DISEASE:
Postaxial polydactyly-anterior pituitary anomalies-facial dysmorphism syndrome

NAME:	Postaxial polydactyly-anterior pituitary anomalies-facial dysmorphism syndrome
DESCRIPTION:	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.
ORPHACODE:	420584
SYNONYMS:	Culler-Jones syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	GLI2
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