BAYLOR GENETICS

Outpatient

Prenatal

PreSeek®

Cell-Free Fetal DNA Screening

Non-Invasive Fetal DNA Assessment
Across 30 Clinical Relevant Genes.

PreSeek®

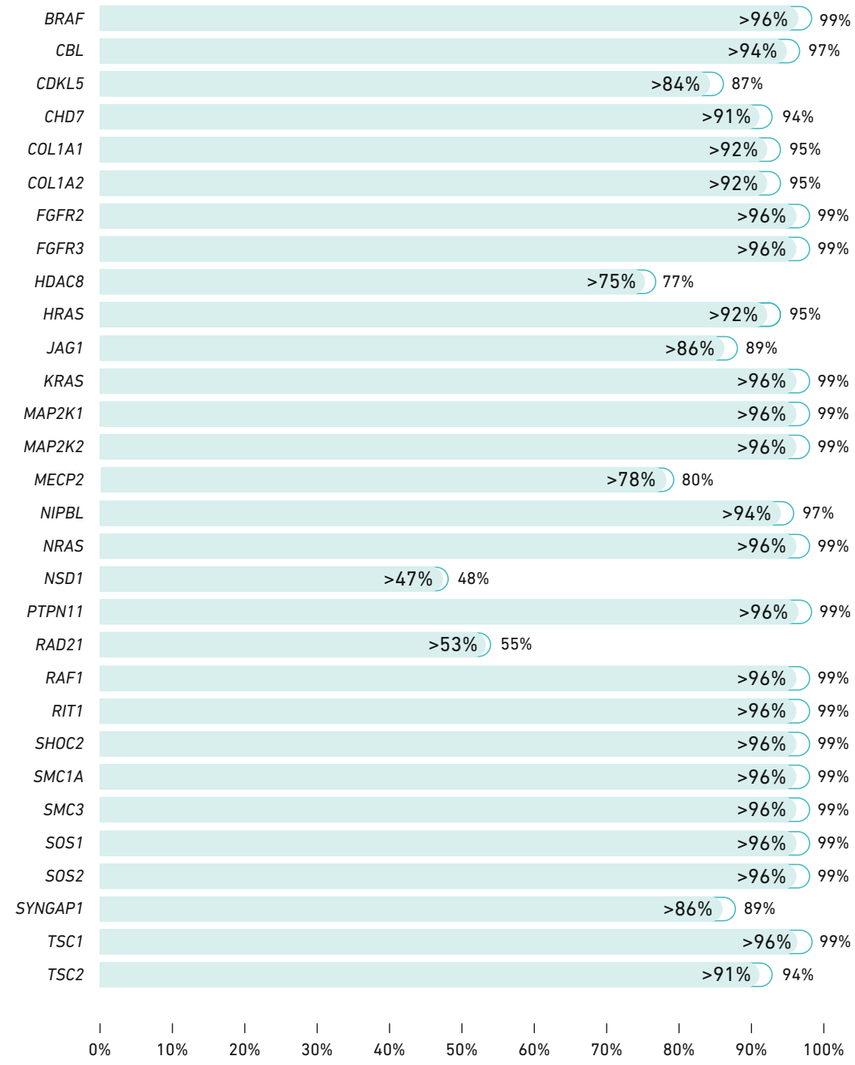
PreSeek® is a comprehensive single gene cell-free fetal DNA screen.

PreSeek® screens for various clinically significant and life-altering genetic disorders that are not screened for with current cell-free fetal DNA screening technology. Disorders screened by this innovative test can occur in the absence of a family history of the condition. The screen, developed by the genomic experts at Baylor Genetics in conjunction with Baylor College of Medicine, assesses fetal DNA for pathogenic and likely pathogenic variants in 30 genes. PreSeek® is the next step in the evolution of screening for genetic disorders during pregnancy, providing information that can affect medical decisions, preparation, and peace of mind for families and physicians.

30 Assessed Genes

Some disorders in PreSeek® are not typically associated with abnormal prenatal ultrasound findings (especially in the first trimester), or may not be evident until late second/third trimester or after delivery. Although the occurrence of each disorder is relatively rare, the cumulative rate of occurrence of these conditions is similar to that of Down syndrome. Knowing whether or not a fetus has one of these significant, and often devastating, genetic disorders can allow for healthcare providers and families to form a plan of care including, but not limited to, genetic counseling, specialist referrals, confirmatory studies, and delivery care. The difference in detecting a significant genetic disorder in the first/second trimester versus late in pregnancy, or in the neonatal period, can be of immeasurable benefit to healthcare providers and families.

PreSeek® Detection Rates[1]



Detection Rate = **Sequencing Detection Rate** (% of mutations caused by small sequence changes)[2] × **Analytical Sensitivity** (>99%) × **Next-Generation Sequencing Coverage** (>97%)

1 Detection rates vary due to size of deletions and/or duplications. Large deletions and/or duplications are not detected by the sequencing technology.

2 Sequencing detection rate for mutations caused by small sequence changes is based on current literature.

Disorders Screened by PreSeek®

Syndromic Disorders

Gene	Disorder
JAG1	Alagille Syndrome
CHD7	CHARGE Syndrome
NIPBL	Cornelia de Lange Syndrome 1
SMC1A	Cornelia de Lange Syndrome 2
SMC3	Cornelia de Lange Syndrome 3
RAD21	Cornelia de Lange Syndrome 4
HDAC8	Cornelia de Lange Syndrome 5
CDKL5	Epileptic Encephalopathy, Early Infantile, 2
SYNGAP1	Intellectual Disability
MECP2	Rett Syndrome
NSD1	Sotos Syndrome 1
TSC1	Tuberous Sclerosis 1
TSC2	Tuberous Sclerosis 2

Noonan Spectrum Disorders

Gene	Disorder
BRAF	Cardiofaciocutaneous Syndrome 1
MAP2K1	Cardiofaciocutaneous Syndrome 3
MAP2K2	Cardiofaciocutaneous Syndrome 4
HRAS	Costello Syndrome/Noonan Syndrome
PTPN11	Noonan Syndrome 1/LEOPARD Syndrome/ Cancers
SOS1	Noonan Syndrome 4
RAF1	Noonan Syndrome 5/LEOPARD Syndrome 2
NRAS	Noonan Syndrome 6/Cancers
RIT1	Noonan Syndrome 8
SOS2	Noonan Syndrome 9
SHOC2	Noonan Syndrome-Like Disorder with Loose Anagen Hair
CBL	Noonan Syndrome-Like Disorder with or without Juvenile Myelomonocytic Leukemia (NSLL)
KRAS	Noonan Syndrome/Cancers

Craniosynostosis Syndromes

Gene	Disorder
FGFR2	Antley-Bixler Syndrome without Genital Anomalies or Disordered Steroidogenesis
	Apert Syndrome
	Crouzon Syndrome
	Jackson-Weiss Syndrome
	Pfeiffer Syndrome Type 1/2/3

Skeletal Disorders

Gene	Disorder
FGFR3	Achondroplasia
	CATSHL Syndrome
	Crouzon Syndrome With Acanthosis Nigricans
	Hypochondroplasia
	Muenke Syndrome
	Thanatophoric Dysplasia, Type I
	Thanatophoric Dysplasia, Type II
COL1A1	Ehlers-Danlos Syndrome, Classic
	Ehlers-Danlos Syndrome, Type VIIA
	Osteogenesis Imperfecta, Type I
	Osteogenesis Imperfecta, Type II
	Osteogenesis Imperfecta, Type III
	Osteogenesis Imperfecta, Type IV
COL1A2	Ehlers-Danlos Syndrome, Cardiac Valvular Form
	Ehlers-Danlos Syndrome, Type VIIB
	Osteogenesis Imperfecta, Type II
	Osteogenesis Imperfecta, Type III
	Osteogenesis Imperfecta, Type IV

PreSeek® is a screening test. Pregnancy decisions should not be based solely on the results of PreSeek®. The purpose of PreSeek® is to indicate if the fetus is at increased risk for a genetic disorder allowing for follow-up invasive prenatal studies or newborn studies.

Performing this screening allows for an assessment for known pathogenic and likely pathogenic variants in select genes associated with select disorders. PreSeek® should be offered in conjunction with genetic counseling, including a review of family history, to help determine the most appropriate prenatal studies for any pregnant woman.

Gestational Age

9 Weeks
or Greater

Samples Needed

Peripheral Blood
in Two 10ml
Cell-Free DNA BCT®
Streck Tubes

Turnaround Time

14 Calendar Days

Test Code

21200

45+ YEARS OF INNOVATION

4 MILLION+ CLINICAL TESTS PERFORMED

1 MILLION+ FAMILIES HELPED

1 MISSION EMPOWERING YOU WITH ANSWERS THAT MATTER

Baylor Genetics pioneered the history of genetic testing. Now, we're leading the way in precision diagnostics.

A pioneer of precision medicine for nearly 50 years, Baylor Genetics is a leading diagnostic genomics partner offering a full spectrum of clinically relevant genetic testing, including Whole Genome Sequencing, Whole Exome Sequencing, and focused panels. Baylor Genetics couples the fastest and most comprehensive precision diagnostics options with the support of genetic counselors to help clinicians and patients avoid a lengthy diagnostic odyssey, guide medical management, and make sure no patient with a genetic disorder gets left behind. Our test menu spans from family planning, pregnancy, neonatal and pediatric testing, oncology, and beyond.

Baylor Genetics is located in Houston's Texas Medical Center.

1.800.411.4363 | BAYLORGENETICS.COM

05.08.26