

Externally Led Patient Focused Drug Development in Peroxisomal Disorder (PD) Patient Survey

Consent Online Consent Form

Please read below with care. You can ask questions at any time, now or later. You can talk to other people before you click on the link to consent.

Title: Patient-Focused Drug Development in Peroxisomal Disorders (PD)

Study Number: IRB-FY22-23-2922

Why is this study being done? The purpose of this study is to understand how patients with peroxisomal disorders (PD) are affected by their disease, what they want to improve in their daily experience, and what they need to take part in a clinical trial for PD. Your input in this study will be important to all families affected by a PD.

You may be eligible to participate in this study if you are 18 years of age or older and you are either:

- 1) a person diagnosed with a PD
- 2) a family caregiver to a person diagnosed with a PD. This includes parents, legal guardians, stepparents, grandparents, siblings or any other relative that provides care to a person with a PD.

What will happen while you are in the study? You will be asked to complete a confidential online survey about your or your family member's experience with symptoms, treatments, and clinical trial participation related to PD's.

Time: You will be asked to complete the survey one time. The survey will take up to an hour to complete.

Risks: You may feel distress or confusion from recalling sensitive issues related to your PD or your experience caring for the individual with a PD. If you feel any discomfort, you are free to take a break or stop taking the survey altogether. *Additionally, you may contact Katie Sacra, Director of Family Programs at the GFPD at Katie@thegfpd.org for further support.*

Data will be collected using the internet; we expect that your participation offers no greater risk than everyday use of the internet. Confidentiality will be kept to the extent allowed by the technology used. Please note that email communication is neither private nor secure. Though we are taking precautions to protect your privacy, you should be aware that information sent through email or internet could be read by a third party. We strongly advise that you do not use an employer-issued device, laptop, phone or WIFI to take this survey, as many employers monitor use of all devices.

Benefits: By sharing your experiences or the experiences of your family member with a PD, you may help advance healthcare for you or your family member. Furthermore, this study will set the foundation for tools that may be used in clinical trials of therapies for PD's.

Compensation: You will not be compensated for your participation in this study.

Who will know that you are in this study? Your name will not be linked to any presentations or publications related to this study. We will make sure that your name and any personal or medical information you give us will be kept private. All computer data files will be password-protected. Although we will keep who you are confidential, we strongly advise that you do not use an employer issued electronic device, laptop, phone or WIFI to respond to these surveys, as many employers monitor use of all devices. *You should know that New Jersey requires that any person having reasonable cause to believe that a child has been subjected to child abuse or acts of child abuse should report immediately to the Division of Youth and Family Services.*

Do you have to be in the study? You do not have to be in this study, your participation is voluntary. It is okay if you want to stop at any time and not be included in the study. You do not have to answer any questions you do not want to answer.

Do you have any questions about this study? Phone or email the principal investigator, Dr. Mousumi Bose, at 973-655-3358 or bosem@mail.montclair.edu.

Do you have any questions about your rights as a research participant? Phone or email the IRB Chair, Dr. Dana Levitt, at 973-655-2097 or reviewboard@montclair.edu.

Confirmation of consent

By clicking "I agree to participate in this study" below, I confirm that I have read this form and will participate in the project described. Its general purposes, the involvement, and possible risks and inconveniences have been explained to my satisfaction. I understand that I can leave the study at any time. My consent also indicates that I am at least 18 years of age.

A) I agree to participate in this study

B) I decline to participate in this study

By clicking "It is okay to audio and video record me during the 2024 GFPD conference", I confirm that I will consent to having my participation recorded during the conference. (Please select "Not Applicable" if you are not attending the 2024 GFPD conference)

A) It is okay to audio and video record me during the 2024 GFPD conference

B) It is NOT okay to audio and video record me during the 2024 GFPD conference

C) Not applicable

Demographics & Diagnosis

Please answer the following questions about your demographic details as well as your PD diagnosis.

Q1 What is your diagnosis?

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

Q2 How were you diagnosed with the PD? Please select all that apply.

- A) Newborn screening for X-linked adrenoleukodystrophy
- B) Elevated plasma VLCFA levels
- C) Elevated plasma pristanic acid
- D) Elevated plasma phytanic acid
- E) Decreased RBC plasmalogen levels
- F) Elevated C20 bile acids
- G) Elevated plasma and /or urine pipecolic acid levels
- H) Genetic diagnostic testing (sequencing of PEX genes)
- I) Exome sequencing
- J) Genome sequencing
- K) Not sure/ I don't remember
- L) Other not listed (Please Describe)

Q3 What symptom led your medical care team to test for a PD diagnosis? Please select all that apply

- A) Low muscle tone
- B) Inability/difficulty to feed
- C) Failed or abnormal hearing test
- D) Failed or abnormal vision test
- E) Respiratory distress
- F) Liver dysfunction
- G) Distinctive facial features
- H) Seizures
- I) Failure to thrive
- J) Developmental delay
- K) Adrenal dysfunction
- L) Leukodystrophy
- M) Peripheral neuropathy and ataxia
- N) Other (Please specify)
- O) Not applicable (I was diagnosed prenatally, newborn screening etc...)
- P) I don't know/ I don't remember

Q4 Are you:

- A) Male
- B) Female
- C) Nonbinary
- D) Prefer not to answer
- E) Not listed, Please describe your gender

Q5 Are you currently:

- A) Single
- B) Married/Domestic Partnership
- C) Widowed
- D) Divorced/Separated
- E) Other (Please explain)
- F) Prefer not to answer

Q6 What is the highest level of education you completed?

- A) Elementary (grade 5 or less)
- B) Middle School (grade 6-8)
- C) High School or equivalent (grade 9-12)
- D) Some college
- E) Associates Degree
- F) Bachelor's Degree or equivalent
- G) Masters Degree or equivalent
- H) Doctoral degree (PhD, MD, PharmD, JD, etc...)
- I) Other licensure, certification or diploma not listed
- J) Prefer not to answer

Q7 Are you currently employed or working?

- A) Yes B) No

Q8 Which of the following best describes you? (please choose one option)

- A) Hispanic, Latino(a/x), or Spanish
- B) American Indian or Alaska Native
- C) Black or African-American
- D) East Asian
- E) Middle Eastern
- F) Native Hawaiian or Other Pacific Islander
- G) South Asian
- H) Southeast Asian
- I) White
- J) Multiracial or Multi-ethnic
- K) Race/Ethnicity not listed
- L) Unknown/Prefer not to answer

Q9 Please describe your specific ethnic heritage:

Q10 Which of the following best describes your current annual household income?

- A) Less than \$25,000
- B) \$25,000 - \$49,999
- C) \$50,000 - \$74,999
- D) \$75,000 - \$99,999
- E) \$100,000 - \$149,999
- F) \$150,000 - \$199,999
- G) \$200,000 and above
- H) Don't know
- I) Prefer not to answer

Q16 Where do you live?

- A) Midwest - IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, SD, WI
- B) Northeast – CT, DC, DE, MA, MD, ME, NH, NJ, NY, PA, RI, VT
- C) Southeast – AL, AR, FL, GA, KY, LA, MS, NC, SC, TN, VA, WV
- D) Southwest – AZ, NM, OK, TX
- E) West – AK, CA, CO, HI, ID, MT, NV, OR, UT, WA, WY
- F) United States Territory – Puerto Rico, Guam, US Virgin Islands, American Samoa, Northern Mariana Islands
- G) Outside United States, please indicate which country

Symptoms and Impact

Please answer the following questions on symptoms and the impact of your PD.

Q1 Do you currently have or previously had any of the following symptoms of PD? Please select all that apply.

- A) Vision loss
- B) Hearing loss
- C) Deaf-blindness
- D) Sensory processing disorder
- E) Low muscle tone
- F) Ataxia
- G) Balance problems
- H) Tremors
- I) Gross motor delays
- J) Bone density issues
- K) Seizures
- L) Leukodystrophy
- M) Orthopedic symptoms (Mobility issues, pain, bone deformities etc...)
- N) Inability to feed orally
- O) Frequent vomiting/reflux symptoms
- P) Bowel movement difficulty
- Q) Anemia
- R) Difficulty swallowing
- S) Choking on food
- T) Gastroparesis
- U) Liver dysfunction (elevated liver enzymes, liver fibrosis etc...)
- V) Dental symptoms (delayed tooth eruption, enamel issues, etc...)
- W) Behavioral symptoms (anxiety, aggressiveness, obsessive compulsive disorder, mood shifts etc...)
- X) Respiratory distress
- Y) Bladder/urinary symptoms
- Z) Developmental/cognitive delays
- AA) Adrenal insufficiency
- BB) Other symptoms (Please specify)

Q2 Please rank your symptoms from most impactful to least impactful in your life by moving the boxes below from top to bottom. (1 = most impactful, highest # = least impactful, only answer choices selected in Q1 will carry forward to Q2)

- _____ Vision loss
- _____ Hearing loss
- _____ Deaf-blindness
- _____ Sensory processing disorder
- _____ Low muscle tone
- _____ Ataxia
- _____ Balance problems
- _____ Tremors

- ☐ Gross motor delays
☐ Bone density issues
☐ Seizures
☐ Leukodystrophy
☐ Orthopedic symptoms (Mobility issues, pain, bone deformities etc...)
☐ Inability to feed orally
☐ Frequent vomiting/reflux symptoms
☐ Bowel movement difficulty
☐ Anemia
☐ Difficulty swallowing
☐ Choking on food
☐ Gastroparesis
☐ Liver dysfunction (elevated liver enzymes, liver fibrosis etc...)
☐ Dental symptoms (delayed tooth eruption, enamel issues, etc...)
☐ Behavioral symptoms (anxiety, aggressiveness, obsessive compulsive disorder, mood shifts etc...)
☐ Respiratory distress
☐ Bladder/urinary symptoms
☐ Developmental/cognitive delays
☐ Adrenal insufficiency
☐ Other symptoms (Please specify)

Q3 If you have anything else to add about **these symptoms** please indicate which symptoms and share here: _____

Q4 If you have any comments on any additional symptoms please add them here: _____

Q5 Based on your observation, how much have each of the following symptoms impacted your daily life? Please indicate on the scale provided, where a score of 0 indicates no importance and a score of 10 indicates greatest importance. If you do not have a symptom that is listed, please select Not Applicable for that symptom in order to move to the next section.

	0 = No Importance	10 = Greatest Importance	Not Applicable
Cognitive delay			
Verbal speech/communication impairment			
Behavioral and learning disorders (ADHD, OCD, Oppositional defiant disorder, etc.)			
Sensory processing disorder (sensitivity to bright light, loud sounds, scratchy clothing, food textures, etc.) ()			

Gross motor impairment (walking, balance, coordination issues)	
Fine motor impairment (problems with writing, tying shoes, buttoning shirts)	
Seizures	
Feeding issues (picky eater, trouble swallowing, etc.)	
Sleep/Fatigue issues (sleep disturbances, needing increased naps/breaks)	
Symptom not listed, please describe and rate another symptom that has had an impact on your life (please select Not Applicable if there is no additional symptom)	

Q6 When thinking about a potential treatment for peroxisomal disorders, what one activity of your daily life would you find most important to preserve or to restore? Please rank the activities that apply to your life by moving the boxes below from top to bottom (1 = most impactful, highest # = least impactful)

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

Q7 Based on your observation, how much has the PD impacted the following activities in your daily life? Please indicate on the scale provided, where a score of 0 indicates no importance and a score of 10 indicates greatest importance. If you do not participate in an activity listed, or you cannot comment on that activity, please select Not Applicable for that activity in order to move to the next section.

0 = No Importance 10 = Greatest Importance Not Applicable

Social interactions (family, friends, and meeting new people)	
School/academic performance	
Executive functioning (decision-making, staying focused, bringing tasks to completion)	

Independent activities (mobility, feeding, dressing, personal hygiene)	
Activity not listed, please describe and rate another activity (please select Not Applicable if there is no additional activity)	

Q8 How important is it to improve the following aspects of your daily life activities? Please indicate on the scale provided, where a score of 0 indicates no importance and a score of 10 indicates greatest importance. If you do not participate in an activity listed, or you cannot comment on that activity, please select Not Applicable for that activity in order to move to the next section.

0 = No Importance 10 = Greatest Importance Not Applicable

Walking	
Seizures	
Feeding	
Communicating effectively	
Playing/interacting with friends	
School performance, grades, work	
Control of sensory stimulation and other behavioral symptoms	
Sleep patterns	
Improve Independence	
Not listed, please describe and rate something else you would like to see improved (please select Not Applicable if there is nothing more to improve)	

Treatments for PD's

Please answer the following questions regarding treatments for PD's.

Q1 Have you ever received a:

- A) Liver/hepatocyte transplant
- B) Stem cell transplant
- C) Other transplant (Please specify) _____
- D) I have not undergone any transplant procedures

Q2 Have you ever been treated for any of the following symptoms associated with your PD? (please select all that apply)

- A) Treatment for sensory impairment (vision loss, hearing loss...
- B) Treatments or seizures
- C) Treatment for adrenal insufficiency
- D) Treatment for bleeding issues and anemia
- E) Treatment for feeding and gastrointestinal symptoms (Feeding problems, Vomiting, Bowel movement difficulties, Gastroparesis, Liver dysfunction...)
- F) Treatment of ataxia and mobility symptoms (Low muscle tone, Balance problems, Ataxia, Gross motor delays...)
- G) Treatment for bone disease
- H) Treatment for bladder issues
- I) Treatment of breathing issues
- J) Treatment for behavioral symptoms (Mood shifts, Anxiety, Aggressiveness...)
- K) Treatment for intellectual disability
- L) Other Conditions (Please describe the condition)
- M) I have never been treated for symptoms associated with my PD

Q3 Please select all medications/ treatments you have used to treat sensory impairments (Hearing and vision impairments)(if applicable)

- A) Physical devices (Hearing aids, cochlear implants...)
- B) Medications (acetazolamide ...)
- C) Therapies (Vision therapy, speech therapy...)
- D) I don't know/ I don't remember
- E) Other (Please state medication/therapy and what symptom it's treating)_____

Q4 Please select all medications / treatments you have used to treat seizures or spasms (if applicable)

- A) Benzodiazepine anticonvulsants (Clonazepam, Lorazepam, Diazepam, Misazolam/Buccolam, Clobazam...)
- B) Carbamazepine anticonvulsants (Oxcarbazepine...)
- C) Barbiturate anticonvulsants (phenobarbital (Luminal), Primidone...)
- D) Carbonic anhydrase inhibitor anticonvulsants (Topiramate (Trokendi, Qudexy, Topamax), zonisamide (Zonegran), Diamox...)
- E) Fatty acid derivative anticonvulsants (Valproate/valproic acid...)
- F) Gamma-aminobutyric acid analogs (vigabatrin, gabapentin, pregabalin...)
- G) pyrrolidine anticonvulsants (levetiracetam (Keppra)...)
 - H) Marijuana derivatives (CBD oil, THC...)
- I) Miscellaneous anticonvulsants (lacosamides...)
- J) Phenyltriazine (Lamotrigine (Lamictal)...)
 - K) Trihexyphenidyl (Artane, Pacitane, Parkin)
 - L) AMPA receptor antagonists (Perampanel (Fycompa)...)
 - M) I don't know/ I don't remember
 - N) Other (Please specify _____)

Q5 Please select all medications / treatments you have used to treat adrenal insufficiency (if applicable)

- A) Steroids (Hydrocortisone, Cortef, Fludrocortisone, Fluticasone...)
- B) I don't know/ I don't remember
- C) Other (please specify _____)

Q6 Please select all medications / treatments you have used to treat bleeding issues and anemia. (if applicable)

- A) Vitamin K
- B) Iron
- C) Blood transfusions
- D) I don't know/ I don't remember
- E) Other (please specify _____)

Q7 Please select all medications/treatments you have used to treat feeding and gastrointestinal symptoms (if applicable)

- A) Anti-Constipation medications (Senecot, Miralax, Dulcolax...)
- B) Anti- Constipation devices (Enema...)
- C) Probiotics
- D) Fiber supplement
- E) Feeding therapy
- F) Medications for GI motility (Erythromycin, lactulose...)
- G) Physical devices for GI motility (baclofen pump...)
- H) Antireflux medications (Antacids, Omeprazole, Esomeprazole, Lansoprazole, Rabeprazole, Pantoprazole, Dexlansoprazole...)
- I) Appetite stimulant (Cyproheptadine ...)
- J) Enteral tube placement (Gastrostomy tube, jejunostomy tube...)

- K) Cholic acid (Cholbam, Ursodiol)
 - L) I don't know/ I don't remember
 - M) Other (Please state the medication/therapy and indicate what symptoms it's treating)
-

Q8 Please select all medications/treatments you have used to treat ataxia and mobility symptoms (if applicable)

- A) Therapies (Physical therapy, occupational therapy, orientation and mobility O&M...)
 - B) Orthopedic surgery
 - C) Orthotic footwear
 - D) Beta-Blockers (Propanolol...)
 - E) Wheelchair
 - F) I don't know/ I don't remember
 - G) Other (Please state the medication/therapy and indicate what symptoms it's treating)
-

Q9 Please select all medications/treatments you have used to treat bone disease (if applicable)

- A) Calcium
- B) Vitamin D
- C) Bisphosphonate medications
- D) I don't know/ I don't remember
- E) Other (please specify)_____

Q10 Please select all medications/treatments you have used to treat bladder/urinary issues (if applicable)

- A) Prophylactic antibiotics
- B) Urinary retention medication (Flomax / tamsulosin)
- C) I don't know/ I don't remember
- D) Other (please specify)_____

Q11 Please select all medications/treatments you have used to treat breathing issues (if applicable)

- A) Breathing treatment (oxygen support., Saline inhalation treatment...)
- B) Bronchodilators (Albuterol...)
- C) Nebulizers
- D) I don't know/ I don't remember
- E) Other (Please state the medication/therapy and indicate what symptoms it's treating)_____

Q12 Please select all medications/treatments you have used to treat behavioral symptoms *(if applicable)*

- A) Anti-anxiety medications (buspirone...)
- B) Antidepressants (Prozac, Fluoxetine...)
- C) Antipsychotics (risperidone...)
- D) Benzodiazepines (Ativan...)
- E) Multiple behavior symptoms medications (Zoloft...)
- F) I don't know/ I don't remember
- G) Other (Please state the medication/therapy and indicate what symptoms it's treating)_____

Q13 Please indicate all treatments / therapies that you have used to treat intellectual disability *(if applicable)*

Q14 Please select all dietary supplements that you have used to manage your condition

(Please indicate in the text box the specific supplement) (if applicable)

- A) Fat soluble vitamins (vitamin A, D, E, K) _____
- B) Water soluble vitamins (folate, thiamine, riboflavin, niacin, pantothenic acid, biotin, vitamin B6, and vitamin B12 and vitamin C) _____
- C) Vitamin K only
- D) Mineral supplements (calcium, phosphorus, magnesium, sodium, potassium, chloride, sulfur, iron, zinc, copper, selenium, manganese, iodine, chromium and molybdenum)_____
- E) Food fortifiers (carbohydrate or protein supplement) _____
- F) DHA, fish oil, cod liver oil _____
- G) Medium chain triglycerides supplements (MCT)
- H) Multivitamin
- I) Other dietary supplements (creatine, collagen, probiotic, fiber, betaine...) _____
- J) I don't know/ I don't remember
- K) I do not take any dietary supplements to manage my PD

Q15 Please list all medications / therapies that you have used to treat other conditions *(if applicable)*

Clinical Trial Participation

Please answer the following questions about clinical trial participation. Please note that all questions about clinical trials are referring to **interventional clinical trials** (where a new treatment or therapy may be administered) unless otherwise stated.

Q1 Has you ever participated in a research study for PDs?

- A) Yes B) No C) I don't know/I am unsure/I cannot remember

Q2 Please select the research studies in which you have participated in (select all that apply if applicable):

- A) DHA study by Dr Martinez - Barcelona, Spain
 B) DHA study by Dr Raymond-Maryland, USA
 C) Cholic acid therapy by Dr Bwee Tien Poll -Amsterdam , Netherlands
 D) Cholic Acid Therapy in Zellweger Spectrum Disorders Dr Heubi - Cincinnati, USA
 E) Betaine and Peroxisome Biogenesis Disorders Dr Rizzo & Dr Braverman - Quebec, Canada
 F) Other (Please specify)_____

Q3 If you had the opportunity to participate in a new clinical trial to study an experimental treatment for PD's, would you consider enrolling them in the study?

- A) Yes
 B) Maybe (Please describe the factors, for example, it depends on the type of drug, risks, time involved, etc....) _____
 C) No, not at this time, maybe in the future
 D) Definitely No

Q4 If you were to consider your participation in a new clinical trial, how important would each of the factors listed below be in making the decision to enroll or not to enroll? Please indicate on the scale provided, where a score of 0 indicates no importance and a score of 10 indicates greatest importance.

0 = No Importance 10 = Greatest Importance Not Applicable

Goals/outcomes of the trial (study of a drug that may improve motor skills, speech, seizures, behavior, sleep...) ()	
Types of assessments and tests performed during the study (blood work, surveys, MRI, spinal tap...) ()	
Visit schedule (number of tests during the day, accounting for fatigue and anxiety...)	
Risks of serious side effects (worsening of symptoms, unanticipated hospitalization, sudden death...)	
Risks of common side effects (headaches, nausea, gastrointestinal upset, skin irritation)	
Risk of worsening other symptoms	
How the test drug is administered (oral, G tube, i.v., i.m., intrathecal, inhalation, intranasal, implant, rectal, etc) and how often	

the treatment is given (once or multiple times a day, long i.v. perfusion...)	
Having to discontinue your current treatment or management plan (stopping a medication, supplement, or therapy)	
Your confidence in the Clinical Research team (their ability to listen to and accommodate your needs, to communicate and give you feedback, to empathize ...)	
Distance to travel to the clinical study site	
Having to coordinate accommodations for travel (booking flights, lodging, transportation, meals)	
Length of visits to the clinical study site	
Timing of visits to the clinical study site (during school year, during the summer, etc.)	
Frequency of visits to the clinical study site	
Option to have testing done at home	
Accessibility and cost	
Risk of being in the placebo group and not benefiting from the treatment ()	

Q5 If a new therapy is associated with severe or life threatening risk would you still consider enrolling in a clinical trial?

- A) It would depend on the potential benefit of the clinical trial B) No

Q6 If a new therapy or treatment in a clinical trial is associated with severe or life-threatening risks, what is the minimal benefit on symptoms or impact of PD that you would find worthwhile to accept those risks and participate in a clinical trial?

- A) Improvement of sensory impairment
 B) Improvement of seizures
 C) Improvement of adrenal insufficiency
 D) Improvement of bleeding issues and anemia
 E) Improvement of feeding and gastrointestinal symptoms
 F) Improvement of ataxia and mobility symptoms
 G) Improvement of bone disease
 H) Improvement of bladder issues
 I) Improvement of breathing issues
 J) Improvement of behavioral symptoms
 K) Improvement of intellectual disability
 L) Improvement of other symptoms (Please specify) _____

Q7 Would you participate in a randomized clinical trial, in which you could receive a placebo instead of drug, but in which they would also have the opportunity to receive the drug at a later time (e.g., a cross-over or open-label extension trial)?

- A) Yes
 B) No
 C) I'm not sure
 D) Depends on other factors (Please describe) _____

Q8 Are there certain symptoms that you would be willing to tolerate in order to address your most urgent symptom in a clinical trial?

- A) Yes
 B) No
 C) Maybe
 D) It depends (Please specify) _____

Q9 Is there anything else that you consider in thinking about enrolling in participating in a clinical trial?

Q10 Are there any ways that clinical trials could be better tailored for patients?

Q11 Is there anything else that was not mentioned that you would like to share with the researchers, the GFPD, medical product developers, health care providers and federal partners (like the FDA)?
