ABCG2 gene

ATP binding cassette subfamily G member 2 (Junior blood group)

Normal Function

The *ABCG2* gene belongs to a group of genes called the ATP-binding cassette family; genes in this family provide instructions for making proteins that transport molecules across cell membranes. In the intestines, the ABCG2 protein helps release (secrete) a substance called urate into the urine. Urate is a byproduct of certain normal biochemical reactions in the body. In the bloodstream it acts as an antioxidant, protecting cells from the damaging effects of unstable molecules called free radicals. Urate levels are regulated by the kidneys and, to a lesser extent, by the intestines.

The ABCG2 protein also transports certain drugs out of cells. For example, this protein clears some chemotherapy drugs from organs and tissues. Transport of these drugs allows them to have their intended effects and be eliminated from the body.

Health Conditions Related to Genetic Changes

Gout

Genetic changes in the *ABCG2* gene are associated with a condition called gout, which is a form of arthritis that causes painful joint inflammation.

ABCG2 gene changes associated with gout decrease the protein's ability to release urate. One variant replaces the protein building block (amino acid) glutamine with the amino acid lysine at position 141 in the protein (written as Gln141Lys or Q141K). This change reduces the protein's ability to secrete urate by half. Another variant creates a premature stop signal in the instructions for making the ABCG2 protein (written as Gln126Ter or Q126X), which results in no functional ABCG2 protein. Variants in the *ABCG2* gene reduce the removal of urate from the blood, which causes the blood level of urate to rise. The excess urate can accumulate in the body's joints in the form of crystals, triggering an inflammatory response from the immune system and leading to gout.

While changes in the *ABCG2* gene can alter urate levels in the body, they are not enough to cause gout by themselves. A combination of dietary, genetic, and other environmental factors play a part in determining the risk of developing this complex disorder.

Other Names for This Gene

- ABC15
- ABCP
- ATP-binding cassette transporter G2
- ATP-binding cassette, sub-family G (WHITE), member 2 (Junior blood group)
- BCRP
- BCRP1
- BMDP
- breast cancer resistance protein
- CD338
- CDw338
- EST157481
- mitoxantrone resistance-associated protein
- MRX
- multi drug resistance efflux transport ATP-binding cassette sub-family G (WHITE) member 2
- MXR
- MXR-1
- MXR1
- placenta specific MDR protein
- placenta-specific ATP-binding cassette transporter
- UAQTL1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ABCG2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9429[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ABCG2%5BTI%5D%29+OR+%28ATP+binding+cassette+subfamily+G+member+2%5BTI%5D%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+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ATP-BINDING CASSETTE, SUBFAMILY G, MEMBER 2; ABCG2 (https://omim.org/entry/603756)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/9429)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ABCG2[gene])

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Genomic Location

The *ABCG2* gene is found on chromosome 4 (https://medlineplus.gov/genetics/chromosome/4/).

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