GENETIC TESTING OPTIONS AND CONSENT FORM

The American College of Obstetricians and Gynecologists (ACOG) recommends that all pregnant women be offered options for genetic testing during pregnancy. There are two broad categories of genetic testing: screening and diagnostic.

1. **Screening tests** involve one or two blood samples from the expectant mother possibly combined with an abdominal ultrasound at 11-13 weeks gestation. All these tests "screen" or determine a woman's **risk** of carrying a fetus with a genetic abnormality. These tests do not give a yes or no answer, but rather a risk ratio. For example, the risk of your fetus having Down's Syndrome is 1 in 1000. Although low, this risk is not zero. In addition, all screening tests have a risk of both false positives (the test shows an increased risk of a fetal genetic abnormality and the fetus is normal) and false negatives (the test shows a very low risk of a fetal genetic abnormality and the fetus does indeed have a genetic abnormality). Any patient who has a screening test that reveals an increased risk of having a chromosomally abnormal fetus is offered additional testing, including diagnostic testing.
2. A **diagnostic test** is an invasive procedure to obtain cells that reflect the genetic make-up of your fetus. Depending on the test chosen, a needle is passed into the placenta or the intrauterine cavity (bag of water). There are two diagnostic tests: CVS (chorionic villus sampling) and amniocentesis. With each of these tests, the results are 99.9% accurate. Due to the invasive nature of these tests, there is a small risk of pregnancy loss associated with the procedures ranging from 1 in 450 to 1 in 900, depending on the procedure chosen and the experience of the provider performing the test.

Although there is a risk of chromosomal abnormalities with any pregnancy, as the age of the expectant mother increases, so does the risk of carrying a chromosomally abnormal fetus. As such, mothers who are 35 years of age or more at the time of delivery are generally offered both screening and diagnostic testing.

It is important to understand that every pregnancy has some risk (3-5%) for other kinds of birth defects that cannot be diagnosed during pregnancy, such as autism, non-specific intellectual disability, some genetic diseases, and some types of anatomic/physical birth defects.

Genetic testing can evaluate for several different chromosomal abnormalities including Down Syndrome (Trisomy 21), Trisomy 18, Trisomy 13, Turner's Syndrome, Klinefelter Syndrome, and others. Testing can also screen for open neural tube defects (ONTD). ONTD are abnormalities that arise from the lack of closure of the neural tube in early pregnancy (prior to 10 weeks pregnant). The neural tube develops into a baby's spinal cord and brain and therefore abnormalities can result in paralysis of the legs, lack of bladder/bowel control, club feet, a build-up of spinal fluid in the head or incomplete brain development. The most common ONTD is spina bifida. Depending on the test performed, various abnormalities are evaluated for and the rates of detection and risk to the baby vary (see chart).

	When Performed (Weeks Gestation) and How	Down Syndrome Detection Rate	Trisomy 18 Detection Rate	Trisomy 13 Detection Rate	ONTD Detection Rate	Risk to Baby (Fetal Loss Rate)	False Positive Rate	Can Reveal Gender or Sex of Baby
Screening Tests								
Noninvasive Prenatal Testing (NIPT) CPT code: 81420	Any Time After 10 Weeks (Blood test)	99%	98%	80-90%	Does Not Screen; Can be Done Separately (Via Blood)	0%	<1%	Yes
Integrated or Sequential CPT code: 84163	11-13 Weeks (Blood test plus ultrasound); 15-22 Weeks (Blood Test)	92-95%	90-92%	Screens Sometimes Based on Lab: 72-80%	80%	0%	3.5%	No
Quad/Penta Screen CPT codes: 82105 ,82677, 84702, 86336	15-22 Weeks (Blood test)	81%	80%	Does Not Screen	80%	0%	5%	No
Diagnostic Tests								
Amniocentesis	15-20 Weeks (Needle in Bag of Water)	>99%	>99%	>99%	95%	1 in 500 to 900	<1%	Yes
Chorionic Villus Sampling (CVS)	10-13 Weeks (Needle in Placenta)	>99%	>99%	>99%	Does Not Screen; Can be Done Separately (Via Blood)	1 in 450	<1%	Yes

General recommendations for women **under 35 years old** at the time of delivery with no history of a previously affected child are as follows:

- NIPT Testing after 10 weeks and ONTD testing after 15 weeks. NIPT screening may **not** be covered by your insurance carrier and may be an out-of-pocket expense
    - NIPT or diagnostic testing is recommended if integrated or sequential testing reveals an increased risk of a chromosomally abnormal fetus **OR**
- Integrated or sequential testing with an ultrasound and blood test between 11-13 weeks and another blood test between 15-20 weeks **OR**
- Quad or penta testing if prenatal care begins after 13 weeks (it will be too late for integrated/sequential testing) and you opt out of NIPT screening

General recommendations for women **35 years old or more** at the time of delivery or with a history of a fetus/child with a chromosomal abnormality/ONTD are as follows:

- NIPT after 10 weeks gestation and another blood test to screen for ONTD after 15 weeks
- May consider diagnostic testing, if desired

Every woman has the right to decline all genetic testing based on personal or religious convictions.

After a thorough discussion and working together my provider, I opt for the following genetic testing as I believe this best reflects the clinical evidence and my own values and perspective (shared decision-making):

___ NIPT (if not covered by insurance, cash price is $249 and includes fetal sex identification)

___ Integrated/Sequential Testing

___ Quad/Penta Testing

___ Amniocentesis by my Provider or a Perinatologist (High Risk Pregnancy Doctor)

___ Referral to a Perinatologist for Chorionic Villus Sampling (CVS)

___ No Genetic Testing

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Patient Name

_____ _____/_____/_____

Patient/Health Care Agent/Guardian/Relative Signature Date