Short Communication

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Progressive Familial Intrahepatic Cholestasis

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Progressive Familial Intrahepatic Cholestasis (PFIC) or Byler disease is a disorder that can cause liver disease and, subsequently, liver failure. In children with Byler disease, cells in the liver cannot release bile, a fluid that helps the body digest food. Without enough of this fluid present, the body can't break down and absorb fats. The unreleased bile accumulates in the liver cells known as cholestasis. Cholestasis can damage the liver. The condition typically develops before adulthood, and it can progress very rapidly over a few months. PFIC is a rare disorder which affects around 1 in 50,000 – 1 in 100,000 births and it affects males and females equally [1-3].

Types of PFIC

There three main types of PFIC and the symptoms that they cause vary.

FIC 1 deficiency (PFIC 1): Mutations in the ATP8B1 gene cause deficiency of the FIC1 protein. PFIC1 often causes symptoms such as jaundice and itching in the first year of life.

BSEP deficiency (PFIC 2): Mutations in the ABCB11 gene are responsible for deficiency of the BSEP protein. There is an increased risk of liver cancer associated with BSEP deficiency.

MDR3 deficiency (PFIC 3): Mutations in the ABCB4 gene cause deficiency of the MDR3 protein. PFIC 3 can occur during infancy, childhood and even into young adulthood.

Symptoms

All three can cause liver disease and related complications, including: Jaundice (the yellow colouring of the whites of the eyes and the skin), Itching (pruritus), Failure to thrive (poor weight gain), Vitamin deficiencies (the fat soluble vitamins A, D, E and K), Enlarged liver, Gall stones, Portal hypertension, Swollen blood vessels in the lining of the oesophagus, Ascites (fluid retention in the abdomen), Liver failure and Increased risk of liver cancer.

Diagnosis

To tell if a patient has PFIC, some tests need to be done.

- Liver function tests are blood tests used to check the state of the liver or biliary system.
- Ultrasound, CT Sc an or MRI may be done to check the liver or biliary system.
- A liver biopsy may be done to check the liver tissue.
- A highly specialized test measuring bile salt levels may be done to pinpoint PFIC.

Treatment

There is no cure for PFIC but medicines, surgical treatments and changes to the diet can reduce the effects and complications of the condition. The treatments to be used will be recommended by the specialist depending on the features and severity of the condition and its effects. These include dietary treatments, medicines, potential operations, Partial external biliary diversion, Partial internal biliary drainage, internal ileal exclusion and liver transplant.

Dietary treatments: Dietary treatments used in children/young people with PFIC will depend on the specific symptoms and characteristics of the individual child.

Partial external biliary diversion: This is an operation which is only suitable in patients who have not developed cirrhosis. It is usually only considered when all medical treatments have failed to control itching (pruritus).

Liver transplant: This is a major operation with risks but a successful transplant can restore a good quality of life. Most children with PFIC will require a transplant at some point in their lives, whether as a child or in the future when they are adults.

References

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3 https://www.tgh.org/bylers-syndrome