DISEASE:
IMAGe syndrome

NAME:	IMAGe syndrome
DESCRIPTION:	A rare genetic disease characterized by intrauterine growth restriction, metaphyseal dysplasia, congenital adrenal hypoplasia, and genital anomalies (such as cryptorchidism, posterior hypospadias, and micropenis). Patients may present shortly after birth with severe adrenal insufficiency. Additional manifestations include postnatal growth failure and delayed bone age, mild developmental delay, macrocephaly, mild facial dysmorphism (with frontal bossing, wide nasal bridge, and small, low-set ears), epiphyseal dysplasia, and hypercalcemia/hypercalciuria, among others.
ORPHACODE:	85173
SYNONYMS:	Intrauterine growth retardation-metaphyseal dysplasia-adrenal hypoplasia congenita-genital anomalies syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	CDKN1C POLE
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- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

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