Opis choroby *

Definicja

A rare developmental defect during embryogenesis, resulting from partial or total deletion of the short arm of chromosome 5, classically characterized by a high-pitched, monotone, cat-like cry (cri du chat) present since birth, associated with varying degrees of intellectual disability, developmental delay, microcephaly, and facial dysmorphism.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Cri du chat syndrome
Delecja 5p
Zespół kociego płaczu
Deletion 5p

Kod ORPHA
281

Kod OMIM
123450

Kod ICD10
Q93.4

Kod ICD11
LD44.51

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*Źródło

orphanet