SPECIALTY LABORATORIES
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Specialty is pleased to introduce several new FISH assays used to evaluate several important lymphoid malignancies: Burkitt's lymphoma and variants with related c-MYC translocations, Anaplastic large cell lymphoma (ALK translocations), Large cell lymphomas and variants with bcl-6 translocations, and Chronic Lymphocytic Leukemia (CLL) related loss of Chromosome 13q. In addition other tests for common aneusomies of Chromosome 1, 7 and 18 are now available as separately orderable tests.

- **5890 FISH c-MYC 8q24**
- **5892 FISH ALK 2p23**
- **5894 FISH BCL6 3q27**
- **5896 FISH del(13)(q14.3) D13S25**
- **5898 FISH Chromosome 1 Aneuploidy**
- **5856 FISH Chromosome 7 Aneuploidy**
- **5858 FISH Chromosome 18 Aneuploidy**

We are also very pleased to offer a new array based Comparative Genomic Hybridization (array CGH) test used to detect over 40 different genetic syndromes and all unbalances of the subtelomeric regions of human chromosomes in a single reaction:

- **5888 Array CGH**

Array CGH is especially valuable for further evaluating causes of congenital disorders where conventional karyotyping may be normal or non-conclusive.

Finally, we are also introducing an important test for genotyping patients with suspected alpha-Thalassemia, a hemoglobinopathy which predominantly affects patients with familial ties or origins from Southeast Asian or Mediterranean regions.

- **5220 Alpha-Thalassemia GenotypR™**

For additional information on these tests, please visit our Web site at www.specialtylabs.com or contact Client Services at 800-421-4449.

Regards,

Michael C. Dugan, M.D.
Vice President and Laboratory Director

New and Recent Assays Available from *Specialty*

Effective date as noted

5220 Alpha-Thalassemia GenotypR™

(effective 10/17/2006)

Component		Method	Reference Range	Units
Alpha-Thalassemia		PCR	By Report	
	Specimen/Stability	5.0 (3.0) mL Whole Blood EDTA, Ambient 3 days, Refrigerated 3 days		
	Unacceptable Specimens	Frozen specimens		
	Collection Instructions	EDTA is the preferred anticoagulant, but ACD (A or B) and Heparin is also acceptable. Refrigerated specimens are also acceptable but not preferred.		
	Clinical Utility	Alpha thalassemia, the most prevalent of all thalassemias, is the deficient or absent production of alpha-globin. The imbalance of alpha and beta-globin chain production creates the red blood cell abnormalities of alpha thalassemia. The genetic abnormalities of alpha thalassemia primarily affect Southeast Asian and Mediterranean populations.		
	Performance Schedule	Set-up: Friday	Reported: 7 days	
	CPT Code	83891, 83900, 83901, 83894, 83909, 83896x21, 83912		
	Notes	Assay detects 21 mutations and covers more than 95% of all alpha-thalassemia mutant alleles commonly found in Mediterranean countries and more than 99% of all those reported in the Middle East and South East Asia. Not offered to New York patients at this time.		

5888 Array CGH

(effective 10/03/2006)

Component		Method	Reference Range	Units
Array CGH		CGH	By report	
	Specimen/Stability	5.0 (3.0) mL Whole Blood EDTA, Ambient 2 weeks, Refrigerated 2 weeks		
	Unacceptable Specimens	Frozen or clotted specimens		
	Collection Instructions	Draw aseptically; avoid contamination, hemolysis or clotting. Store at room temperature		
	Clinical Utility	Array Comparative Genomic Hybridization (CGH) testing is designed to detect chromosome deletions (losses) or duplications (copy gains) and is recommended in individuals with normal routine cytogenetic studies who have unexplained developmental delay/mental retardation, multiple congenital anomalies, dysmorphic features or other suspected genomic imbalances.		
	Performance Schedule	Set-up: Monday, Wednesday	Reported: 5 Days	
	CPT Code	Suggested: 83891, 88385		

5890 FISH c-MYC 8q24

(effective 10/17/2006)

Component		Method	Reference Range	Units
c-MYC 8q24		FISH	By Report	
	Specimen/Stability	3.0 (1.0) mL Bone Marrow Heparinized; Ambient 72 hours		
	Alternate Specimens	5.0 (3.0) mL Whole Blood Heparin; Ambient 72 hours		
	Unacceptable Specimens	Lithium heparinized specimens are not acceptable. Do not refrigerate or freeze.		
	Clinical Utility	To detect MYC translocations involving the MYC region at 8q24, including t(8;14) involving IGH at 14q32, the most frequently observed MYC region translocation, or one of the variant MYC translocations, either t(8;22)(q24;q11.2) or t(2;8)(p11.2;q24) that involve one of the two light chain immunoglobulin loci (kappa on chromosome 2 or lambda on 22). These 3 translocations characterize certain types of B-cell neoplasia and are found in both leukemias and lymphomas, in particular in Burkitt's lymphoma, high grade Burkitt-like lymphomas and some leukemic variants of other non-Hodgkin lymphomas (NHL).		
	Performance Schedule	Set-up: Sunday-Saturday	Reported: 7 days	
	CPT Code	88237, 88271x2, 88275, 88291		

5892 FISH ALK 2p23

(effective 10/17/2006)

Component		Method	Reference Range	Units
ALK 2p23		FISH	By Report	
	Specimen/Stability	3.0 (1.0) mL Bone Marrow Heparinized; Ambient 72 hours		
	Alternate Specimens	5.0 (3.0) mL Whole Blood Heparin; Ambient 72 hours		
	Unacceptable Specimens	Lithium heparinized specimens are not acceptable. Do not refrigerate or freeze.		
	Clinical Utility	Assist in the detection of the known 2p23 rearrangements that occur in t(2;5) and its variants. The t(2;5) has been shown to fuse the nucleophosmin (NPM) gene located on chromosome 5q35 with the receptor tyrosine kinase gene, named anaplastic lymphoma kinase (ALK) on chromosome 2p23.2. This NPM/ALK gene fusion gives rise to a chimeric protein that is overexpressed. The translocation t(2;5)(p23;q35) is a recurring abnormality in anaplastic large cell lymphoma (ALCL) a well-recognized clinicopathologic entity accounting for 2% of all adult non-Hodgkin's lymphomas (NHL) and about 13% of pediatric NHL; 46% of ALCL patients bear this signature translocation.		
	Performance Schedule	Set-up: Sunday-Saturday		Reported: 7 days
	CPT Code	88291, 88237, 88271x2, 88275		

5894 FISH BCL6 3q27

(effective 10/17/2006)

Component		Method	Reference Range	Units
BCL6 3q27		FISH	By Report	
	Specimen/Stability	3.0 (1.0) mL Bone Marrow Heparinized; Ambient 72 hours		
	Alternate Specimens	5.0 (3.0) mL Whole Blood Heparin; Ambient 72 hours		
	Unacceptable Specimens	Lithium heparinized specimens are not acceptable. Do not refrigerate or freeze.		
	Clinical Utility	Assist in detecting chromosome breaks associated with a number of different translocations that involve the BCL6 gene located on chromosome 3q27. The t(3;14)(q27;q32) is the most common translocation involving BCL6 in B-cell lymphoma. Although this translocation was predominantly associated with diffuse large B-cell lymphoma (DLBCL), recent studies have shown that it can also be found in follicular lymphomas (FL), often associated with a large cell component. Several t(3;14) variants have been described, e.g. t(3;22)(q27;q11.2) and t(2;3)(p12;q27).		
	Performance Schedule	Set-up: Sunday-Saturday		Reported: 7 days
	CPT Code	88237, 88271x2, 88275, 88291		

5896 FISH del(13)(q14.3) D13S25

(effective 10/17/2006)

Component		Method	Reference Range	Units
del(13)(q14.3) D13S25		FISH	By Report	
	Specimen/Stability	3.0 (1.0) mL Bone Marrow Heparinized; Ambient 72 hours		
	Alternate Specimens	5.0 (3.0) mL Whole Blood Heparin; Ambient 72 hours		
	Unacceptable Specimens	Lithium heparinized specimens are not acceptable. Do not refrigerate or freeze.		
	Clinical Utility	Assist in identifying deletions in the 13q14.3 region. A high incidence abnormality in B-cell CLL is deletion of chromosome 13 (13q14) occurring in 51% of CLL patients and in as much as 70% of mantle-cell lymphoma patients. The deletion of 13q14.3 affects a locus telomeric to the RB1 gene (retinoblastoma gene) and the marker D13S25 which bears a relation to a candidate tumor suppressor gene.		
	Performance Schedule	Set-up: Sunday-Saturday		Reported: 7 days
	CPT Code	88237, 88271, 88275, 88291		

5898 FISH Chromosome 1 Aneuploidy

(effective 10/17/2006)

Component	Method	Reference Range	Units
Chromosome 1 Aneuploidy	FISH	By Report	
Specimen/Stability	3.0 (1.0) mL Bone Marrow Heparinized; Ambient 72 hours		
Alternate Specimens	5.0 (3.0) mL Whole Blood Heparin; Ambient 72 hours		
Unacceptable Specimens	Lithium heparinized specimens are not acceptable. Do not refrigerate or freeze.		
Clinical Utility	Assist in the identification and enumeration of chromosome 1. Chromosome 1 aberrations are frequently described, the short arm being preferentially involved in deletions and the long arm in gains. Trisomy 1 (partial or complete) is a frequent and consistent chromosome change in human malignancies.		
Performance Schedule	Set-up: Sunday-Saturday	Reported: 7 days	
CPT Code	88237, 88271, 88275, 88291		

5856 FISH Chromosome 7 Aneuploidy

(effective 10/17/2006)

Component	Method	Reference Range	Units
Chromosome 7 Aneuploidy	FISH	By Report	
Specimen/Stability	3.0 (1.0) mL Bone Marrow Heparinized; Ambient 72 hours		
Alternate Specimens	5.0 (3.0) mL Whole Blood Heparin; Ambient 72 hours		
Unacceptable Specimens	Lithium heparinized specimens are not acceptable. Do not refrigerate or freeze.		
Clinical Utility	Assist in the identification and enumeration of chromosome 7. Trisomy 7 not only occurs frequently as an anomaly additional to other karyotypic changes in tumors, but has been known as the sole anomaly in some tumors and, most interestingly, in the alleged normal tissues near tumors. It is mostly found as an additional aberration in malignant lymphomas. Whereas the overall frequency of +7 in non-Hodgkin's lymphomas (NHL) is 10-15%, only a few cases have been published in which +7 was the sole anomaly. Partial or total loss of chromosome 7 represents a common chromosomal abnormality associated with clonal myeloid disorders, in particular myeloproliferative disorders (MPD), myelodysplastic syndromes (MDS) and acute myeloid leukemia (AML).		
Performance Schedule	Set-up: Sunday-Saturday	Reported: 7 days	
CPT Code	88237, 88271, 88275, 88291		

5858 FISH Chromosome 18 Aneuploidy

(effective 10/17/2006)

Component	Method	Reference Range	Units
Chromosome 18 Aneuploidy	FISH	By Report	
Specimen/Stability	3.0 (1.0) mL Bone Marrow Heparinized; Ambient 72 hours		
Alternate Specimens	5.0 (3.0) mL Whole Blood Heparin; Ambient 72 hours		
Unacceptable Specimens	Lithium heparinized specimens are not acceptable. Do not refrigerate or freeze.		
Clinical Utility	Assist in the identification and enumeration of chromosome 18. Trisomy 18 occurs in 10% of all Non-Hodgkin's lymphoma (NHL) and is almost always accompanied by other numerical aberrations or structural changes. A nonrandom association between trisomy 18 and trisomy 3 has been noticed.		
Performance Schedule	Set-up: Sunday-Saturday	Reported: 7 days	
CPT Code	88237, 88271, 88275, 88291		