DEPARTMENT OF GENETIC SCIENCES

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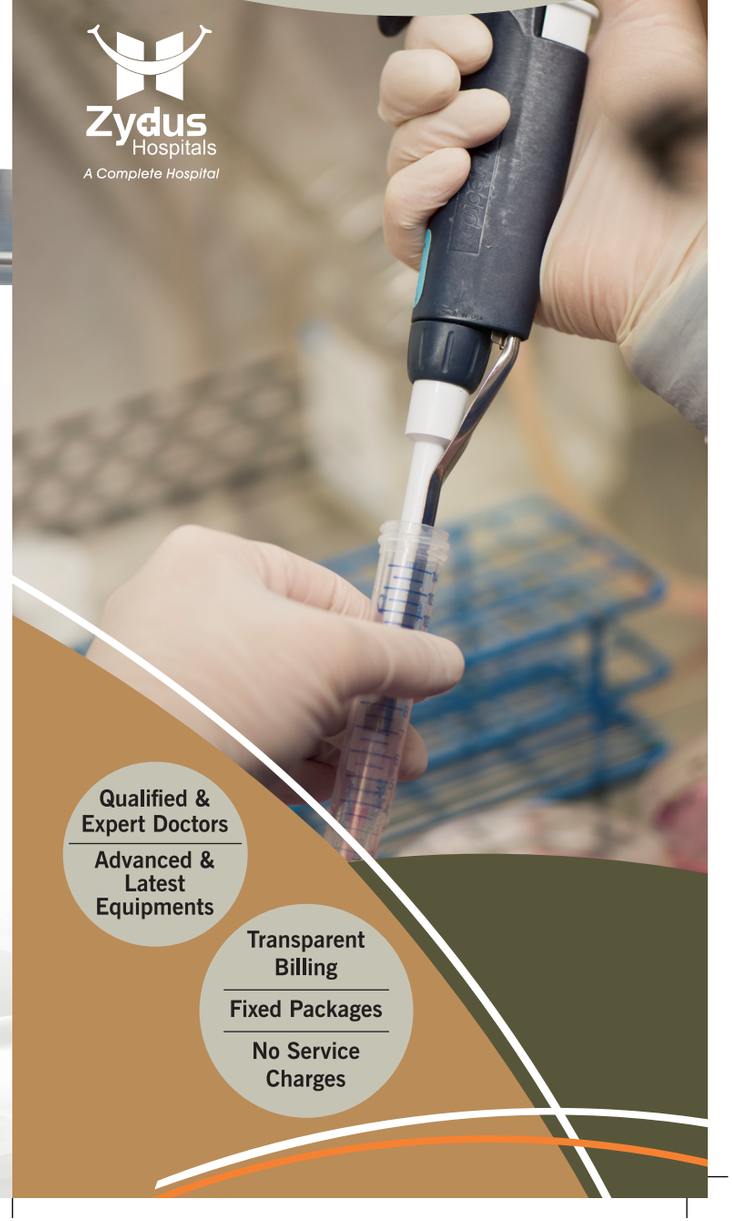

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The department of Genetic Sciences specializes in the investigation, risk assessment, management & prevention of genetic disorders. It is a one stop place which provides guidance for all types of genetic disorders beginning from fetal life to adult onset diseases, including cancers.

Genetic disorders are caused due to a defect in one's genes or chromosomes. A number of common symptoms like developmental delay, muscle weakness, recurrent abortions etc. could be because of an underlying genetic abnormality. When these symptoms are due to a genetic etiology, they not only affect the quality of life of an individual but the family is also at risk of having other similarly affected children / individuals.

Accurate and timely diagnosis is important to ensure proper management of the disease, prevent complications, identify other at-risk individuals and to prevent birth of other affected offsprings. A confirmed genetic diagnosis and expert guidance is the way forward to personalized medicine.

ABOUT GENETICS

WHO NEEDS IT?

- Family history of an affected individual (even single) with any of the conditions listed below.
- Previous affected offspring: if you have/had a child with any of the disorders listed below.
- Patients with any of the symptoms listed below even in the absence of a family history.

SPECTRUM (SYMPTOMS) OF GENETIC DISORDERS

- Neurology: Developmental Delay, Autism, Epilepsy, Ataxia, Chorea and Dystonia
- Neuro Muscular: Hypotonia or Muscle Weakness
- Metabolic including Lysosomal Storage Disorders and Mitochondrial Disorders
- Congenital Malformations: Heart Defects, Cleft Lip/Palate, Polydactyly, Club Foot etc.
- Dysmorphology: a term used when the person/child looks different - like Down Syndrome
- Children with Skeletal Dysplasia, unexplained Short Stature and Failure to Thrive
- Ophthalmology: Retinitis Pigmentosa and Congenital Cataract
- Deafness
- Hematology: Thalassemia, Sickle Cell Anemia and Leukemia
- Skin Conditions: Albinism, Icthysosis and Epidermolysis Bullosa
- Familial Cancer Syndromes: Breast, Ovarian, Colorectal, Thyroid Cancer, etc.
- Obstetric Problems: Infertility, Recurrent Abortions, Abnormal Biochemical Screening or Ultrasound and Drug Teratogenicity
- Cardiovascular: Congenital Heart Defects and Cardiomyopathy
- Respiratory: Cystic Fibrosis and Surfactant Deficiency
- Miscellaneous: Wilson Disease, Osteogenesis Imperfecta, Osteopetrosis, Marfan Syndrome, Polycystic Kidneys, etc.

The above list is limited to only common genetic disorders.

Note: It is not always necessary to have multiple affected members to label a symptom as being probably genetic.

WE OFFER

- Comprehensive management for all forms of genetic disorders like Chromosomal Abnormalities, Single Gene Disorders, Multi-gene Disorders and Inborn Errors of Metabolism
- Testing facilities include FISH, Karyotyping, Microarray, Enzyme Analysis, Mass Spectrometry and Sequencing- Sanger and Next Generation Sequencing
- Pre-conceptional and pre-marital counselling along with pre-natal diagnostic facilities for all forms of genetic disorders
- Guidance in pregnancies complicated by teratogenic exposure to drugs or radiation as well as those with associated co-morbidities (e.g. Diabetes, SLE)
- Screening during pregnancy to determine the risk of having a child with Down Syndrome, Thalassemia, Neural Tube Defects, etc.