

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

Kod ORPHA	Kod OMIM	Kod ICD10
281	123450	Q93.4

Kod ICD11
LD44.51

*Źródło

orphanet