

FLT3 Mutation Detection

FLT3 mutations have been identified in hematologic neoplasms, particularly in acute myeloid leukemia (AML) with an incidence of 20-30%. *FLT3* internal tandem duplication (ITD) mutations have been associated with an unfavorable outcome. *FLT3* tyrosine kinase domain mutations affecting codon D835 are also common (7%) but have a less clear prognostic significance. Early identification of these markers may provide better prognostication and aid in the determination of the most effective therapeutic regimen for the patient.

INDICATIONS FOR ORDERING

- Refine classification and determine prognosis in patients with AML.
- Monitor diagnosis and prognosis in patients with documented *FLT3*-mutated disease.

DISEASE OVERVIEW

Treatment Issues

- 50% of cases are cytogenetically normal AML (CN-AML) and are considered to be intermediate risk.
- Mortality varies significantly among patients within the intermediate risk group.
- Mutational testing may help in AML prognostication.
 - Presence of mutations may alter therapeutic decisions.

GENETICS

Structure/function

FLT3

- ITDs on exon 14/15; D835 mutation on exon 20
- Tyrosine kinase receptor regulates cell survival and maturation.

RELATED INFORMATION

[Acute Myeloid Leukemia - AML Tumor Markers](#)

TESTS TO CONSIDER

Initial Prognostication in AML

[FLT3 ITD and TKD Mutation Detection 3001161](#)

Method: Polymerase Chain Reaction

Genomic DNA is extracted and amplified in multiplex with primers targeting *FLT3* mutations in AML

- *FLT3* mutation detection by polymerase chain reaction (PCR) amplification of exon 14 ITDs and D835
- PCR products are *EcoRV*-digested and analyzed by capillary electrophoresis
- ITDs are reported with a signal ratio and D835 variants as Detected or Not Detected

[Myeloid Malignancies Mutation Panel by Next Generation Sequencing 2011117](#)

Method: Massively Parallel Sequencing

[NPM1 Mutation Detection by RT-PCR, Quantitative 3000066](#)

Method: Quantitative Reverse-Transcription Polymerase Chain Reaction

[CEBPA Mutation Detection 2004247](#)

Method: Polymerase Chain Reaction/Sequencing

[IDH1 and IDH2 Mutation Analysis, exon 4 2006444](#)

Method: Polymerase Chain Reaction/Sequencing