Dubin-Johnson syndrome

Description

Dubin-Johnson syndrome is a condition characterized by jaundice, which is a yellowing of the skin and whites of the eyes. In most affected people jaundice appears during adolescence or early adulthood. Jaundice is typically the only feature of Dubin-Johnson syndrome, but some people can experience weakness, mild abdominal pain, nausea, or vomiting. In most people with Dubin-Johnson syndrome, certain deposits build up in the liver but do not seem to impair liver function. The deposits make the liver appear black when viewed with medical imaging.

Rarely, jaundice develops soon after birth in individuals with Dubin-Johnson syndrome. Affected infants typically also have enlarged livers (hepatomegaly) and a severely reduced ability to produce and release a digestive fluid called bile (cholestasis). As these children get older, their liver problems go away and they usually do not have any related health problems later in life.

Frequency

The prevalence of Dubin-Johnson syndrome is unknown. It appears to be most common in Iranian and Moroccan Jews living in Israel, with 1 in 1,300 individuals affected. Additionally, several people in the Japanese population have been diagnosed with Dubin-Johnson syndrome. This condition appears to be less common in other populations.

Causes

Dubin-Johnson syndrome is caused by changes in a gene known as *ABCC2*. The *ABCC2* gene provides instructions for making a protein that transports certain substances out of cells so they can be released (excreted) from the body. For example, this protein 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.

ABCC2 gene mutations result in the production of a protein with reduced or absent activity that cannot effectively transport substances out of cells. These mutations particularly affect moving bilirubin into bile. As a result, bilirubin accumulates in the body, causing a condition called hyperbilirubinemia. The buildup of bilirubin in the body

causes the yellowing of the skin and whites of the eyes in people with Dubin-Johnson syndrome. The black liver in affected individuals is due to a buildup of different substance normally transported out of the liver by the protein produced from the *ABCC2* gene.

Learn more about the gene associated with Dubin-Johnson syndrome

- ABCC2

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Black liver-jaundice syndrome
- Chronic idiopathic jaundice
- Chronic idiopathic jaundice with pigmented liver
- DJS
- Dubin-Sprinz syndrome
- Hyperbilirubinemia II
- Hyperbilirubinemia, Dubin-Johnson type
- Jaundice, chronic idiopathic

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Dubin-Johnson syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0022350/)

Genetic and Rare Diseases Information Center

- Dubin-Johnson syndrome (https://rarediseases.info.nih.gov/diseases/2793/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Dubin-Johnson syndrome%22)

Catalog of Genes and Diseases from OMIM

- DUBIN-JOHNSON SYNDROME; DJS (https://omim.org/entry/237500)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Dubin-Johnson+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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