DIAGNOSTIC EVALUATION

Each patient will undergo a diagnostic evaluation tailored to their own particular presenting signs and symptoms. The geneticist will establish a differential diagnosis and recommend appropriate testing.

1. Chromosome Studies

Chromosome studies are used in the general genetics clinic to determine a cause for developmental delay/mental retardation, birth defects, dysmorphic features, and/or autism. A large number of different methods have been developed for chromosome analysis:

- Karyotype
- Fluorescence in situ hybridization (FISH)
- Array comparative genomic hybridization.

2. Basic Metabolic Studies

Biochemical studies are performed to screen for imbalances of metabolites in the bodily fluid, usually the blood (plasma/serum) or urine, but also in cerebrospinal fluid (CSF).

3. Molecular Studies

- DNA sequencing
- DNA methylation analysis
- Next generation sequencing

SERVICES OFFERED

- Diagnostic evaluation.
- Management of inborn errors of metabolism,skeletal dysplasia, or lysosomal storage diseases.
- A prenatal genetics clinic to discuss risks to the pregnancy (advanced maternal age, teratogen exposure, family history of a genetic disease), test results (abnormal maternal serum screen,abnormal ultrasound), and/or options for prenatal diagnosis (typically amniocentesis or chorionic villus sampling).
- Support of a clinical geneticist or genetic counselor (cancer genetics, cardiovascular genetics, craniofacial or cleft lip/palate, hearing loss clinics, muscular dystrophy / neuro degenerative disorder clinics).

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GENETICS

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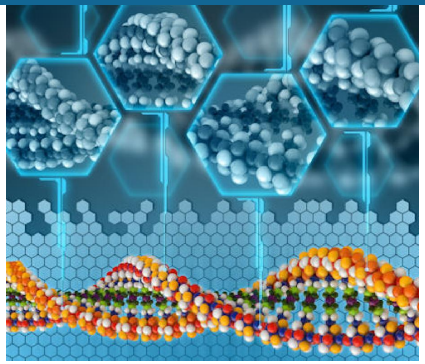
CIMS GENETICS



CIMS Genetics team works into the causes and inheritance of genetic disorders. We treat birth defects and dysmorphology, mental retardation, autism, and mitochondrial disorders, skeletal dysplasia, connective tissue disorders, cancer genetics, teratogens, and prenatal diagnosis. We treat or advice regarding neurologic, endocrine, cardiovascular, pulmonary, ophthalmologic, renal, psychiatric and dermatologic conditions.

Our clinical geneticists advice with particular attention to hereditary disorders. Examples of genetic syndromes that are commonly seen in patients include chromosomal rearrangements, Down syndrome, DiGeorge syndrome (22q11.2 Deletion Syndrome), Fragile X syndrome, Marfan syndrome, Neurofibromatosis, Turner syndrome, and Williams's syndrome.

GENETIC COUNSELLING

Genetic counseling is the process of providing information about genetic conditions, diagnostic testing, and risks in other family members, within the framework of nondirective counselling. Our genetic counsellors guide in family risk assessment and counselling of patients regarding genetic disorders.

CANCER AND GENETICS

If you have a family and/or personal history of the following, you should consider having a genetics evaluation.

- Two or more relatives with breast cancer on the same side of the family with at least one diagnosed before age of 50
- A blood relative with ovarian cancer
- A close blood relative with breast cancer before age of 45
- A blood relative with male breast cancer
- Breast cancer at or before the age of 50
- Triple-negative breast cancer at or before the age of 60
- Ovarian, fallopian tube, or primary peritoneal cancer at any age
- Male breast cancer at any age
- A second primary breast cancer

PAEDIATRICS/ PAEDIATRIC CARDIOLOGY AND GENETICS

Genetics evaluation is helpful in cases of

- Developmental delay
- Mental retardation
- Congenital abnormalities
- Dysmorphology (unusual physical features)
- Growth problems
- Certain syndromes (e.g., mental retardation, distinct facial features, and a heart defect or heart defects)
- Lysosomal Storage Diseases (LSDs)
- Inborn errors of metabolism

PHYSICIAN/ NEUROLOGY AND GENETICS

If you have a family and/or personal history of the following, you should consider having a genetics evaluation

- Known genetic conditions (e.g., Huntington disease, Myotonic dystrophy, Muscular Dystrophy, Charcot-Marie Tooth, Thrombophila)
- Genetic mutation carrier in the family (e.g., sickle cell anaemia, cystic fibrosis, Tay-Sachs disease)
- Known family history of genetic or developmental conditions

CARDIOVASCULAR GENETICS

If you have a family and/or personal history of the following, you should consider having a genetics evaluation.

- Cardiovascular condition diagnosis at a young age
- Two or more family members with the same type of cardiovascular condition
- Arrhythmia (heart beat abnormalities)
- Cardiomyopathy (enlarged heart)
- Enlarged aorta or aortic aneurysm in the chest at a young age (<60)
- Family member who died from a sudden cardiac death
- Family member with a gene mutation related to a cardiovascular condition

OBSTETRICS AND GENETICS

The geneticist's advice is helpful in cases of

- Advanced maternal age (age 35 or older)
- First trimester/nuchal screening pre-test education/ counselling
- Abnormal maternal serum screening
- Concerns about genetic disease because of ethnicity – Thalassemia, Sickle Cell Disease
- Abnormal ultrasound findings
- Previous child with a genetic condition, birth defect(s) and/or mental retardation
- Recurrent miscarriages
- Family/personal history of genetic condition
- Family/personal history of birth defect(s)
- Family/personal history of mental retardation
- Medication exposures during pregnancy
- Considering Preimplantation Genetic Diagnosis (PGD)