North American Indian childhood cirrhosis

Description

North American Indian childhood cirrhosis is a rare liver disorder that occurs in children. The liver malfunction causes yellowing of the skin and whites of the eyes (jaundice) in affected infants. The disorder worsens with age, progressively damaging the liver and leading to chronic, irreversible liver disease (cirrhosis) in childhood or adolescence. Unless it is treated with liver transplantation, North American Indian childhood cirrhosis typically causes life-threatening complications including liver failure.

Frequency

North American Indian childhood cirrhosis has been found only in children of Ojibway-Cree descent in the Abitibi region of northwestern Quebec, Canada. At least 30 affected individuals from this population have been reported.

Causes

North American Indian childhood cirrhosis results from at least one known mutation in the *UTP4* gene. This gene provides instructions for making a protein called cirhin, whose precise function is unknown. Within cells, cirhin is located in a structure called the nucleolus, which is a small region inside the nucleus where ribosomal RNA (rRNA) is produced. A chemical cousin of DNA, rRNA is a molecule that helps assemble protein building blocks (amino acids) into functioning proteins. Researchers believe that cirhin may play a role in processing rRNA. Studies also suggest that cirhin may function by interacting with other proteins.

Cirhin is found in many different types of cells, so it is unclear why the effects of North American Indian childhood cirrhosis appear to be limited to the liver. Researchers are working to determine how a *UTP4* gene mutation causes the progressive liver damage characteristic of this disorder.

Learn more about the gene associated with North American Indian childhood cirrhosis

- UTP4

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

- NAIC

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Hereditary North American Indian childhood cirrhosis (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1858051/)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- NORTH AMERICAN INDIAN CHILDHOOD CIRRHOSIS; NAIC (https://omim.org/entry/604901)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28north+american+indian%5BTIAB%5D%29+AND+%28childhood%5BTIAB%5D%29+AND+%28cirrhosis%5BTIAB%5D%29%29+OR+%28CIRH1A%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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