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Prenatal Screening and Diagnosis



Testing at ARUP Laboratories



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BD-CS-014, Rev 1, April 2019





ARUP's comprehensive genetics laboratory performs **prenatal screening and diagnostic testing** in one location.

Prenatal screening and diagnosis refers to a group of tests used to assess the presence of fetal disease prior to birth.

Prenatal screening tests include maternal serum screening and non-invasive prenatal testing (NIPT).

Prenatal diagnostic tests include amniocentesis and chorionic villus sampling (CVS), which are invasive procedures that allow for the collection of cells that can be used for biochemical testing, chromosome analysis, or DNA testing.

Diagnostic Testing	Sample type		Performed in		Methodology		
	Amniocentesis	CVS	First trimester	Second trimester	Chromosome analysis	FISH	Microarray
Chromosome Analysis, Amniotic Fluid (2002293)	●			●	●		
Chromosome Analysis, Amniotic Fluid, with Reflex to Genomic Microarray (2008367)	●			●	●		●
Chromosome FISH, Amniotic Fluid with Reflex to Chromosome Analysis or Genomic Microarray (2011130)	●			●	●	●	●
Chromosome FISH, Prenatal (2002297)	●			●		●	
Cytogenomic SNP Microarray—Fetal (2002366)	●	●	●	●			●
Chromosome Analysis, Chorionic Villus (2002291)		●	●		●		
Chromosome FISH, Chorionic Villus with Reflex to Chromosome Analysis or Genomic Microarray (2011131)		●	●		●	●	●
Chorionic Villus, FISH (0040203)		●	●			●	

Prenatal Aneuploidy/NTD Screening	Performed in		Screens for			
	First trimester	Second trimester	Trisomy 21	Trisomy 18	Other chromosome abnormalities	Open neural tube defect
Non-Invasive Prenatal Testing for Fetal Aneuploidy (Panorama) (2007537)	●	●	●	●	Trisomy 13, triploidy, sex chromosome abnormalities	
Non-Invasive Prenatal Testing for Fetal Aneuploidy (Panorama) with Microdeletions (2010232)	●	●	●	●	Trisomy 13, triploidy, sex chromosome abnormalities, 5 microdeletion syndromes	
Non-Invasive Prenatal Testing for Fetal Aneuploidy with 22q11.2 Microdeletion (2013142)	●	●	●	●	Trisomy 13, triploidy, sex chromosome abnormalities, 22q11.2 deletion syndromes	
Maternal Serum Screen, First Trimester (3000145)	●		●	●		
Maternal Screening, Sequential, Specimen #1 (3000146)	●		●	●		
Maternal Screening, Sequential, Specimen #2 (3000148)		●	●	●		●
	Result based on first- and second-trimester samples.					
Maternal Serum Screening, Integrated, Specimen #1 (3000147)	●		●	●		
Maternal Serum Screening, Integrated, Specimen #2 (3000149)		●	●	●		●
Maternal Serum Screen, Alpha Fetoprotein, hCG, Estriol, and Inhibin A (3000143)		●	●	●		●
Maternal Serum Screen, Alpha Fetoprotein (Only) (3000144)		●				●

Products of Conception Testing
Chromosome Analysis, Products of Conception (2002288)
<ul style="list-style-type: none"> Standard chromosome analysis is performed on fetal tissue or villi.
Chromosome Analysis, Products of Conception, with Reflex to Genomic Microarray (2005762)
<ul style="list-style-type: none"> Ensures the highest chance of obtaining meaningful results from POC specimens. If tissue culture is unsuccessful or chromosome analysis results are normal, testing reflexes to genomic microarray.
Genomic SNP Microarray, Products of Conception (2005633)
<ul style="list-style-type: none"> Detects copy-number alterations and loss of heterozygosity in unfixed fetal tissue or villi.
Cytogenomic Molecular Inversion Probe Array, FFPE Tissue—Products of Conception (2010795)
<ul style="list-style-type: none"> For the detection of copy number alterations and loss of heterozygosity in FFPE fetal tissue or villi