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THE CONCEPTUAL STUDY OF THE CONGENITAL ANOMALIES AND GENETIC ANOMALIES W.R.S. TO SHAD GARBHAKAR BHAVAS AND ADHIDAIVIK VYADHI

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INTRODUCTION

The health of the nation depends on the health of the citizens. Throughout history, the birth of malformed fetuses has been well documented and the attitude towards the infants and their parents varied according to the cultural state of the people and ranged from admiration to rejection and hostility.[6] Advanced modern medical science has results in very critical diseased conditions and also extended the life span of the human, but the new upcoming health problems are also awaiting their solutions. The medical world is really worried about increasing rate of birth defects. They are minor, major, anatomical, physiological and even challenging to the nature.

Every 4 1/2 minutes, a baby is born with a birth defect in the United States. Birth defects affect 1 in every 33 babies born in the United States each year. That translates into about 120,000 babies.[10]

An estimated 240 000 newborns die worldwide within 28 days of birth every year due to birth defects. Birth defects cause a further 170 000 deaths of children between the ages of 1 month and 5 years.[11] Data reveals that 3 – 5% of all births result in congenital malformations, 20 – 30% of all infant deaths are due to genetic disorders, and 30 – 50% of postneonatal deaths are due to congenital malformations, 11.1% of pediatric hospital admissions are for children with genetic disorders, 18.5% are children with other congenital malformations, 12% of adult hospital admissions are for genetic causes, and 50% of mental retardation has a genetic basis. 15% of all cancers have an inherited susceptibility. 10% of the chronic diseases (heart, diabetes, arthritis), which occur in the adult population have a significant genetic component.

Robert Brent estimated incidences of Genetic Disorders Recessive (0.1%), X linked(1%), Irregularly inherited (9%), and Chromosomal aberrations (0.6%). The Congenital anomalies contributes an estimated 276000 neonatal death per-anum worldwide. Some of the common congenital anomalies are heart defects, neural tube defects and Down syndrome, Congenital Talus Equinovarus etc. Those may be due to genetic, infectious, nutritional or environmental factors but often it is difficult to identify the exact cause.[6]

Ayurveda first of all laid principles of Genetics in developing ideal Supraja (offspring) based on the theory of parental imprints. Acharya had told that when the progeny is born in a geographical region where people are naturally healthy, when the climatic factors are favorable, both the germ cells & reproductive tract are healthy as well as in a healthy mother; then the progeny will definitely be healthy. Acharya strictly discourage marriage between people belonging to same clan (tulyagotra) or any disease running family. Acharya had even named a chapter as ―Atulyagotreeya‖ in ShareeraSthana of CharakaSamhita. Due consideration was given to marriage to other clan in ancient time because consanguineous marriage increase the risk of congenital anomalies. One of the known examples is the prevalence of sickle cell anemia in Victorian family. AcharyaCharaka and Sushruta both recorded fundamentals of Genetics in respect of heredity. The etiology of congenital anomalies is based on the core principle of state of Dosha at the time of fertilization. The Ayurvedic perspective of congenital anomalies has its foundation on Adibalapravritha (due to defect in somatic cells in the intra-uterine environment). Adibalapravrithavyadhi are because of defective sperm (Shukra) & ovum (Shonita) and are determined right at the time of fertilization. Janmabalapravrithavyadhi are because of improper maternal diet & regimen (maturapacharaja) and has its impact throughout the intrauterine period.[8] The factors responsible for congenital anomalies in the fetus are defects in sperm & ovum, the actions associated with the Soul, the uterine environment, climate as well as diet & regimen of the mother.[5] According to SusrutaSamhita the factors are new generation life style (without spiritual thought), the unwholesome activities done in the past by both the parents . The three technical terminologies in case of congenital anomalies as Acharya have mentioned are defect (Dosha) in either Beeja, Beejabhaga or Beejabhagaavayava[2], which in modern parlance may be germ cell, chromosome or gene.

Again in this science all diseases(affecting man) have been described in brief, since it is common to all branches of Ayurveda. It is given in Samhitas that diseases are association of

miseries. Such miseries are of three kids viz.-Adhyatmika, Adhibhautika, Adhidaivikvyadhi. Out of these Adhudaivikvyadhi are related with super human beings(gods, demons, etc.)[9]

The effect of what is done during the previous life is known as Daiva. The unrighteous deeds of the previous life induces one to diseases; if however, they are righteous, then the individual remains free from diseases.[1]

For meeting the objective of a healthy progeny, Ayurveda scholars felt the importance of Six Procreative Factors (Shadgarbhakarabhavas) such as Matrija (maternal), Pitrija (paternal), Atmaja (Soul), Rasaja (Nutritional), Satmyaja (Wholesomeness), and Sattvaja (Psych / Mind).[3] The conglomarance of these procreative factors is a must for healthy progeny.[4]

With this background of the gravity of congenital and Genetic anomalies, as well as, the knowhow of the birth defects from the ancient scholars of Ayurveda; a study was planned as follows:

Aim

To study congenital anamolies and genetic anamolies w.r.s. Shad Garbhakarbhavas and Adhidaivikvyadhi.

Objectives

Conceptual study of the Congenital and Genetic anomalies.

Conceptual study of the Shad GharbhakaraBhavas

Canceptual study of the Adhidaivikvyadhi

To study and see the relation between GarbhakaraBhavas and Adhidaivikvyadhi with Congenital and Genetic Anomalies

Materials

Classical literature of Ayurveda as well as Modern medical science.

Ayurvedic journals and Published Article

Methods

This was purely a Literary study where the explored literature was critically analysed and interpreted.

Concepts in the Literature

As per the Ayurvedic concept of Sharir (Embryogenesis), perfection of all the Procreative factors in turn of their assigned structures and functions leads to a healthy offspring. The abovementioned Matrija, Pitrija, and Aatmaja Bhavas cannot be changed as they come from the parents and Poorvajanma Samskaras (as a result of the code of conduct), respectively, but the other three Bhavas-factors, namely Satmyaja, Rasaja and Sattvaja Bhavas, practiced properly can modify the intrauterine environment and psychosomatic health of the mother, producing a healthy impact on the fetus.[3] It is a known fact now that environmental factors can influence the genome.

According to modern medical science, there are three phases of intrauterine growth as following :

1. The Zygote Phase (Period-I): week 1-2 after fertilization consist of Cell division and implantation of this Cell mass in the Uterus. 2. The Embryonic Phase (Period-II): week 3-8 and most of the Organs developed. 3. The fetal phase (Period-III): week 9-38 and further growth as well as elaboration of the Organ systems takes place.[7]

The birth defects occur mainly due to abnormal formation of tissues, abnormal forces on normal tissues or destruction of normal tissues. Some of these defects may have a cascade effect and result in a group of related anomalies or multiple anomalies (syndromes). A congenital disorder is any medical condition that is present at birth. A congenital disorder can be recognized before birth (prenatally), at birth, or many years later. Congenital disorders can be a result of genetic abnormalities, the intrauterine environment or unknown factors. A congenital malformation is a deleterious physical anomaly, a structural defect perceived as a problem. A recognizable combination of malformations or problems affecting more than one body part is referred to as a malformation syndrome.[6]

Genetic disorders are all congenital, although they may not be expressed or recognized until later in life. Genetic diseases may be divided into single-gene defects, multiple gene disorders or chromosomal defects. Singlegene defects may arise from abnormalities of both copies of an autosomal gene (a recessive disorder) or from only one of the two copies (a dominant disorder). Some conditions result from deletions or abnormalities of a few genes located contiguously on a chromosome. Chromosomal disorders involve the loss or duplication of larger portions/total chromosome containing hundreds of genes. Large chromosomal abnormalities always affect many different body parts and organ systems.[6]

A mutation is a permanent change in the DNA sequence of a gene. Sometimes mutations in DNA can cause changes in the way a cell behaves. Mutations can be inherited; this means that if a parent has a mutation in his or her DNA, then the mutation is passed on to his or her children. This type of mutation is called germ line mutation. Mutations can be acquired; can occur when environmental agents damage DNA or when mistakes occur when a cell copies its DNA prior to cell division. Mutations can occur in every cell of the body; when they occur in somatic cells there is a risk of cancer development, when they occur in the germ line there is a risk of the offspring inheriting astructural or functional disability. Many mutations are benign or silent; others explain variation in the severity of a genetic disease (polymorphisms), and there are others that produce serious consequences.[6]

The novel germ line mutation arises in a parent‘s germ cell — either the father‘s sperm cell or the mother‘s egg cell. The child conceived through the union of sperm and egg carries the novel germ line mutation. Other than these mutations, epigenetics is also responsible for the congenital and genetic abnormalities. Epigenetics refers to changes in phenotype (appearance) or gene expression caused by mechanisms other than changes in the DNA sequence. These changes may remain through cell divisions for the remainder of the cell‘s life and may also last for multiple generations.[6]

Six procreative factors have an important role as causative factors of congenital, hereditary, and genetic anomalies (by mutation and epigenetics) — before conception, at the time of conception, and after conception, that is, during pregnancy.[3] Concepts and details of congenital anomalies have been described by almost all the scholars of Ayurveda with the opinion that congenital anomalies can occur due to the diet and lifestyle of the mother, deeds in the previous life of the fetus, vitiation of vayu, beeja (ovum and sperm), beejabhaga (chromosome), and beejabhagavyava (genes) in parents, a detailed view point in the light of the present knowledge is discussed herewith.[2]

DISCUSSION

1. Matrija Bhavas

Kula or Gotra of parents, maternal age at the time of conception, health of the reproductive organs of the female, time of conception, bija of mother, maternal diet during pregnancy, drugs-medicines taken by a woman during her pregnancy, and any infection in the mother during her pregnancy, can affect the health and normalcy of a fetus. If a mother is affected by

rubella during organogenesis of the fetus the new born may have a congenital abnormality, tjhat is, CRS triad – PDA, Blindness or Senorineural deafness.

Early age or very late age conception may lead to unhealthy or defective child birth. If a woman below 16 yrs is impregnated by a man below the age of 25, either she will not conceive, or either she will not conceive, or if at all she conceives, she will have intrauterine death of the fetus; if the child is born, it will not live or will have weak organs, ill health, deformed body parts, and so on. Younger woman…..give birth to a majority (80%) of children with Down Syndrome. Advanced maternal age, more than 35 years, is associated with the presence of abnormal chromosome number such as trisomy 21, 13, and 18. 45, X is not associated with advanced maternal age.

Due to the abnormalities of bija (ovum and sperms), Atmakarma (deeds of previous life), ashaya (uterus), kala (time factor orabnormality of ritukala), and dietetics, along with the mode of life of the mother, the vitiated doshas produce abnormalities in the fetus, affecting its appearance, complexion, and indriyas. These factors create an environment formutation and epigenetic changes in the ovum, leading to abnormalities in the fetus.[6,4]

2. Pitrija Bhavas

If a beeja (Sperm) coming from a male is afflicted, a progeny may have congenital or genetic anomalies. Abnormalities of shukra and vayu, as well as vitiated vayu located in the shukra are also believed to produce congenital anomalies. Acharya Bhavamishrahas also mentioned the abnormality of Shukra as a cause of congenital blindness, and so on.

Advanced paternal age is well documentedto be associated with new dominant mutations. The assumption is that the increased mutation rate is due to the accumulation of new mutation from many cell divisions. The more the cell divisions the more chances of an error (mutation) occuring. The four most common new autosomal dominant mutations are, Achondroplasia, myositis ossificans, and Marfan syndrome. The male germ cell exposure to drugs or environmental agents may alter genomic imprinting or cause other changes in gene expression.[6,4]

3. Atmaja Bhavas

The soul undergoes a series of births and deaths depending upon his own good or bad actions. The effects of the actions of the previous life are carried by the soul to his next life, which are

the results of good or bad actions. He has to get rid of these afflictions by following a proper code of conducting his given life, otherwise he goes into the cycle of births and deaths. This life and death cycle is achieved instantaneously at the time of the union of shukra—male reproductive element vis-à-vis the spermatozoon contained inthe semen and the Artava—female reproductive element, vis-à-vis the ovum produced by theovary.

The effect of what is done during the previous life is known as daiva. If the daiva is unrighteous sufferings are shared in the present life; if however, they are righteous then the individual enjoys a happy and healthy life. On the contrary unrighteous Purushartha is due to the sufferings of the present and future life. Indian mythology further explains and believes that righteous Purushartha also acts as an remedy for the unrighteous daiva. This is likely the law of probability, for example, if there is an autosomal dominant trait running in the family and only one partner is affected, 50% of the offsprings are expected be affected. The remaining 50% may escape unaffected. Even if it had been considered a mythological concept, it is a guiding path toward the righteous path for a happy and healthy present and future life if any.[6,4]

4. Satmyaja Bhavas

Kalasatmya: Ayurveda that in the course of the union of parents for progeny, they present an opportunity for the soul to attain a body; therefore the Vedic studies consider the time of conception eminent. That is why due consideration is given to proper time of gharbhadhanasanskara for achieving a healthy baby. Improper time, season, age of conception; all these periodical factors can influence the health of the fetus by creating a mutogenic or epigenetic influence, probably.

Karmaj/Sahaja: Tribal groups of India have their distinctive genetic makeup. They serve as a unique gene pool, which has evolved in the natural setting over thousands of years. Therefore, they have special health problems and genetic abnormalities like Sickle cell anemia, Thalassemia, G-6 PD, red cell enzyme deficiencies, and soon. The practice of endogamy and consanguinity among tribals is likely to be one of the influencing factors for the high prevalence of genetic disorders among tribals.

It can be enumerated that the Satmyaja (wholesome) procreative factor is responsible for conception, normal inheritance, and growth and development of the fetus leading to the birth of healthy, happy, active, and productive citizen of generations to come.

5. Rasaja Bhavas

If the couple consumes ruksha (dry) and the like, vata vitiating diet during ritukala and suppresses the natural urges, then the aggravated vayu vitiates Rakta and the other dhatus of the fetus and produces hoarse or nasal voice, deafness, and other disorders of vata. Also, vata produces baldness, premature graying of hair, absence of hair on face, tawny color of skin, nail, and hair and other abnormalities of vata. When a pregnant woman continuously consumes a diet capable of aggravating Kapha, it produces kustha (leprosy), kilasa (a type of skin disorder), switra (Leucoderma) and pandu (anemia) arise due to consumption of a dietcapable of vitiating kapha. Whatever diet and regimen thepregnant woman adopts, the childwill develop the same qualities.[6,4]

6. Sattvaja Bhavas

Human birth is a very rare privilege, for only man has the possibility of living a conscious, wide-awake, controlled life.Human being possess instinct and intelligence. All these things may not happen without the presence of Manasa (psyche). The Sattva of the fetus is moulded by three factors, namely:

1. Sattva of parents – Genetic derivatives
2. Garbhini Uparjita Karma - Gestation derivatives
3. Janmantara Vishesha Abhyasa- Environmental derivatives

Among these three, the one that is stronger, affects the psychology of the child more. Although it has been stressed that the psychic factors remain present from the preembryonic life and are associated in the embryo since the process of fertilization, yet apparently the psychic tendencies of the fetus manifest when the indriyas (special sensory faculties) develop in the fetus. Therefore, with the emergence of the indriyas, the mana of the fetus begins to feel Vedana (perception) and earns for the things experienced in the previous life and this phenomenon is called Dou-hridya. That is whythe second factor, that is, Garbhini Uparjita Karma has a very practical significance in relation to our context. In ancient Ayurvedic classics, special preference has been given to the Saumanasya of Mana (calm psychostatus) during the antenatal period. They have even stressed the negative results in the fetus, if followed otherwise.

The activity of the mother during the gestation period up to the delivery will result in the same Manobhavas (psycho-makeup) inthe fetus as well. Dauhrida Avastha of Garbhini (special desires of a pregnant woman) is avery evident manifestation of the Sattvaja Bhava.

Acharyas have clearly specified that the suppression of desires of the Dauhridi (pregnant woman) may influence the psychology of both the mother and fetus.[6,4]

CONCLUSION

GarbhakaraBhavas are not only the factors that bring the similar new one into this universe, but they are the carriers of the organogenesis and other traits to the fetus. These traits are similar to the traits carried by chromosomes/genes as per contemporary concepts, embryogenesis, fetal growth, and development. Garbhakarbhavas and Adhidaivikvyadhi give strong support in Ayurved point of view for preconception counseling.

And thus the Preconception counseling and Ayurved can play a vital role not only in achieving the goal of ahealthy progeny, but also in preventing congenital and genetic anomalies.

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