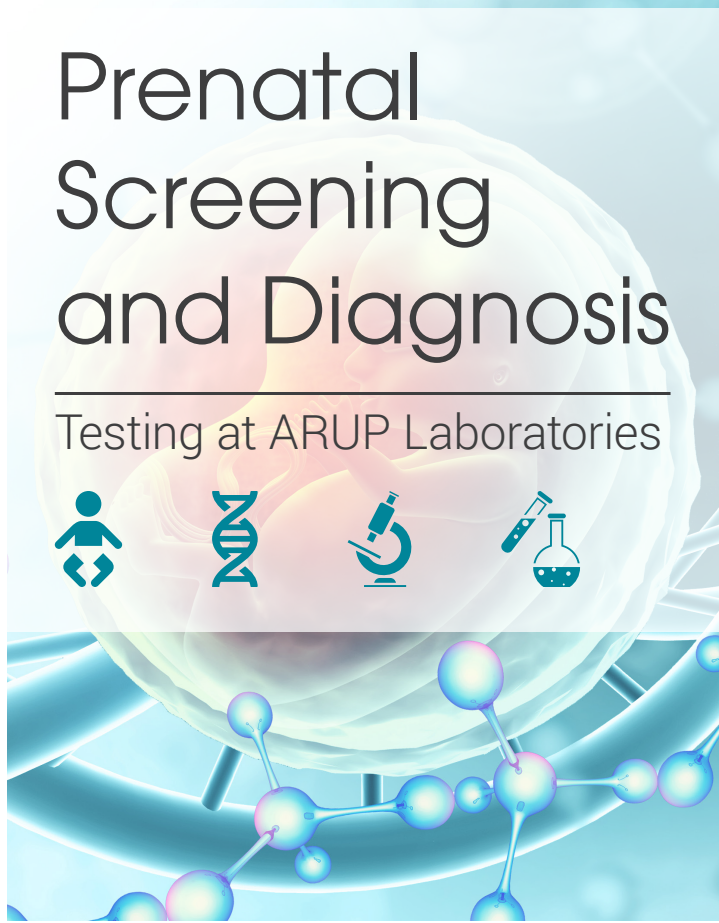


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Prenatal Screening and Diagnosis

Testing at ARUP Laboratories



aruplab.com

ARUP LABORATORIES
500 Chipeta Way
Salt Lake City, UT 84108-1221
Phone: 800-522-2787

Keyword: prenatal

*A nonprofit enterprise of the University of
Utah and its Department of Pathology*

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BD-CS-014, Rev 2, July 2024





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 before birth.

Prenatal screening tests include maternal serum screening and prenatal cell-free DNA (cfDNA) screening, previously referred to as noninvasive 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 During		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/ Neural Tube Defect Screening	Performed During		Conditions Screened			
	First Trimester	Second Trimester	Trisomy 21	Trisomy 18	Other Chromosome Abnormalities	Open Neural Tube Defect
Non-Invasive Prenatal Aneuploidy Screen by cell-free DNA Sequencing (3003043)	●	●	●	●	Trisomy 13, triploidy, sex chromosome abnormalities	
Fetal Aneuploidy Screening with Microdeletions (3004781)	●	●	●	●	Trisomy 13, triploidy, sex chromosome abnormalities, 5 microdeletion syndromes	
Fetal Aneuploidy Screening with 22q11.2 Microdeletion (3004778)	●	●	●	●	Trisomy 13, triploidy, sex chromosome abnormalities, 22q11.2 deletion syndromes	
Maternal Serum Screen, First Trimester, hCG, PAPP-A, NT (3000145)	●		●	●		
Maternal Screening, Sequential, Specimen #1, hCG, PAPP-A, NT (3000146)	●		●	●		
Maternal Screening, Sequential, Specimen #2, Alpha Fetoprotein, hCG, Estriol, and Inhibin A (3000148)	Result based on first- and second-trimester samples		●	●		●
Maternal Serum Screening, Integrated, Specimen #1, PAPP-A, NT (3000147)	●		●	●		
Maternal Serum Screening, Integrated, Specimen #2, Alpha Fetoprotein, hCG, Estriol, and Inhibin A (3000149)		●	●	●		●
Maternal Serum Screen, Alpha Fetoprotein, hCG, Estriol, and Inhibin A (Quad) (3000143)		●	●	●		●
Maternal Serum Screen, Alpha Fetoprotein (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 specimens for products of conception testing 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 (3004273) <ul style="list-style-type: none"> For detection of copy number alterations and loss of heterozygosity in FFPE specimen



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