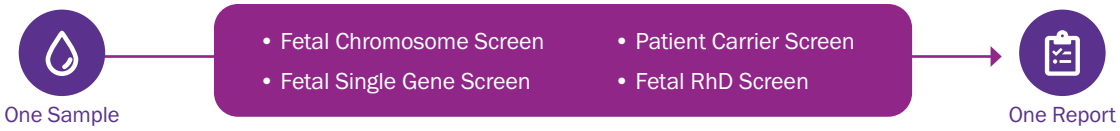
FirstGene®

Multiple Prenatal Screen

Multiple prenatal screen conditions list

The FirstGene® Multiple Prenatal Screen delivers a comprehensive view of common genetic risks in pregnancy from a single sample, combining fetal chromosome, single-gene, carrier, and RhD screening into one streamlined report. It focuses on clinically meaningful conditions, including those aligned with ACOG and ACMG guidelines, to help you provide clear, actionable information to your patients.



Screen Element	Screens for	Gene	ACOG Guidelines	ACMG Guidelines
Fetal chromosome screen[1,2]	Trisomy 21: Down syndrome		✓	
	Trisomy 18: Edwards syndrome		✓	
	Trisomy 13: Patau syndrome		✓	
	Fetal Sex			
	Sex Chromosome Aneuploidies		✓	
	22q11.2 Microdeletion: DiGeorge syndrome			✓
Fetal single gene screen + Patient carrier screen[3,4]	Cystic Fibrosis	*CFTR*	✓	✓
	Spinal Muscular Atrophy	*SMN1*	✓	✓
	Fragile X syndrome*	*FMR1*	✓	✓
	Beta Globin-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease)	*HBB*	✓	✓
	Hb Bart syndrome	*HBA1 / HBA2*	✓	✓
	Hexosaminidase A Deficiency (including Tay-Sachs Disease)	*HEXA*	✓	✓
	Canavan Disease	*ASPA*	✓	✓
	Familial Dysautonomia	*ELP1*	✓	✓
	Smith-Lemli-Opitz syndrome	*DHCR7*		✓
	Phenylalanine Hydroxylase Deficiency	*PAH*		✓
	Medium Chain Acyl-CoA Dehydrogenase Deficiency	*ACADM*		✓
	Galactosemia	*GALT*		✓
	Methylmalonic Acidemia, MMUT-Related	*MMUT*		✓
	Methylmalonic Aciduria and Homocystinuria, cblC Type	*MMACHC*		✓
	Homocystinuria, CBS-Related	*CBS*		✓
	Pompe Disease	*GAA*		✓
	Carnitine Palmitoyltransferase II Deficiency	*CPT2*		✓
	Congenital Disorder of Glycosylation, *PMM2*-Related	*PMM2*		✓
	GNPTAB-Related Disorders	*GNPTAB*		✓
	Autosomal Recessive Polycystic Kidney Disease, *PKHD1*-Related	*PKHD1*		✓
Fetal RhD screen[5]	Feto-Maternal Blood Compatibility		✓	

*Fragile X status is determined for the carrier status of the pregnant person only

1. NIPS Guidelines ACMG: Dungan JS, Klugman S, Darilek S, et al. Noninvasive prenatal screening (NIPS) for fetal chromosome abnormalities in a general-risk population: An evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine. 2023;25(2):100336. doi:10.1016/j.gim.2022.11.004. **2.** NIPS Guidelines ACOG: Screening for fetal chromosomal abnormalities. Obstetrics & Gynecology. 2020;136(4). doi:10.1097/aog.0000000000004084. **3.** Carrier Screen guidelines ACOG: Committee Opinion No. 690 Summary: Carrier Screening in the Age of Genomic Medicine. 2017. Obstetrics and Gynecology 129 (3): 595–96. **4.** Carrier screening guidelines ACMG: P Gregg, A.R., Aarabi, M, Klugman, S et al. Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource of the American College of Medical Genetics and Genomics (ACMG). Genet Med 23, 1793–1806 (2021). https://doi.org/10.1038/s41436-021-01203-z. **5.** ACOG guidelines RhD: American College of Obstetricians and Gynecologists. Practice Bulletin No. 181: Prevention of Rh D Alloimmunization. Obstet Gynecol. 2017;130(2):e57–e70.

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