DISEASE:
Inherited acute myeloid leukemia

NAME:	Inherited acute myeloid leukemia
DESCRIPTION:	Inherited acute myeloid leukemia (AML) is a rare, malignant hematopologic disease characterized by clonal proliferation of myeloid blasts, primarily involving the bone marrow, in association with congenital disorders (e.g. Fanconi anemia, dyskeratosis congenita, Bloom syndrome, Down syndrome, congenital neutropenia, neurofibromatosis, etc.) and genetic defects predisposing to AML. Patients present with signs and symptoms related to ineffective hematopoesis (fatigue, bleeding and bruising, recurrent infections, bone pain) and/or extramedullary site involvement (gingivitis, splenomegaly, etc.). Depending on the underlying genetic defect, there may be additional cancer risks and other health problems present.
ORPHACODE:	319465
SYNONYMS:	Familial AML Inherited AML Pure familial AML Pure familial acute myeloid leukemia
XREF(S):	Orphanet ICD-10 OMIM

ANALYTE(S):	CEBPA TGM6 ERCC6L2 GATA2
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest-acc.healthdata.be/disease/1752

RELATED CONTENT

Related Genetic Tests

- Myeloid neoplasms with germline predisposition (Hereditary MDS/Acute Leukemia) (gene panel)
- Myeloid/lymphoid neoplasms with germline predisposition (gene panel)
- « Inherited bone marrow failures syndromes » with or without organ dysfunction

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman

Related Analytes

- CCAAT enhancer binding protein alpha
- ERCC excision repair 6 like 2
- GATA binding protein 2
- transglutaminase 6

Related Gene Panels

- Hematologic Familiar Forms - ULG