Opis choroby *

Definicja

A rare, genetic, neurodevelopmental disorder characterized by global developmental delay, congenital heart defects, generalized hypertrichosis and dysmorphic facial features, most commonly triangular face, thick arched eyebrows, widely spaced eyes, posteriorly rotated low set ears, depressed nasal bridge, broad nasal root and tip, and pointed chin.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
562569

Kod OMIM
618316

Kod ICD10
Q87.8

Kod ICD11
-

---

*Źródło

orphanet