Opis choroby *

Definicja

A rare multiple congenital anomalies syndrome, reported in the offsprings of a consanguineous couple and characterized by multiple congenital skeletal (dolichocephaly, skull asymmetry, camptodactyly, clubfoot), muscular (muscle hypoplasia), ocular (anophthalmia, buphthalmos, retinal detachment, aniridia (see this term)) and cardiac (prolapse of tricuspid valves, mitral and tricuspid insufficiency) abnormalities. An autosomal recessive inheritance with variable expressivity was suspected. There have been no further descriptions in the literature since 1992.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Cassia Stocco dos Santos syndrome
Zespół Cassia Stocco dos Santos

Kod ORPHA
1101

Kod OMIM
-

Kod ICD10
Q87.8

Kod ICD11
-

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*Źródło

orphanet