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Jalili Syndrome: The Rare retinopathy with Amelogenesis Imperfecta, Clinical Insights, novel variant and genetic Perspectives in four patients

Background

Jalili syndrome is a rare progressive retinal-ciliopathy disorder displayed with cod-rod dystrophy and amelogenesis imperfecta (AI) caused by CNNM4 gene. The onset of visual impairment starts in infancy or early childhood.

Purpose

We report on four Egyptian patients affected by Jalili syndrome, highlighting the clinical presentation and implications of the disorder. Additionally, we expand the molecular characterization of the CNNM4 gene by reporting a novel pathogenic variant, contributing to the understanding of its role in this rare condition.

Methodology

Four patients (P1-P4) from two unrelated families complained of eye problems and amelogenesis imperfecta were included in the study. The patients were subjected to detailed clinical and dental examination. WES was used for the molecular identification of the causative gene.

Results

Exome sequencing identified two different variants in CNNM4 gene of Jalili syndrome, one novel homozygous missense variant c.1423G>A (p.Val475Met) (P1) and a homozygous deletion of the entire exon 1 of the gene c.1-?_1403þ?del (P2-P4).

Conclusion

This study is focusing on Jalili syndrome, highlighting and expanding its clinical and genetic phenotypes. Moreover, the identification of a novel CNNM4 mutation enriches the genomic landscape of the syndrome, helping to guide future genetic testing and counseling efforts. Amelogenesis imperfecta is a hallmark evidence in the diagnosis of rare disorders. Finally, this research emphasizes the essential role of specialists, particularly dental geneticists, in the diagnosis and management of rare disorders.

Keywords: Jalili syndrome, CNNM4, amelogenesis imperfecta, retinopathy, cone-rod dystrophy

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Biography

Professor of Orodental Genetics department, National Research Centre. Her expertise lasts for 20 years dealing with dental genetics, particularly inherited tooth structure defects, their associated syndromes, and underlying molecular mechanisms. She is the principal investigators of many national projects related to oral abnormalities in rare disorders. She is a member in many national and international societies serving the rare inherited disorders. She has different international publications reporting various rare orodental anomalies in syndromes.

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