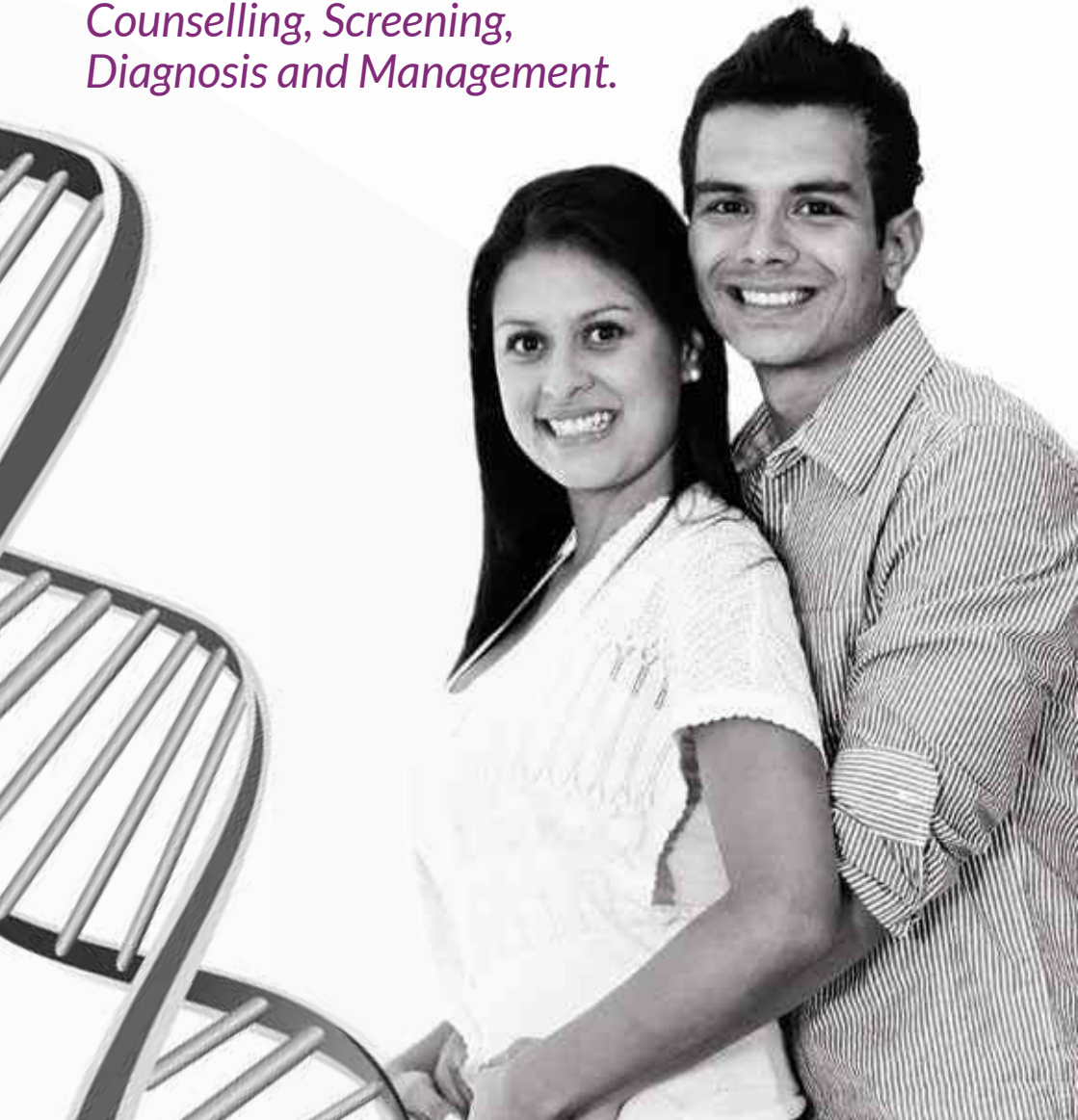
MEDGENOME

MedGenome
Center For Genetic Health Care

Leaders in Genetic Testing, Counselling, Screening, Diagnosis and Management.



MedGenome offers the most comprehensive range of Reproductive Genetic Testing service

- Non Invasive Prenatal Test
- Carrier Screening Test
- Preimplantation Genetic Screening/Diagnosis
- Products of Conception
- Karyotyping
- FISH
- Prenatal Diagnostic Services
- Genetic Counselling

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About us

Medical genetics plays an important role in all aspects of medical practice: prevention, screening, diagnosis and management. About 60% of pregnancy losses, 2-3% of all neonate and 50% of childhood deafness, blindness, mental retardation and 1 to 10% of all malignancies are directly due to genetic factors.

Dr. Hema Purandary, Director eminently qualified for conducting the centre, has been trained in all aspects of medical genetics at the most advanced centres/laboratories over the world (USA, UK and Europe). A team of physicians adds to the credit of our centre. Our skilled, trained and dedicated scientific officers and technical staff with state-of-the-art equipment ensure the output of quality work. The centre has vast experience of more than 5000 referrals/year for genetic testing.

Accreditations

The Centre has been accredited by nation and international bodies.

- NABL (National Accreditation Board for Testing and Calibration of Laboratories), Government of India.
- PCPNDT (Preconception and Prenatal Diagnostic Technique Act) – BMC /542.
- CAP Accredited (College of American Pathologists)

Benefits of choosing MedGenome - Centre for Genetic Health Care

- Fully accredited laboratory
- State-of-the-art equipment
- Range of genetic testing and counselling services
- Qualified, experienced and dedicated team of physicians, scientists and technologists
- Competitive fees
- All results interpreted by qualified medical geneticists
- Minimum turn-around time
- Report by email is default and printed on request
- Collection facilities – local and outstation
- All the facilities are under one roof

Services

Genetic Counselling: This is a critical support that assists affected and/or at – risk individuals, for themselves and their pregnancy to understand the nature of the genetic disorder, its risk of recurrence and the tests available to them for management & family planning. The cost effectiveness is also discussed. Counselling can be offered for a large spectrum of genetic disorders for individuals in any age group - preconception, prenatal, pediatric and oncology.

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Common indication for genetic testing/counselling

Preconception, preimplantation & prenatal.

- Consanguineous marriage
- Member of a high risk ethnic group
- Maternal disease
- Single gene disorder (Thalassemia, DMD)
- Infertility or patients on IVF programme
- Two or more pregnancy losses
- A previous child / family with birth defect
- Positive maternal serum screening test (Double / Triple / Quadruple)
- Abnormal ultrasound findings
- Women 35 years of age or older

Infants, children, adolescents and adults

- Development delay
- Ambiguous genitalia; puberty disorders
- Dysmorphic features, mental retardation
- Growth retardation
- Suspected metabolic disorder
- Blindness or deafness
- Primary and secondary amenorrhoea
- Cancer

Cytogenetic testing (Karyotyping, Chromosomal studies)

Cytogenetic testing (Karyotyping) can be done on various body tissues in prenatal or postnatal life. Numerical or structural abnormalities can lead to various phenotypic abnormalities.

Blood

Karyotyping on peripheral blood samples is done for confirmation of clinical diagnosis, management, recurrence risk estimation, for various reproductive options and prenatal diagnosis

Specimen: 2ml venous blood in sodium heparin vial. (transport at ambient temperature)

Bone marrow

Chromosomal pattern from the bone marrow in hematological malignancy helps the physician in giving confirmation of diagnosis, prognosis and management. Most of the abnormalities are acquired and show characteristics changes in remissions / relapses.

Specimen: 0.5ml of bone marrow in a sterile sodium heparinized vial. Include routine blood count and patient medication. In case sample is not adequate, an additional 2ml of peripheral blood in sterile sodium heparin vial is requested. (transport at ambient temperature).

Products of conception

60% of early fetal loss is due to chromosomal defects in the conceptus. Karyotyping of the products of conception assists in identifying the chromosomal factor, if any. And counselling and management for future pregnancies.

Specimen: Chorionic villi, or placental villi in tissue culture medium / balanced salt solution / normal saline or Cord / cardiac blood in sterile sodium heparin vial. attach clinical history and USG reports (Transport at ambient temperature). If Karyotype is normal chromosomal microarray can be offered.

Prenatal diagnosis

The tissues generally studied for prenatal diagnosis are chorionic villi, amniotic fluid and cord blood. All prenatal fetal tissue samplings are done as outpatient procedures, under ultrasound guidance. Patient needs to be adequately counselled for the choice, safety, specificity and sensitivity of the test. An informed written consent is a prerequisite. In-house facility for the obstetric procedure and sonography is available at the centre. The samples are also accepted through centres licensed for fetal tissue sampling. Prior discussion with MedGenome- Centre For Genetic Health Care before sampling is important.

For indications refer to genetic counseling section.

Chorionic villi

The tissue sampling is done by transcervical or transabdominal route between 10-12 weeks gestation. At a later gestation, placental biopsy (late CVS) is offered. (For enzyme assays and molecular diagnosis, carrier testing in the couple and / or mutations analysis in the affected child is essential.) Do not send these sample unless the case is discussed with Medical team of MedGenome- Centre For Genetic Health Care.

Specimen: 20mg of chorionic villi in tissue culture media (transport at ambient temp).

Amniotic fluid

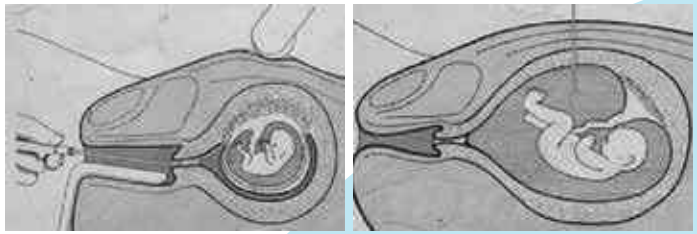
Obtained by amniocentesis between 15-18 weeks gestation.

Specimen: 20ml of amniotic fluid in provided sterile tubes (transport at ambient temperature).

Cord blood

Is indicated for rapid karyotyping during 2nd and 3rd trimester of pregnancy with abnormal ultrasound findings or when confirmation of karyotype of CVS or amniotic fluid is needed.

Specimen: 3ml cord blood in sodium heparinized vial. Verification at source that the blood is of fetal and not maternal origin (transport at ambient temperature).



Obstetrics procedures for prenatal diagnosis

Genetic screening

Prenatal screening

Different strategies are being devised to screen populations of pregnant mothers to help identify those at potential risk of a genetically abnormal fetus, targeting them of invasive diagnostic procedures if needed. These procedures include 1st and 2nd trimester.

1st and 2nd trimester sonography can pick up at-risk babies for genetic and non genetic syndromes. Genetic counselling and a follow-up test is suggested by doctor / genetic professional. Maternal serum screening is routinely suggested for trisomy 21, trisomy 18, NTD in 1st and 2nd trimester in conjunction with ultrasound evaluation. Pre and post test genetic counselling is advised. Specimen: 2ml maternal blood (information required:Maternal age, LMP, Assisted or normal conception) Ultrasound biometry and Number of fetuses, diabetic status and weight of the patient (transport at ambient temperature).

Claria from MedGenome

MedGenome is driven to enable clinicians to deliver the best outcomes to their patients. Our passion to deliver actionable insights to clinicians has resulted in the development of "Claria" - a suite of NGS (Next-Generation Sequencing) technology-based solutions for reproductive testing. Claria offers the most accurate Non-Invasive Prenatal Screening Test (NIPT), the Genetic Carrier Screening Test and the Preimplantation Genetic Screening/Diagnosis (PGS/PGD).

Claria NIPT

MedGenome Claria NIPT is a simple, safe and non-invasive prenatal screening test that provides assurance to expectant parents with accurate genetic information about their baby. This screening test can be performed from as early as the 9th week of a pregnancy. Compared to first trimester screening, Claria NIPT screens for more chromosomal abnormalities, has a higher sensitivity and lower false positive rate for the conditions screened.

Claria NIPT screens for microdeletions

Claria NIPT now covers clinically relevant microdeletions that occur in 1-1.7% of all structurally normal pregnancies. These sub-chromosomal abnormalities which collectively have a population incidence of approximately 1 in 1000 will result in severe physical and/or intellectual impairments.

Claria NIPT can screen for Twins, egg donor and s urrogate pregnancies

For the first time in India, Claria NIPT determines

- Zygosity information
- Individual foetal fractions for dizygotic twins
- Monosomy X risk for monozygotic twins

Claria Carrier Screening Test

Carrier detection and diagnosis for single gene disorders

The Claria Carrier Screening Test is based on Next-Generation Sequencing (NGS) and Multiplex Ligation Probe Amplification (MLPA) technologies. This enables you to detect disease-causing mutations in over 2000 genes which are responsible for the Autosomal Recessive and X-Linked Recessive genetic disorders.

The Claria Carrier Screening Test leverages the Indian population genetic variant database created by Sir Ganga Ram Hospital over the last twenty years. This has enabled MedGenome to develop a highly focused and cost-effective test to screen for diseases and genetic.

Molecular cytogenetics

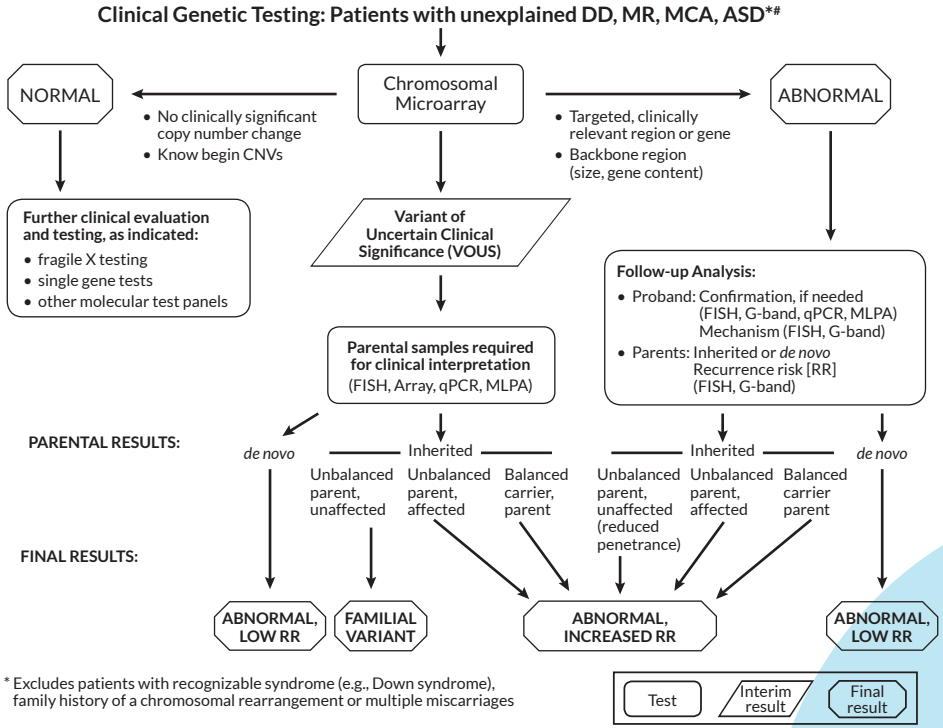
Fluorescent In Situ Hybridization (FISH)

FISH technique is used to detect numerical as well as minor structural re arrangements on all the above tissues as per the indications (prior discussion with the lab is essential). Currently it is available for fetal aneuploidy, micro deletion syndromes and oncology.

Chromosomal Microarray (CMA)

CMAs are a high resolution molecular genetic test which screen for very small gains and losses (copy number variants, in kilobase ranges) as well as loss of heterozygosity in the DNA of an individual. CMAs are commonly used to test for genetic abnormalities for individuals with unexplained developmental delay/intellectual disability (DD/ID), autism spectrum disorder (ASD) or multiple congenital anomalies (MCA).

Algorithm for CMA Testing in Patients with Unexplained DD, MR, MCA, and ASD.



* Excludes patients with recognizable syndrome (e.g., Down syndrome), family history of a chromosomal rearrangement or multiple miscarriages

Genetics of organ system diseases

Many organ system disorders have genetic basis, involving many medical specialities. This affect systematic development and functioning. Appropriate diagnosis helps in management of risk to family and progeny.

#Miller DT et al, "Consensus Statement: Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies" The American Journal of Human Genetics, 2010, May

Various panel tests are available with appropriate genetic counselling

Cardiology	Haematology	Neurology	Oncology	Immunology	Panorama™ Non invasive prenatal screening test
Metabolic disorders	Endocrinology	Nephrology	Ophthalmology	Connective tissue disorders & skeletal dysplasias	Preimplantation Genetic Screening

Training facility

The center provides didactic and hands-on training courses to science and medical graduates in Leukocyte Culture Basis Course, prenatal diagnosis (with approval from PCNDT), abortus study, advanced courses in cytogenetic of hematological disorders with FISH, advanced course in fluorescence in SITU hybridization, genetic counselling. The course includes directions for setting up a laboratory, quality control and management of a genetic centre.

Chromosome microarray (CMA), next generation sequencing (NGS), whole genome sequencing (WSG), whole exome sequencing (WES)

The continuing evolving technologies in genetics have introduced tests like chromosome microarray (CMA) which can detect small deletions and duplications (100kb resolution) as compared to conventional karyotypes (5MB). Tests like next generation sequencing (NGS), whole genome sequencing (WGS), whole exome sequencing (WES) is useful in cases where genetic etiology is suspected but phenotype does not match, or there is genetic heterogeneity of the disease, where multiple genes can be analyzed.

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MedGenome - Center for Genetic Health Care
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For further details
Call: 1800 103 3691 to learn more
Email: diagnostics@medgenome.com

Certificate Number MC-2054 (in lieu of M-0019)

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