

Open Peer Review on Qeios

## **EEM** syndrome

**INSERM** 

## Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>EEM</u> <u>syndrome</u>. ORPHA:1897

EEM syndrome is characterised by the association of ectodermal dysplasia, ectrodactyly, and macular dystrophy. So far, it has been described in individuals from seven families. Hypotrichosis, dental anomalies and absent eyebrows have also been reported. EMM syndrome appears to be transmitted as an autosomal recessive trait and may be caused by mutations in the cadherin-3 gene (CH3, 16q22.1).

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