A short introduction to genetics and genetic analysis

The instruction for the form and function of the human is embedded in the genetic material. This genetic material is in the form of a sequence of alphabets corresponding to four nucleotides adenine (A), thymine (T), guanine (G) and cytosine (C). This genetic material is densely packed into thread-like compact structures in the cell called chromosomes. We inherit genetic material from our parents. Each of our parents contributes one half of the genetic material. The total genetic material is called the genome.

Genetic diseases are caused due to deleterious changes in the genetic sequence in the human body. The human genome, encompasses 3.3 billion alphabets of A, T, G and C, organized into 23 pairs of chromosomes (making it a total of 46). The pairs look similar for 22 chromosomes and are called autosomes, while the remaining two may be different in case of males and females and are called sex chromosomes.

As stated before, the genome sequence contains all the codes and instructions, which are required for the functioning of the organism. This functioning is expressed through proteins, which are encoded in the genes. The genes in the genome are transcribed. The transcribed sequences are further translated or decoded to make specific proteins. This flow of information is summarized in Figure 1. The region of the human genome, which contains these codes for creation of protein, is called gene. It is estimated that the human genome has approximately ~22,000 genes. Proteins coded by genes have defined functional role in the cell.

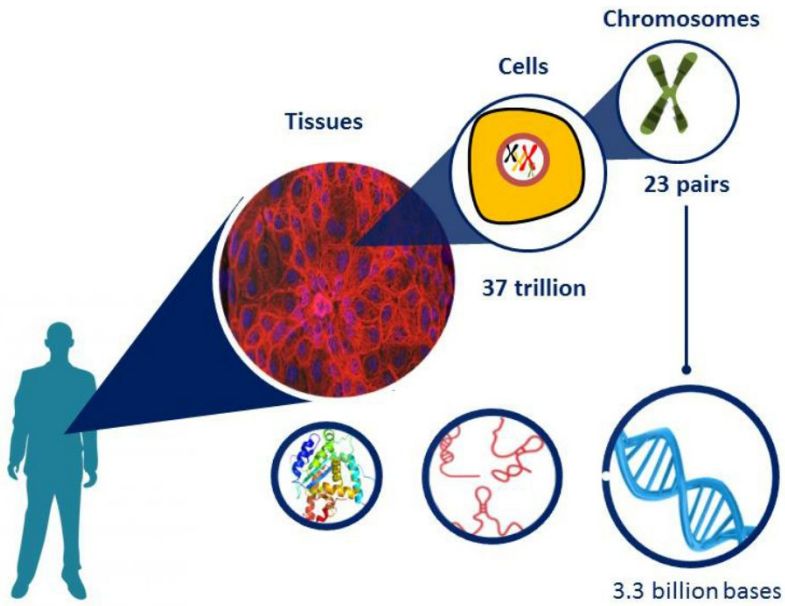

Figure 1. Schematic organization of the information flow in the genome.

Not all changes in the genome result in changes in functional consequences, but a small subset of genetic changes, which render a protein nonfunctional could potentially result in disease state. The manifestation of the disease is based on the particular function of the protein.

What is genetic testing ?
Genetic testing involves a variety of approaches to identify genetic mutations/defects in the genome of an individual. Genetic testing would provide clues to the genetic mutation and in case of genetic diseases offer a confirmation of the diagnosis. By performing a genetic test one would also hope to learn the possible genetic mutation (alteration) which may be causing the disease in you or your family.

Which genetic tests can be done ?
As part of the GOMED Programme at the CSIR Institute of Genomics and Integrative Biology, a variety of tests are currently offered for patients referred by practicing clinicians. A comprehensive listing of the test is available at http://gomed.igib.in

Why is genetic diagnosis important ?
Genetic diagnosis is important, because it helps to identify the causal genetic defect and confirm the clinical diagnosis. This would also enable screening individuals in the family for carrier status and help in tracking inheritance of the genetic defect by performing prenatal diagnosis on fetal samples. In addition, a genetic diagnosis helps in genetic counselling for risks on bearing other children and helps in estimating in how the disease might progress.

Your rights as a patient
The decision to get a genetic test is entirely upon your informed consent. Your questions will be answered clearly and to your satisfaction by the referring clinician. If you have any further doubts, please feel free to contact us at the address provided below. You have the right to refuse to allow your blood sample to be studied now or saved for future study. By providing samples, you do not waive any of your legal rights to revoke your consent and withdraw from the genetic testing at any time.

Risks, Discomfort and inconvenience
There is no major risk involved in providing blood for the analysis if you do not suffer from a bleeding disorder, in which case, you would need to inform the clinician regarding the disease. Blood would be collected by venepuncture and the analysis would require approximately 10ml of blood.

Data confidentiality
Information collected for this study will be kept confidential. Your records, to the extent of the applicable laws and regulations, will not be made publicly available. Only your Investigator(s) will have access to the confidential information being collected.

Why is genetic analysis provided investigative and not diagnostic ?
Genetic tests are relatively new and are being improved and expanded continuously. Genetic testing has its limitations. In other words, no test is 100% accurate. In fact most genetic tests would be accurate only for the specific test requested and the genomic loci tested. In most cases, genetic analysis/testing directly detects the most common disease-causing changes in a specific gene or sets of genes, and the test results are highly accurate (~98%). In other cases, an indirect method called linkage analysis is used which may produce an uncertainty in predicting carrier status or diagnosis due to naturally occurring rearrangements in the DNA (recombination). The linked markers may not always be informative in all families. Therefore the test cannot provide results for the family, or for some members of that family. In many cases the diagnosis cannot be reached due to paucity of information and inaccuracy of diagnosis or test requested. For example, rare genetic variations can also cause uncertainty in the results and in the interpretations of results. Additionally the testing is

complex and utilizes specialized materials, knowledge and skills. There is therefore always a very small possibility that the test will not work properly or that an error in the test will occur. In addition, in some cases. the genetic analysis of the mutation in the specific loci is in the genome would not always provide a diagnosis as there are still grey areas in interpreting novel genetic variations and their functional implications. In such cases, though the genetic test provides clues on the diagnosis, would not really be clinically actionable, due to the gaps in knowledge.

Cost of the genetic test

The genetic test is provided free of charge as the genetic analysis is supported by funding from the Council of Scientific and Industrial Research (CSIR), India through grants. You will not be compensated for the genetic analysis.

Whom to contact if I have a query

We would be happy to address your specific queries or offer clarifications on any of your queries related to genetic testing. Please feel free to contact us:

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