EXPANDED FLUORESCENCE *IN-SITU* HYBRIDIZATION PANEL FOR ACUTE LEUKEMIAS

Fluorescence in-situ hybridization (FISH) testing complements conventional chromosome analysis in the diagnosis, prognosis, risk stratification, selection of treatment modalities and monitoring the treatment response in acute leukemias. The Cytogenetics laboratory has added seven new tests to expand the current FISH panel for acute leukemias.

Acute Myeloid Leukemia (AML): Acute myeloid leukemia with recurrent genetic abnormalities is a WHO defined distinct category. LSI *CBFB1-MYH11* dual color dual fusion (DCDF), LSI *DEK-NUP214* DCDF and LSI MECOM triple color break-apart (TC-BAP) probes will be used to confirm gene rearrangements and disease monitoring in AML with inversion (16)(p12q22)/t(16;16), t(6;9)(p23;q34.1) and inv (3)(q21.3q26.2)/t(3;3). The *CBFB1-MYH11* probes will replace currently used, LSI *CBFB1* DC-BAP probes in routine practice; *CBFB1 DC-BAP* probes will only be used for reflex testing to delineate variant rearrangements. LSI *NUP98* (11p15.4) probes will be employed to identify *de novo* and therapy related AML with chromosomally cryptic NUP98 translocations, a poor prognostic category. The *CBFA2T3-GLIS2* [inv(16)(p13.3q24.3)] rearrangement is prevalent in ~30% of non-DS AMKL and is associated with a worse outcome. The LSI *CBFA2T3/ GLIS2* DCDF probes will be utilized to identify this chromosomally cryptic inversion and variant translocation of *GLSI2* in pediatric and adult AMLs.

Acute Lymphocytic Leukemia (ALL) LSI *ABL1* (9q34.1) dual color break-apart (DC- BAP) and LSI *ABL2* (1q25.2) DC- BAP probes will be used to identify a subcategory of high risk acute lymphocytic leukemias with ABL1 and/ or ABL2 gene rearrangements that constitutes a group of recently described high risk ALL i.e. Philadelphia like acute lymphocytic leukemia. Timely and accurate identification of ALL with ABL1 and/ or ABL2 rearrangement is important to design effective treatment modalities as these patients respond to and benefit from tyrosine kinase inhibitors (TKIs) therapy, in an otherwise high risk ALL.

Acute Lymphocytic Leukemia FISH Panel

Translocation Probes	Enumeration/deletion Probes
LSI *BCR-ABL1* DC ES	CEP 4 (*D4Z1)*
LSI *ETV6-RUNX1* DC ES	CEP 10 (*D10Z1)*
LSI *KMT2A* DC- BAP	LSI *CDKN2A*/ CEP 9 (*D9Z1)*
LSI *ABL1* DC- BAP	
LSI *ABL2* DC-BAP	
LSI *ETV6* DC -BAP	
LSI *PDGFR-B* DC- BAP	
LSI *TRA/D* DC- BAP	

For any questions, please contact Mickey Muscolino at 464-6789 or Dr. Shiphali Gupta at guptashi@upstate.edu

Acute Myeloid Leukemia FISH Panel

Translocation Probes	Enumeration/ deletion Probes
LSI *PML- RARA* DCDF	LSI EGR1/ 5p15.2 DC
LSI *RUNX1T1-RUNX1* DCDF	CEP 7 (D7Z1)/ LSI D7S486 DC
LSI *CBFB1- MYH11* DCDF	CEP 8 (D8Z2)
LSI *DEK- NUP214* DCDF	LSI D20S108
LSI *MECOM* TC BAP	
LSI *NUP98* DC BAP	
LSI *KMT2A* DC- BAP	
LSI *CBFB1* DC - BAP	
LSI *RARA* DC- BAP	
LSI *GLIS2*/CBFA2T3 DCDF	