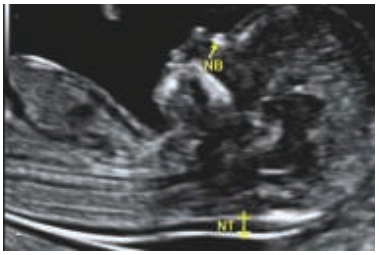
CARNEGIE IMAGING
The Leading Edge in Women's Ultrasound

Nuchal Translucency and Nasal Bone Assessments

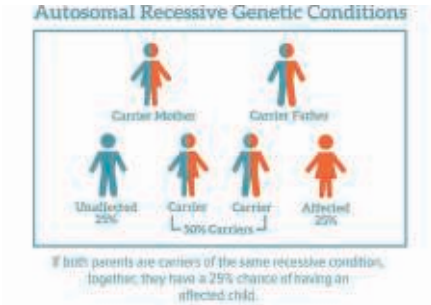
Measurement of the Nuchal Translucency (NT) is part of a first trimester risk assessment for non-inherited genetic conditions (eg. trisomy 21, 13,18). The ultrasound can also be used to detect birth defects.



www.mountsinai.org/health-library/tests/nuchal-translucency-test

Carrier Screening

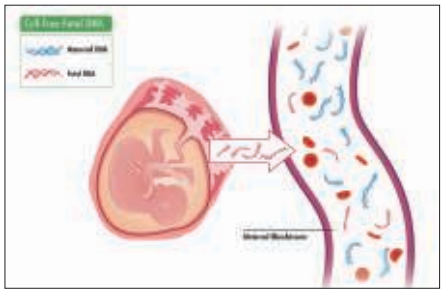
Carrier screening is a test that analyzes parental DNA to see if they are carriers of genetic variants that could cause an inherited disease in their children.



www.sema4.com/products/expandedcarrierscreen/patients/

Non-Invasive Prenatal Screening/Testing (NIPS/NIPT)

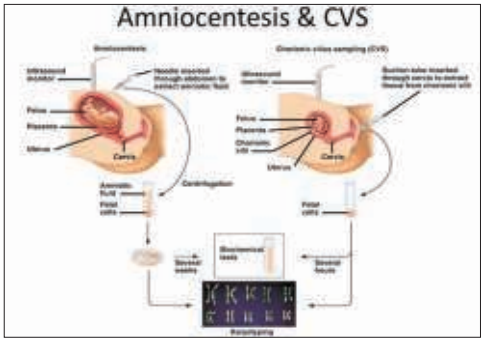
NIPT/NIPS determines risk that the fetus will be born with certain usually non-inherited genetic abnormalities (trisomy 21, 13, 18). Testing analyzes small fragments of placental DNA that are circulating in maternal blood. NIPS/NIPT also tests the presence, absence, and total number of X and Y chromosomes. Babies can be born with a change in the total number of sex chromosomes, such as only one X (X), an extra X (XXX or XXY), or an extra Y (XYY). Microdeletions affect pregnancies equally, regardless of maternal age, and they occur when a small piece of a chromosome is lost. Common microdeletions include DiGeorge syndrome, Angelman syndrome, Prader-Willi and Cri-du-chat syndrome.



www.natera.com/womens-health/panorama-nipt-prenatal-screening/

Invasive Testing

The only way to diagnose Down syndrome (trisomy 21) or other chromosomal abnormalities is by having an invasive diagnostic test - either a chorionic villus sampling (CVS) or an amniocentesis -and testing the placental or fetal cells. When CVS or amniocentesis are performed by a skilled operator, the risk of subsequent miscarriage is very low. (Salomon LJ, et al. UOG 2019) Microarray using array comparative genomic hybridization with single nucleotide polymorphism (array CGH and SNP) testing is a comprehensive analysis to examine key locations across the entire genome for deletions and duplications that may be clinically significant and can ONLY be obtained through invasive testing.



www.cdn1.sema4.com/wp-content/uploads/Chromosomal-Microarray-and-Microdeletion-FISH-Panel_062017.pdf

Healthful Woman: www.healthfulwoman.com

Womens Health Podcast for Pregnancy, Fertility, Gynecology & Nutrition

"Fragile X Screening" – with Dr. Tamar Goldwaser; Published on: April 19, 2021
"Invasive Testing: CVS and Amniocentesis" – with Dr. Andrei Rebarber; Published on: July 9, 2020
"We're All Mutated: Carrier Screening" – with Dr. Tamar Goldwaser; Published on: July 6, 2020
"Looking into our Baby's Genes: Aneuploidy Screening and Testing" – with Dr. Tamar Goldwaser; Published on: July 2, 2020