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Medi Quest BRS Hospital

A monthly News letter from BRS Hospital

GENETIC TESTING FROM BENCH TO ~~BEDSIDE~~ CLINIC

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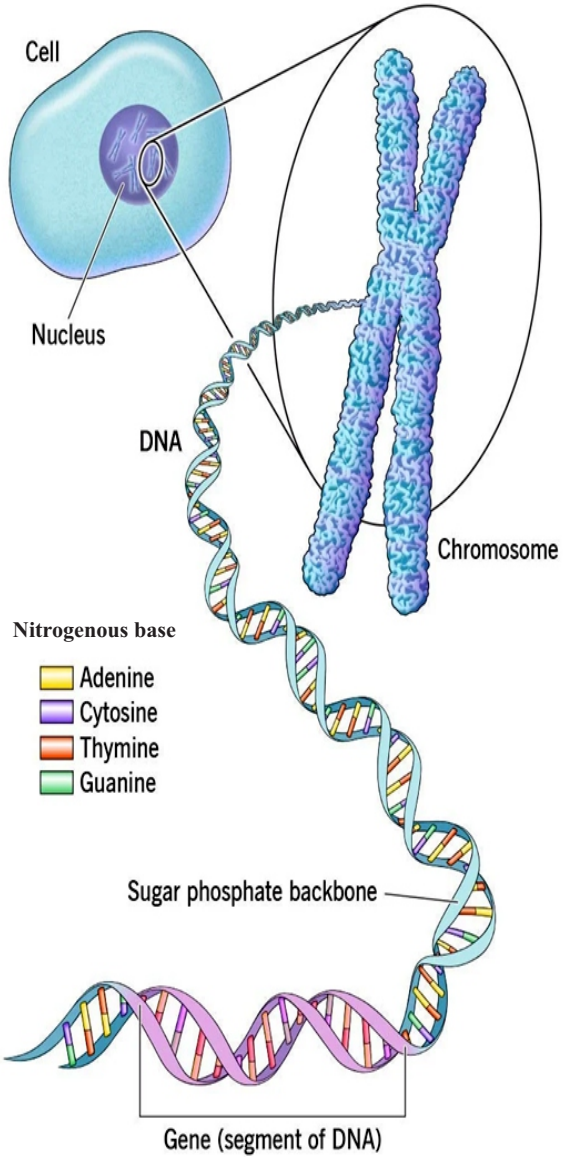
A bewildering list of tests in genetics has become available today. As the title suggests, no longer are the genetic tests confined to realm of geneticists, research laboratories and hospitals. Today the technology is available to a general practitioner. Genetic laboratories can collect blood sample from one's OP clinic and 5 ml of blood in Heparin or EDTA tube is all that is needed to perform a karyotype or next generation sequencing. There has been an explosion of knowledge in genetics , and this speciality has found its place under the sun , a DM course in Genetics is testimony to that statement .

Genetic testing can be defined as the analysis of human chromosomes, DNA , RNA ,in order to detect disease related karyotypes, genotypes, and mutations , for clinical purposes .

Such purposes include making a diagnosis of heritable diseases , identifying carriers , offering prenatal testing , for predicting risk of disease and for prognosis as in Oncology.

In this issue and forthcoming issues the focus will be on genetic testing in heritable disease in Pediatrics .

Fig: 1 - DNA, GENES, CHROMOSOMES



The broad classification of genetic disorders includes

1. Chromosomal disorders
2. Single gene disorders
3. Multifactorial disorders

For multifactorial disorders there is no specific diagnostic tests as the etiology includes multiple genes and environment.

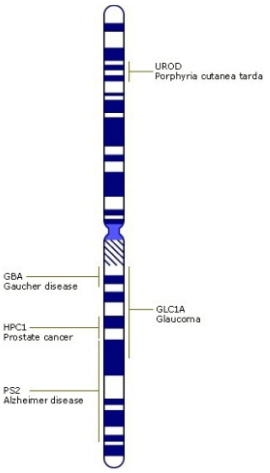
There is separate language in genetics, glossary of the commonly terms and their explanation are given below

1. Chromosomes
2. DNA
3. Nucleotide
4. Gene
5. Genome
6. Exome
7. Clinical Exome

1. Chromosomes

Chromosomes reside in the nucleus of cells. Humans have 22 pairs of numbered chromosomes (autosomes) and one pair of sex chromosomes (XX or XY), for a total of 46. Each pair contains two chromosomes, one coming from each parent, which means that children inherit half of their chromosomes from their mother and half from their father. Chromosomes can be seen through a microscope when the nucleus dissolves during cell division.

Fig 2: Chromosome 1 - Largest Chromosome



- Contains over 3000 genes
- Contains over 240 million base pairs, of which ~90% have been determined

2. DNA, Nucleotides and Base pairs

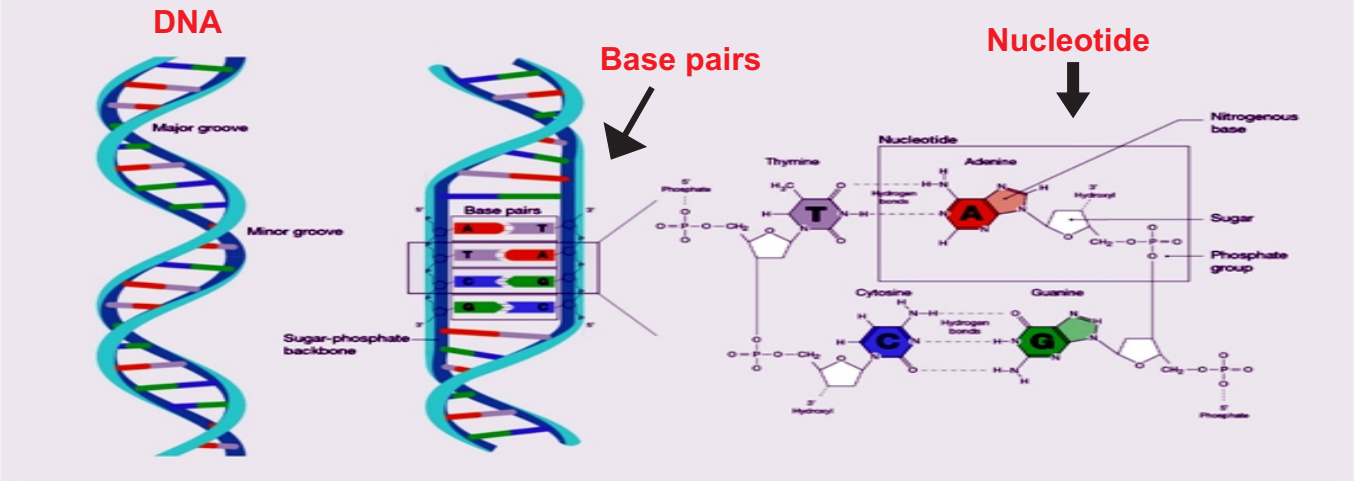
A DNA molecule is made up of two linked strands that wind around each other to resemble a twisted ladder in a helix-like shape. Each strand has a backbone made of alternating sugar (deoxyribose) and phosphate groups. Attached to each sugar is one of four bases: adenine (A), cytosine (C), guanine (G) or thymine (T). **The triad of base, deoxyribosugar and phosphate is known as nucleotide**. The two strands are connected by chemical bonds between the bases: adenine bonds with thymine (**Apple** in the **tree**) , and cytosine bonds with guanine (**Car** in the **Garage**). **This is base pairing and a base pair can be considered to the unit of DNA .** Refer Fig.3

There are 3 billion base pairs in the human genome. Because the bases exist as pairs, and the identity of one of the bases in the pair determines the other member of the pair, scientists do not have to report both bases of the pair — which is why DNA sequence is typically represented as single strings of letters. DNA sequencing involves determining the exact order of the base pairs across a DNA segment of interest or across an entire genome.

DNA from any two people is 99.9% identical, with that shared blueprint guiding our development and forming a common thread across the world. The differing 0.1% contains variations that influence our uniqueness,

which when combined with our environmental and social contexts give us our abilities, our health, our behavior.

Fig 3: DNA, Base pairs and Nucleotides



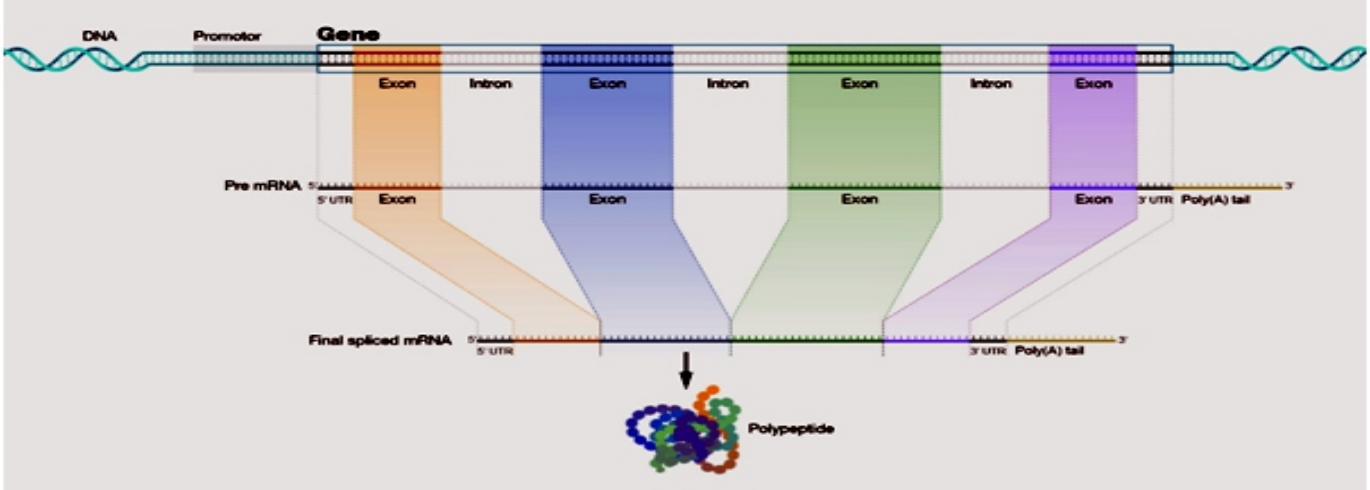
Nucleotide consists of **a sugar molecule (either ribose in RNA or deoxyribose in DNA) attached to a phosphate group and a nitrogen-containing base**. The bases used in DNA are adenine (A), cytosine (C), guanine (G) and thymine (T). In RNA, the base uracil (U) takes the place of thymine.

3. Gene

The exact definition of the word gene has long been a source of scientific debate. A simple way to think about it is as follows. Proteins are the brick and mortar that make up our cells and tissues. And genes are the part of our genome that encodes the information for making those proteins.

The human genome has roughly 20,000 protein-coding genes. Interestingly, all of the information for those 20,000 protein-coding genes is encoded by only 1.5% of the entire human genome.

Fig 4: Gene, Exon and Interon



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4. Genome :

In humans, the genome consists of 23 pairs of chromosomes located in the cell's nucleus, as well as a small chromosome in the cell's mitochondria. A genome contains all the information needed for an individual to develop and function. The genome is the entire set of DNA instructions found in a cell.

5. Exon, Exome : Proteins are encoded by genes. Genes consist of two major components, exons and introns.

Exons contain the nucleotides that directly encode for proteins, whereas introns are stretches of DNA between the exons and do not encode for proteins. The entire collection of all the exons from all the genes in a genome is called an exome. Ref Fig. 4

In the case the human genome, the exome only corresponds to about 1.5% of the genome's roughly 3 billion nucleotides.

Genome scientists have developed laboratory methods that allow them to just sequence a genome's exome; in other words, just the part of the genome that directly encodes for proteins. So, you will often hear genomicists talk about an exome sequence, which is a very small part of the overall genome (or whole-genome) sequence.

6. Clinical Exome : Clinical exome consists of only the genes known to cause heritable disease.

7.Cytogenetics is a branch of biology focused on the study of chromosomes and their inheritance, especially as applied to medical genetics.

Cytogenetics will be discussed in the next issue.

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