Hematologics, Inc.

• Molecular Analysis •

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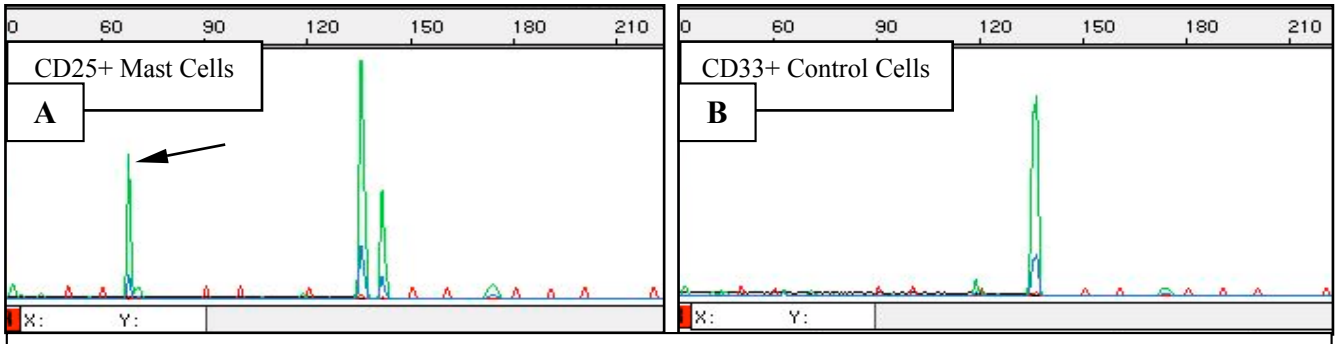
c-Kit D816V
Point Mutation Detection

Clinical Significance: **AML – t(8;21)**
Mastocytosis

The D816V c-Kit point mutation has been associated as a prognostic indicator with shorter event-free survival in core binding factor (CBF) acute myeloid leukemia (AML) [Boissel N et al. Leukemia 2006]. In mastocytosis the c-Kit mutation has been associated with both aggressive systemic disease and increased bone marrow mast cell content [Pardanani et al. Leukemia Research 27 2003: 739-742]. In addition, it has been shown that the D816V c-kit activation loop mutation is highly resistant to Imatinib (Gleevec). Therefore identification of this mutation might be informative for therapeutic decisions in systemic mast cell disease (SMCD) and acute myeloid leukemia (AML) [Krystal GW, Leuk Res 2004: S53-S59; Growney JD et al. Blood 2005 106(2): 721-4].



Detection of the D816V c-kit point mutation in sorted CD25+/117+ mast cells from bone marrow.
(A) *A 68 bp digestion product (indicative for the D816V c-kit point mutation) was detected in the CD25+ sorted Mast Cell population but not in the CD33+ Control Cell population* ***(B)****.*

Methodology:

Utilizes PCR amplification in combination with sequence specific restriction enzyme digestion and fluorescence-based capillary electrophoresis to identify the D816V c-kit point mutation. An intact 138 base pair product will be generated after PleI restriction enzyme digest from normal samples without the c-kit mutation. Samples harboring a homozygous D816V mutation have an active PleI restriction site and a 68 bp product will be generated.

The Hematologics Difference:

- Unsurpassed turn-around time.
- Only one specimen required for High Resolution Flow Cytometry™, Molecular Analysis & complementary DNA Archiving.
- Cell Sorting combined with Molecular Analysis can be used for highly sensitive and patient specific Minimal Residual Disease detection and for confirmatory testing in difficult cases.