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Chapter 16

Concept of Genetics and Genetic Disorders in Ancient Indian Medical Science Ayurveda

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Abstract

Ayurveda is an ancient medical science of India. The roots of many concepts of modern medicine are deep seated in Ayurveda. Genetics is one such concept which has its roots in Ayurveda. The word 'Gene' probably have developed from the word 'Genesis' which in turn might have evolved from the Sanskrit word *'Janana'* which literally means to produce. Since Sanskrit being Indo-European language, many Sanskrit medical words can be traced in modern medicine. Due to evolution of individual specific treatment in present era, it becomes necessary to study the genetic concept in Ayurveda. Ayurveda had used the word *'Sahaj Vyadhi or Aadibalpravrutta Vyadhi'* to denote all genetic abnormalities. The list of genetic diseases given in Ayurveda includes Hereditary Diabetes (*Sahaj Prameha*), Hereditary Piles (*Sahaj Arsha*), Hereditary Impotency (*Sahaj Klaibya*), Hereditary leukoderma (*Sahaj Kushtha*), Hereditary obesity (*Sahaj Sthaulya*) and reproductive diseases such as (*Garbha Prastravati* (Hereditary tendency for habitual abortion), *Alpa-ayu Prajayate* (Short lived sibling like Aicardi syndrome), *Virupa Prajayate* (Sibling with Structural deformity), *Sahaj Karnapali Vikar* (Hereditary disorders of Ear Pinna) and *Khanda Aushtha (Cleft lip)* etc. Ayurveda also talks about the genetically determined body power (Sahaj Bala) and natural immunity (*Sahaj Vyadhikshamatva*). The present study aims to study the concept of genetics in Ayurveda. It is observed and concluded that the concept of genetics is very well mentioned in Ayurveda and probably the modern genetics have evolved from it.

Keywords: Genetics, *Sahaj, Prakriti, Vyadhi*.

1. Introduction

As per Ayurveda the smallest unit of the body is the *Parmanu* (atom) which refers to the modern concept of cell. As per Ayurveda, after sexual intercourse, the *parmanu swarupa* (atomic) *Shukra Beeja* (sperm) and *Stree Beeja* (ovum) unite togive rise to the Sanyukta Beeja which is referred as zygote in modern science. Ayurveda also says that as per the maternal and paternal genetic influence during embryogenesis, organs with maternal and paternal make up shapes up. Ayurveda had also mentioned that during embryogenesis, these body *Parmanus* divides and redivides to give rise to different organs, systems etc. Ayurveda is of the view that during this process of embryogenesis, if any part of the Beeja (zygote) gets affected, then the genetic abnormality of that part is noticed in later life. As per ayurveda the individual resembling like the mother or father also due to inheritance of genetic maternal or paternal traits. *Prakriti* (Individual constitution) is one such concept which is genetically determined during the process of embryogenesis.

In Ayurveda, hereditary disorders are referred as *"Adibala Pravrutta Vikara* or *Sahaj Vikara"*. The list of genetic diseases given in Ayurveda includes *Sahaj Prameha* (*Hereditary* Diabetes), Hereditary obesity (*Sahaj Sthaulya*), *Sahaj Arsha* (Piles), *Sahaj Klaibya* (Impotency), *Sahaj Kushtha* (leukoderma), And reproductive diseases such as (*Garbha Prastravati* (Hereditary tendency for habitual abortion), *Alpa-ayu Prajayate* (Short lived sibling like Aicardi syndrome), *Virupa Prajayate* (Sibling with Structural deformity),*Sahaj Karnapali Vikar* (Hereditary disorders of Ear Pinna) and *Khanda Aushtha* (Cleft lip).

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2. Material & Method

Literary descriptive method of research is adopted in the present study. Critical study of Ayurvedic and modern literature is done to come to logical conclusions.

Review Of Literature

Genetic Determination of Sex As Per Ayurveda

The sex determining factors depends upon the past deeds of the individua as well as on the degree of involvement of *Panchabhautik* constitution of the semen and ovum and the factors that determines the sex of the baby. If the feminine factors are predominant in the genetic make-up of zygote, then female baby will be born. If there is predominance of masculine characters in the genetic makeup of zygote then male baby will be born. If there is equality of male and female characters in the genetic makeup of zygote then eunuch baby will be born [1].

Here, the predominance of sexual characters in the in the genetic makeup of zygote stated by acharya Charaka might be referring to the genetic information on chromosome. As per modern genetic studies, it is proved that XX combination of chromosomal pair result in female sex baby and XY combination of chromosomal pair result in male sex baby. Whereas XXY combination of chromosomal pair result in eunuch baby which is neither a male nor female.

Although this is true, the particular set of combination of chromosomes may be occurring due to dominance of masculine or feminine characters in parents. Female have XX pair of sex determining chromosome whereas male have XY set of pair sex determining chromosome. How the particular combination should take place may be attributed to the Ayurvedokta theory of dominance of masculine or feminine characters. For e.g. dominance of masculine characters during mixing of semen and ovum may cause XY combination of chromosomes resulting in male progeny whereas dominance of feminine characters during mixing of semen and ovum may cause XX combination of chromosomes resulting in female progeny.

Concept of Sex Chromosomes & Genetic Abnormalities In Ayurveda

The sex determining part of the ovum (*Streekaranam Sharir Beeja Bhaga*) and the sex determining part of the sperm (*Purushkaranam Sharir Beej Bhaga*) mentioned by ancient Ayurvedic acharyas definitely indicate towards the X and Y chromosomes proved by modern studies which are responsible for the sex determination of the child.

When a woman consumes the dosha vitiating faulty diet, the vitiated morbid doshas traverses to her reproductive organs and partially vitiates the uterus and the ovum. In such condition when that woman conceives then any or many maternal organ of the zygote may get affected. The organ which is going to be evolved from the zygote, if gets vitiated by the morbid doshas then in later life that organ exhibit the pathology. If the genes in the ovum expressing the uterus gets affected by morbid doshas then such woman may deliver the sterile child. If the gene in ovum expressing any part of the reproductive tract gets vitiated by morbid doshas then such woman delivers the child with foul smelling reproductive organ. If the gene in ovum expressing the secondary sexual characters in woman gets affected by the morbid doshas then such a woman delivers the child which is deficient in female secondary sexual characters. This child in adult life may appear like female but it is not the complete female and is unable to deliver the child. Such an offspring is called as *'Varta Stree Vyapad'* (abnormal woman) [2].

This could be modern Turner's syndrome which occurs due to missing or partial X chromosome. Females have only one X chromosome instead of two [45X0]. This is characterized with short stature, delayed puberty, undeveloped breasts, puffy hands and feet, inability to reproduce.

Likewise, in males if the entire sperm is affected with morbid doshas then offspring which is developed remains infertile (male infertility). When only part of the sperm is affected with morbid doshas then offspring which is developed has foul smelling body (*Puti Praja*). When the morbid dosha affects only the part of sperm and the sex determining part of the sperm (Y chromosome) then eunuch naming *'Trinaputrik'* is delivered which in adult life appears like female with masculine characters. Such child has the desire to perform sex but unable to do so. This is referred as *Purush Vyapad* (Abnormal male).

Trinaputrik purush Vyapada could be modern Klinefelter Syndrome in which affected male has extra X chromosome [47XXY]. They have enlarged breast, small testicles, low libido, less body hair. Often symptoms may be subtle and many peoples do not realize that they are affected.

Hereditary/Genetic Disorders In Ayurveda

In Ayurveda, hereditary disorders are referred as *"Adibala Pravrutta Vikara or Sahaj Vikara"* (Diseases passed down from genes of parents). And are mentioned as follows :

Causes of Hereditary Disorders As Per Ayurveda [3]

Shukra Beeja Doshanwaya - Genetic defects in sperm
Shonit Beeja Doshanwaya - Genetic defects in ovum

Classification of Hereditary Disorders As Per Ayurveda [4]

Matruja Sahaj Vikarani - Maternal / X linked hereditary disorders.
Pitruja Sahaj Vikarani - Paternal / Y linked hereditary disorders.

Examples of Hereditary Disorders As Per Ayurveda

Sahaj Arsha (Hereditary Piles) [5]
Sahaj Madhumeha (Hereditary Diabetes mellitus) [6]
Sahaj Sthaulaya (Hereditary Obesity) [7]
Sahaj Kushtha (Skin diseases like leukoderma) [8]
Alpa-ayu Prajayate (Short lived sibling like Aicardi syndrome) [9]
Virupa Prajayate (Sibling with Structural deformity) [10]
Sahaj Klaibya (Hereditary Impotency/ infertility) [11]
Garbha Prastravati (Hereditary tendency for habitual abortion) [11]
Klibam Prajayate - Hereditary Impotency in offsprings [12]
Na Jayate Garbha - Hereditary Infertility [12]
Shukra Avisarga – Offsprings with Ejaculatory failure [12]
Sahaj Karnapali Vikar (Hereditary disorders of Ear Pinna) [13]
Khanda Aushtha (Cleft lip) [14]

Inheritance of Maternal & Paternal Characters as Per Ayurveda

The human gametocyte in itself contains the seeds of self like different organs from which the self like human being is generated with all the organs on germination [15].

As per Ayurveda, the autosomal genetic expression from mother's side gives rise to the *'Matruj Avayava'* like heart, lungs, liver, spleen, intestine, kidneys and muscles whereas somatic genetic expression from father's side gives rise to the *'Pitruj Avayava'* (organs) like bones, hairs, dentures, nails, blood, vessels, ligaments and sperm (sex determinant) [16].

The genetic defects in the organs to be expressed (*upatapta Beejabhaga*) of the ovum or sperm will lead to the abnormality of those organs in later life [17].

The genetic defect in the organs to be expressed (*upatapta beejabhaga*) of the sperm (pitru beeja) will cause abnormality of those organs [18]. For e.g. if there is genetic defect in the reproductive part expression of the sperm then offspring will have male infertility problem. And the same has been listed while enumerating the genetic reproductive disorders. In this context acharya Charaka in sutrasrthana chapter 28, verse no. 18 and Sushruta in sutrasthana chapter 24, verse no. 9 have mentioned the paternal genetic defects in the sperm and the resultant anomaly in the individual.

Modern Genetic Diseases

This refers to the diseases passed from the parent's genes either due to the abnormalities in genes or mutations in the genes or due to changes in the DNA [19].

Classification

A. Mendelian Disorders (Caused by Single Gene Defects) [20]

Autosomal dominant disorders
Autosomal recessive disorders
X- Linked disorders
Y linked disorders
Single gene defect with atypical pattern of inheritance (Fragile X syndrome)
Diseases by mutations in mitochondrial genes (Hereditary optic neuropathy causing blindness)
Cytogenic disorders involving sex chromosomes (Turners's, Klienfelter's syndrome)
Cytogenic disorders involving autosomes (Down's syndrome)

It occurs due to the single muted gene. It can be passed by several ways. It can be carried by single or both the parents. It is further divided into following 2 types :

1. **Autosomal dominant disorders**
   A single somatic muted gene may be passed by either of the parent. Though parents are carriers, but it's not necessary that gene will express in offspring. It may or may not express to cause a disease.
   Examples
   Marfan's syndrome
   Polycystic Kidney disease
   Neurofibromatosis type 1
   Huntington's disease
   Spherocytosis of RBCs
   Familial Hypercholesteremia
2. **Autosomal recessive disorders**
   In this type affected person gets two copies of muted gene passed separately from the carrier but unaffected mother and father.
   Examples
   Sickle cell disease
   Glycogen Storage disease
   Galactosemia

Phenylketonuria
Cystic fibrosis
Tay Sachs disease
Pyruvic Kinase deficiency
Niemann Pick disease

3. **X- Linked disorders**
   In this group, females are the carriers i.e. mutant gene is present on X chromosome only.

   (a) **X- Linked dominant**
   Males and female siblings both are affected in this disorder with male preponderance.
   Examples
   Aicardi syndrome
   Rett Syndrome

   (b) **X linked recessive**
   In this group, male offspring is affected mainly and females are relatively spared. **Examples**
   Duchenne's muscular dystrophy
   Hemophilia-A
   G6PD deficiency
   Colour blindness

4. **Y-linked disorders**
   They are caused by mutations on the Y chromosomes. These disorders can be passed only by males to their male offspring. Females can never be affected as they don't have Y chromosome.
   Examples
   Hereditary Infertility
5. **Diseases by mutations in mitochondrial genes**
   As only ovum contribute mitochondria to developing embryo, these disorders are inherited from mothers only.
   Examples
   Hereditary optic neuropathy causing blindness

B. Disorders with multifactorial inheritance [21]

They occur in combination with lifestyle and environmental factors. These types of polygenic disorders tend to run in families but their inheritance do not follow simple pattern of Mendelian diseases.**Examples**

Hypertension
Diabetes
Asthma
Cancers
Cleft Palate
Heart diseases
Infertility
Refractive errors
Mood disorders like Schizophrenia
Intellectual disability
Autoimmune disease such as multiple sclerosis

Hereditary obesity [22]

It can be classified as monogenic, polygenic or syndromic.

Monogenetic Obesity – It is due to mutations in the leptin signalling pathways causing suppression of anorexigenic pathways and activation of orexigenic pathways. It may be autosomal recessive or dominant.

Polygenic Obesity – 60% of the inherited obesity is polygenic. It is associated with the mutations in the gene CYP27A1, TFAP2B, PARK2, IFNGR1, UCP 2 & 3 which codes for uncoupling of proteins in brown adipose tissue.

Syndromic obesity – It may be caused by chromosomal rearrangements like Prader Willi Syndrome, WAGR Syndrome, Bardet Biedl Syndrome or mutation syndrome like Alstrom syndrome, Carpenter Syndrome, Cohen Syndrome.

Diagnosis Of Hereditary diseases

It can be done through following techniques:
Aminocentesis
USG
Family history
Chromosomal study
DNA study

3. Observations

Hereditary Disorder Category As Per Ayurveda

Table 1: Metabolic Disorder Examples

Sr	Ayurvedic Name	Modern Name
1	*Sahaj Madhumeha*	Hereditary Diabetes mellitus
2	*Sahaj Sthaula*	Hereditary obesity

Table 2: Skin Disorder Examples

Sr	Ayurvedic Name	Modern Name
1	*Sahaj Arsha*	Hereditary Piles
2	*Sahaj Kushtha*	leukoderma

Table 3: Reproductive Disorder Examples Cytogenic disorders involving sex chromosomes

Sr	Ayurvedic Name	Modern Name
1	*Sahaj Klaibya*	Hereditary Impotency
2	*Alpa-ayu Prajayate*	Short lived sibling [Aicardi syndrome]
3	*Virupa Prajayate*	Sibling with Structural deformity
4	*Garbha Prastravati*	Hereditary tendency for habitual abortion
5	*Varta Stree Vyapad*	Turner's syndrome
6	*Trinaputrik purush Vyapada*	Klinefelter Syndrome

Table 4: Structural abnormality Examples

Sr	Ayurvedic Name	Modern Name
1	*Sahaj Karnapali Vikar*	Hereditary disorders of Ear Pinna
2	*Khanda Aushtha*	*Cleft lip*

Table 5: Endocrinal Disorders

Sr	Ayurvedic Name	Modern Name
1	*Ati Hrasva* (Dwarfism)	*Ati Deergha* (Giantism)
2	*Ati Sthula* (Hereditary Obesity)	*Ati Krusha* (Hereditary leanness)
3	*Ati Gaura* (Hereditary Albinism)	*Ati Krushna* (Hereditary Hypermelanosis)
4	*Ati Loma* (Hereditary Hypertrichosis)	*A Loma* (Hereditary Hypotrichosis)

4. Discussion

Bija refers to the male and female gamete; i.e. *Shukra* (sperm) and *Shonit* (ovum) respectively. As per Ayurveda, the fusion of male gametocyte (*Shukra*) and female gametocyte (Shonita) in the uterus (*Garbhashaya*) gives rise to the germinating embryo in which life force (*Jiva*) generates to call it as foetus (*Garbha*). The foetus is called 'Garbha' as it develops in *Garbhashaya* (uterus).

Acharya Chakrapani refers *Bijabhaga* as that part of the sperm or ovum from which organogenesis starts. For e.g. the part in the ovum from which the uterus is expressed, is called as *'Garbhashaya Beejabhaga'* In this sense the' *Janak Bijabhaga'* refers to the chromosomal part of the sperm or ovum on which the specific expressive genes are located.

The fusion of male gametocyte and female gametocyte in the uterus to form zygote is mainly the fusion of maternal and paternal nuceli causing haploid nucleus to become diploid with chromosomal exchange and linkage. The 23 chromosomes from the nucleus of ovum and 23 chromosomes from the nucleus of sperm combine together to have joint genetic information of mother and father. The embryonic stem cell express specific sets of gene that determines their ultimate organ cell type.

As per modern science, Organogenesis refers to the formation of organs from the germ layers. Each germ layer gives rise to specific tissue type. Organs develop from the germ layers through the process of differentiation during which the embryonic stem cell express specific sets of gene that will determine their ultimate cell type.

Acharya Charaka is of the view that the autosomal genetic expression from mother's side gives rise to the *'Matruj Avayava'* like heart, lungs, liver, spleen, intestine, kidneys and muscles whereas somatic genetic expression from father's side gives rise to the *'Pitruj Avayava'*(organs) like bones, hairs, dentures, nails, blood, vessels, ligaments and sperm (sex determinant). This concept of maternal and paternal organs is obscure in modern science and needs to be explored with modern technical advancement.

As per acharya Sushruta the genetic disorders result due to genetic defects in sperm or ovum.

Acharya Charaka is of the view that genetic defects in the organs to be expressed (*upatapta Beejabhaga*) of the ovum or sperm will lead to the abnormality of those organs in later life. This is evident from the list of genetic reproductive disorders given by acharya Charaka and Sushruta such as (*Garbha Prastravati* (Hereditary tendency for habitual abortion), *Alpa-ayu Prajayate* (Short lived sibling like Aicardi syndrome), *Virupa Prajayate* (Sibling with Structural deformity),*Sahaj Klaibya* (Hereditary erectile dysfunction) *Sahaj Karnapali Vikar* (Hereditary disorders of Ear Pinna) and *Khanda Aushtha* (Cleft lip).

'Varta Stree Vyapad' (abnormal woman) mentioned by acharya Charaka, could be modern Turner's syndrome which occurs due to missing or partial X chromosome. Females have only one X chromosome instead of two [45X0]. This is characterized with short stature, delayed puberty, undeveloped breasts, puffy hands and feet, inability to reproduce.

Trinaputrik purush Vyapada could be modern Klinefelter Syndrome in which affected male has extra X chromosome [47XXY]. They have enlarged breast, small testicles, low libido, less body hair. Often symptoms may be subtle and many peoples do not realize that they are affected.

In Ayurveda, we do not find classification of genetic disorders like which is mentioned in modern science. However, we do find the Sex linked classification in the form of Maternal (*Matruj*) and Paternal (*Pitruj*) genetic (*Sahaja*) diseases. The other classification of genetic diseases that can be traced in Ayurveda is the disorders with multifactorial inheritance. The genetic reproductive disorders mentioned in Ayurveda can be included in either X-linked (e.g. *Garbha Prastravati* (Hereditary tendency for habitual abortion, *Alpa-ayu Prajayate* (Short lived sibling like Aicardi syndrome)) or Y linked genetic disorders(e.g. *Klibam* (Hereditary erectile dysfunction). The list of other genetic diseases given by acharya Charaka and Sushruta such as *Sahaj Prameha* (*Hereditary* Diabetes), Hereditary obesity (*Sahaj Sthaulya*), *Sahaj Arsha* (Piles), *Sahaj Kushtha* (leukoderma), may be included in the list of disorders with multifactorial inheritance. These disorders occur in combination with lifestyle and environmental factors. These types of polygenic disorders tend to run in families but their inheritance do not follow simple pattern of Mendelian diseases. Though the credit of evolving genetics as separate branch of science is given to Gregor Johann Mendel (1822-1884) but the concept of genetics and genetic disorders can very well be traced in ancient Indian medical science that is Ayurveda.

5. Conclusions

The concept of genetics and genetic disorders can very well be traced in ancient Indian medical science that is Ayurveda. The Ayurvedic classification of genetic disorders include X linked (*Matruja*) and Y linked (*Pitruja*) genetic diseases along with the multifactorial inheritance of diseases.

The Ayurvedic mentioned disease*Garbha Prastravati* (Hereditary tendency) for habitual abortion, *Alpa-ayu Prajayate* (Short lived sibling like Aicardi syndrome) can be classified under X-linked genetic disorders.

The Ayurvedic mentioned disease *Klibam* (Hereditary erectile dysfunction) can classified under Y-linked genetic disorder.

The Ayurvedic mentioned diseases such as *Sahaj Prameha* (Hereditary Diabetes), *Sahaj Sthaulya* (Hereditary obesity), *Sahaj Arsha* (Piles), *Sahaj Kushtha* (leukoderma), can classified under disorders with multifactorial inheritance.

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