Trisomy/Down's Syndrome Screening Tests

These tests are offered to all of our patients but it is your choice to have done and no testing is also an option. The purpose of genetic screening is to help identify those at risk for certain birth defects and chromosomal abnormalities. Knowing if your baby is at risk or has a particular condition would allow you to prepare for having a child with this disorder and organize the medical care and resources your child may need. To help you decide the best genetic screening test for you if desired we've made a comparison chart to help guide you. These are currently the best tests that science has to offer.

	1st Trimester Screen	Cell free fetal DNA	Penta
Screens for:	Down's syndrome, Trisomy 18	Down's syndrome, Trisomy 18, 13, Turner's/Kleinfelter's syndrome	Down's Syndrome, Trisomy 13/18, includes ms-afp
Involves:	Ultrasound + mom's blood test	Mom's blood test	Mom's blood test
Accuracy:	82-87% some false positive	>99% low false positive and negative	81% Some false positive
Timing:	12-14 weeks	10+ weeks	15-22 weeks
Insurance coverage:	Almost always covered	If mom >35 at delivery, almost always. If <35, Sometimes	Almost always
Codes for insurance:	Procedure code: 84704, 84163, 76813	Procedure code: 81420	Procedure code: 81511

Carrier Screening Tests

Below are Carrier Screenings we offer. They are performed by a mom's mouthwash swish in office and test to see if Mom carries the gene. They are >99% accurate and have an occasional false negative. They can be done at anytime in the pregnancy and if done in a prior pregnancy DO NOT need to be repeated. They are all usually covered by insurance but as always, as we cannot guarantee this, please check prior to your appointment as if not they can be expensive.

Cystic Fibrosis: A disease which causes problems with digestion and breathing. In most people it causes serious risk to health and has an average lifespan of 37. Mild forms have a lifespan in the 50s. 1 in 25 Caucasians are carriers (somewhat less common in other ethnicities) and 1 in 2,500 babies are born with CF

Spinal Muscular Atrophy: A disease of muscle wasting and weakness, can range from mild (normal lifespan, may need wheelchair at some point) to severe (death prior to 2 years old). 1 in 40-60 people are carriers and 1 in 6,000-10,000 babies are born with a form of this disease.

Fragile X: The most common form of inherited intellectual disability. Can also be a cause of autism. About 1:157 women have pre-mutation (more if family history of disability, less without) and 1 in 3,600 males are born with fragile X and 1 in 4,000-6,000 females.

	Cystic Fibrosis	Fragile X	Spinal Muscular Atrophy
Procedure Code	81222	81243	81400

Please check with your insurance company prior to your doctor's appointment for coverage so you are able to have them ordered at that time if you wish to proceed with any of the above testing. It testing if done through your insurance and not covered it can be very expensive.

Diganosis codes for any of above: All Use: Z36 1st Trimester- up to 14 weeks, 2nd is > 14 weeks

1st Trimester, 1st Pregnancy: Z34.01

2nd Trimester, 1st Pregnancy: Z34.02

1st Trimester, 2 + Pregnancy: Z34.81

2nd Trimester, 2+ Pregnancy: Z34.81

1St Trimester, 1st Pregnancy, 35+ at delivery: O 09.511

2nd Trimester, 1st Pregnancy, 35+ at delivery: O 09.512

1st Trimester, 2+ Pregnancy, 35+ at delivery: O 09.521

2nd Trimester, 2+ Pregnancy, 35+ at deliver: O 09.522

If your insurance company does not cover a test you are interested in the self pay costs are listed below. If you do choose to self pay please let us know so we do not put your insurance information on the order sheet.

Cell Free Fetal DNA: $150

Cystic Fibrosis (CF): $75

Spinal Muscular Atrophy: $35

Fragile X: $40