ALBERTA PRECISION LABORATORIES
Leaders in Laboratory Medicine

Familial Acute Myeloid Leukemia (AML) Panel: Information for Ordering Providers

Most cases of acute myeloid leukemia (AML) are sporadic; however, families with an inherited predisposition to hematological cancers have been reported.

Indications for Testing

Patients with AML and a family history of AML are eligible for testing. In addition, individuals who have developed AML at an early age (<50 years old) are eligible for testing.

Limitations

In individuals with a hematological malignancy, genetic testing may reveal a variant that is acquired rather than inherited. Confirmatory testing on another tissue (buccal, skin or urine) or a family member may be required. For patients who have undergone bone marrow transplantation, please submit a skin, urine or buccal sample.

Ordering Privileges

Please refer to the APL Test Directory (http://ahsweb.ca/lab/apl-td-lab-test-directory) for specific ordering restrictions.

The genes included on the familial AML panel are:

Gene	Associated Condition	Features
CEBPA[1]	Familial acute myeloid leukemia	AML with a family history of AML AML diagnosed before age 50
GATA2[2]	GATA2 deficiency (includes phenotypes previously known as MonoMAC syndrome and Emberger syndrome)	Hematological malignancies including myelodysplastic syndrome (MDS)/AML Recurrent infections Lymphedema Cytopenia Sensorineural hearing loss
RUNX1[1]	Familial platelet disorder with associated myeloid malignancies	Thrombocytopenia Prolonged bleeding and/or easy bruising Increased risk of hematological malignancy Eczema

The conditions described above follow an autosomal dominant pattern of inheritance.

Associated Disorders

These genes are not currently known to be associated with other disorders.

When can I expect results?

Results may take up to 4 months.

How are results reported?

Results are sent to the ordering provider and available in Netcare and Connect Care.

Requisition forms, contact information and other resources can be found at:
http://ahsweb.ca/lab/if-lab-genetics-and-genomics

Contact Information

Genetic Counsellors, Genetics & Genomics
Edmonton: 780-407-1015

References

1. Adam MP, Mirzaa GM, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2022. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1116/ (accessed [2022 August])
2. Fabozzi F, Mastronuzzi A, Ceglie G, Masetti R and Leardini D (2022) GATA 2 Deficiency: Focus on Immune System Impairment. Front. Immunol. 13:865773.