GENEPLANET

NIPT PREMIUM
List of tested conditions

- Down (trisomy 21), Edwards (trisomy 18), and Patau (trisomy 13) syndrome
- Trisomies 9, 16, and 22
- Aneuploidies of sex chromosomes (XO, XXX, XXY, XYY)
- 92 deletion/duplication syndromes (see below for a complete list)
- All other autosomal aneuploidies and deletions/duplications*
- 202 single-gene conditions (see below for a complete list)
- Gender information

*In case the patient chooses incidental findings (deletions and duplications bigger than 5 M base pairs).

92 Deletion/Duplication Syndromes (PRO)

Chromosome 1p36 deletion syndrome, Chromosome 1q41-q42 deletion syndrome, Chromosome 1p32-p31 deletion syndrome, Chromosome 2p16.1-p15 deletion syndrome, Chromosome 2q33.1 deletion syndrome, Chromosome 2q31.1 duplication syndrome, Chromosome 2q37 deletion syndrome, Chromosome 2q31.1 microdeletion syndrome, Chromosome 2q duplication, Chromosome 3pter-p25 deletion syndrome, Dandy-Walker syndrome, Chromosome 3q13.31 deletion syndrome, Distal chromosome 3p duplication, Chromosome 3q duplication, Chromosome 4p16.3 deletion syndrome, Chromosome 4q21 deletion syndrome, Chromosome 4p duplication, Distal chromosome 4q duplication, Distal chromosome 4q deletion, Cri-du-Chat syndrome, Chromosome 5q14.3 deletion syndrome, Chromosome 5q12 deletion syndrome, Chromosome 5p13 duplication syndrome, Chromosome 5p duplication, Chromosome 6pter-p24 deletion syndrome, Chromosome 6q24-q25 deletion syndrome, Chromosome 6q11-q14 deletion syndrome, Chromosome 6p deletion, Chromosome 6q15-q23 deletion syndrome, Chromosome 6q25-qter deletion syndrome, Chromosome 6q26-q27 deletion syndrome, Chromosome 7q deletion, Chromosome 7q11.23 deletion syndrome, Chromosome 7q21-q32 deletion, Chromosome 7q31-q32 deletion, Chromosome 8p23.1 deletion syndrome, Chromosome 8p23.1 duplication syndrome, Langer-Giedion syndrome, Chromosome 8q22.1 deletion syndrome, Chromosome 8q22.1 duplication syndrome, Chromosome 8p duplication, Chromosome 8q duplication, Chromosome 9p deletion syndrome, Chromosome 9p duplication, DiGeorge syndrome 2, Chromosome 10q22.3-q23.2 deletion syndrome, Chromosome 10q26 deletion syndrome, Chromosome 10p12-p11 deletion syndrome, Chromosome 10p duplication, Chromosome 11p13 deletion syndrome, Chromosome 11p11.2 deletion syndrome, Jacobsen syndrome, Chromosome 11q23 deletion syndrome, Chromosome 12q14 microdeletion syndrome, Chromosome 12p12.1 microdeletion syndrome, Chromosome 12p duplication, Chromosome 13q14 deletion syndrome, Distal chromosome 13q deletion, Chromosome 14q11-q22 deletion syndrome, Chromosome 14q22 deletion syndrome, Proximal chromosome 14q deletion, Chromosome 14q duplication, Angelman syndrome, Prader-Willi syndrome, Chromosome 15q26-qter deletion syndrome, Levy-Shanske syndrome, Chromosome 15q14 deletion syndrome, Chromosome 15q24 microdeletion syndrome, Chromosome 15q26 overgrowth syndrome, Distal chromosome 15q deletion, Chromosome 16p12.2-p11.2 deletion syndrome, Chromosome 16p12.2-p11.2 duplication syndrome, Chromosome 16p13.3 deletion syndrome, Chromosome 16p13.3 duplication syndrome, Proximal chromosome 16q duplication, Smith-Magenis syndrome, Chromosome 17p13.3 deletion syndrome, Potocki-Lupski syndrome, Chromosome 17p13.3 duplication syndrome, Yuan-Harel-Lupski syndrome, Chromosome 17p duplication, Chromosome 18p deletion syndrome, Distal chromosome 18q deletion syndrome, Alagille syndrome 1, Chromosome 20p duplication, Chromosome 21q22 deletion, Chromosome 22q11.2 deletion syndrome, Chromosome Xp11.23-p11.22 duplication syndrome, Chromosome Xp21 deletion syndrome, Chromosome Xq27.3-q28 duplication syndrome, Chromosome Xq21 deletion syndrome, Chromosome Xq22.3 deletion syndrome

202 Single-Gene Conditions (MONO)

GENE	SINGLE-GENE CONDITION
ACTB	Baraitser-Winter syndrome 1
ACTG1	Baraitser-Winter syndrome 2
ACTG2	Visceral myopathy 1
ACVR1	Fibrodysplasia Ossificans Progressiva
ADNP	Autosomal Dominant Mental Retardation 28
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2
ANKRD11	KBG Syndrome
ARID1A	Autosomal Dominant Mental Retardation 14
ARID1B	Coffin-Siris syndrome 1
ASXL1	Bohring-Opitz Syndrome
ASXL3	Bainbridge-Ropers Syndrome
ATP1A2	Alternating Hemiplegia of Childhood 1
ATP1A2	Developmental and epileptic encephalopathy 98
ATP1A3	Developmental and epileptic encephalopathy 99
BCL11A	Dias-Logan syndrome
BICD2	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant
BRAF	Noonan Syndrome 7
BRAF	Cardiofaciocutaneous Syndrome
BRAF	LEOPARD syndrome 3
CACNA1A	Developmental and epileptic encephalopathy 42
CAMTA1	Cerebellar dysfunction with variable cognitive and behavioral abnormalities
CASK	Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia
CDKL5	Developmental and epileptic encephalopathy 2
CHD2	Developmental and epileptic encephalopathy 94
CHD7	CHARGE syndrome

CHD8	Intellectual developmental disorder with autism and macrocephaly
COL11A1	Stickler syndrome, type II
COL1A1	Osteogenesis Imperfecta type I
COL1A1/COL1A2	Osteogenesis Imperfecta type II
COL1A1/COL1A2	Osteogenesis Imperfecta type III
COL1A1/COL1A2	Osteogenesis Imperfecta type IV
COL2A1	Stickler syndrome, type I
COL2A1	Platyspondylic Lethal Skeletal dysplasia, Torrance type
COL2A1	Achondrogenesis, type II or hypochondrogenesis
COL2A1	SED congenita
COL4A1	Porencephaly 1
COL9A2	Epiphyseal dysplasia, multiple, 2
COL9A3	Epiphyseal dysplasia, multiple, 3, with or without myopathy
COMP	Pseudoachondroplasia
COMP	Epiphyseal dysplasia, multiple, 1
CREBBP	Rubinstein-Taybi Syndrome 1
CREBBP	Menke-Hennekam syndrome 1
CTCF	Intellectual developmental disorder, autosomal dominant 21
CTNNB1	Neurodevelopmental disorder with spastic diplegia and visual defects
DNM1	Developmental and epileptic encephalopathy 31
DYNC1H1	Intellectual developmental disorder, autosomal dominant 13
DYRK1A	Intellectual developmental disorder, autosomal dominant 7
EBP	Chondrodysplasia punctata, X-linked dominant
EFNB1	Craniofrontonasal dysplasia
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type
EHMT1	Kleefstra Syndrome 1
EP300	Rubinstein-Taybi Syndrome 2
EP300	Menke-Hennekam syndrome 2
ERF	Craniosynostosis 4
ERF	Chitayat syndrome
FBN1	Marfan Syndrome
FGFR1/FGFR2	Jackson-Weiss Syndrome
FGFR1	Trigonocephaly 1
FGFR1/FGFR2	Pfeiffer syndrome
FGFR2	Saethre-Chotzen Syndrome
FGFR2	Bent Bone Dysplasia Syndrome
FGFR2	Beare-Stevenson Cutis Gyrata syndrome
FGFR2	Antley-Bixler Syndrome Without Genital Anomalies Or Disordered Steroidogenesis
FGFR2	Crouzon syndrome
FGFR2	Apert syndrome
FGFR3	Thanatophoric Dysplasia, type II
FGFR3	SADDAN (severe achondroplasia with developmental delay and acanthosis nigricans)
FGFR3	Muenke Syndrome
FGFR3	Crouzon syndrome with acanthosis nigricans

FGFR3	Thanatophoric Dysplasia, type I
FGFR3	Hypochondroplasia
FGFR3	Achondroplasia
FLNA	Otopalatodigital syndrome, type II
FLNB	Atelosteogenesis, type I
FLNB	Larsen Syndrome
FLNB	Atelosteogenesis, type III
FLNB	Boomerang dysplasia
FOXG1	Rett syndrome, congenital variant
FOXP1	Intellectual developmental disorder with language impairment with or without autistic features
FREM1	Trigonocephaly 2
GABRA1	Developmental and epileptic encephalopathy 19
GABRB2	Developmental and epileptic encephalopathy 92
GATAD2B	GAND syndrome
GFAP	Alexander Disease
GNAO1	Developmental and epileptic encephalopathy 17
GNAO1	Neurodevelopmental disorder with involuntary movements
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant
GRIN2B	Developmental and epileptic encephalopathy 27
GRIN2B	Intellectual developmental disorder, autosomal dominant 6, with or without seizures
HDAC8	Cornelia de Lange syndrome 5
HNRNPK	Au-Kline Syndrome
HNRNPU	Developmental and epileptic encephalopathy 54
HRAS	Costello Syndrome
IFITM5	Osteogenesis imperfecta, type V
JAG1	Alagille Syndrome 1
KANSL1	Koolen-De Vries Syndrome
KAT6B	SBBYS syndrome
KAT6B	Genitopatellar syndrome
KCNB1	Developmental and epileptic encephalopathy 26
KCNJ2	Andersen syndrome
KCNQ2	Developmental and epileptic encephalopathy 7
KCNT1	Developmental and epileptic encephalopathy 14
KIF1A	NESCAV syndrome
KMT2A	Wiedemann-Steiner syndrome
KMT2D	Kabuki Syndrome 1
KRAS	Cardiofaciocutaneous Syndrome 2
KRAS	Noonan Syndrome 3
LMNA	Muscular dystrophy, congenital
LMNA	Hutchinson-Gilford Progeria Syndrome
LZTR1	Noonan Syndrome 10
MAP2K1	Cardiofaciocutaneous Syndrome 3
MAP2K2	Cardiofaciocutaneous Syndrome 4
MECP2	Rett syndrome
MED13L	Impaired intellectual development and distinctive facial features with or without cardiac defects

MEF2C	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language
MSX2	Craniosynostosis 2
MSX2	Parietal Foramina With Cleidocranial Dysplasia
NALCN	Congenital Contractures Of The Limbs And Face, Hypotonia, And Developmental delay
NF1	Neurofibromatosis 1
NF2	Neurofibromatosis 2
NFIX	Marshall-Smith Syndrome
NIPBL	Cornelia de Lange syndrome 1
NOTCH2	Hajdu-Cheney Syndrome
NOTCH2	Alagille Syndrome 2
NR2F1	Bosch-Boonstra-Schaaf Optic Atrophy Syndrome
NRAS	Noonan syndrome 6
NSD1	Sotos Syndrome 1
NSDHL	CHILD syndrome
PACS1	Schuurs-Hoeijmakers syndrome
PIK3CA	Cowden syndrome 5
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1
PPP2R1A	Intellectual developmental disorder, autosomal dominant 36
PPP2R5D	Intellectual developmental disorder, autosomal dominant 35
PRKAR1A	Acrodysostosis 1, with or without Hormone Resistance
PTPN11	Noonan Syndrome 1
PURA	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties
RAD21	Cornelia de Lange syndrome 4
RAF1	Noonan Syndrome 5
RERE	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart
RIT1	Noonan Syndrome 8
RPS6KA3	Coffin-Lowry Syndrome
RUNX2	Metaphyseal Dysplasia with Maxillary Hypoplasia with or without Brachydactyly
RUNX2	Cleidocranial dysplasia
SATB2	Glass Syndrome
SCN1A	Early Infantile Epileptic Encephalopathy 6
SCN1A	Developmental and epileptic encephalopathy 6B, non-Dravet
SCN2A	Developmental and epileptic encephalopathy 11
SCN2A	Episodic ataxia, type 9
SCN8A	Developmental and epileptic encephalopathy 13
SCN8A	Cognitive Impairment With Or Without Cerebellar Ataxia
SETBP1	Schinzel-Giedion syndrome
SETBP1	Intellectual developmental disorder, autosomal dominant 29
SETD2	Luscan-Lumish Syndrome
SETD5	Intellectual developmental disorder, autosomal dominant 23
SHANK3	Phelan-McDermid Syndrome
SHOC2	Noonan-Like Syndrome with Loose Anagen Hair
SKI	Shprintzen-Goldberg Syndrome
SLC25A24	Fontaine Progeroid Syndrome
SMAD3	Loeys-Dietz syndrome 3

SMAD4	Myhre Syndrome
SMARCA2	Nicolaides-Baraitser Syndrome
SMARCA2	Blepharophimosis-impaired intellectual development syndrome
SMARCA4	Autosomal Dominant Mental Retardation 16
SMARCB1	Autosomal Dominant Mental Retardation 15
SMARCE1	Coffin-Siris Syndrome 5
SMC1A	Cornelia de Lange syndrome 2
SMC3	Cornelia de Lange syndrome 3
SOS1	Noonan Syndrome 4
SOS2	Noonan Syndrome 9
SOX9	Campomelic Dysplasia
SPECC1L	Opitz GBBB Syndrome, Type II
SPTAN1	Developmental and epileptic encephalopathy 5
SRCAP	Floating-Harbor Syndrome
SRCAP	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities
STAT3	Hyper-IgE recurrent infection syndrome
STXBP1	Developmental and epileptic encephalopathy 4
SYNGAP1	Intellectual developmental disorder, autosomal dominant 5
TBL1XR1	Pierpont Syndrome
TBL1XR1	Intellectual developmental disorder, autosomal dominant 41
TBX5	Holt-Oram Syndrome
TCF4	Pitt-Hopkins syndrome
TGFB2	Loeys-Dietz syndrome 4
TGFBR1	Loeys-Dietz syndrome 1
TGFBR2	Loeys-Dietz syndrome 2
TRAF7	Cardiac, Facial, and Digital Anomalies with Developmental Delay
TRPS1	Trichorhinophalangeal syndrome, type I
TSC1	Tuberous Sclerosis-1
TSC2	Tuberous Sclerosis-2
TUBA1A	Lissencephaly 3
TUBB	Symmetric circumferential skin creases, congenital, 1
TUBB	Cortical Dysplasia, Complex, with Other Brain Malformations 6
TUBB2A	Cortical Dysplasia, Complex, with Other Brain Malformations 5
TUBB4A	Leukodystrophy, hypomyelinating, 6
TWIST1	Saethre-Chotzen syndrome with or without eyelid anomalies
TWIST1	Craniosynostosis 1
TWIST1	Sweeney-Cox syndrome
TWIST1	Robinow-Sorauf Syndrome
WDR45	Neurodegeneration with brain iron accumulation 5
ZBTB20	Primrose syndrome
ZC4H2	Wieacker-Wolff syndrome, female-restricted
ZEB2	Mowat-Wilson syndrome

NIPT PREMIUM STATISTICS

SYNDROME		SENSITIVITY	SPECIFICITY	PPV
Trisomy 21		99.17%	99.95%	92.19%
Trisomy 18		98.24%	99.95%	76.61%
Trisomy 13		>99.9%	99.96%	32.84%
CNV	**≥10 Mb**	>99.9%	99.97%	NA
	<10 Mb	>99.9%	99.86%	NA
Single-gene conditions		>99%	>99%	NA
Fetal sex		99,53%	99.20%	NA
SEX CHROMOSOME ANEUPLOIDIES		**SENSITIVITY**	**PPV**	**NPV**
XYY		>99.9%	50.00%	>99.9%
XXY		>99.9%	42.86%	>99.9%
XXX		>99.9%	70.00%	>99.9%
XO		>99.9%	40.00%	>99.9%

Note: The data in the table is based on historical literature and internal data, and only reflects past detection, not the actual condition of the tested sample nor the promised value.

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