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The Secret of Certain Human Diseases

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Our medical science has beautifully overcome almost all the diseases, even then etiology of some of the diseases remains unnoticed. Every one of us might have come across somewhere with certain human beings who are quite different from the normal human beings is morphology.

Our knowledge regarding such diseases remains shallow in the past, but recently extensive studies have thrown much light on this aspect. Today out of 1000 children born, at least three are mentally retarded or abnormal babies.

Now, what accounts for their abnormalities? The fact is that we inherit our normal characteristics from our ancestors. The characteristics come in coded forms, called genes, situated on string like structures known as chromosomes. Thus, genes are the passengers and chromosomes are the vehicles. A normal human cell has 46 chromosomes, half of which are derived from mother and the other half from the father through the germ cells called ovum and sperm respectively. On fertilization, the two germ cells fuse and the resulting zygote have 46 chromosomes or 23 pairs like those of the parents. Of these 22 pair is common to both male and female cells and these are called autosomes. The 23rd pair is of the sex chromosomes which is homomorphic in female and is termed as XX, and heteromorphic is male where it is called XY. Thus all the ova formed by female had 22 autosomes and one X chromosome, but in case of males two types of sperms are formed, half with 22 autosomes. The sex of the child is determined by the fertilizing sperm which bear either X or Y chromosomes, a female child is born and if the ovum is fertilized and a sperm with Y chromosomes, then a male child is born.

As mentioned earlier, the genes lie on the chromosomes. These genes like chromosomes exist in pairs are got from each parent. Now what a gene is ? Chemically, a gene is a segment of Deoxyribonucleic acid (DNA) molecule which codes for the synthesis of a specific sequence amino acids of a single protein. Any alteration in a gene leads to the synthesis of an abnormal protein. Thus, the function of the normal which may be an enzyme or a hormone or a receptor or a structural protein, gets disrupted. The result could be a deficiency of the product and an accumulation of the substrate. Lack of sufficient product would affect the function of the system and abnormality arises. The accumulation of substrate with its by - products cause toxicity which manifests itself as a disease. Clearing, the background, we can say that each gene has some specific function for the normal functioning and development of an individual and any change in the chromosomes, which are the carriers of the gene, would lead to the abnormalities. The chromosomal changes may be with respect to their structure or their number. If the chromosomes increase from 46 or decreases, abnormalities would result.

For each character, at least there are two genes, one from either parent. Out of these one may be dominant over the other example - the gene for the brown eyes is dominant over the gene of the blue eyes. If an individual has one gene coding for brown eye colour and a gene for the blue colour, then he/she will have brown eyes because the genes for the blue eye is recessive. Thus, to have blue eyes the genes obtained from both parents must code for blue eye trait i.e. recessive trait needs double dose genes to express.

Marriage relation

By detailed studies and experimental work, it has been shown that consanguineous (between blood relations) marriages often lead to the transmission of defective genes.

An unrelated couple generally has a three percent risk of giving birth to a baby with some genetic defect. In consanguineous the risk becomes almost double (5-6%). It is because of the fact that the genes carrying recessive traits are not common in a population. But the chance that, both parents have the

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same rare abnormal gene increases when they have a common ancestry. Consanguineous marriages are common in certain sections. In these, sections genetic disease are comparatively more common. To quote an example - Phenylketonuria is the one such disease (PKU), which is more frequent in Karnataka, where consanguineous marriages are common in certain sections. The patients of PKU have fair skin and hair with mild to severe degree of mental retardation. This condition is due to the deficiency of enzyme, which converts the amino acid phenylalanine to Tyrosine. Tyrosine is necessary for the formation of melanin. The black pigment in our product is toxic to the developing brain. By reducing phenylalanine in the diet and by supplementing Tyrosine, the child may have normal growth.

In man, phenotypic changes studies so far have been related to the sex chromosomes or autosomes. These may be numeral or structural. Phenotypic changes due to abnormalities is the sex chromosomes may not be recognized at birth because there are at times no physical abnormalities and if there are, they are so minor that they can be missed. They become noticeable at puberty when sexual maturation becomes abnormal. But the phenotypic abnormalities in autosomes are well pronounced even at birth.

The word syndrome has been used to call such individual, which are abnormal up to his time many kinds of syndrome have been reported. But at present we shall discuss only the syndromes arising through some changes in the sex chromosomes. Two most common types of syndrome that arise due to some changes in the number of sex chromosomes especially the X chromosomes are:

Turner syndrome

Turner (1938) came across with certain female individuals with short statures. Low I.Q., webbing of neck and cubitus vulgus was prominent and they had poorly developed secondary sexual characters. The cause of these abnormalities was known in 1959 when "Ford" and his associates study the chromosomes number in females having above cited features. These females had 45 chromosomes whereas, in normal ones we have 46 chromosomes. These are present only 44 autosomes and only one X chromosome. Thus, the cause of morphological changes in Turner syndrome was discovered. Birth rate of these estimated to be 2/ 1000 live births.

Klinefelter syndrome

Klinefelter (1942) reported a male who had some unusual features like gynaecomastia, a tall in statue and dull. These male were sterile and called as Klinefelter syndrome. The chromosomal constitution studies by Jacob and Strong in 1959 in these males revealed that they had 47 chromosomes with one extra X chromosomes (47, XXY). All the phenotypic in these males were therefore attributed to the extra X chromosomes. Birth rate of these males is estimated to be 2/1000 live births.

Intersexes

Beside the Turner and Klinefelter syndrome, the intersexes are equally common. We do see some individuals who share the male as well as female characters. They are between males and females. Such individuals are called intersexes (*Hijaras*). Such individuals have the chromosomes both for male and female. In these individuals the sex hormone for male and femaleness are produced at the same time and they don't let each hormone to express and, thus an intermediate situation arises. These are sterile.

Why such individuals after all born? It is a question that generally arises in mind in each one of us. These individuals are born due to some error in the division of sex chromosomes. Thus, resulting in the formation of abnormal gametes, which on fusion forms the abnormal zygote and this zygote develops into either a Turner syndrome or Klinefelter Syndrome or in intersex. Birth of these individuals should never be considered a cruse of God. We had started doing work on Human Genetic Diseases of Thalassaemia with collaboration to Human Genetic Research cum Counselling Centre (Jammu) and Delta RIA Pathology Lab Pvt. Ltd. (Bhopal).

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