ISSN 2456-3110
Vol 9 · Issue 12
December 2024

JAiMS
INTERNATIONAL

Journal of Ayurveda and Integrated Medical Sciences

www.jaims.in

JAIMS

An International Journal for Researches in Ayurveda and Allied Sciences

Maharshi Charaka
Ayurveda

Indexed

Journal of
Ayurveda and Integrated Medical Sciences
ISSN 2456-3110
REVIEW ARTICLE December 2024

Ayurgenomics - A Conceptual Study

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ABSTRACT

Ayurveda, one of the oldest medical systems, provides a detailed and systematic understanding of life processes. The concept of genetics, as described by various ancient Acharyas, is a prominent aspect of Ayurvedic literature. Ayurvedic genetics is an emerging interdisciplinary field that integrates traditional Ayurvedic principles with modern genetic science. In Ayurveda, the concept of *Beeja* (seed) and related terms in classical texts align with the modern understanding of genes and chromosomes. The foundational principles of inheritance, as described in Ayurvedic texts, can be correlated with contemporary genetic theories. This article aims to systematically compare the genetic concepts outlined in ancient Ayurvedic literature with their modern genetic counterparts, highlighting parallels and potential integrations.

Key words: *Heriditary, genetics, Beeja, Bandhya, Putipraja, Varta, Trinuputrik*

INTRODUCTION

Modern Genetic entities such as chromosomes, genes, nucleotide sequence referred in our *Samhitas* with different terminologies as 3 genetic units in the form of *Beeja Beejabhaag, Beejabhagavayav, Beejabhaga Avayav Ekdesh* which are responsible for creating new healthy individual. But if these entities get vitiated, then these are responsible for Heriditary & Congenital anomalies.[1]

Concept of *Beeja*

In both ancient & modern sciences, *Beeja* is considered as fundamental unit for creating new life. It carries entire set of chromosomes necessary for reproduction.

बीज - बीज इति शुक्रशोणिते । (च.शा.३/१७ चक्रपाणि टीका)

Beeja is considered as *Shukra & Shonita* & are initiators for creating new life.[2]

Shonita is also termed as *Artava* & has ability for fertilization when female reached to 14 years of age.

शुक्रशोणितजीवसंयोगे तु खलु कुक्षिगते गर्भसंज्ञा भवति ॥ Ch.Sha.4

When *Shukra* of male meets with *Shonita* of female in her *Kukshi* along with *Jeeva*, then it will lead to formation of *Garbha* (fetus).[3]

Concept of *Beejabhaag*

Beejabhaag is the part of *Beeja* which is related to formation of body parts.[4]

Conept of *Beejabhaagavyav*

Beejabhaagavyav is the component lying inside *Beeja*. Chromosomes are passed on as units from generation to generation. Chromosomes carry hereditary information in the form of *genes*.[5]

From term *Garbhashyabeejabhaag Avyav*, men's smaller part of genetic material i.e. chromosomes concerned with formation of *Garbhashya & Artava.*

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Submission Date: 05/11/2024 Accepted Date: 21/12/2024

Access this article online

Quick Response Code

Website: www.jaims.in

DOI: 10.21760/jaims.9.12.18

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Concept of *Beejabhaagavyavekdesh*

Genes are the basic physical & functional unit of heredity. These are responsible for expression of particular trait in an individual, that are transmitted from one generation to another

यश्चोतं-यदि व मनुष्यो मनुष्यप्रभवः, कस्मान्न जडादिभ्यो जाताः पितृसहशरूपा भवन्तीति ||

As it is considered in Samhitas, life comes from life (man from man) then why progeny from deformed body parts of parents not always have deformed body parts.[6]

यस्य यस्य ह्यवयवस्य बीजे बीजभागे वा दोषाः प्रकोपमापद्यन्ते, तं तमवयवं विकृतिराविशति । Ch.Sha 4/30

It is said that in the *Beeja*, the part of body which is damaged in its genetic source gets abnormality otherwise not.

Even it is said, leprosy suffered parents tranmit disease to offspring, if genes are affected otherwise not.

Causes of congenital anomalies

- Idiopathic (60-70 %)
- Genetic Factors
- Chromosomal abnormalities
- Single-gene defects
- Autosomal dominant disease: Achondroplasia, Marfan syndrome, neurofibromatosis
- Autosomal recessive disease: Cystic fibrosis, galactosemia, sickle cell anemia
- Environmental Agents (Teratogens)
- Drugs: Alcohol, anticonvulsants (phenytoin sodium, valproate, carbamazepine, anticoagulants (e.g. warfarin), anticancer drugs and androgens
- Chemicals: Organic mercurial and organic solvents
- Maternal / placental / intrauterine infections: Rubella (German measles), cytomegalovirus, toxoplasmosis, human immunodeficiency virus (HIV), herpes simplex, varicella zoster, influenza.
- Mumps
- Physical agent: Ionizing radiation and hyperpyrexia
- Maternal illness/disorders: Epilepsy, diabetes, phenylketonuria, iodine deficiency
- Multifactorial: Neural tube defects, congenital heart disease, cleft lip and cleft palate.[7]

Beejabhaagdushti

When *Beeja* or *Beejabhaaga* involved in reproduction is extremely vitiated, then there will be production of male & female sterile child (*Bandhya*). It includes ovular & spermatic defects.[8]

Ovular defects

- Defective oogenesis
- Blighted ovum
- Luteal phase defect
- Anovulation
- Oocyte maturation defect
- Luteal unruptured follicle syndrome
- Autoimmune oophoritis
- Mullerian duct anomaly

Spermatic defects

- Defective spermatogenesis
- Cryptorchidism
- Wolffian duct anomaly
- Persistent mullerian duct syndrome

Beejabhaagavyav Dushti

Putipraja Offspring with modern perspective as chromosomal defects leads to deformed limbs & organs.

When *Beejabhaagavyav* involved in reproduction is vitiated, then there will be production of *Putipraja* offspring

पूतिप्रजामितिम्रियमाणापत्याम्; अन्ये तु क्लिन्नाङ्ग्गप्रत्यङ्गां पूतिमाहुः ।

Puti word implies to child having deformed limbs & organs[9]

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Numerical abnormalities	Structural abnormalities
Aneuploidy (extra/missing chromosome) Monosomy Trisomy Tetrasomy	Inversion Translation Ring chromosome Isochromosome

- **Down Syndrome (Trisomy 21):**

This condition is caused by an extracopy of chromosome 21.

Individuals with Down syndrome may have congenital heart defects, gastrointestinal issues, and can have a variety of physical and developmental abnormalities including shorter limbs.

- **Turner Syndrome:** This occurs in females who have only one X chromosome instead of two (45,X).

It can lead to short stature, heart defects, and sometimes skeletal abnormalities.

- **Ring Chromosome Syndromes** (e.g., Ring Chromosome 14 Syndrome, Ring Chromosome 20 Syndrome)
- Formation of a ring chromosome due to the loss of both ends of a chromosome, such as chromosome 14 or 20.
- Intellectual disability, seizures, growth delays, and sometimes limb deformities depending on the affected genes in the deleted portions of the chromosome.
- **Robertsonian Translocations -** Chromosomal Abnormality, Fusion of two acrocentric chromosomes (such as 13, 14, 15, 21, 22), leading to either partial monosomy or trisomy in the offspring.
- Depending on which chromosomes are involved, deformities may include heart defects, limb abnormalities (e.g., shortened or extra limbs), and organ malformations such as kidney or brain defects.

- **Trisomy 13 (Patau Syndrome)**
- Extra copy of chromosome 13.
- Severe limb deformities such as polydactyly, rocker-bottom feet, cleft lip/palate, severe heart defects, and abnormalities in the brain, kidneys, and other organs.

- **Trisomy 18 (Edwards Syndrome)**
- Extra copy of chromosome 18.
- Severe developmental delays, rocker-bottom feet, clenched hands with overlapping fingers, heart defects, kidney malformations, and other organ abnormalities.

- **Cri-du-Chat Syndrome (5p Deletion Syndrome) -**
- Deletion of the short arm of chromosome 5 (5p)
- Intellectual disability, microcephaly (small head size), low birth weight, heart defects, and characteristic facial features. Limb deformities are not as common but developmental delays can affect motor skills.

- **Wolf-Hirschhorn Syndrome**
- Deletion of the short arm of chromosome 4 (4p)
- Severe growth retardation, intellectual disability, seizures, characteristic facial appearance (Greek helmet-like face), heart defects, and skeletal abnormalities, including limb deformities such as clubfoot or hip dislocation.

- **Cat Eye Syndrome -** Duplication or triplication of a segment of chromosome 22
- Anal atresia, coloboma (eye abnormality), heart and kidney malformations, and sometimes skeletal deformities like shortened limbs or polydactyly (extra fingers or toes)

- **Tetrasomy 12p (extra isochromosome 12p) -**
- Intellectual disability, distinctive facial features, diaphragmatic hernia, and skeletal abnormalities, including shortened limbs or abnormal joint development.[10]

Beejabhaagavyavekdesh Dushti

Varta and *Trinuputrik*

Genes are the finer structures that determine specific hereditary traits. Variation in these genes lead to different expression of genes (mutation of genes)

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वार्तातृणपुत्रिकयोर्व्यवायेच्छा परं भवति, नतु व्यवायसामर्थ्यमिति ब्रुवते | (च शा ४/३१ चक्रपाणि टीका)

If there is genetic defect in female *Beejabhaagavyav Ekdesh*, progeny will be predominantly female shaped (phenotypically) but actually not female, named as *Varta*.

अस्त्रियमिति असंपूर्णस्त्रीलक्षणाम् । (च शा ४/३० चक्रपाणि टीका)

Varta Offspring with modern perspective as phenotypically female but genetically male[11]

- **Androgen Insensitivity Syndrome (AIS)** - Mutations in the **__AR gene__* (Androgen Receptor gene), located on the X chromosome. -

Genotype: 46, XY (genetically male).

In AIS, the body is unable to respond to male hormones (androgens) due to a defective androgen receptor.

Individual has male XY chromosomes and internal testes, they develop female external characteristics such as breasts and female genitalia.

They do not develop male traits or internal female reproductive organs (like a uterus or ovaries).

- **Swyer Syndrome (46, XY Gonadal Dysgenesis)-** Mutations in genes involved in sex determination, such as the **__SRY gene* on the Y chromosome or other genes like *NR5A1, **DHH, or **MAP3K1*.**

***Genotype*: 46, XY (genetically male).**

In Swyer Syndrome, despite having XY chromosomes, the testes do not develop properly, and the body develops as phenotypically female.

Individuals have typical female external genitalia but lack ovaries, and their gonads remain underdeveloped (streak gonads).[12]

If there is genetic defect in male *Beejabhaagavyavekdesh*, progeny will be predominantly male shaped(phenotypically) but actually not male named as *Trinuputrik.*

अपुरुषमिति असमस्तपुरुषलक्षणयुक्तम्

Trinuputrik offspring with modern perspective as phenotypically male but genetically female[13]

- **Congenital Adrenal Hyperplasia (CAH)** due to 21-Hydroxylase Deficiency*
  - Mutations in the **__CYP21A2 gene__* affecting steroid hormone production.
  - **Genotype*: 46, XX (genetically female**)
  - CAH causes the adrenal glands to produce excess androgens (male hormones), which can cause the external genitalia to appear male even though the person is genetically female (46, XX). Individuals may develop male-like features such as an enlarged clitoris that can resemble a penis, but they still have female reproductive organs (ovaries and uterus).
- **XX Male Syndrome (De la Chapelle Syndrome)**
  - The **__SRY gene__* (Sex-determining Region Y gene), which is normally on the Y chromosome, gets translocated onto one of the X chromosomes during sperm formation.
  - **Genotype: 46, XX (genetically female, but with the SRY gene**).
  - In this condition, individuals have two X chromosomes (which is typical for females), but because they possess the SRY gene, they develop male physical characteristics. They are phenotypically male (develop male genitalia), but since they lack the rest of the Y chromosome, they are sterile and typically do not have functioning testes.
- **Ovotesticular Disorder (True Hermaphroditism)**
  - Genetic defect: Various genetic causes, including mosaicism, chimerism, or mutations in genes such as **__SOX9* or *RSPO1__*.
  - ***Genotype*: Can be 46, XX, 46, XY, or mosaic (with both XX and XY cells).**
  - In ovotesticular disorder, individuals can have both ovarian and testicular tissue. Phenotypically, they may have male genitalia or ambiguous genitalia, but their genetic makeup may be female (46, XX) or mixed (mosaic). The external appearance may be male, but internally, they may have both male and female reproductive structures.[14]

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Genetic Disorders

- Monogenic Disorder - Sex Chromosome Related Disorders
- Polygenic Disorder - Autosomal Disorders

Sex Chromosome Related Disorders

X linked inheritance		Y linked inheritance
Dominant	**Recessive**	Swyer syndrome
Fragile X syndrome Rett's syndrome Hypophosphatemia	Colour blindness Haemophillia A Duchenne muscular Dystrophy	Holandric inheritance

Discussion

By taking whole modern concepts of genetics into consideration, following Ayurvedic inferences that can be drawn are:

Genes are the basic & fundamental unit of all the living organisms. *Beeja* carries all the genetic material that is transmitted from parents to offspring. It has also been noticed that not only genetic factors, but also the epigenetics plays important role in genetic defects.

In Ayurveda, it is considered that not all the genetic traits are passed from one generation to other, but it is only the part of body which is damaged in its genetic constitution gets abnormality in fetus otherwise not.

Beejadushti in the form of *Beejabhaag, Beejabhagavyav, Beejabhaagavyav Ekdesh Dushti* as *Bandhya, Putipraja (Varta & Trinuputrik)* respectively can be directly or indirectly correlated with various genetic disorders or syndromes as:

Beejabhaagdushti: Germ cell defects, which includes spermatic & ovular defects.

Beejabhaagavy Dushti: Chromosomal defects resulting in deformed limbs & organs.

Beejabhaagavyavekdesh Dushti: Genetic defects (either monogenic or polygenic disorders) resulting in difference in phenotypical & genotypical constitution in same individual.

Conclusion

By considering above references, we can conclude that genetic hereditary disorders explained in today's modern science is very widespread topic. But our *Acharyas* explained the hereditary genetic disorders beautifully in ancient literatures in terms of *Beejadushti*, which can be very well correlated with modern genetics as *Beejabhaagdushti* as Germ cell defects, *Beejabhaagavyav Dushti* as Chromosomal defects, *Beejabhaagavyavekdesh Dushti* as Genetic defects.

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How to cite this article: Shubhangi Sood, Soni Kapil. Ayurgenomics - A Conceptual Study. J Ayurveda Integr Med Sci 2024;12:151-156. http://dx.doi.org/10.21760/jaims.9.12.18

Source of Support: Nil, **Conflict of Interest:** None declared.

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