

GFPD EL PFDD Meeting Polling and Discussion Questions

Tuesday, May 28, 2024

9:00 AM – 12 PM **Polling Questions:**

1. Are you a:

- Patient with a peroxisomal disorder
- Caregiver (Parent/Stepparent/Legal guardian) of a patient with a peroxisomal disorder
- Other family member of a patient with a peroxisomal disorder

2. What is your/your loved ones' diagnosis?

- Zellweger spectrum disorder (ZSD)
- Acyl-CoA oxidase 1 deficiency (ACOX 1)
- D-bifunctional protein deficiency
- Methylacyl-CoA racemase deficiency (AMACR)
- Adult Refsum disease (ARD)
- X-linked adrenoleukodystrophy (X-ALD)
- Other (Please specify) _____

3. What symptom led the doctor to the diagnosis of peroxisomal disorder in you or your loved one?

- Deaf/Blindness
- Low muscle tone
- Inability to feed orally
- Liver dysfunction
- Frequent Vomiting/Reflux symptoms
- Seizures
- Other, please specify

4. What is the age category of you or your loved one? If your loved one has passed away, please enter the age category of your child when they passed away.

- 0-10 years
- 11-20 years
- 21 years or above

Panel #1: Symptoms and Daily Impacts Panel

- **Sensory Loss Polling Question (5 min):**

5. Are there specific activities that are important to you but that you/your loved one cannot do at all or fully because of your/their sensory losses? (Select up to three that apply the best)

- Independent activities (mobility, feeding, dressing, personal hygiene)
- Social interactions (family, friends, and meeting new people)
- School/academic and work performance
- Executive functioning (decision-making, staying focused, bringing tasks to completion)
- Other, please specify

6. How has your or your loved ones sensory losses changed over time?

- Increased over time
- Decreased over time
- Not changed over time

- **Large-Group Facilitated Discussion on Sensory Loss (10 min)**

7. Do you have anything else to add about your/your loved one's sensory loss? (open-ended)

- **Mobility Polling Question (5 minutes):**

8. Are there specific activities that are important to you but that you/your loved one cannot do at all or fully because of your/their mobility issues? (Select up to three that apply the best)

- Independent activities (mobility, feeding, dressing, personal hygiene)
- Social interactions (family, friends, and meeting new people)
- School/academic and work performance
- Executive functioning (decision-making, staying focused, bringing tasks to completion)
- Other, please specify

9. How have your or your loved ones' mobility changed over time?

- Improved over time
- Worsened over time
- Not changed over time

- **Large-Group Facilitated Discussion on Mobility Symptoms (10 min)**
 - 10. Do you have anything else to add about your/your loved one's mobility issues? (open-ended)**
- **Intellectual Disability Polling Question (5 minutes):**
 - 11. Are there specific activities that are important to you but that you/your loved one cannot do at all or fully because of your/their intellectual disability? (Select up to three that apply the best)**
 - Independent activities (mobility, feeding, dressing, personal hygiene)
 - Social interactions (family, friends, and meeting new people)
 - School/academic and work performance
 - Executive functioning (decision-making, staying focused, bringing tasks to completion)
 - Other, please specify
 - 12. How has your or your loved ones cognitive ability changed over time?**
 - Improved over time
 - Worsened over time
 - Not changed over time
- **Large-Group Facilitated Discussion (10 min)**
 - 13. Do you have anything else to add about your/your loved one's intellectual disability? (open-ended)**
- **Seizures, GI, Respiratory and Other Systems Polling Question (5 minutes):**
 - 14. Of all the symptoms due to you/your loved ones condition, which symptoms (up to 3) have the most significant impact on you/your loved ones life?**
 - Deaf/ blindness
 - Seizures/Neurological Symptoms
 - Low muscle tone
 - Developmental/cognitive delays
 - GI/Feeding Difficulties
 - Behavioral Symptoms
 - Respiratory Distress
 - Kidney/Bladder/Urinary Symptoms
 - Adrenal Insufficiency
 - Other

15. Are there specific activities that are important to you but that you/your loved one cannot do at all or fully because of these symptoms? (Select up to three that apply the best)

- Independent activities (mobility, feeding, dressing, personal hygiene)
- Social interactions (family, friends, and meeting new people)
- School/academic and work performance
- Executive functioning (decision-making, staying focused, bringing tasks to completion)
- Other, please specify

- **Large-Group Facilitated Discussion (10 min)**

Polling Question

16. Do you have anything else to add about the symptoms that your/your loved one faces or has faced? (open-ended)

- **Leukodystrophy and Disease Progression Polling Question (5 minutes):**

17. How has your or your loved ones' condition and symptoms changed over time?

- Improved over time
- Worsened over time
- Not changed over time

- **Large- Group Facilitated Discussion (5 min)**

18. When thinking about the progression of PD what worries you most about this condition?

9:30 – 10:45 AM

- **Large-Group Facilitated Discussion on Symptoms and Impact of PD (5 min)**

- **Discussion Questions: 20 min**

19. How do/did you/your loved ones symptoms and their negative impacts affect your/their daily life on the best days?

20. How do you/your loved ones symptoms and their negative impacts affect your/their daily life on the worst-days?

- **Polling Question: 2 min**

21. Would you define your or your loved ones' condition today as being well managed?

- Yes, it is well managed
- No, it is not well managed

- I am not sure

12:00 – 2:00 PM

2:00 – 3:20 PM

Panel #2: Current Approach to Treatments Panel

- **Treatment Polling Question:**

22. What are you currently doing to help treat your/your loved ones' condition?

- Treatment for sensory impairment (hearing/vision loss)
- Treatment for feeding or gastrointestinal symptoms
- Treatment of mobility symptoms
- Treatment for seizures
- Treatment for adrenal insufficiency
- Treatment for bleeding issues and anemia
- Treatment for intellectual disability
- Treatment for respiratory symptoms
- Treatment for behavioral symptoms
- Treatment for bone disease
- Treatment for another symptom (please specify)

- **Large-Group Facilitated Discussion (5 min)**

23. How well does you or your loved ones' current treatment regimen treat the most significant symptoms of your/their disease?

- **Polling Question (5 min)**

24. How has your or your loved ones' treatment regimen changed over time?

- Increased over time
- Decreased over time
- Not changed over time

- **Discussion Question (5 min)**

25. How well have these treatments worked for you/your loved ones as your/their condition has changed over time?

Large-Group Facilitated Discussion on Topic #2 (34 min)

- **Polling Questions (5 min)**

- 26. **What are the most significant downsides to your/your loved ones current treatments, and how do they affect your/their daily life?** (open-ended question on poll everywhere)
- **Large-Group Facilitated Discussion (8 min)**
- **Discussion Question**
 - 27. **How have these downsides affected your/your loved ones daily life?**
 - 28. **Short of a complete cure, what specific things would you look for in an ideal treatment for your/your loved ones condition?**
- **Polling Question (5 min)**
 - 29. **What would you consider to be a meaningful improvement in your/your loved ones condition that a treatment could provide?** (open-ended question on poll everywhere)

3:20 – 3:25 PM

Panel #3: Clinical Trial Readiness Panel

3:25 – 5:00 PM

Large-Group Facilitated Discussion on Topic #3 (45 min)

- **Discussion Question (10 min)**
 - 30. **What do/did you consider when thinking about participating in a clinical trial?**
- **Polling Question (5 min)**
 - 31. **Would you participate in a randomized clinical trial, in which your loved one could receive a placebo instead of a drug, but in which they would also have the opportunity to receive the drug at a later time?**
 - Yes, I would participate
 - No, I would not participate
 - I'm not sure if I would participate
 - It would depend on several factors
- **Large-Group Facilitated Discussion**
 - 32. **What are some of these factors to consider enrolling you/your loved one in a clinical trial for PD?**
- **Other Discussion Questions**
 - 33. **In the event that a new treatment/therapy is associated with severe or life-threatening risks, what is the minimal benefit that you find worthwhile to accept those risks and**

enroll yourself/your child in an interventional clinical trial?

(Prompt: 50% reduction in seizures, O2 saturation above 90%, etc)

34. **How could clinical trials be better tailored for the patients, parents, and caregivers?**
35. **Is there anything else that was not mentioned that you would like to share with the FDA and other regulatory agencies?**