WORLD JOURNAL OF PHARMACEUTICAL RESEARCH
SJIF Impact Factor 8.453
Volume 13, Issue 5, 202-211. Review Article ISSN 2277–7105

UNDERSTANDING THE GENETICS AND HEREDITARY DISORDERS THROUGH AYURVEDA A. REVIEW

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Article Received on
13 January 2024,

Revised on 02 Feb.2024,
Accepted on 23 Feb. 2024

DOI: 10.20959/wjpr20245-31402

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ABSTRACT

Genetics is the science of inheritance; the blossom of scientific revolution in human genetics has been started since ancient period. Heredity, the sum of all biological processes by which particular characteristics are transmitted from parents to their offspring. There is a high prevalence of genetic disorders in India. An estimated 495,000 infants with congenital malformations, 390,000 with G6PD deficiency, 21,400 with Down syndrome, 9,000 with ß-thalassaemia, 5,200 with sickle cell disease, and 9,760 with amino acid disorders are born each year. The Ayurveda acharyas had fundamental knowledge of genetics much before the modern genetic scientist. Ayurveda has taken up its applied aspect scientifically i.e the role of tridosha, prakruti and panchamahabhuta in formation of different organs. The concept of basic unit of genetics like Beeja–Beejabhaga–Beejabhagavayava, mutation, inheriting factors, fertilization, and description of congenital diseases like Madhumeha, Arsha, Kustha due to shukra and shonitadushti and how to prevent birth of a physically and mentally handicapped child. Sahaja, kulajavikaras are outcome of a beejadosa and intrauterine environment which are Asadhya in nature. Charaka in this context explained due to vikriti of beeja, bijabhaga and bijabhagavayava of the couple, there will be vikriti or vyapada in the child depending on gender. Hence Ayurveda advised shodhana and rasayanato restore health which prevents the appearance of genetic disorders.

KEYWORDS: Genetics, Heredity, Beeja, Beejabhag, Beejabhagavayava.

INTRODUCTION

Genetics is the science of inheritance. Heredity, the sum of all biological processes by which particular characteristics are transmitted from parents to their offspring. Around 65% of people have some kind of health problems as a result of congenital genetic mutation. There are well over 6,000 known genetic disorders,[1] More than 600 genetic disorders are treatable.[2] Around 1 in 50 people are affected by a known single-gene disorder, while around 1 in 263 are affected by a chromosomal disorders.[3] In this modernized era due to lifestyle changes and environmental hazards there is increase in the number of genetic diseases like genetic defect in childlike lameness. In our classical texts genetics is best described by Acharya Susruta and Acharya Caraka in Sharira Sthana as concepts related to genetics like pakruti, matrujadi shadbhavas and beejabhaga avayava are described. They knew the factors determining the sex of a child, genetic defect in a childlike lameness or, blindness; he said it was not due to any defect in the mother or the father but in the ovum or sperm of the parents.[4] Thus Ayurveda and genetics if attempted in systemic manner along with the upcoming technologies would be used to prevent and cure various genetic disorders.

AIMS AND OBJECTIVES

To study the aspects of genetic disorders described in Ayurveda.

MATERIALS AND METHODS

The concept of beejabhaga, matrujadi shadbhavas and prakruti etc. which closely explains about genetics and its disorders in sharira sthana of both charaka and sushruta are explained.

Concept of prakruti

Prakriti is the basic factor which is determined at the very time of the conjugation of Sukranu and Andanu in the presence of Beeja and Kshetra. The concept of the Prakriti plays a central role in understanding health and disease in Ayurveda. Prakriti has a genetic connotation that can provide knowledge for classifying human population based on phenotype characteristics. Ayurvedic literature classifies all individuals into different Prakriti types based on relative proportion of each Dosa. The human phenotype can provide a genetic basis for the three major constitutions. The concept of Prakriti in Ayurveda should be considered from genomic perspective. Permutation and combination of Vayu, Pitta and Kapha attributes characters along with other factors like different types of Purusa.

Thus Ayurveda determined the Prakriti on the basis of the individual variation and it is proven to have strong genetic component. Prakriti assessment evaluates each Dosas degree of dominance. It gives an important idea for diagnosis, prognosis and therapeutics. Subtle combination of the three Vata, Pitta, Kapha Prakriti types are specific and individualized as the DNA sequence based genetic background.

The human phenomena based on Ayurveda can provide a genetic basis for the three major constitutions or Prakriti. Prakriti of the foetus is also determined by the following factors.[5]

1. Sperm and ovum (Sukrasonita prakriti).
2. Season and condition of uterus (Kal garbhasaya prakriti).
3. Food and regimen of the mother (Matur ahar-vihar prakriti).
4. Nature of the Mahabhutas comprising the foetus (Mahabhuta vikar prakriti)

Fertilization and Sex Determination

Ayurveda considered two basic factors in the development of human beings i.e Sukra and Sonita to resemble that of sperm and ovum in modern medical science and which are responsible for the fertilization in the human beings. In these context Charak clearly stated that dominance of Sonita during conception resulting procreation of female child and dominance of Sukra (sperm) leads to male child. The sex of an individual is determined by the X and Y chromosome as female and male respectively.[6] regarding the mother diet, life style or adverse psychological state at time of conception or development of fetus play significant role in genetic predisposition of disease, sushruta in terms of ksetra, ambu, bija and Rtu rightly conceives this concept.

Concept of inheritance pattern – shadbhavas

The factors responsible for the procreation of foetus or human being derived from the following sources like one is matrija[7], pitrija[8], Atmaja, Satmyaja, Rasaja, Sattvaja.[9] These Matrija (maternal) and Pitrija (paternal) Bhavas are responsible for different organogenesis like- Twak, Rakta, Mamsa, Meda, Majja; Nabhi, Hridayam, Kloma, Yakrit, Pleeha, Vrikka, Vasti, Purishadhanam, Amashaya, Pakvashaya, Uttara Guda, Adhara Guda, Kshudrantra, Sthulantra, Vapa, Vapavahanam are deriverd from Matrija (maternal) bhavas and Sukra, Kesha, Smasru, Nakha, Loma, Danta, Asthi, Sira, Snayu, Dhamani are derived from Pitrija (paternal) Bhavas. This organogenesis is controlled by gene or chromosomes. In human being half of chromosome come from maternal and half from paternal chromosomes or genes, that is similarly to Matrija (maternal) and Pitrija (paternal) Bhavas. If any changes in these factors

they result congenital deformities or Adibala Privratta Rogas. Any changes Matrija (maternal) and Pitrija (paternal) Bhavas are depends on health status of parents.

Concept of Beeja–Beejabhaga–Beejabhagavayava and its vikriti

Ayurveda considered three genetic units in the form of Beeja (Germinal cell), Beejbhaga (Chromosome) and Beejbhagavyava (Gene). Acarya Charaka speaks first about the component of Beeja whether of male or female and designated them as Beejbhaga and Beejbhagavayava.[10] Commenting on these terms, Acarya Chakrapani has clearly stated that the smallest unit founding in Shukra (Sperm) & Shonita (Ovum) can be considered as Beeja of male and female respectively, which may compare with the male and female gametes i.e Sperm and ovum. The Beejbhaga[11] is the component lying inside the Beeja and holding responsibility of development of different body organs and tissues of the body and it may compare with the Chromosomes which are passed on as units from one generation to other generation one from each of the parents. Beejbhagavayava should be taken as further more subtle stage of Beejbhaga carrying hereditary characters and it may compare with the gene which is the functional unit of heredity and mainly responsible for expression of a particular trait in an individual.

Acarya Caraka has explained further that teratologic abnormalities depend upon the condition of beeja, not on the physical status of the couple. In other words, what so ever part of Beeja is defective, the body part developing from that portion of beeja will be abnormal.[12]

Genetic diseases described in Ayurveda

1. As per Charaka Samhita, obesity could also be attributed to genetic imperfections. Prameha (urological diseases such as diabetes mellitus) has been described as a hereditary disorder.[13]
2. Acharya Sushruta cited two types of Prameha: Sahaja (hereditary/congenital), which would be acquired from mother and father due to vitiation of Shukra and Shonita (seed defects), respectively. Patients who are diabetic from birth (congenital) and those born of diabetic parents (hereditary) are not curable due to indisposition in their genes.[14]
3. As per Charaka Samhita, there are two categories of piles: hereditary piles and nonhereditary piles. The vitiation of seeds (sperm and ovum), specifically the part of the seed involved in the generation of the anal sphincter, induces hereditary piles. Vitiation of seed is affected by two factors: (a) a poor diet and regimen of the father and mother and (b) the morally reprehensible acts supposedly committed by them.[15]

4. Shukragata Kustha, according to Madhav Nidan, is one of the genetically inherited underlying factors implicated in the inherent disease condition in their descendants.[16]

Charak has described that due to vikriti of bija (gametes), bijabhaga (chromosome) and bijabhagavayava (gene) of the couple, there will be vikriti or vyapada in the child depending on gender. Vitiation of beejabhagavayava of mother leads to **putipraja** and in case of same condition in sperm, it leads ro **putipraj**. When the Beejbhag in beeja is responsible for the development of Garbhashaya which is excessively vitiated, then woman gives birth to a **Vandhya**, while same in sperm condition called as **vandhy.**[17]

When Beejbhagavayava which is responsible for the production of uterus and organ that characterize a female e.g. breast, genital organs etc. were excessively vitiated then she gives birth to a child who is not completely female but only having feminine characteristic in abundance, known as varta and similarly when the Beejbhagavayava which is responsible for the production of sperm and organs that characterize a male, are excessively vitiated, then this gives birth to a child who is not complete male but only having masculine characteristics in abundance, known as Trinputrika.[18]

Concept of chromosome, gene, chromosomal abnormality

In this context charak describes regarding sex abnormality like Dwireta which relates to hermaphrodities(46XX Karyotype) in which an individual has both testicular and ovarian tissues. Other like pavanendriya it may consider as klinfelters syndrome (47 chromosome 44XXY) having involuntary infertility and evidence of azoospermia in male partner causing complete failure of gametogenesis occurs. Whereas narashanda and narishanda can be consider as pseudohermaphoridism. characteristics mentioned in Shandi Yonivyapad correspond closely to Turner's syndrome(45XO) and its occurrence due to Beejadosha. The others are Samskarvahi (anaphrodisia), Vakri (hypospada), Irsyabhirati (mixoscopia) and Vatikshanda (eviration).[19]

MANAGEMENT

The following measures have been described in Ayurvedic classics to remediate the Beejpushti and other underlying factors for procreation. The approaches that must be executed can be used both before and during pregnancy. Ayurveda aims at physical and mental health. Therefore, good counselling using the Ayurvedic principle is required to maintain mental and physical health. There is strong evidence that an optimal diet reduces the

frequency of unsuccessful pregnancy complications and severe congenital malformations during the reproductive years, particularly preconception. The male should consume Shali rice with Ghrita and milk. Tila Taila and Masha should be consumed by the female during the preconception phase. Black grams included in the pre conceptional diet is a rich source of folic acid, proteins, and fibers. Thus, it helps prevent neural tube defects. As it is a potent antioxidant, it helps reduce oxidative stress. Rice contains more carbohydrates, and rice bran contains Vitamin B complex, which is easily digestible. Milk provides calcium. Ghee has properties of imparting strength, improving tonicity, and nourishing the body.[20]

After successful conception, replenishing the woman's with garbhini paricharya.[21]

Ayurveda Panchakarma therapy provides physical and mental fitness. Recent updates on Panchakarma therapies show significant evidence in curing many disorders as well as for the prophylactic purpose. It also helps keep the hormones at a normal level. These would also help in the removal of endotoxins. The couple should first undergo Shodhana (purification) therapy, starting with Purvakarma (preparatory measures), Snehana (oleation), Swedana, then Vamana (therapeutic vomiting), Virechana (therapeutic purgation), Asthapana (decoction enema), Anuvasana Basti (oil enema), and Uttar Basti for females.[22-23]

Muscular dystrophy occurs due to vitiation of vata dosha and it decays the sapta dhatu by improper digestion and absorption of nutrients. vata hara chikitsa as Snehana, Swedana, vasti and lepana which may slow down muscle destruction other than this gentle yoga and breathing exercises gives strength to muscle and improves the digestion. Herbs like ashwagandha, guggulu, guduchi, shallaki, Punarnava and triphala support over all wellbeing of MD.

DISCUSSION

In Ayurvedic classics, genetic disorders are described as Sahaja Roga (prameha), Kulaja Roga, or Adibala Pravritta Roga, which are precipitated by flaws in Beeja. Sahaja Roga means that which is apparent at birth, Kulaja Roga means that which runs in families, and Adibala Pravritta Roga means that which arises from defects in the male and female reproductive attributes. While attempting to explain the stree vyapadha and purush vyapadh the terms Beeja, Beejabhaga, and Beejabhagavayava were used. Acharya Charaka also explains Astha Nindita Purusha may consider as genetic disabilities. In Atulyagotriya

Adhyaya it has been mentioned that marriages in two similar “Gotras” should be spared because they contribute to autosomal recessive disorders to prevent genetic abnormalities.

Early age or very late age conception may lead to unhealthy or defective childbirth. If a woman below 16 years is impregnated by a man below the age of 25, either she will not conceive, or if at all she conceives, she will have intrauterine death of the fetus. Younger women give birth to a majority (80%) of children with Down syndrome.[24]

For congenital disorders Maternal pregestational diabetes, hypothyroidism, different acquired maternal infections, prescription drugs, and high-dose radiation during pregnancy are also some of the prominent factors other than chromosomal factors in genetic and heredity disorders.In the 4th month of development, the Garbhini is called as Dauhrida at that time, whose wishes and desires, not being honored and gratified lead to the birth of a paralyzed, hump-backed, crooked-armed, lame, dwarfed, defect-eyed, and a blind child. Other Gharbhaja Vikar like- Pangu, Sheershambu / hydrocephalous, Hraday Rog/ congenital heart disease etc.

Gharbhaja Vikrati are mainly cause by Shadbhava & diet of mother during pregnancy, during developmental stage of fetus Ayurveda explain Garbhini Paricharya for fetal wellbeing. various other Concept of sex determination by punsavana karma, formation of twins or multiple pregnancies have been described in classics. Ayurveda aims at physical and mental health. Panchakarma in role of keeping the hormone at normal level and removal of endotoxins. Discription of diet for both male and female in preconception phase helps prevent neural tube defects. As it is a potent antioxidant, it helps reduce oxidative stress. Early detection of diseases, prevention, the natural history of reoccurrences, and prenatal diagnostic opportunities are some of the measures that can be practiced to deal with the situation.

CONCLUSION

Whole world is looking towards Ayurveda for better life style and preventive method of congenital anomalies and hereditary disorders. Our classical Scholars have explained the facts that genetic disorders are not due to any defect in the mother or the father but in the ovum or sperm of the parents. A malformed alive fetus is one of the worst aspects of pregnancy. These diseases affect the life of parents, society & nation. Ayurveda advised shodhana and rasayana to restore health which prevents the appearance of genetic disorders. The Ayurveda suggest that the prevalence of congenital or genetic disorders controlled by

various approaches like; Dietary regimen related to Garbhini Paricharya, garba sanskar, avoiding Garbhopghatkar Bhava and Tridosha vitiating. Sahaja and kulajavikaras are outcome of a beejadosa and intrauterine environment which are Asadhya in nature in this condition prevention is better than cure. An attempt is made to understand the concept of genetics and heredity through ayurveda with beejadosha and its vikriti, shadbhavas and prakruti and prevention. Although more research is needed to support this assertion made in Ayurvedic classics, the current study provides a framework for future research directions.

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