MATERNAL GENETIC CARRIER TESTING

The physicians at Haugen OB/GYN are offering carrier screening for genetic diseases in women (and their partners) who are thinking about starting a family or are already pregnant. This test can screen for many genetically inherited conditions. Individuals may not have a family history of a genetic disease but they can still be a carrier of a genetic condition that can be passed on to their children. Whether or not they have an affected child depends on the genetics of the father of the child as well.

Some of the tests included are cystic fibrosis, tay-sachs disease, sickle cell disease, and spinal muscular atrophy (SMA). Certain ethnicities have a higher likelihood of being a carrier of some of these conditions. This test can also be useful to couples undergoing in vitro fertilization (IVF) to see who might benefit from preimplantation genetic diagnosis (PGD). The physicians at Haugen OB/GYN can discuss this with you in greater detail. For those individuals interested in testing, we are currently recommending the ACOG (American College of Obstetrics and Gynecology)/ACMG (American College of Medical Genetics) panel. "Tandom testing" of the partner/spouse can also be coordinated to provide you with the most detailed information possible.

PRENATAL TESTING SUMMARY
ALL PRENATAL TESTING IS OPTIONAL

All couples have a 3-4% chance of having a baby with a birth defect. Your risk may be less or more than this depending on many factors including age, medical conditions, medication use, and family history. These tests are looking for problems with your baby. Prenatal testing for certain disorders is optional for all couples. There are screening tests and diagnostic tests to give you more information about your specific risk in this pregnancy. Each test has benefits and risks and the options can be confusing. Prenatal testing is an area of medicine that is changing rapidly and there are new variations of tests coming out frequently. There is no test available that can identify every possible birth defect or medical condition of a baby. There is no guarantee despite any testing, that any condition can be diagnosed or that you will have a healthy baby. To follow is a brief summary of conditions that can be tested for and the types of testing available.

Types of conditions tested for.
Neural Tube Defects: this category of birth defects comprises problems with how the brain and/or spinal cord of the fetus formed. These structures develop from the neural tube of the embryo. If this tube does not close appropriately (usually occurs 30 days after conception), this type of defect can occur. These problems range from spina bifida (an opening in the spine and spinal cord causing a range of disabilities) to anencephaly (an absence of the brain). The risk of having this condition is 1/500 and does not change with maternal age.

Chromosomal abnormalities: Chromosomes are structures in every cell that carry genetic information. Normally a person has 23 pairs of chromosomes or 46 total. If a pregnancy gets too many or not enough chromosomes, problems arise. Usually these pregnancies miscarry. If miscarriage does not occur, a baby may be born with mental and physical disabilities, sometimes incompatible with life. One of the most common chromosome abnormalities is Down Syndrome, or Trisomy 21, which is an extra chromosome 21. In this condition, there are 3 chromosome 21s instead of 2. Additional chromosome abnormalities that can be sometimes detected with prenatal testing are Trisomy 18 and Trisomy 13, these can be lethal complications resulting in fetal or neonatal death. The risk of having a baby with a chromosomal abnormality increases with maternal age. However, because younger women have more babies, 80% of all chromosomal abnormalities are found in mothers less than 35 years of age. Please see the attached table at the end of this information with your age related risk.

OPTIONAL SCREENING TESTS

These tests DO NOT diagnose or prove there is a problem. They can simply inform you of your risk of having a baby with a certain disorder. There are false positive and false negatives with these tests, so even normal tests do not guarantee the birth of a healthy baby. If you have abnormal results from a screening test, one of the diagnostic tests would be offered to give you further information.

Test	*When Done*	*Type of Test*	*What can this test screen for?*	*Benefits*	*Risks*	*Special notes about this test*
First trimester screening	11-13 weeks. Results in 7-10 days.	Ultrasound and maternal blood test	Down syndrome (Trisomy 21) Trisomy 18 Nuchal translucency (fluid area behind the neck)	Detects 82-87% of babies with Down Syndrome.	5% false positive rate. More false positives with increasing maternal age. If the ultrasound measurement is abnormal, but the blood results are normal, there is still an increased risk for fetal heart defects (40% risk).	This test does not assess for other birth defects. Optional neural tube defect screening may be offered at 16 weeks. Anatomy ultrasound is recommended for all patients.
Cell-free DNA Screening (NIPT)	10+ weeks. Results in 7-10 days.	Maternal blood test	Down syndrome Trisomy 18 Trisomy 13 Sex chromosomes	Detects 99% of babies with Down Syndrome and Trisomy 18 Detects 91% of babies with Trisomy 13 Detects 96%of problems with sex chromosomes 99% accuracy of baby's sex	There can be false positives with this test. Rates vary based on age. There is a <1% risk of inconclusive results (no result is able to be given). This test is validated in women age 35 and older though it is likely that it performs as well in women under age 35.	This test does not assess for other birth defects. Optional neural tube defect screening may be offered at 16 weeks. Anatomy ultrasound is recommended for all patients. **Insurance coverage varies for this test. You MUST check Insurance coverage prior to having this test done.**
Quad Screen	15-22 weeks. Results in 7-10 days.	Maternal blood test	Down syndrome Trisomy 18 Neural tube defect	Detects 80-85% of babies with Down Syndrome. Detects 90% of babies with Trisomy 18	5% false positive rate. There are more false positives with increasing maternal age.	This test does not assess for other birth defects. Anatomy ultrasound is recommended for all patients.
AFP Screen	15-22 weeks. Results in 7-10 days.	Maternal blood test	Neural tube defects. Abdominal wall defects.	Detects 65-90% of neural tube defects.	1-3% false positive rate.	An elevated value may represent conditions other than neural tube defects. Specialized anatomy ultrasound and MFM consultation may be recommended.

ANATOMY ULTRASOUND IS RECOMMENDED FOR ALL PATIENTS

This is performed between 19-21 weeks. This ultrasound assesses for birth defects, placental concerns, and screens the cervical length. It detects 50% of babies with Down Syndrome. There is a 10-15% false positive rate. For example, there is risk of finding "soft markers" on ultrasound which can be associated with a chromosomal problem but usually are not. A normal ultrasound does NOT rule out all birth defects. Patients with high risk pregnancies may be recommended to undergo their anatomy ultrasound with our maternal fetal medicine consultants. This is for level II. This recommendation is made on an individual basis.

OPTIONAL DIAGNOSTIC TESTS:

These tests are more accurate at finding a fetal problem but they are looking for specific problems. Even a normal result of one of these tests does not guarantee a healthy baby. The trade off with improved accuracy of these tests is having a risk to the fetus. Some women do these diagnostic tests after abnormal screening test results and some women do diagnostic testing without previous screening tests. These tests are performed by a high risk obstetric specialist and we will refer you if you desire one of these tests.

Test	*When Done*	*Type of Test*	*Benefits*	*Risks*
Chorionic Villus Sampling (CVS)	10-13 weeks. Results in 7-14 days.	Ultrasound and biopsy of placenta	99.9% accurate at diagnosing fetal chromosome disorders. Can diagnose single gene disorders if known family risk.	1/450 miscarriage rate. Can find placental chromosome problems which are not found in the fetus (3% risk).
Amniocentesis	15-20+ weeks. Results in 7-14 days.	Ultrasound with sample of amniotic fluid around fetus via needle	99.4% accurate at diagnosing fetal chromosome problems. Increases detection of neural tube defects. Can diagnose single gene disorders if known family risk.	1/900 miscarriage rate

SUMMARY

There are risks and benefits to these tests and it is an individual decision for each family whether to do any of these tests and which ones. You do not need to do any screening or diagnostic testing. The benefits of any prenatal testing include reassurance, or in the event of a problem, preparation, optimal medical management, or termination of the pregnancy. The risks including additional worrying if you have abnormal screening tests but don't do diagnostic testing and the miscarriage risks associated with diagnostic testing. This risk of not doing any testing is not knowing about a birth defect, or a higher risk of one, before delivery.

Any ultrasound requires a specific ultrasound appointment with one of our ultrasonographers. No ultrasound is done as part of any routine prenatal visit. Please ensure that you have your appointments scheduled appropriately. Our front desk staff would be happy to confirm your appointments.

***Please check on your insurance coverage before deciding to do any test. In addition, some insurance only pays for a certain number of ultrasounds so if you decide to do first trimester screening, you may find that later ultrasounds are not paid for. It is YOUR responsibility to know your insurance coverage and pay any costs over and above that coverage.*

RISK OF CHROMOSOMAL ABNORMALITIES IN LIVEBORNS

Maternal age at delivery	*Trisomy 21 (Down Syndrome)*	*All chromosome abnormalities*
20	1/1667	1/526
21	**1/1667**	**1/526**
22	**1/1429**	**1/500**
23	**1/1429**	**1/500**
24	**1/1250**	**1/476**
25	**1/1250**	**1/476**
26	**1/1176**	**1/476**
27	**1/1111**	**1/455**
28	**1/1053**	**1/435**
29	**1/1000**	**1/417**
30	**1/952**	**1/384**
31	**1/909**	**1/384**
32	**1/769**	**1/322**
33	**1/625**	**1/317**
34	**1/500**	**1/260**
35	**1/385**	**1/204**
36	**1/294**	**1/164**
37	**1/227**	**1/130**
38	**1/175**	**1/103**
39	**1/137**	**1/82**
40	**1/106**	**1/65**
41	**1/82**	**1/51**
42	**1/64**	**1/40**
43	**1/50**	**1/32**
44	**1/38**	**1/25**
45	**1/30**	**1/20**
46	**1/23**	**1/15**
47	**1/18**	**1/12**
48	**1/14**	**1/10**
49	1/11	1/7

47XXX excluded for ages 20-32 (data not avail). Data from Hook: Rates of chromosomal abnormalities at different maternal ages. Obstet Gynecol 58:282. 1981