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Abstract Title:	**Jalili Syndrome - Cone-Rod Dystrophy (CRD) and Amelogenesis Imperfecta (AI); Six Families and Consistent Linkage to 2q11**
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Purpose: We ascertained six families with autosomal recessive AI, a defect of enamel formation, and CRD leading to loss of central vision and blindness. We propose to name this condition Jalili syndrome. This study aims to describe the phenotype more completely and determine whether this is a single condition.
Methods: Affected individuals were subject to direct ophthalmoscopy. Deciduous teeth were examined by electron microscopy, energy dispersive X-ray analysis and microradiography. Microsatellite genotypes were obtained by PCR from genomic DNA and size fractionation on an ABI sequencer. SNP genotypes were obtained using the Affymetrix 50k SNP chips.
Results: Primary and secondary teeth show a generalised mixed hypomaturation/hypoplastic form of AI. Ultrastructural analyses of deciduous teeth from one individual confirm poor enamel structure and hypomineralisation. Impaired central vision is evident in all cases in infancy and progresses with loss of colour vision and photophobia. Typically patients have poor vision from the first decade of life and are blind before the age of 20. Genetic analyses of four families confirm linkage to 2q11, a fifth is consistent with linkage while a sixth is too small to test.
Conclusions: With only two published families it remained possible that this condition resulted from the coincidence of two closely linked mutations, and that this could involve different genes in each family. However the observation of six such families, with confirmation of 2q11 linkage in four, confirms the existence of a single, genetically homogeneous syndrome and points to the intriguing possibility of a link between tooth and eye development.

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