

Opis choroby *

Definicja

Microphthalmia-ankyloblepharon-intellectual disability syndrome is characterized by microphthalmia, ankyloblepharon and intellectual deficit. It has been described in seven male patients from two generations of a Northern Ireland family. The causative gene is localized to the Xq27-q28 region. The syndrome is transmitted as an X-linked recessive trait.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	MCOPS4
	MCOPS4
	Syndromiczne małowocze typu 4
	Syndromic microphthalmia type 4

Kod ORPHA	Kod OMIM	Kod ICD10
85275	-	Q11.2

Kod ICD11
LD21.0

*Źródło

orphanet