



**Micro
Health**
Laboratories

.....for precious life



Genetic answers for precious life

Trisomies **21,18,13**
All chromosome aberration



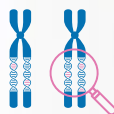
Sex
Chromosome Aneuploidies



116
Microdeletions



100
Monogenic diseases



MICROGen™ - Non Invasive Prenatal Screening Test (NIPT)



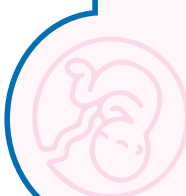
MicroGen Basic - NIPT Screening Test



- ✓ Trisomy 21,18,13
- ✓ Sex chromosome aneuploidy
- ✓ Presence of Y chromosome



MicroGen Premium - NIPT Screening Test



- ✓ All chromosome aberration
- ✓ Sex chromosome aneuploidy
- ✓ Presence of Y chromosome
- ✓ 116 Microdeletions



MicroGen Monogenic - NIPT Screening Test

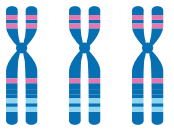


- ✓ Trisomy 21,18,13
- ✓ Sex chromosome aneuploidy
- ✓ Presence of Y chromosome
- ✓ 4 Microdeletions
- ✓ 100 Monogenic diseases
- ✓ Inclusive of confirmatory test



TRISOMIES

- Trisomy 21 (Down Syndrome)
- Trisomy 18 (Edward Syndrome)
- Trisomy 13 (Patau Syndrome)

21, 18, 13
Trisomies 

SEX CHROMOSOME ANEUPLOIDIES

- Turner syndrome (Monosomy X)
- Triple X syndrome (XXX)
- Klinefelter syndrome (XXY)
- Jacobs syndrome (XYY)

Sex
Chromosome Aneuploidies 

116 DELETION / DUPLICATION SYNDROMES

- 1p36 deletion syndrome
- 2q33.1 deletion syndrome
- 4p16.3 Wolf-Hirschhorn syndrome
- 5p- Cri Du Chat syndrome
- 7q11.23 Williams-Beuren syndrome
- 11q23 Jacobsen syndrome
- 15q11.2-q13 Prader-willi / Angelman syndrome
- 22q11.2 DiGeorge syndrome
- + 108 microdeletion/duplication syndromes

116
Microdeletions 

116 Deletion/Duplication
Syndromes
(Annexure I)

100 MONOGENIC DISEASES

- | | |
|-----------------------------|--------------------------------------|
| • Cystic Fibrosis | • Alstrom syndrome |
| • Sickle-Cell disease | • Abetalipoproteinemia |
| • Beta Thalassemia | • Bardet-Biedl syndrome |
| • Tay-Sachs disease | • Alport syndrome, X-linked |
| • Gaucher disease | • Pendred syndrome |
| • Phenylketonuria | • Familial Dysautonomia |
| • Autosomal Recessive | • Joubert syndrome, Type 2 |
| • Polycystic Kidney disease | • Isovaleric Acidemia |
| • Canavan disease | • Glutaric Acidemia, Type 2A |
| • Fanconi Anemia, Type C | • Maple Syrup Urine disease, Type 1B |
| • Usher syndrome, Type 1F | • Factor XI Deficiency |

100
Monogenic diseases 

Panel of
100 autosomal and X-linked
monogenic diseases
(Annexure II)



S NO	SYNDROME
1	1p36 1p36 deletion syndrome
2	2q33.1 2q33.1 deletion syndrome
3	4p16.3 Wolf-Hirschhorn syndrome
4	5p- Cri Du Chat syndrome
5	7q11.23 Williams-Beuren syndrome
6	11q23 Jacobsen syndrome
7	15q11.2-q13 Prader-willi/Angelman syndrome
8	22q11.2 DiGeorge syndrome
9	1p32-p31 deletion syndrome
10	1p41-q42 deletion syndrome
11	1q43-q44 deletion syndrome
12	2p12-p11.2 deletion syndrome
13	2p15-p16.1 deletion syndrome
14	2q13 deletion syndrome
15	2q13 duplication syndrome
16	2q31.1 microdeletion syndrome
17	2q31.1 duplication syndrome
18	2q35 duplication syndrome
19	3p25.3 deletion syndrome
20	3pter-p25 deletion syndrome
21	3q13.31 deletion syndrome
22	Dandy- Walker syndrome (DWS)
23	3q26 microduplication syndrome
24	3q29 deletion syndrome
25	4q21 deletion syndrome
26	Axenfeld-Rieger syndrome, type 1 (RIEG1)
27	5p13 duplication syndrome
28	5q12 deletion syndrome
29	5q14.3 deletion (proximal) syndrome
30	Sotos syndrome
31	6p22 microdeletion syndrome
32	6q11-q14 deletion syndrome
33	6q24-q25 deletion syndrome
34	Coffin-Siris syndrome 1 (CSS1)
35	Chordoma
36	Greig cephalopolysyndactyly syndrome (GCPS)
37	7p22.1 microduplication syndrome
38	7q11.23 deletion (distal) syndrome
39	Williams-Beuren syndrome (WBS)
40	Currarino syndrome
41	8q12 microduplication syndrome
42	Nablis mask-like facial syndrome (NMLFS)
43	7q36.3 duplication syndrome
44	8p11.2 deletion syndrome
45	8p23.1 deletion syndrome
46	Trichorhinophalangeal syndrome type 2 (TRPS2)
47	9p deletion syndrome
48	9p13 microdeletion syndrome
49	9p24.3 deletion syndrome
50	9q33.3q34.11 microdeletion syndrome
51	Early infantile epileptic encephalopathy 4 (EIEE4)
52	Kleefstra syndrome 1 (KLEFS1)
53	10p11.21-p12.31 microdeletion syndrome
54	DiGeorge syndrome/ velocardiofacial syndrome complex 2 (DSG2)
55	10q22.q23.2 deletion syndrome
56	Split-hand/foot malformation 3 (SHFM3)
57	10q26 deletion syndrome
58	Potocki-Shaffer syndrome
59	WAGR syndrome

S NO	SYNDROME
60	WAGRO syndrome
61	11q13.2-q13.4 deletion syndrome
62	11q22.2q22.3 microdeletion syndrome
63	11q23 deletion syndrome
64	12p12.1 microdeletion syndrome
65	12q14 microdeletion syndrome
66	12q15q21.1 microdeletion syndrome
67	13q14 deletion syndrome
68	14q11-q22 deletion syndrome
69	Frias syndrome
70	14q24.1-q24.3 microdeletion syndrome
71	15q13.3 deletion syndrome (BP4 to BP5) (loss)
72	15q13.3 deletion syndrome (BP4 to BP5) (gain)
73	15q14 microdeletion syndrome
74	15q25.2 deletion (proximal) syndrome
75	15q26-qter deletion syndrome
76	16p11.2-p12.2 microduplication syndrome
77	16p12.2 deletion (proximal) syndrome
78	16p13.11 duplication syndrome
79	16p13.11 deletion syndrome
80	Polycystic kidney disease, infantile severe,with tuberous sclerosis (PKDTS)
81	Rubinstein- Taybi syndrome
82	Alpha-thalassemia/intellectual disability syndrome,chromosome 16-related (ATR-16 syndrome)
83	16q22 deletion syndrome
84	Smith-Magenis syndrome
85	Yuan-Harel-Lupski syndrome (YUHAL)
86	17p12 deletion syndrome
87	17p12 duplication syndrome
88	17p13.1 deletion syndrome
89	Miller-Dieker lissencephaly syndrome (MDLS) (loss)
90	Miller-Dieker lissencephaly syndrome (MDLS) (gain)
91	17p13.3 telomeric duplication syndrome
92	17q12 deletion syndrome
93	17q21.3 deletion syndrome
94	17q23.1-q23.2 deletion syndrome
95	Tetrasomy 18p syndrome
96	18p deletion syndrome
97	18q deletion syndrome
98	19p13 duplication syndrome
99	19q13.11 microdeletion syndrome
100	20p13 microdeletion syndrome
101	21q22.11-q22.12 microdeletion syndrome
102	22q11.2 deletion syndrome
103	22q11.2 deletion syndrome (LCR22 B/C-D)
104	22q13 deletion syndrome
105	22q13 duplication syndrome
106	Xp11.22 duplication syndrome
107	Xp11.22-p11.23 duplication syndrome
108	Xp11.23 microdeletion syndrome
109	Xp11.3 deletion syndrome
110	Xp21 microdeletion syndrome
111	Xp21.2 microdeletion syndrome
112	Xp22.31 microdeletion syndrome
113	Xq21 microdeletion syndrome
114	Xq22.3 telomeric deletion syndrome
115	Xq27.3-q28 duplication syndrome
116	Xq28 deletion syndrome

S NO	DISEASE	GENE	CLASSIFICATION
1	3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	HMGCL	Metabolic
2	3-Methylcrotonyl-CoA Carboxylase Deficiency 1	MCCC1	Metabolic
3	3-Methylcrotonyl-CoA Carboxylase Deficiency 2	MCCC2	Metabolic
4	Abetalipoproteinemia	MTTP	Digestive, Neurological, Ophthalmological, Hematological
5	Acyl-CoA Oxidase I Deficiency	ACOX1	Neurological
6	Aicardi-Goutières Syndrome	SAMHD1	Neurological
7	Alport Syndrome, X-Linked	COL4A5	Renal, Ophthalmological, Hearing
8	Alstrom Syndrome	ALMS1	Ophthalmological, Hearing, Renal, Cardiac
9	Andermann Syndrome	SLC12A6	Muscular, Neurological
10	Aromatase Deficiency	CYP19A1	Sexual Development
11	Arthrogryposis Mental Retardation Seizures	SLC35A3	Metabolic
12	Asparagine Synthetase Deficiency	ASNS	Neurological
13	Aspartylglycosaminuria	AGA	Metabolic, Neurological
14	Autosomal Recessive Polycystic Kidney Disease	PKHD1	Renal
15	Bardet-Biedl Syndrome (BBS1-related)	BBS1	Ophthalmological, Metabolic, Endocrine
16	Bardet Biedl Syndrome (BBS12-related)	BBS12	Ophthalmological
17	Beta Thalassemia	HBB	Hematological
18	Biotinidase Deficiency	BTD	Metabolic
19	Canavan Disease	ASPA	Neurological
20	Carpenter Syndrome	RAB23	Skletal
21	Choreacanthocytosis	VPS13A	Neurological
22	Choroideremia, X-Linked	CHM	Ophthalmological
23	Citrin Deficiency	SLC25A13	Metabolic
24	Combined Oxidative Phosphorylation Deficiency 3	TSFM	Neurological, Metabolic, Cardiac
25	Congenital Disorder of Glycosylation, Type 1A (PMM2-related)	PMM2	Metabolic
26	Congenital Neutropenia (HAX1-related)	HAX1	Immunological
27	Crigler Najjar Syndrome, Type I	UGT1A1	Metabolic
28	Cystic Fibrosis *	CFTR	Respiratory, Digestive
29	Factor XI Deficiency	F11	Hematological
30	Familial Dysautonomia	IKBKAP	Neurological
31	Fanconi Anemia, Type C	FANCC	Immunological
32	Fanconi Anemia, Type G	FANCG	Hematological
33	Gaucher Disease	GBA	Neurological, Hepatic, Cardiac
34	Glutaric Acidemia, Type 2A	ETFA	Metabolic
35	Glycine Encephalopathy (GLDC-related)	GLDC	Metabolic
36	Glycogen Storage Disease, Type 1A	G6PC	Metabolic
37	Glycogen Storage Disease, Type 1B	SLC37A4	Metabolic
38	Glycogen Storage Disease, Type 3	AGL	Metabolic
39	Glycogen Storage Disease, Type 7	PFKM	Metabolic
40	GRACILE Syndrome	BCS1L	Metabolic
41	Hereditary Fructose Intolerance	ALDOB	Metabolic
42	Homocystinuria, Type cbLE	MTRR	Metabolic
43	Hydroletharus Syndrome	HYLS1	Neurological, Cardiac
44	Inclusion Body Myopathy, Type 2	GNE	Muscular
45	Isovaleric Acidemia	IVD	Metabolic
46	Joubert Syndrome, Type 2	TMEM216	Neurological
47	Junctional Epidermolysis Bullosa, Herlitz Type	LAMC2	Skin
48	Lamellar Ichthyosis, Type 1	TGM1	Metabolic
49	Leber Congenital Amaurosis (LCA5-related)	LCA5	Ophthalmological
50	Leigh Syndrome, French-Canadian Type	LRPPRC	Neurological, Muscular
51	Leukoencephalopathy with Vanishing White Matter	EIF2B5	Neurological
52	Leydig Cell Hypoplasia [Luteinizing Hormone Resistance]	LHCGR	Sexual Development
53	Limb Girdle Muscular Dystrophy, Type 2E	SGCB	Muscular
54	Lipoamide Dehydrogenase Deficiency [Maple Syrup Urine Disease, Type 3]	DLD	Metabolic
55	Lipoprotein Lipase Deficiency	LPL	Metabolic
56	Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADHA	Metabolic
57	Lysinuric Protein Intolerance	SLC7A7	Metabolic
58	Maple Syrup Urine Disease, Type 1B	BCKDHB	Metabolic
59	Methylmalonic Acidemia (MMAA-related)	MMAA	Metabolic
60	Methylmalonic Aciduria, Type Mut(0)	MUT	Metabolic
61	Methylmalonic Aciduria and Homocystinuria, Type cbLC	MMACHC	Metabolic
62	Methylmalonic Aciduria and Homocystinuria, Type cbLD	MMADHC	Metabolic
63	Mucopolysaccharidosis, Type II [Hunter Syndrome], X-Linked	IDS	Respiratory, Cardiac
64	Mucopolysaccharidosis, Type IIIC [Sanfilippo C]	HGSNAT	Metabolic, Neurological, Ophthalmological
65	Multiple Sulfatase Deficiency	SUMF1	Metabolic

S NO	DISEASE	GENE	CLASSIFICATION
66	Myotubular Myopathy, X-Linked	MTM1	Muscular
67	Navajo Neurohepatopathy [MPV17-related Hepatocerebral Mitochondrial DNA Depletion Syndrome]	MPV17	Neurological
68	Neuronal Ceroid Lipofuscinosis (CLN8-related)	CLN8	Neurological
69	Neuronal Ceroid Lipofuscinosis (MFSD8-related)	MFSD8	Neurological
70	Neuronal Ceroid Lipofuscinosis (TPP1-related)	TPP1	Neurological
71	Nijmegen Breakage Syndrome	NBN	Neurological
72	Omenn Syndrome (RAG2-related)	RAG2	Immunological
73	Ornithine Aminotransferase Deficiency	OAT	Ophthalmological
74	Ornithine Translocase Deficiency [Hyperomithinemia-Hyperammonemia -Homocitrullinuria (HHH) Syndrome]	SLC25A15	Metabolic
75	Pendred Syndrome	SLC26A4	Hearing, Endocrine
76	Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX1-related)	PEX1	Metabolic
77	Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX2-related)	PEX2	Metabolic
78	Phenylketonuria	PAH	Metabolic
79	Pontocerebellar Hypoplasia, Type 1A	VRK1	Neurological, Muscular
80	Pontocerebellar Hypoplasia, Type 2D	SEPSECS	Neurological
81	Pontocerebellar Hypoplasia, Type 2E	VPS53	Neurological
82	Primary Ciliary Dyskinesia (DNAH5-related)	DNAH5	Respiratory, Infertility
83	Primary Ciliary Dyskinesia (DNAI1-related)	DNAI1	Respiratory, Infertility
84	Primary Hyperoxaluria, Type 3	HOGA1	Renal, Metabolic
85	Pycnodysostosis	CTSK	Metabolic
86	Pyruvate Dehydrogenase Deficiency (PDHB-Related)	PDHB	Neurological, Metabolic
87	Retinal Dystrophy (RLBP1-related) [Bothnia Retinal Dystrophy]	RLBP1	Ophthalmological
88	Retinitis Pigmentosa 25 (EYS-related)	EYS	Ophthalmological
89	Retinitis Pigmentosa 59 (DHDDS-related)	DHDDS	Ophthalmological
90	Sanfilippo Syndrome, Type D [Mucopolysaccharidosis IIID]	GNS	Metabolic
91	Severe Combined Immunodeficiency, Type Athabaskan	DCLRE1C	Immunological
92	Severe Combined Immunodeficiency, X-Linked	IL2RG	Immunological
93	Sickle-Cell Disease	HBB	Hematological
94	Sjögren-Larsson Syndrome	ALDH3A2	Metabolic
95	Steroid-Resistant Nephrotic Syndrome	NPHS2	Renal
96	Stuve-Wiedemann Syndrome	LIFR	Skletal
97	Tay-Sachs Disease	HEXA	Metabolic
98	Usher Syndrome, Type 1F	PCDH15	Hearing
99	Usher Syndrome, Type 3	CLRN1	Hearing, Ophthalmological
100	Wolman Disease	LIPA	Metabolic, Hepatic

MicroGen NIPT - About the Test

MicroGen NIPT is a new-generation non-invasive prenatal test that utilizes cell-free DNA analysis of the mother's blood to detect genetic syndromes, such as trisomies and sex chromosome aneuploidies in the fetus. The test can be conducted after the 10th week of pregnancy and uses Next-Generation Sequencing (NGS) to evaluate fetal chromosomal abnormalities. MicroGen NIPT is currently among the most comprehensive NIPTs available, with the ability to screen all 23 pairs of chromosomes, identify Down syndrome, Edward syndrome, Patau syndrome, sex chromosome aneuploidies, 116 microdeletions, and 100 monogenic diseases.

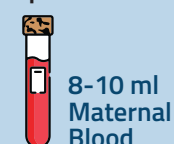
NIPT analysis process from maternal blood collection to reporting of test results



TEST OVERVIEW:

- Suitable after the 10th week of pregnancy
- Single pregnancy
- Twin pregnancy
- IVF pregnancies using self-eggs, donor eggs, or surrogate pregnancies
- Reports Fetal fraction
- Reports gender

Blood specimen:



8-10 ml
Maternal
Blood

Test method:

Cell-Free DNA extraction, massively parallel sequencing, and analysis of sequencing results to determine fetal aneuploidy.

Specimen:

8-10ml Maternal blood, STRECK BCT tube (18~25°C)

+ in case of **Monogenic** on request, **required 2 buccal swabs (from biological father)**

TAT:
7-10
Working days



WHO NEEDS PRENATAL TESTING?

- Pregnant women of any age who want to gain insight into their baby's development.
- Women who are 35 years or older (advanced maternal age).
- Women who have abnormal or positive serum screening results.
- Women with ultrasound findings of chromosomal abnormality.
- Family history of chromosomal abnormality or birth defects.
- Couples who have had a child with a chromosomal disorder.
- Couples with a history of infertility or pregnancy loss (miscarriages or stillbirths).

American College of Obstetricians and Gynecologists (ACOG), support the use of NIPT as the first line of screening for all pregnancies, irrespective of maternal age or baseline risk. [Obstet Gynecol. 2007;109(1):217-227. Obstet Gynecol. 2007;110(6):1459-1467].

TEST CRITERIA & LIMITATIONS:

	Trisomies	All Chromosome aberration	Sex Chromosome Aneuploidies	Microdeletions/ Duplications (4 or 116)	Presence of Y Chromosome	100 Monogenic Diseases
REGULAR PREGNANCY						
👶 Singleton	✓	✓	✓	✓	✓	✓
👶👶 Twin	✓	✗	✗	✓*	✓*	✓
👶👶 Vanishing Twins	✓	✓	✓	✓	✓	✓
IVF PREGNANCY (Self Egg Used)						
👶 Singleton	✓	✓	✓	✓	✓	✓
👶👶 Twin	✓	✗	✗	✓*	✓	✓
👶👶 Vanishing Twins	✓	✓	✓	✓	✓	✓
IVF PREGNANCY (Donor Egg Used or Surrogate)						
👶 Singleton	✓	✓	✓	✓	✓	✗
👶👶 Twin	✓	✗	✗	✗	✓	✗
👶👶 Vanishing Twins	✓	✓	✓	✓	✓	✗

Note: * For Twin pregnancies:

- Testing for 4 microdeletions can be done after the 10th week of pregnancy.
- Testing of comprehensive that covers 116 microdeletions and duplications is available, **After the 12th week of pregnancy.**
- For twin pregnancies, detection of Y chromosome can be done only after the 12th week. Please note that gender cannot be differentiated.

1. Singleton, vanishing twin, and IVF singleton pregnancies (using either the mother's own eggs, donor eggs, or a surrogate) are eligible for testing after the 10th week of gestation for all 23 autosomal chromosome aneuploidies, sex chromosome aneuploidies, and 116 microdeletions. For pregnancies resulting from vanishing twins, testing should occur four weeks after the vanishing event.
2. However, twin pregnancies after the 10th week of gestation are only eligible for testing for trisomies 21, 18, and 13, as well as selected 4 microdeletions. After the 12th week of gestation, they are eligible to undergo a comprehensive test for 22 autosomal chromosomes for aneuploidies, the 116 microdeletions/duplications, and Y chromosome detection. Nevertheless, they are not eligible for testing for sex chromosome aneuploidies.
3. **MicroGen Monogenic** offers testing for a selected number of pathogenic and likely pathogenic mutations associated with 100 monogenic diseases listed in the annexure (II) for singleton, twin, and vanished twin pregnancies, including in-vitro fertilization (IVF) pregnancies using the mother's own eggs, after the 10th week of gestation.
 - 3.1 The test is available for trisomies 21, 18, and 13, as well as selected 4 microdeletions. However, it is not eligible for testing for sex chromosome aneuploidies for twin and vanished twin gestations. Testing for pregnancies resulting from vanishing twins should occur four weeks after the vanishing event.
 - 3.2 Pregnancies achieved with egg/sperm donation or surrogacy cannot be tested with **MicroGen Monogenic**. Patients with malignancy or a history of malignancy, bone marrow or organ transplant, or recent transfusion are also ineligible for the test.
 - 3.3 Samples from both biological parents are required for the test to be performed, and the test result is only valid if the samples are collected from the biological parents. In some cases, the amount of fetal DNA present in maternal blood (fetal fraction) may be insufficient for analysis (less than 4%), and a redraw may be required.
4. **MicroGen** is a screening test, and its positive predictive value is not 100% reliable. Therefore, confirmatory testing is necessary before making any irreversible decisions about the pregnancy. Moreover, it does not rule out the possibility of other chromosomal abnormalities, birth defects, or complications.
5. There are various factors that can lead to false positive and false negative results, including chromosomal or sub chromosomal abnormalities, birth defects such as open neural tube defects or other conditions like autism, as well as maternal, fetal, and placental mosaicism, which refer to the presence of both normal and abnormal cells in the pregnancy. Other possible sources of inaccuracy include malignancies, prior history of cancer, bone marrow or organ transplant, recent blood transfusions. In addition, false negative or false positive results can occur in cases of fetal reduction, vanishing twin syndrome, or fetal demise.
6. Test failure can happen due to the low fetal fraction (less than 4%). It is also important to note that this test does not screen for polyploidy (such as triploidy).
7. For a definitive diagnosis, chorionic villus sampling, or amniocentesis would be necessary.

MicroGen NIPT - Genetic Conditions

Trisomies and monosomies:

A trisomy and a monosomy are types of numerical chromosome abnormalities that can result in certain birth defects. Normally, a person has 23 pairs of chromosomes or a total of 46 chromosomes in each cell, with one set of chromosomes inherited from the mother and the other from the father. However, in the case of numerical chromosome abnormalities, there may be either 45 or 47 chromosomes in each cell. These conditions can lead to various health issues and developmental disorders.

Monosomies:

Monosomy is a term used to describe the absence of one member of a pair of chromosomes. Therefore, there are 45 chromosomes in each cell of the body instead of the usual 46. Monosomy X, or Turner syndrome, occurs when a baby is born with only one X sex chromosome, rather than the usual pair (either two XX or one X and one Y sex chromosome).

Microdeletion/duplication syndrome:

Microdeletion and microduplication syndromes are disorders caused by submicroscopic deletions or duplications of contiguous genes on particular parts of chromosomes. The size and position of the deletion/duplication determine which clinical features are manifested and how severe they are. e.g., developmental delays, growth differences, behavioral problems, cardiac defects, neurological malformations etc.

