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GENETIC SCREENING: INFORMED CONSENT/REFUSAL

There are many options for prenatal testing, that is, testing that parents can choose to find out more information about their baby, including estimated due date, baby's gender, and gathering information about baby's overall health. Some parents choose to "test for everything," others choose just the minimum, and some parents choose to decline all testing.

All pregnant women have a small chance (3-5%) of delivering a baby with a physical and/or mental birth defect. Most birth defects (65-75%) have an unknown cause, whereas others are genetic (15-20%) or caused by environmental exposures (10%). Optional testing is available during pregnancy to detect some genetic disorders in the baby, including certain chromosomal abnormalities (e.g., Down Syndrome) and neural tube defects (e.g., *spina bifida*). Families may choose to use this information to decide to terminate a pregnancy, or to become emotionally and financially prepared for a higher needs baby. Other parents, knowing they would do nothing different if they had the information, choose to forego all or most prenatal testing options.

Insurance coverage of the tests varies. Please call your insurance company to find out what is covered. *Established clients* can call Ingrid at Island Medical Billing to verify (**360-632-4435**).

Quad Screen/Sequential Screen:

What is the purpose of the Quad and Sequential screening tests?

These screening tests are intended to measure the chance that your baby has Down syndrome, trisomy 18, or neural tube defects. Before birth, all normal babies make a substance called alpha-fetoprotein or AFP. Small amounts of AFP pass into the amniotic fluid surrounding the baby. An even smaller amount of AFP crosses the placenta and passes into the mother's blood. The screen hopes to pick up any abnormalities in the baby by looking at this fluid in the mother's blood.

How is the Quad screening done?

The Quad screen is one blood test that is offered to the mother between 15-22 weeks of pregnancy. *Due to its high false positive rates, the Quad screening is rarely done by itself, but often combined with the Sequential screen.*

What does a negative result mean?

A negative result means that you have less than a 1:270 chance of having a baby with a condition. It is important to keep in mind that the incidence of these defects is very low (1:800 for Down syndrome and 1:3,000 for trisomy 18).

What does a positive result mean?

A positive result means that the chance of these three birth defects is higher and further testing could be considered. Keep in mind that most women (98%) with positive results have normal, healthy babies.

How is the Sequential screening done?

The Sequential screen is doing the Quad screen plus an additional earlier blood draw and an ultrasound. It is more accurate than the Quad screen alone. The first blood draw is done between 10-14 weeks, with the ultrasound performed between 11-14 weeks, followed by the Quad screen blood draw between 15-22 weeks.

What is the next step if I have a positive result?

If you decide to do the screen and the screen comes back positive, we will have a discussion about the results and help you decide if you want to follow up with a level II ultrasound and/or amniocentesis.

Panorama Screening

What is Panorama Screening?

Panorama is a DNA screening test that can tell you important information about your pregnancy. You can find out if your baby is at risk for having Down Syndrome or other chromosomal abnormalities. Panorama can also tell you the sex of your baby, if you choose to find out. Panorama has the lowest false positive rate of any prenatal screening test for the commonly screened chromosomal abnormalities: Trisomy 21, 18, or 13. And, Panorama can be done as early as nine weeks into your pregnancy using a simple blood draw. Panorama cannot detect neural tube defects. Some women, wanting the neural tube information, also do a 20-week ultrasound.

What kind of results will I get from the Panorama screening?

The reports sent to your midwife will have one of these results:
LOW-RISK RESULTS: A low-risk result means the chance that your baby has one of the chromosome conditions that was screened for is very low.
HIGH-RISK RESULTS: A high-risk result means there is an increased risk that your baby has that condition. Your midwife will talk to you about follow-up testing options.

How accurate is the Panorama screening?

Panorama is a screening test, not a diagnostic test. A positive test does not necessarily mean that your baby is affected with that condition. Although rare, false positives and false negatives may occur. Panorama has a false positive rate of 8.6% and a false negative rate of 0.7%. Follow up testing is recommended if the screen comes back positive.

Is there anything that can make the screening more accurate?

A cheek swab is offered to the known biological father of the baby; this can help gather more genetic information for the baby, but does not impact accuracy. It cannot determine paternity and should not be added to the test if there is any question about paternity.

Ultrasound Screening:

- **Early Ultrasound / "Dating" Ultrasound:** Helpful if your dates of last menstrual period or conception dates are vague/unknown. Performed between 7-10 weeks of pregnancy.
- **Nuchal Translucency (NT) ultrasound.** It is also used to determine the chances your baby has a chromosomal problem. This ultrasound is performed between 11-14 weeks of pregnancy.
- **Fetal Survey Ultrasound/"Anatomy Screen" or "20 week Ultrasound":** Uses sound waves to create an image of the baby. A wide variety of structural abnormalities, the presence of Neural Tube Defects (92-95%), and "soft signs" of Trisomy 21 can be identified. Additionally, ultrasound can provide information about the baby's sex, placenta, amniotic fluid, and the mother's anatomy. It is performed between 18-22 weeks of pregnancy.

INFORMED CONSENT/REFUSAL FOR GENETIC SCREENING:

I have read all of the above information and have had the opportunity to ask questions.
My choice(s) regarding genetic testing for this pregnancy is indicated below:

I ELECT (select one or more):

[ ] Quad Screen
[ ] Sequential Screen
[ ] Panorama Screen
[ ] Early Ultrasound ("Dating Ultrasound")
[ ] Nuchal Translucency Ultrasound
[ ] Fetal Survey Ultrasound ("Anatomy Screen" or "20 Week Ultrasound")
[ ] I DECLINE ALL GENETIC TESTING

_____ _____
Client Signature Date