

## 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

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych Fryns	Fryns microphthalmia syndrome Mikroftalmia z rozszczepem twarzy Zespół mikroftalmii Frynsa Microphthalmia with facial clefting

<b>Kod ORPHA</b> 1104	<b>Kod OMIM</b> 600776	<b>Kod ICD10</b> Q87.8
<b>Kod ICD11</b> LD21.0		

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### \*Źródło

orphonet