Tracing India’s Invisible Threads

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Each one of us belongs to a group of people called family. Our family is part of a family-tree, where we trace out our common ancestors. We all experience and are aware of, that we have some common invisible threads amongst every member of our family tree. But our tracing back for our ancestors remains limited for 100 or may be 200 years ago. What about years much before that? Do we, all Indians, share common ancestors? Do we all belong to one common family tree? How to find out that?

Research using modern technologies in the field of biology is of great help to find out answers to these questions. How is it possible to study a family tree of having ancestors almost thousands of years ago? Let us try to understand some basic parameters of the research.

Chromosomes and genes:

All organisms consist of cells. In human beings and most other organisms, chromosomes are found in the part of a cell known as the nucleus. Chromosomes are tiny threadlike structures made up of Deoxyribonucleic Acid, DNA. DNA is a chain like large molecule that has a double helical structure means spiral structure having pair of strands. Every gene is a segment of a DNA molecule. Gene is a tiny biochemical structure that determines the characteristics of living things inherit from their parents. Genes influence chemical and physical processes during growth and aging.

Every cell has thousands of genes. Most genes occupy a specific place on a certain chromosome. Genes are like blueprints for building a house, except that they carry the plans for building cells, tissues, organs, and bodies. They have the instructions for making the thousands of chemical building blocks in the body. These building blocks are called proteins. Some proteins are responsible for the size, shape, and structure of the parts making up our body. Other proteins, known as enzymes, make possible the thousands of chemical reactions that occur constantly in our body. The process by which the cell makes a protein according to the instructions carried by a gene is known as gene transcriptions.

Each species of animal and plant has a characteristic number of chromosomes in its body cells. Body cells, often called somatic cells, are the cells that make up such body parts as muscles and bones. Body cells differ from sex cells. In male animals, the sex cells are sperm, in female animals, the sex cells are eggs. Human beings typically have 46 chromosomes, arranged in 23 pairs, in their body cells.

Every species of plant and animal has a certain number of genes on its chromosomes. Scientists estimate that human beings have from 50,000 to 100,000 different genes. A set of all the genes that a species has on its chromosomes is called its genome.

Genetics:

Genetics is the scientific study of heredity, the passing on of characteristics of living organisms from one generation to the next. Geneticists investigate the structure, function, and transmission of genes. Each of the cells in the human body has 50,000 to 100,000 genes. Genetics can be divided into three major areas of study as molecular genetics, transmission genetics and population genetics.

Advances in molecular genetics led to the development of genetic engineering (techniques for altering the structure of genes). Transmission genetics is the traditional approach to the study of heredity. Transmission geneticists locate and describe where genes are arranged on chromosomes, a process called gene mapping.

Population genetics focuses on processes that change the relative frequency of genes in a population through time. A population is a group of individuals of the same species that live within the same area. Population geneticists often study how mutations and other processes of evolution, such as natural selection, interact with one another. They try to understand how such interactions affect the frequency with which certain genes occur within a population. Population geneticists believe that an understanding of the processes of evolution and genetic transmission helps explain the diversity of life on the earth--and within our own species.

Gene flow and genetic drift:

Gene flow is the transfer of genes from one population to another. When separate populations come into contact and interbreed, combinations of genes are introduced into each group. Consequently, the gene pool of each group comes to include genes from pools of the other populations. In this manner, the allele (different forms of the same gene are called alleles. Some alleles are dominant, and others are recessive.) frequencies of populations may change over time.

Genetic drift refers to a chance to increases or decreases in allele frequencies of a population from generation to generation. The genes of each generation represent only a sample of the previous generation's gene pool. As a result, the allele frequencies of each generation of individuals tend to vary randomly within the limitations of the preceding generation's gene pool. Such changes will probably have little effect on each generation in large populations. But they can lead to major genetic changes in small ones in a short period.

Mitochondrial DNA:

During fertilisation only the nuclear DNA of the sperm penetrates the nucleus of the egg. The two sets of chromosomes, one from the father and the other from the mother, come together to form pairs. All the nutrients necessary for nurturing an embryo during gestation period are provided by the mother. Even all the energy needed for the set of biochemical reactions during the process is also provided by the powerhouse of the egg cell. This powerhouse is called mitochondria. Interestingly the mitochondria too have their own DNA called as mitochondrial DNA. mtDNA is different from the nuclear DNA. Therefore, all humans have two copies of each chromosome, and therefore each gene, with one copy from the mother and one from the father. But mitochondria, and the genes in them, are almost exclusively inherited from the mother.

Mitochondrial DNA replicates only in mitochondria, and doesn't interact with the rest of the genome. This means that not only do mutations, that are changes in the mtDNA, occur completely separately, but they also won't be covered over by recombination with nuclear DNA. The other thing is that sexual reproduction involves two gametes, or sex cells. One, the ovum or the female cell, is very very large. The other, the sperm or the male cell, is very very small. So small, in fact, that it really doesn't contribute anything to the next generation other than its DNA. Mitochondria are only present and passed down in the female gamete, which, in turn, implies that every mitochondria in our body right now is shared with our mother and mother only, and not our father.

Y- Chromosomes:

Human Y chromosomes are male-specific sex chromosomes; nearly all humans that possess a Y chromosome will be morphologically male. Y chromosomes are therefore passed from father to son; although Y chromosomes are situated in the cell nucleus, they only recombine with the X chromosome at the ends of the Y chromosome; the vast majority of the Y chromosome (95%) does not recombine. When mutations arise in the Y chromosome, they are passed on directly from father to son in a direct male line of descent. The Y chromosome and mtDNA therefore share specific properties.

Other chromosomes, autosomes and X chromosomes in women, share their genetic material (called crossing over leading to recombination) during meiosis (a special type of cell division that occurs for the purposes of sexual reproduction). Effectively this means that the genetic material from these chromosomes gets mixed up in every generation, and so any new mutations are passed down randomly from parents to offspring.

The special feature that both Y chromosomes and mtDNA display is that mutations can occur along a certain segment of both molecules and these mutations remain fixed in place on the DNA. Furthermore the historical sequence of these mutations can also be inferred. For example, if a set of ten Y chromosomes (derived from ten different men) contains a mutation, A, but only five of these chromosomes contain a second mutation, B, it must be the case that mutation B occurred after mutation A. Furthermore all ten men who carry the chromosome with mutation A are the direct male line descendants of the same man who was the first person to carry this mutation. The first man to carry mutation B was also a direct male line descendant of this man, but is also the direct male line ancestor of all men carrying mutation B. Series of mutations such as this, forms molecular lineages.

Haplogroups:

Each mutation defines a set of specific Y chromosomes called a haplogroup. All men carrying mutation A form a single haplogroup, all men carrying mutation B are part of this haplogroup, but mutation B also defines a more recent haplogroup (which is a subgroup or subclade) of its own which men carrying only mutation A do not belong to. Both mtDNA and Y chromosomes are grouped into lineages and haplogroups; these are often presented as tree like diagrams. Molecular evolution is the process of evolution at the scale of DNA, RNA, and proteins. In the study of molecular evolution óhaplogroupsô play an important role. A haplogroup consists of similar haplotypes, thus making it possible to predict a haplogroup from haplotypes. Y-chromosome and mitochondrial DNA haplogroups have different haplogroup designations. Haplogroups pertain to deep interesting features. Most prominent is, there is extensive sharing of oncestral origins dating back thousands of years.[1]

Research and society:

Lalji Singh, David Reich and their colleagues have documented high level population structure in India. They have shown that, todayøs Indian population is the model of mixture between two ancestral populations, named as ANI and ASI. ANI means óAncestral North Indiansô is genetically close to Middle Easterners, Central Asians and Europeans, and ASI, óAncestral South Indiansö, is as distinct from ANI and East Asians is. They analyzed some 25 diverse groups in India, to provide evidence for these two genetically divergent, ancient populations. Authors drew few more conclusions in addition to the above mentioned one. One is that Indian populations bear the genetic imprint of European, Asian and even African genomes. Secondly, they found that diversity is 3 to 4 times greater than that observed within Europe. Thus they concluded that many Indian populations were founded by small numbers of individuals with subsequent limited migrations. These events can be dated by genomic data between 750 years to 2,500 years ago. Each Indian population had random genetic drift. The drift has four implications. First, one can predict a high burden of genetically recessive disorders in India, many unique to each population, estimated to be greater. Second, some diseases will have elevated frequencies in many regions of India owing to shared ANI and ASI ancestry.

Further more, Deepa Edwin and P. Majumder studied DNA samples from 160 unrelated individuals belonging to five Dravidian tribal populations of southern India. They were analysed for ten mtDNA polymorphism. There is extensive sharing of mtDNA haplotypes among all the tribal populations studied, indicating that there was a small female founding populations in India. The wide spread history of founder events in India is also medically significant because it predicts a high rate of recessive disease. Haldane wrote decades ago that if inter caste marriages in India become common, recessive diseases will become rarer. However, it has been generally appreciated that this applies to groups throughout India.

Susanta Roychoudhary, P. Majumder and their colleagues studied mitochondrial profiles of 23 ethnic populations of India from diverse cultural, linguistic and geographical background. There is extensive sharing of small number of mtDNA haplotypes, among different populations. This indicates that Indian populations were founded by a small number of females. Ethnic differentiation took place subsequently thru a series of demographic expansions, geographic dispersal and social groupings.

The results of comparison of haplotype data across populations within India helps to trace out Indiaôs invisible threads. Research reveals fundamental unity of mtDNA lineages in India, in spite of extensive cultural and linguistic diversity.
