DISEASE:
Hirschsprung disease

NAME:	Hirschsprung disease
DESCRIPTION:	A rare congenital intestinal motility disorder that is characterized by signs of intestinal obstruction due to the presence of an aganglionic segment of variable extent in the terminal part of the colon.
ORPHACODE:	388
SYNONYMS:	Aganglionic megacolon Colonic aganglionosis Congenital intestinal aganglionosis HSCR

XREF(S):	Orphanet MeSH OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM MedDRA ICD-10
ANALYTE(S):	ATP7A ABCD1 ERBB2 ERBB3 SMO RET ECE1 EDN3 EDNRB GDNF NRTN SEMA3C SEMA3D SREBF1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/392

RELATED CONTENT

Related Genetic Tests

- Hirschsprung disease
- Hirschsprung disease
- Hirschsprung disease
- Multiple Endocrine Neoplasia type 2A and 2B / Familial medullary thyroid carcinoma

Related Laboratories

- Centre de Génétique Médicale UCL
- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- ATP binding cassette subfamily D member 1
- ATPase copper transporting alpha
- endothelin converting enzyme 1
- endothelin 3
- endothelin receptor type B
- erb-b2 receptor tyrosine kinase 2
- erb-b2 receptor tyrosine kinase 3
- glial cell derived neurotrophic factor
- neurturin
- ret proto-oncogene

- semaphorin 3C
- semaphorin 3D
- smoothened, frizzled class receptor
- sterol regulatory element binding transcription factor 1

Related Gene Panels

- Hirschsprung disease - Ugent
