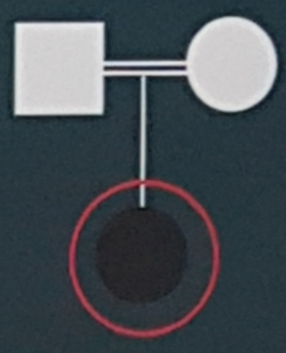
JALILI SYNDROME

A Novel Mutation in CNNM4

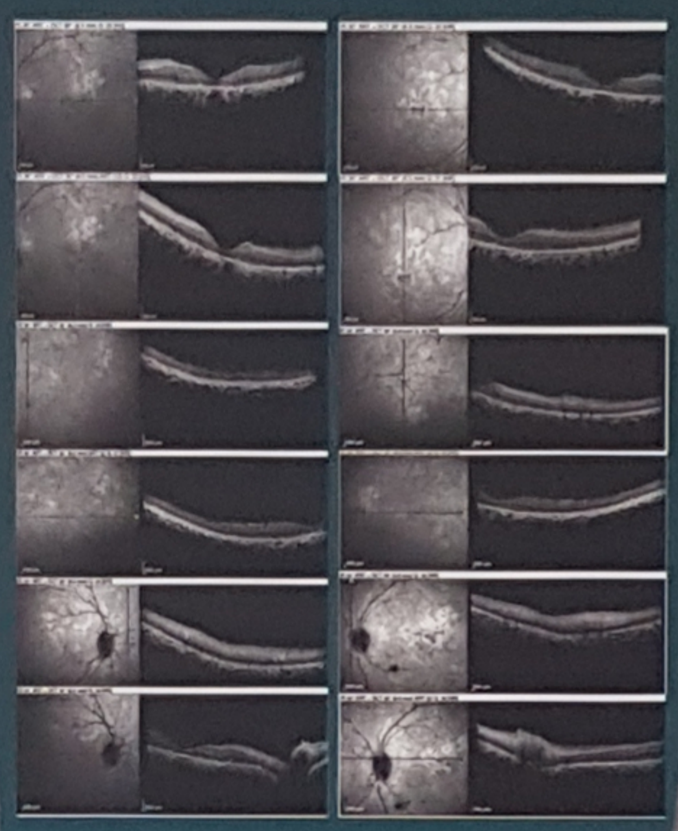
c.1936_1934dup,p.(Ser649Argfs*88)

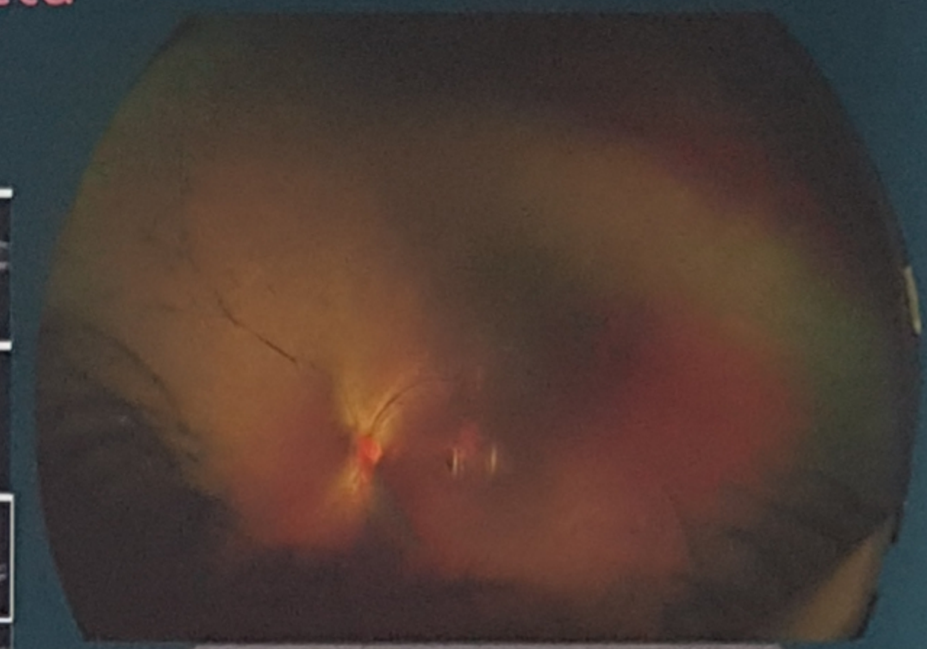
Recessive Cone-Rod Dystrophy with Amelogenesis Imperfecta

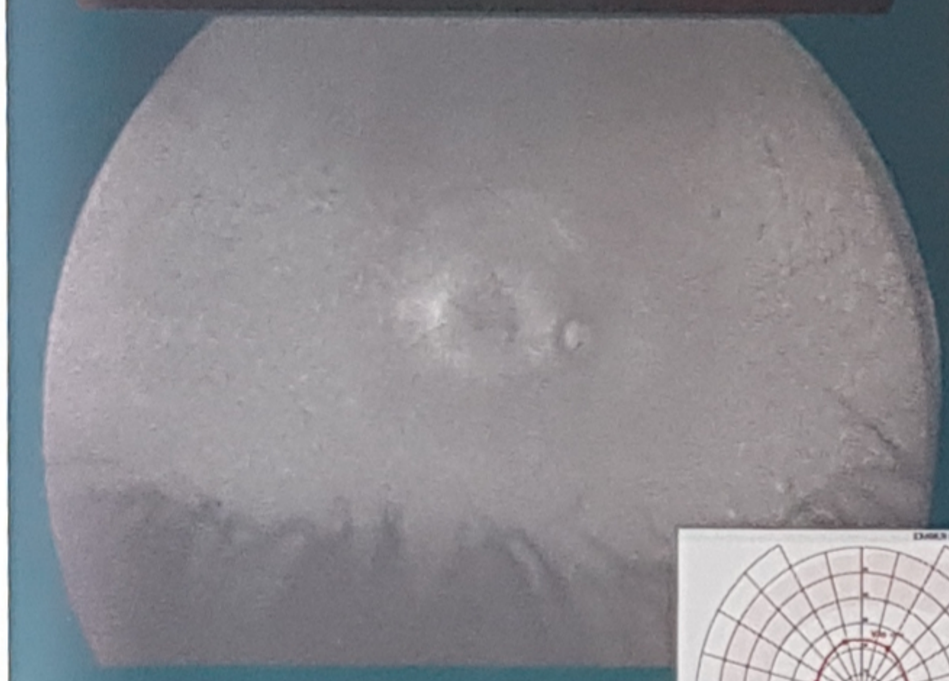
Indeed, the Jalili syndrome is a rare multisystem disorder with the most prominent features consisting of cone-rod dystrophy and amelogenesis imperfecta.

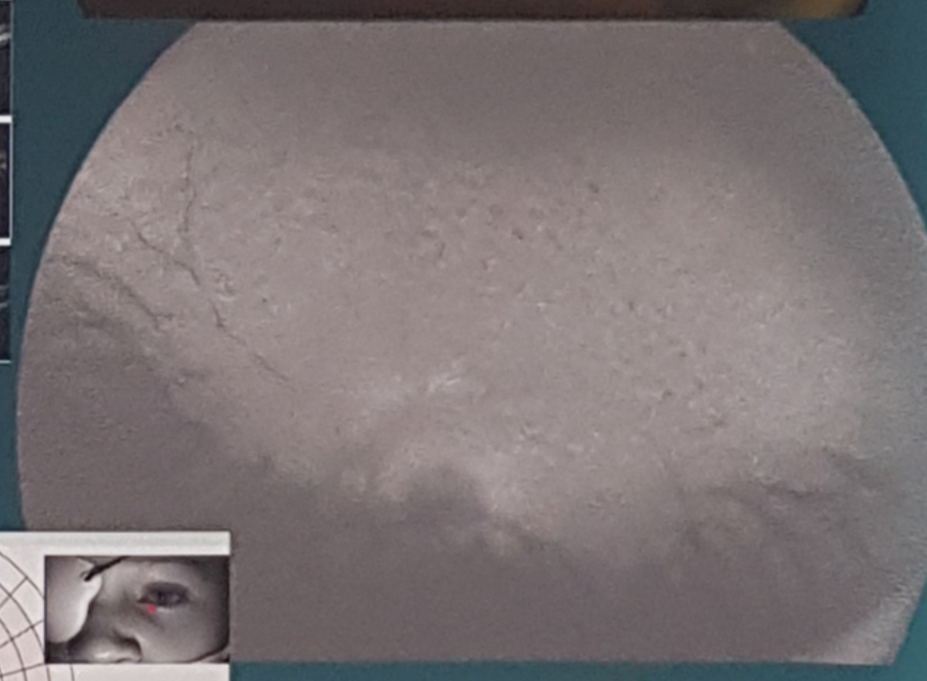


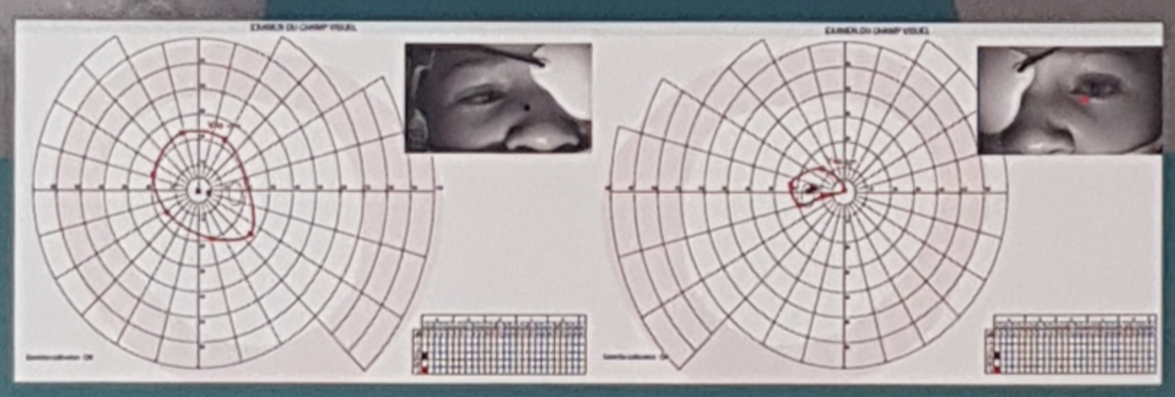
Consanguinity between the parents with a consanguinity coeficient close to **1/32** which means that the parents are **little cousins.**











NGS study of a **230 genes Inherited Retinal Distrophy genes**

Novel mutation

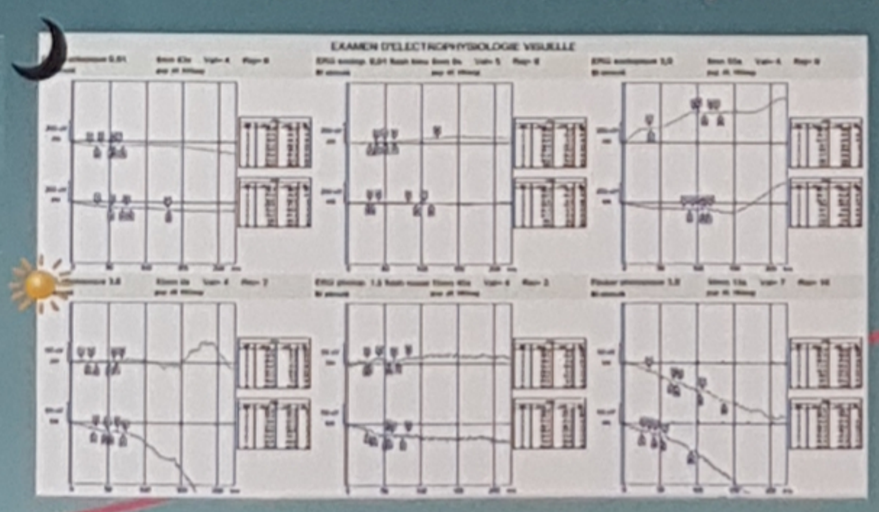
This young 11 years old girl from Chad is suffering from an early and serious recessive cone-rod dystrophy assossiated with amelogenesis Imperfecta.

→ Presence of the variation **c.1936_1934dup,p.(Ser649Argfs*88)** in homozygous state located in **exon 5 of CNNM4 gene**

→ The duplication of 8 nucléotides is responsible for a frameshift inducing a truncated protein or the absence of protein synthesis by NMD (nonsense mediated decay) mechanism.

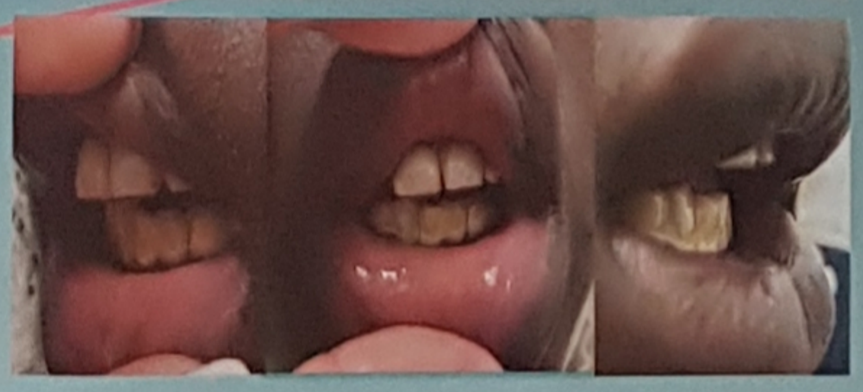
→ This variant is classified as pathogenic according to the ACMG 2015 criteria.

RE		LE
Near blindness (0.016)	VA logMAR	Near Blindness (0.016)
+2.00 (-1.25) 49°	AR	+1.50 (-1.25) 135°



Congenital Nystagmus

Night Blindness



Dental Lesion

Walking delay around 2 years old

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