



North American Indian childhood cirrhosis

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.

Inheritance Pattern

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

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
[/primer/testing/genetic-testing](#)
- Genetic Testing Registry: North American Indian childhood cirrhosis
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1858051/>

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22North+American+Indian+childhood+cirrhosis%22+OR+%22Liver+Cirrhosis%22>

Other Diagnosis and Management Resources

- Children's Organ Transplant Association
<https://cota.org/>

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Cirrhosis
<https://medlineplus.gov/ency/article/000255.htm>
- Encyclopedia: Jaundice - Yellow Skin
<https://medlineplus.gov/ency/article/000210.htm>
- Health Topic: Cirrhosis
<https://medlineplus.gov/cirrhosis.html>

Additional NIH Resources

- National Institute of Diabetes and Digestive and Kidney Diseases
<https://www.niddk.nih.gov/health-information/liver-disease/cirrhosis>

Educational Resources

- MalaCards: north American Indian childhood cirrhosis
https://www.malacards.org/card/north_american_indian_childhood_cirrhosis
- Merck Manual Home Edition for Patients and Caregivers
<https://www.merckmanuals.com/home/liver-and-gallbladder-disorders/fibrosis-and-cirrhosis-of-the-liver/cirrhosis-of-the-liver>
- Orphanet: Hereditary North American Indian childhood cirrhosis
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=168583

Patient Support and Advocacy Resources

- American Liver Foundation
<https://liverfoundation.org/for-patients/about-the-liver/diseases-of-the-liver/cirrhosis/>
- Canadian Liver Foundation
<https://www.liver.ca/>
- Children's Liver Association for Support Services (CLASS)
<https://www.classkids.org/what-we-do>
- Children's Organ Transplant Association
<https://cota.org/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?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>

Catalog of Genes and Diseases from OMIM

- NORTH AMERICAN INDIAN CHILDHOOD CIRRHOSIS
<http://omim.org/entry/604901>

Medical Genetics Database from MedGen

- North american indian childhood cirrhosis
<https://www.ncbi.nlm.nih.gov/medgen/387974>

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Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/condition/north-american-indian-childhood-cirrhosis>

Reviewed: March 2011

Published: June 11, 2019

Lister Hill National Center for Biomedical Communications

U.S. National Library of Medicine

National Institutes of Health

Department of Health & Human Services