

Opis choroby *

Definicja

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.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

2475

Kod OMIM

277740

Kod ICD10

Q87.8

Kod ICD11

-

*Źródło

orphanet