ABCC2 gene

ATP binding cassette subfamily C member 2

Normal Function

The *ABCC2* gene provides instructions for producing a protein called multidrug resistance protein 2 (MRP2). This protein is one of a family of multidrug resistance proteins involved in the transport of substances out of cells. For example, MRP2 clears certain drugs from organs and tissues, playing a part in drug metabolism. Drug metabolism involves the breakdown of drugs into different chemical components allowing the drugs to have their intended effects and eventually be eliminated from the body. MRP2 also transports a substance called bilirubin out of liver cells and into bile (a digestive fluid produced by the liver). Bilirubin is produced during the breakdown of old red blood cells and has an orange-yellow tint.

MRP2 is primarily found within the outer membrane that surrounds cells in the liver, with smaller amounts in the kidneys, intestine, and placenta.

Health Conditions Related to Genetic Changes

Dubin-Johnson syndrome

More than 40 mutations in the *ABCC2* gene have been found to cause Dubin-Johnson syndrome. This condition is characterized by jaundice, which is a yellowing of the skin and whites of the eyes, that typically appears during adolescence or early adulthood. Most of the mutations change single protein building blocks (amino acids) in MRP2. A common mutation in Iranian Jews living in Israel who have Dubin-Johnson syndrome replaces the amino acid isoleucine with the amino acid phenylalanine at position 1173 in MRP2 (written as Ile1173Phe or I1173F). Another mutation that is seen more frequently in those affected in Israel's Moroccan-Jewish population replaces the amino acid arginine with the amino acid histidine at position 1150 in MRP2 (written as Arg1150His or R1150H).

ABCC2 gene mutations that cause Dubin-Johnson syndrome have a variety of effects on the structure and function of MRP2. Mutations may alter how the protein is made, impair transport of the protein to the cell surface, or cause the protein to be broken down too quickly. All of these mutations result in a decrease or absence of MRP2 activity at the cell membrane. As a result, the body's ability to release (excrete) bilirubin is impaired. A buildup of bilirubin causes jaundice in people with Dubin-Johnson

syndrome. The accumulation of other substances that usually get transported out of tissues by the MRP2 protein can cause additional signs and symptoms in people with Dubin-Johnson syndrome, but these features usually do not cause health problems.

Other Names for This Gene

- ATP-binding cassette, sub-family C (CFTR/MRP), member 2
- canalicular multispecific organic anion transporter
- CMOAT
- cMRP
- MRP2
- MRP2_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ABCC2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1244[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ABCC2%5BTI%5D%29+OR+%28MRP2%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ATP-BINDING CASSETTE, SUBFAMILY C, MEMBER 2; ABCC2 (https://omim.org/entry/601107)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1244)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ABCC2[gene])

References

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Genomic Location

The *ABCC2* gene is found on chromosome 10 (https://medlineplus.gov/genetics/chromosome/10/).

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