HCP / Caregiver(s): Dr. Brown

Brief Summary - Rare Diagnosis

Name	Bob Doe
DOB	11 May 2010
Medical record number	123445657
Location	General Hospital - Springfield
Date	16/02/2022

Evaluations

Major clinical findings	IUGR/growth delay (symmetrically small for age) Transient neonatal Transient neonatal hyperinsulinism /hypoglycaemia Idiopathic hepatic fibrosis Idiopathic intermittent metabolic acidosis Multiple ventricular septal defects (spontaneous closure) Ectodermal abnormalities sparse/unruly hair and fine eyebrows, numerous café-au-lait lesions
Development	Growth delay
Behavioral characteristics	Normal gross and fine motor skills development

What we knew before UDP assessment

Family History	healthy parents, Polish/English ancestry, non-consanguineous Unaffected sibling
Previous key evaluations	SNP microarray 1st July 2013 Gene panel 28th September 2016

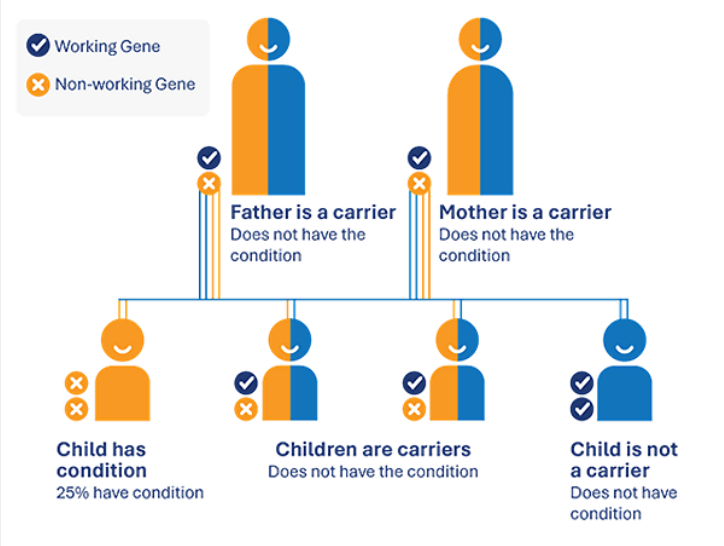
What we found

We have identified the cause of firstname's condition - their diagnosis is	Tricho-hepato-enteric syndrome
Pathogenic variants were identified	Pathogenic variants were identified (gene changes) were identified in the SKIV2L gene
Clinical features include	Tricho-hepato-enteric syndrome is autosomal recessive condition with varied clinical severity. Clinical features in this condition typically include problems with intestinal function characterized by chronic diarrhea/failure to gain weight, cirrhosis of the liver, wooly/brittle hair, a weakened immune system, cafe-au-lait spots and congenital heart defects.

Single Gene

Predicted Class/s	Genes	Chromosome	Nucleotide Change	Protein Change	Zygosity	Effect	Inheritance
Pathogenic	SKIV2L	6	c.904C>T c. 2662_2663delAG	.Gin302* p.Arg888Glyfs*12	homozygous	Stopgain Frameshift	Autosomal recessive

Genetic information

Autosomal recessive	 Testing of both parents has shown that each carries a copy of the SINGLE GENE change and have passed this down to BOB DOE. This inheritance pattern is called autosomal recessive. Autosomal recessive inheritance (genetics.edu.au)

Conclusions

Ongoing Management

Based on BOB DOE's assessment, we have the following recommendations for ongoing management:
Adult genetic clinic- Bob Doe may wish to discuss recurrence risks of his condition if/when he is planning a family.

Resource and support links

Home - Rare Is Everywhere
MyTime - Supporting parents and carers of children with disability
Raising Children with Disability | Raising Children Network
Children with Multiple Disabilities, Rare Conditions or are Undiagnosed Navigate Life Texas
Extraordinary! A Book for Children with Rare Diseases
Rareminds | Mental Health Services for the Rare Disease Community