THE LIVER A to ZSD

Peroxisomal biogenesis disorder-Zellweger spectrum disorder (PBD-ZSD), the liver, and what to discuss with your child's doctor

If your child was diagnosed with PBD-ZSD, it can leave you with a lot of questions. Use the Doctor Discussion Guide to have an important discussion about your child's liver health, any necessary testing, and management plans for PBD-ZSD.

Complete the PBD-ZSD Liver Symptom Checker

To help identify signs of liver involvement related to PBD-ZSD in your child.

PBD-ZSD Liver Symptom Checker

1 Which of the following symptoms has your child experienced? Check all that apply.

- ☐ Yellowing skin or white parts of eyes (jaundice)
- ☐ Oily, pale, foul-smelling stools (steatorrhea)
- ☐ Dark (tea-colored) urine
- ☐ Difficulties with feeding
- ☐ Poor growth delays (not meeting weight or height milestones for age)
- ☐ Reports of enlarged liver (hepatomegaly) from previous doctor visits

2 How old was your child when you first noticed symptoms?

- ☐ Infancy (0–1 year)
- ☐ Childhood (2–9 years)
- ☐ Pre-teen or older (10+ years)

3 Have these symptoms gotten worse, better, or stayed the same?

- ☐ Worse
- ☐ Better
- ☐ Stayed the same
- ☐ Not sure

4 Has your child been seen by a hepatologist (doctor specializing in the liver) before?

- ☐ Yes
- ☐ No

5 Are you aware of any tests your child's doctor has run that monitor your child's liver function?

- ☐ Yes (If yes, please list the tests below)
- ☐ No

Notes:

If you've selected any of these symptoms, it may be a sign of liver involvement. Talk to your child's doctor about testing for your child's liver health.

Doctor Discussion Guide

Questions to ask your child's hepatologist or gastroenterologist

Symptoms, treatment, and management

How rare is PBD-ZSD? Are you managing any other patients with PBD-ZSD?

How might my child's symptoms change over time?

Will my child develop any other liver symptoms? If so, can they be prevented or managed?

Is my child meeting height and weight benchmarks compared to other similarly aged children?

Is PBD-ZSD treatable? If so, what treatments are available?

Are you familiar with the recommended guidelines for treating and managing PBD-ZSD?

Which specialists will I need to work with, based on my child's symptoms? Can you provide a referral?

Should I modify my child's diet?

Monitoring the liver

Routine liver function tests are recommended for people with PBD-ZSD. How would I go about getting my child's liver tested?

How often should we run tests to check on the health of my child's liver?

Does my child need an atypical bile acid test? Are you familiar with atypical bile acid testing? Does my child need this test? If so, there is a free test available for patients with PBD-ZSD.

Write down any other questions below:

For more information about free atypical bile acid testing for PBD-ZSD patients, visit **TheLiverAtoZSD.com**

Doctor Discussion Guide (continued)

Which doctors should I talk to?

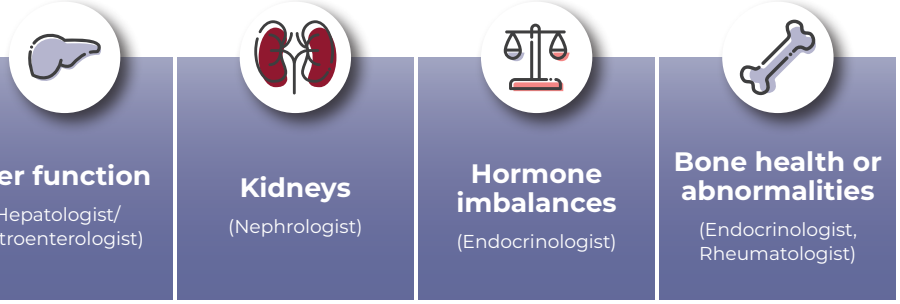
Since PBD-ZSD affects many parts of the body, you may need to work with different specialists to provide the best possible care for your child. Depending on your child's symptoms, you may need to talk to doctors who specialize in:

Nutrition and growth
(Hepatologist, Gastroenterologist, and Nutritionist)

Vision
(Ophthalmologist)

Hearing
(ear, nose, and throat [ENT] or Otolaryngologist/ Audiologist)

Nervous system
(Neurologist)

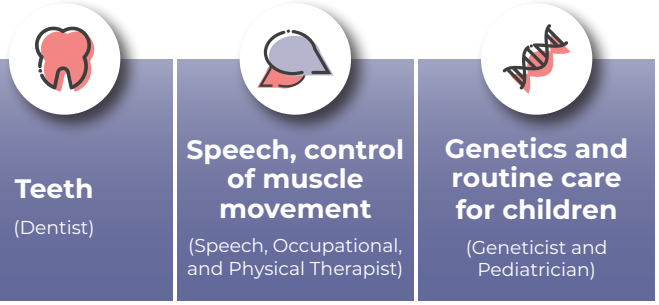


Liver function
(Hepatologist/ Gastroenterologist)

Kidneys
(Nephrologist)

Hormone imbalances
(Endocrinologist)

Bone health or abnormalities
(Endocrinologist, Rheumatologist)



Teeth
(Dentist)

Speech, control of muscle movement
(Speech, Occupational, and Physical Therapist)

Genetics and routine care for children
(Geneticist and Pediatrician)

Use the questions below as a guide when discussing your child's condition with a particular specialist.

Ask the following questions for each body system/function that affects your child.

When was the last time my child had his/her _____ checked? What were the results?

Are there any changes in my child's _____ we should address?

When should my child get his/her next evaluation for_____?

Write down any other questions below:

_____

_____

Bring this guide to your child's doctor's appointment

Talk to your child's doctor about running an atypical bile acid test to see if your child's liver is impacted.* For more information about testing, visit **TheLiverAtoZSD.com**.

*This test is offered for free through a program sponsored by Cincinnati Children's Hospital and Travere Therapeutics, Inc.

Key terms for PBD-ZSD

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Atypical: Not typical; irregular; abnormal.

Atypical Bile Acid Test: A less routine liver test that detects the levels of harmful bile acids known to cause liver damage.

Bile: Contains water, certain minerals, bile acids, cholesterol, and bilirubin (an orange-yellow pigment). Bile helps to digest dietary fats, vitamins, and nutrients, and eliminates excess cholesterol, bilirubin, waste, and toxins from the body.

Bile acids: Compounds that aid in digestion and absorption of dietary fats, vitamins, and other nutrients, and in the elimination of excess cholesterol, bilirubin, waste, and toxins from the body.

Jaundice: When a person's skin and the whites of the eyes are discolored yellow due to an increased level of bile pigments in the blood resulting from liver disease.

Liver Function Test: A group of blood tests that detect inflammation and damage to the liver. They can also check how well the liver is working.

Peroxisome: (pronounced: puh-ROK-suh-SOHMs) are in nearly every cell in the body; they help to make bile acids, which digest fats and fat-soluble vitamins that are important nutrients for growth and development.

Steatorrhea: The excretion of abnormal quantities of fat with the feces caused by reduced absorption of fat by the intestine.

Zellweger Spectrum Disorders: Also known as PBD-ZSD; a spectrum of disorders ranging from mild to moderate to severe, is caused by defects in peroxisome function.