DISEASE:
CHARGE syndrome

NAME:	CHARGE syndrome
DESCRIPTION:	CHARGE syndrome is a multiple congenital anomaly syndrome characterized by the variable combination of multiple anomalies, mainly Coloboma; Choanal atresia/stenosis; Cranial nerve dysfunction; Characteristic ear anomalies (known as the major 4 C's).
ORPHACODE:	138
SYNONYMS:	CHARGE association Coloboma-heart defects-atresia choanae-retardation of growth and development-genitourinary problems-ear abnormalities syndrome Hall-Hittner syndrome
XREF(S):	Orphanet OMIM MedDRA ICD-10 MeSH
ANALYTE(S):	SEMA3E CHD7
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RELATED CONTENT

Related Genetic Tests

- CHARGE syndrome
- Hypogonadotropic hypogonadism (33 genes)
- Renal or urinary tract malformation (CAKUT) (gene panel)

Related Laboratories

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

Related Analytes

- chromodomain helicase DNA binding protein 7
- semaphorin 3E

Related Gene Panels

- CHARGE (2 genes) - UZA
- Cakut (congenital anomalies of the kidney and urinary tract-1) (69 genes) - IPG
- Hypogonadotropic Hypogonadism/Kallmann (61 genes) - ULG
- Hypogonadotropic hypogonadism (33 genes) - VUB