Molecular Diagnostic Tests Performed

February, 2009

Genetics		
Test	**Methodology**	**Purpose**
Factor V Leiden (R506Q) mutation	PCR-ARMS	APC Resistance
MTHFR A233V mutation	PCR-ARMS	Hyperhomocysteinemia
Prothrombin G20210A mutation	PCR-ARMS	Hypercoagulability
Hemochromatosis (HFE) C282Y and H63D mutations	PCR/RFLP	Iron overload
Cystic Fibrosis- neonate screen- 23+ mutations	PCR/OLA – fragment analysis	Neonatal screen for CF
Cystic Fibrosis- adult screen- 23+ mutations	PCR/OLA – fragment analysis	Adult screen for CF carriers
Mixed Chimerism	PCR/fragment analysis	Engraftment status in allogeneic transplant patients
UGT1A polymorphism	PCR/DNA sequencing	Pharmacogenetics- Irinotecan dosing
Hematopathology		
IgH Gene Rearrangement	PCR	B-cell clonality
T-Cell Receptor Gene Rearrangement, gamma/beta chains	PCR	T-cell clonality
t(9;22) (BCR/ABL) Screen for p210, p190 and p230 encoded transcripts	RT-PCR	Diagnosis of CML
Quantitiative t(9;22) (BCR/ABL)	Quantitative RT-PCR	Minimal residual disease
Cyclin D1 mRNA quantitation	Quantitative RT-PCR	Diagnosis of Mantle cell lymphoma
FLT-3 ITD and D835 mutations	PCR	Association with poor prognosis in AML
JAK2 V617F mutation	PCR	Association with polycythemia vera
Leukemia Translocation Screen	RT-PCR	Various common translocations found in AML and ALL
Infectious Disease		
Epstein-Barr Virus viral load	Quantitative PCR	EBV viral load in blood
Epstein-Barr Virus	In situ hybridization	Presence EBV in tissue to determine disease association

Bacteria Identification	PCR/DNA sequencing	Species identification