

Opis choroby *

Definicja

Microphthalmia with brain and digit anomalies is characterised by anophthalmia or microphthalmia, retinal dystrophy, and/or myopia, associated in some cases with cerebral anomalies. It has been described in two families. Polydactyly may also be present. Linkage analysis allowed identification of mutations in the <I> BMP4</I> gene, which has already been shown to play a role in eye development.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Bakrania-Ragge syndrome MCOPS6 Mikroftalmia syndromiczna typu 6 Zespół Bakrania i Ragge MCOPS6 Syndromic microphthalmia type 6

Kod ORPHA	Kod OMIM	Kod ICD10
139471	607932	Q11.2

Kod ICD11

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*Źródło

orphanet