

**Services Offered By
GeneCare Medical Genetics Center**

Lab Testing

- ? Chromosome Analyses
 - Amniotic Fluid - 7 Day Avg Turnaround
 - Routine Bloods - 6 Day Avg Turnaround
 - STAT Bloods - 2 Day Avg Turnaround
 - Bone Marrow - 4 Day Avg Turnaround
 - Abortus or Skin - 8 Day Avg Turnaround
- ? Biochemical Analyses
- ? MSAFP/freeBeta Screening with ScreenSpot Plus (13wk 4d - 22wk 3d)
- ? freeBeta/PAPP-A Screening with ScreenSpot Plus (9wk 0d - 13wk 3d)
- ? DNA Analyses
- ? Paternity DNA

Genetic Counseling

- ? Amniocentesis
- ? Targeted Fetal Ultrasound
- ? AFP Screening Counseling and Testing
- ? Prior Reproductive Losses
- ? Family History of Genetic Disorders
- ? Drug and/or Alcohol Abuse
- ? Exposure to Toxic Materials or Infections

Service Highlights

- ? Early Amniocentesis Offered at 9 -14 Weeks
- LMP Timely, Courteous Patient Scheduling of Counseling, Amniocentesis and Targeted Ultrasound, As Appropriate
- ? ABMG Medical Geneticists Available for Consult 365 Days/Year
- ? Fast, Accurate Reports Telephoned, Mailed and Faxed
- ? Supplies for Simple, Safe Overnight Transport of Samples from Anywhere in the U.S.
- ? HMO, Physician, University, Hospital and Campus
- ? Private Referral Center

Research and Continuing Education

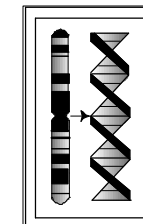
- ? Educational Materials for Physicians and Patients
- ? Genetic Seminars for Physicians, Nurses, Practices and Hospitals

Fetal Hemolytic Disorders

Rh *Kidd*
Duffy *MN*
Kell *Platelet**

*(Neonatal Alloimmune Thrombocytopenia Purpura)

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GeneCare®
Medical Genetics Center

www.genecare.com

Hemolytic disease of the fetus (unborn baby) and newborn occurs when the maternal immune system is sensitized to the baby's red blood cells. The surfaces of the red blood cells are covered with antigens, categorized as Rh, Kell, ABO, Duffy, Kidd, platelet, and MN antigens. When the fetus's antigens are different from those of the mother, the mother's immune system recognizes the baby's red blood cells as foreign and becomes sensitized to them. In a subsequent pregnancy, there is risk for hemolytic disease. If the fetus carries antigens different from the mother's, the mother's immune system responds by destroying the baby's red blood cells, a process known as hemolysis. This results in anemia, and if left untreated, death.

Who Should Be Tested?

Pregnant sensitized women may elect DNA analysis of amniocytes (baby's cells) to determine the fetal Rh, Kell, ABO, Duffy, Kidd, platelet, or MN type. DNA analysis can determine what antigens compose the fetus's red blood cells. Knowledge of the fetal antigen type will assist the physician in obstetric management. A negative test result, meaning that the fetus's cells are not in conflict with the mother's cells, allows discontinuation of serial amniocentesis and fetal blood sampling.

How Is Testing Performed?

Amniotic fluid cells are retrieved through a procedure known as amniocentesis. Amniocentesis involves removal of a small amount of the amniotic fluid that surrounds the baby using a fine needle inserted through the mother's abdomen. Fetal cells (amniocytes) float within the amniotic fluid. Once the amniocytes are retrieved, PCR (polymerase chain reaction) analysis is performed on the baby's DNA to determine what antigens compose the baby's red blood cells. To analyze for Kell, Duffy, Kidd, MN antigens and platelets, parents' bloods must also be tested.

How Soon Can This Test Be Completed?

The analysis is completed in about 1 week.

Specimen Collection and Transport

- ‡ Call GeneCare at 1-800-277-4363 to discuss clinical indications, current testing, and informed consent.
- ‡ Have patient complete Consent Form.
- ‡ Complete Laboratory Request Form for Special DNA tests.
- ‡ Label each specimen tube with patient's name, birth date and collection date.
- ‡ Specimen collection: For prenatal chromosome analysis, amniotic fluid AFP, and Rh, ABO, Duffy, Kell, Kidd, MN, or platelets DNA analysis, collect 30-40cc of amniotic fluid. For ABO, Duffy, Kell, MN, Kidd or platelets, also draw 10ml EDTA (purple top) tube on each parent. The blood draw must be performed Monday through Wednesday. Specimen must be received by GeneCare no later than Thursday. The test must be performed within 72 hours.
- ‡ **Ship at room temperature in our styrofoam kit to:**

**GeneCare Medical Genetics Center
201 Sage Road, Suite 300
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- ‡ **DHL/Airborne (800) 247-2676) Priority Overnight** (or by our local courier). Delivery required within 24 hours. For Friday shipments, mark **Saturday Priority Overnight** delivery.
- ‡ Notify GeneCare of shipment date and DHL/Airborne airbill/tracking number.

What Is The Cost?

Please call (866) 485-3336 to discuss fee and billing. Transport is paid by GeneCare.

References

Leggat HM, Gibson JM, et.al. Anti-Kell in pregnancy. Brit J of Obstet and Gyn, 1991; 98: 162-165.

Corfied VA, Moolman JC, Brink PA. Polymerase chain reaction-based detection of MN blood group-specific sequences in the human genome. Transfusion, 1993; 33: 119-124.

Bennett, P., Le Van Kim, et.al. Prenatal determination of fetal Rh type by DNA amplification. N Engl J Med 1993; 329:607-610.

Luban, N. Editorial: The new and the old - molecular diagnostics and hemolytic disease of the newborn. N. Engl J Med 1993; 329:658-660.

Spence, W., Maddalena, A., et.al. Molecular analysis of the Rh genotype in fetuses at risk for Rh hemolytic disease. Obstet Gynecol 1995; 85:296-298.

Lee, S., Wu, X., et.al. Molecular Basis of the Kell (K₁) Phenotype. Blood 1995; 85:912-916.

Sagot P, Bonneville F, et.al. Management of Platelet and RhD Maternal Immunizations by PCR Phenotypings after Early Amniocentesis. Fetal Diagn Ther, 1995; 10:373-380.

Goodrick MJ, Hadley AG, Poole G. Haemolytic disease of the fetus and newborn due to anti-FY(a) and the potential clinical value of Duffy genotyping in pregnancies at risk. Transfus Med 1997; 7(4): 301-4

Duerbeck NB, Chaffin DG, Coney P. Platelet and Hemorrhagic Disorders Associated with Pregnancy: A Review. Part I and II. Obstet and Gyn, 1997; 52(9) 575-595.

Rose NC, Hurwitz C, Silberstein LE, Andovalu R, Stoerker J. Prenatal analysis of rehesus CcDEe blood groups by heteroduplex generator. Amer J Obstet Gynecol 1997;176:1084-9.

Pearson SL, Hessner MJ. A^{1,2}BO^{1,2} genotyping by multiplexed allele-specific PCR. Brit J of Haem, 1998. 100. 229-234.

Hessner MJ, Pircon RA, Johnson ST, Luhm RA. Prenatal genotyping of Jk^a and Jk^b of the human kidd blood group system by allele-specific polymerase chain reaction. Prenat. Diagn. 1998; 18: 1225-1231.