Opis choroby *

Definicja

A rare disorder of sex development characterized by primary amenorrhea and ambiguous external genitalia (enlarged clitoris with marked fusion of the labioscrotal folds) in association with skeletal anomalies (such as hypoplasia of the mandibular condyles and the maxilla, and ulnar dislocation of the radial heads), in the presence of a 46,XX karyotype and regular ovaries, fallopian tubes, and uterus. There have been no further descriptions in the literature since 1972.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Pseudohermafrodytyzm żeński - wady szkieletu
	46,XX disorder of sex development-skeletal anomalies syndrome

Kod ORPHA	**Kod OMIM**	**Kod ICD10**
2975	264270	Q56.2

Kod ICD11
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*Źródło

orphanet