

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

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| Klasyfikacja | Synonimy |
| Zespół wad wrodzonych Fryns | Fryns microphthalmia syndrome Mikroftalmia z rozszczepem twarzy Zespół mikroftalmii Frynsa Microphthalmia with facial clefting |

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| Kod ORPHA 1104 | Kod OMIM 600776 | Kod ICD10 Q87.8 |
| Kod ICD11 LD21.0 | | |

*Źródło

orphanel