

Open Peer Review on Qeios

## Ichthyosis-oral and digital anomalies syndrome

**INSFRM** 

## Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Ichthyosis-oral and digital anomalies syndrome</u>. ORPHA:2272

Ichthyosis-oral and digital anomalies syndrome is characterised by ichthyosis, unusual facies (small mouth with a thin upper lip and lower lip with a midline groove) and digital anomalies (tapered fingers with a lack of distal flexion creases and wide spacing between the second and third fingers). It has been described in two sibs born to first cousin parents. Transmission appears to be autosomal recessive.

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