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CONCEPT OF AMBIGUOUS GENITALIA AND DISORDERS OF GONADAL DIFFERENTIATION IN AYURVEDA

Kumar Vijendra*[1] and Gupta Shilpy[2]

[1]Assistant Professor, Department of Prasuti Tantra Evum Stree Roga, Uttaranchal Ayurvedic College, Rajpur, Dehradun, Uttarakhand, 248009 (India).

[2]Assistant Professor, Department of Kaumarbhritya/Balroga, Uttaranchal Ayurvedic College, Rajpur, Dehradun, Uttarakhand, 248009 (India).

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***Corresponding Author**
Dr. Kumar Vijendra
Assistant Professor, Department of Prasuti Tantra Evum Stree Roga, Uttaranchal Ayurvedic College, Rajpur, Dehradun, Uttarakhand, 248009 (India).

ABSTRACT

Description of ambiguous genitalia has been laid down very first time in *Ayurveda*. The abnormalities concerned with ambiguity of sex includes *dwireta* (hermaphrodite), *pavanendriya* (azoospermic), *samskarvahi* (anaphrodisiac), *vakri* (hypospadiac) *narshand* (sterile male), *narishand* (sterile female), *irshand* (voyeurism), *vatikshand* (anorchidism). Disorder of sexual differentiation (DSD) is being used now a day for ambiguous genitalia synonymously. Many researches are going on day by day to explore the facts. Modern medical literature has explained the genetic and endocrinal basis of sex determination and its abnormality. The Genotype of an individual is determined by the chromosomes while the phenotype is dependent on genotype and its environmental influence. Ayurveda deals about two types of *beeja* (gametes); *shukra* (male gametes) and *artava* (female gametes). Depending upon their predominance during fertilization they are responsible for determination of sex in *garbha* (fetus). Further abnormalities in sex determination have been explained in terms of vitiation of *beeja* and *beejabhaga* (genotype). Not only the genetic makeup (genotype) but also environmental factors like *ashaya dosha* (abnormalities of reproductive organ), *kal dosha* (improper timing), *atmakarma dosha* (idiopathic or unexplained factors), and *matur ahar vihar dosha* (maternal dietetics and regimens) are equally responsible for expression of anomalies in the fetus i.e., phenotype.

KEYWORDS: *Beeja*, *Beejabhaga*, Ambiguous Genitalia, Gonadal Differentiation, DSD.

INTRODUCTION

Ayurvedic literatures contain almost all dimensions of medical diversity including eight major branches.[1] Apart from these major divisions, subject matters of super specialty of present era has also been described briefly e.g., embryology, genetics, plastic surgery etc. *Sharira* (anatomy and physiology) not only explain the role of *shukra* (male gametes), *artava* (female gamete) with other components contributing in procreation of organism but also their qualitative and quantitative significance in reproductive biology.[2] Sex determination in fetus is dependent on role of male gamete or female gamete quantitatively; that is dominance of male gamete results male sex whereas that of female gamete in to female sex. As per concept of genetics there is restoration of diploid set of chromosomes during fertilization which is contributed as haploid set by both parents. XY chromosome determines male sex while XX chromosome determines female sex.[3] Description of ambiguity of sex has been first time mentioned by *Charaka.* There are eight types of disorder of sexual differentiation; these are *Dwireta, Pavanendriya, Samskarvahi, Vakri, Narshand, Narishand, Irshand and Vatikshand.*[4]

In *dwireta* (hermaphrodite) the sperm and ovum of parents responsible for the formation of the germ cells of the fetus is vitiated qualitatively and their proportion becomes equal quantitatively then the offspring becomes a *dwireta*.[5] Such an offspring will have the characteristic features of both the sexes. Due to affliction with *vata*, reservoirs of sperms i.e., testicles of the fetus becomes aspermic and offspring is called *pavanendriya* (azoospermic). Under influence of *vata* there is obstruction of the *shukravaha srotasa* (ejaculatory ducts) resulting in to *samskarvahi* (conditioned anaphrodisiac) offspring.[6] In case of *narshand* (sterile male), they have testicles with sperm reserve but these are vitiated. Similarly, *narishand* (sterile female) have ovaries with ova reserve but these are vitiated. The congenital lack of strength and passion result in impairment and insufficiency of sperm and ovum which in turn give rise to male and female sterility respectively. During cohabitation, irregular or abnormal posture of the female partner and weakness in sperms of the male partner results in *vakri* (hypospadiac) type of offspring. Reduced passion along with jealous parental cohabitation produces mixoscopia in the offspring and the condition is termed as *irshand* (voyeurism). Destruction of fetal testicles by vitiated *vayu* (*vata*) and *agni* (*pitta*), results in to *vatikshand* (anorchidism) type of offspring.[7]

In another context *Charaka* has also described about some of factors which lead to deformity and malformation of male and female fetuses. These disorders actually arise due to abnormal change occurring at the level of gene due to which three types of abnormalities occur in female and male offspring. *Vandhya, Putipraja* and *Varta* in female whereas *Vandhya, Putipraja* and *Trinaputrika* in male.[8]

In Ayurveda there is description of *beeja, beejabhaga* and *beejabhagavayava* which probably resemble pronucleus of gametes, nuclear material (chromosome) and gene (specific length of DNA) responsible for organogenesis respectively. In terms of vitiation of *doshas* which can affect either *beeja*, or *beejabhaga* or *beejabhagavayava* results in to the disorder of specific organ or its constituent part either structural or functional. Similarly the reproductive organ formed as a result of abnormal *beeja*, results into *vandhya* (infertile) offspring, *beejabhaga* into *putipraja* (neonate likely to die very soon or born with congenital anomalies resulting in to degenerated body parts) and *beejabhagavayava* into *varta* (individual with incomplete feminine features) in female and *trinaputrika* (individual with incomplete masculine features) in male progeny.[9]

DISCUSSION

The ancient concept of ambiguous genitalia is not a hypothetical context but it is a very scientific and challenging issue even in the present era. Detection of genital defects in neonate requires tremendous evaluation by experts before assignment of sex. To diagnose sex of such a neonate it requires clinical methods, as well as laboratory tools and techniques e.g. hormonal, molecular, genetic, and radiographic investigations. Now a days the Quigley's scale is used to classify degree of ambiguity of genitalia.[10] The eight types of ambiguity of genitalia as per *Charaka* can be studied in the following way.

Dwireta resembles hermaphroditism, which may be either true or pseudo. Individual having both ovaries and testes is true hermaphrodite whereas pseudo hermaphrodites are the individual with either ovarian or testicular tissues along with ambiguous sexual anatomy. Another form is called as ovotestes who may have either ovary on one side and testes on another side (lateral type) or ovotestes on one side and normal gonad on another side (unilateral type) or having two ovotestes (bilateral type). The *Dwireta* (hermaphroditism) has close resemblance with Stage IV of Quigley's scale. They are the individuals with ambiguous phenotype and severely limited masculinization evidenced by a phallic structure that is

intermediate between a clitoris and a penis, generally accompanied by a urogenital sinus with perineal orifice and labioscrotal folds.

In the same scale stage-I have close resemblance with *Pavanendriya*. These individual suffers from azoospermia, they have normal male external genitalia but reduced virilization at puberty and become infertile due to hormonal insufficiency i.e. minimal androgen resistance. *Vakri* are independently male phenotypically, but have mild defect in fetal masculinization, with clinical features of hypospadias and, or micropenis and they have been categorized under stage-II of Quigley's scale. *Vatikshand* can be included under stage-III and they are predominantly male but with severely defect in fetal masculinization and clinically manifested by perineal hypospadias, micro penis, cryptorchidism and, or bifid scrotum. *Narishand* have essentially female phenotype due to minimal fetal androgen action with separate urethra and vaginal introitus. Minimal androgenization results in to mild clitoromegaly or a small form of posterior labial fusion which is stage-V of Quigley's scale. *Narshand*, resembles individuals who have no fetal androgen action in intra uterine life and are likely to develop androgen dependent pubic and or axillary hair at puberty as per stage-VI of Quigley's scale. *Samskarvahi* individuals are male genotypically without ambiguity of genitalia and suffer from ejaculatory dysfunction of congenital or acquired origin. *Irshand* are individuals who suffer from psychosexual disorder called voyeurism, a variant of paraphilia such individuals get sexual gratification by observing sexual activities of others. *Varta* resembles Turner's female with XO genotype and they are individual with incomplete feminine features. *Trinaputrika* may have resemblance with Klinfilter male with XXY genotype and they are individual with incomplete masculine features. *Putipraja* (still born with degenerated body parts or congenitally malformed baby) may occur due to salt wasting nephropathy and death due to vascular collapse.

CONCLUSION

Diagnosis of ambiguous genitalia plays a vital role as a key towards its optimal management. A multidisciplinary team of pediatric endocrinologists, psychiatrists, obstetricians, radiologists, surgeons, geneticists and biologists working in concert is essential in such a condition. Similarly several contexts have been described briefly or elaborately in Ayurvedic literatures which can be the source for worthy researches in the present era.

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