

Patient diagnosed with variegate porphyria after detecting a bi-allelic mutation in the PPOX gene

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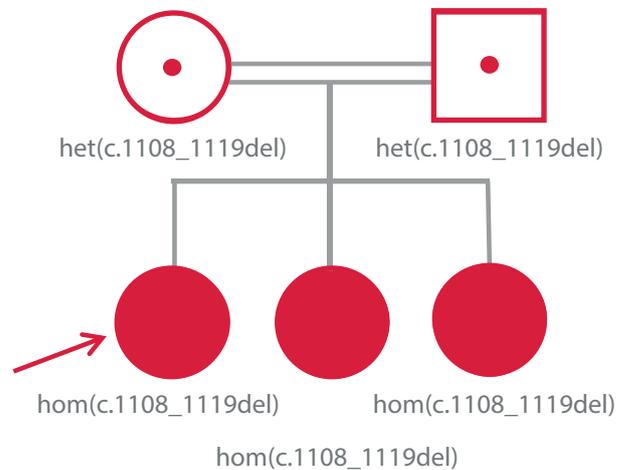
Geographic region

Libya, North Africa

Clinical information

The case in point is a newborn baby girl. She had symptoms of mild birth asphyxia, nystagmus, skin discoloration and seizures. Leukodystrophy and ichthyosis later manifested from the age of 1 year.

Her parents were reported consanguineous and healthy, but had two other affected daughters who presented with similar symptoms.



Diagnostic procedure

Patient symptoms were highly heterogeneous and a large number of genes could have been associated with each of the symptoms. For this reason, CentoXome[®] Trio Advanced was recommended for this patient.

End-to-end bioinformatics analysis and variant prioritization found a homozygous variant in the **PPOX gene** of the patient; c.1108_1119del(p.Gly370_Trp373del). Her parents were found to be heterozygous carriers.

This variant is an in frame deletion that results in the loss of 4 amino acid residues. This is the first time we have detected this pathogenic variant based on CENTOGENE's mutation database CentoMD[®].

Bi-allelic mutations in PPOX lead to variegate porphyria. Presentations consist of nystagmus, developmental delay and ataxia, combined with a photosensitive eruption. However, there are many atypical presentations that can make a clinical diagnosis difficult. The variant was also identified in the patient's affected sisters.

Variegate porphyria is initially managed in an intensive care unit (ICU). The presence of seizures, motor neuropathy, and pain is controlled using anti-convulsive therapy, narcotic analgesics, and phenothiazines for nausea, agitation, and hallucinations.

CentoXome[®] Trio Advanced analysis helped identify the cause of the disease, select the most optimal treatment, and further help with genetic counselling for the family.

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