

DISEASE:**Acute myeloid leukemia with CEBPA somatic mutations**

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| NAME: | Acute myeloid leukemia with CEBPA somatic mutations |
| DESCRIPTION: | A subtype of acute myeloid leukemia with recurrent genetic abnormalities, characterized by clonal proliferation of myeloid blasts harboring somatic mutations of the CEBPA gene in the bone marrow, blood and, rarely, other tissues. It can present with anemia, thrombocytopenia, and other nonspecific symptoms related to ineffective hematopoiesis (fatigue, bleeding and bruising, recurrent infections, bone pain) and/or extramedullary site involvement (gingivitis, splenomegaly). |
| ORPHACODE: | 319480 |
| SYNONYMS: | AML with CEBPA somatic mutations |
| XREF(S): | Orphanet OMIM ICD-10 |
| ANALYTE(S): | CEBPA |
| CREATED: | 13 May 2019 - 01:02 |
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Source URL: <http://gentest.healthdata.be/disease/1750>