

CHARLES STREET OB/GYN
Genetic Testing Options

<i>Genetic Test</i>	What is this testing for?	What does the procedure involve?	What is the risk of miscarriage?	When is the test performed?	What is the detection Rate?	What do we recommend for you?
First Trimester Screen **	Down's Syndrome, Trisomy 18 & Trisomy 13	Sonogram (nuchal translucency) & maternal blood work	None	12-13 weeks	90%	
CVS ++	Down's Syndrome, Trisomy 18, Trisomy 13 & any chromosomal abnormality	Chorionic villi is obtained from the placental bed (either through the cervix or abdominally) using a small needle.	1/200	10-14 weeks	> 99%	
Quad Screen **	Down's Syndrome, Trisomy 18 & Neural Tube Defects	Maternal blood work	None	16-18 weeks	75% for Down's Syndrome 75-90% for NTD	
MSAFP **	Neural Tube Defects	Maternal blood work	None	16-18 weeks	75-90%	
Amniocentesis ++	Down's Syndrome, Trisomy 18, Trisomy 13, Neural Tube Defects & any chromosomal abnormality	Amniotic fluid is obtained (through the abdomen) using a small needle.	1/300-1/500	16-18 weeks	> 99%	

** These are screening tests – they only provide an estimate of the risk for a condition in the pregnancy.

++ These are diagnostic tests (CVS and Amnio) – they provide a definitive answer about chromosome abnormalities.