

## 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

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych	Cassia Stocco dos Santos syndrome Zespół Cassia Stocco dos Santos

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
1101	-	Q87.8

**Kod ICD11**  
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### \*Źródło

orphanet