Opis choroby *

Definicja

A rare, genetic multiple congenital anomalies syndrome characterized by genitourinary malformations (hydrometrocolpos in females and in males, glanular hypospadias and prominent scrotal raphe) , postaxial polydactyly that may affect only one or several limbs, and to a lesser extent cardiac defects. Hydrometrocolpos is due to either a congenital obstruction, imperforate hymen or vaginal atressia, and causes a palpable mass and possibly hydronephrosis. Other anomalies occasionally reported include choanal atresia, pituitary dysplasia, esophageal atresia and distal tracheoesophageal fistula, Hirschsprung disease, vertebral anomalies, and hydrops fetalis. The disorder is allelic with Bardet-Biedl, and as some phenotypic overlap has been observed, patients should be reevaluated in later childhood for retinistis pigmentosas and other signs of Bardet-Biedl syndrome.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Hydrometrocolpos-postaxial polydactyly syndrome
Hydrometrocolpos - polidaktylia zaosiowa
Zespół Kaufmana i McKusicka
Kaufman-Mckusick syndrome

Kod ORPHA
2473

Kod OMIM
236700

Kod ICD10
Q87.8

Kod ICD11
9B70

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*Źródło

orphanet