PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS (PFIC)

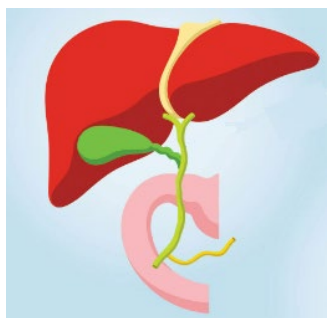
WHAT IS PFIC?

PFIC is a group of rare cholestatic liver diseases that impact at least 3,000 people in the U.S.[1]

It reduces the body's ability to eliminate bile, a liquid produced in the liver that helps rid the body of toxins and absorb important fats and vitamins. PFIC can lead to liver disease and ultimately liver failure.[2]

There are three main subtypes of PFIC: **PFIC1, PFIC2,** and **PFIC3**

Each is caused by a different change in one of the genes that govern the production and movement of bile throughout the body.[2]



HOW IS IT DIAGNOSED?

Most people living with PFIC experience liver failure before they reach adulthood.[2]

Symptoms caused by bile buildup, such as ongoing severe itching called pruritus, jaundice, and failure to grow and develop normally often begin in infancy and progress over time.

People with PFIC may also experience a variety of other symptoms, including:

- Short stature
- Deafness
- Diarrhea
- Inflammation of the pancreas (pancreatitis)
- Low levels of certain vitamins in the blood[2]

OFTEN DIAGNOSED IN INFANCY

Genetic testing can also provide information about the severity of the disease and subtype.[3]

Several tests are used to determine a PFIC diagnosis and identify which subtype is present, including blood tests to look at the levels of liver enzymes, as well as bile acid tests and a liver biopsy.

LIVING WITH PFIC

PFIC is much more than an itch.

The majority of people with PFIC have severe pruritus, which is associated with intense and often constant itching, and can lead to skin damage, hemorrhages, and scarring.

Severe pruritus can make it challenging to:

SLEEP
due to itching throughout the night.

PARTICIPATE
at school or in daily activities.[4]

Parents and children with PFIC can face a childhood marked by growth delays, developmental challenges and a variety of symptoms that require ongoing careful monitoring by a team of physicians.

Early diagnosis can help inform treatment options and focuses on supporting growth and development, and alleviating symptoms of pruritus, aiming to help children with the condition develop as normally as possible.[5] In severe cases, liver failure may require a liver transplant.[2]

For more support information and resources, visit:
PFIC Network

References

1. https://www.ncbi.nlm.nih.gov/books/NBK559317/
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3. https://www.pfic.org/learn-about-pfic-disease/pfic-diagnosis/
4. https://childrennetwork.org/Clinical-Studies/Progressive-Familial-Intrahepatic-Cholestasis
5. https://liverfoundation.org/liver-diseases/pediatric-liver-information-center/pediatric-liver-disease/progressive-familial-intrahepatic-cholestasis-pfic/