Pugh: I have strawberry blonde, slash, red hair that's pulled back in a clip, and I have freckles.

37
00:04:06.610 --> 00:04:09.490
Jenn Banna MTF2FHIC: Thank you. Dr. Wood. Can you introduce yourself.

38
00:04:10.630 --> 00:04:27.439
E. Lynne Wood: Yes, sorry I had to log off. My connection was bad for a second. I am Dr. Wood. I am a Billings clinic, and I'm a physician. I am a middle-aged, blonde woman with long messy hair, and I am wearing a black polka dot tank top.

39
00:04:29.040 --> 00:04:32.359
Jenn Banna MTF2FHIC: Thank you. Okay, Deb. Gibson, can you introduce yourself.

40
00:04:37.145 --> 00:04:41.290
Debbie Gibson: Yeah, well, I'm trying to get my video, am I on yet?

41
00:04:41.820 --> 00:04:42.429
Debbie Gibson: Can hear you?

42
00:04:42.430 --> 00:04:45.129
Debbie Gibson: I can't get my video to. Oh, there we go.

43
00:04:45.420 --> 00:04:46.310
Debbie Gibson: Okay?

44
00:04:46.690 --> 00:04:54.829
Debbie Gibson: Yeah. So I'm Debbie Giventon. I'm the Laboratory Services Bureau chief. I'm a non voting member here. I'm

45
00:04:55.520 --> 00:05:02.339
Debbie Gibson: white female, longer blonde hair, blue eyes. And I'm wearing a long sleeve, black shirt.

46
00:05:03.680 --> 00:05:05.219
Jenn Banna MTF2FHIC: Thank you, Jeannie.

47
00:05:08.290 --> 00:05:19.889
Jeanne Lee: Hi, there! I'm Jeannie Lee. I'm the newborn screening and serology Supervisor in the Montana Public Health Laboratory. I'm a non voting member.

48
00:05:20.596 --> 00:05:36.360
Jeanne Lee: I am a white woman with kind of collarbone length, brown hair, and today I'm a little bit chilled. So I'm wearing my mauve jacket.

49
00:05:37.960 --> 00:05:50.339
Jenn Banna MTF2FHIC: Thank you, Jeannie, Stephanie and Michaela. You did not get a chance to introduce yourselves when we got started today, I don't think. And after the 2 of you. I think we've covered all of the voting and nonvoting members and the internal committee.

50
00:05:53.790 --> 00:06:00.750
Stephanie Burkholder: Okay. I'm Stephanie Burkholder. I'm a white female, with brown and gray hair

51
00:06:00.870 --> 00:06:09.289
Stephanie Burkholder: and a black shirt. I work with Michaela for Yarrow, and we're facilitating this meeting as well as kind of the entire committee.

52
00:06:12.170 --> 00:06:27.850
Mikaela Miller: Thank you, Stephanie. Yeah, you've pretty much covered it. My name is Michaela. Once again we work for Yarrow, and we're here to facilitate this meeting. I am a white female with brown hair pulled back in a hair tie today with a teal top on.

53
00:06:28.940 --> 00:06:32.119
Mikaela Miller: Thank you. Everyone for providing your

54
00:06:32.270 --> 00:06:38.429
Mikaela Miller: introductions. Let's go ahead and move on to the next slide here.

55
00:06:38.640 --> 00:06:49.489
Mikaela Miller: And we're gonna just read this acknowledgement. We've decided to go ahead and include this with our meetings going forward. Jen, if you'd like to go ahead and read this for us, that would be great.

56
00:06:50.130 --> 00:07:13.130
Jenn Banna MTF2FHIC: Thank you. I'm excited to read this. I'm hoping this will help contribute to a more robust discussion for all of us, because I know this was one of the things that's dear to my heart. So we thank the families, caregivers, committee members and advocates for their contributions to the Montana newborn screening program. We recognize that each condition

reviewed affects children and families in Montana, and we strive to balance the emotions and vulnerabilities shared with the need

57
00:07:13.130 --> 00:07:22.120
Jenn Banna MTF2FHIC: for careful, sometimes difficult discussions on logistics and finances. Our goal is to ensure the process is publicly accessible, transparent, and carefully examined. Thanks.

58
00:07:23.430 --> 00:07:24.809
Mikaela Miller: Wonderful. Thank you.

59
00:07:26.830 --> 00:07:53.759
Mikaela Miller: Next, I would just like to take a quick moment here to go over an acronym. We normally try to avoid acronyms in these meetings, but I have a feeling due to the nature of today's meeting. You'll hear Asmd quite often, and that just stands for acid sphingomyelinase deficiency, which is also known as Niemann-pick disease. And that is the condition that we will be hearing today.

60
00:07:55.730 --> 00:08:18.599
Mikaela Miller: All right. So here is our agenda for today. We're a little ahead of schedule, which is great. That'll allow for plenty of time for discussion later in the meeting. So from until about 1220 we had welcome roll call. Next, we're going to do some brief updates that the committee has to share.

61
00:08:18.600 --> 00:08:35.630
Mikaela Miller: The Asmd nomination packet review will come next, and then we have a chunk of time to go ahead and discuss that packet, and then we're going to hear from the subject matter expert we have presenting at our meeting today, Dr. Gerald Raymond.

62
00:08:35.640 --> 00:08:44.639
Mikaela Miller: Then we're going to take just a short 10 min break before we jump into the family presentation by Dr. Justin Hopkin.

63
00:08:45.160 --> 00:08:50.099
Mikaela Miller: Then we are going to hear from Jeannie with the Montana State laboratory.

64

00:08:50.450 --> 00:09:08.549
Mikaela Miller: just a short presentation on some of the logistics of testing, and then we have a chunk of time for the Asmd discussion where members can discuss amongst each other voting members primarily. But

65
00:09:09.001 --> 00:09:16.729
Mikaela Miller: just to be able to ask questions to the presenters, or discuss any other things they would like to bring up.

66
00:09:17.020 --> 00:09:36.339
Mikaela Miller: And then we always have a 10 min public comment period. During these meetings I don't see any members from the public here today, but if anyone happens to hop on they will have that 10 min towards the end of the meeting, and then we'll have kind of just a brief next steps and closing at the end of the meeting.

67
00:09:38.140 --> 00:10:03.950
Mikaela Miller: All right. So just to note that again, that public comment period, just to give everyone a brief reminder how we run those. We do reserve that for members of the public, not the committee members. So just keep in mind that we do try to reserve that time. If anyone does happen to hop on, each person gets 2 min to ask their questions.

68
00:10:03.950 --> 00:10:10.080
Mikaela Miller: And I can show this again later on, if anyone happens to hop on. But that's how we run it.

69
00:10:11.320 --> 00:10:30.490
Mikaela Miller: Next, just some ground rules, and we always like to read through these just to make sure that we can have a effective and respectful meeting. These ground rules include please making sure you mute your microphone. If you're not talking just to help minimize any kind of background noise that you may not have control over.

70
00:10:30.490 --> 00:10:43.840
Mikaela Miller: please have your video on unless there's distractions in your background. The chat will be used for asking any questions during the meeting, either Stephanie or I. The moderators will go ahead and read those aloud when it's appropriate.

71
00:10:44.000 --> 00:11:08.550

Mikaela Miller: feel free to ask any clarifying questions. If you don't understand something, but just make sure to avoid talking over or interrupting other speakers. Just please be clear and avoid any acronyms when we're discussing, just to ensure that everyone knows all the relevant information. There's a lot of people here from various backgrounds. So we just like to keep, make sure that

72
00:11:09.200 --> 00:11:16.870
Mikaela Miller: information is understandable. Try using any specific examples. When you're explaining any points.

73
00:11:17.160 --> 00:11:42.149
Mikaela Miller: remember to focus on the collective interests and goals of the committee rather than individual positions or opinions due to the time bound nature of these meetings. Not all disagreements may be able to be solved within the meeting time. Any additional meetings or communications may be scheduled to continue the conversation. Just to make sure we leave room in the scheduled agenda.

74
00:11:42.150 --> 00:11:49.840
Mikaela Miller: So, for example, if something is left unresolved during those discussion periods, and we can always come back to those at a later time.

75
00:11:51.230 --> 00:11:58.189
Mikaela Miller: Next steps or action items will be assigned to each individual just to help ensure any accountability

76
00:11:58.660 --> 00:12:08.209
Mikaela Miller: in order to ensure equity of voice and engagement facilitators. So Stephanie or I may call on attendees for their input.

77
00:12:08.470 --> 00:12:33.170
Mikaela Miller: And lastly, this meeting space is intended to be a safe space just to help guide the determination of screening for newborn conditions. So if you do not feel comfortable sharing in the meeting space, please let Stephanie or I know, and we can communicate with you in another way, whether it be an individual chat or over email.

78
00:12:36.090 --> 00:12:42.994
Mikaela Miller: Alright. So we're just gonna kind of roll right into the updates.

79
00:12:43.640 --> 00:13:12.240
Mikaela Miller: we just have a really brief chunk of time scheduled for this one quick update to share here. So 1st of all, thank you to all of you who were involved in the meetings related to gaucher disease. As you may recall at our November 2024, meeting to vote on adding gaucher disease to the Montana newborn screening panel. The advisory committee voted not to include it on the panel.

80
00:13:12.290 --> 00:13:29.910
Mikaela Miller: just to let everyone know the committee's recommendation was then forwarded on to the Dphhs, or Director of Public Health and Human Services Director for the State of Montana, Charlie Breton, and he concurred with the Committee's decision.

81
00:13:30.000 --> 00:13:59.360
Mikaela Miller: or, I should say, the committee's recommendation, and that was on December 20th of 2024, so approximately a month later, thus moving forward, gaucher disease will officially not be included on our Montana newborn screening panel. However, we do have stipulations in those condition nomination packets that it can be submitted for again at a future time. Should something change in the

82
00:13:59.420 --> 00:14:05.919
Mikaela Miller: condition, whether it be a new treatment or new information found out.

83
00:14:08.368 --> 00:14:12.041
Mikaela Miller: So that's somewhat how the timeline and process works.

84
00:14:12.690 --> 00:14:40.140
Mikaela Miller: Next, we're going to go ahead and just roll right into that nomination packet review for Asmd. I just want to make sure to note that this is quite a broad overview of some of the items from the nomination packet that we ask for. But just know that you will hear more details from the subject matter expert and the family presenter later in the meeting.

85
00:14:41.490 --> 00:14:44.699
Mikaela Miller: So kind of a broad look at

86
00:14:45.320 --> 00:15:06.320

Mikaela Miller: the 2 main types. If we were to overview quickly here the signs and symptoms. Just know that they're highly variable between the 2 conditions at age of onset for or I'm sorry. Age of onset signs and symptoms of type. A disease include

87
00:15:06.500 --> 00:15:29.870
Mikaela Miller: on the slide here. So we've got age of onset. For type A is in early infancy. Signs and symptoms include enlarged liver and or spleen, accumulation of fluid in the abdomen, jaundice, feeding difficulties, constipation, nausea, vomiting, significant gastrointestinal reflux, failure to thrive

88
00:15:29.870 --> 00:15:39.209
Mikaela Miller: irritability loss of reflexes, and progressive loss of muscle, tone and respiratory issues. So keep in mind. That's for type A

89
00:15:39.440 --> 00:16:08.429
Mikaela Miller: type B kind of a brief overview. We have age of onset, infancy to adulthood signs and symptoms include some similar to type, a, but not necessarily as severe enlarged liver and or spleen. Increased infections, prolonged bleeding, abdominal pain, liver disease, respiratory issue. Neurological issues delayed growth, slash, puberty and bone thinning.

90
00:16:08.980 --> 00:16:15.119
Mikaela Miller: And once again keep in mind. We'll go into detail in those a little bit later in the presentation.

91
00:16:16.450 --> 00:16:39.109
Mikaela Miller: How is the disorder currently identified? So this primarily starts out with symptomatic presentation, followed by a blood test. Why should it be screened for at birth, early detection and management can help mitigate some of the symptoms or some of these serious health risks, and include or an improve quality of life.

92
00:16:39.440 --> 00:16:56.490
Mikaela Miller: How's the disorder treated enzyme replacement therapy or ert. I'm sure we'll hear more detail about the treatments later today. There is an FDA approved treatment, and it is not in the experimental phase.

93
00:16:56.640 --> 00:17:01.590
Mikaela Miller: The proposed screening test method is the dried blood spot test

94
00:17:05.859 --> 00:17:26.629
Mikaela Miller: next up status of the condition in the United States. So we found that States currently screening for the condition include Illinois and New Jersey. The condition has been reviewed by the rusp and the registries or databases that are currently established for the condition include 2.

95
00:17:31.300 --> 00:17:54.580
Mikaela Miller: Next, we have the selection criteria. So these are used by the voting members to help assess their decision on whether or not to include the condition on the panel, so it can be identified at a period of time within 24 to 48 h after birth, at which would not ordinarily be clinically detected.

96
00:17:55.170 --> 00:18:00.299
Mikaela Miller: A test with appropriate sensitivity and specificity is available.

97
00:18:00.750 --> 00:18:10.319
Mikaela Miller: There is significant risk of illness, disability, or death. If babies are not treated promptly within the recommended timeframe for the condition

98
00:18:10.460 --> 00:18:18.330
Mikaela Miller: effective treatment is available, and access to follow up care and counseling is generally available.

99
00:18:18.890 --> 00:18:27.650
Mikaela Miller: There are demonstrated benefits of early detection, timely intervention, and efficacious treatment.

100
00:18:28.320 --> 00:18:39.020
Mikaela Miller: Number 6 is the benefits to babies and to society outweigh the risks and burdens of screening and treatment. That is also true.

101
00:18:40.790 --> 00:18:45.740
Mikaela Miller: The last few. Here there's minimal financial impacts on the family.

102

00:18:46.560 --> 00:19:06.090
Mikaela Miller: There is a public health benefit to conducting the test. There exists responsible parties who will follow up with families and implement necessary interventions, and the conditions case, definition and spectrum are well described. We'll hear from the lab

103
00:19:06.821 --> 00:19:08.830
Mikaela Miller: later in the presentation today.

104
00:19:12.490 --> 00:19:30.790
Mikaela Miller: So that are is some of those like selection criteria for the packet feel free. If you want me to go back and share any of those slides for this discussion, but I'm going to go ahead and pass it over to Jen to go ahead and discuss that packet with the voting members.

105
00:19:34.140 --> 00:19:40.489
Jenn Banna MTF2FHIC: Thank you. Michaela. So I have one question about the information about the rus.

106
00:19:41.014 --> 00:19:47.109
Jenn Banna MTF2FHIC: That wasn't clear. If the rest had considered it, or if it was added to the rest in the way that question was answered.

107
00:19:47.970 --> 00:19:52.640
Jenn Banna MTF2FHIC: and I was trying to look through the background, because that just says reviewed.

108
00:19:53.810 --> 00:20:03.640
E. Lynne Wood: I think, in the packet there was a letter from back in 2,008 about the review, and I I don't think that it was accepted, based on what I saw. But okay.

109
00:20:03.640 --> 00:20:05.349
Jenn Banna MTF2FHIC: I was trying to find it. Thank you.

110
00:20:07.470 --> 00:20:10.580
Mikaela Miller: Yes, in your email there should be a letter.

111

00:20:11.568 --> 00:20:15.159
Mikaela Miller: I believe it's called Asmd addendum from Montana.

112
00:20:15.800 --> 00:20:16.440
Jenn Banna MTF2FHIC: Got it.

113
00:20:18.886 --> 00:20:24.749
Mikaela Miller: Actually, I think the file is called Neiman pick letter, and then it says, Dash committee.

114
00:20:27.830 --> 00:20:30.529
Jenn Banna MTF2FHIC: Got it. I was looking through things, but not in the right order.

115
00:20:33.570 --> 00:20:36.239
Jenn Banna MTF2FHIC: and this is from 22,008.

116
00:20:36.620 --> 00:20:37.390
Mikaela Miller: Yes.

117
00:20:37.720 --> 00:20:38.880
Jenn Banna MTF2FHIC: Okay, got it.

118
00:20:39.610 --> 00:20:40.540
Jenn Banna MTF2FHIC: Thank you.

119
00:20:48.110 --> 00:20:49.680
Jenn Banna MTF2FHIC: Was there anything else that

120
00:20:53.580 --> 00:20:56.869
Jenn Banna MTF2FHIC: came up for anybody, or questions specific to that? But.

121
00:20:57.070 --> 00:21:13.090

E. Lynne Wood: I'm not sure if this is the time for me to ask. But sort of to your point about that letter from 2,008. That seems like a long time ago. I don't think the Ert was out by then. Does anyone know if Ert was available back when they did the 1st application.

122
00:21:13.910 --> 00:21:14.560
Justin Hopkin: I can answer.

123
00:21:14.933 --> 00:21:15.680
Jenn Banna MTF2FHIC: Dr. Hoppin.

124
00:21:15.680 --> 00:21:16.080
Justin Hopkin: Yeah, yeah.

125
00:21:16.080 --> 00:21:16.959
Jenn Banna MTF2FHIC: Yeah. Go ahead.

126
00:21:16.960 --> 00:21:27.870
Justin Hopkin: So yeah, it it was not worthy of a rust. Nomination at that point. In time it was approved in August. 2 years ago. So the it's a recently approved ert good question.

127
00:21:28.130 --> 00:21:29.020
E. Lynne Wood: Thank you.

128
00:21:31.336 --> 00:21:38.010
Jenn Banna MTF2FHIC: And I. This might not be the right time for this question, either. But I know when we're talking about types A and B.

129
00:21:38.370 --> 00:21:42.890
Jenn Banna MTF2FHIC: Those are. That's the same test, or they come back separately.

130
00:21:43.140 --> 00:21:47.085
Jenn Banna MTF2FHIC: Same test. No, go ahead, Dr. Hopkin. Go ahead and.

131
00:21:47.480 --> 00:22:03.990

Justin Hopkin: It's a spectrum disorder, and the name were, I would say, they were outlined by clinicians like 4 decades ago. It's the same gene and the same disease. It's just the severity of symptoms, and I'll talk about that.

132
00:22:05.360 --> 00:22:07.008
Jenn Banna MTF2FHIC: I thought you might.

133
00:22:09.040 --> 00:22:31.890
Abe Elias: Yeah, just also I wanted to with in the earlier table. So the the slides it mentions that a treatment effective treatment is available. And I know this will be addressed today as well, of course, but I just wanted to mention that you know that the treatment that is effective treatment is is really

134
00:22:32.030 --> 00:22:40.609
Abe Elias: that is effective is related to the visceral aspect and not the neurological aspects. I think that is an important point.

135
00:22:40.720 --> 00:22:47.880
Abe Elias: The table, of course, doesn't allow for a qualification of that, but I think we'll be hearing about that.

136
00:22:49.210 --> 00:22:51.430
Jenn Banna MTF2FHIC: Abe, you said. Did you say visceral.

137
00:22:51.430 --> 00:23:07.019
Abe Elias: Yeah. Oh, sorry. Yeah. So, for you know the effect how the disease affects, especially the liver, the spleen, and the lungs, but not how it affects the nervous system.

138
00:23:09.090 --> 00:23:15.750
Jenn Banna MTF2FHIC: Thank you. Anything else about the application or the selection criteria that

139
00:23:18.050 --> 00:23:22.790
Jenn Banna MTF2FHIC: I think we'll understand more after we hear from both of our experts.

140
00:23:31.520 --> 00:23:32.880
Steven Shapero: I had a question.

141
00:23:33.490 --> 00:23:39.789
Steven Shapero: I by the way, is there a hand raise somewhere that I should be using when I have a question.

142
00:23:40.580 --> 00:23:45.750
Mikaela Miller: I think there's a hand raise in there somewhere, but you're just fine. If you wanna speak up.

143
00:23:46.260 --> 00:23:55.029
Steven Shapero: Yeah, simple question. So it's, I believe there's 2 states that are currently testing for this. Do we know how long they've been testing? For?

144
00:23:56.830 --> 00:23:57.930
Steven Shapero: Did anyone know.

145
00:23:57.930 --> 00:24:06.880
Justin Hopkin: I'm gonna present that data. As far as Illinois is concerned. New Jersey's been testing for 5 years, and I'll review Illinois, who's been testing for over a decade.

146
00:24:06.880 --> 00:24:07.949
Steven Shapero: Perfect. Thank you.

147
00:24:12.690 --> 00:24:18.270
Jenn Banna MTF2FHIC: And I'm not afraid to have a little bit of quiet time where people think about. If there's questions that they have.

148
00:24:28.560 --> 00:24:36.640
Jenn Banna MTF2FHIC: okay, well, we'll have an opportunity later to discuss more. So seems like we can go ahead and move on. Michaela.

149
00:24:37.110 --> 00:24:37.800
Mikaela Miller: Okay.

150
00:24:39.610 --> 00:24:40.830
Mikaela Miller: Well.

151
00:24:42.310 --> 00:24:54.880
Mikaela Miller: that then, brings us to the presentation from Dr. Raymond. I'll go ahead and let you, Jen, if you'd like to introduce our presenter.

152
00:24:55.530 --> 00:25:14.109
Jenn Banna MTF2FHIC: Hey? Thanks for being here today. Dr. Raymond. Dr. Raymond is a professor of genetic medicine and neurology at Johns Hopkins, in Baltimore, where he serves as the director of the Lysosomal Storage Disease program. He graduated from the University of Connecticut School of Medicine and did speciality.

153
00:25:14.110 --> 00:25:30.070
Jenn Banna MTF2FHIC: specialty training in pediatrics, child neurology and genetics. He's been involved in the clinical care and research in neurogenetic disorders. He has also been involved in newborn screening, especially that of x-linked adrenolucodystrophystrophia

154
00:25:30.490 --> 00:25:32.040
Jenn Banna MTF2FHIC: close consulting with

155
00:25:32.430 --> 00:25:43.929
Jenn Banna MTF2FHIC: New York's program in other States, and advocated for its inclusion on the rest, he presently serves as the chair of the Maryland State Advisory Council on hereditary and congenital disorders.

156
00:25:44.350 --> 00:25:46.889
Jenn Banna MTF2FHIC: Thank you, Dr. Raymond. I'll turn it over to you.

157
00:25:47.610 --> 00:25:48.310
Gerald Raymond: Okay.

158
00:25:48.760 --> 00:25:50.040
Gerald Raymond: You want me to share.

159
00:25:50.290 --> 00:25:53.339
Mikaela Miller: Yeah, go ahead and share your slides. If that works for you.

160
00:25:53.500 --> 00:25:55.979
Gerald Raymond: It's not turned on, so I'm sending a request

161
00:26:00.870 --> 00:26:01.850
Gerald Raymond: share.

162
00:26:02.330 --> 00:26:06.180
Mikaela Miller: Looks like you should be good to go. I'll see if it lets you.

163
00:26:07.470 --> 00:26:08.530
Gerald Raymond: Share that.

164
00:26:08.800 --> 00:26:09.669
Stephanie Burkholder: There it goes!

165
00:26:09.670 --> 00:26:12.189
Gerald Raymond: Okay, and let me put it in presenter mode.

166
00:26:15.090 --> 00:26:18.450
Gerald Raymond: And can you see it in presenter mode? Or is it still a slides.

167
00:26:18.930 --> 00:26:21.470
Mikaela Miller: Looks like it's still the the slides.

168
00:26:21.810 --> 00:26:24.650
Gerald Raymond: Yeah, hold on there.

169
00:26:25.230 --> 00:26:28.490
Gerald Raymond: Okay. And which screen are you seeing.

170
00:26:28.690 --> 00:26:30.060
Mikaela Miller: The presenter mode.

171
00:26:30.060 --> 00:26:31.560
Gerald Raymond: Okay, very good. Okay.

172
00:26:31.680 --> 00:26:42.840
Gerald Raymond: All right. So I'm going to talk today about acid single myelinatis deficiency as we've already started to hear, it's also referred to as Niemann-pick disease.

173
00:26:43.260 --> 00:26:58.009
Gerald Raymond: and I'm going to try and continue to refer to assassin single myelinase deficiency, because Niemann Pick is an older term. Niemann was a pediatrician in Germany, and Pick was a pathologist, and over oh, probably 80 years ago, it was 1st described.

174
00:26:58.140 --> 00:27:19.719
Gerald Raymond: And over the years there's been multiple forms of demon pick described. And that's where it gets confusing. If you go into the historical literature, there is type A type B's type, C's and type D's. And it actually turns out that there's actually 3 diseases buried into this. But we're going to talk about

175
00:27:20.010 --> 00:27:32.330
Gerald Raymond: type A and type B, and as we're going to talk a little bit about A and B. Meld into one another, and sometimes there's a type A and B separated out of that, but making it all a little bit more confusing.

176
00:27:34.150 --> 00:27:45.760
Gerald Raymond: but all types, A, A, B+B are secondary to acid sphingomyelinase deficiency, which is due to a deficiency in the sphingomyelin phosphodiesterase one. G,

177
00:27:47.970 --> 00:27:51.120
Gerald Raymond: okay, so this is everything

178
00:27:51.330 --> 00:28:02.190

Gerald Raymond: that the as me, as I as the expert kind of talk about acid myelin deficiency or asmd is due to variants in Smp. One.

179
00:28:02.480 --> 00:28:22.970
Gerald Raymond: This provides instructions for the enzyme acid single myelinase. This enzyme is found in the lysosomes. It's involved in converting single myelin ceramide. It's an autosomal recessive condition. It has an incidence of about one in 250,000. And actually, I think that's holding up in about one in 40,000 in the Ashkenazi Jewish population.

180
00:28:24.850 --> 00:28:41.289
Gerald Raymond: This is my cartoon about what a lysosomal disorder is, because we just said Asmd is a lysosomal disorder. The lysosome is the recycling center within the cells, and so I don't know if you can see my pointer. But these are the Lysosomes.

181
00:28:41.410 --> 00:29:00.320
Gerald Raymond: They're involved in sort of nutrient regulation and digesting a variety of very complex compounds, and there are lipids, proteins, but one of them is the single myelins, which over here is, we have single myelin being broken down to ceramide

182
00:29:00.450 --> 00:29:17.600
Gerald Raymond: the enzymatic step. Here is the sphingomyelinase, and when that doesn't work, you get accumulation of sphingomyelin, which is seen in this, as you can see in this pathologic cell. So you see this ballooning of cells with very characteristic chemical compounds that were subsequently identified as sphingomyelin.

183
00:29:21.210 --> 00:29:22.070
Gerald Raymond: The

184
00:29:22.180 --> 00:29:37.320
Gerald Raymond: accumulation and severity is a continuum. We're going to continue to talk about this being a continuum. But type A is classically, our infants who develop enlargement of the liver and spleen by around 3 months of age.

185
00:29:37.670 --> 00:29:54.869
Gerald Raymond: With this enlargement of the liver and spleen, you start to get failure to thrive. They're not growing. And by 12 months of age you see progressive loss in sort of neuromotor

developmental issues. And so you start to see. And then you start to see loss of skill, psychomotor regression.

186
00:29:55.170 --> 00:30:04.650
Gerald Raymond: You're also seeing interstitial lung disease which looks like in recurrent infections, pneumonias, and ultimately leads to respiratory failure.

187
00:30:05.130 --> 00:30:05.760
Gerald Raymond: If

188
00:30:06.580 --> 00:30:14.340
Gerald Raymond: the ophthalmologist or eye doctor, and see these cherry red spots, which is this little red bull's eye here on the eye exam.

189
00:30:14.440 --> 00:30:34.650
Gerald Raymond: and that's not the red spot is abnormal. But what is actually happening is, the cells in the retina are dying around this part of the eye, which is called the Maculu, which is actually where we do most of our vision. And so that's the last remnant of the remnant of the retina that's being preserved.

190
00:30:34.780 --> 00:30:39.649
Gerald Raymond: and it's a progressive disorder without without any treatment. And there's usually death in childhood.

191
00:30:40.820 --> 00:30:46.400
Gerald Raymond: This is contrasted to type B, which typically has an onset a little bit later

192
00:30:46.780 --> 00:31:02.430
Gerald Raymond: can present infancy, but usually in mid childhood you start to see similar findings, but not as severe. So the liver starts to get enlarged. You start to see field enlargement of the spleen, and, as you can imagine initially, this is often thought to be secondary to cancer

193
00:31:02.780 --> 00:31:09.880
Gerald Raymond: or a childhood neoplasms. You start to see recurrent lung infections. A fall on the platelet count

194
00:31:10.110 --> 00:31:18.679
Gerald Raymond: short stature and slow mineralization of the bone, which is also seen as delayed bone age, and about a 3rd have that cherry respon.

195
00:31:20.260 --> 00:31:29.660
Gerald Raymond: There is adult survival. And so there are individuals who are would have this condition in adulthood.

196
00:31:31.650 --> 00:31:43.209
Gerald Raymond: As I said, it's a continuum. We have a severe early onset form, which is the neural visceral form, because it has involvement of the brain, which is sometimes labeled as type A,

197
00:31:43.320 --> 00:31:50.219
Gerald Raymond: but we have this later onset, chronic visceral form type B, which typically does not have any brain involvement.

198
00:31:50.550 --> 00:32:11.190
Gerald Raymond: But there is a subgroup of individuals who didn't read the textbook and have what is sometimes referred as chronic, neuro, visceral, Asmd or a slash B, so they have some neurologic involvement, not as severe as the infantile form, but may present with ataxia, motor issues and other manifestations.

199
00:32:15.430 --> 00:32:45.379
Gerald Raymond: There is therapy, as we've heard. There's an enzyme replacement therapy lipidase, which goes by the brand name Zenpozyme, was approved in August of 2022. The little cartoon on the right shows what we are used to seeing for lysosomal storage diseases, and those of us of a certain age can imagine that the little Pac-men are the administered enzyme which gets taken up to the bloodstream, gets taken up by the cells and actually finds its way to the lysosome

200
00:32:46.159 --> 00:32:51.949
Gerald Raymond: and helps to digest and act on the accumulating material.

201
00:32:52.650 --> 00:33:19.089

Gerald Raymond: 4 reasons that are a little complex. It does not impact the neurocognitive issues. Basically, these enzymes which are administered are large molecules, and they cannot cross a part of our body we call the blood brain barrier. The blood brain barrier keeps out large molecules from crossing into into the nervous system, and therefore all of our enzyme replacement therapies do not impact the nervous system.

202
00:33:19.710 --> 00:33:32.490
Gerald Raymond: apparently hematopoietic stem cell therapy or bone marrow transplant. And I'm not going to go through the mechanisms of that. But I can watch poetic about it does correct the metabolic defect, it improves, the blood, counts.

203
00:33:32.620 --> 00:33:38.520
Gerald Raymond: it reduces the liver and spleen volumes, but it. It also does not stabilize neurologic disease.

204
00:33:39.100 --> 00:33:45.370
Gerald Raymond: bone, marrow, transplant or hematopoietic stem cell therapy also has a significant morbidity. Mortality.

205
00:33:45.760 --> 00:33:57.180
Gerald Raymond: significant is in the eyes of the beholder. It's a fairly aggressive therapy, but even in the best of conditions it has a mortality of about 5%.

206
00:33:57.500 --> 00:34:09.239
Gerald Raymond: I do wonder if some of this. Not stabilization of neurologic disease is the fact that many of these individuals come to attention symptomatic. But I'll leave that as a speculation.

207
00:34:12.590 --> 00:34:33.649
Gerald Raymond: As I said, enzyme replacement therapy is available. This was one of the early papers by the group, a multi-center group, led by Melissa Wasserstein, and this paper was published in 2022. And it looked at the use of ellipidase alpha in adults.

208
00:34:33.650 --> 00:34:46.419
Gerald Raymond: I'm just going to share with you one slide of the results, but also comment that about some of the other aspects. And so I'm not going to delve deeply into this slide. But basically we're seeing

209
00:34:46.469 --> 00:35:13.930
Gerald Raymond: one aspect. The red here is the placebo, and the green is those individuals who receive the lipidase, and you can see reductions in spleen volumes. So you see that the placebo individuals. Their spleen stayed about the same. And then for a large group who received the therapy, we see a reduction in the spleen volumes, and with that reduction in the spleen volumes we see increases in the platelet counts.

210
00:35:14.040 --> 00:35:35.679
Gerald Raymond: When you look at some of the other manifestations, you also see reductions in the liver size and interstitial lung disease improve, and so all of the key visceral aspects of this disorder improve in a placebo-controlled fashion or placebo control trial on therapy.

211
00:35:35.920 --> 00:35:41.369
Gerald Raymond: indicating that it was highly. It is highly effective therapy for the visceral manifestations.

212
00:35:42.980 --> 00:35:45.250
Gerald Raymond: Oops excuse me

213
00:35:45.390 --> 00:35:53.679
Gerald Raymond: so that brings me to newborn screening, for as a single myelinase deficiency, and I will try to avoid. Use the eponyms. But

214
00:35:54.430 --> 00:35:56.059
Gerald Raymond: it's not always going to be possible.

215
00:35:56.310 --> 00:36:19.709
Gerald Raymond: It is available. It is possible, through tandem mass spec, you can measure the enzyme levels in the dried blood spots. You basically measure the activity in the dry blood spots. You can also do second tier quantification of lysosphingomyelin, or you can do asmpd 0 1 gene analysis.

216
00:36:19.980 --> 00:36:29.940
Gerald Raymond: It is not on the rust. I did not. I was not even aware it had been submitted to the rust. It's presently only being screened in Illinois and New Jersey, but it's

217
00:36:30.050 --> 00:36:55.440
Gerald Raymond: and that's and there's a little bit of wiggle in here because it's also being piloted in New York through something that's called screen, plus, which is sort of New York's way of getting around trying to get everything, added they, you can opt screen plus is, you can opt into new one screen for a variety of conditions including including asnd and a variety of other things in New York State by opting into the screen. Plus.

218
00:36:57.160 --> 00:37:00.950
Gerald Raymond: So what is the experience? For newborn screening?

219
00:37:01.140 --> 00:37:08.090
Gerald Raymond: There's a published paper out of the experience of the group in Illinois, this was published in 2024

220
00:37:08.410 --> 00:37:16.190
Gerald Raymond: Illinois piloted it from about 11 1 2014, through the end of May, in 2015, and they

221
00:37:16.700 --> 00:37:42.570
Gerald Raymond: examined at that time in the pilot 93,792 infants, with one infant identified and then expanded it to statewide shortly thereafter, one day after 6, 1, 2,015, through July 31, st 2023 is when they closed their examination for this paper, and they screened approximately 1,000,100 individuals, and they had a 9 additional infant screen. Positive.

222
00:37:42.570 --> 00:37:52.729
Gerald Raymond: interestingly for those of us who deal with a lot of like sole storage diseases and in newborn screening. They had no borderline results. So there was no.

223
00:37:52.900 --> 00:37:59.839
Gerald Raymond: there was no one sort of you were kind of equivocal about you, were you either had it or you didn't.

224
00:38:00.100 --> 00:38:13.210
Gerald Raymond: 8 out of 10 were determined to be type B, which I think is pretty impressive. 6 infants had bialelic pathogenic variants. At least 4 of them had at least one bus

225
00:38:13.630 --> 00:38:23.990
Gerald Raymond: and and kind of working against that idea of doing a second tier using else. Lsm, 6 out. The 6 had normal Lsm zipper.

226
00:38:24.240 --> 00:38:32.750
Gerald Raymond: This was a disease in incidence of about one in 126,000 or 0 point 7 9 in 100,000 births.

227
00:38:35.010 --> 00:38:42.080
Gerald Raymond: So so the next question, I think, as a newborn, screening person, we need to ask, is.

228
00:38:42.190 --> 00:38:51.718
Gerald Raymond: what is the what difference does it make if we identify them early versus identifying them a little later when they become symptomatic and

229
00:38:52.270 --> 00:38:56.931
Gerald Raymond: this paper here tries to get at that. So this is 2 siblings.

230
00:38:57.780 --> 00:39:07.079
Gerald Raymond: one was identified at slightly earlier age than than 7. I think he was identified actually around 5 years of age. But don't quote me because I didn't.

231
00:39:07.544 --> 00:39:22.530
Gerald Raymond: I'm just forgetting at the moment. But enzyme replacement wasn't available until he was 7 years of age. He had a younger sib who could meet, who, when identified, could immediately go on enzyme replacement at 3 years of age.

232
00:39:22.550 --> 00:39:37.790
Gerald Raymond: So the younger sib had no deceleration in growth, and the older sib, because he was a little bit delayed, actually developed some lung disease and some organomegaly. However, when I was reviewing the paper, it wasn't very clear to me

233
00:39:37.840 --> 00:39:40.020

Gerald Raymond: that the older individual

234
00:39:40.210 --> 00:40:06.860
Gerald Raymond: suffered any irreparable harm in the sense that at least in the window, that that individual was identified and went with treatment. They both came to essentially the same place. Their spleen sizes are the same, their liver sizes are the same, and the older and the older one actually had ketchup growth. So it's it's not 100% clear to me that identify them, identifying the visceral forms.

235
00:40:07.510 --> 00:40:10.700
Gerald Raymond: Overall results in irreparable harm.

236
00:40:11.430 --> 00:40:15.849
Gerald Raymond: So in summary, oh, sorry in summary, hey?

237
00:40:15.930 --> 00:40:28.550
Gerald Raymond: Acid singomyelin disease, deficiency is a lysosomal storage disease that presents as a continuum of severe early onset, infantile, neurovisceral type, a. Through a later onset, chronic visceral form. Type. B,

238
00:40:28.610 --> 00:40:45.250
Gerald Raymond: there's an approved therapy. FDA approved available lipidase or Zenpazine for visceral manifestations does not get into the Cns. It is not going to correct those aspects. I do wonder if identification of

239
00:40:45.300 --> 00:40:54.470
Gerald Raymond: some of these, the type A's at an earlier age would allow you to do bone marrow transplant. But that's a research question.

240
00:40:54.510 --> 00:41:20.030
Gerald Raymond: I find it an interesting research question. But it is still a research question. It is detectable through newborn screening, using available technologies. Experience suggests it's accurate with low levels of false positives. And there's a limited perspective experience suggesting early detection. Treatment offers moderate benefits in terms of preventing growth, lung disease and hepatosplenomegaly, and with that I will stop sharing.

241

00:41:23.240 --> 00:41:25.110
Gerald Raymond: Did I truly stop sharing.

242
00:41:25.860 --> 00:41:27.340
Mikaela Miller: You did.

243
00:41:27.350 --> 00:41:30.000
Gerald Raymond: All right. It's successful always.

244
00:41:30.460 --> 00:41:34.032
Gerald Raymond: It's like, how long have I been using zoom.

245
00:41:34.430 --> 00:41:55.430
Mikaela Miller: I know you just can never plan for issues sometimes, though, you just never know let me go ahead and check here. We? Normally, we're going to have a break here. But I think, since we're a little bit ahead of schedule, let's go ahead and listen to the family story first, st if that works okay for everyone.

246
00:41:56.320 --> 00:42:07.749
Mikaela Miller: I see some head nods. All right. Well, I will go ahead and let you start sharing your screen, Dr. Hopkin. If you want to go ahead and take over the introduction.

247
00:42:07.990 --> 00:42:08.630
Mikaela Miller: Jen.

248
00:42:08.630 --> 00:42:30.169
Jenn Banna MTF2FHIC: I'll go ahead and introduce him. Yeah, so thank you for being with us today. Dr. Hopkin. Dr. Justin Hopkin is a rare disease parent, physician and advocate. As chief of the Hospital Medicine Division at the University of Rochester. He provides strategic and operational leadership with responsibility across the division's missions. He is vice chair of the Board for uplifting Athletes.

249
00:42:30.170 --> 00:42:56.740
Jenn Banna MTF2FHIC: Chief Scientific Officer for the International Niemann Pick Disease Registry and an Emeritus Board member for the National Niemann Pick Disease Foundation, the patient support organization for patients and families. With Niemann pick disease as an

advocate he works to support and empower patient communities. He's particularly interested in promoting collaboration between patients industry and regulators with a focus on drug development, clinical trial design, patient-owned registries and newborn screening.

250
00:43:02.690 --> 00:43:23.250
Justin Hopkin: Thanks, Jen, I'll come off mute. Yes, I've been using zoom a little while. I appreciate the opportunity to talk to the group. I am a Wyoming native still. Consider myself a Wyoming native. Have the Yellowstone photo in the background, and then have some fish over here that I used to fly fish, for in Montana and Wyoming

251
00:43:23.250 --> 00:43:47.760
Justin Hopkin: I'm a Wyoming whammy. I don't know if everyone on the call has heard of the Whammy program, for which you have many outstanding physicians in Montana. I loved hanging out with the Montana group, actually was married in Bozeman, and so I feel like I'm among my people. I thought I would be practicing primary care and being the county health officer in this community, which is Lander Wyoming.

252
00:43:47.760 --> 00:44:11.549
Justin Hopkin: near Jackson for a number of years. This is where I started my career after completing my training in Denver and Seattle. But life happens. This was me. Many years ago I was officiating my brother's wedding on this day, and this was the day after I found out that the child that I was holding in my hand. Who is Garrett, who's a little over 12 months of age?

253
00:44:12.033 --> 00:44:21.126
Justin Hopkin: Was diagnosed with the disease we're going to be discussing today. There are 5 carriers listed in this photo as well. My parents and my brother and

254
00:44:21.590 --> 00:44:45.719
Justin Hopkin: my other 2 kids and my wife are in this photo. The carrier status was just known by my kids because we looked at the stem cell transplant plant that was just discussed as a possible treatment for someone with a more severe type of Niemann pick disease. It was actually my lovely bride who noticed that Garrett was not on the same trajectory of growth and development as his siblings around 5 or 6 months of age.

255
00:44:45.730 --> 00:45:06.259
Justin Hopkin: He wasn't sitting up at the same age. He wasn't eating well, he was not a good sleeper at all, and we blame this on several things, including ear infections, reflux, and the like.

Our diagnostic journey was much shorter than those in the community because of an outstanding clinician who thought outside the box a little bit.

256
00:45:06.260 --> 00:45:16.429
Justin Hopkin: he closely examined Garrett, identified his spleen, was enlarged. We thought we were dealing with the cancer because of the low platelet count and the enlarged spleen, as was discussed earlier.

257
00:45:16.530 --> 00:45:43.090
Justin Hopkin: But we ran into a fantastic clinician at Colorado children's a couple weeks later, who said, with interstitial lung disease, with this neurologic abnormality, the enlarged plane along enlarged liver, I think you have a metabolic disorder. And so the day before this picture was taken, my wife and I learned that Garrett had the neurologic form of Niemann-pick disease likely type A is what we were told

258
00:45:43.327 --> 00:45:47.599
Justin Hopkin: and that his lifespan would likely be on the order of a couple of years, and we should

259
00:45:47.600 --> 00:45:57.569
Justin Hopkin: think about engaging with palliative care at that point. So neiman-pick disease. This slide comes off of Nord's website.

260
00:45:57.630 --> 00:46:15.769
Justin Hopkin: and it's been there quite a while, and it talks about the acid sphingomyelinase enzyme activity as potentially being the differentiator between the type A, which they say is a complete absence of enzyme, especially in the central nervous system versus type B, which is, you have some residual amount.

261
00:46:15.770 --> 00:46:29.270
Justin Hopkin: the enzyme can be secreted. So you don't need a lot of enzyme to avoid significant neurologic symptoms. So that may be the case. But, as I mentioned earlier in my talk, it's a spectrum disorder.

262
00:46:29.270 --> 00:46:54.080
Justin Hopkin: And because lysosomes and cells exist in all parts of our body, all parts of the body are impacted by this disease. The ones that stand out are the ones you can see on

physical exam and just looking at a patient, which is what I'll show you here in a minute. But the enlarged liver and the large spleen are really incredibly remarkable, and for somebody who's been in medicine now for a couple of decades. I've never seen anyone with a liver spleen

263
00:46:54.080 --> 00:46:55.580
Justin Hopkin: as big as my son's.

264
00:46:55.600 --> 00:47:08.920
Justin Hopkin: The lung disease is marked. The amount of neurologic disease is variable across all patients, and we saw pictures of the back of the eye, the retina, which is the gateway into the central nervous system, and even those patients

265
00:47:08.920 --> 00:47:31.070
Justin Hopkin: who don't have a lot of neurologic disease that maybe adults often have abnormalities there or peripheral neuropathy. And so I think we're understanding some of the spectrum of the disorder as far as neurologic disease is concerned. But there's definitely a patient population with very significant disease throughout the body, including the brain that have a shortened lifespan, and we'll talk a little bit more about them.

266
00:47:31.870 --> 00:47:53.909
Justin Hopkin: My son probably lands in the middle of this graph, now that I know him and have seen him develop in that sort of didn't read the textbook Ab, or chronic neurovisceral presentation, the type A, or what we now call infantile neurovisceral. We discussed earlier, and the type B is in the middle.

267
00:47:54.390 --> 00:48:07.189
Justin Hopkin: After knowing many of the patients in the community and seeing virtually all the the patients that were born, not only in the Us. But probably in a large part of the the western part of Europe.

268
00:48:07.190 --> 00:48:16.450
Justin Hopkin: There are very few patients that fit neatly into 2 or 3 of these categories, and it's really a spectrum disorder. So I led a group that

269
00:48:16.450 --> 00:48:39.399
Justin Hopkin: looked at changing the name from type A or type. B, as far as the Icd codes are concerned, to make it more acid sphingomyelinase deficiency, one is so. Clinicians would be

able to differentiate it from the 5 other types, because it's very confusing. But the other is that this is a spectrum disorder, and we wanted treatments to be available to all patients because it was really hard, clinically.

270
00:48:39.600 --> 00:48:52.400
Justin Hopkin: for even the most astute clinicians like Melissa Wasserstein, to identify which patient had the most severe form, had an intermediate form or a different form, especially early on in life. And so we were very happy that this made it through.

271
00:48:52.550 --> 00:49:04.720
Justin Hopkin: This is a study that was done primarily in Europe, looking at patients excluding those type, a or severe patients that looked at the age of onset.

272
00:49:04.790 --> 00:49:34.340
Justin Hopkin: which is on the vertical axis and the age of death in those patients that had passed, and you can see that the age of onset of symptoms is very, very low in many of the patients who pass from this order regardless of when they pass in the 1st 1020, or 30 years of life. But even those without the significant neurologic symptoms, there are many who die before the age of 10, most who die before the age of 20, and symptom onset is almost always in the 1st few years of life.

273
00:49:34.460 --> 00:49:52.789
Justin Hopkin: And there's a lot of other studies that have been done, including those in the Us. That replicate this sort of scenario for those patients with the severe form of the disorder. This is the list of all of the symptoms they have. I'm not going to go through this entire chart, but I'll just tell you that in addition to the neurologic symptoms.

274
00:49:52.790 --> 00:50:09.019
Justin Hopkin: almost all of them have significant visceral disease. So the lungs are heavily involved. The gi tract is heavily involved in those that pass, and so ameliorating those symptoms will probably be helpful, not only in symptom management, but hopefully prolonging life.

275
00:50:09.300 --> 00:50:17.050
Justin Hopkin: So this is Garrett, this is a little while later. This is a video of him in our living room. And there's a few things I would point out.

276
00:50:17.730 --> 00:50:42.739

Justin Hopkin: Sleep was a real challenge, and one of the reasons sleep was a challenge was because nutrition was a challenge, and that was mostly because he has this huge spleen and huge liver that take up his entire abdomen. So whenever he tried to eat solid food he vomited. So we, after our 1st visits, learned that a liquid diet may stay down a little bit better, and so he lived off of insure pediasure for his 1st several years of life.

277
00:50:43.356 --> 00:51:03.553
Justin Hopkin: And he would wake up multiple times just to try and keep up with the calories he needed to grow, but also the calories to keep his liver and spleen functioning because they were 20. His spleen was many, many times its normal range, and so was the liver. You can see that as he walks around here, and I'll try and start the video.

278
00:51:04.090 --> 00:51:14.309
Justin Hopkin: his ribs are are still showing quite a bit, not very big, despite having really big organs and moving around is a challenge. We didn't know if it was the ataxia from the neurologic disease.

279
00:51:14.820 --> 00:51:23.260
Justin Hopkin: Nothing, or or if it was re related to just carrying that huge, those huge organs around

280
00:51:23.260 --> 00:51:46.039
Justin Hopkin: that exact spot is where he fell about a year later, playing with his brother, and ended up in this condition in this contraption, which is a spicas, I'd never heard or seen a spicas, but fractures and bone thinning was brought up. This is one of the many hospitalizations we had. Unfortunately, we had to use life flight a few times, living in rural Wyoming. We know what that's like.

281
00:51:46.040 --> 00:52:07.119
Justin Hopkin: and his cast, as you can see, covered his abdomen as it does in most kids with this disease. But because of the way he was built he couldn't breathe with that cast on, and you'll notice the photo. On the right was a window was cut into that cast by my local orthopedic surgeon when we got him home, and he went back into respiratory failure after being discharged from children's.

282
00:52:07.120 --> 00:52:20.009

Justin Hopkin: A quick, thinking, orthopedic surgeon probably saved his life. And so we're very appreciative of that. We did also find that a Cpap was helpful in trying to help a kid breathe who really had no room to move his lungs because

283
00:52:20.090 --> 00:52:22.419
Justin Hopkin: everything was full of organs for him.

284
00:52:22.760 --> 00:52:45.189
Justin Hopkin: I spent most of the early years advocating for him to get access to this lifesaving therapy that I had heard about and read about, flown out to try and convince Genzyme to give my son access to. It's this enzyme. And this study was dated Pre. 2,000. So this was 20 years in the making of a enzyme replacement therapy that offered promise to my son.

285
00:52:45.530 --> 00:53:01.279
Justin Hopkin: The trial enrollment was graded in different age groups, and I know the 2 siblings that were involved in the case study that was just described, and the younger sibling actually got into the trial, and the older sibling didn't the 1st time around. So that's why the younger sibling got got treated first.st

286
00:53:01.350 --> 00:53:24.710
Justin Hopkin: We were trying to get in the trial, and the only trial site in North America was at Mount Sinai, in New York City, where Melissa Wasserstein and George Diaz were practicing. And so we flew out multiple times. This is a flight we had to take back across the country when the radiologist thought the MRI was too blurry to interpret after we completed the initial screening, and so we had just taken a red eye flight.

287
00:53:25.988 --> 00:53:34.060
Justin Hopkin: at this point we met some friends in New York City, and Garrett looked a little peak at hanging out with some of my friends. I didn't think much of it, but on the flight back home.

288
00:53:34.750 --> 00:53:35.510
Justin Hopkin: hey?

289
00:53:36.530 --> 00:54:01.289
Justin Hopkin: He looked a little bit worse and less than 24 h after that photo on the 1st slide was taken. This was Garrett, sitting in the Icu at Colorado children's with influenza and those of

you that remember the last time we had the nasal form of the flu shot that was this year. It didn't work so well, and it didn't work so well for Garrett. He went into respiratory failure.

290
00:54:01.750 --> 00:54:20.559
Justin Hopkin: He had a C. Diff infection. His bone marrow went into failure, so he was what we call pancytopenic and neutropenic, and it was during this hospital stay that we learned that he was accepted into the clinical trial. So we worked really hard with the team there.

291
00:54:26.230 --> 00:54:33.915
Justin Hopkin: talked about this a few times. You think I'd be able to get through this? They worked very hard to get him through this hospital. Stay,

292
00:54:34.210 --> 00:54:57.070
Justin Hopkin: and we're very fortunate to get him out of the hospital a few days before his 6th birthday, when we were supposed to fly from from Jackson Hole out to New York City to get him in the trial. We're very fortunate that once you land at sea level the partial pressure of oxygen is better because he needed oxygen for the flight. But when we took it off for the enrollment he no longer needed it.

293
00:54:57.346 --> 00:55:05.089
Justin Hopkin: I think I just wanted to show the video, showing just how hard it is for him to breathe. And we talked about the the pulmonary disease, often

294
00:55:05.410 --> 00:55:14.420
Justin Hopkin: being the reason that many of these kids pass, and you can kind of see why he had interstitial lung disease. And he just was having a really tough time fighting this. So he was on

295
00:55:14.530 --> 00:55:21.799
Justin Hopkin: pressure ventilation for a week through this illness. But a great team took care of him. And 2 weeks later.

296
00:55:22.732 --> 00:55:23.325
Justin Hopkin: He

297
00:55:25.690 --> 00:55:35.739

Justin Hopkin: He got into the clinical trial, and which was great, but the clinical trial required us to be present in New York City every 2 weeks for what we thought would be a year

298
00:55:36.400 --> 00:55:39.640
Justin Hopkin: and so we kept our house

299
00:55:39.670 --> 00:56:04.949
Justin Hopkin: in Lander. I kept my place in our practice, and we found a place that we thought would be temporary, which was Rochester, New York, because we had friends here that I trained with, and we traveled every 2 weeks to New York City. I was told that a country boy like myself would not do well living in Manhattan with a family of 5, so we settled on the Midwest feel of Rochester, New York, thinking that this would be a short experience.

300
00:56:05.271 --> 00:56:26.458
Justin Hopkin: So that the photo in the middle is our family as we took the move out to New York City or to Rochester. Sorry after I drove the kids across the country. That's a picture of Garrett 6 months into the trial, and you can already see his abdomen is markedly improved from from where it was before. And then I took this photo

301
00:56:26.860 --> 00:56:53.810
Justin Hopkin: at the World Symposium this year, which is the Research Conference for enzyme or for lysosomal storage disorders, and that one year clinical trial turned into 6 years before we would get an approval. And so, in order to maintain Garrett on a life sustaining therapy, the short one year that we thought we would spend in Rochester has turned into 8 years to maintain access to a life-saving therapy for my son

302
00:56:53.920 --> 00:57:06.820
Justin Hopkin: as a side hustle. If anyone's interested, my older 2 kids just were featured in an article about the sibling experience of what that's been like for them to be moved across the country away from friends and family. It's a good read.

303
00:57:07.440 --> 00:57:26.249
Justin Hopkin: This is what Garrett looks like with 3 other people who live with this disorder. You see, you read about a spectrum in the presentation earlier. The person on the far left has been part of the original clinical trial, and on the therapy for 12 years she now competes in triathlons.

304
00:57:26.280 --> 00:57:54.919

Justin Hopkin: The person with red hair is an occupational therapist in Toronto. She had her aortic valve replaced, has terrible lung disease and cirrhosis, and is our president of our international Neiman-pick Disease Alliance. She's an amazing human. And then one of our most amazing advocates is on the far right. He's Garrett's good friend Everen, who is now getting his master's in special education, and he gives amazing talks on what it's like to live with this disorder

305
00:57:56.142 --> 00:58:01.240
Justin Hopkin: Garrett's living his best life. We are in an amazing supportive community.

306
00:58:01.780 --> 00:58:11.819
Justin Hopkin: and the enzyme therapy is probably among the best. From what I'm told, as far as treatments for lysosomal storage disorders.

307
00:58:11.820 --> 00:58:40.909
Justin Hopkin: It literally melts the disease outside the central nervous system. It actually wasn't studied to look at anyone with Cns disease, because they were all excluded from the trial. My son is among probably the 2 sickest patients with the most significant neurologic disease to be included in the trial, and his neurologic disease has been stable for 8 years receiving enzyme replacement therapy. So there are some things we don't understand, but it doesn't. It's not supposed to cross the blood brain barrier because it's too big. But he's done pretty well with this.

308
00:58:42.040 --> 00:59:01.959
Justin Hopkin: I'm going to spend just a little bit of time. You can cut me short if you need to. But I want to talk about some of my advocacy priorities and some of the things I'm working on in particular, global access to this therapy is desperately needed, so it's an expensive therapy. Fortunately, everybody in America who wants access to this therapy has it. It's been very well covered.

309
00:59:01.960 --> 00:59:17.510
Justin Hopkin: I think understanding. The sibling experience is important. I won't talk as much about that, but we need to identify treatments for the brain, and I will talk about that expanding newborn screening is really important. And we have an amazing patient registry which I'll go into just briefly.

310
00:59:17.730 --> 00:59:33.880

Justin Hopkin: this is the test that is being used in states like New Jersey and Illinois, but many other states that are screening for other diseases, like Pompeii, Krabe, and Mps, one that are Rusp included.

311
00:59:33.880 --> 01:00:01.889
Justin Hopkin: So this is sold by 2 companies, Revedy, and then Gelp chem. Michael Gelp's an amazing clinician who did the original studies on this test, and, as was said, it works really well, it's got high sensitivity and specificity. Most states that use this product are actually blinding the results for Niemann pick disease. So they're using it. It's no additional cost to just look at the results. They just want to make sure it's a high quality test and they have support to use it afterward.

312
01:00:02.170 --> 01:00:22.089
Justin Hopkin: We've already talked about the Illinois experience. I'm not going to waste any time other than to let you know that a few of those patients are going to be evaluated, or are likely on therapy, and an update should be coming soon on their decade of screening for the disorder. But it's a very effective test at this point.

313
01:00:22.618 --> 01:00:31.840
Justin Hopkin: Having good clinical guidelines is really important if you have a treatment. And so I was fortunate to be part of the group that pulled these international guidelines together.

314
01:00:31.990 --> 01:00:57.969
Justin Hopkin: and in that guideline we clearly identify how a patient should be diagnosed, and then who should be considered for treatment, and among many other things. And this is just the graph that is included in that guideline that talks about how you diagnose a patient. Usually we recommend biomarkers and gene sequencing, but based on the Illinois experience. That test seems to be a very predictive test as far as who has the disease

315
01:00:57.970 --> 01:01:07.320
Justin Hopkin: and limits the number of false positives from what we know so far. But obviously we would need to do more studying as hopefully, more states and more places. Look at screening for this

316
01:01:09.000 --> 01:01:16.820
Justin Hopkin: as was discussed. The the after 20 years, 22 years exactly in the pipeline. This

317

01:01:18.055 --> 01:01:20.049
Justin Hopkin: therapy was approved.

318
01:01:20.130 --> 01:01:38.959
Justin Hopkin: It was approved, not for certain types of the disease, but it was approved to treat symptoms of the disease, and this is in contrast to the other countries that are looking at this, and we believe that the reason that there is a contrast was because of the work we did as an advocacy organization

319
01:01:38.970 --> 01:01:47.709
Justin Hopkin: to explore the experience of patients and families who who took part in this study, and we included patients who were

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01:01:47.710 --> 01:02:11.320
Justin Hopkin: included in the trial, but also those who are on managed access programs, and of the patients that were included, there were 3 with significant neurologic disease that were included, some who had seizures and considered to be classic type, a patients. And what we found is that the amelioration of symptoms of the lung disease, the Gi disease, to allow patients to get off oxygen to allow them to tolerate tube feeds

321
01:02:11.320 --> 01:02:31.800
Justin Hopkin: was a marked improvement for those patients, and we actually captured that cohort in a poster that we presented at an international meeting in Portugal this last year, and those 3 patients who have pretty severe disease, all of whom probably would have been expected to pass are alive after the age of 5.

322
01:02:33.020 --> 01:02:58.790
Justin Hopkin: we're capturing their disease and a study that's being done by a nonprofit known as the Wilder Nation Foundation. There's a picture of Wilder there. There's an online registry through picnic health that we're using. And we're gathering more data on these patients to better understand the disorder and hopefully identify good ways. We can treat them. And here's a few of those warriors who passed before there was any sort of treatments available, and those are the ones that are on my mind as we think about

323
01:02:58.790 --> 01:03:03.069
Justin Hopkin: newborn screening and what we can do for those with significant symptoms. Early on in life.

324
01:03:05.460 --> 01:03:14.620
Justin Hopkin: the FDA. When they approved this, they approved the enzyme replacement therapy starting at age 0 or birth for those visceral symptoms.

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01:03:14.620 --> 01:03:38.759
Justin Hopkin: and they want to continue to capture what the experience is for those that are young with significant symptoms. So we're recruiting a trial to see what enzyme replacement therapy looks like. And this just started within the last year. We've already recruited 60% of the patients they hope to recruit in the 1st 2 years. And so there's more information coming on those with severe symptoms that have early age of onset, and we look forward to hearing about those patients that are being captured

326
01:03:38.760 --> 01:03:40.680
Justin Hopkin: outside of newborn screening.

327
01:03:40.680 --> 01:03:59.429
Justin Hopkin: I have 3 quick slides to talk about the clinical programs that are out there that are looking at treating the Cns disease. This is looking at the Endocannabinoid system. And again, research that was funded by the Wilder Nation Foundation identified that if we

328
01:03:59.600 --> 01:04:24.589
Justin Hopkin: I give the mice a blocker of this endocannabinoid system. They had better lifespans and neurologic outcomes compared to those that don't. And there was a lot of research that went into identifying that there are at least 6 patients that have had access to this therapy that appears safe. There is not an active clinical program that has published any of the data on this. It's hard, because these are all n of one studies.

329
01:04:24.590 --> 01:04:35.320
Justin Hopkin: But the animal data and the basic science data appear to be very promising for a potential therapy to address the Cns symptoms. This slide was presented

330
01:04:35.410 --> 01:04:53.250
Justin Hopkin: on a drug that was approved for the other type of Niemann pick disease type C, as potentially a platform therapy, meaning that it's not a therapy that is mechanistically meant just to treat Npc. It's just to treat inflammation in the lysosome and hopefully help the mitochondria and the lysosome work better.

331
01:04:53.250 --> 01:05:09.779
Justin Hopkin: And in theory this should work just as well in Asmd as it should in Npc. And they're looking at it in another couple of different drugs. And there are multiple Asmd patients who are now trying to get access to this newly approved therapy in Npc. Which we anticipate to work in Asmd.

332
01:05:09.810 --> 01:05:27.799
Justin Hopkin: And the last thing I'll say is that Regeneron presented at the same conference in February that they're looking to use gene therapy. This is a very fancy slide, a lot of science here. But essentially, they're going to use a Crispr technology and nanoparticles to inject something

333
01:05:27.800 --> 01:05:41.609
Justin Hopkin: that will cause the liver to produce the protein that can cross the blood, brain, barrier and deliver enzyme into the brain. And this would be just amazing for patients to be able to do. And again, they have mice data to look at that.

334
01:05:42.100 --> 01:05:58.970
Justin Hopkin: A couple last comments. We think we have a biomarker that may be helpful in identifying patients with more severe disease. It's lysosphingomyelin. Melissa, Wasserstein, and others worked on this paper a number of years ago that showed that patients with more severe disease had higher lysosphingomyelin levels.

335
01:05:59.020 --> 01:06:12.180
Justin Hopkin: those levels also all improved with treatment of the enzyme replacement therapy in the patients that were included in the clinical trial that was discussed earlier. Here's that paper. Here's the older 2 kids.

336
01:06:13.030 --> 01:06:32.589
Justin Hopkin: And so if you want to, Google, rare evolution and the sibling experience, it's a pretty interesting. Read, it'll take you all of about 15 or 20 min, but I'm pretty proud of their honesty and their transparency about what their experience has been living like having to move across the country and living in a family that deals with a rare disease.

337
01:06:33.490 --> 01:06:46.659

Justin Hopkin: There's the crew in Idaho this last year, when we were out for a family wedding. As you can see, Garrett looks pretty good and much better than the 1st few photos I shared, so we're very thankful to have a

338
01:06:46.930 --> 01:06:54.999
Justin Hopkin: a therapy that is very effective. And so with that I will stop sharing, appreciate the extra time that you probably gave happy to answer questions.

339
01:06:59.340 --> 01:07:06.581
Mikaela Miller: Yeah, thank you for being here. And sharing all of that with us. It's perfectly fine. You went a little over. We usually try to

340
01:07:06.860 --> 01:07:31.849
Mikaela Miller: schedule some buffer time there, because we're so grateful for the information you're able to provide for us. We were normally going to take a break here in about 10 min. So I say, let's go ahead and do the lab presentation. We can take a 10 min break, and then we'll go into the discussion. If that works for everyone, Dr. Hopkin, Dr. Raikman, you're welcome to go. If you

341
01:07:31.850 --> 01:07:40.040
Mikaela Miller: have other pressing matters to get to. Otherwise we're happy to have you here for that discussion. In the event anyone has any questions.

342
01:07:42.850 --> 01:07:49.612
Mikaela Miller: Alright, I'm gonna go ahead and share my screen so that we can see the information

343
01:07:50.180 --> 01:07:53.230
Mikaela Miller: Jeannie is going to share with us.

344
01:07:54.470 --> 01:08:02.832
Jeanne Lee: Yeah, Hi, everyone. Thank you. Dr. Raymond and Dr. Hopkins, for your presentations today

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01:08:03.900 --> 01:08:20.249

Jeanne Lee: I'm Jeannie Lee, and I'm the newborn screening and serology supervisor in the Montana Public Health Laboratory. My role for this committee is to provide you with information on how testing can be accomplished

346
01:08:20.260 --> 01:08:39.930
Jeanne Lee: for conditions that are brought before you, and the cost implications for that testing. But before I talk about that today, and since we have a few new folks, I wanted to briefly describe and give you some background information about newborn screening in Montana.

347
01:08:42.729 --> 01:09:00.460
Jeanne Lee: all of Montana's newborn screening samples come to the Montana Public Health laboratory by our courier service from 16 facilities, or by overnight shipping through carrier services like Ups or Fedex.

348
01:09:00.939 --> 01:09:14.630
Jeanne Lee: the newborn blood spot. Samples are received Monday through Saturday, and testing begins here in the laboratory for 7 of the 31 blood spot conditions on the Montana panel.

349
01:09:15.029 --> 01:09:25.790
Jeanne Lee: That means 24 conditions are tested at the Wisconsin State Laboratory of hygiene, where we ship samples daily excluding Sundays.

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01:09:26.300 --> 01:09:31.360
Jeanne Lee: You may be questioning why we send testing to Wisconsin

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01:09:31.770 --> 01:09:46.039
Jeanne Lee: through the nineties and early 2 thousands. There was an explosion of newborn screening conditions to screen for, and the Montana Public Health Laboratory was not able to bring on testing because of funding

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01:09:46.710 --> 01:09:59.749
Jeanne Lee: Montana. Public Health Laboratory is not funded through State legislature, but rather is a fee for service laboratory meaning. We cover our costs through testing fees.

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01:10:00.010 --> 01:10:13.320

Jeanne Lee: So in 2,007. When the Montana Legislature mandated additional tests to be added to the panel. The Montana public health lab contracted with Wisconsin

354
01:10:13.460 --> 01:10:21.600
Jeanne Lee: to do this testing for fees that would cost less than we could then we could bring on ourselves

355
01:10:22.540 --> 01:10:35.160
Jeanne Lee: when bringing on new tests. There are many things to consider like instrumentation, expertise and methods whether there's an FDA approved test and what the cost will be.

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01:10:36.000 --> 01:10:39.030
Jeanne Lee: So here we are with Asmd.

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01:10:39.520 --> 01:10:59.199
Jeanne Lee: We currently have Pompeii disease in the rulemaking process to add to Montana's newborn screening panel, and we have decided that we will have Wisconsin perform this test for us. This is because we're not in a position to purchase instrumentation.

358
01:10:59.360 --> 01:11:03.589
Jeanne Lee: Nor do we have the laboratory space to bring on new instruments.

359
01:11:04.200 --> 01:11:10.840
Jeanne Lee: With the remodel that is happening. Now, however, this is something we can consider for the future.

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01:11:11.470 --> 01:11:38.670
Jeanne Lee: So later, this year, or early 2026, with Pompei disease added to the panel, Wisconsin will begin testing. Asmd is a test that can be multiplexed with Pompei disease. Multiplexing means that Asmd can be tested alongside Pompei disease. Using the same blood spot punch.

361
01:11:39.320 --> 01:11:49.100
Jeanne Lee: I reached out to Wisconsin, and even though they don't currently run this test on their panel, they could validate it and perform it for us.

362
01:11:49.280 --> 01:11:55.840
Jeanne Lee: I didn't get a straight answer on the cost for adding Asmd alongside Pompeii.

363
01:11:56.540 --> 01:12:06.179
Jeanne Lee: even though it shouldn't be much of an addition to the price. I estimate that it might cost up to $15 per test.

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01:12:07.440 --> 01:12:16.390
Jeanne Lee: Since Montana's newborn screening panel is currently $150 and 50 cents without Pompei

365
01:12:16.860 --> 01:12:27.869
Jeanne Lee: adding Pompeii disease, and Asmd would mean that the new cost could make the panel as much as a hundred $80 and 50 cents.

366
01:12:28.250 --> 01:12:43.359
Jeanne Lee: I want to note here that newborn screening is covered by insurance and medicaid. So families that don't have insurance or medicaid Medicaid would be paying this amount out of pocket.

367
01:12:44.206 --> 01:12:49.230
Jeanne Lee: So I think that's all I have, unless any of you have questions.

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01:12:52.120 --> 01:12:59.099
Justin Hopkin: I, it may be worth noting that I'm also. We're also looking at Wisconsin. I actually presented to their committee this last week.

369
01:12:59.418 --> 01:13:16.160
Justin Hopkin: And they provided some feedback to me. They actually said it would be very little additional cost to run the test, because they're already running it with Pompeii and and the others that they're running on that multiplex as well. So it's interesting to me that they would charge more if you're already doing Pompeii.

370
01:13:16.160 --> 01:13:34.429

Justin Hopkin: And they're already running the test. I'm not sure where the additional cost would be on their end. But I have no idea about the economics of testing and things like that. But we are. We are applying to Wisconsin, May Baker, and and others are giving some great feedback. And so it may be hopefully another state that's considering it.

371
01:13:35.850 --> 01:13:38.799
Gerald Raymond: Are you screening for Mps. One and Mps. 2.

372
01:13:39.870 --> 01:13:44.019
Jeanne Lee: Montana is not screening for Mps. One and Mps. 2.

373
01:13:44.530 --> 01:13:48.319
Gerald Raymond: So are you, would you consider adding them for the since they're on the rust?

374
01:13:49.356 --> 01:13:51.340
Gerald Raymond: Oh, your Wisconsin panel.

375
01:13:52.000 --> 01:14:00.529
Jeanne Lee: We? We are waiting for nominations so that they can be brought before our advisory committee.

376
01:14:05.980 --> 01:14:26.790
Jenn Banna MTF2FHIC: It might be worth noting, Dr. Raymond, that our advisory committee has only been active for less than 2 years, and our legislature just created us as the body to bring recommended conditions forward and to listen to these. So we're we haven't had a lot of time to listen to a lot of conditions, and there hasn't been another way to really nominate conditions until now.

377
01:14:30.000 --> 01:14:33.209
Gerald Raymond: What's 1 final question? Then I'll go.

378
01:14:33.470 --> 01:14:36.049
Gerald Raymond: So are you a rusp mandated state.

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01:14:37.840 --> 01:14:41.269

Jenn Banna MTF2FHIC: But I think we were rusp mandated in 2014.

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01:14:41.777 --> 01:14:56.460
Jenn Banna MTF2FHIC: I think our state in. I don't know. Someone can correct me if he is wrong. But more than 10 years ago Montana passed legislation to bring us up to date to the rusp, and since then there hasn't been any changes until this committee was created and started talking about it. So.

381
01:15:00.020 --> 01:15:01.790
Abe Elias: Yeah, I think that's 1 of the

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01:15:03.050 --> 01:15:08.460
Abe Elias: one of the limitations for our committee. We we need to

383
01:15:09.240 --> 01:15:24.869
Abe Elias: wait for nominations that come from the public through that system in order to consider which has led to us having to consider conditions that you know there's several conditions that we that were not under rusp

384
01:15:25.270 --> 01:15:28.810
Abe Elias: in including Mps. One, for example.

385
01:15:29.090 --> 01:15:42.460
Abe Elias: So the and and not instead of Mps. One. So we had considered, and then recently. Gosh! As well. So we're we're dependent on

386
01:15:43.307 --> 01:15:52.372
Abe Elias: getting a nomination. And those nominations are on a 1st serve 1st looked at basis, basically.

387
01:15:53.580 --> 01:15:55.170
Abe Elias: which is a limitation.

388
01:16:04.890 --> 01:16:10.880

Jenn Banna MTF2FHIC: Okay, Michaela. Then are we ready to take a 10 min break? Come back at 1 30.

389
01:16:10.880 --> 01:16:11.290
Mikaela Miller: Yeah.

390
01:16:11.290 --> 01:16:13.060
Jenn Banna MTF2FHIC: 11 min. Is that okay?

391
01:16:13.060 --> 01:16:20.259
Mikaela Miller: Yeah, I believe. So I was gonna just double check. Dr. Raymond. Did you say that you would be leaving the meeting.

392
01:16:21.991 --> 01:16:24.698
Gerald Raymond: I can stay. I will.

393
01:16:25.540 --> 01:16:27.169
Gerald Raymond: Let's see if there's any questions.

394
01:16:27.200 --> 01:16:36.797
Mikaela Miller: Yeah, I just figured, instead of making you wait through the 10 min break I could open the floor if anyone had any questions for Dr. Raymond.

395
01:16:37.330 --> 01:16:39.089
Mikaela Miller: before he has to go.

396
01:16:46.110 --> 01:16:53.630
Abe Elias: I, if nobody has a question. I wanted to follow up Dr. Raymond on you mentioned

397
01:16:54.570 --> 01:17:09.819
Abe Elias: that paper where the question was, when you what is the difference in outcome when you start Ert at an earlier age versus a later childhood age

398
01:17:09.940 --> 01:17:30.369

Abe Elias: are there, and this was just a, you know, an anecdotal or example. There, do we know a little bit more about this? And and or what's the timeframe when we knew that? And and I asked, because, you know, I think earlier, the question or the

399
01:17:31.050 --> 01:17:56.627
Abe Elias: idea of a diagnostic Odyssey come came up. You know. I think today we would be able to diagnose Niemann. Pick early onset fairly quickly. The way, how our diagnostic in Montana, the diagnostic workflow goes, and and so so their question then is really, if you start treatment

400
01:17:57.700 --> 01:18:06.400
Abe Elias: at age, you know, at a few years earlier, or right, you know, at after birth versus

401
01:18:06.620 --> 01:18:13.570
Abe Elias: a few months or a few years later. Do we know anything more about this in terms of the.

402
01:18:13.570 --> 01:18:18.119
Gerald Raymond: That's the only that's the only I know. There's probably Dr. Hopkin probably has

403
01:18:18.520 --> 01:18:29.600
Gerald Raymond: abstracts or posters out there, but it's the only paper I found that's in the that I could find and sort of the time I allowed to prepare this talk the and so

404
01:18:30.440 --> 01:18:38.249
Gerald Raymond: I think that you know, I agree that if someone presented with Splenomegaly and went through the usual things and

405
01:18:39.270 --> 01:18:44.010
Gerald Raymond: we, we quickly get to an exome or genome, and we get the diagnosis.

406
01:18:44.823 --> 01:18:46.130
Gerald Raymond: The the.

407

01:18:47.210 --> 01:18:48.950
Gerald Raymond: The one thing I am

408
01:18:51.520 --> 01:18:54.699
Gerald Raymond: wondering about is the fact that

409
01:18:54.830 --> 01:19:00.489
Gerald Raymond: if we start to diagnose type A's right in the newborn period.

410
01:19:01.511 --> 01:19:11.060
Gerald Raymond: Whether they would be better candidates for bone marrow transplant. And the reason I bring this up is, if you look at the experience with, say, Mps. One

411
01:19:11.440 --> 01:19:17.030
Gerald Raymond: and Ps. One is with the neuropathic form, the severe end of the spectrum.

412
01:19:17.530 --> 01:19:30.059
Gerald Raymond: severe Mps. One s. They don't do particularly well with enzyme replacement therapy. Yes, their their livers and spleens, their airways get better, but they still have neuronal disease. But we know that

413
01:19:30.770 --> 01:19:34.149
Gerald Raymond: bone marrow transplant in that population does make it.

414
01:19:34.390 --> 01:19:37.899
Gerald Raymond: And so, if we were now to transplant type A's

415
01:19:38.413 --> 01:19:42.169
Gerald Raymond: the severe end of the spectrum in the 1st months of life.

416
01:19:42.300 --> 01:19:45.380
Gerald Raymond: with a little bit of enzyme replacement before, but it

417
01:19:45.560 --> 01:19:52.000

Gerald Raymond: they may actually start to look like Mps. Well, the severe Mps ones that we have been transplanting for almost

418
01:19:52.130 --> 01:20:01.872
Gerald Raymond: 20 years. And having relatively reasonable developmental outcomes. And so I I that's, I think that's probably my

419
01:20:02.510 --> 01:20:08.180
Gerald Raymond: I you know I'm I'm a sort of on the fence about

420
01:20:08.310 --> 01:20:19.579
Gerald Raymond: how much it makes for the later forms, because I do think the spleen, the spleen size, and the livers. The lung disease gets better. And so I think it's it's re. It's it's a it's

421
01:20:19.730 --> 01:20:25.489
Gerald Raymond: probably better to be prospectively monitoring an individual rather than hoping that they fall into your lap.

422
01:20:26.020 --> 01:20:30.020
Gerald Raymond: and I think the type A's may actually benefit from this. The most.

423
01:20:33.953 --> 01:20:34.859
Abe Elias: Thank you.

424
01:20:37.570 --> 01:20:39.090
Mikaela Miller: Steven. I see your.

425
01:20:41.020 --> 01:20:59.480
Steven Shapero: Yeah, I just had a quick question for for anybody really, do we know if other States are considering adopting this as part of their new one screening? Or if other States are waiting? Or is this the tip of the spear? What do we know about what other States are doing? If anything.

426
01:21:01.390 --> 01:21:05.780
Gerald Raymond: I'm go ahead, Mr. Hopkins. Dr. Hopkins, you probably know better than I do.

427
01:21:05.780 --> 01:21:23.959
Justin Hopkin: It's a it's a little bit of a longer answer, and if Dr. Raymond's time is pressed, I could answer that maybe after the break because most of it is advocacy based including the rust. But I can tell you sort of where we were at with a refiling of the rust, even though there's the Achdnc being disbanded at all.

428
01:21:23.960 --> 01:21:44.869
Gerald Raymond: The the committee that advises for things to be added to the rust has been has been disbanded. There are actually and because I'm actually on our on our State side, commit Statewide committee. You know, we're there's presently a variety of things being considered to sort of substitute for a rust

429
01:21:44.870 --> 01:22:00.610
Gerald Raymond: to the rusp hasn't gone away, it is still. It is still in regulation, however, getting things added to. It is sort of in limbo at the moment, and so I think it's going to be a little bit of a

430
01:22:01.800 --> 01:22:05.246
Gerald Raymond: and I apologize for using this term. But a Wild West.

431
01:22:05.930 --> 01:22:19.707
Gerald Raymond: to try and it's for the next couple years, couple of years. Until we have something in place that helps us codify how to get things back on the rust, or something or something in the

432
01:22:20.410 --> 01:22:49.839
Gerald Raymond: again for the state from the stakeholders, and to be brought back into sort of a list of recommended findings. I think this would be. This. Disorders would be high. It's just that the States are going to have to be doing this disease by disease and making the determinations what works for them. And there's a lot of things sort of in the queue at the moment that we have we at the State level have to sort of juggle.

433
01:22:51.520 --> 01:22:52.260
Steven Shapero: Thank you.

434
01:22:53.350 --> 01:22:54.349

Jenn Banna MTF2FHIC: Yeah, thank you.

435
01:22:55.590 --> 01:22:58.940
Jenn Banna MTF2FHIC: Sorry I got us excited about the break. A little early. Good call, Michaela.

436
01:22:58.940 --> 01:23:05.050
Mikaela Miller: No, no worries. I just wanted to make sure we use Dr. Raymond's time as efficiently as possible.

437
01:23:05.438 --> 01:23:10.459
Mikaela Miller: It looks like Shanalia. Did you have one more question for Dr. Raymond before he goes. Okay, please.

438
01:23:10.910 --> 01:23:15.339
Shawnalea Chief Goes Out: I'm not sure if it's a Dr. Raymond question or a Doctor Hopkins. Question.

439
01:23:15.550 --> 01:23:16.040
Mikaela Miller: Oh!

440
01:23:16.040 --> 01:23:16.390
Shawnalea Chief Goes Out: But on.

441
01:23:16.390 --> 01:23:16.980
Gerald Raymond: You can jump in.

442
01:23:16.980 --> 01:23:17.910
Shawnalea Chief Goes Out: Vacation.

443
01:23:18.533 --> 01:23:26.536
Shawnalea Chief Goes Out: It's on the follow up. Care and counseling are generally available. And you talked about the National Nyman Pick Foundation, you know.

444

01:23:27.020 --> 01:23:36.379
Shawnalea Chief Goes Out: connects to local providers to provide education and services by chance. Do you know if there are any of those located within Montana

445
01:23:37.000 --> 01:23:38.140
Shawnalea Chief Goes Out: at this time.

446
01:23:38.140 --> 01:23:55.210
Justin Hopkin: It's a good question. They're not so the National Niemann Pick Disease Foundation has identified comprehensive care centers, and we do that on a volunteer basis. So we look for medical centers that have experience in treating patients. It could even be a patient

447
01:23:55.210 --> 01:24:20.669
Justin Hopkin: with acid sphingomyelinase deficiency and have a few of the specialists that are recommended, a pulmonologist and gastroenterologist that have seen these patients. And if so, we connect with them and provide that resource to the community. We're part of screenplus, which is a New York State based Nih program which has developed protocols for how we manage patients that are newly diagnosed in newborn screening. So we have

448
01:24:20.860 --> 01:24:46.980
Justin Hopkin: a family services manager who connects the patients as soon as they're diagnosed and provides support to the families, but also services to clinicians, families, and patients about opportunities for where they can seek care if they want care, but also providing educational materials to their team on the ground, because ultimately, we don't want patients to have to travel a really long ways to to continue to receive care. So that's all been protocolized over the last several years

449
01:24:54.560 --> 01:25:00.239
Justin Hopkin: we'd love to start one in Montana, so we'll find some interested clinicians and a patient, and we'll get one going.

450
01:25:01.990 --> 01:25:11.650
Jenn Banna MTF2FHIC: That's a great question, Shannelia. We talk about that a lot with the real nature of our state. It says like a resources readily, we're like. Hmm! And you know that Dr. Hopkins.

451
01:25:18.110 --> 01:25:18.440

Mikaela Miller: Oh!

452
01:25:18.440 --> 01:25:19.460
Gerald Raymond: What is the birth?

453
01:25:20.180 --> 01:25:22.060
Gerald Raymond: What is the birth rate for Montana?

454
01:25:24.230 --> 01:25:26.450
Abe Elias: About 12,000 per year.

455
01:25:26.760 --> 01:25:28.239
Gerald Raymond: 12,000 births.

456
01:25:28.240 --> 01:25:43.299
Abe Elias: Yeah. So that was another question I had here. So it sounds like sort of the incidence is one out of 22, and 50,000. But, Dr. Raymond, when you you you cited the Illinois experience, and there were like 9

457
01:25:43.590 --> 01:25:48.899
Abe Elias: positives over. I think they screened about a million, so that.

458
01:25:48.900 --> 01:25:51.360
Gerald Raymond: It was about a hundred 25,000

459
01:25:52.350 --> 01:25:54.509
Gerald Raymond: was one in a hundred 25,000.

460
01:25:54.740 --> 01:25:55.820
Abe Elias: Over.

461
01:25:56.810 --> 01:25:57.939
Gerald Raymond: Couple of years.

462
01:25:57.940 --> 01:26:01.970
Abe Elias: Oh, oh! But they started already in 2,015, now 2,015! So.

463
01:26:01.970 --> 01:26:12.889
Gerald Raymond: Yeah, so they. But they they piloted, they piloted, and then they. And then they think they said they started screening in 2019, you know. So yeah, you'll have one every 10 years.

464
01:26:13.240 --> 01:26:19.590
Abe Elias: Right. That was a so that would be right.

465
01:26:27.750 --> 01:26:30.660
Gerald Raymond: But don't worry. You won't ever have it from a from what I can tell.

466
01:26:41.010 --> 01:26:48.250
Abe Elias: And then just to follow up on that. So the New York is also there's a 3rd site that is currently

467
01:26:48.560 --> 01:26:49.480
Abe Elias: so, yeah.

468
01:26:49.480 --> 01:26:54.259
Gerald Raymond: They have a. They have a program, and Dr. Hopkin can also is called screen plus.

469
01:26:54.290 --> 01:27:15.370
Gerald Raymond: that is, sort of a pilot program. I think it's only it's only in certain hospitals in New York City. But correct me if I'm wrong. And Melissa, it's Melissa. Waste is the pi. And so in conjunction, they they screen for you, can

470
01:27:15.370 --> 01:27:30.279
Gerald Raymond: they? You can screen for a sort of a this, additional disorders that people are eager to consider adding new one screening. I'm aware of cache and even pick, and I forget what the I don't know what the other ones are. Quite honestly.

471
01:27:30.660 --> 01:27:38.250
Justin Hopkin: The. It's 9 of the highest birth rate hospitals in New York State. Syracuse actually made the list. So they made it all the way north to Syracuse.

472
01:27:38.540 --> 01:27:39.240
Gerald Raymond: Right.

473
01:27:39.240 --> 01:27:42.089
Justin Hopkin: And they're they're also doing a lot of

474
01:27:42.200 --> 01:28:03.800
Justin Hopkin: ethical questionnaires to patients and or to parents, I guess, to better understand their perspectives on newborn screening, and with a lot of the political climate and cases that have been brought to court regarding blood spot management and newborn screening and consents. It's going to provide some really valuable information. So we're lucky to be part of that.

475
01:28:09.000 --> 01:28:38.539
Abe Elias: So I don't want to monopolize this and perseverate on this, but I do struggle a little bit in this in this case, with the you know. On the one hand, you have clearly effective treatment. For some aspects. The testing, I think, is not a problem. When I struggle with this, what we know about the difference about starting treatment

476
01:28:39.360 --> 01:28:45.940
Abe Elias: that's triggered by newborn screening. The experience that we have versus a treatment that is

477
01:28:46.230 --> 01:28:51.280
Abe Elias: the outcomes that is started after a clinical diagnosis.

478
01:28:52.020 --> 01:29:02.820
Abe Elias: And here, really, I mean, given that newborns, even though I think even later on, that even adults potentially could benefit from this. If this is diagnosed, I think newborn screening being really focused on.

479

01:29:03.050 --> 01:29:12.010
Abe Elias: on, on the, on the early period here. So I'm not completely clear yet what the data tells us.

480
01:29:12.170 --> 01:29:17.919
Justin Hopkin: I think if I could say 2 things, one, that patient reported, outcomes paper, speaks to

481
01:29:17.950 --> 01:29:42.749
Justin Hopkin: the morbidity associated with the early years of patients who participated in the clinical trial and what their lives were like, I can tell you, even being a clinician and a physician having the delayed diagnosis, even though we had it very quickly, those times where we were living without a treatment, if that could be avoided, and the cost of the healthcare system and things like that. It would be really nice

482
01:29:42.750 --> 01:29:55.259
Justin Hopkin: for that sibling that had those many years of gi symptoms before he started treatment. The older sibling of the 2. There's a lot of morbidity that came out in that patient reported outcomes paper.

483
01:29:55.350 --> 01:30:11.930
Justin Hopkin: And then for those with significant neurologic disease, whether it's bone, marrow, transplant, or any of the 3 or 4 therapies I talked about that are in the pipeline. What we know from other lysosomal storage disorders is that as soon as neurons are degenerating to the point where there's clinical symptoms.

484
01:30:12.020 --> 01:30:21.080
Justin Hopkin: you've had irreversible damage, and so diagnosing these disorders to enroll patients in a treatment to treat the brain as early as possible.

485
01:30:21.130 --> 01:30:41.210
Justin Hopkin: is going to be critical in order to identify a treatment that might be efficacious because once neurons are gone. And we knew this in Niemann pick type C, you can't recover them. And so enrolling those patients that have significant neurologic disease is really hard to tell a difference because of the damage that's already been done. You essentially need a cure to try and just stabilize the disease at that point.

486

01:30:41.210 --> 01:30:52.580
Justin Hopkin: So identifying these patients early on is going to be really important, because we have therapies that are going to become available, and we'll want to enroll them in clinical trials as best we can.

487
01:30:57.660 --> 01:31:06.009
E. Lynne Wood: Please forgive me if this was mentioned. The clinical trials that you mentioned. Where where are those happening.

488
01:31:07.190 --> 01:31:21.709
Justin Hopkin: These are all preclinical programs that we haven't started clinical trials. This is data that's being presented in different places. But none of these are to a patient-centered clinical trial. There are patients that have access to these therapies that are off label

489
01:31:22.083 --> 01:31:49.339
Justin Hopkin: so, for instance, right now, I'm trying to get my son access because he has ataxia, cognitive delays, and things like that to the therapy that's approved for Niemann pick type C, the end which was just recently approved for Niemann pick type C, so we need to learn more and gather more data about all of these in this space. And so trying to make the diagnosis early. So we can identify those and enroll patients is the point I was making. And you could make that for a lot of disorders.

490
01:31:49.340 --> 01:31:51.559
E. Lynne Wood: Sure, sure, but it sounds like

491
01:31:52.334 --> 01:31:59.920
E. Lynne Wood: there may be a decent amount of movement in the clinical trial space in the near future.

492
01:32:00.180 --> 01:32:00.720
Justin Hopkin: Yeah.

493
01:32:01.050 --> 01:32:06.709
Justin Hopkin: we'd like to believe. So we have to convince the companies to continue to invest and move forward with the climate we're in. But

494

01:32:06.710 --> 01:32:09.709
Justin Hopkin: sure there's some promising science. We'll put it that way.

495
01:32:23.340 --> 01:32:23.920
Mikaela Miller: Hey?

496
01:32:25.410 --> 01:32:55.319
Mikaela Miller: Well, hopefully, this doesn't feel too disjointed here. I just wanted to make sure everyone had a chance to ask Dr. Raymond their questions. So let's go ahead and take a 10 min break as promised. We do have about an hour left scheduled in this meeting. It's questionable whether or not, it'll take that long, but we'll go ahead and take a break. Feel free to brainstorm any other discussion items or questions that you have. We'll come back for discussion.

497
01:32:55.380 --> 01:33:22.379
Mikaela Miller: Maybe take a few moments for the public comment period and then go ahead and wrap up the end of the meeting. I just wanna make sure, since we're going a little bit longer than the promise break time. I just wanna make sure everyone has a chance. So let's come back here at 3, 45, or I'm sorry. 1, 45 for those of you in Montana. 3, 45 for the rest of us over more on the east coast.

498
01:33:22.640 --> 01:33:24.960
Mikaela Miller: Thank you, Dr. Raymond, for being here.

499
01:33:25.240 --> 01:33:29.679
Gerald Raymond: I'm gonna step away. If anyone has any additional questions they can certainly reach out. Thank you.

500
01:33:30.600 --> 01:33:31.210
Mikaela Miller: Sounds good.

501
01:33:31.210 --> 01:33:32.338
Abe Elias: Yeah, thanks, so much.

502
01:33:32.850 --> 01:33:34.120
Gerald Raymond: By a.

503
01:33:34.120 --> 01:33:34.860
Abe Elias: In banks.

504
01:33:36.650 --> 01:33:39.209
Mikaela Miller: All right. We'll see you all back here in about 9 min.

505
01:42:36.990 --> 01:42:41.820
Mikaela Miller: Great! It looks like everyone's starting to

506
01:42:42.090 --> 01:43:05.559
Mikaela Miller: kind of roll back in here. So just to kind of briefly catch everyone up where we're at in the agenda. We are a little ahead of schedule, so we can still take that full 40 min for the discussion. And then, like, I said, we're going to go into quick public comment period and then wrap things up.

507
01:43:05.620 --> 01:43:27.350
Mikaela Miller: So I'd like to go ahead and open the floor back for discussion. Jen, I'll let you go ahead and take over. But I just did want to note quickly. Here there was something earlier that was kind of left unresolved. I believe Steven had a question, and just, or Dr. Hopkin you mentioned you could kind of speak to that a little bit more if you would like to do so.

508
01:43:27.350 --> 01:43:40.880
Justin Hopkin: Yeah, Steven, great question. So as far as newborn screening in the Us. We have the original rusp application. That was done almost 16 years ago, before treatment was available.

509
01:43:40.940 --> 01:44:03.480
Justin Hopkin: After the approval of this therapy, in 2022, we started working on another rusp application. Ruspap actually changed their application process about a year and a half ago, I think it was, and we had started to work through that application with them with 4 just original screening questions. So we were working with them when they were still taking nominations. I think

510
01:44:03.480 --> 01:44:23.070

Justin Hopkin: we got some encouragement from them. But now, as you all know, that process is sort of uncertain at this point in time. So we're taking an approach of going State by State and progressive states like Montana, that have an application we presented to Georgia 2 year and a half ago. I think they're very excited about it. They moved it into Pilot.

511
01:44:23.080 --> 01:44:39.340
Justin Hopkin: So Dr. Cox is leading things down there. We hear that Missouri is actually moving forward with Asmd screening, though I cannot confirm this. So I think from what I'm told they're going to start screening for it as well. They had contemplated it many years ago, and are going to pick it back up

512
01:44:39.748 --> 01:45:04.169
Justin Hopkin: Wisconsin like I said. I've been on 2 or 3 of their meetings. we're continuing that discussion, and I'm hopeful that they'll be one of the States that adds, and then Indiana and Pennsylvania also have applications. That they are looking at, but they don't have a formal hearing process like Montana does, and then from a legislative perspective. We've been close in Massachusetts.

513
01:45:04.470 --> 01:45:20.699
Justin Hopkin: They're close to considering it, and they screen for 4 or 5 other States around New England, which would be a big catchment, I guess, as far as patients are concerned. So that's where we're at. As far as expanding newborn screening. In addition to those places in New York State that we talked about.

514
01:45:21.140 --> 01:45:24.229
Steven Shapero: Have you? Have you approached California at all at this point.

515
01:45:24.620 --> 01:45:37.619
Justin Hopkin: They don't have a an, a process by which we can do this from an application perspective. We would like to do that, but I think it's going to be legislative approach at this point. In Texas. We are also looking at a legislative approach, too.

516
01:45:37.620 --> 01:45:38.710
Steven Shapero: Gotcha. Thanks.

517
01:45:43.940 --> 01:45:49.469

Justin Hopkin: And when I say we, there's like 2 or 3 of us working on this, it is a small we, but we're we're moving it forward.

518
01:45:53.420 --> 01:46:00.620
Jenn Banna MTF2FHIC: Okay, so it's time for us to be able to move into our discussion period.

519
01:46:02.700 --> 01:46:15.129
Jenn Banna MTF2FHIC: yeah. So I'm gonna have. Stephanie put the criteria that we're using up on the screen for us, because I think that's 1 of the things that's been a little difficult in our discussions in in the past is trying to go back and remember

520
01:46:16.720 --> 01:46:32.490
Jenn Banna MTF2FHIC: as we're discussing what our selection criteria is, and then sometimes also, this is off the application. And so, as we know before, this is off the application. So we may have things to add that are unique for our state, and I think Abe brought up some of that

521
01:46:32.610 --> 01:46:39.630
Jenn Banna MTF2FHIC: earlier you know the floor is really

522
01:46:39.900 --> 01:46:48.239
Jenn Banna MTF2FHIC: all of ours to talk about this right now, so we could talk about it thing by thing. Or if people have comments or questions they just want to ask. We can do that, too.

523
01:46:51.270 --> 01:47:02.774
Jenn Banna MTF2FHIC: I I know Dr. Hopkin. I'm gonna Re, ask my question again, because the late onset thing has been this curiosity we've had where some people are like. I was tested for that on the newborn screen. And then I got it as an adult

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01:47:03.420 --> 01:47:08.259
Jenn Banna MTF2FHIC: where we've heard that with some of like late onset. So I just want to ask again

525
01:47:08.540 --> 01:47:10.709
Jenn Banna MTF2FHIC: if this particular test

526

01:47:11.020 --> 01:47:16.739
Jenn Banna MTF2FHIC: does does it end up catching that whole group when we've talked about the A's and the B's and the C's and the Laters and.

527
01:47:17.760 --> 01:47:43.959
Justin Hopkin: So to clarify this, we're just looking at the I think the 3 diseases that Dr. Raymond said were A, A, B, and B, so the C is a different disease. It's a different newborn screen test they're doing in that New York specific to the Asmd group, which is those 3. And I think the Illinois data suggests that yes, we do capture those with a less severe phenotype, because it appears that of the 10 or so that have tested positive in Illinois.

528
01:47:43.960 --> 01:47:50.889
Justin Hopkin: Almost all of them don't have significant neurologic diseases they're following. And so they are picking up

529
01:47:51.297 --> 01:47:55.349
Justin Hopkin: all of the the phenotypes, including those that are less severely affected.

530
01:47:55.970 --> 01:48:13.769
Jenn Banna MTF2FHIC: Okay? And then that less severely affected or late Onset group. Then are they watched for symptoms, or are they treated? And I come from a non medical background. I'm a parent of a rare, with a rare neurogenetic disability. So I have a non medical background. So I may ask the questions in an unusual way, but.

531
01:48:13.770 --> 01:48:14.480
Justin Hopkin: That's an amazing.

532
01:48:14.480 --> 01:48:27.030
Jenn Banna MTF2FHIC: Are they? Yeah, are they watched? Are they treated like what happens? Because as a parent with that group, if you get a baby and they're like, well, you know, it's like, now, what do we do? We're looking, watching, waiting. And so I'm wondering while that's working for those families.

533
01:48:27.030 --> 01:48:47.730
Justin Hopkin: It's a hot topic. That clinical guideline that we created was meant to be expert opinion on what to do, and anyone with symptoms of the disease should be considered for

treatment. So if you're asymptomatic essentially, if you're diagnosed because a sibling had it, or something like that, the therapy likely is probably not going to provide significant benefit.

534
01:48:47.730 --> 01:49:10.910
Justin Hopkin: What we're looking at doing, probably in the next 6 months is developing guidance specifically for patients who are diagnosed to newborn screening, because at birth they're all probably going to be relatively asymptomatic to start with, and so trying to identify when that right timeframe is to treat, and how to have that conversation of informed decision making with parents and families is something that

535
01:49:10.910 --> 01:49:27.969
Justin Hopkin: you know Josh Baker, who's the person who authored that paper in Illinois. He and I have talked about this quite a bit like there's not a lot of guidance on on when to start therapy. He's looking at spleen size and following it closely, and looking at blood counts and talking about, tolerating, of eating

536
01:49:27.970 --> 01:49:48.480
Justin Hopkin: respiratory symptoms, watching growth curves really closely, and things like that to try and determine what that trigger is to start enzyme replacement therapy. But it's thought that earlier treatment probably leads to better outcomes. But starting it in the asymptomatic period. We just don't have enough data yet to figure out that exact right time. But symptoms is kind of the key thing.

537
01:49:48.940 --> 01:49:50.329
Jenn Banna MTF2FHIC: Okay. Thank you.

538
01:50:04.540 --> 01:50:18.799
Jeanne Lee: Dr. Hopkin. I have a question. I know the Revedy assay is an FDA. Approved assay, but I was wondering about the geld chem assay is that FDA approved.

539
01:50:18.800 --> 01:50:33.250
Justin Hopkin: It is. Yeah, as you can guess by the name. Michael Gelbs kind of has a lab. He did the original studies in Oregon on this test, and there's been a lot of other countries and states that have done it as well. They both screen for the same 6 disorders.

540
01:50:33.524 --> 01:50:56.829

Justin Hopkin: And they're just 2 different companies. So the revedy is much more frequently used. I don't know the price of each but as far as states that I've talked with in the labs. They tend to to gravitate toward that over the other. But in in fairness I try to let people know that there are 2 that are out there. Especially since Michael did so much of the science and all of that. He's the one that told me about it. So.

541
01:50:57.990 --> 01:50:58.900
Jeanne Lee: Thank you.

542
01:51:02.340 --> 01:51:15.299
Steven Shapero: Dr. Hopkin. One question you mentioned that there are people around who are carriers who don't, I guess, don't show symptoms at all, or very minor symptoms.

543
01:51:15.470 --> 01:51:16.140
Steven Shapero: and.

544
01:51:16.140 --> 01:51:45.359
Justin Hopkin: I should have explained that a little bit better in the when I said that. So I showed my family. This is an autosomal recessive disease. So in order to have the disease, you need to inherit an abnormal gene from both parents. And so I'm a carrier, and my wife is a carrier. We don't have the disease, and our enzyme levels were checked. The idea of a bone marrow transplant was discussed by Dr. Raymond.

545
01:51:45.360 --> 01:51:55.140
Justin Hopkin: and what we would do in that scenario is to replace the original bone marrow with someone else's bone marrow. Who can make the enzyme

546
01:51:55.390 --> 01:52:11.409
Justin Hopkin: that's been looked at in this disease, and my family went to Duke to have that evaluated, and the best candidates usually for bone marrow transplants are siblings, because the rest of the genome is very similar. And so both of my other kids were tested. They're also

547
01:52:11.410 --> 01:52:34.679
Justin Hopkin: they carry one of the 2 genes each, and you don't want to have someone who's a carrier be someone who donates bone marrow because the amount of enzyme they make probably isn't as much as somebody who's not a carrier at all. But you don't need to make a lot of enzyme to not have symptoms of this disease. So you need both genes to be abnormal in

order to have the disorder so sorry for any confusion that caused hopefully, that clears it up a little bit.

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01:52:34.880 --> 01:52:43.869
Steven Shapero: It. It does, and I guess that it kind of kind of explains. The rarity of this disease is that the likelihood of of 2 carriers meeting whatever is fairly rare.

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01:52:43.870 --> 01:53:13.810
Justin Hopkin: Exactly one in 200. In the Us. Population is the carrier rate, so I found one of the other one in 200 S. Neither one of us come from the Ashkenazi Jewish population, which is often what's known in the literature to have this. But we're these are the sort of random things that can pop up with these genetic disorders. And so we have a 1 in 4 chance of having a child that has the disease with each of us being carriers. And so that's what happened to my son.

550
01:53:38.537 --> 01:53:44.412
Abe Elias: Jeannie, I I wanted to go back to you know, when you talked about the

551
01:53:45.250 --> 01:53:46.560
Abe Elias: the costs?

552
01:53:48.150 --> 01:53:49.110
Abe Elias: So

553
01:53:49.250 --> 01:54:00.549
Abe Elias: based. If you think about based on on our birth rate, it's I mean, it's I think it's a fair. It's an estimate, at least, that we would have to screen 10 years to get one

554
01:54:00.780 --> 01:54:07.490
Abe Elias: positive. So if you few, and you mentioned that the

555
01:54:08.270 --> 01:54:17.089
Abe Elias: you didn't get a clear answer from Wisconsin, what the costs were so currently based on $15 per test. That would be.

556
01:54:18.870 --> 01:54:23.400

Abe Elias: you know, per year per year, about $180,000

557
01:54:25.060 --> 01:54:30.890
Abe Elias: for this, and then, and and over 10 years would be 1.8 million

558
01:54:31.420 --> 01:54:47.859
Abe Elias: is there? I guess. I think in this case it would be helpful to actually try to get from Wisconsin to get a little bit of better, perhaps a more precise estimate of what it is because it it is a significant cost. I mean, it's basically 10% of our current

559
01:54:48.260 --> 01:54:48.950
Abe Elias: current.

560
01:54:52.410 --> 01:55:04.209
Abe Elias: you know, the newborn to a total newborn screening costs. Is that right? Well, no, not not 10%. It's it's a little less. It's not 10% but but so it is significant.

561
01:55:04.400 --> 01:55:16.239
Abe Elias: So if if they'll if Wisconsin would be able to do it, and I mean, I assume that that if they were to start screening, that it would get cheaper. But I don't know.

562
01:55:16.770 --> 01:55:18.705
Abe Elias: Do you have any.

563
01:55:19.570 --> 01:55:35.010
Jeanne Lee: I. You know I was kind of disappointed, Dr. Elias, when I had reached out to May Baker and said that you know our advisory committee would be listening to this condition.

564
01:55:35.120 --> 01:55:53.739
Jeanne Lee: And would they be able to do testing for us? And if she had an idea of what the cost would be. She didn't give me an answer. She just said, Let's talk when it's if it's added to your panel, and so So

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01:55:53.960 --> 01:56:02.920

Jeanne Lee: that's why I I don't have a feel for what it would be. I I do think. You know, when we add Pompeii.

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01:56:03.030 --> 01:56:20.190
Jeanne Lee: I think it's been like a year now, since we 1st considered Pompei disease, she had told us at that time, you know, $11 to add Pompei disease to the panel, I think.

567
01:56:20.340 --> 01:56:36.249
Jeanne Lee: with, like the recent kind of rise in costs and tariffs. We can expect that to be a little bit more than $11 per test. And so with Asmd.

568
01:56:36.440 --> 01:56:47.239
Jeanne Lee: You know it is multiplex it. I don't think it will be another $15 to add Asmd. But it'll be

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01:56:47.640 --> 01:57:00.860
Jeanne Lee: somewhere up to. That is what I think just, but it it will depend on like her staffing, and being able to to cover their costs as well.

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01:57:02.500 --> 01:57:10.739
Justin Hopkin: It's interesting in my discussion there, and at Georgia they both use the multi, the assay that that I showed with Revedy, and both

571
01:57:10.860 --> 01:57:21.310
Justin Hopkin: lab said, there's no additional cost. They're actually blinding the results right now, because if you run Pompeii or you run Mps, one on either of those or crab A, now that it's been approved

572
01:57:21.480 --> 01:57:39.889
Justin Hopkin: that you actually have to blind the results. And so I'm not sure where the extra cost that you're having, in addition to Pompeii would come from. But there's definitely cost considerations if you have either true, positive, or false, positive to the system. And so that's I think that's worth considering

573
01:57:39.960 --> 01:57:55.160

Justin Hopkin: how many of these tests are going to require. Follow up. Who's going to do that? Follow up in the public health system, and those sorts of things should definitely be considered. But from a lab perspective, I'm not sure why. Why, on a per test, if you're testing for one, why, there would be extra test. But I'm not a lab person.

574
01:57:55.910 --> 01:58:05.390
Jeanne Lee: And I agree with that. So I I don't know. I'll just. We'll have to find out from May Baker at that time.

575
01:58:06.870 --> 01:58:26.709
Abe Elias: Yeah, I, Dr. Hopkins, I do think that there is. So when you run this an additional test, you have to run. Qc, you have to have additional control samples, and Ginny meant staffing. So I think that in fact, those are the part, especially in these scale tests.

576
01:58:26.710 --> 01:58:39.290
Abe Elias: Those are the kind of the costs that often are more significant than the consumables. So I think if a lab director tells me that it wouldn't cost to add anything.

577
01:58:39.290 --> 01:58:59.820
Abe Elias: I think that's more a conceptual, not a practical answer. So I think. But $15 is quite a bit. And I could imagine, if they actually had it implemented in their workflow. You know they have a staff there. They have to work the Ftes for that. And Qc. Is significant that you have to do there.

578
01:59:00.110 --> 01:59:08.409
Abe Elias: I couldn't imagine that that that would go down, but I don't know.

579
01:59:14.880 --> 01:59:18.400
Jeanne Lee: Oh, sorry. Jen.

580
01:59:18.600 --> 01:59:35.939
Jenn Banna MTF2FHIC: Oh, go ahead, Jeannie. No, I was just. It was what we were talking about yesterday, with like the economic changes and the tariffs. And we were like, oh, we probably don't need to talk about that quite yet. But we were just talking about how that changes all of these things, and I don't think we've ever talked about how, if we added one test that Wisconsin was testing.

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01:59:35.950 --> 01:59:48.709
Jenn Banna MTF2FHIC: and then, like what's happening right now with Pompeii, we're now we're in a situation where, if we add another test that can be done on the same thing, then our cost stuff changes between our meetings and our votes as we're trying to consider all that information.

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01:59:48.760 --> 01:59:53.959
Jenn Banna MTF2FHIC: And I I don't think that's something we've ever really like addressed or talked about before.

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01:59:57.100 --> 02:00:07.509
Jeanne Lee: Yes, thank you, Dr. Hopkins. I was going to ask a little bit more about Illinois and New Jersey, and

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02:00:07.650 --> 02:00:28.879
Jeanne Lee: the assays that they're using, and like the cutoffs, you know, laboratory practice would be kind of establishing your cutoff for your population. Do you have a feel based on from package insert, if they had to change their cutoffs for their assays.

585
02:00:31.480 --> 02:00:57.000
Justin Hopkin: I don't know what the package inserts. Say, I do have the cutoffs written down. I think it's 15% of the daily, is it? Daily I forget the the measurement. But daily mean, or something like that average daily. Mean? It's not language I I'm used to. But I have it written down. And if you want to give me your email, I can follow up with you. I haven't looked at that data in a couple of years, but I can pull up.

586
02:00:59.120 --> 02:01:00.020
Jeanne Lee: Thank you.

587
02:01:00.020 --> 02:01:01.270
Justin Hopkin: I think they sort of

588
02:01:01.460 --> 02:01:18.550
Justin Hopkin: made it up as they went, because they were the pilot, and so they sort of said, Let's try this one, and it's been a good fit for them for not having false positives, but feeling

they're capturing most of the patients, and they rerun samples as part of all of that to for their own Qa. To make sure that it's on, repeat samples that they see it.

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02:01:19.000 --> 02:01:26.819
Jeanne Lee: And and so do you know, Dr. Hopkins, if Illinois had a lab developed test when they began, or or.

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02:01:27.540 --> 02:01:44.639
Justin Hopkin: Oh, I I'm it's a good question. I believe that they used the the product that was present. I had a conversation, but as far as when they started with the pilot, they published their pilot data, and I can't remember if they use their own, or if they're they're using the commercially available one, so I don't know.

591
02:01:45.120 --> 02:01:46.500
Jeanne Lee: Okay. Thank you.

592
02:01:49.790 --> 02:01:54.319
E. Lynne Wood: I don't know, Dr. Hopkin, if this is a question that is even

593
02:01:54.990 --> 02:02:00.940
E. Lynne Wood: appropriate or reasonable to ask you. But with the discussions that we were having a few minutes ago about

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02:02:01.740 --> 02:02:19.589
E. Lynne Wood: the potential cost for the number of patients we'd be benefiting in a 10 year period. Did Wisconsin give you a sense of when they might consider adding it to theirs, particularly if we're thinking, oh, well, look! Wisconsin adds it, our cost might go down, and it might be more feasible like. Did they give a timeframe at all?

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02:02:19.590 --> 02:02:44.640
Justin Hopkin: So. These are publicly available minutes. It was a meeting literally, just like 2 weeks ago. So I don't think the minutes are out yet. I am going to to resubmit my packet in full, because there were people that were on the call that didn't get all of the packet, and I submitted over a year ago. And there's a lot of new data that I referenced that wasn't in the original packet, but that I showed in a presentation.

596

02:02:44.880 --> 02:03:03.619
Justin Hopkin: And so they wanted a chance to review more of the information, particularly any posters or anything that came up in regard to those other therapies. The sibling data, I thought, was interesting, and they wanted that as well. So, rather than vote on what was a

597
02:03:03.830 --> 02:03:22.789
Justin Hopkin: old and somewhat incomplete, I offered to just redo the entire pack, redo the entire application for them to review and hold so they could have all the information they needed to make. A more precise measurement. As far as if they wanted to add it or not. So so that's what I'm going to do soon.

598
02:03:22.790 --> 02:03:23.380
E. Lynne Wood: Yeah.

599
02:03:24.190 --> 02:03:29.109
Justin Hopkin: No, I'm hoping it'll be in the next 6 months or so that they will be able to evaluate that. Make a decision.

600
02:03:29.730 --> 02:03:30.689
E. Lynne Wood: Okay, so.

601
02:03:30.690 --> 02:03:31.960
Justin Hopkin: Timing of their meetings.

602
02:03:31.960 --> 02:03:34.889
E. Lynne Wood: Yeah, decision would be in 6 months, and then

603
02:03:35.070 --> 02:03:53.540
E. Lynne Wood: I don't. This is my 1st like real meeting. So I'm very, very new at all this. So please forgive me if these are ignorant questions, but my impression from the meetings that I've shadowed in has been that even once a condition gets added, it takes quite a while for it to actually be rolled out. So

604
02:03:54.160 --> 02:04:01.199
E. Lynne Wood: who knows but maybe reasonable to say that Wisconsin might have it if they decide to approve it. In a year or 2.

605
02:04:02.000 --> 02:04:20.489
Justin Hopkin: I think it's a little different with this test, and that they own the equipment. Then they have the reagent, and they're already running the test. So I think it would happen more quickly than in other places, because the funding needs to be appropriated, the lab needs to get the equipment. And I think. And Jean, is it, Jeannie? Sorry.

606
02:04:20.797 --> 02:04:38.329
Justin Hopkin: Can probably tell us a better timeframe, but my sense is that they could likely institute this at least a pilot if they want to do a pilot 1st versus just expanding the screening more quickly than if it was a new test they had to bring in new equipment and and make sure that the test was

607
02:04:38.480 --> 02:04:41.000
Justin Hopkin: appropriate. But that's just a guess.

608
02:04:42.410 --> 02:04:59.130
Jeanne Lee: And then Dr. Wood, it also, you know, does take a little bit time to go through that rulemaking process to add it to the panel. And so when we added Xald to Montana's panel, it did take about 9 months to do that.

609
02:04:59.290 --> 02:04:59.890
E. Lynne Wood: Yeah.

610
02:05:01.000 --> 02:05:26.369
E. Lynne Wood: Part of why I'm asking all the questions is I'm trying to figure out well, is there a sweet spot to try and add it to where we're diagnosing these kids, and it sounds like, and Dr. Hopkins, correct me. If I'm wrong, it sounds like a lot of the clinical trials we'd be getting them into as far as potential therapies. And then the specific data about what it means for maybe neurologic outcomes.

611
02:05:26.370 --> 02:05:39.089
E. Lynne Wood: We still don't know yet. We're optimistic, but not sure that any of the clinical trials are going to be ready for prime time just yet, but maybe soon, so I don't know if there's an ideal time for us to like

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02:05:39.510 --> 02:05:55.019
E. Lynne Wood: consider approving, adding, so it's at Wisconsin, and it's cost effective. And then we actually have places where we can send these kids if they screen positive. But gosh! That's so many variables, it'd be hard for me to even calculate in my head what that perfect little

613
02:05:55.440 --> 02:05:56.830
E. Lynne Wood: spot is

614
02:05:57.381 --> 02:06:03.020
E. Lynne Wood: but yeah, I don't know. Sorry, guys. Now I'm thinking out loud. Probably not a good use of your time.

615
02:06:05.050 --> 02:06:28.270
Jenn Banna MTF2FHIC: I thought those were great questions. This is the 1st time I think we've been up against this Dr. Wood, where we've had this, where we're actually, we've had someone on like Dr. Hopkin, who could say Wisconsin was doing in the background. So I mean, this is these are those unique things we're running up against that that we need to talk about. So I actually appreciate there's no really, all the questions are really good, because some people still have the question, and they don't ask it. So that ask away.

616
02:06:36.830 --> 02:06:39.800
Jenn Banna MTF2FHIC: Do you want to go to the second page, Michaela, of the

617
02:06:40.860 --> 02:06:43.310
Jenn Banna MTF2FHIC: selection criteria, just to remind people

618
02:06:46.710 --> 02:06:47.850
Jenn Banna MTF2FHIC: about that.

619
02:06:52.520 --> 02:06:54.360
Jenn Banna MTF2FHIC: I guess there's only a few on there, but

620
02:06:59.490 --> 02:07:03.779
Jenn Banna MTF2FHIC: I think we've discussed these ones pretty well. Go ahead, Steven.

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02:07:04.270 --> 02:07:06.759
Steven Shapero: I was just gonna say,

622
02:07:08.130 --> 02:07:13.090
Steven Shapero: we did talk about Number 9, but I trying to pull back from my memory

623
02:07:13.710 --> 02:07:18.880
Steven Shapero: we think there would be support available fairly quickly if we

624
02:07:19.130 --> 02:07:21.929
Steven Shapero: had a positive, or we weren't sure.

625
02:07:22.090 --> 02:07:23.230
Steven Shapero: In Montana.

626
02:07:25.240 --> 02:07:41.599
Abe Elias: Yeah, I can speak to that. Yeah, we have a newborn screening follow up metabolic, newborn screening follow-up program. And and that would be not. And so the way, if it was positive the patient would be referred, and then there would be a genetic evaluation, and then we could

627
02:07:41.980 --> 02:07:42.655
Abe Elias: Nope

628
02:07:47.250 --> 02:08:03.580
Abe Elias: and then could be evaluated. Yeah, that's I think that's not. That would be available just similar to other metabolic conditions. As Dr. Hopkins mentioned earlier. You know, if there was the need, then later on, especially if if

629
02:08:03.790 --> 02:08:16.099
Abe Elias: manifestations get more severe and there's they can also be referred into A to a center. But you know I

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02:08:16.140 --> 02:08:41.339

Abe Elias: given that. These are, you know, the infusions of those for the enzyme replacement therapy is every 2. It's an enzyme replacement that you start low, and you and you ramp up fairly quickly, and there are every 2 week infusions. So you would have to organize the infusion here in Montana, and that is usually not a problem we have. We do that for other so much storage disorders.

631
02:08:43.220 --> 02:08:44.262
Steven Shapero: Okay. Thank you.

632
02:08:54.200 --> 02:09:04.520
Abe Elias: Maybe I can follow up and kind of frequenting Dr. Hopkin here quite a bit. So if

633
02:09:04.780 --> 02:09:05.889
Abe Elias: you know it.

634
02:09:06.390 --> 02:09:18.200
Abe Elias: we we know that this is a therapy. Early therapy is is beneficial, I think, and and and you know, Nieman pick, even though it's such a rare disorder, is, is a

635
02:09:18.500 --> 02:09:28.700
Abe Elias: very, I think, distinct, you know disease, and and and I think the ert is is the enzyme. Replacement. Therapy is is very

636
02:09:29.214 --> 02:09:30.559
Abe Elias: you know, I think if we

637
02:09:31.790 --> 02:09:45.530
Abe Elias: it's a beneficial and and I think exciting. The last, you know, couple of years, especially. A development. Is it fair to say, though, that that we were still in terms of

638
02:09:46.140 --> 02:10:04.879
Abe Elias: numbers, and you know we can't even talk about larger numbers, because even in the Us. The number of patients that are every year diagnosed is limited. So to enroll them into studies that all takes some time, of course. But is it fair to say that we're still in terms of numbers. How many

639
02:10:05.350 --> 02:10:07.869
Abe Elias: you know? How much do we know about

640
02:10:08.050 --> 02:10:17.769
Abe Elias: treatment that was initiated shortly after birth, for example, versus later meaning. At 2 years or 3 years of age

641
02:10:18.000 --> 02:10:23.729
Abe Elias: we have limited data, but that link that data is being produced as we speak is that is that a fair statement.

642
02:10:24.126 --> 02:10:28.089
Justin Hopkin: Think you're spot on and I think for these.

643
02:10:28.310 --> 02:10:43.450
Justin Hopkin: I would say 60% or so of patients. And I'm shooting from the hip that don't have the more severe form of the disease very early on in life, 50, 60%. As you said, the drug is almost too good.

644
02:10:43.480 --> 02:11:02.050
Justin Hopkin: and that it melts away the disease so you could start it potentially later on in life. And you don't see significant long-term sequelae after you've started therapy, I think the question that is at the table is for those more severe patients that have the severe phenotype

645
02:11:02.080 --> 02:11:30.350
Justin Hopkin: is early initiation of enzyme is early diagnosis needed to start early, ert important. And from a time perspective, do we need to have newborn screening to do that? And then that's the question. I think we're trying to ask ourselves, and that's what's been posed to me at Wisconsin and the other places is for the more severe phenotypes does this matter, and that's 1 of the reasons, you know. I thought it was interesting that there are some severe phenotype kids that really shouldn't be with us, based on how they presented

646
02:11:30.350 --> 02:11:54.229
Justin Hopkin: that started therapy late, and they started with severe neurologic symptoms. They're still alive, but very much impacted by their disease. Their visceral symptoms have gotten better so. Really, I think the question is, for those that have severe disease is early

initiation for them, going to be helpful to prevent sequelae and or enroll in a clinical trial, and that would be the key, because for the others

647
02:11:54.230 --> 02:12:11.060
Justin Hopkin: we don't have good information on that. We're hoping to. There's about 20 or so patients that were treated worldwide on a compassionate use plan under the age of 2, over the last 7 or 8 years, and we're trying to encourage the

648
02:12:11.300 --> 02:12:29.599
Justin Hopkin: the reporting and the sharing of them with posters and publications. So we can better understand how we can identify those patients and what they may look like. But that would be the argument otherwise, for many of the pediatric and even the adult patients, they got significantly better after a couple of years of Ert

649
02:12:30.910 --> 02:12:35.169
Justin Hopkin: hopefully. That answers, or I, I would say, strengthens the point that you're making right.

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02:13:07.620 --> 02:13:10.630
Jenn Banna MTF2FHIC: Do you want to take us back to the previous slide, Michaela?

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02:13:13.590 --> 02:13:17.199
Jenn Banna MTF2FHIC: And if there's other slides that the committee wants to see, or other things that

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02:13:18.860 --> 02:13:20.569
Jenn Banna MTF2FHIC: we want to review. We can.

653
02:13:21.010 --> 02:13:23.809
Jenn Banna MTF2FHIC: We can ask for somebody to try to find that for us, too.

654
02:13:31.780 --> 02:13:45.640
E. Lynne Wood: You know, I feel like we've been over things pretty thoroughly, you know. I feel like I have the most of the information that that we can have. I don't have any other slides that I I feel like I need to see again, or anything like that.

655
02:13:45.910 --> 02:13:46.590
Jenn Banna MTF2FHIC: Okay.

656
02:13:55.800 --> 02:13:58.239
Jenn Banna MTF2FHIC: I just wanna make sure everybody's had enough time to.

657
02:13:58.390 --> 02:14:02.990
Jenn Banna MTF2FHIC: But I also don't want to take up too much of your time. What do you think, Michaela? Are we ready to

658
02:14:04.500 --> 02:14:06.980
Jenn Banna MTF2FHIC: Move on? It would be public comment next correct.

659
02:14:07.610 --> 02:14:10.730
Mikaela Miller: Yeah, so we would normally.

660
02:14:11.950 --> 02:14:13.760
Jenn Banna MTF2FHIC: Oh, Abe, just something to say.

661
02:14:14.290 --> 02:14:15.020
Jenn Banna MTF2FHIC: Come on, Abe.

662
02:14:15.020 --> 02:14:34.259
Abe Elias: It's not I. Just one last question, maybe among those. And it's again, I agree. It's nice to have the Hopkins here is involved in different. You know, activities in different states. Among those States that are currently considering how many are

663
02:14:35.410 --> 02:14:38.889
Abe Elias: starting this as a pilot versus implementing it right away.

664
02:14:40.050 --> 02:15:09.610
Justin Hopkin: It's a great question. I think it's up to the committees when they make the decision. Just what they want to do. I think Georgia just standardly want creates a pilot out of all of their additions to their own newborn screening panel. Because they want to monitor it more

closely, and probably bring it back to the committee rather than just just adding it to their panel. So that was the sense I got when they were evaluating you know, when the call I was on and they decided to move forward.

665
02:15:09.915 --> 02:15:29.159
Justin Hopkin: I I don't know. Other committees and other other States structural process. But I thought it was a unique way to do things, to start with a pilot to see how it goes, and then allow the committee the opportunity to sort of check in and see the queue quality assurance on the test, and what the results were.

666
02:15:31.260 --> 02:15:40.619
Jenn Banna MTF2FHIC: I'm actually glad you asked that because I was. We haven't ever talked about a pilot, and I was, think I so I I couldn't even figure out how that would even apply in our situation. Because I don't think we've piloted

667
02:15:41.030 --> 02:15:42.440
Jenn Banna MTF2FHIC: like that's not really.

668
02:15:42.970 --> 02:15:57.929
Jenn Banna MTF2FHIC: No, Jeannie's shaking her head. No, and I suppose if you can't already do the test, then you don't necessarily want to be piloting a test that's going to cost. If it were to cost extra money. Right? You would, pilot. Am I right, Dr. Hopping? You would be more likely to pilot something that you already had the testing

669
02:15:58.230 --> 02:15:59.980
Jenn Banna MTF2FHIC: things to do. Is that right?

670
02:16:01.910 --> 02:16:03.939
Justin Hopkin: Up for Jean, who who runs live.

671
02:16:03.940 --> 02:16:04.510
Jenn Banna MTF2FHIC: No.

672
02:16:04.510 --> 02:16:24.330
Justin Hopkin: Yeah, but that makes sense to me. That I mean, we run pilots in the hospital all the time, and I think they're just ways of saying we're not committing to doing this long term.

We're gonna start at the short term. We're gonna analyze it. Make sure it. It's that we understand the test itself. What the results are that we're getting. We're happy with it before we decide that we're gonna

673
02:16:24.430 --> 02:16:32.940
Justin Hopkin: commit to doing this long term. So it's it's an engagement. I would refer to it as just a not a commitment.

674
02:16:33.700 --> 02:16:43.009
Abe Elias: Yeah. And I think one of the limitations we have. I think you know you could. There are many ways how to organize. A pilot is the the numbers.

675
02:16:43.010 --> 02:17:10.510
Abe Elias: I think a pilot makes most sense. If you have a state with a large birth rate where you can say we commit this much money for this amount of time we can expect these results. Given that we are on our birth rates. It's difficult to do these for these type of scenarios. Unfortunately. So we're kind of, in a way, to some extent dependent on pilots on the information from other States.

676
02:17:10.840 --> 02:17:11.970
Abe Elias: To some extent.

677
02:17:15.959 --> 02:17:18.259
Jenn Banna MTF2FHIC: Thank you, Jeannie. Did you have anything you wanted to add to that?

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02:17:21.119 --> 02:17:23.999
Jenn Banna MTF2FHIC: Alright, Michaela? I think think we're good.

679
02:17:24.919 --> 02:17:25.769
Jenn Banna MTF2FHIC: Maybe.

680
02:17:27.440 --> 02:17:45.279
Mikaela Miller: Well, I was just going to say, I don't see any members from the public present during the meeting today, so I think we can take this time just kind of a final call. If anyone has anything else that they would like to share or ask.

681
02:17:55.559 --> 02:18:05.979
Abe Elias: I just want to thank. I mean, it was great to have really 2 2 great experts here today on the call. Both Dr. Hopkins and Dr. Raymond. So I really appreciate that.

682
02:18:09.440 --> 02:18:10.299
Jenn Banna MTF2FHIC: Paid.

683
02:18:17.639 --> 02:18:19.479
Mikaela Miller: It's well.

684
02:18:20.619 --> 02:18:46.249
Mikaela Miller: then some final steps here, or I guess I should point out for anyone who does have any additional comments. If you would like those on the public record, and you think of it within the next hour you're welcome to send an email to the newborn advisory committee@mt.gov. And we can include those within the formal meeting minutes

685
02:18:47.609 --> 02:19:15.729
Mikaela Miller: which brings us to follow up. We'll go ahead and wrap up these notes, Stephanie, do you want to go ahead and put a feedback form in the chat? We have a link for all of the voting members? If you could fill that out and just give us your feedback as far as how the meeting went for today. If there's anything we can improve for the next meeting, we will go ahead and

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02:19:16.625 --> 02:19:29.769
Mikaela Miller: go through that feedback. We'll get all the materials put together, all of the slides. The minutes, the recording all of this will be put on the website for the public. We usually try to do it within

687
02:19:29.809 --> 02:19:49.059
Mikaela Miller: a couple of weeks, and then we will start getting everything prepared for our next meeting in the fall. Doodle Poll should be going out to all of you committee members here within the next month or so to determine when we can hold that meeting

688
02:19:49.059 --> 02:20:09.849
Mikaela Miller: which will include a vote on whether or not we want to include Asmd on the Montana panel. So we do try to, or we need to ensure that we have a quorum for that meeting.

So that just means that we need to have at least 5 of 9 voting members present. So once we find a date that works for

689
02:20:10.269 --> 02:20:14.479
Mikaela Miller: at least 5 of the members, we can go ahead and get that meeting scheduled

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02:20:15.909 --> 02:20:23.809
Mikaela Miller: alright. Well, I'll let everyone go just a little bit early here today. But once again, thank you all for being here.

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02:20:23.810 --> 02:20:24.640
Justin Hopkin: Mikaela, could I?

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02:20:24.640 --> 02:20:25.670
Jenn Banna MTF2FHIC: Thank you. Everyone.

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02:20:25.670 --> 02:20:54.639
Justin Hopkin: Thank everybody for the opportunity, but also for the work that you're doing, especially having patient engagement and the volunteers from the community. I think it's great. The questions were fantastic. Everybody was spot on with identifying, I think, really important aspects of Asmd. The treatment the program. So thanks for allowing me to to present today. Thanks for the great questions, and thank you for doing really important work and probably volunteering a lot of your time. It's appreciative.

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02:20:55.640 --> 02:21:04.379
Jenn Banna MTF2FHIC: Thank you for sharing your story. I know you said you've done that a lot, but I'm a parent that shared my story, too, so I know how our stories are, so thank you for doing that.

695
02:21:04.380 --> 02:21:05.050
Justin Hopkin: Thank you.

696
02:21:06.590 --> 02:21:08.080
Jenn Banna MTF2FHIC: Have a great day. Everyone.

697

02:21:09.250 --> 02:21:09.799
Abe Elias: Yeah, I think.

698
02:21:09.800 --> 02:21:10.590
Mikaela Miller: Bye, everyone.

699
02:21:10.590 --> 02:21:12.660
Abe Elias: Bye-bye, bye.