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AYURVEDIC INSIGHT TO MODERN GENETICS W.S.R CHROMOSOMAL DISORDERS

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ABSTRACT

Chromosomal disorders represent a significant aspect of genetic abnormalities that can lead to a range of medical conditions, affecting individual health, development, and quality of life. These disorders typically arise from structural abnormalities, such as deletion, duplications, or translocations of chromosomes, or from numerical abnormalities like aneuploidy. While the scientific understanding of chromosomal disorders has advanced significantly through the lens of modern genetics, ancient systems of medicine like Ayurveda offers valuable perspectives on health, disease, and human variability. This paper explores the concept of chromosomal disorders within the framework of Ayurveda, examining its principles related to genetic health. Ayurveda emphasizes the importance of balancing "*Doshas*'' (biological energies), maintaining the integrity of "*Dhatus*" (tissues), and protecting the "*Beeja*" (reproductive essence) to ensure optimal health. The implications of chromosomal disorders can be understood through Ayurvedic concepts such as "*Beeja Bhaga Dushti*", which refers to the potential dysfunctions in the reproductive elements, ultimately influencing progeny health. Discussion elaborates on how these chromosomal abnormalities can be understood under the influence of doshas and '*Beeja Bhaga Dushti*' In conclusion, integrating Ayurvedic insights with contemporary genetic understanding can foster a more comprehensive approach in managing

chromosomal disorders. This intersection of ancient wisdom and modern science not only enhances preventive strategies but also opens avenues for personalized therapeutic interventions that address genetic health within a holistic framework, ultimately contributing to improved reproductive outcomes and overall wellness.

KEYWORDS: Chromosomal disorders, *beeja bhaga dushti*, Genetics, Inheritence.

INTRODUCTION

Beeja Bhaga Dushti, a concept rooted in the ancient science of Ayurveda, refers to the potential dysfunctions or disturbances affecting the reproductive elements, or *beejas*, that contribute to the formation and health of progeny. The term *Beeja* translates to seed, symbolizing the fundamental building block of life, while *Bhaga* denotes a portion or aspects, and *Dushti* signifies a disorder or impairment. In Ayurveda, human beings are viewed as a composite of physical, mental, and spiritual elements, with reproduction considered a critical aspect of human life and continuity. Chromosomes are merely compared with *beeja bhaga* which carries genetic information in genes referred as *beeja bhaga avayava*. We encounter the disorders which occurs due to abnormality in the structure and number of chromosomes such as Down syndrome, Patau syndrome, Edward syndrome, Klinefelter syndrome, Turner syndrome here is an attempt of understanding the causative factors pathogenesis with *Doshic* involvement under the lens of ayurveda, *beeja bhaga dushti* to make the diagnosis, management and assess the *sadhya asadhyata* effectively. This understanding will open up the new ideas to explore the different genetic disorders and correlate with the various *vyadhis* explained in our *samhitas*. Ancient Ayurveda with its roots extending back thousands of years, posits that the diversity in *prakriti,* or individual constitution, arises from variations in genetic determinants. In *Mahatigarbhavakranti adhyaya* of the *sharirasthana acharya charaka* elucidates the concept of three fundamental genetic units: *Beeja, Beejabhag, Beejabhagavayava* . Ayurvedic scholars draws an analogy between *Beeja* and germinal cells, specifically linking it to *Shonita* (the ovum) in females and *Shukra* (the sperm) in males.[1] *Beejabhaga* as constituent of *Beeja*, is aptly compared to modern chromosomes, as it plays a pivotal role in transmission of traits across generations. The third unit, *Beejabhagaavayava*, represents the most elemental entity involved in the inheritance of characteristics, drawing parallels with contemporary understanding of genes and DNA.

According to *Acharya Charaka*, an exacerbation of *doshas* may adversely affect the ovum and sperm, which are integral to the development of specific organs.[2] Consequently, any

vitiation of *Beeja* and *Beejabhaga* could lead to deformities in the organs of the progeny, manifesting both as somatic and genetic anomalies. Factors contributing to this vitiation, as outlined in Ayurveda, include advanced maternal age, premature aging, consanguineous marriages, chronic ailments in parents, and various environmental influences. Further more the *charaka Samhita* indicates that impaired *Beeja, Beejabhaga, and Beejabhagavayava* can result in sexual and reproductive dysfunctions in offspring, leading to the conditions known as *Putipraja* and *Varta* within the context of *Streevyapad*. Similarly, issues such as sterility in children, alongside the conditions of *Putipraja* and *Trinputrika*, fall under the domain of *Purushvyapad*.[3]

Importance of *tridosha* in formation of *Garbha*

In Ayurveda, the concept of *Tridosha* (the three doshas: *Vata, Pitta*, and *Kapha*) plays a crucial role in the process of *garbha* formation (conception and foetal development). According to Ayurvedic principles, the balance of these *doshas* is essential for healthy reproduction and the development of a foetus.

Vata: Responsible for movement and activity, *Vata* governs the processes of conception, fertilization, and early embryonic development. It controls the flow of energy and vital forces, ensuring the proper initiation of pregnancy.[4]

Pitta: Governing transformation and metabolism, *Pitta* helps in the formation of tissues, digestion, and the growth of the foetus. It influences the development of the body's heat, and when balanced, it ensures proper nourishment and metabolic functions.[5]

Kapha: Responsible for stability and nourishment, *Kapha* ensures that the foetus receives adequate nutrition and is provided with a stable and nurturing environment in the womb. It supports the formation of bodily structures and tissues, facilitating the growth and development of the child.[6]

Together, the *Tridosha* must be in harmony for proper *garbha* formation. If there is an imbalance in any of the *doshas*, it can lead to *beeja, beejabhaga, beejabhagavayava dushti* and causes complications in conception, pregnancy, or foetal development.

Genetic inheritance in Ayurveda

The determinants implicated in the genesis of the human embryo or foetal entity can be attributed to various sources, prominently inclusive of maternal essence (*Matrijabhava*), paternal essence (*Pitrijabhava*), the soul (*Atmaja*), the principle of

wholesomeness(*Satmyaja*), the nutritive fluid known as chyle (*Rasaja*), and the cognitive faculties associated with the mind (*satva*).[7]

The contributions deriving from the maternal source encompass a plethora of vital soft tissues, including but not limited to integumentary systems such as skin, circulatory constituents like blood, corporeal substances including flesh and adipose tissues, along with essential visceral components such as the heart and stomach. The paternal contributions may be delineated as follows, the scalp, hair, nails, teeth, and bones, which represent the predominant hard structures, as well as the vascular components such as veins, ligaments, and arteries. Additionally, certain functional attributes derived from the soul (*Atma*) include the innate desires for happiness, courage, intellect, memory, and vocal expression. Factors arising from the principle of wholesomeness (*Satmya*) encompass growth, strength, contentment, and enthusiasm. Moreover, the emotional dimensions attributed to *Sattva* give rise to feelings such as fear, anger, gentleness, vigor, and recollection.

Shukra and *Shonita dushti* as a cause for genetic abnormalities

Shukra and *Shonita Dushti* refer to the imbalance or impurity of *shukra* (semen) and *shonita* (blood) which can affect fertility and reproductive health. Both of these substances carriers the beeja which are fundamental in the process of conception and foetal development.

1. *Shukra Dushti* (Impurity of Semen): According to *Charaka Samhita*, *Shukra* consists of sperms, *beeja* essential for the formation of a healthy offspring. When *Shukra* becomes vitiated due to factors like poor digestion or an unhealthy lifestyle, it leads to problems in conception and foetal development.[8]
2. *Shonita Dushti* (Impurity of Blood**)-** it is mentioned that the purity of blood is vital for the formation of a healthy embryo. Impure blood can lead to complications like miscarriage, foetal abnormalities, or problems during delivery due to abnormal ovum.[9]

Exploring the Intricacies of *Beeja Bhaga Dushti* in chromosomal Anomalies to cause *Vikruti* in *Garbha*

Due to *Dushti* of *Shrukra* and *Shonita* conataining the *Beeja* along with *Beejabhaga avayava* leads to autosomal and Sex chromosomal anomalies and are acknowledged as congenital deviations with pronounced genetic predispositions. *Acharya Charakas* discourse references conditions such as *Dwireta*, typified by true hermaphroditism, where in the majority of the cases reflect a 46XXkaryotype, manifesting both testicular and ovarian tissues. The *Pavanendriya* condition aligns with Klinefelter syndrome (47XXY), marked by unexplained

infertility and azoospermia due to meiotic disruptions, culminating in gametogenic failure. *Narashanda* embodies a male karyotype (XY) yet displays phenotypic female characteristics, exemplifying male pseudo hermaphroditism, while *Narishanda* denotes a female karyotype (XX) with externally male genitalia. Additional ailments encompass *samskarvahi(*anaphrodisia), *Vakri* (hypospadias), *Irsyabhirati* (Mixoscopia), and *Vatikshanda* (eviration) on the other side also mention about the *Putipraja* which relates with defective child.[10]

Chromosomal abnormalities

Chromosomal abnormalities constitutes mainly of two types abnormal in number of chromosomes and alteration in structure of chromosomes.

Non disjunction in chromosome -Nondisjunction is a genetic phenomenon that occurs when chromosomes fail to separate properly during meiosis or mitosis, leading to an abnormal distribution of chromosomes in daughter cells .This error can result in the formation of gametes with an abnormal number of chromosomes, either too many (trisomy) or too few (monosomy).

Alteration in structure of chromosomes includes Deletion, Inversion, Ring chromosomes, Isochromosomes, Translocation – Balanced reciprocal and Robertsonian translocation.[11]

Down's syndrome

Most common type of autosomal trisomy that surviving to birth and one of the commonest cause of intellectual disability

Molecular basis of Downs syndrome has 3 mechanisms

1. Meiotic nondisjunction (maternal non disjunction) – Parents are normal in all aspect this meiotic error mostly occurs in ovum and a maternal age contributes an influence of incidence of trisomy 21
2. Robertsonian Translocation- Extra chromosomal material derives from presence of Robertsonian translocation of long arm of chromosome 21 to another acrocentric chromosome 22/14.
3. Mosaicism – due to mitotic non disjunction of chromosome 21 during early embryogenesis Mixture of cells of 46/47 chromosomes are seen.

Clinical features- flat facial profile, epicanthal folds, Mental retardation, simian crease, hypotonia, cardiac disorders, Alzheimer's disease.[12]

Edward syndrome- trisomy 18 presents with clinical features of prominent occiput, micrognathia, Intellectual disability, rocker bottom feet, cardia disorders.[13]

Patau syndrome- trisomy 13 presents with Mental retardation, cardiac and renal disorders, microcephaly.[14]

Chromosomal abnormality involving sex chromosome

These are much more common than autosomal disorders and are better tolerated due to lyonization / inactivation of one X chromosome and modest amount of genetic material carried by the Y chromosome.

Klinefelter's Syndrome – Male hypogonadism due to meiotic non disjunction in either father or mother presents with clinical features of Eunuchoid body habitus, lack of secondary sexual characteristics, Gynaecomastia, small atrophic testis with small penis, mitral valve prolapse.

Turner Syndrome – Results from complete or partial monosomy of X chromosome and is characterised by hypogonadism in phenotypic female. Presents with Primary Amenorrhea, infertility, webbing of neck, broad chest, failure to develop normal secondary sexual characters, short stature, obesity.[15]

DISCUSSION

The various chromosomal abnormalities arising due to *beeja, beejabhaga avayava dushti* results in different kind of syndromes involving autosomes and sex chromosomes where in there is influence of vitiation of *doshas* during *garbha* formation resulting in different characteristic features.

Down's syndrome- A condition arised due to meiotic non disjunction, Robertsonian Translocation, Mosaicism leading to trisomy 21 can be analysed that doshas like *Vata* is a prime and a major cause for any mitotic or meiosis which underwent abnormality during the Garbha formation in *beeja* (ovum) along with *kapha* and *pitta* which can be taken as *putipraja* (defective child).

Kapha – Imbalance of *kapha* in *beeja* leads to rounded facial, hypotonia, epicanthal folds, delayed motor skills.

Vata- Imbalance of *vata* in *beeja* (ovum) causes Mental retardation, inability to judge and accomplish the action, emotional instability.

Pitta – *Pitta* also contributes to one's memory retention which gets hampered due to its vitiation in Down's syndrome, compromised immune regulation leading to inflammation of tissues and variable appetite.

Edward syndrome – A condition due to trisomy 18 also shows variable *doshic* influences due to *dushti* in *beeja* to give a progeny of *Putipraja* (defective child)
Kapha – Imbalance of *kapha* leads to delayed motor skills, heaviness and stagnation
Pitta- less immune function leading to increased risk of inflammation involving cardiac and renal systems, irritability, anxiety
Vata – Intellectual disability, short neck, micrognathia, rocker bottom feet, irritability.

Patau syndrome – Due to trisomy 13 the vitiation of *doshas* at the time of fertilization to cause one extra chromosome at 13 where is Vata due to its defective karma fails to equally separate the chromosomes to give rise to a child with various defects.
Kapha – *kapha* qualities of growth and solidity reflects in abnormal tissue development to cause cleft lip, cleft palate, polydactyly, hypotonia, sluggishness and heaviness of body.
Pitta – Immune dysregulation causing inflammation in renal and cardiac systems.
Vata – microphthalmia, microcephaly, mental retardation, various other neurological abnormality.

Klinefelter syndrome – Male hypogonadism when two or more X chromosome and one or more Y chromosome due to meiotic non disjunction in ovum or sperm. Here genetic defects in male *beeja bhaga avayava* will produce offspring looks like male but is not actually male known as *Trunputrika*
Vata- Imbalance small atrophic testis with small penis, gynaecomastia, minimal mental retardation, mitral valve prolapses
Pitta – Imbalance of *Pitta* causes increased FSH, Estrogen harmones with decreased Testosterone levels, osteoarthritis, osteoporosis
Kapha- lack of secondary sexual male characters, increased incidence of type 2 diabetic mellitus.

Turner syndrome- complete or partial monosomy of X chromosome and is characterised by hypogonadism in phenotypic female, abnormalities occurring in female *beej bhaga* the progeny resembles female but not a actual female known as *Varta.*

Kapha – Edema of dorsum of hands and feet due to lymph statis and swelling of nape of neck, bilateral neck webbing.

Pitta – failure to develop secondary sexual characters, infertility, amenorrhoea, pigmented nevus.

Vata- short stature, severely fibrotic and atrophic ovaries, broad chest with widely spaced nipples.

Hypothetical Pathogenesis of nondisjunction related to *tridosha* vitiation

Imbalance of *tridosha- Vata* – excessive *Vata* may lead to condition such as stress, anxiety and neuroticism. This negatively impact hormonal and nervous systems, potentially affecting ovarian function and the regulation of ovulation.

Pitta imbalance – lead to high levels of stress hormones which intern causes oxidative stress, which may compromise cellular integrity and influence the processes of cell division. Which lead to errors in chromosome alignment and separation during meiosis and mitosis.

Kapha imbalance – excessive *kapha* may result in sluggishness, obesity, and metabolic disorders. Such conditions can be correlated with insulin resistance and hormonal imbalances, potentially affecting ovarian and testicular function in order to produce effective gametogenesis

CONCLUSION

Through the lens of modern genetics we discussed various chromosomal disorders due to non disjunction during oogenesis and spermatogenesis with a reference in mind of *beeja*, *beejabhaga* and *beejabhaga avayava dushti* in ayurveda. Ayurveda eloborates various abnormalities in *sharirasthana* regarding the causative factors and leading symptoms for a *beeja dushti* dated back to ages. From ayurvedic perspective, these conditions can be understood not only through the genetic lens but also in relation how the imbalances in *tridoshas* will lead to different disorders predominating different conditions in each syndrome. Hence ayurveda gives a collaborative understanding of all *beeja*, *beejabhaga*, *beejabhagaavayava*, *doshas*, *dhatus* in genesis of human genome makeup. It is by detailed analysis one can understand the specific causative factor for viatiation of tridoshas involved in *Beeja Dushti* and there severity by which it causes the abnormalities.

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