Opis choroby *

Definicja

A very rare multiple congenital anomaly syndrome characterized by the presence of anophthalmia or severe microphthalmia, cleft lip/palate, facial cleft and sacral neural tube defects, along with various additional anomalies including congenital glaucoma, iris coloboma, primary hyperplastic vitreous, hypertelorism, low-set ears, clinodactyly, choanal atresia/stenosis, dysgenesis of sacrum, tethering of spinal cord, syringomyelia, hypoplasia of corpus callosum, cerebral ventriculomegaly and endocrine abnormalities. An autosomal recessive inheritance has been suggested.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Fryns microphthalmia syndrome
Mikroftalmia z rozszczepem twarzy
Zespół mikroftalmii Frynsa
Microphthalmia with facial clefting

Kod ORPHA
1104

Kod OMIM
600776

Kod ICD10
Q87.8

Kod ICD11
LD21.0

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*Źródło

orphanet