

## Opis choroby \*

### Definicja

Postaxial polydactyly-anterior pituitary anomalies-facial dysmorphism syndrome is a rare, genetic developmental defect during embryogenesis disorder characterized primarily by congenital hypopituitarism and/or postaxial polydactyly. It can be associated with short stature, delayed bone age, hypogonadotropic hypogonadism, and/or midline facial defects (e.g. hypotelorism, mild midface hypoplasia, flat nasal bridge, and cleft lip and/or palate). Hypoplastic anterior pituitary and ectopic posterior pituitary lobe are frequent findings on MRI examination.

### Dane

|                       |  |
|-----------------------|--|
| <b>Klasyfikacja</b>   | <b>Synonimy</b>                                |
| Zespół wad wrodzonych | Culler-Jones syndrome<br>Culler-Jones syndrome |

|                  |                 |                  |
|------------------|-----------------|------------------|
| <b>Kod ORPHA</b> | <b>Kod OMIM</b> | <b>Kod ICD10</b> |
| 420584           | 615849          | Q87.8            |

### Kod ICD11

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### \*Źródło

orphanet