ABCB11 gene

ATP binding cassette subfamily B member 11

Normal Function

The *ABCB11* gene provides instructions for making a protein called the bile salt export pump (BSEP), which is found in the liver. Bile salts are a component of bile that help the body digest fats. Bile salts are produced by liver cells and then transported out of the cell by the BSEP to make bile. The release of bile salts from liver cells is critical for the normal secretion of bile.

Health Conditions Related to Genetic Changes

Benign recurrent intrahepatic cholestasis

Variants (also called mutations) in the *ABCB11* gene can cause benign recurrent intrahepatic cholestasis type 2 (BRIC2). Benign recurrent intrahepatic cholestasis is characterized by episodes of liver dysfunction. People with BRIC2 have occasional episodes of impaired bile secretion that lead to severe itching (pruritus) and yellowing of the skin and whites of the eyes (jaundice). On occasion, people with BRIC2 have received a diagnosis for a more severe condition called progressive familial intrahepatic cholestasis type 2 (described below) when their symptoms worsened.

Affected individuals have a variant in both copies of the *ABCB11* gene. Variants in the *ABCB11* gene that cause BRIC2 lead to a 40 to 50 percent reduction of bile salt transport. The resulting buildup of bile salts in the liver leads to the signs and symptoms of BRIC2. It is unclear what causes the episodes to begin or end.

Progressive familial intrahepatic cholestasis

Many variants in the *ABCB11* gene have been found to cause a severe form of liver disease called progressive familial intrahepatic cholestasis type 2 (PFIC2). PFIC2 is a disorder that causes progressive liver disease, which often leads to liver failure. To develop this condition, a person needs to have a disease-causing variant in both copies of the *ABCB11* gene. Variants in the *ABCB11* gene that cause PFIC2 result in either a significant reduction or a complete absence of bile salt transport out of the liver. This causes bile salts to build up in liver cells, leading to liver disease and its associated signs and symptoms.

Variants that cause the cell to produce short, nonfunctional proteins or no proteins at all tend to be associated with severe liver disease that appears earlier in life. People with no functional BSEP protein also seem to be at a greater risk of developing a type of liver cancer called hepatocellular carcinoma.

Other Names for This Gene

- ABC16
- ATP-binding cassette, sub-family B (MDR/TAP), member 11
- bile salt export pump
- BRIC2
- BSEP
- PFIC2
- PGY4
- progressive familial intrahepatic cholestasis 2
- sister of p-glycoprotein
- SPGP

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ABCB11 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8647[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ABCB11%5BTIAB%5D%29+OR+%28BSEP%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ATP-BINDING CASSETTE, SUBFAMILY B, MEMBER 11; ABCB11 (https://omim.org/entry/603201)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/8647)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ABCB11[gene])

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Genomic Location

The *ABCB11* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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