

## 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	Kod OMIM	Kod ICD10
562569	618316	Q87.8
<b>Kod ICD11</b>		
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### \*Źródło

orphanet