

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 atresia, 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

Synonimy

Zespół wad wrodzonych 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

*Źródło

orphanet