Micro Health Laboratories

.....for precious life

MICROGEN™

PREIMPLANTATION GENETIC TESTING

- PGT-A
- PGT-SR
- PGT-M

MICROGEN™ PrecisionDx

The Future of Targeted Treatment

MicroGen PGT Panels

- ☑ PGT- A
- ☑ PGT-SR
- ☑ PGT- M

1 MicroGen
Preimplantation Genetic Testing for Aneuploidies (PGT-A)

2 MicroGen
Preimplantation Genetic Testing for Structural Rearrangements (PGT-SR)

3 MicroGen
Preimplantation Genetic Testing for Monogenic disorders (PGT-M)

1 MicroGen Preimplantation Genetic Testing for Aneuploidies (PGT-A)

PGT-A (previously known as preimplantation genetic screening or PGS) involves checking embryos created through IVF or ICSI for chromosomal abnormalities. This process does not screen for specific genetic diagnoses but focuses on detecting aneuploidies, or abnormalities in the number of chromosomes, across all 24 chromosomes (22 autosomes and the X and Y chromosomes).

During MicroGen PGT-A analysis, a single cell or a small number of cells are removed from the embryo, and the DNA of these cells is tested to identify any chromosomal abnormalities. Only embryos without chromosomal abnormalities, such as trisomy 21 (Down syndrome) or monosomy X (Turner syndrome), are transferred back into the womb.

MicroGen PGT-A can detect a range of conditions including:

- Turner Syndrome (Monosomy X)
- Klinefelter Syndrome (XXY)
- Down Syndrome (Trisomy 21)
- Edwards Syndrome (Trisomy 18)
- Patau Syndrome (Trisomy 13)
- Other trisomies and monosomies that may increase the risk of implantation failure and miscarriage

2 MicroGen Preimplantation Genetic Testing for Structural Rearrangements (PGT-SR)

The MicroGen PGT-SR analysis only allows for blastocyst biopsy on Days 5, 6, or 7 of development and deferred transfer. A biopsy of 4-8 trophectoderm cells is required. Day 3 biopsy and fresh transfer are available only for Robertsonian translocations.

MicroGen PGT-SR can be used to identify various types of structural chromosomal rearrangements, including:

- **Inversion**
- **Reciprocal translocation**
- **Robertsonian translocation**

Individuals with balanced translocation have the correct number of chromosomes, but they are not in their usual positions. This condition may lead to:

- **Difficulty conceiving**
- **Recurrent pregnancy loss**
- **The possibility of having a child without the translocation or with a balanced form of the translocation (which is phenotypically healthy)**
- **The risk of having a child with an unbalanced form of the translocation, potentially causing extra or missing chromosomal material and resulting in physical or intellectual impairments.**

MicroGen PGT-SR can be used to identify embryos that are euploid or unbalanced forms of a rearrangement, thereby improving the chances of achieving a successful pregnancy and live birth.

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3 MicroGen Preimplantation Genetic Testing for Monogenic disorders (PGT-M)

MicroGen PGT-M is an advanced genetic testing method that significantly reduces the risk of having a child with an inherited genetic condition. By analyzing the DNA of each embryo, it allows for the selection of embryos with a lower risk of developing the condition, increasing the likelihood of a healthy outcome. MicroGen PGT-M linkage analysis identifies relevant alleles for single gene disorders within the family. Each MicroGen PGT-M test is performed on a case-by-case basis.

The MicroGen PGT-M analysis permits biopsy only during the blastocyst stage on Days 5, 6, or 7 of development. A biopsy of 4-8 trophectoderm cells is required.

MicroGen PGT-M is suitable for individuals at high risk of passing on a single-gene disorder, and may be considered if:

- **Both partners are carriers of the same autosomal recessive condition (e.g. cystic fibrosis)**
- **One partner is a carrier of an X-linked condition (e.g., Duchenne muscular dystrophy)**
- **One partner has an autosomal dominant condition (e.g., Huntington's disease)**
- **One partner has a mutation linked to a hereditary cancer syndrome (e.g., BRCA1 or BRCA2)**
- **You have had a previous child or pregnancy affected by a single-gene disorder**
- **You wish to perform HLA matching for tissue compatibility**

MICROGEN

PREIMPLANTATION GENETIC TESTING

- PGT-A
- PGT-SR
- PGT-M

MicroGEN PGT-M : List of gene panels for monogenic diseases / single-gene disorders.

Gene	Diesase research area
12q15	12q15 deletion syndrome
12q24.33	12q24.33 duplication syndrome
15q26.3	15q26.3 deletion syndrome
16p11.2	16p11.2 microduplication
17q12	17q12 deletion syndrome
1q21.1	1q21.1 microdeletion
22q11.2	22q11.2 deletion syndrome
2p25.3	2p25.3 duplication syndrome
MCCC2	3-methylcrotonyl-CoA carboxylase 2 deficiency
TRIP11	Achondrogenesis type 1A
FGFR3	Achondroplasia
CNGA3	Achromatopsia 2
PDE6C	Achromatopsia/cone-rod dystrophy
NOTCH1	Adams-Oliver syndrome 5
APRT	Adenine phosphoribosyltransferase deficiency
MUTYH	Adenomas, multiple colorectal
ADA	Adenosine deaminase deficiency
ABCD1	Adrenoleukodystrophy X-Linked
ABCD1+HLA	Adrenoleukodystrophy+ HLA histocompatibility
CSF1R	Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia (ALSP)
TREX1	Aicardi-Goutières syndrome
ADAR	Aicardi-Goutières syndrome 6
LARP7	Alazami syndrome
OCA2	Albinism
SLC16A2	Allan-Herndon-Dudley syndrome
POLG	Alpers-Huttenlocher syndrome
HBA1, HBA2	Alpha thalassemia
SERPINA1	Alpha-1 antitrypsin deficiency
COL4A5	Alport syndrome
COL4A3	Alport syndrome 2
COL4A4	Alport syndrome 2, autosomal recessive
ALMS1	Alstrom syndrome
FAM83H	Amelogenesis imperfecta, type IIIA
ALS2	Amyotrophic lateral sclerosis (ALS)
FUS	Amyotrophic lateral sclerosis (ALS)
UBE3A	Angelman syndrome
SERPING1	Angioedema, hereditary, 1 and 2
PAX6	Aniridia
PRKG1	Aortic aneurysm, familial thoracic
ACTA2	Aortic aneurysm, familial thoracic 6
FGF10	Aplasia of lacrimal and salivary glands
ASL	Argininosuccinic Aciduria
PKP2	Arrhythmogenic right ventricular dysplasia 9
NEB	Arthrogryposis multiplex congenita 6
ATM	Ataxia-telangiectasia
NPPA	Atrial fibrillation, familial
STAT3	Autoimmune disease, multisystem, infantile-onset, 1
LIPH	Autosomal recessive hypotrichosis
ASPM	Autosomal recessive primary microcephaly (ASPM)
WDR62	Autosomal recessive primary microcephaly (WDR62)
BBS1	Bardet-Biedl syndrome 1
BBS10	Bardet-Biedl syndrome 10
BBS4	Bardet-Biedl syndrome 4
TAZ	Barth syndrome

MicroGEN PGT-M : List of gene panels for monogenic diseases / single-gene disorders.

Gene	Diesase research area
SLC12A1	Bartter syndrome type I
HBB	Beta thalassemia
HBB+ HBA 1-2	Beta thalassemia and alpha thalassemia
HBB, HLA	Beta thalassemia and histocompatibility
UPB1	Beta-ureidopropionase deficiency
BTD	Biotinidase deficiency
FLCN	Birt-Hogg-Dube syndrome
FY	Blood group, Duffy system
RH	Blood group, RH system
OPN1MW	Blue cone monochromacy
SMCHD1	Bosma arhinia microphthalmia syndrome
TFAP2A	Branchio-oculofacial syndrome
EYA1	Branchiootic syndrome 1
SIX5	Branchio-oto-renal syndrome 2
BRCA1	Breast-ovarian cancer syndrome
BRCA2	Breast-ovarian cancer syndrome 2
SLC52A2	Brown-Vialetto-Van Laere syndrome 2
BCHE	Butyrylcholinesterase deficiency
NOTCH3	CADASIL
CPS1	Carbamoyl phosphate synthetase I deficiency
TNNI3K	Cardiac conduction disease with or without dilated cardiomyopathy
DSP	Cardiomyopathy
NEXN	Cardiomyopathy, dilated, 1CC
DES	Cardiomyopathy, dilated, 1I
TNNC1	Cardiomyopathy, dilated, 1Z
FLNC	Cardiomyopathy, familial hypertrophic, 26
LDB3	Cardiomyopathy, hypertrophic, 24
SLC22A5	Carnitine deficiency
COMP	Carpal tunnel syndrome 2
RMRP	Cartilage-hair hypoplasia
GJA8	Cataract
TDRD7	Cataract 36
LRP4	Cenani-Lenz syndactyly syndrome
RYR1	Central core disease
MTM1	Centronuclear myopathy
PDCD10	Cerebral cavernous malformation
CCM2	Cerebral cavernous malformations
SLC6A8	Cerebral creatine deficiency syndrome 1
SLC6A8	Cerebral creatine deficiency syndrome 1
SLC6A8	Cerebral creatine deficiency syndrome 1
MFN2	Charcot-Marie-Tooth disease type 2A2
SLC12A6	Charcot-Marie-Tooth disease, axonal, type 2II
PNKP	Charcot-Marie-Tooth disease, type 2B2
SH3TC2	Charcot-Marie-Tooth disease, type 4C
GJB1	Charcot-Marie-Tooth neuropathy 1
PMP22 (CMT1A)	Charcot-Marie-Tooth type 1A and 1E
MPZ (CMT1B)	Charcot-Marie-Tooth type 1B
ARSE	Chondrodysplasia punctata, X-linked recessive
VPS13A	Choreoacanthocytosis
CHM	Choroideremia
15q13.3	Chromosome 15q13.3 microdeletion syndrome
16p13.11	Chromosome 16p13.11 duplication syndrome
22q11.1	Chromosome 22q11.1q11.21 duplication
Xq21.1	Chromosome Xq21.1 duplication syndrome

MicroGEN PGT-M : List of gene panels for monogenic diseases / single-gene disorders.

Gene	Diesase research area
10q26	Chromosomic region
1p36	Chromosomic region
DNAH11	Ciliary dyskinesia, primary, 7, with or without situs inversus
DNAI2	Ciliary dyskinesia, primary, 9
RUNX2	Cleidocranial dysplasia
CLN3	CLN3 disease
CC2D2A	COACH syndrome 2
ERCC8	Cockayne syndrome, type A
COG5	COG5-congenital disorder of glycosylation
VPS13B	Cohen syndrome
COL12A1	COL12A1 disorder-related
RAG1	Combined immunodeficiency due to RAG1 deficiency
ACSF3	Combined malonic and methylmalonic aciduria
FARS2	Combined oxidative phosphorylation deficiency 14
CYP21A2	Congenital adrenal hyperplasia
PMM2	Congenital disorder of glycosylation
COG6	Congenital disorder of glycosylation, type III
RYR1	Congenital myopathy 1B, autosomal recessive
SLC26A3	Congenital secretory diarrhea, chloride type
PRRT2	Convulsions, familial infantile, with paroxysmal choreoathetosis
RAD21	Cornelia de Lange syndrome 4
PTEN	Cowden syndrome 1
ERF	Craniosynostosis
5p	Cri-du-chat syndrome
FGFR2	Crouzon syndrome
CFTR	Cystic fibrosis
SLC3A1	Cystinuria
ATP2A2	Darier disease
GJB6	Deafness
MYO7A	Deafness, autosomal dominant 11
OTOA	Deafness, autosomal recessive 22
SLC26A4	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct
TMIE	Deafness, autosomal recessive 6
PPP1R21	Decreased viability
IGFALS	Deficiency of acid-labile subunit
15q11.2	Deletion 15q11.2 (PWS/AS region)
CLCN5	Dent disease 1
ATN1	Dentatorubral-pallidoluysian atrophy
DSPP	Dentin dysplasia, type II
DEPDC5	Developmental and epileptic encephalopathy 111
SCN8A	Developmental and epileptic encephalopathy 13
SZT2	Developmental and epileptic encephalopathy 18
KCNA2	Developmental and epileptic encephalopathy 32
CACAN1A	Developmental and epileptic encephalopathy 42
UBA5	Developmental and epileptic encephalopathy 44
SCN1B	Developmental and epileptic encephalopathy 52, atrial fibrillation, familial, 13
GABBR2	Developmental and epileptic encephalopathy 59
ARHGEF9	Developmental and epileptic encephalopathy 8
OTOF	DFNB9 nonsyndromic hearing loss
HNF1A	Diabetes mellitus insulin-dependent
WNT2B	Diarrhea 9
SLC26A2	Diastrophic dysplasia
CDH1	Diffuse gastric and lobular breast cancer
DPYD	Dihydropyrimidine dehydrogenase deficiency

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Gene	Diesase research area
LMNA	Dilated cardiomyopathy
TNNT2	Dilated cardiomyopathy
SCN5A	Dilated cardiomyopathy 1E
TTN	Dilated cardiomyopathy 1G
ECEL1	Distal arthrogryposis type 5D
SALL4	Duane-radial ray syndrome
DMD	Duchenne muscular dystrophy
15q11	Duplication of 15q 11 region
ELP1	Dysautonomia, familial
KMT2B	Dystonia 28, childhood-onset
GCH1	Dystonia, DOPA-responsive
COL7A1	Dystrophic epidermolysis bullosa
CYP1B1	Early-onset glaucoma
TNXB	Ehlers-Danlos syndrome, classical-like
COL3A1	Ehlers-Danlos syndrome, vascular type
EVC, EVC2	Ellis-van Creveld syndrome
EMD	Emery-Dreifuss muscular dystrophy 1, X-linked
NR2E3	Enhanced S-cone syndrome
LAMA3	Epidermolysis bullosa
KRT14	Epidermolysis bullosa simplex (KRT14)
KRT5	Epidermolysis bullosa simplex (KRT5)
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg)
SLC13A5	Epileptic encephalopathy, early infantile, 25
MATN3	Epiphyseal dysplasia, multiple, 5
SLC1A3	Episodic ataxia, type 6
KCNA1	Episodic ataxia/myokymia syndrome
SCN10A	Episodic pain syndrome, familial, 2
EXT1	Exostoses type 1
EXT2	Exostoses type 2
GLA	Fabry disease
D4Z4	Facioscapulohumeral muscular dystrophy (FSHD)
F5	Factor V deficiency
F11	Factor XI deficiency, autosomal recessive
APC	Familial adenomatous polyposis (FAP)
STXBP2	Familial hemophagocytic lymphohistiocytosis
MEFV	Familial Mediterranean fever
BRIP1	Fanconi anemia
FANCA	Fanconi anemia
SLC2A2	Fanconi-Bickel syndrome
RAPSN	Fetal akinesia deformation sequence 2
RASPN	Fetal akinesia deformation sequence 2
KIF21A	Fibrosis of extraocular muscles
FKRP	FKRP-related muscular dystrophy
FMR1	Fragile X
C9orf72	Frontotemporal dementia and/or ALS 1
GRN	Frontotemporal lobar degeneration with ubiquitin-positive inclusions
ALDOB	Fructose intolerance
GALT	Galactosemia
GBA	Gaucher disease
GORAB	Geroderma osteodysplasticum
SLC12A3	Gitelman syndrome
ITGA2B	Glanzmann thrombasthenia
SATB2	Glass syndrome
G6PD	Glucose-6-phosphate dehydrogenase deficiency

MicroGEN PGT-M : List of gene panels for monogenic diseases / single-gene disorders.

Gene	Diesase research area
FTCD	Glutamate formiminotransferasa deficiency
GCDH	Glutaric acidemia type I
ETFDH	Glutaric acidemia type II
GLDC	Glycine encephalopathy
AMT	Glycine encephalopathy 2
SLC37A4	Glycogen storage disease I
GAA	Glycogen storage disease II
GBE1	Glycogen storage disease IV
AGL	Glycogen storage disease type III
PHKB	Glycogen storage disease type IX
PHKA2	Glycogen storage disease, type IXa2
GLB1	GM1 ganglioside
GHR	Growth hormone insensitivity
ATAD3A	Harel-Yoon syndrome
UNC13D	Hemophagocytic lymphohistiocytosis 3
F8	Hemophilia A
F9	Hemophilia B
F12	Hereditary angioedema type 3
MSH6	Hereditary colorectal cancer
MSH6	Hereditary colorectal cancer
HFE	Hereditary hemochromatosis type 1
FH	Hereditary leiomyomatosis and renal cell cancer (HLRCC)
FGA	Hereditary renal amyloidosis
ANTXR2	Hereditary systemic hyalinosis
MMP21	Heterotaxy, visceral, 7, autosomal
HLA	Histocompatibility
HMGCL	HMGCL deficiency
SHH	Holoprosencephaly 3
TBX5	Holt-Oram syndrome
CBS	Homocystinuria
MTHFR	Homocystinuria
MTR	Homocystinuria-megaloblastic anemia
HPA 1, 5 and 15	Human platelet antigen system 1, 5 and 15
HTT	Huntington
L1CAM	Hydrocephalus
MPDZ	Hydrocephalus, congenital, 2
LDLR	Hypercholesterolemia familial 1
APOB	Hypercholesterolemia, familial, 2
AGXT	Hyperoxaluria, primary, type 1
PRODH	Hyperprolinemia, type I
ELAC2	Hypertrophic cardiomyopathy
MYBPC3	Hypertrophic cardiomyopathy
MYL2	Hypertrophic cardiomyopathy 10
ANOS1	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1)
WDR11	Hypogonadotropic hypogonadism 14 with or without anosmia
SEMA3A	Hypogonadotropic hypogonadism 16 with or without anosmia
FGFR1	Hypogonadotropic hypogonadism 2 with or without anosmia
FGF8	Hypogonadotropic hypogonadism 6 with or without anosmia
GNRHR	Hypogonadotropic hypogonadism 7 without anosmia
EDA	Hypohidrotic ectodermal dysplasia
CACNA1S	Hypokalemic periodic paralysis, type 1
CLDN19	Hypomagnesemia 5, renal, with ocular involvement
ALPL	Hypophosphatasia
PHEX	Hypophosphatemic rickets

MicroGEN PGT-M : List of gene panels for monogenic diseases / single-gene disorders.

Gene	Diesase research area
TBCK	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3
FLG	Ichthyosis vulgaris
ABCA12	Ichthyosis, congenital
IL7R	Immunodeficiency 104, severe combined
NFKB1	Immunodeficiency, common variable, 12
IKBKG	Incontinentia pigmenti
NBAS	Infantile liver failure syndrome 2
GRIA3	Intellectual developmental disorder
SYNGAP1	Intellectual developmental disorder
ELP2	Intellectual developmental disorder, autosomal recessive 58
DelXq24	Intellectual developmental disorder, X-linked syndromic, Cabezas type
HUWE1	Intellectual developmental disorder, X-linked syndromic, Turner type
THOC2	Intellectual disability
ABCB4	Intrahepatic cholestasis type 3
IVD	Isovaleric acidemia
DYNC2H1	Jeune syndrome
CPLANE1 (C5orf42)	Joubert syndrome
TCTN2	Joubert syndrome
TMEM216	Joubert syndrome 2 / Meckel syndrome 2
AHI1	Joubert syndrome 3
NPHP1	Joubert syndrome 4
TMEM67	Joubert syndrome 6
LAMB3	Junctional epidermolysis bullosa
KDM6A	Kabuki syndrome
KMT2D	Kabuki syndrome (KMT2D)
NAGA	Kanzaki disease
KCNT1	KCNT1-Related epilepsy
KEL	Kell Blood group
AR	Kennedy disease
TBCE	Kenny-Caffey syndrome type 1
GALC	Krabbe disease
LAMA2	LAMA2-related muscular dystrophy
TGM1	Lamellar ichthyosis
RPGRIP1	Leber congenital amaurosis
TULP1	Leber congenital amaurosis 15
RPE65	Leber congenital amaurosis 2
LCA5	Leber congenital amaurosis 5
HPRT1	Lesch-Nyhan syndrome
RUNX1	Leukemia, acute myeloid
UFM1	Leukodystrophy, hypomyelinating, 14
TP53	Li-Fraumeni syndrome
ECM1	Lipoid proteinosis
TGFBR1	Loeys-Dietz syndrome
TGFB2	Loeys-Dietz syndrome 4
KCNQ1	Long QT syndrome 1
SCN5A	Long QT syndrome 3
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency
TSC2	Lymphangioleiomyomatosis, somatic
PRF1	Lymphoma, non-Hodgkin
MSH2	Lynch syndrome
PMS2	Lynch syndrome 4
EPCAM	Lynch syndrome 8
TBC1D7	Macrocephaly/megalencephaly syndrome, AR
POLD1	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome

MicroGEN PGT-M : List of gene panels for monogenic diseases / single-gene disorders.

Gene	Diesase research area
BCKDHB	Maple syrup urine disease, type Ib
FBN1	Marfan syndrome
MKS1	Meckel Gruber syndrome
CEP290	Meckel-Gruber syndrome
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency
STK11	Melanoma, malignant, somatic
MTTL1	MELAS syndrome
ADAT3	Mental retardation
SETD5	Mental retardation, autosomal dominant 23
TUSC3	Mental retardation, autosomal recessive 7
ATRX	Mental retardation-hypotonic facies syndrome, X-linked
PTPN11	Metachondromatosis
ARSA	Metachromatic leukodystrophy
MMAB	Methylmalonic Acidemia
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II
CENPJ	Microcephaly
RTTN	Microcephaly, short stature, and polymicrogyria with seizures
OTX2	Microphthalmia, syndromic 5
MYH7	Miopathy
MLH1	Mismatch repair cancer syndrome
MLH1	Mismatch repair cancer syndrome
RFX6	Mitchell-Riley syndrome
NDUFS8	Mitochondrial complex I deficiency, nuclear type 2
NUBPL	Mitochondrial complex I deficiency, nuclear type 21
SCO2	Mitochondrial complex IV deficiency, nuclear type 2
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
ECHS1	Mitochondrial syndrome
GALNS	Mucopolysaccharidosis IV
IDUA	Mucopolysaccharidosis type I (MPS I)
IDS	Mucopolysaccharidosis type II (MPS II–Hunter syndrome)
SGSH	Mucopolysaccharidosis type IIIA
ARSB	Mucopolysaccharidosis type VI
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3
MEN1	Multiple endocrine neoplasia
RET	Multiple endocrine neoplasia type 2
NFU1	Multiple mitochondrial dysfunctions syndrome 1
CHRNG	Multiple pterygium syndrome
NOG	Multiple synostoses syndrome 1
CAPN3	Muscular dystrophy, limb-girdle
DYSF	Muscular dystrophy, limb-girdle, autosomal recessive 2
SGCA	Muscular dystrophy, limb-girdle, autosomal recessive 3
POMK	Muscular dystrophy-dystroglycanopathy
POMT1	Muscular dystrophy-dystroglycanopathy
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11)
CHRNE	Myasthenic syndrome, congenital
SLC5A7	Myasthenic syndrome, congenital, 20, presynaptic
CLCN1	Myotonia congenita
CNBP	Myotonic dystrophy 2
ABCC8, KCNJ11	Neonatal diabetes mellitus
NPHS2	Nephrotic syndrome, type 2
PLA2G6	Neurodegeneration with brain iron accumulation 2B
WDR45	Neurodegeneration with brain iron accumulation 5

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Gene	Diesase research area
BRAT1	Neurodevelopmental disorder with cerebellar atrophy
LNPK	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum
SPATA5	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities
DEAF1	Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures
RERE	Neurodevelopmental syndrome
NF1	Neurofibromatosis type 1
ELANE	Neutropenia, severe congenital 1
NPC1	Niemann-Pick disease (NPC1)
SMPD1	Niemann-Pick disease (SMPD1)
GNE	Nonaka myopathy
GJB2	Nonsyndromic hearing Loss, DFNB1/DFNA3
LZTR1	Noonan syndrome 10
LEPR	Obesity, morbid, due to leptin receptor deficiency
SLC45A2	Oculocutaneous albinism
TYR	Oculocutaneous albinism
GJA1	Oculodentodigital dysplasia
CPABPN1	Oculopharyngeal muscular dystrophy
GIPC3	Oculopharyngodistal myopathy 2
WNT10A	Odonto-onycho-dermal dysplasia
RAG2	Omenn syndrome
OTC	Ornithine transcarbamylase deficiency
COL1A1	Osteogenesis imperfecta
COL1A1	Osteogenesis imperfecta (COL1A1)
COL1A2	Osteogenesis Imperfecta (COL1A2)
IFITM5	Osteogenesis imperfecta, type V
P3H1	Osteogenesis imperfecta, type VIII
AMER1	Osteopathia striata with cranial sclerosis
OSTM1	Osteopetrosis, AR 5
TNFSF11	Osteopetrosis, autosomal recessive 2
KRT9	Palmoplantar keratoderma, epidermolytic
PTF1A	Pancreatic agenesis 2
SDHC	Paraganglioma and gastric stromal sarcoma
SDHD	Paraganglioma and gastric stromal sarcoma
SDHB	Paragangliomas
SCN4A	Paramyotonia
PLP1	Pelizaeus-Merzbacher disease
PEX1	Peroxisome biogenesis disorder 1A (Zellweger)
PKD1	Polycystic kidney disease 1
PKD2	Polycystic kidney disease 2
PKHD1	Polycystic kidney disease AR
ADGRG1	Polymicrogyria, bilateral frontoparietal
BMPR1A	Polyposis, juvenile intestinal
TSEN54	Pontocerebellar hypoplasia
RARS2	Pontocerebellar hypoplasia, type 6
IRF6	Popliteal pterygium syndrome 1
DNAH5	Primary ciliary dyskinesia/heterotaxy
COQ4	Primary coenzyme Q10 deficiency
ABCB11	Progressive familial intrahepatic cholestasis
AIMP2	Progressive neurodevelopmental disorder
PEPD	Prolinasa deficiency
GNAS	Pseudohypoparathyroidism 1A
ABCC6	Pseudoxanthoma elasticum
PAH	Pulmonary arterial hypertension
ABCA3	Pulmonary surfactant dysfunction due to ABCA3 deficiency

MicroGEN PGT-M : List of gene panels for monogenic diseases / single-gene disorders.

Gene	Diesase research area
PKLR	Pyruvate kinase deficiency
RAB3GAP1	RAB18 deficiency
STS	Recessive X-linked ichthyosis
HNF1B	Renal cysts and diabetes syndrome
ACE	Renal tubular dysgenesis
CRB1	Retinitis pigmentosa
RHO	Retinitis pigmentosa
FAM161A	Retinitis pigmentosa 1
RP1	Retinitis pigmentosa 1
EYS	Retinitis pigmentosa 25
RPGR	Retinitis pigmentosa 33
SNRNP200	Retinitis pigmentosa 33
SNRNP200	Retinitis pigmentosa 33
RB1	Retinoblastoma
RS1	Retinoschisis
GNPAT	Rhizomelic chondrodysplasia punctata, type 2
TWIST1	Saethre-Chotzen syndrome
HEXB	Sandhoff disease, infantile, juvenile, and adult forms
TRPV4	Scapuloperoneal spinal muscular atrophy
SMARCAL1	Schimke immuno-osseous dysplasia
JAK3	SCID, T-negative/B-positive type
ALOX12B	Self-healing collodion baby
KCNH2	Short QT syndrome
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis
ACADS	Short-chain acyl-CoA dehydrogenase (SCAD) deficiency
SBDS	Shwachman-Diamond syndrome 1
GPC3	Simpson-Golabi-Behmel syndrome, type 1
ABCG5	Sitosterolemia
ABCG8	Sitosterolemia 1
MTOR	Smith-Kingsmore syndrome
DHCR7	Smith-Lemli-Opitz syndrome
NSD1	Sotos syndrome
KIF1C	Spastic ataxia 2, autosomal recessive
AP4M1	Spastic paraplegia
BSCL2	Spastic paraplegia
SACS	Spastic paraplegia
ZFYVE26	Spastic paraplegia 15, autosomal recessive
ATL1	Spastic paraplegia type 3A
SPAST	Spastic paraplegia type 4
SMN1	Spinal muscular atrophy
PPP2R2B	Spinocerebellar ataxia 12
AFG3L2	Spinocerebellar ataxia 28
ATXN3	Spinocerebellar ataxia 3
CACNA1A	Spinocerebellar ataxia 6 (SCA6)
ATXN-7	Spinocerebellar ataxia 7
ATXN8OS	Spinocerebellar ataxia 8
ATXN1	Spinocerebellar ataxia type 1
ATXN2	Spinocerebellar ataxia type 2
WWOX	Spinocerebellar ataxia, AR 12
COL2A1	Spondyloepiphyseal dysplasia
TRAPPC2	Spondyloepiphyseal dysplasia tarda
PRPH2	Stargardt disease
ABCA4	Stargardt disease 1
DMPK	Steinert disease (Myotonic dystrophy type 1)

MicroGen PGT-M : List of gene panels for monogenic diseases / single-gene disorders.

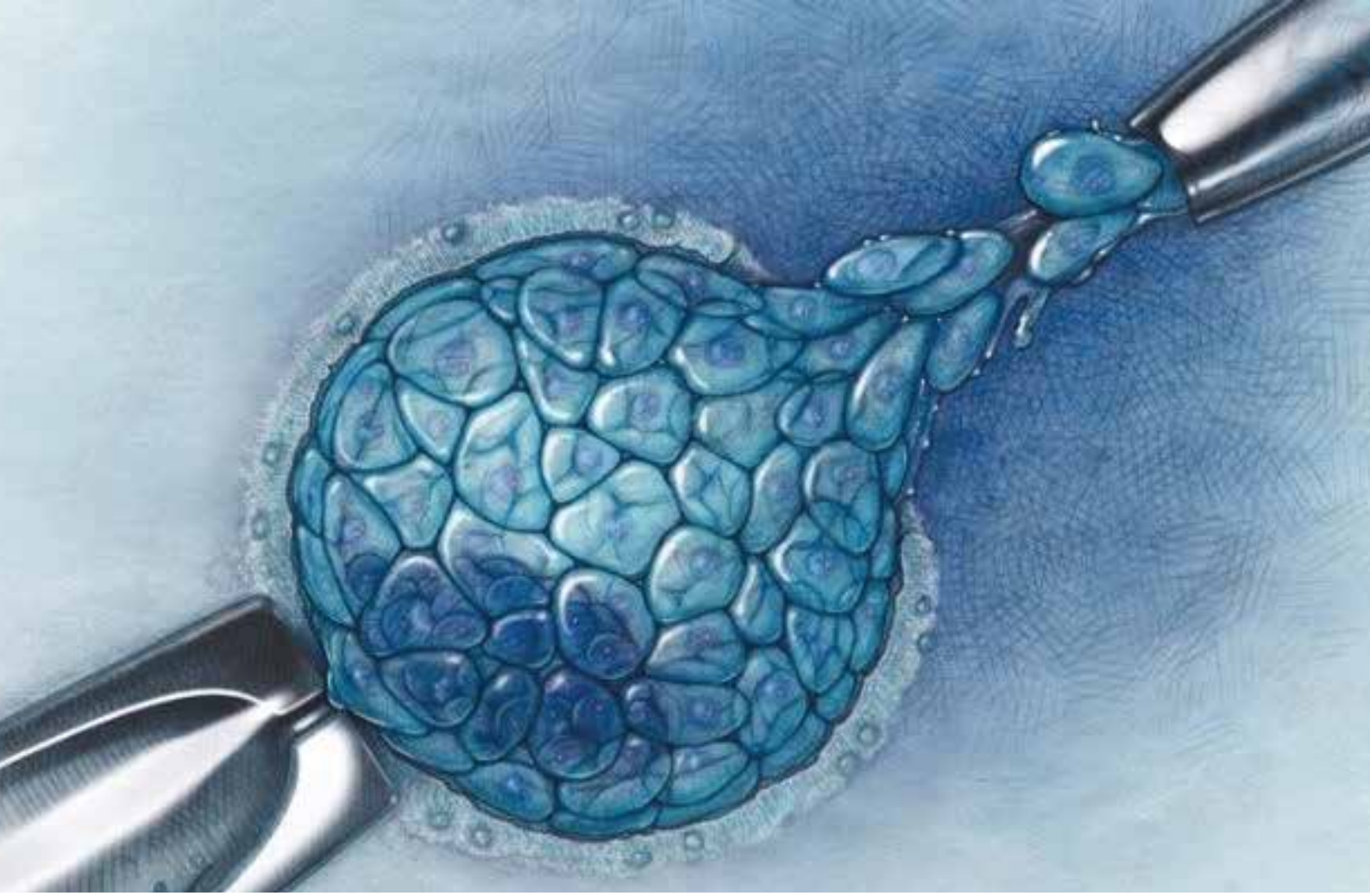
Gene	Diesase research area
COL11A1	Stickler syndrome
ELN	Supravalvular aortic stenosis and cutis laxa
PALB2	Susceptibility to breast and pancreatic cancer
CHEK2	Susceptibility to breast cancer
RAD51C	Susceptibility to breast-ovarian cancer
RAD51C	Susceptibility to breast-ovarian cancer, familial
ANXA5	Susceptibility to/recurrent pregnancy loss
HEXA	Tay-Sachs disease
ENG	Telangiectasia, hereditary hemorrhagic, type 1
ACVRL1	Telangiectasia, hereditary hemorrhagic, type 2
C12orf57	Temtamy syndrome
RBM8A	Thrombocytopenia-absent radius syndrome
F2	Thrombophilia 1 due to thrombin defect
DUOX2	Thyroid dyshormonogenesis 6
SOX6	Tolchin-Le Caignec syndrome
PAX9	Tooth agenesis, selective, 3
TTR	Transthyretin amyloidosis
TCOF1	Treacher-Collins syndrome
TSC1	Tuberous sclerosis
UMOD	Tubulointerstitial kidney disease, autosomal dominant, 1
COL6A2	Ullrich congenital muscular dystrophy 1
COL6A3	Ullrich congenital muscular dystrophy 1
COL6A1	Ullrich myopathy
USH1C	Usher syndrome, type 1C
USH2A	Usher syndrome, type 2A
ADGRV1	Usher syndrome, type 2C
RYR2	Ventricular tachycardia
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
ACTG2	Visceral myopathy 1
BEST1	Vitelliform macular dystrophy
VHL	Von Hippel-Lindau syndrome
POMGNT2	Walker-Warburg syndrome
WT1	Wilms tumor, type 1
ATP7B	Wilson disease
WAS	Wiskott-Aldrich syndrome
DCAF17 (C2orf37)	Woodhouse-Sakati syndrome
BTK	X-linked agammaglobulinemia (XLA)
IL2RG	X-linked combined immunodeficiency
Xp21.1	Xp21.1 deletion syndrome
Xp22.2	Xp22.2 duplication syndrome
Xp22.3	Xp22.3 duplication syndrome
PEX16	Zellweger syndrome
PEX2	Zellweger syndrome (PEX2)

Technology

MicroGen PGT is carried out using advanced **Next Generation Sequencing (NGS)** technology

TEST TAT

MicroGen PGT-A	Day 3	24 - 30 hours
	Day 5	7 - 10 Business Days
MicroGen PGT-SR		7 - 10 Business Days
MicroGen PGT-M		4 - 6 Weeks



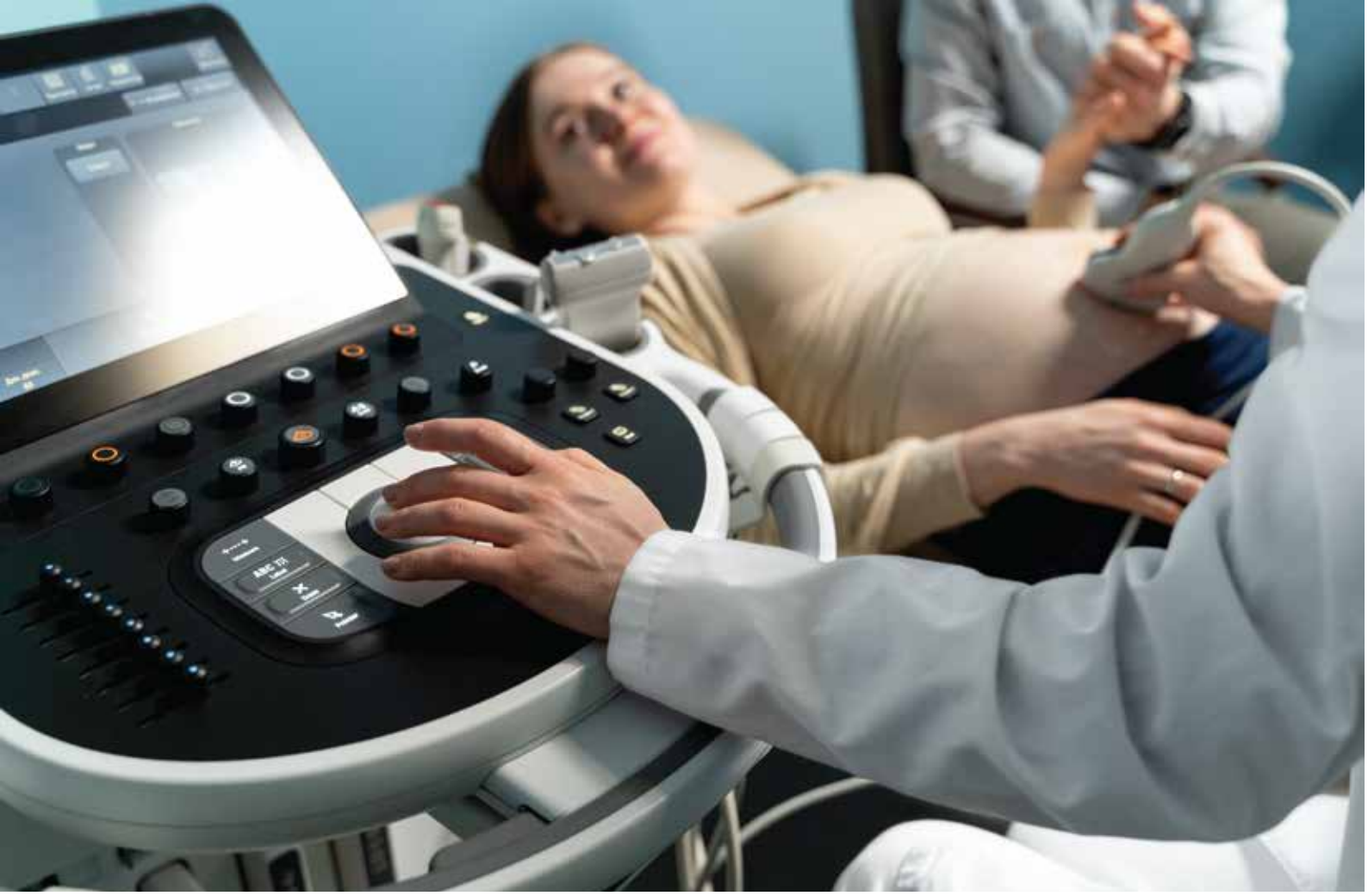
When Should a **Biopsy Be Performed?**

A biopsy can be conducted on the following days of embryo development.

Day 3 (Blastomere) or **Day 5 (Trophectoderm)**

However, Day 5 biopsy is generally preferred for PGT for Aneuploidy (PGT-A) due to several advantages:

- **Sufficient Cell Quantity:**
  By Day 5, there are enough cells available to isolate DNA, which enhances the accuracy of the test.
- **Detection of Mosaicism:**
  This stage allows for the detection of mosaicism in aneuploidies, improving the reliability of the results.
- **Vitrification:**
  Embryos can be rapidly frozen (vitrified) after biopsy, providing the clinician with the opportunity to optimize implantation conditions for the future transfer.



How to order the **Embryo Biopsy Kit**

Contact: Customer Support at **Micro Health Laboratories.**

Notice: Make your request at least **48 hr / 2 days** in advance.

Quantity: Specify the number of **Kits / Tubes** required.

Transportation: Kits will be delivered to the provided address at **room temperature.**

Check Contents: Verify that all kit components are intact before **proceeding with the biopsy.**

How to order the
Embryo Biopsy Kit - ☑ **TRF** (Test Request Form)

Micro Health Laboratories
.....for precious life

Test Request Form
International V-0.4

MICRO GEN™

PREIMPLANTATION GENETIC TESTING

*accurate information is necessary for a valid interpretation | please (✓) boxes

PATIENT INFORMATION GENERAL*

Name | Sex | QID | MRN / File No. | Ph No. | Date of Birth | Nationality

REFERRED BY*

Doctor / Hospital / Clinic / Lab - Account Code | Mail Id. | Ph No. | Seal / Signature

TEST REQUESTED | **CLINICAL INDICATIONS**

☐ PGT-A
Preimplantation Genetic Testing for **aneuploidy**

- ☐ Advanced Maternal Age
- ☐ Recurrent Miscarriage (No of failures) _____
- ☐ Recurrent Implantation failure (No of failures) _____
- ☐ Aneuploidy testing
- ☐ Sex-linked Disorder
- ☐ Male factor
- ☐ Gender Selection (please specify)
- ☐ Male ☐ Female

☐ PGT-SR
Preimplantation Genetic Testing for **Structural Rearrangements**

Note: The karyotype of the carrier will be required for PGT-SR

Karyotype(s): Patient ☐ Normal ☐ Abnormal
Partner ☐ Normal ☐ Abnormal

ISCN of Known Structural rearrangement (Abnormal Karyotype): _____

☐ PGT-M [Monogenic Disorders]
Preimplantation Genetic Testing for **Monogenic Disorders**

Monogenic Disease Prevention (Disease Name) :

Gene of interest :

Patient Condition Status : ☐ has the condition ☐ Carrier ☐ Non-carrier

Partner Condition Status : ☐ has the condition ☐ Carrier ☐ Non-carrier

Report : ☐ Report sex of Embryo(s) ☐ Do NOT report sex of Embryo(s)

Additional family history relating to the condition :

Others (Please Specify) :

PATIENT ETHNIC GROUP (Please select all that apply)

☐ Caucasian ☐ East Asian ☐ South Asian ☐ Arab / Middle East ☐ Ashkenazi J. ☐ Hispanic ☐ Romani ☐ Afro - _____

☐ Unknown ☐ Other _____

SPECIMEN INFORMATION

Donor Details: ☐ Donor eggs ☐ Donor sperm ☐ No Donor* ☐☐☐ Egg retrieval date (if using own eggs):

No. of Fertilised eggs: _____ No. of Biopsied embryos: _____

Fertilisation method: ☐ IVF ☐ ICSI BLANK PROVIDED: ☐ Yes ☐ No

Date of biopsy: ☐☐☐ Date/time planned for embryo transfer: ☐☐☐ (Only mandatory for transfers in the same cycle)

Embryo transfer: ☐ Fresh cycle (transfer in the same cycle) ☐ Frozen cycle (transfer in other cycle)

Type of biopsy: ☐ Day-3 Blastomere ☐ Day 5 / 6 / 7 Trophectoderm

FOR LABORATORY USE ONLY		ACKNOWLEDGEMENT
TRIAGE REMARKS	TRANSPORT TEMPERATURE ☐ Ambient Temperature ☐ Refrigerated (2° to 8°C) ☐ Frozen (-20°C)	SAMPLE COLLECTED / RECEIVED @ LAB Date ☐☐ ☐☐ ☐☐☐☐ Time ☐☐ ☐☐ am / pm	SAMPLE RECEIVED / VALIDATED BY Technician Name / Signature

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Diagnosis Education Research | Joint Commission International JCI Accredited Laboratory | Micro Health Laboratories

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PREIMPLANTATION GENETIC TESTING

- ☑ PGT-A
- ☑ PGT-SR
- ☑ PGT-M

In in vitro fertilization (IVF), with or without intracytoplasmic sperm injection (ICSI), the selection of the best embryo(s) for transfer is primarily based on morphological assessment, which includes evaluating the number of cells, cell regularity, and the presence of cell fragments. However, despite transferring 'good quality' embryos, nearly two-thirds of couples do not achieve pregnancy. One suspected cause is that these embryos may have an abnormal number of chromosomes (aneuploidy).

Preimplantation Genetic Testing for Aneuploidy (PGT-A) is a technique used to analyze the chromosomal number in IVF embryos. During PGT-A, a polar body (a byproduct of maternal meiosis) or a few cells from the embryo are obtained through biopsy and tested. Only embryos with a normal chromosomal count, known as 'euploid embryos,' are selected for transfer into the uterus. This approach aims to increase the live birth rate per IVF cycle. While earlier studies using fluorescence in situ hybridization (FISH) found PGT-A ineffective in improving live birth rates, advancements in PGT-A techniques—such as testing at different stages of embryo development using array comparative genomic hybridization (aCGH) or next-generation sequencing (NGS).

At MHL, we offer NGS-based PGT, "MicroGen PGT," which is used to screen in vitro fertilized embryos for chromosomal abnormalities and genetic mutations. This testing can determine whether an embryo created through IVF is chromosomally normal (euploid). Selecting an euploid embryo for implantation can increase the likelihood of a healthy pregnancy and live birth while potentially reducing the number of IVF cycles and the time needed to achieve pregnancy.

MicroGen PGT is designed for

- **Individuals or couples undergoing in vitro fertilization (IVF)**
- **Those with a history of unsuccessful fertility treatments**
- **Women who have experienced one or more miscarriages**
- **Individuals with structural chromosomal rearrangements**
- **Women who have had a previous pregnancy involving chromosomal aneuploidy**
- **Women over the age of 35 undergoing IVF**
- **Patients undergoing assisted reproduction who have a personal or family history of high-risk single-gene conditions.**

Why recommend MicroGen PGT ?

MicroGen PGT identifies chromosomally normal embryos, which can:

- **Increase the chances of successful implantation and the likelihood of healthy live births**
- **Up to 40% of embryos that appear morphologically normal may still harbor aneuploidies.**
- **Lower the miscarriage rate.**
- **Reduce the number of IVF cycles a patient needs to undergo.**
- **Help achieve pregnancy earlier compared to cycles without PGT.**
- **Promote safer pregnancies with fewer maternal & perinatal complications by supporting single embryo transfers (SET).**

Limitations of
MicroGen PGT-A

MicroGen PGT-A is a screening test rather than a diagnostic test. This distinction arises because PGT-A examines only a small subset of cells that will develop into the placenta, from an embryo that has approximately 100 cells in total. Consequently, it cannot provide a definitive picture of the entire embryo's chromosomal composition. However, PGT-A offers the most accurate estimate available. As a screening test, we advise using Microgen NIPT (non-invasive prenatal testing) for any pregnancy resulting from an embryo that has undergone MicroGen PGT-A.

MicroGen PGT-A does not evaluate birth defects, inherited single-gene disorders like cystic fibrosis, multifactorial conditions such as autism, adult-onset diseases including diabetes or Alzheimer's disease, physical and mental traits such as intelligence or athleticism, or microdeletions and microduplications.
The key limitations of MicroGen PGT-A include an accuracy of approximately 98%, with potential risks of false positives, where an embryo might be incorrectly identified as abnormal, and false negatives, where an embryo deemed normal might still have chromosomal abnormalities. MicroGen PGT-A only tests cells obtained from an embryo biopsy, not the entire embryo, and cannot identify structural abnormalities (gains or losses of genetic material), chromosome losses or gains smaller than 10Mb, low levels of mosaicism (less than 30%), uniparental disomy (UDP), or complete chromosome set defects like haploidy or triploidy.

In rare instances, factors like human error or external conditions (such as weather or logistical issues) may prevent testing or result in the inability to obtain results. Even in such cases, only the biopsied sample is affected. In some cases, genetic testing may be affected by technical issues like improper biopsy techniques, loss of biopsied cells, or poor DNA quality.

Limitations of
MicroGen PGT-SR

MicroGen PGT-SR is a screening test rather than a diagnostic test. The accuracy of PGT-SR is estimated to be 98%. However, the test has several limitations. It cannot detect balanced translocation and chromosomal imbalances smaller than 6MB. Additionally, PGT-SR does not detect uniparental disomy (UPD) or assess for birth defects directly. For babies born through IVF, there is a general risk of birth defects ranging from 4-6%, which may arise from both genetic and non-genetic factors. Furthermore, PGT-SR does not identify single-gene disorders such as cystic fibrosis, sickle cell anemia, nor does it evaluate multifactorial conditions like autism, schizophrenia, or diabetes.

Limitations of
MicroGen PGT-M

MicroGen PGT-M is a screening test, not a diagnostic one. It cannot guarantee that a child will be chromosomally or genetically normal and should not be used as a substitute for prenatal testing. We recommend that prenatal testing be considered for all pregnancies resulting from embryos tested with MicroGen PGT-M.

PGT-M involves analyzing cells from the trophectoderm (which will develop into the placenta) rather than the inner cell mass (which will form the fetus). As a result, there may be differences between the genetic profile of the tested cells and that of the cells that will become the fetus.

It is crucial to understand the benefits, risks, and alternative options before deciding to proceed with MicroGen PGT-M. While there are risks, advances in technology have made MicroGen PGT-M a valuable tool for improving IVF success rates. In rare instances, factors like human error or external conditions (such as weather or logistical issues) may prevent testing or result in the inability to obtain results. Even in such cases, only the biopsied sample is affected.

Additionally, engaging in sexual intercourse during treatment may lead to a spontaneous pregnancy. Sperm from intercourse could potentially fertilize and implant an egg, either alongside or instead of the treatment process, which would invalidate the PGT results.