



Title of Article

“Conceptual Study On The Congenital And Genetic Anomalies W.S.R. To Shad Garbhakara Bhavas”

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ABSTRACT:

Despite the advancements in diagnostic techniques and therapeutic interventions, medical science has failed to keep the incidence of congenital malformations under control. Ayurveda, the ancient Indian medical system has given due emphasis on this and postulated various measures to minimize the risks. Ayurvedic Acharya's has great knowledge about genetics. The conglomeration of the six procreative factors (*Shad Garbhakara Bhavas*) such as *Matrija*, *Pitrija*, *Aatmaja*, *Rasaja*, *Satmyaja*, and *Sattvaja* is must for healthy progeny. The physical, mental, social, and spiritual well-being of the person, proper nutrition of the mother during pregnancy, and practice of a wholesome regimen, play a prime role in achieving a healthy offspring, thus structuring a healthy family, society, and nation. Present article focuses on various congenital and genetic anomalies in Ayurveda on the basis of modern scientific knowledge.

KEYWORD: *Atmaja, Matrija, Pitrija, Rasaja, Sattmyaja, Sattvaja, Shad Ggarbhakar Bhavas*, procreative factors.

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INTRODUCTION:

The health of the nation depends on the health of its 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. Advanced modern medical science has no doubt extended the life span of the human, but the new upcoming health problems are also awaiting their solution. The medical world is really worried about the increasing rate of inborn defects in the new born. These inborn defects are seen as minor, major, anatomical, physiological and even latent in nature.

Data reveals that 3 – 5% of all births result in congenital malformations¹, 20 – 30% of all infant deaths are due to genetic disorders², and 30 – 50% of post-neonatal deaths are due to congenital malformations³, 11.1% of pediatric hospital admissions are for children with genetic disorders, 18.5% are children with other congenital malformations⁴, 12% of adult hospital admissions are for genetic causes, and 50% of mental retardation has a genetic basis⁵. 15% of all cancers have an inherited susceptibility⁶. 10% of the chronic diseases (heart, diabetes, arthritis), which occur in the adult population have a significant genetic component⁷. Robert Brent estimated incidences of Genetic Disorders Recessive (0.1%)⁸, AD and X-linked (1%), Irregularly inherited (9%)⁹, and Chromosomal aberrations (0.6%)⁹. The Congenital anomalies contributes an estimated 276000 neonatal death per-anum worldwide¹⁰. 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.

According to Acharya Charaka, body is congregation of *Aparisankheya* (millions and trillions) of *Deha Parmanu* which means they are innumerable and uncountable.

पुरुषावयवविशेषा अपरिसंख्येयाः॥
(च.शा.५/४)

शरीरावयवास्तु
परमाणुभेदेनापरिसंख्येया भवन्ति॥
(च.शा.७/१७)

Parental imprints theory (Genetics) is a science of hereditary prevalent for atleast past 4000 years. 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 (*tulya gotra*) or any disease running family¹². Acharya had even named a chapter as “*Atulya gotreeya*” in *Shareera Sthana* of Charaka Samhita. 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. Acharya Charaka 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 *Adibala pravrittha* (due to defect in germ cells) & *Janmabala pravrittha vyadhi* (due to defect

in somatic cells in the intra-uterine environment). *Adibala pravrittha vyadhi* are because of defective sperm (*Shukra*) & ovum (*Shonita*) and are determined right at the time of fertilization. *Janmabala pravrittha vyadhi* are because of improper maternal diet & regimen (*maturapacharaja*) and has its impact throughout the intrauterine period¹¹. According to Charaka Samhita, 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 mother¹². According to Susruta Samhita the factors are new generation life style (without spiritual thought), the unwholesome activities done in the past by both the parents¹¹. The three technical terminologies in case of congenital anomalies as Acharya have mentioned are defect (*Dosha*) in either *Beeja*, *Beeja bhaga* or *Beeja bhaga avayava*¹², which in modern parlance may be germ cell, chromosome or gene.

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). The conglomerance of these procreative factors is a must for healthy progeny¹³.

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:

Aims and Objectives :

- Conceptual study of the Congenital and Genetic anomalies.
- Conceptual study of the *Shad Gharbhakara Bhavas*.

- To study and see the relation between *Gharbhakara Bhavas* with Congenital and Genetic anomalies.

Materials :

- Classical literature of Ayurveda as well as Modern medical science.
- Ayurvedic journals and Published Articles.

Methods :

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

Literary Study :

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 (Table-1). The above-mentioned *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. 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.

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.

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. Single-gene 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.

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 a structural 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.

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.

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. 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*, *bija* (ovum and sperm), *bijabhaga* (chromosome), and *beejabhagavyava* (genes) in parents, a detailed view point in the light of the present knowledge is discussed herewith.

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, that is, CRS triad-PDA, Blindness or Sensorinural deafness.
- Early age or very late age conception may lead to unhealthy or defective child birth. 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; if the child is born it will not live long or will have weak organs, ill health, deformed body parts, and so on. Younger women give birth to a majority (80%) of children with Down Syndrome¹⁵. 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 age¹⁶.
- Due to the abnormalities of *bija* (ovum and sperms), *Atmakarma* (deeds of previous life), *ashaya* (uterus), *kala* (time factor or abnormality 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 for mutation and epigenetic changes in the ovum, leading to abnormalities in the fetus.

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 Bhavamishra has also mentioned the abnormality of *Shukra* as a cause of congenital blindness, and so on¹⁷.
- Advanced paternal age is well-documented to 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) occurring. The four most common new autosomal dominant mutations are, Achondroplasia, Ape's syndrome (acrocephalosyndactyly), myositis ossificans, and Marfan syndrome¹⁸. The male germ cell exposure to drugs or environmental agents may alter genomic imprinting or cause other changes in gene expression¹⁹.

3. *Aatmyaja 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 conduct

in 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-a-vis the spermatozoon contained in the semen and the *Artava* — female reproductive element, vis-a-vis the ovum produced by the ovary. *Lingashareera* is the carrier of these deeds.

- 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 life²⁰. 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.

4. *Satmyaja Bhavas*:

- *Kalasadmya*: Ayurveda believes 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 *gharbhadhana sanskara* 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 so on. 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 diet capable of vitiating *kapha*²¹. Whatever diet and regimen the pregnant woman adopts, the child will develop the same qualities [Table-2]²².

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 Vishesh 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 yearns for the things experienced in the previous life and this phenomenon is called *Dou-hridya*²³. That is why the 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) in the fetus as well. *Dauhrida Avastha* of *Garbhini* (special desires of a pregnant woman) is a very 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.

Conclusion:

- *Garbhakara Bhavas* 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.
- “Pregnancy should be by choice not by chance”; preconception counseling can play a vital role not only in achieving the goal of a healthy progeny, but also in preventing congenital and genetic anomalies.

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Tables:

Table-1 : Features developed from six Procreative factors¹⁴

Procreative Factors	Features
<i>Matrija</i> – Maternal	Soft structures are inherited by <i>Matrija Bhava</i> . <i>Twak, Rakta, Mamsa, Meda, Majja; Nabhi, Hridayam, Kloma, Yakrit, Pleea, Vrikka, Vasti, Purishadhanam, Amashaya, Pakvashaya, Antra, Uttara Guda, Adhara Guda, Kshudrantra, Sthulantra, Vapa, Vapavahanam</i> Garbhashaya
<i>Pitrija</i> – Paternal	Hard structures by <i>Pitrija Bhava</i> . <i>Sukra, Kesha, Smasru, Nakha, Loma, Danta, Asthi, Sira, Snayu, Dhamani</i> and all <i>Sthira Angas</i>
<i>Aatmaja</i> – Soul	<i>Buddhi</i> /intellect/sensory/Psychological development by <i>Atmaja Bhava</i> . <i>Taasu Taasu, Yonishu Utpatti (Birth in specific species), Ayu (life span), Atmagnanam, Vignanam, Prerana of Prana and Apana, Swara, Sukha, Duhkha, Ichcha, Dvesha, Chetana, Dhriti, Buddhi, Smriti, Ahankara, Prayatna, Kama, Krodha, Lobha, Bhaya, Harsha, Dharmadharmaeelata, Upachaya; Mana, Indriyas, Akriti, Varna</i>
<i>Satmyaja</i> – Wholesomeness	Development of acquired immunity through congenial (favorable) diet consumed by mother. <i>Arogyam, Analasyam, Alolupatvam, Indriya Prasadnam, Svara Varna Beeja Sampat, Praharsha, Veeryam, Balam, Medha, Ayu, Ojas, Prabha, Uthanam, Santosham</i>
<i>Rasaja</i> – nutrition factor	<i>Ahara Rasa</i> , which helps in the proper formation of <i>Sapta Dhatus</i> /physical growth mainly. <i>Sharirasya Abhinivritti Sharirasya Abhivridhi, Pranandubandhata, Tripti, Pushti, Utsaham, Balam, Varnam, Sthiti Hani, Aloulyam, Buddhi, Vritti</i>
<i>Sattvaja</i> – Mind	Psychological makeup and development. <i>Bhakti, Sheelam, Saucham, Dvesham, Smriti, Moham, Tyagam, Matsaryam, Souryam, Bhayam, Krodham, Tandra, Utsaham, Taikshnyam, Mardavam, Gambhiryam, Anavasthitatvam</i>

Table-2: Effects of various dietetics and mode of life upon the fetus²²

Dietetics or mode of life	Effects on the fetus or Child
Using daily wine	Over thirsty, short memory, and fickle mind
Using meat of iguana often	Suffers from bladder stone, gravel or dysuria (<i>Shanairmeha</i>)
Using Pork often	Red eyes, rough body hair, and obstructed breathing and snoring
Daily use of Fish	Fixed eyes or delayed dropping of eyelids
Daily(excessive) use of sweet articles except milk	Suffers from <i>prameha</i> (urinary disorders) obese and dumb
Daily use of (excessively) sour articles	Suffers from raktavata (bleeding diathesis), and skin and eye disorders.
Daily (excessive) use of salty articles	Early wrinkling, graying of hair, and baldness
Excessive use of hot (<i>katu</i>) articles daily	Weak, possess less quantity of <i>shukra</i> , and infertile.
Excessive use of bitter (<i>tikta</i>) articles daily	Suffers from emaciation (<i>Shosha</i>) or edema (<i>Shopha</i>), weak, scraggy and less digestive powers
Excessive use of astringent (<i>kashaya</i>) articles daily	Blackish (of dark complexion), suffers from <i>anaha</i> (flatulence), and <i>udavarta</i> (eructation)
Use of articles likely to produce disease	Disease according to cause

Tip: *Shlokas* are written with 'Baraha Pad'