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Critical Evaluation of Concept of Genetics WSR to Hereditary Diseases (*Sahaj Vikaras*) in Ayurveda

Subhash D Waghe*

M.D. (Diagnostics & Pathology) Dept. of Roganidana - Sarvepalli Radhakrishnan College of Ayurveda & Research Center, SRK University, India

***Corresponding author:** Subhash D Waghe, M.D. (Diagnostics & Pathology) Dept. of Roganidana - Sarvepalli Radhakrishnan College of Ayurveda & Research Center, SRK University, Bhopal – 462 047 M.P, India

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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

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 to give 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 Ar-*

sha (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).

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

Hereditary/Genetic Disorders in Ayurveda

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

Causes of Hereditary Disorders as Per Ayurveda [1]

1. Shukra Beeja Doshanwaya - Genetic defects in sperm
2. Shonit Beeja Doshanwaya - Genetic defects in ovum.

Classification of Hereditary Disorders as Per Ayurveda [2]

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

Examples of Hereditary Disorders as Per Ayurveda

1. *Sahaj Arsha* (Hereditary Piles) [3].
2. Sahaj Madhumeha (Hereditary Diabetes mellitus) [4].
3. Sahaj Sthaulaya (Hereditary Obesity) [5].
4. *Sahaj Kushtha* (Skin diseases like leukoderma) [6].
5. Alpa-ayu Prajayate (Short lived sibling like Aicardi syndrome) [7].
6. Virupa Prajayate (Sibling with Structural deformity) .
7. *Sahaj Klaibya* (Hereditary Impotency/ infertility) .
8. Garbha Prastravati (Hereditary tendency for habitual abortion) .
9. Sahaj Karnapali Vikar (Hereditary disorders of Ear Pinna) [8].
10. Khanda Aushtha (Cleft lip) [9].

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. [10] 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) [11].

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 [12]. The genetic defect in the organs to be expressed (upatapta beejabhaga) of the sperm (pitru beeja) will cause abnormality of those organs. 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 [13].

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 .

Classification

A. Mendelin Disorders (Caused by Single Gene Defects)

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

B. Disorders with multifactorial inheritance (DM, HTN, Schizophrenia)

a Mendelin Disorders (Caused by Single Gene Defects)

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 type1
- 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.

1. X- Linked Dominant

Males and female siblings both are affected in this disorder with male preponderance.

Examples :

- Aicardi syndrome
- Rett Syndrome

2. 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

3. Y Linked Lisorders

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

4. 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

They occur in combination with lifestyle and environmental factors. These type of polygenic disorders tend to run in families but their inheritance do not follow simple pattern of Mendelian diseases. (Tables 1-5)

Examples :

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

Table 1.

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

Table 2.

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

Table 3.

Sr	Reproductive Disorder Examples	
	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

Table 4.

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

Table 5.

Sr	Endocrinal Disorders	
1	Ati Hrasva(Dwarfism)	Ati Deergha (Giantism)
2	*Ati Sthula* (HereditaryObesity)	*Ati Krusha* (Hereditary leanness)
3	*Ati Gaura* (Hereditary Albinism)	*Ati Krushna* (Hereditary Hypermelanosis)
4	*Ati Loma* (Hereditary Hypertrichosis)	*A Loma* (Hereditary Hypotrichosis)

Diagnosis of Hereditary Diseases

1. Aminocentesis
2. USG
3. Family history
4. Chromosomal study
5. DNA study

Discussion

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). 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.

Conclusion

1. The concept of genetics and genetic disorders can very well be traced in ancient Indian medical science that is Ayurveda.
2. The Ayurvedic classification of genetic disorders include

X linked (Matruja) and Y linked (Pitruja) genetic diseases along with the multifactorial inheritance of diseases.

3. 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.
4. The Ayurvedic mentioned disease Klibam (Hereditary erectile dysfunction) can classified under Y-linked genetic disorder.
5. 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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