PRENATAL

BAYLOR GENETICS

PRESEEK™
Noninvasive Prenatal Sequencing Screen

Expecting a baby is an exciting and mysterious time. And it's natural for your patient to wonder if they have a healthy pregnancy. Introducing PreSeek, the most comprehensive, single gene, cell-free fetal DNA screen available. This means we can distinguish between the baby's DNA from the mother's DNA, allowing for genetic analysis that is revolutionary.

Get a more complete picture into your patient's pregnancy with PreSeek

Introducing PreSeek, the first clinical noninvasive prenatal multigene sequencing screen.

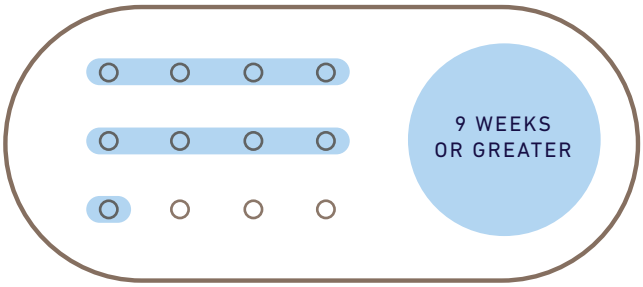
PreSeek screens for various clinically significant and life-altering genetic disorders that are not screened for with current NIPT technology. Disorders screened by this innovative test can occur in the absence of a family history of the condition. The screen, developed by the genomic experts at Baylor 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.

ASSESSES
30
GENES

Simply put, PreSeek is the most comprehensive single gene cell-free fetal DNA screen available.

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 baby 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.

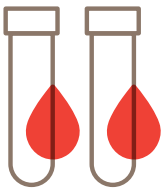
GESTATIONAL AGE



SAMPLES NEEDED

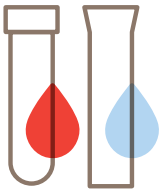
BOTH BIOLOGICAL MOM
& BIOLOGICAL DAD

MOM



PERIPHERAL BLOOD IN
TWO 10ML CELL-FREE
DNA BCT® STRECK TUBES

DAD

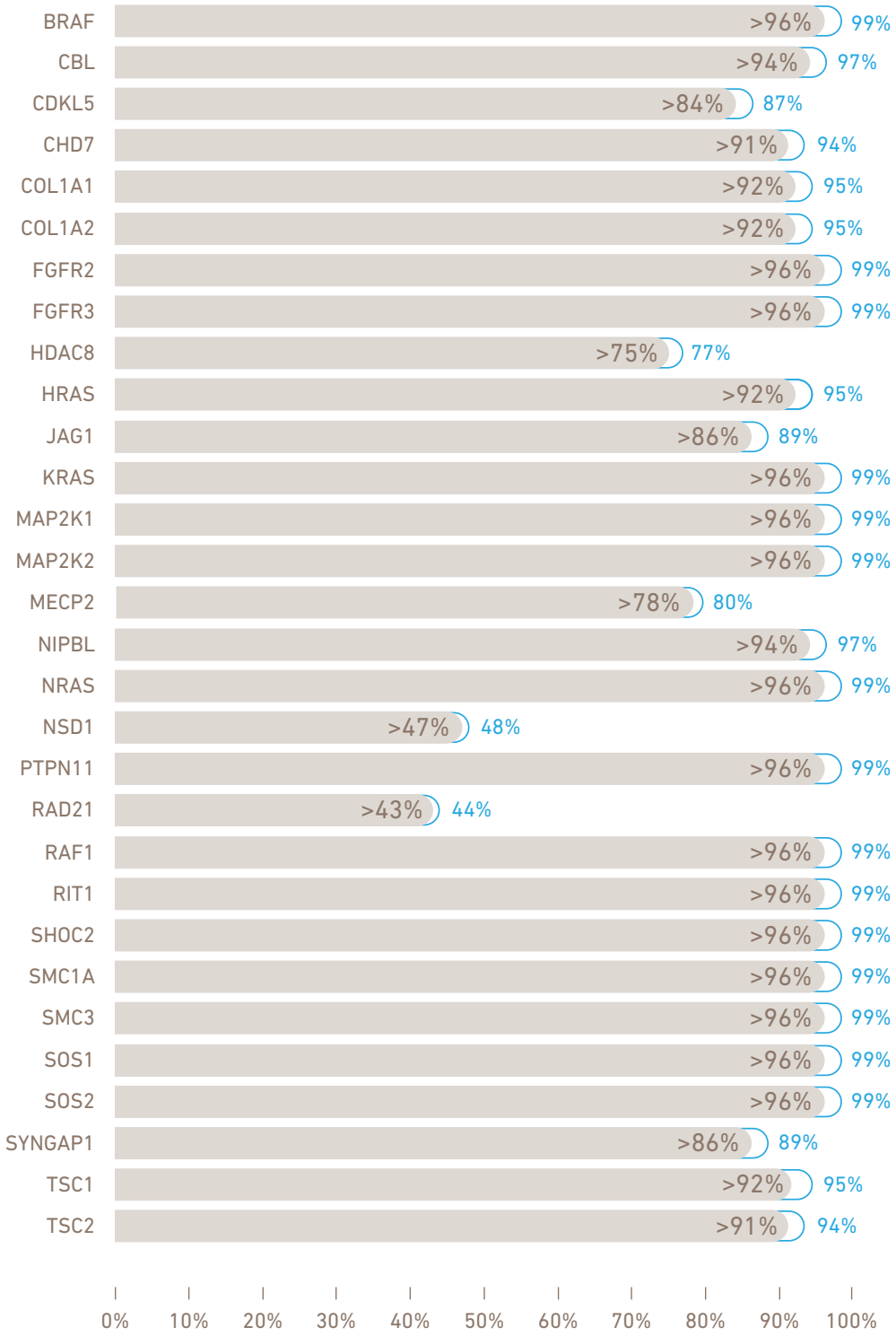


PERIPHERAL BLOOD IN
AN EDTA TUBE OR SALIVA
USING SALIVA COLLECTION
KIT PROVIDED BY BAYLOR
GENETICS

TURNAROUND TIME

TEST CODE

PreSeek Detection Rates



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

*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

Disclaimer: 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 baby 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.

40 YEARS OF INNOVATION

4 MILLION+ CLINICAL TESTS PERFORMED

1 MILLION+ FAMILIES HELPED

3 THOUSAND+ TESTS OFFERED

1 MISSION IMPROVE HEALTHCARE THROUGH GENETICS

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

Baylor Genetics is a joint venture of Miraca Holdings, Inc. and Baylor College of Medicine, including the #1NIH funded Department of Molecular and Human Genetics. A pioneer of precision medicine for nearly 40 years, Baylor Genetics now offers a full spectrum of clinically relevant genetic testing, access to world-renowned experts, and the confidence to provide patients with the best care.

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09.24.18

APA Insert

PreSeek™ The only prenatal screen that analyzes for conditions associated with Advanced Paternal Age (APA).

While traditional NIPT screens for conditions associated with Advanced Maternal Age (AMA), PreSeek™ screens for conditions associated with Advanced Paternal Age ensuring a more complete screen for parents of advanced age.

Advanced paternal age (APA) refers to the age at which a male is considered to be at an increased risk of having children with certain genetic disorders. Typically, men who are 40 years old or greater at the time of conception are considered to be of APA (Toriello, et al 2008). Approximately 4% of babies are conceived by men of APA, and the percentage is increasing (Martin, et al 2013).

Disorders associated with APA typically are caused by changes, or errors, in just one copy of a gene. Sperm are continuously being produced throughout a man's lifetime. During the copying process of DNA some errors might arise and as a man ages, the chance for these errors to occur increases. Some genetic disorders show a stronger association with APA than others such as specific skeletal disorders caused by changes in different genes that can range from mild to severe. Some skeletal disorders, such as Achondroplasia, are up to 8 times more likely in fathers of APA (Toriello, et al 2008). In addition to Achondroplasia, a few other disorders associated with APA that are screened on PreSeek™ are Pfeiffer syndrome, Crouzon syndrome, Apert syndrome, achondroplasia, thanatophoric dysplasia, and Osteogenesis Imperfecta. Currently PreSeek™ is the only prenatal screen available that analyzes for conditions associated with APA.

PreSeek™ is the next step in the evolution of screening for genetic disorders during pregnancy. Knowing whether or not a baby has one of the genetic conditions related to APA can allow for planning and care during pregnancy and at delivery.

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