

## 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 retinitis pigmentosa and other signs of Bardet-Biedl syndrome.

### Dane

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych	Hydrometrocolpos-postaxial polydactyly syndrome Hydrometrocolpos - polidaktylia zaosiowa Zespół Kaufmana i McKusicka Kaufman-Mckusick syndrome

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
2473	236700	Q87.8

**Kod ICD11**  
9B70

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### \*Źródło

orphanet