SPECIALTY LABORATORIES
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Valencia, CA 91355 www.specialtylabs.com

September/October, 2005

Dear Colleague:

New Tests

We are introducing a number of new tests in this letter including several related to Ashkenazi Jewish genetic diseases:

- Ashkenazi Jewish GenotypR™– Carrier Panel tests for 31 mutations associated with 8 diseases for pre-pregnancy evaluation of disease risk.
- Individual GenotypR™ assays for Bloom Syndrome, Canavan Disease, Familial Dysautonomia, Fanconi Anemia GrpC, Gaucher Disease, Mucolipidosis Type IV, Niemann-Pick Disease, Tay-Sachs Disease with separate carrier, diagnostic and fetal study testing.
- Heat Shock Protein 70 (Hsp70) Autoantibodies – human hsp are elevated in a number of autoimmune, infectious and inflammatory conditions.
- *Mycobacterium tuberculosis* Complex DNA DetectR™ by real time PCR detects the MTb complex of *M. tuberculosis, M. bovis , M. bovis* BCG, *M. africanum, M. microti, M. canettii*. This discrete group of organisms share a DNA homology of greater than 95%.
- N-Telopeptides Serum is used in assessing antiresorptive therapy in osteoporosis.

We have also created a new panel MTHFR C677T/A1298C GenotypR™ combining MTHFR mutations C677T and A1298 for assessing homocysteine metabolism. These two mutations are associated with neural tube defects and cardiovascular disease due to hyperhomocysteinemia and lowered plasma folate levels. This new panel will replace the individual and reflex panels for the MTHFR mutations (see Test Discontinuations).

Fee for Attempted Assay Performance That Does Not Generate a Reportable Result

Specialty has determined that it is not necessary at this time to implement the fee for attempted assay performance previously announced as effective September 1, 2005. We have introduced internal production methods to help alleviate the need for these fees. If you would like more information, please call Client Services at 800-421-4449.

Cystic Fibrosis Report Changes

In order to assure faster, more efficient reporting for Cystic Fibrosis GenotypR™, *Specialty* is now issuing the results and interpretive analysis via the standard electronic interface, fax and mail routes. The specialized color reports will no longer be issued; risk revision tables will still be posted on our Web site www.specialtylabs.com. Because the report contains information on contingencies that would affect the interpretive analysis such as ethnicity and family history of disease, *Specialty* will no longer delay result release and attempt to obtain any data not included on the initial requisition. We will, however, continue to review results in light of new information when it is received. If you have any questions regarding this change in procedure, please call Client Services at 800-421-4449.

Maternal Cell Contamination (MCC)

All prenatal amniotic fluid specimens for genetic testing must be accompanied by a maternal blood sample for analysis of possible maternal cell contamination. If necessary to clarify a result interpretation (e.g., the fetal sample has the same mutation as the mother), *Specialty* will perform a Maternal Cell Contamination test (#5365) for an additional fee. This testing will extend the TAT about 2 weeks. If you have any questions, please contact Client Technical Services at 800-421-4449 or ask to speak to Milhan Telatar, Ph.D. our Scientific Director of Molecular Genetics.

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

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

New from *Specialty*

Effective Tuesday, October 18, 2005 or as noted

5206 Ashkenazi Jewish GenotypR™ - Carrier Panel (includes common mutations for Bloom Syndrome, Canavan Disease, Familial Dysautonomia, Fanconi Anemia GrpC, Gaucher Disease, Mucolipidosis Type IV, Niemann-Pick Disease, Tay-Sachs Disease)

effective 10/18/05

Component	Method	Reference Range	Units
AJ Panel carrier study	PCR	By report	

Specimen/Stability	5.0 (3.0) mL Whole Blood EDTA (Lavender top) Ambient 3 days; Refrigerated 3 days
Collection Instructions	EDTA is preferred but ACD is also acceptable. Heparinized whole blood is not acceptable. Ambient is preferred, refrigerated is acceptable; frozen is not acceptable.
Clinical Utility	Detects 31 mutations/polymorphisms in 8 genes (Tay - Sachs disease, Canavan Disease, Familial Dysautonomia, Gaucher Disease, Bloom Syndrome, Fanconi Anemia GrpC, Niemann-Pick Disease, and Mucolipidosis Type IV) responsible for conditions that are predominantly found in persons of Ashkenazi ancestry.
Schedule	Thursday
Turnaround Time	3-10 days
CPT Code	83891, 83892x2, 83896x31, 83901, 83912
Notes	Carrier detection will be performed only for adult patients and pregnant minors. Prior carrier risk (the risk, before testing, that an asymptomatic patient is carrier) and revised carrier risk (the risk that the same patient is a carrier after a negative test), based on family history and ethnicity are provided in a patient-specific report. For specific diagnostic workup, refer to individual tests below. Please use specialized AJ GenotypR™ Requisition Form, which requests the clinical information necessary for interpretation of results, (patient ethnicity and family history). Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com

The following tests are all related to the Ashkenazi-Jewish Panel offering and have the same effective date, schedule and assay time as the panel listing. Specimen requirements and collection instructions are the same as for the panel listing for all Carrier and Diagnostic testing. Instructions for fetal studies are included with the individual test descriptions.

5216 Bloom Syndrome GenotypR™ - Carrier Testing

Component	Method	Reference Range	Units
Bloom Syndrome Carrier Testing	PCR	By report	

Specimen/Stability	same as A-J Panel above
Collection Instructions	same as A-J Panel above
Clinical Utility	Detects a single mutation (2281del6/ins7) in the Bloom Syndrome gene for carrier assessment in adult patients. The 2281del6/ins7 mutation is responsible for ~97% of mutant alleles in Ashkenazi Jewish Population.
Schedule/TAT	same as A-J Panel above
CPT Code	83891, 83892x2, 83896, 83901, 83912
Notes	same as A-J Panel above

5217 Bloom Syndrome GenotypR™ - Diagnostic Testing

Component	Method	Reference Range	Units
Bloom Syndrome Diagnostic Testing	PCR	By report	

Specimen/Stability	same as A-J Panel above
Collection Instructions	same as A-J Panel above
Clinical Utility	Detects a single mutation (2281del6/ins7) in the Bloom Syndrome gene for support of diagnosis in a symptomatic patient. The 2281del6/ins7 mutation is responsible for ~97% of mutant alleles in Ashkenazi Jewish Population.
Schedule/TAT	same as A-J Panel above
CPT Code	83891, 83892x2, 83896, 83901, 83912
Notes	Please use specialized AJ GenotypR™ Requisition Form. Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com

5218 Bloom Syndrome GenotypR™ - Fetal Study reflex to MCC

Component	Method	Reference Range	Units
Bloom Syndrome Fetal Study	PCR	By report	

Specimen/Stability	20 (10) mL Amniotic Fluid; Ambient 24 hr 5 (3) mL of Maternal Whole Blood EDTA; Ambient 3 days, Refrigerated 3 days
Collection Instructions	Amniotic Fluid: Ship immediately, do not refrigerate or freeze. Maternal WholeBlood must accompany amniotic fluid specimen.
Alternate Specimens	Cultured Amniocytes: Do not refrigerate or freeze. Ship flask at confluency, topped off with culture media. An internal code will be added at the laboratory for cell culture. For chorionic villus specimens, please contact Technical Services. Maternal blood is required for potential studies of maternal cell contamination (MCC) of the fetal DNA.
Clinical Utility	Detects a single mutation (2281del6/ins7) in the Bloom Syndrome gene for prenatal diagnosis of an at-risk fetus. The 2281del6/ins7 mutation is responsible for ~97% of mutant alleles in Ashkenazi Jewish Population.
Schedule/TAT	same as A-J Panel above (when cell culture required, add ~2 weeks)
CPT Code	83891, 83892x2, 83896, 83901, 83912
Notes	Please use specialized AJ GenotypR™ Requisition Form. Informed consent is required for residents of New York. Consent form is available on our website: www.specialtylabs.com. Maternal blood should accompany any prenatal specimen. If warranted, test will reflex to #5365 Maternal Cell Contamination for an additional fee and extended turnaround time.

5226 Canavan Disease GenotypR™ - Carrier Testing

Component	Method	Reference Range	Units
Canavan Disease Carrier Testing	PCR	By report	

Specimen/Stability	same as A-J Panel above
Collection Instructions	same as A-J Panel above
Clinical Utility	Detects 4 Canavan Disease mutations (433(-2)A>G, Y231X (C>A), E285A, A305E) and a benign variant, Y231Y (C>T) for carrier assessment in adult patients. These mutations are responsible for ~99% of mutant Canavan alleles in Ashkenazi Jewish Population.
Schedule/TAT	same as A-J Panel above
CPT Code	83891, 83892x2, 83896x5, 83901, 83912
Notes	same as A-J Panel above

5227 Canavan Disease GenotypR™ - Diagnostic Testing

Component	Method	Reference Range	Units
Canavan Disease Diagnostic Testing	PCR	By report	

Specimen/Stability	same as A-J Panel above
Collection Instructions	same as A-J Panel above
Clinical Utility	Detects 4 Canavan Disease mutations (433(-2)A>G, Y231X (C>A), E285A, A305E) and a benign variant, Y231Y (C>T) for support of diagnosis in a symptomatic patient. These mutations are responsible for ~99% of mutant Canavan alleles in Ashkenazi Jewish Population.
Schedule/TAT	same as A-J Panel above
CPT Code	83891, 83892x2, 83896x5, 83901, 83912
Notes	Please use specialized AJ GenotypR™ Requisition Form. Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com

5228 Canavan Disease GenotypR™ - Fetal Study reflex to MCC

Component	Method	Reference Range	Units
Canavan Disease Fetal Study	PCR	By report	

Specimen/Stability	20 (10) mL Amniotic Fluid; Ambient 24 hr 5 (3) mL of Maternal Whole Blood EDTA; Ambient 3 days, Refrigerated 3 days
Collection Instructions	Amniotic Fluid: Ship immediately, do not refrigerate or freeze. Maternal Whole Blood must accompany amniotic fluid specimen.
Alternate Specimens	Cultured Amniocytes: Do not refrigerate or freeze. Ship flask at confluency, topped off with culture media. An internal code will be added at the laboratory for cell culture. For chorionic villus specimens, please contact Technical Services. Maternal blood is required for potential studies of maternal cell contamination (MCC) of the fetal DNA.
Clinical Utility	Detects 4 Canavan Disease mutations (433(-2)A>G, Y231X (C>A), E285A, A305E) and a benign variant, Y231Y (C>T) for prenatal diagnosis of an at-risk fetus. These mutations are responsible for ~99% of mutant Canavan alleles in Ashkenazi Jewish Population.
Schedule/TAT	same as A-J Panel above (when cell culture required, add ~2 weeks)
CPT Code	83891, 83892x2, 83896x5, 83901, 83912
Notes	Please use specialized AJ GenotypR™ Requisition Form. Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com. Maternal blood should accompany any prenatal specimen. If warranted, test will reflex to #5365 Maternal Cell Contamination for an additional fee and extended turnaround time.

5236 Familial Dysautonomia GenotypR™ - Carrier Testing

Component	Method	Reference Range	Units
Familial Dysautonomia Carrier Testing	PCR	By report	
Specimen/Stability	same as A-J Panel above		
Collection Instructions	same as A-J Panel above		
Clinical Utility	Detects 2 Familial Dysautonomia mutations (R696P, IVS20(+6)T>C) for carrier assessment in adult patients. These mutations are responsible for ~99% of mutant Familial Dysautonomia alleles in Ashkenazi Jewish Population.		
Schedule/TAT	same as A-J Panel above		
CPT Code	83891, 83892x2, 83896x2, 83901, 83912		
Notes	same as A-J Panel above		

5237 Familial Dysautonomia GenotypR™ - Diagnostic Testing

Component	Method	Reference Range	Units
Familial Dysautonomia Diagnostic Testing	PCR	By report	
Specimen/Stability	same as A-J Panel above		
Collection Instructions	same as A-J Panel above		
Clinical Utility	Detects 2 Familial Dysautonomia mutations (R696P, IVS20(+6)T>C) for support of diagnosis in a symptomatic patient. These mutations are responsible for ~99% of mutant Familial Dysautonomia alleles in Ashkenazi Jewish Population.		
Schedule/TAT	same as A-J Panel above		
CPT Code	83891, 83892x2, 83896x2, 83901, 83912		
Notes	Please use specialized AJ GenotypR™ Requisition Form. Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com		

5238 Familial Dysautonomia GenotypR™ - Fetal Study reflex to MCC

Component	Method	Reference Range	Units
Familial Dysautonomia Fetal Study	PCR	By report	
Specimen/Stability	20 (10) mL Amniotic Fluid; Ambient 24 hr 5 (3) mL of Maternal Whole Blood EDTA; Ambient 3 days, Refrigerated 3 days		
Collection Instructions	Amniotic Fluid: Ship immediately, do not refrigerate or freeze. Maternal Whole Blood must accompany amniotic fluid specimen.		
Alternate Specimens	Cultured Amniocytes: Do not refrigerate or freeze. Ship flask at confluency, topped off with culture media. An internal code will be added at the laboratory for cell culture. For chorionic villus specimens, please contact Technical Services. Maternal blood is required for potential studies of maternal cell contamination (MCC) of the fetal DNA.		
Clinical Utility	Detects 2 Familial Dysautonomia mutations (R696P, IVS20(+6)T>C) for prenatal diagnosis of an at-risk fetus. These mutations are responsible for ~99% of mutant Familial Dysautonomia alleles in Ashkenazi Jewish Population.		
Schedule/TAT	same as A-J Panel above (when cell culture required, add ~2 weeks)		
CPT Code	83891, 83892x2, 83896x2, 83901, 83912		
Notes	Please use specialized AJ GenotypR™ Requisition Form. Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com. Maternal blood should accompany any prenatal specimen. If warranted, test will reflex to #5365 Maternal Cell Contamination for an additional fee and extended turnaround time.		

5246 Fanconi Anemia GrpC GenotypR™ - Carrier Testing

Component	Method	Reference Range	Units
Fanconi Anemia GrpC Carrier Testing	PCR	By report	
Specimen/Stability	same as A-J Panel above		
Collection Instructions	same as A-J Panel above		
Clinical Utility	Detects 2 Fanconi Anemia GrpC mutations (322delG, IVS4(+4)A>T) for carrier assessment in adult patients. These mutations are responsible for ~99% of mutant Fanconi Anemia GrpC alleles in Ashkenazi Jewish Population.		
Schedule/TAT	same as A-J Panel above		
CPT Code	83891, 83892x2, 83896x2, 83901, 83912		
Notes	same as A-J Panel above		

5247 Fanconi Anemia GrpC GenotypR™ - Diagnostic Testing

Component	Method	Reference Range	Units
Fanconi Anemia GrpC Diagnostic Testing	PCR	By report	

Specimen/Stability	same as A-J Panel above
Collection Instructions	same as A-J Panel above
Clinical Utility	Detects 2 Fanconi Anemia GrpC mutations (322delG, IVS4(+4)A>T) for support of diagnosis in a symptomatic patient. These mutations are responsible for ~99% of mutant Fanconi Anemia GrpC alleles in Ashkenazi Jewish Population.
Schedule/TAT	same as A-J Panel above
CPT Code	83891, 83892x2, 83896x2, 83901, 83912
Notes	Please use specialized AJ GenotypR™ Requisition Form. Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com

5248 Fanconi Anemia GrpC GenotypR™ - Fetal Study reflex to MCC

Component	Method	Reference Range	Units
Fanconi Anemia GrpC Fetal Study	PCR	By report	

Specimen/Stability	20 (10) mL Amniotic Fluid; Ambient 24 hr 5 (3) mL of Maternal Whole Blood EDTA; Ambient 3 days, Refrigerated 3 days
Collection Instructions	Amniotic Fluid: Ship immediately, do not refrigerate or freeze. Maternal Whole Blood must accompany amniotic fluid specimen.
Alternate Specimens	Cultured Amniocytes: Do not refrigerate or freeze. Ship flask at confluency, topped off with culture media. An internal code will be added at the laboratory for cell culture. For chorionic villus specimens, please contact Technical Services. Maternal blood is required for potential studies of maternal cell contamination (MCC) of the fetal DNA.
Clinical Utility	Detects 2 Fanconi Anemia GrpC mutations (322delG, IVS4(+4)A>T) for prenatal diagnosis of an at-risk fetus. These mutations are responsible for ~99% of mutant Fanconi Anemia GrpC alleles in Ashkenazi Jewish Population.
Schedule/TAT	same as A-J Panel above (when cell culture required, add ~2 weeks)
CPT Code	83891, 83892x2, 83896x2, 83901, 83912
Notes	Please use specialized AJ GenotypR™ Requisition Form. Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com. Maternal blood should accompany any prenatal specimen. If warranted, test will reflex to #5365 Maternal Cell Contamination for an additional fee and extended turnaround time.

5256 Gaucher Disease GenotypR™ - Carrier Testing

Component	Method	Reference Range	Units
Gaucher Disease Carrier Testing	PCR	By report	

Specimen/Stability	same as A-J Panel above
Collection Instructions	same as A-J Panel above
Clinical Utility	Detects 8 Gaucher Disease mutations (84G>GG, IVS2(+1)G>A, N370S, Del55bp, V394L, D409H, L444P, R496H) for carrier assessment in adult patients. These mutations are responsible for ~96% of mutant Gaucher alleles in Ashkenazi Jewish Population.
Schedule/TAT	same as A-J Panel above
CPT Code	83891, 83892x2, 83896x8, 83901, 83912
Notes	same as A-J Panel above

5257 Gaucher Disease GenotypR™ - Diagnostic Testing

Component	Method	Reference Range	Units
Gaucher Disease Diagnostic Testing	PCR	By report	

Specimen/Stability	same as A-J Panel above
Collection Instructions	same as A-J Panel above
Clinical Utility	Detects 8 Gaucher Disease mutations (84G>GG, IVS2(+1)G>A, N370S, Del55bp, V394L, D409H, L444P, R496H) for support of diagnosis in a symptomatic patient. These mutations are responsible for ~96% of mutant Gaucher alleles in Ashkenazi Jewish Population.
Schedule/TAT	same as A-J Panel above
CPT Code	83891, 83892x2, 83896x8, 83901, 83912
Notes	Please use specialized AJ GenotypR™ Requisition Form. Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com

5258 Gaucher Disease GenotypR™ - Fetal Study reflex to MCC

Component	Method	Reference Range	Units
Gaucher Disease Fetal Study	PCR	By report	

Specimen/Stability	20 (10) mL Amniotic Fluid; Ambient 24 hr 5 (3) mL of Maternal Whole Blood EDTA; Ambient 3 days, Refrigerated 3 days
Collection Instructions	Amniotic Fluid: Ship immediately, do not refrigerate or freeze. Maternal Whole Blood must accompany amniotic fluid specimen.
Alternate Specimens	Cultured Amniocytes: Do not refrigerate or freeze. Ship flask at confluency, topped off with culture media. An internal code will be added at the laboratory for cell culture. For chorionic villus specimens, please contact Technical Services. Maternal blood is required for potential studies of maternal cell contamination (MCC) of the fetal DNA.
Clinical Utility	Detects 8 Gaucher Disease mutations (84G>GG, IVS2(+1)G>A, N370S, Del55bp, V394L, D409H, L444P, R496H) for prenatal diagnosis of an at-risk fetus. These mutations are responsible for ~96% of mutant Gaucher alleles in Ashkenazi Jewish Population.
Schedule/TAT	same as A-J Panel above (when cell culture required, add ~2 weeks)
CPT Code	83891, 83892x2, 83896x8, 83901, 83912
Notes	Please use specialized AJ GenotypR™ Requisition Form. Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com. Maternal blood should accompany any prenatal specimen. If warranted, test will reflex to #5365 Maternal Cell Contamination for an additional fee and extended turnaround time.

5266 Mucolipidosis Type IV GenotypR™ - Carrier Testing

Component	Method	Reference Range	Units
Mucolipidosis Type IV Carrier Testing	PCR	By report	

Specimen/Stability	same as A-J Panel above
Collection Instructions	same as A-J Panel above
Clinical Utility	Detects 2 Mucolipidosis Type IV mutations (Del6.4kb, IVS3(-2)A>G) for carrier assessment in adult patients. These mutations are responsible for ~95% of mutant Mucolipidosis Type IV alleles in Ashkenazi Jewish Population.
Schedule/TAT	same as A-J Panel above
CPT Code	83891, 83892x2, 83896x2, 83901, 83912
Notes	same as A-J Panel above

5267 Mucolipidosis Type IV GenotypR™ - Diagnostic Testing

Component	Method	Reference Range	Units
Mucolipidosis Type IV Diagnostic Testing	PCR	By report	

Specimen/Stability	same as A-J Panel above
Collection Instructions	same as A-J Panel above
Clinical Utility	Detects 2 Mucolipidosis Type IV mutations (Del6.4kb, IVS3(-2)A>G) for support of diagnosis in a symptomatic patient. These mutations are responsible for ~95% of mutant Mucolipidosis Type IV alleles in Ashkenazi Jewish Population.
Schedule/TAT	same as A-J Panel above
CPT Code	83891, 83892x2, 83896x2, 83901, 83912
Notes	Please use specialized AJ GenotypR™ Requisition Form. Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com

5268 Mucolipidosis Type IV GenotypR™ - Fetal Study reflex to MCC

Component	Method	Reference Range	Units
Mucolipidosis Type IV Fetal Study	PCR	By report	

Specimen/Stability	20 (10) mL Amniotic Fluid; Ambient 24 hr 5 (3) mL of Maternal Whole Blood EDTA; Ambient 3 days, Refrigerated 3 days
Collection Instructions	Amniotic Fluid: Ship immediately, do not refrigerate or freeze. Maternal Whole Blood must accompany amniotic fluid specimen.
Alternate Specimens	Cultured Amniocytes: Do not refrigerate or freeze. Ship flask at confluency, topped off with culture media. An internal code will be added at the laboratory for cell culture. For chorionic villus specimens, please contact Technical Services. Maternal blood is required for potential studies of maternal cell contamination (MCC) of the fetal DNA.
Clinical Utility	Detects 2 Mucolipidosis Type IV mutations (Del6.4kb, IVS3(-2)A>G) for prenatal diagnosis of an at-risk fetus. These mutations are responsible for ~95% of mutant Mucolipidosis Type IV alleles in Ashkenazi Jewish Population.
Schedule/TAT	same as A-J Panel above (when cell culture required, add ~2 weeks)
CPT Code	83891, 83892x2, 83896x2, 83901, 83912
Notes	Please use specialized AJ GenotypR™ Requisition Form. Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com. Maternal blood should accompany any prenatal specimen. If warranted, test will reflex to #5365 Maternal Cell Contamination for an additional fee and extended turnaround time.

5276 Niemann-Pick Disease GenotypR™ - Carrier Testing

Component	Method	Reference Range	Units
Niemann-Pick Disease Carrier Testing	PCR	By report	
Specimen/Stability	same as A-J Panel above		
Collection Instructions	same as A-J Panel above		
Clinical Utility	Detects 4 Niemann-Pick Disease mutations (L302P, 1bp del P330fs, R496L, DelR608) for carrier assessment in adult patients. These mutations are responsible for ~92% of mutant Niemann-Pick alleles in Ashkenazi Jewish Population.		
Schedule/TAT	same as A-J Panel above		
CPT Code	83891, 83892x2, 83896x4, 83901, 83912		
Notes	same as A-J Panel above		

5277 Niemann-Pick Disease GenotypR™ - Diagnostic Testing

Component	Method	Reference Range	Units
Niemann-Pick Disease Diagnostic Testing	PCR	By report	
Specimen/Stability	same as A-J Panel above		
Collection Instructions	same as A-J Panel above		
Clinical Utility	Detects 4 Niemann-Pick Disease mutations (L302P, 1bp del P330fs, R496L, DelR608) for support of diagnosis in a symptomatic patient. These mutations are responsible for ~92% of mutant Niemann-Pick alleles in Ashkenazi Jewish Population.		
Schedule/TAT	same as A-J Panel above		
CPT Code	83891, 83892x2, 83896x4, 83901, 83912		
Notes	Please use specialized AJ GenotypR™ Requisition Form. Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com		

5278 Niemann-Pick Disease GenotypR™ - Fetal Study reflex to MCC

Component	Method	Reference Range	Units
Niemann-Pick Disease Fetal Study	PCR	By report	
Specimen/Stability	20 (10) mL Amniotic Fluid; Ambient 24 hr 5 (3) mL of Maternal Whole Blood EDTA; Ambient 3 days, Refrigerated 3 days		
Collection Instructions	Amniotic Fluid: Ship immediately, do not refrigerate or freeze. Maternal Whole Blood must accompany amniotic fluid specimen.		
Alternate Specimens	Cultured Amniocytes: Do not refrigerate or freeze. Ship flask at confluency, topped off with culture media. An internal code will be added at the laboratory for cell culture. For chorionic villus specimens, please contact Technical Services. Maternal blood is required for potential studies of maternal cell contamination (MCC) of the fetal DNA.		
Clinical Utility	Detects 4 Niemann-Pick Disease mutations (L302P, 1bp del P330fs, R496L, DelR608) for prenatal diagnosis of an at-risk fetus.		
Schedule/TAT	same as A-J Panel above (when cell culture required, add ~2 weeks)		
CPT Code	83891, 83892x2, 83896x4, 83901, 83912		
Notes	Please use specialized AJ GenotypR™ Requisition Form. Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com. Maternal blood should accompany any prenatal specimen. If warranted, test will reflex to #5365 Maternal Cell Contamination for an additional fee and extended turnaround time.		

5286 Tay-Sachs Disease GenotypR™ - Carrier Testing

Component	Method	Reference Range	Units
Tay-Sachs Disease Carrier Testing	PCR	By report	
Specimen/Stability	same as A-J Panel above		
Collection Instructions	same as A-J Panel above		
Clinical Utility	Detects 5 Tay-Sachs Disease mutations (Del7.6kb, G269S, IVS9(+1)G>A, 1278insTATC, IVS 12(+1)G>C) and 2 benign variants (R247W, R249W) for carrier assessment in adult patients. These mutations are responsible for ~98% of mutant Tay-Sachs alleles in Ashkenazi Jewish Population.		
Schedule/TAT	same as A-J Panel above		
CPT Code	83891, 83892x2, 83896x7, 83901, 83912		
Notes	same as A-J Panel above		

5287 Tay-Sachs Disease GenotypR™ - Diagnostic Testing

Component	Method	Reference Range	Units
Tay-Sachs Disease Diagnostic Testing	PCR	By report	
Specimen/Stability	same as A-J Panel above		
Collection Instructions	same as A-J Panel above		
Clinical Utility	Detects 5 Tay-Sachs Disease mutations (Del7.6kb, G269S, IVS9(+1)G>A, 1278insTATC, IVS 12(+1)G>C) and 2 benign variants (R247W, R249W) for support of diagnosis in a symptomatic patient. These mutations are responsible for ~98% of mutant Tay-Sachs alleles in Ashkenazi Jewish Population.		
Schedule/TAT	same as A-J Panel above		
CPT Code	83891, 83892x2, 83896x7, 83901, 83912		
Notes	Please use specialized AJ GenotypR™ Requisition Form. Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com		

5288 Tay-Sachs Disease GenotypR™ - Fetal Study reflex to MCC

Component	Method	Reference Range	Units
Tay-Sachs Disease Fetal Study	PCR	By report	
Specimen/Stability	20 (10) mL Amniotic Fluid; Ambient 24 hr 5 (3) mL of Maternal Whole Blood EDTA; Ambient 3 days, Refrigerated 3 days		
Collection Instructions	Amniotic Fluid: Ship immediately, do not refrigerate or freeze. Maternal Whole Blood must accompany amniotic fluid specimen.		
Alternate Specimens	Cultured Amniocytes: Do not refrigerate or freeze. Ship flask at confluency, topped off with culture media. An internal code will be added at the laboratory for cell culture. For chorionic villus specimens, please contact Technical Services. Maternal blood is required for potential studies of maternal cell contamination (MCC) of the fetal DNA.		
Clinical Utility	Detects 5 Tay-Sachs Disease mutations (Del7.6kb, G269S, IVS9(+1)G>A, 1278insTATC, IVS 12(+1)G>C) and 2 benign variants (R247W, R249W) for prenatal diagnosis of an at-risk fetus. These mutations are responsible for ~98% of mutant Tay-Sachs alleles in Ashkenazi Jewish Population.		
Schedule/TAT	same as A-J Panel above (when cell culture required, add ~2 weeks)		
CPT Code	83891, 83892x2, 83896x7, 83901, 83912		
Notes	Please use specialized AJ GenotypR™ Requisition Form. Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com. Maternal blood should accompany any prenatal specimen. If warranted, test will reflex to #5365 Maternal Cell Contamination for an additional fee and extended turnaround time.		

Additional New Tests from *Specialty*

3266 Heat Shock Protein 70 (Hsp70) Autoantibodies (effective 11/01/05)

Component	Method	Reference Range	Units
Hsp 70 Autoantibodies	EIA	<1.2	Index
Specimen/Stability	1.0 (0.5) mL Serum Ambient 7 days; Refrigerated 14 days; Frozen 30 days		
Collection Instructions	Collect whole blood by standard venipuncture technique. Allow blood to fully clot and remove the serum from the clot promptly. Specimens collected in serum separation tubes (SST) should be centrifuged and serum removed from the gel separator into a different tube and shipped. Plasma samples will be rejected.		
Clinical Utility	Hsp70 autoantibodies have been found in patients with autoimmune inner ear diseases including Ménière' disease.		
Schedule	Wednesday		
Turnaround Time	1-8 days		
CPT Code	83520		

Immunohistochemistry Stain Update (effective immediately)

IHC203 MART-1

Specimen Req	Formalin-fixed, paraffin-embedded tissue; Ambient; unstained, positively charged slides (4) are also acceptable.
Clinical Utility	IHC stains are useful for identification of antigens present in paraffin-embedded tissues. Properly selected panels may aid in the identification of tumor type and subclassification. Additional markers may be useful in assessing proliferation (Ki67) or prognosis (e.g., bcl-2 or HER-2/*neu*) in selected tumor types.
Collection Notes	Shipping on cold pack is recommended during the warm weather months. Ship by overnight courier to *Specialty*.
Schedule	Monday-Sunday
Turnaround Time	1-4 days
CPT Code	88342 times the number of antibodies selected
Notes	Diagnostic code required for third party reimbursement. Please send copy of pathology report and other applicable tests.
Order Codes	**1857 IHC Stain & Diagnostic Interpretation: Pathologist Chooses 1-3 Stains** **1859 IHC Stain & Diagnostic Interpretation: Pathologist Chooses 1-6 Stains** **1854 IHC Stain & Interpretation: Client Chooses Stains** **1856 IHC Stain Only: Client Chooses Stains**

4562 MTHFR C677T/A1298C GenotypR™ (effective 11/01/05)

Component	Method	Reference Range	Units
MTHFR C677T/A1298C	Invader	By report	

Specimen/Stability	5.0 (3.0) mL EDTA Whole Blood (Lavender top) Ambient 72 hours; Refrigerated 72 hours
Collection Instructions	EDTA is preferred but ACD is also acceptable. Heparinized whole blood is not acceptable. Ambient is preferred, refrigerated is acceptable; frozen is not acceptable.
Clinical Utility	Two mutations in the MTHFR gene have been associated with an increased risk for neural tube defects (NTD) and cardiovascular disease (C677T and A1298C). Homozygosity for the C677T mutation or compound heterozygosity for C677T and A1298C is associated with reduced MTHFR activity. Decreased MTHFR activity leads to hyperhomocysteinemia and lowers plasma folate levels.
Schedule	Monday
Turnaround Time	3-10 days
CPT Code	83891, 83892x4, 83896x10, 83903x2, 83912
Notes	Informed consent is required for residents of New York. Consent form is available on our Web site: www.specialtylabs.com. See Thrombotic AssessR™ for Obstetric Complications (#4555) for evaluation of recurrent fetal loss.

7468 *Mycobacterium tuberculosis* Complex DNA DetectR™ (effective 10/18/05)

Component	Method	Reference Range	Units
***M. tuberculosis* Complex DNA**	RT-PCR	Not detected	

Specimen/Stability	2.0 (1.0) mL Sputum (Sterile tube) Ambient 24 hrs; Refrigerated 7 days
Collection Instructions	Collect sputum early in the morning from a deep, productive cough on 3 consecutive days. Place in sterile, leakproof container and ship ambient or on cold pack
Alternate Specimens	Whole Blood or Bone Marrow; Bronchoalveolar Lavage CSF, Culturette/Swab, Tissue, Fluid
Alternate Collection Instructions	Call Client Services 800-421-4449 for collection instructions on alternate specimens
Clinical Utility	A variety of sterile and non-sterile clinical specimen sources for direct detection of *M. tuberculosis* complex DNA can be tested. This PCR assay provides a lower limit of sensitivity at 400 cells/mL.
Schedule	Tuesday, Friday
Turnaround Time	2-5 days
CPT Code	87556
Notes	*M. tuberculosis* complex consists of the following: *M. tuberculosis, M. bovis , M. bovis* BCG, *M. africanum, M. microti, M. canettii*. This discrete group of organisms share a DNA homology of greater than 95%.

4266 N-Telopeptide Serum
(effective 11/01/05)

Component	Method	Reference Range	Units
N-Telopeptide Serum	EIA	Females 25-49 y 6.2 – 19.0	nm BCE
		Males 31-80 y 5.4 – 24.2	nm BCE

Specimen/Stability	1.0 (0.5) mL Serum Frozen 2 months
Collection Instructions	Collect whole blood by standard venipuncture technique. Allow blood to fully clot and remove the serum from the clot promptly. Specimens collected in serum separation tubes (SST) should be centrifuged and serum removed from the gel separator into a different tube and shipped. Specimens sent in SST tubes will be rejected. Plasma samples will also be rejected.
Clinical Utility	Serum N-Telopeptide (NTx) levels may be used in predicting skeletal response (bone mineral density) to antiresorptive therapy and in monitoring bone resorption changes following initiation of antiresorptive therapy. Prior to initiating antiresorptive therapy, a serum NTx level is used to determine the probability for a decrease in bone mineral density (BMD) after one year in postmenopausal women treated with hormonal antiresorptive therapy relative to those treated with calcium supplementation.
Schedule	Wednesday
Turnaround Time	1-8 days
CPT Code	82523
Notes	For urine specimens, please order #4266U; N-Telopeptides with Creatinine.

Test Changes

Allergen Name Changes effective immediately

F93	Allergen-Cacao IgE	F93	Allergen-Cacao (Chocolate) IgE
F93G	Allergen-Cacao IgG	F93G	Allergen-Cacao (Chocolate) IgG
E1	Allergen-Cat Dander IgE	E1	Allergen-Cat Epithelium/Dander IgE
E1G	Allergen-Cat Dander IgG	E1G	Allergen-Cat Epithelium/Dander IgG
G1	Allergen-Sweet Vernal IgE	G1	Allergen-Sweet Vernal Grass IgE
G13	Allergen-Velvet IgE	G13	Allergen-Velvet Grass IgE
G6	Allergen-Timothy IgE	G6	Allergen-Timothy Grass IgE
M6	Allergen-*Alternaria alternata* IgE	M6	Allergen-*Alternaria alternata/tenuis* IgE
M6G	Allergen-*Alternaria alternata* IgG	M6G	Allergen-*Alternaria alternata/tenuis* IgG
T6	Allergen-Mountain Juniper IgE	T6	Allergen-Mountain Juniper/Cedar IgE
W9	Allergen-English Plantain IgE	W9	Allergen-Plantain (English), Ribwort IgE

Allergen Test Code Changes effective 10/18/05

RF202G	Allergen-Cashew Nut IgG	F202G	Allergen-Cashew Nut IgG
RF203G	Allergen-Pistachio IgG	F203G	Allergen-Pistachio IgG
RF260G	Allergen-Broccoli IgG	F260G	Allergen-Broccoli IgG
RM207	Allergen-*Aspergillus niger* IgE	M207	Allergen-*Aspergillus niger* IgE
RM207G	Allergen-*Aspergillus niger* IgG	M207G	Allergen-*Aspergillus niger* IgG

Test Changes

Test Code	Effective Date	Test Name	Specific Change	Also Affected
5368	10/04/05	CF70™ Fetal Study	Name and Reflex CF70™ Fetal Study w/ Reflex to MCC A maternal blood specimen must be sent with the fetal sample so that MCC testing can be performed if a possibility of maternal cell contamination exists.	
5363	10/04/05	Fragile X Fetal Study	Name and Reflex Fragile X Fetal Study w/ Reflex to MCC A maternal blood specimen must be sent with the fetal sample so that MCC testing can be performed if a possibility of maternal cell contamination exists.	
7581	10/18/05	Herpes Simplex Virus DNA DetectR™	Alternate Specimen 5 (1.0) mL Whole Blood EDTA; Ambient is acceptable	
4859	Immediately	Copper Liver	Specimen Stability Tissue - Ambient 28 days, Refrigerated 28 days, Frozen 2 months Trace Metal Tissue - Ambient 28 days, Refrigerated 28 days, Frozen 2 months	

Test Code	Effective Date	Test Name	Specific Change	Also Affected
3536	Immediately	Iron Liver	Specimen Stability Tissue - Ambient 28 days, Refrigerated 28 days, Frozen 2 months Trace Metal Tissue - Ambient 28 days, Refrigerated 28 days, Frozen 2 months	
4866U	Immediately	Magnesium 24 hr Urine	Collection Instructions Collect a 24 hour urine specimen. Add 10 mL of 6N HCl at start of collection. Record total volume in mL on both the container and requisition.	
4866UR	Immediately	Magnesium Urine Random	Collection Instructions After voided midstream urine collection is complete, add 1 mL of 6N HCl per 100 mL urine. Mix the specimen and transfer a 10 mL aliquot of urine to a clean screw cap leakproof container. Refrigerated specimen is preferred. Ship within 24 hours of collection by overnight courier.	
1369	10/18/05	HLA A, B, C, DR DetectR™	CPT Codes The code for each molecular probe (83896) was not calculated consistently. Other CPT codes for the tests listed are correct. The appropriate numbers of probes used for each HLA test are listed below: HLA-A 83896x66 HLA-B 83896x28 HLA-C 83896x62 HLA-DR 83896x43 HLA A, B C 83896x156 HLA A, B, C, DR 83896x199	1376 HLA A DetectR™ 1377 HLA B DetectR™ 1378 HLA C DetectR™ 1379 HLA DR DetectR™ 1368 HLA A, B, C DetectR™

Test Discontinuations

The following test(s) are no longer routinely available from *Specialty*. Whenever possible, alternate tests are recommended. Please note that if a test is designated as a "replacement," contractual pricing will be copied from discontinued test to replacement test. Contractual pricing does not apply to alternate tests or sendout tests. Please contact Client Services or your Sales Representative if you have any questions.

Test Code	Test Name	Reason	Alternate or Replacement Tests
1697	FC Myeloma/Plasma Cell Disorder Panel (effective immediately)	Low volume.	No replacement identified to date
4559	MTHFR GenotypR™ (A1298C Mutation) (effective 11/01/05 as are tests below)	Offered as a panel - C677T/ A1298C	4562 MTHFR C677T/A1298C GenotypR™
4560	MTHFR GenotypR™ reflex to A1298C Mutation	Offered as a panel - C677T/A1298C	4562 MTHFR C677T/A1298C GenotypR™
4558	MTHFR GenotypR™ (C677T)	Offered as a panel - C677T/A1298C	4562 MTHFR C677T/A1298C GenotypR™

Referral Testing Changes

Discontinue Sendout S48673 and use #5206 Ashkenazi-Jewish GenotypR™ - Carrier Panel (effective 10/18/05) (includes common mutations for Bloom Syndrome, Canavan Disease, Familial Dysautonomia, Fanconi Anemia GrpC, Gaucher Disease, Mucolipidosis Type IV, Niemann-Pick Disease, Tay-Sachs Disease)

Discontinue Sendout S49197 and use #5216 Bloom Syndrome GenotypR™ - Carrier Testing (effective 11/01/05)
Discontinue Sendout S41345 and use #5256 Gaucher Disease GenotypR™ - Carrier Testing (effective 11/01/05)
Discontinue Sendout S49198 and use #5246 Fanconi Anemia GrpC GenotypR™ -Carrier Testing (effective 11/01/05)
Discontinue Sendout S48728 and use #RT600 Allergen-Hackberry IgE (effective 10/10/05)
Discontinue Sendout S46825 and use #3266 Heat Shock Protein 70 (Hsp 70) Autoantibodies (effective 11/15/05)
Discontinue Sendout S50918 and use #4266 N-Telopeptide, Serum (effective 11/15/05)
Discontinue Sendout S49574 and use #7468 *M. tuberculosis* Complex DNA DetectR™ (effective 11/15/05)

Please visit our website at www.specialtylabs.com or call Client Services at 800-421-4449 for more information.