

DISEASE:
Chédiak-Higashi syndrome

NAME:	Chédiak-Higashi syndrome
DESCRIPTION:	Chédiak-Higashi syndrome (CHS) is a rare severe genetic disorder generally characterized by partial oculocutaneous albinism (OCA, see this term), severe immunodeficiency, mild bleeding, neurological dysfunction and lymphoproliferative disorder. A classic, early-onset form and an attenuated, later-onset form (Atypical CHS; see this term) have been described.
ORPHACODE:	167
SYNONYMS:	Chédiak-Higashi disease Chédiak-Higashi-Steinbrink syndrome
XREF(S):	Orphanet MeSH OMIM MedDRA ICD-10
ANALYTE(S):	LYST
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: <http://gentest.healthdata.be/disease/178>

RELATED CONTENT

Related Genetic Tests

- [Trombosis - Hemostasis \(gene panel\)](#)

Related Laboratories

- [Centrum Menselijke Erfelijkheid - KUL](#)

Related Analytes

- [lysosomal trafficking regulator](#)

Related Gene Panels

- [Trombosis - Hemostasis \(107 genes\) - KUL](#)

Source URL: <http://gentest.healthdata.be/disease/178>