

Patient diagnosed with Hyaline fibromatosis syndrome after detecting the c.134T>C variant in the ANTXR2 gene

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Geographic region

Kuwait, Middle East

Clinical information

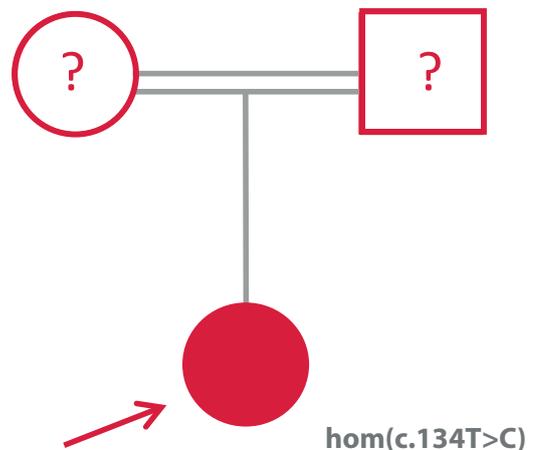
2 year old girl with mild dysmorphic facial features, distal arthrogryposis, hypotonia with positive deep tendon reflexes.

The parents are cousins with a history of repeated neonatal and infant deaths with similar features and polycystic kidneys. No parental samples available.

Diagnostic procedure

Patient symptoms were highly heterogeneous and a large number of genes could have been associated with each of the symptoms. The family history with polycystic kidney disease additionally increased the number of possible candidate genes. As parents did not provide samples, CentoXome® Solo Advanced was immediately recommended for this patient.

End-to-end bioinformatics analysis and variant prioritization, identified a homozygous pathogenic variant in the **ANTXR2 gene** of the patient; c.134T>C that results in amino-acid change p.Leu45Pro and subsequent change of protein function. This pathogenic variant has previously been described as disease-causing for infantile hyaline fibromatosis syndrome (HGMD Professional 2015.3 - PMID: 19191226, 25458638). The variant does not explain the family history of polycystic kidney disease.



Pathogenic biallelic variants in the **ANTXR2 gene** cause Hyaline fibromatosis syndrome.

Presentations are characterized by abnormal growth of hyalinized fibrous tissue usually affecting subcutaneous regions on the head, neck and limbs, as well as motor disability, thickened skin, and progressive joint contractures.

Early diagnosis and treatment for joint contractures, nutrition problems and pain (anti-inflammatory drugs) could improve the quality of a patient's life.

CentoXome® Solo 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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