

DISEASE:
Hereditary isolated aplastic anemia

NAME:	Hereditary isolated aplastic anemia
DESCRIPTION:	Hereditary isolated aplastic anemia is a rare, genetic, constitutional aplastic anemia disorder characterized by severe peripheral blood pancytopenia and bone marrow hypoplasia in multiple individuals of a family, in the absence of any somatic symptoms. Abnormal bleeding, as well as erythrocyte macrocytosis, is reported and patients usually become transfusion-dependent.
ORPHACODE:	397692
XREF(S):	<u>Orphanet</u> <u>ICD-10</u> <u>OMIM</u>
ANALYTE(S):	<u>THPO</u> <u>MPL</u> <u>ACD</u>
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: <http://gentest.healthdata.be/disease/2376>

RELATED CONTENT

Related Genetic Tests

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

Related Laboratories

- [Centrum Menselijke Erfelijkheid - KUL](#)

Related Analytes

- [ACD shelterin complex subunit and telomerase recruitment factor](#)
- [MPL proto-oncogene, thrombopoietin receptor](#)
- [thrombopoietin](#)

Related Gene Panels

- [Dyskeratosis Congenita \(18 genes\) - KUL](#)
- [Trombosis - Hemostasis \(107 genes\) - KUL](#)

Source URL: <http://gentest.healthdata.be/disease/2376>