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White forelock with malformations

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. White forelock with malformations. ORPHA:2475

White forelock with malformations is a multiple congenital anomalies syndrome characterized by poliosis, distinct facial features (epicanthal folds, hypertelorism, posterior rotation of ears, prominent philtrum, high-arched palate) and congenital anomalies/malformations of the eye (blue sclera), cardiopulmonary (atrial septal defect, prominent thoracic and abdominal veins), and skeletal (clinodactyly, syndactyly of the fingers and 2nd and 3rd toes) systems. There have been no further descriptions in the literature since 1980.

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