Frequently Asked Questions about Peroxisomal Biogenesis Disorders (PBD)

(Compiled by The Global Foundation for Peroxisomal Disorders- www.thegfpd.org)

You have been told that your child has or may have a peroxisomal biogenesis disorder (PBD). This is a scary and difficult time for you and your family. All parents have questions about the diagnosis and what it means for their child and their family. We wish we could tell you it will all go away. Unfortunately it won't. You may have some really difficult days ahead of you. But please remember, there are many of us who have been there, done that. Please do not hesitate to come to us for advice and support. Here are some questions and responses from other parents whose families have been impacted by PBD. Keep in mind these are responses of parents, not doctors. They are not a replacement for your doctor's advice.

1.What is a peroxisomal biogenesis disorder (PBD)? Are there different types?
Peroxisomal Biogenesis Disorder (PBD) refers to disorders in the Zellweger Spectrum, which include: Infantile Refsum Disease (IRD), Neonatal Adrenoleukodystrophy (NALD), and Zellweger Syndrome. There are also two single enzyme peroxisomal disorders: acyl-CoA oxidase deficiency and D-bifunctional protein deficiency that are closely related to PBDs. These are all rare, genetic, metabolic, terminal conditions affecting all major systems of the body.

2. Why do they think my child has a PBD?
Global developmental delay is a hallmark characteristic of PBD. This is often accompanied by severe craniofacial abnormalities, including a high forehead, a flat occiput (the bone forming the rear and rear bottom of the skull), a large fontanelle (the child's "soft spot"), a broad nasal bridge, shallow orbital ridges (the ridge beneath the eyebrow), and a high arched palate. Another typical feature of the Zellweger Spectrum involves abnormalities of the eye, such as Brushfield spots (speckled iris, although this can occur in normal children), cataracts, and glaucoma. Hepatomegaly (enlargement of the liver), renal cysts, impaired adrenocortical function, and hypotonia (poor muscle tone, or "floppiness") are also common features. Many children, but not all, also fail their newborn hearing screening.

Biochemical assays can determine definitively whether an individual has a PBD. The measurement of plasma very-long-chain fatty acid (VLCFA) concentrations is the most commonly used and most informative initial screen. Elevation of the plasma concentrations of C26:0 and C26:1 and the ratios of C24/C22 and C26/C22 is consistent with a defect in peroxisomal fatty acid metabolism. The degree of VLCFA plasma concentration elevation may vary, with a small percentage of individuals demonstrating only modest elevations. For more information visit the GENE Review for Peroxisomal biogenesis disorders at http://www.ncbi.nlm.nih.gov/books/NBK1448/

3. How does a child become affected by a PBD? Will it happen again?
PBD is inherited in an autosomal recessive manner, meaning each parent is an unaffected (asymptomatic) carrier. Any additional children conceived by parents who are both asymptomatic carriers have a 25% chance of being affected, a 50% chance of being an asymptomatic carrier, and a 25% chance of being unaffected and not a carrier. (GENE Reviews - http://www.ncbi.nlm.nih.gov/books/NBK1448/)

4. What type of pediatric medical specialists should my child see?
In addition to a primary care physician/pediatrician your child may need the following pediatric specialists: Audiologist, Endocrinologist, Gastroenterologist (GI), Geneticist, Neurologist, Ophthalmologist, Orthopedic, Otolaryngologist (ENT**)** and Pulmonologist.

5. What additional services does my child need?
Early intervention and ongoing therapy services such as: Physical therapy, occupational therapy, speech, feeding therapy/nutrition, hearing and vision services are necessary and prescribed for a child with a PBD throughout his/her lifetime.

6. What treatments exist for PBD?
While there is currently no cure for PBD, treatment is symptomatic. It is recommended that all children diagnosed with PBD take a multivitamin supplement which includes higher levels of vitamins A, D, E, and K (ex. Source CF and AquADEKs). Some children experience Coagulopathy (impaired blood clotting) and therefore require additional supplementation of vitamin K (ex. Mephyton). Many children also take a DHA supplement. Children with adrenal insufficiency will require daily steroid treatments as prescribed by their endocrinologist. Children experiencing seizures will require anti-seizure medicine as prescribed by their neurologist. Children also may experience spontaneous bone fractures as well as osteopenia. A pediatric orthopedic specialist can advise families on the best treatment for osteopenia. **Reminder: This information is not to take the place of your child's primary care physician and medical team.**

7. What does my child's future look like?
We often say that each child affected by PBD "writes their own story," because each one is unique. Even children with the exact same mutations are often impacted very differently by PBD. Children at the most severe end of the spectrum, often referred to as classic Zellweger Syndrome, typically do not survive their first year. Approximately half of all children diagnosed with PBD have this most severe form. Children on the mild end of the spectrum (NALD/IRD) may live into early childhood, late childhood, teen years or beyond; although the majority of children with PBD do not live past the age of ten.

8. How does a PBD affect my child's physical and cognitive abilities?
PBD affects each child differently, however nearly all children experience global delays along with combined hearing and vision impairments. Children who are at the most severe end of the spectrum typically meet very few developmental milestones. Children at the most mild end of the spectrum often learn to walk, talk, and a few can even read. Some children have suffered a regression resulting in the loss of skills previously gained. Therefore, it is difficult to determine where your child may ultimately fall along the spectrum.

9. How can I find out more about PBDs and connect with other parents who understand what I am going through? The Global Foundation for Peroxisomal Disorders, a 501(c)(3) public charity, is a resource for families who have received a diagnosis of Zellweger Syndrome, Neonatal Adrenoleukodystrophy (NALD), Infantile Refsum Disease (IRD), and D-Bifunctional Protein Deficiency through connections to medical & scientific professionals and family support networks.

Additionally, GFPD organizes family support and informational conferences, connects families through an online support group, and provides an equipment exchange program. GPFD also shares objective and credible information to families and caregivers of patients with PBDs and is a voice in the public arena for children affected by the disorders.

Please visit our website, www.thegfpd.org for more information. To be connected with our online family support groups, please contact Pamela Marshall, GFPD Family Registry and Support Group Coordinator by email: registry@thegfpd.org . For other inquiries or questions pleasecontact Shannon Butalla, GFPD President, by email: shannon@thegfpd.org or by phone: 402-429-5650.