Prenatal Screening Tests

	First Trimester Screening	Quad Marker Screen	Chorionic Villus Sampling (CVS)	Amniocentesis	Second Trimester Ultrasound (Sonogram)
Purpose of Test	Correlates maternal blood tests and fetal ultrasound to identify increased risk for several chromosomal abnormalities	Maternal blood for increased risk for fetal Down Syndrome, Trisomy 18, open abdominal or neural tube defects	Placental tissue is tested for diagnosis of genetic and chromosomal abnormalities	Sample of amniotic fluid is removed and tested for genetic and chromosomal abnormalities	Non-invasive procedure to scan a woman's abdomen and pelvis to produce imaging of the ovaries, uterus, placenta, and baby
When Performed	Between 11 and 14 weeks of pregnancy	Between 15 and 22 weeks of pregnancy	Between 10 and 14 weeks of pregnancy	Between 15 and 20 weeks of pregnancy	Between 18 and 22 weeks of pregnancy
What is Tested	Maternal blood for free beta-hCG and PAPP-A Ipregnancy-associated plasma protein-A); Fetal ultrasound for nucal translucency	Maternal blood for four "markers" associated with Down Syndrome, Trisomy 18, open abdominal or neural tube defects	Ultrasound-guided needle insertion into placenta to obtain tissue	Ultrasound-guided needle insertion into amniotic sac surrounding the fetus	Sound waves travel from the transducer and reflect off the internal structures of the baby and mother
Test Accuracy	80% accurate for detection of Down Syndrome and Trisomy 18	Detection rates are approximately 80% for Down Syndrome, 90% for Trisomy 21, and 85% for abdominal or neural tube defects	99% accurate for detection of abnormalities tested for	99% accurate for detection of abnormalities tested for	Detection rates of fetal anomalies are variable; diagnosis usually in conjunction with other tests
Considerations for Test	ACOG recommends all pregnant women be offered fetal chromosomal screening regardless of age Women or partners with family history of birth defects, mental deficits, Trisomy 13, 18, 21	Positive family history of birth defects, Trisomy 18, Down Syndrome, open abdominal/neural tube defects Women with diabetes Women with a viral infection during pregnancy	ACOG recommends women 35+ at time of delivery be offered testing (CVS or amniocentesis) Positive family history for chromosomal or genetic abnormalities Women with increased risk based on first trimester screening	ACOG recommends women 35+ at time of delivery be offered testing (CVS or amniocentesis) Positive family history for chromosomal or genetic abnormalities Women with increased risk based on first trimester screening	All pregnant women
Advantages	Early knowledge of increased risk for Trisomy 13, 18, 21 among other abnormalities Ability to pursue counseling and time to make informed decisions or preparations	Early knowledge of increased risk for Trisomy 13, 18, 21 among other abnormalities Ability to pursue counseling and time to make informed decisions or preparations May be predictive of pregnancy outcomes such as low birth weight infant, preterm labor, IUGR, IUFD	Detection of conditions include Tay-Sachs, thalassemia, sickle cell anemia, cystic fibrosis, and Down Syndrome Testing tailored to the individual based on maternal age, couple's ethnic background, medical history	Detection of conditions include Tay-Sachs, thalassemia, sickle cell anemia, cystic fibrosis, and Down Syndrome Testing tailored to the individual based on maternal age, couple's ethnic background, medical history Amniotic fluid alpha-fetoprotein (AFP) can help rule out open neural tube defect	Early detection of some congenital abnormalities Visualization of multiple organ systems Verifies gestational age, due date, gender, presentation and placental location
Disadvantages	Results are risk estimate, NOT diagnosis False positives/negatives are possible	Results are risk estimate, NOT diagnosis False positives/negatives are possible	Small risk (less than 1 in 300-500) of miscarriage Diagnostic only for tests ordered, does not eliminate chances of offspring with mental deficits or birth defects	Small risk (less than 1 in 300-500) of miscarriage Diagnostic only for tests ordered, does not eliminate chances of offspring with mental deficits or birth defects	No significant risks are known to be associated with second trimester ultrasounds
Follow-Up	Genetic Counseling High-level ultrasound, amniocentesis, or CVS may be indicated to clarify diagnosis	Genetic counseling Repeat Quad Screen and Ultrasound Amniocentesis or CVS may be indicated to clarify diagnosis	Genetic counseling Inconclusive CVS may require amniocentesis	Genetic Counseling	Repeat ultrasound Genetic Counseling Amniocentesis or CVS bay be indicated to clarify the diagnosis