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New Test Announcements

May 29, 2007

Dear Colleague:

This announcement contains new testing in the areas of maternal screening, infectious disease, and pharmacogenomics. For maternal screening, three new tests are now available that rapidly detect the most common autosomal trisomies and the sex chromosome aneuploidies by Quantitative Fluorescent Polymerase Chain Reaction (QF-PCR). Trisomies 13, 18, and 21 (which are also known as Patau Syndrome, Edwards Syndrome, and Down Syndrome respectively) and the sex chromosome aneuploidies are the most frequent chromosomal anomalies in live births. Traditionally, prenatal diagnosis of chromosome abnormalities involves karyotyping cells cultured from amniotic fluid or the chorionic villi and takes about 2 weeks. Specialty is pleased to offer three new tests, the Aneuploidy-13-18-21-X-Y DetectR™ (PCR), the Trisomy-13-18-21 DetectR™ (PCR), and the Aneuploidy-X-Y DetectR™ (PCR), which rapidly detect trisomies 13, 18, and 21, and the sex chromosome aneuploidies by Quantitative Fluorescent Polymerase Chain Reaction (QF-PCR), with a turnaround time of 24 hours.

Advantages of Rapid Aneuploidy and Trisomy Detection by Aneuploidy-13-18-21-X-Y DetectR™ *QF-PCR*

- Aneuploidy-13-18-21-X-Y DetectR™by QF-PCR requires less than 100 ng of DNA. By comparison AneuVysion FISH requires roughly double the amount of amniotic fluid needed for testing
- QF-PCR can be easily performed on chorionic villus samples (CVS) or amniocentesis samples, whereas the AneuVysion FISH assay is intended for amniotic fluid only.
- QF-PCR has less potential for misdiagnosis or non-informative tests due to maternal contamination of fetal samples compared to FISH
- QF-PCR takes 24 hours to complete compared to 2-3 days for FISH and 7-10 days for karyotyping, which is sometimes unsuccessful due to culture failure. QF-PCR is much more reliable than screening based on maternal serum biochemistry and fetal ultrasonography.
- QF-PCR in conjunction with fetal ultrasonography misses less than 1% of serious chromosomal abnormalities, whereas maternal serum biochemistry in conjunction with fetal ultrasonography misses 15-20% of the cases of trisomy 21, about 9% of the cases of trisomy 18, and does not consider X, Y, or chromosome 13.

(A complete list of references for Aneuploidy-13-18-21-X-Y DetectR™ are provided on a technical bulletin which is available at www.specialtylabs.com.)

Mycobacterium DNA DetectR™

In infectious disease we have a PCR DNA test for *Mycobacterium* (genus specific only) that offers a report time of only 2 days and a broad selection of sample types acceptable for testing. This test is not designed to differentiate species. Detection is solely genus specific. For suspected *M. tuberculosis* or *M. avium* cases, please contact client relations for PCR assays specific for *M. tuberculosis* complex and/or *M. avium complex*.

Abacavir GenotypR™ (Ziagen®)

In pharmacogenomics we have added the HLA-B*57 (Abacavir) DetectR™ to our menu for patients currently taking or who are considering taking abacavir for HIV antitvirial therapy. HLA-B*5701 is present in 78% of Caucasian patients with Abacavir hypersensitivity and in 2% Abacavir-tolerant patients. Abacavir is a commonly used nucleoside analogue with potent antiviral activity against HIV-1. About 5% (range 0–14%) of caucasian patients treated with abacavir develop a hypersensitivity reaction characterized by fever, rash, gastrointestinal symptoms, and lethargy or malaise within 6 weeks of treatment. These symptoms remit when the drug is discontinued and worsen with re-exposure. Exclusion of HLA-B*5701 positive individuals from abacavir treatment would largely prevent abacavir hypersensitivity.

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

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

1390 Abacavir GenotypR™ (Ziagen®)(HLA-B*57)

(Available 6/12/07)

Component	Method	Reference Range/Units
HLA-B*57 (Abacavir) DetectR™ (Ziagen®)	PCR	By Report

Specimen/Stability	5 (3) mL Whole Blood, Ambient – 1 Week, Refrigerated – 1 Week
Clinical Utility	HLA-B*5701 is present in 78% of Caucasian patients with abacavir hypersensitivity and in 2% abacavir-tolerant patients. Abacavir is a commonly used nucleoside analogue with potent antiviral activity against HIV-1. About 5% (range 0–14%) of Caucasian patients treated with abacavir develop a hypersensitivity reaction characterized by fever, rash, gastrointestinal symptoms and lethargy or malaise within 6 weeks of treatment. These symptoms remit when the drug is discontinued and worsen with re-exposure. Exclusion of HLA-B*5701 positive individuals from abacavir treatment would largely prevent abacavir hypersensitivity.
Schedule	Friday
Report	4 days
CPT Code	83891, 83894x2, 83896x60, 83900, 83901x2, 83909, 83912
Collection	ACD and Heparin are not acceptable. Do not freeze. Refrigerated specimens are also acceptable but not preferred.
Note	This test is not available to NY patients until further notice

5855 Aneuploidy 13-18-21-X-Y DetectR™ (PCR)

(Available 06/12/07)

Component	Method	Reference Range/Units
Chromosomes 13, 18, 21, X & Y Aneuploidy DetectR™	PCR	By Report

Specimen/Stability	5 (2) mL Amniotic Fluid, Ambient – 72 Hours, Refrigerated – 72 Hours 5 (3) mL Mother's Whole Blood EDTA, Ambient – 1 Week, Refrigerated – 1 Week
Alternate Specimens	2 confluent T25 flasks (1 confluent T25 flask) Cultured Amniotic Fluid, Ambient – 72 Hours, Refrigerated – 72 Hours 5 (3) mL Mother's Whole Blood EDTA, Ambient – 1 Week, Refrigerated – 1 Week
Alternate Specimens	30 (20) mg Chronic Villus Specimen, Refrigerated – 48 Hours 5 (3) mL Mother's Whole Blood EDTA, Ambient – 1 Week, Refrigerated – 1 Week
Clinical Utility	This PCR-based assay provides rapid detection of trisomies 13, 18, and 21 and sex chromosome aneuploidies. These conditions account for nearly 2/3 of all abnormalities identified at the time of amniocentesis, and 95-98% of clinically significant chromosomal abnormalities detected in live-born infants. This test is not intended to be used as a stand alone assay for making clinical decisions. The test results are intended to be used in conjunction with conventional karyotyping and other clinical information to confirm a diagnosis of suspected chromosome abnormality and to provide a preliminary result to parents. This assay will not detect chromosome mosaicism, duplications, deletions, or structural rearrangements and does not identify all birth and/or developmental abnormalities.
Schedule	Tuesday – Thursday
Report	2 days
CPT Code	83891, 83900, 83901x24, 83909x2, 83912
Note	Mother's blood should accompany all prenatal specimens. If warranted, test will reflex to #5365 Maternal Cell Contamination (MCC) for an additional fee and extended turnaround time.
Collection	Discard first 2mL of amniotic fluid. Bloody fluid is not acceptable. Collect as usual and ship in a sterile tube. Maintain a back-up culture for the amniotic fluid. Transfer the CVS by using sterile technique to one or two 15-mL centrifuge tube(s) with 15 mL of transport medium.

5857 Trisomy-13-18-21 DetectR™ (PCR)

(Available 06/12/07)

Component	Method	Reference Range/Units
Trisomy 13, 18, and 21 DetectR™	PCR	By Report

Specimen/Stability	3 (2) mL Whole Blood, Ambient – 1 Week, Refrigerated – 1 Week
Clinical Utility	This PCR-based assay provides rapid detection of trisomies 13, 18, and 21. This test is not intended to be used as a stand alone assay for making clinical decisions. The test results are intended to be used in conjunction with other clinical information and further confirmatory karyotyping when requested for rapid identification of suspected chromosomal abnormalities. This assay will not detect chromosome mosaicism, duplications, deletions, or structural rearrangements and does not identify all birth and/or developmental abnormalities.
Schedule	Tuesday, Thursday
Report	2 days
CPT Code	83891, 83900, 83901x14, 83909x2, 83912
Collection	EDTA is the preferred anticoagulant, but ACD (A or B) and Heparin are also acceptable. Do not freeze. Refrigerated specimens are also acceptable but not preferred.

5859 Chromosomes X-Y Aneuploidy DetectR™ (Available 06/12/07)

Component	Method	Reference Range/Units
Chromosomes X and Y Aneuploidy DetectR™	PCR	By Report

Specimen/Stability	3 (2) mL Whole Blood, Ambient – 1 Week, Refrigerated – 1 Week
Clinical Utility	Abnormalities of the sex chromosomes occur frequently, in approximately one in 800 to 1000 male or female live births. This PCR-based assay detects sex chromosome aneuploidies including Turner syndrome (monosomy X) and Klinefelter syndrome (predominantly 47,XXY) and other sex chromosome abnormalities such as 47,XYY and 47,XXX. This test is not intended to be used as a stand alone assay for making clinical decisions. The test results are intended to be used in conjunction with other clinical information and further confirmatory karyotyping when requested for rapid identification of suspected chromosomal abnormalities. This assay will not detect chromosome mosaicism, duplications, deletions, or structural rearrangements and does not identify all birth and/or developmental abnormalities.
Schedule	Tuesday, Thursday
Report	2 days
CPT Code	83891, 83900, 83901x8, 83909, 83912
Collection	EDTA is the preferred anticoagulant, but ACD (A or B) and Heparin are also acceptable. Do not freeze. Refrigerated specimens are also acceptable but not preferred.

7467 Mycobacterium DNA DetectR™ (Available 06/12/07)

Component	Method	Reference Range/Units
Mycobacterium DNA Source	PCR	Not Detected

Specimen/Stability	Tissue Paraffin Block, Ambient – 4 Days, Refrigerated – 4 Days, Frozen – 2 Months
Alternate Specimens	Tissue, Frozen – 1 Month 2 (1) Sputum, Ambient – 24 Hours, Refrigerated – 7 Days 2 (1) Bronchial Lavage, Ambient – 24 Hours, Refrigerated – 7 Days 10 (5) mL Whole Blood ACD, Ambient – 24 Hours 2 (1) mL Fluid, Ambient – 24 Hours, Refrigerated – 7 Days, Frozen – 1 Month
Clinical Utility	This assay supports the diagnosis of mycobacterial infection by detection of *Mycobacterium* DNA in tissue, blood, and other fluids. The absence of detectable DNA does not rule out infection, since pathogen load may be at very low levels.
Schedule	Tuesday, Friday
Report	2 days
CPT Code	87551
Note	This test is not designed to differentiate species. Detection is solely genus specific. For suspected *M. tuberculosis* or *M. avium* cases, please contact client services for PCR assays specific for *M. tuberculosis* complex and/or *M. avium* complex.
Collection	1) Place fresh tissue or needle biopsy in sterile saline and freeze prior to shipment. 2) Collect sputum early in the morning from a deep productive cough. Place in a sterile, leak proof container. 3) Collect bronchoalveolar lavage in a sterile, leak proof container. Bronchial wash specimens are also acceptable if shipped under refrigerated or frozen conditions. 4) Collect pleural, pericardial, or gastric fluid or aspirates in a sterile container. If gastric aspirate cannot be shipped immediately, add 100 mg of sodium bicarbonate as a buffer to neutralize any acid detrimental to Mycobacterium spp. 5) Ship whole blood at ambient temperature within 24 hours of collection. Heparinized blood is not acceptable.