DISEASE:
Combined pituitary hormone deficiencies, genetic forms

NAME:	Combined pituitary hormone deficiencies, genetic forms
DESCRIPTION:	Congenital hypopituitarism is characterized by multiple pituitary hormone deficiency, including somatotroph, thyrotroph, lactotroph, corticotroph or gonadotroph deficiencies, due to mutations of pituitary transcription factors involved in pituitary ontogenesis. Congenital hypopituitarism is rare compared with the high incidence of hypopituitarism induced by pituitary adenomas, transsphenoidal surgery or radiotherapy.
ORPHACODE:	95494
SYNONYMS:	Familial congenital hypopituitarism Multiple pituitary hormone deficiencies, genetic forms
XREF(S):	Orphanet OMIM OMIM OMIM ICD-10

ANALYTE(S):	FOXA2 POU1F1 PROP1 HESX1 OTX2 LHX4 GLI2
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RELATED CONTENT

Related Genetic Tests

- Brain malformations (gene panel)
- Combined pituitary hormone deficiency 1 (CPHD - POU1F1 gene)
- Combined pituitary hormone deficiency 2 (CPHD - PROP1 gene)
- Hypogonadotropic hypogonadism (33 genes)
- Microphthalmia, syndromic 5; Retinal dystrophy, early-onset, and pituitary dysfunction
- Short Stature (gene panel)

Related Laboratories

- Centre de Génétique Humaine - Erasme ULB
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- forkhead box A2
- GLI family zinc finger 2
- HESX homeobox 1
- LIM homeobox 4
- orthodenticle homeobox 2
- POU class 1 homeobox 1
- PROP paired-like homeobox 1

Related Gene Panels

- Hypogonadotropic Hypogonadism/Kallmann (61 genes) - ULG
- Hypogonadotropic hypogonadism (33 genes) - VUB
- Short Stature (46 genes) - IPG

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