

Prenatal Fetal Aneuploidy and Genetic Screening

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Prenatal Genetic Test Ordering

For Denver/Boulder members, prenatal genetic screening tests are ordered through the maternal fetal medicine service.

All patients requesting prenatal chromosome screening or diagnostic testing, other than AFP4 or nuchal translucency plus serum screening, require pre-testing consultation with the Genetics department. We believe this provides the highest quality care to patients because it ensures:

1. Patients are aware of the benefits and potential risks (sensitivity, specificity, results of uncertain significance) of the test,
2. The correct test is ordered, and
3. The patient receives consistent follow up when results are available.

The patient should be instructed to call the Genetics department to discuss the testing options and schedule an appointment. Phone as well as office consultations are available. Additionally a referral to the Genetics department (prenatal not Adult) should be placed if you have questions or wish the counselors to call the patient.

For Southern Colorado and Northern Colorado members, genetic counseling is recommended but not required for NIPT and carrier testing described in this guideline.

Screening and testing for fetal aneuploidy

- The current standard prenatal screening test for a low-risk singleton pregnancy is AFP4 screening.
- The current standard prenatal screening test in women carrying twins is first trimester screening, via serum plus nuchal translucency testing.
- The current standard prenatal screening test for women at high risk for carrying a fetus with a chromosome aneuploidy is cell free fetal DNA screening (aka NIPT, non-invasive prenatal testing).
 - This test is not currently recommended for high risk twin pregnancies.
 - The test will identify increased or decreased amounts of material from chromosomes 21, 18,13, X and Y.
 - The Down syndrome detection rate is 99%, the trisomy 18 detection rate is 98%, and the screen detected 8 out of 10 cases with trisomy 13.
 - The screen can optionally test the sex chromosomes with an approximate 99% detection rate.
 - The Harmony screen does not evaluate the chance for other chromosome abnormalities.

The indications for this test:

- Advanced maternal age (age over 35 at time of delivery)
- Fetal ultrasound abnormality suggestive of trisomy 21, 13, 18 or monosomy X
- Previous pregnancy with trisomy 13, 18, 21 or sex chromosome
- Positive serum screening test.

Diagnostic testing for fetal aneuploidy

- The standard diagnostic testing options are CVS and amniocentesis.
 - Chorionic villus sampling (CVS) will be offered to patients between 11.0 weeks EGA through 13 weeks EGA. This procedure can be performed either through the transabdominal or transcervical route. The estimated risk for pregnancy loss associated with this procedure is approximately 1:200.
 - Amniocentesis is generally performed after the completion of 15 weeks EGA. The loss rate associated with this procedure is estimated to be approximately 1:350.
- All pregnancies with a structural abnormality identified by ultrasound, including chromosome soft markers, should be referred to the Genetics department for follow up. If you wish the genetic counselors will notify the patient of the ultrasound result, discuss appropriate follow-up, and schedule appointments.

The above mentioned chromosome screening and diagnostic testing options as well as carrier screening for genetic diseases are available to all pregnant women; however, these tests may not be a covered benefit for low risk pregnancies. The Genetics department will help determine risk, and explain the self-pay process if indicated.

Screening and Testing for Single Gene Disorders

- Testing for genetic diseases is determined through risk assessment by the genetic counseling service. Clinicians caring for patients whose fetus is at increased risk for carrying a gene mutation, i.e. patient

and/or partner who are affected by a condition such as muscular dystrophy or congenital heart disease should have their charts forwarded to the genetic counseling service.

- Cystic fibrosis carrier screening is available for all patients.
- If the patient and/or her partner have any Ashkenazi Jewish ancestry, screening for Canavan disease, Cystic Fibrosis, Familial Dysautonomia, Tay Sachs, Bloom Syndrome, Fanconi Anemia, Gaucher disease, Glycogen Storage Disease, Maple Syrup Urine Disease, Mucopolysaccharidosis and Niemann Pick Disease is available, and is now a covered benefit for the patient.
- If the patient or her partner has any Black/African American ancestry hemoglobin electrophoresis should be ordered to screen for sickle cell anemia trait on the patient.
- If the patient herself is Southeast Asian (Cambodian, Thai, Laotian, Vietnamese, Hmong) hemoglobin electrophoresis should be ordered to screen for hemoglobinopathies.
- Screening for less common genetic syndromes is available and determined through risk assessment by the Genetics department.

Indications for prenatal Microarray Analysis

- When genetics and MFM determine that it is appropriate, standard karyotyping or FISH with reflex to microarray may be ordered on amniocytes.
- See the [Guideline for Performance of Microarray Testing](#) for more information.

Postnatal testing following fetal demise for underlying karyotype/gene disorders

- Following fetal demise after the first trimester or termination of pregnancy with unexplained anomalies chromosome analysis is recommended. If normal karyotype or chromosome analysis fails then SNP microarray analysis may be recommended in more complicated cases. Amniotic fluid or placenta biopsy are the recommended specimen to obtain for testing.

Member Handouts

- [Screening Options for Chromosome Disorders and Birth Defects](#)

Disclaimer

This guideline is informational only. It is not intended or designed as a substitute for the reasonable exercise of independent clinical judgment by practitioners, considering each patient's needs on an individual basis.

Guideline recommendations apply to populations of patients. Clinical judgment is necessary to design treatment plans for individual patients.