



## Genetic Screening

**There are various tests available to you to determine genetic abnormalities. The purpose is to inform you if there is an increased risk and if other measures of care need to be performed. Please see the options below that we offer you in the office. Please initial next to the test you would like.**

### **1. Nuchal Translucency + First Trimester Screen**

- 82%-87% Detection rate for Trisomy 21 (Down's Syndrome)
- 5% false positivity rate
- Ultrasound for Nuchal should be done with blood work at same time followed by an alpha fetal protein test at 15+weeks
- Done between 11wk6d to 13wk6d
- Nuchal ultrasound alone is 70% for detection rate for Trisomy 21.

### **2. Quad Screen**

- 81% Detection rate for Trisomy 21
- 5% false positivity rate
- Blood work only if missed early detection blood work
- Done at 15+ weeks

### **3. Cell Free DNA testing**

- Most sensitive and specific screening
- Not diagnostic
- 99% Detection rate for Trisomy 21, 98% for Trisomy 18, and 99% for Trisomy 13.
- 2-4% false positivity rate
- Done at 10+ weeks
- Not all insurance cover and may have out of pocket cost
- Can tell you the gender of fetus

### **4. Amniocentesis**

- Diagnostic test
- Invasive test

**I \_\_\_\_\_, consent to have the above test(s) at Women's HealthFirst. I have read the above and understand Insurance may not cover the cost of the test.**

**X \_\_\_\_\_ Date \_\_\_\_\_**