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# Rapid whole genome sequencing identifies a novel homozygous NPC1 variant associated with Niemann-Pick Type C1 Disease in a 7 week old male with cholestasis

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## Abstract

Niemann-Pick Type C disease (NPC; OMIM #257220) is an inborn error of intracellular cholesterol trafficking. It is an autosomal recessive disorder caused predominantly by mutations in NPC1. While characterized as a progressive neurological disorder, it can also cause cholestasis and liver dysfunction due to intrahepatocyte lipid accumulation. We report a 7 week old who was admitted with neonatal cholestasis, who was diagnosed with a novel homozygous stop-gain variant in NPC1 by rapid whole genome sequencing (WGS). WGS results were obtained 16 days prior to return of the standard clinical genetic test results, and prompted initiation of targeted therapy.

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Abnormal cholesterol homeostasis
Clinodactyly of the 5th finger
Foam cells with lamellar inclusion bodies
Generalized neonatal hypotonia
Hepatosplenomegaly
Prolonged neonatal jaundice

Explore HPO Ontology

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