PREQUEL® NIPT
FOR YOUR PEACE OF MIND

MONOGENIC DISEASES

NON-INVASIVE PRENATAL SCREENING

Test performed at an external service laboratory

DOMINANT OR DE NOVO MONOGENIC DISEASES

GENE	DISEASES	
BRAF	Cardiofaciocutaneous syndrome 1, LEOPARD syndrome 3, Noonan syndrome	●
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia	●
CHD7	CHARGE syndrome	●
COL1A1	Osteogenesis imperfecta	●
COL1A2	Osteogenesis imperfecta	●
COL3A1	Ehlers-Danlos syndrome	●
CREBBP	Rubinstein–Taybi syndrome	●
EFNB1	Craniofrontonasal dysplasia	●
EP300	Rubinstein–Taybi syndrome	●
FBN1	Marfan syndrome	●
FGFR1	Pfeiffer syndrome	●
FGFR2	Crouzon syndrome	●
FGFR3	Hypochondroplasia	●
FOXG1	Congenital Rett-like syndrome	
G6PD	Glucose-6-phosphate dehydrogenase deficiency	● ●
HDAC8	Cornelia de Lange syndrome 5	
HRAS	Costello syndrome, Noonan syndrome	●
JAG1	Alagille syndrome	●
KAT6B	Genitopatellar syndrome	●
KMT2D	Kabuki syndrome	●
KRAS	Noonan syndrome	●
MAP2K1	Cardiofaciocutaneous syndrome 3, Noonan syndrome	● ●
MAP2K2	Cardiofaciocutaneous syndrome 4, Noonan syndrome	● ●
MECP2	Rett syndrome	
NIPBL	Cornelia de Lange syndrome 1	● ●

GENE	PATOLOGIE	
NOTCH2	Alagille syndrome 2	●
NRAS	Noonan syndrome	●
NSD1	Sotos syndrome 1	●
OTC	Ornithine transcarbamylase deficiency	●
PAFAH1B1	Lissencephaly 1	●
POLR1C	Treacher Collins syndrome	●
PTPN11	LEOPARD syndrome	●
RAD21	Cornelia de Lange syndrome 4	●
RAF1	Noonan syndrome	●
SCN1A	Dravet syndrome	●
SHH	Holoprosencephaly type 3	●
SHOC2	Noonan-like syndrome with loose anagen hair, Noonan syndrome	●
SMC1A	Cornelia de Lange syndrome 2	●
SMC3	Cornelia de Lange syndrome 3	
SOS1	Noonan syndrome	● ●
STXBP1	Early infantile epileptic	
TBX5	encephalopathy Holt-Oram	●
TCF4	syndrome Pitt-Hopkins syndrome	●
TCOF1	Treacher Collins syndrome	●
TSC1	Tuberous sclerosis 1 Tuberous	
TSC2	sclerosis 2 Lissencephaly	●
TUBA1A		●
TUBB2B	Tubulinopathy with polymicrogyria/lissencephaly	●
TWIST1	Saethre-Chotzen syndrome	●
ZIC2	Nonsyndromic holoprosencephaly	●

● screening test ***NEGATIVE = LOW RISK***

● screening test ***POSITIVE = HIGH RISK***

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RECESSIVE MONOGENIC DISEASES

GENE	DISEASES		GENE	PATOLOGIE	
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency	●	***HGSNAT***	Mucopolysaccharidosis type IIIC	●
ACADVL	Very long-chain acyl-CoA dehydrogenase deficiency	●	***IDUA***	Mucopolysaccharidosis type I	●
ALMS1	Alström syndrome	●	***IFT140***	Jeune syndrome	●
ARSA	Metachromatic leukodystrophy	●	***IVD***	Isovaleric acidemia	●
ARSB	Mucopolysaccharidosis type VI	●	***LAMA2***	Congenital muscular dystrophy MDC1A	●
ASPA	Canavan disease	●	***MEFV***	Familial Mediterranean fever	●
ATP7B	Wilson disease	●	***MMAA***	Methylmalonic aciduria, cblA type	●
BBS2	Bardet–Biedl syndrome	●	***MMACHC***	Methylmalonic aciduria and homocystinuria, cblC type	●
CC2D2A	Meckel/Joubert syndrome	●	***MMUT***	Methylmalonic aciduria, mut(0) type	●
CEP290	Meckel/Joubert syndrome Cystic	●	***NAGLU***	Mucopolysaccharidosis type IIIB	●
CFTR	fibrosis Smith-Lemli-Opitz syndrome	●	***NPC1***	Niemann–Pick disease type C 1	●
DHCR7	Short-rib thoracic dysplasia (ciliopathy)	●	***NPC2***	Niemann–Pick disease type C 2	●
DYNC2H1	Ellis-van Creveld syndrome	●	***PAH***	Phenylketonuria	●
EVC	Ellis-van Creveld syndrome	●	***PCCA***	Propionic acidemia	●
EVC2	Pompe disease	●	***PCCB***	Propionic acidemia	●
GAA	(Glycogen storage disease II)	●	***PEX1***	Peroxisome biogenesis disorder type 1A (Zellweger)	●
GALC	Krabbe disease	●	***PKHD1***	Autosomal recessive polycystic kidney disease	●
GALT	Galactosemia	●	***PMM2***	Congenital disorder of glycosylation type 1A	●
GBA1	Gaucher disease	●	***POMT1***	Walker-Warburg syndrome	●
GJB2	Congenital deafness	●	***RPGRIP1L***	Meckel/Joubert syndrome	●
GLB1	GM1 gangliosidosis	●	***SERPINA1***	Alpha 1 antitrypsin deficiency	●
GNPTAB	Mucolipidosis II/III	●	***SLC26A2***	Diastrophic dysplasia/achondrogenesis IB	●
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	●	***SLC3A1***	Cystinuria	●
HBB	Beta thalassemia	●	***SLC7A9***	Cystinuria	●
HEXA	Tay-Sachs disease	●	***SMPD1***	Niemann–Pick disease type A/B	●
HEXB	Sandhoff disease	●	***TMEM67***	Jouberts syndrome	●

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