

Introducing PreSeek™

The first clinical non-invasive prenatal multi-gene sequencing screen

PRESEEK AT A GLANCE

- Current non-invasive prenatal tests screen for aneuploidies and microdeletions, but **PreSeek** goes further by screening fetal DNA for pathogenic and likely pathogenic variants associated with select single gene conditions.
- **PreSeek** works as a complementary screen to traditional NIPT, allowing a more complete picture of the risk of a pregnancy being affected by a genetic disorder.
- **PreSeek** detects predominantly *de novo* variants in 30 genes.
- **PreSeek** allows for the detection of variants that cause clinically significant and life-altering genetic disorders, such as **Noonan spectrum disorders**, **Cornelia de Lange syndrome**, and **Osteogenesis imperfecta**.
- Per ACMG and ACOG, the risk of having a child with single-gene disorders such as achondroplasia and Crouzon syndrome increases with advanced paternal age. **PreSeek** is now the first non-invasive prenatal screen to detect disorders with an increased prevalence linked to advanced paternal age.

Introducing **PreSeek**, the first clinical non-invasive prenatal multi-gene sequencing screen. **PreSeek** screens for various clinically significant and life-altering genetic disorders that are not screened for with current NIPT technology. Disorders screened by this innovative test often 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. Simply put, **PreSeek** is the most comprehensive single gene cell-free fetal DNA screen available.

Many 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 is a screening test. This means that 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.

TEST DETAILS

Test code: 21200

Specimens needed: Both biological maternal and biological paternal specimens are required for testing.

Specimen collection:

Maternal: peripheral blood in two 10mL Streck tubes.

Paternal: peripheral blood in an EDTA tube or saliva using saliva collection kit provided by Baylor Genetics.

Gestational age: Minimum of 10 weeks.

TAT: 14 calendar days.

Syndromic Disorders	
JAG1	Alagille syndrome
CHD7	CHARGE syndrome
HDAC8	Cornelia de Lange syndrome 5
	Wilson-Turner syndrome
NIPBL	Cornelia de Lange syndrome 1
RAD21	Cornelia de Lange syndrome 4
SMC1A	Cornelia de Lange syndrome 2
SMC3	Cornelia de Lange syndrome 3
TSC1	Tuberous sclerosis 1
TSC2	Tuberous sclerosis 2
CDKL5	Epileptic encephalopathy, early infantile, 2
MECP2	Rett syndrome
NSD1	Sotos syndrome 1
SYNGAP1	Intellectual disability
Craniosynostosis Syndromes	
FGFR2	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis
	Apert syndrome
	Crouzon syndrome
	Jackson-Weiss syndrome
	Pfeiffer syndrome type 1/2/3

Noonan Spectrum Disorders	
BRAF	Cardiofaciocutaneous syndrome 1
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia (NSLL)
HRAS	Costello syndrome/Noonan syndrome
KRAS	Noonan syndrome/cancers
MAP2K1	Cardiofaciocutaneous syndrome 3
MAP2K2	Cardiofaciocutaneous syndrome 4
NRAS	Noonan syndrome 6/cancers
PTPN11	Noonan syndrome 1/ LEOPARD syndrome/cancers
RAF1	Noonan syndrome 5/LEOPARD syndrome 2
RIT1	Noonan syndrome 8
SHOC2	Noonan syndrome-like disorder with loose anagen hair
SOS1	Noonan syndrome 4
SOS2	Noonan syndrome 9
Skeletal Disorders	
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



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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 review of family history, to help determine the most appropriate prenatal studies for any pregnant woman.