

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 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 Macula, 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 lipase, which goes by the brand name Zenposyme, 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 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 lipase, 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 biallelic 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 lipase or Zempazone 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 lcd 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

320

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.

325

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

345

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.

350

01:09:26.300 --> 01:09:31.360

Jeanne Lee: You may be questioning why we send testing to Wisconsin

351

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

352

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.

353

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.

356

01:10:36.000 --> 01:10:39.030

Jeanne Lee: So here we are with Asmd.

357

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.

360

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.

364

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.

368

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.

379

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

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

380

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

382

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 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 persevere 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

524

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 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.

548

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.

549

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 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 10%. 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

565

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.

566

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

569

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.

570

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.

581

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.

582

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.

583

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

584

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.

589

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.

590

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

594

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?

595

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

612

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.

621

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

630

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.

650

02:13:07.620 --> 02:13:10.630

Jenn Banna MTF2FHIC: Do you want to take us back to the previous slide, Michaela?

651

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

652

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 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?

678

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

686

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

690

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.

691

02:20:23.810 --> 02:20:24.640

Justin Hopkin: Mikaela, could I?

692

02:20:24.640 --> 02:20:25.670

Jenn Banna MTF2FHIC: Thank you. Everyone.

693

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 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.

694

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.