






ETHNICITY-BASED GENETIC DISEASE SCREENING FOR INFERTILITY SPECIALISTS

LabCorp Test No.	Test Groups/Names	Clinical Guidelines	Specimen Requirements
Caucasian Descent Test Group			1 yellow-top (ACD) tube, 8.5 mL 1 lavender-top (EDTA) tube, 4.0 mL
450020	CFplus® (Cystic Fibrosis 97-mutation Profile)	ACOG ¹ , ASRM ²	
450010	Spinal Muscular Atrophy (SMA) Carrier Testing	ACOG ^{3,4} , ASRM ² with patient history or by request	
510234	Fragile X Syndrome, PCR With Reflex to Southern Blot		
African, Asian, Hispanic, Mediterranean, and Mixed Descent Test Group			1 yellow-top (ACD) tube, 8.5 mL 3 lavender-top (EDTA) tubes, 4.0 mL each 1 serum gel-barrier (SST) tube, 0.8 mL serum
450020	CFplus® (Cystic Fibrosis 97-mutation Profile)	ACOG ¹ , ASRM ²	
450010	Spinal Muscular Atrophy (SMA) Carrier Testing	ACOG ^{3,4} , ASRM ² with patient history or by request	
510234	Fragile X Syndrome, PCR With Reflex to Southern Blot		
451363	Hemoglobinopathy Profile With Reflex to Alpha-Thalassemia <ul style="list-style-type: none">Complete Blood Count (CBC) Without DifferentialHemoglobinopathy Fractionation by HPLCFerritin, Serum Reflex to Alpha-Thalassemia DNA (Low MCV & normal ferritin)	ACOG ⁵ , ASRM ²	
Jewish Descent (per ACOG guidelines) Test Group			2 yellow-top (ACD) tubes, 8.5 mL each 2 lavender-top (EDTA) tubes, 4.0 mL each
450020	CFplus® (Cystic Fibrosis 97-mutation Profile)	ACOG ¹ , ASRM ²	
450010	Spinal Muscular Atrophy (SMA) Carrier Testing	ACOG ^{3,4} , ASRM ² with patient history or by request	
510234	Fragile X Syndrome, PCR With Reflex to Southern Blot		
343649	Jewish Descent ACOG (3) Add-on Profile <ul style="list-style-type: none">Tay-Sachs Disease, Biochemical, Leukocytes*Canavan Disease, DNA AnalysisFamilial Dysautonomia, DNA Analysis	ACOG ⁶ , ASRM ²	
Jewish Descent Expanded (per ACMG guidelines) Test Group			2 yellow-top (ACD) tubes, 8.5 mL each 2 lavender-top (EDTA) tubes, 4.0 mL each
450020	CFplus® (Cystic Fibrosis 97-mutation Profile)	ACOG ¹ , ASRM ²	
450010	Spinal Muscular Atrophy (SMA) Carrier Testing	ACOG ^{3,4} , ASRM ² with patient history or by request	
510234	Fragile X Syndrome, PCR With Reflex to Southern Blot		
343649	Jewish Descent ACOG (3) Add-on Profile <ul style="list-style-type: none">Tay-Sachs Disease, Biochemical, Leukocytes*Canavan Disease, DNA AnalysisFamilial Dysautonomia, DNA Analysis	ACOG ⁶ , ASRM ²	
339845	Jewish Descent ACMG Expanded (5) Add-on Profile <ul style="list-style-type: none">Bloom Syndrome, DNA AnalysisFanconi Anemia, DNA AnalysisGaucher Disease, DNA AnalysisMucopolysaccharidosis IV MutationNiemann-Pick Disease, DNA Analysis	ACMG ⁷	
Patients with Recurrent Miscarriage			1 green-top (sodium heparin) tube, 4.0 mL
052019	Chromosome Analysis, Blood (Constitutional)	ACOG ⁸	

Note: Additional genetic tests are available for patients with a specific family history of disease or those who request expanded testing.

* Specimens for Tay-Sachs Disease by Enzyme Analysis in Leukocytes must be refrigerated and should be collected and shipped Monday-Thursday only.

For full test information, including CPT codes and specimen collection requirements, visit the online Test Menu at www.LabCorp.com or contact your local account representative.

References

- American College of Obstetricians and Gynecologists. Update on carrier screening for cystic fibrosis. ACOG Committee Opinion No 325. *Obstet Gynecol.* 2005;106:1465-1468.
- Practice Committee of American Society for Reproductive Medicine; Practice Committee of Society for Assisted Reproductive Technology. 2008 Guidelines for gamete and embryo donation: a Practice Committee report. *Fertil Steril.* 2008;90(suppl 5):S30-S44.
- American College of Obstetricians and Gynecologists. Spinal muscular atrophy. ACOG Committee Opinion No 432. *Obstet Gynecol.* 2009;113:1194-1196.
- American College of Obstetricians and Gynecologists. Carrier screening for fragile X syndrome. ACOG Committee Opinion No 469. *Obstet Gynecol.* 2009;116:1008-1010.
- American College of Obstetricians and Gynecologists. Hemoglobinopathies in pregnancy. ACOG Practice Bulletin No 78. *Obstet Gynecol.* 2007;109:229-237.
- American College of Obstetricians and Gynecologists. Preconception and prenatal carrier screening for genetic disease in individuals of Eastern European Jewish descent. ACOG Committee Opinion No 442. *Obstet Gynecol.* 2009;114:950-953.
- Gross SJ, Pletcher BA, Monaghan KG. ACMG Practice Guidelines. Carrier screening in individuals of Ashkenazi Jewish descent. *Genet Med.* 2008;10:54-56.
- American College of Obstetricians and Gynecologists. Management of recurrent early pregnancy loss. ACOG Practice Bulletin No 24. *Int J Gynecol Obstet.* 2002;78:179-190.