

# ETHNICITY-BASED GENETIC DISEASE SCREENING FOR INFERTILITY SPECIALISTS

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

**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](http://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.