DISEASE:
Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome

NAME:	Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome
DESCRIPTION:	Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome is a rare, genetic, syndromic intellectual disability syndrome characterized by mild to moderate intellectual disability, developmental delay (with speech and language development more severely affected) and facial dysmorphism which typically includes full, arched eyebrows, hypertelorism, down-slanting palpebral fissures, long eyelashes, ptosis, low-set, simple ears, bulbous nasal tip, flat philtrum, wide mouth with downturned corners and thin upper lip and diastema of the teeth. Association with infantile hypotonia, seizures, cryptorchidism in males and congenital abnormalities, including cardiac, cerebral or ocular defects, may be observed.
ORPHACODE:	329224
SYNONYMS:	PACS1-related syndrome Schuurs-Hoeijmakers syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PACS1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Kabuki syndrome (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- phosphofurin acidic cluster sorting protein 1

Related Gene Panels

- Kabuki (7 genes) - IPG