

### GESTATIONAL AGE



### SAMPLES NEEDED



PREGNANT MOM



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

#### TURNAROUND TIME



TEST CODE

21200









M = M = M = M + Families Helped

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

Baylor Genetics is a joint venture of H.U. Group 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.

## 1.800.411.4363 BAYLORGENETICS.COM



3 THOUSAND+ TESTS OFFERED

SSION IMPROVE HEALTHCARE THROUGH GENETICS

## BAYLOR GENETICS

10.15.20

## O PRENATAL

Expecting a child comes with a lot of excite r if they have a healthy pregnan lucing PreSeek, the most comprehe ve, single gene, cell-free fetal D e baby's DNA from the moth ing for genetic analysis that is ry. PreSeek provides answers the burden of the unknown. This allows the focus on what matters most – welcom new baby home

## BAYLOR GENETICS

**PRESEEK**<sup>™</sup>

Noninvasive Prenatal Sequencing Screen

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



PreSeek was the first and remains the most comprehensive clinical noninvasive prenatal multigene

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.

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.

ASSESSES 30 GENES  $\models$ 



BRAF	>96%) 99%
CBL	>94%) 97%
CDKL5	>84%) 87%
CHD7	>91%) 94%
COL1A1	>92%) 95%
COL1A2	>92%) 95%
FGFR2	>96%) 99%
FGFR3	>96%) 99%
HDAC8	>75%) 77%
HRAS	>92%) 95%
JAG1	>86%) 89%
KRAS	>96%) 99%
MAP2K1	>96%) 99%
MAP2K2	>96%) 99%
MECP2	>78%) 80%
NIPBL	>94%) 97%
NRAS	>96% ) 99%
NSD1	>47%) 48%
PTPN11	>96% ) 99%
RAD21	>53%) 55%
RAF1	>96% ) 99%
RIT1	>96% ) 99%
SHOC2	>96% ) 99%
SMC1A	>96% ) 99%
SMC3	>96% ) 99%
SOS1	>96% ) 99%
SOS2	>96% ) 99%
SYNGAP1	>86%) 89%
TSC1	>96%) 99%
TSC2	>91%) 94%
0	by small sequence changes!*
	*hased on current literature (>99%) (>97%)



the sequencing technology.

## **PreSeek Detection Rates\***

\*Detection rates vary due to size of deletions and/or duplications. Large deletions and/or duplications are not detected by

\*based on current literature

## Disorders Screened by PreSeek

## SYNDROMIC DISORDERS

## CRANIOSYNOSTOSIS SYNDROMES

GENE	DISORDER	GENE	DISORDER	
JAG1	Alagille syndrome		Antley-Bixler syndrome without genital	
CHD7	CHARGE syndrome		anomalies or disordered steroidogenesis	
NIPBL	Cornelia de Lange syndrome 1	FGFR2	Apert syndrome	
SMC1A	Cornelia de Lange syndrome 2		Crouzon syndrome	
			Jackson-Weiss syndrome	
SMC3	Cornelia de Lange syndrome 3		Pfeiffer syndrome type 1/2/3	
RAD21	Cornelia de Lange syndrome 4			
HDAC8	Cornelia de Lange syndrome 5	SKEL	SKELETAL DISORDERS	
CDKL5	Epileptic encephalopathy, early infantile, 2	CENE	DISORDER	
SYNGAP1	Intellectual disability	GENE	DISORDER	
			Achondroplasia	
MECP2	Rett syndrome		CATSHL syndrome	
NSD1	Sotos syndrome 1		Crouzon syndrome with acanthosis nigrica	
TSC1	Tuberous sclerosis 1	FGFR3	Hypochondroplasia	
TSC2	Tuberous sclerosis 2		Muenke syndrome	

## 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
S0S1	Noonan syndrome 4
RAF1	Noonan syndrome 5/LEOPARD syndrome 2
NRAS	Noonan syndrome 6/cancers
RIT1	Noonan syndrome 8
S0S2	Noonan syndrome 9
SH0C2	Noonan syndrome-like disorder with loose anagen hair
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia (NSLL)
KRAS	Noonan syndrome/cancers

GENE	DISORDER
	Achondroplasia
	CATSHL syndrome
	Crouzon syndrome with acanthosis nigricans
FGFR3	Hypochondroplasia
	Muenke syndrome
	Thanatophoric dysplasia, type I
	Thanatophoric dysplasia, type II
	Ehlers-Danlos syndrome, classic
	Ehlers-Danlos syndrome, type VIIA
COI 1A1	Osteogenesis imperfecta, type I
CULTAT	Osteogenesis imperfecta, type II
	Osteogenesis imperfecta, type III
	Osteogenesis imperfecta, type IV
	Ehlers-Danlos syndrome, cardiac valvular form
	Ehlers-Danlos syndrome, type VIIB
COL1A2	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 pre<mark>gnant woman.</mark>

