Disorders of Sex Development: The Quintessence of Perennial Controversies-III.
DSD, Transgenders & The Judgement by the Hon'ble Supreme Court of India

Transgender issues in children and adolescents

Disorders of Sex Development (DSD) (Mentioned under the Umbrella term - 'Eunuchs' by the Hon'ble Supreme Court of India) have been previously known as Intersex disorders.

Problems in these children often remain unrecognized and if untreated they grow up as adults. During this period they face discrimination. This is the most valuable period of their formative years and puts a deleterious effect on their psychological development.

They are at the risk of getting disillusioned by the environment. Often these children are found in the various subgroups identified by the Hon'ble Supreme Court under the umbrella term TRANSGENDERS as 'Eunuchs'.

These children, born with ambiguous genitalia, have their gonads (testes & ovaries) located at abnormal positions and at risk of damage due to high temperatures losing fertility potential and risk of cancer. Their passage of passing urine (urethral meatus) is abnormally located and leads to infections of kidneys and kidney damage. Some of these children have their uterus in a position where there is blockage of outflow of secretions, leading to various irreversible consequences for their reproductive health.

Infants born with an intersex condition are unable to make an informed decision to consent regarding their treatment, thus leaving the issue to be dealt with by parents in the best interests of the child. Some advocates believe such a decision should be made by the courts as parents should not have to make a potentially irreversible decision.

In the era of assisted reproductive techniques and surrogacy, every attempt should be made to make them cognizant of the technological advances in assisted reproductive techniques through educative awareness programme.

- Educative awareness to them about fertility and endocrine issues
- Sperm storage & Fertility issues are give paramount importance while deciding sex of rearing in DSD
- Same principles should be applied while developing curriculum for educative awareness

There is an urgent need to look into these issues.

(i) Appropriate models for the treatment and care of children born with Disorders of Sex Development (DSD), and
(ii) An appropriate framework for decision-making and consent in relation to the treatment and care of children born with DSD.
Due to wide variability in presentation, DSDs (Disorders of Sex Development) are sometimes identified by etiology and sometimes by phenotype. As a result, overlap in describing some conditions, such as, 46 ovotesticular DSD and mosaicism as in 45, XO/XY is inevitable. All DSDs have the potential to cause psychosocial distress for patients and their families, as often appearance of genitals is in variance from the normal.

Table 1. Clinical Management Guidelines for day to day management of DSD based upon broad consensus

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<thead>
<tr>
<th>Condition</th>
<th>Type of surgery relevant to condition</th>
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<tbody>
<tr>
<td>Genetic females with 46,XX DSD [Congenital Adrenal Hyperplasia (CAH)]</td>
<td>• In cases of CAH, endocrine management is necessary for maintenance of health and fertility and to prevent premature puberty.</td>
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<tr>
<td>• CAH is potentially life-threatening. Until it has been ruled out, prompt diagnosis and treatment should be considered in all children with genital ambiguity.</td>
<td>• Menses require drainage opening (separate from urinary system) to avoid pain and infection. Internalized vagina may be a source of urine pooling and infection if left uncorrected.</td>
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<td>17-beta reductase deficiency (XX or XY) - Appearance female but can’t make estrogen or testosterone; consequently no pubertal changes.</td>
<td>• Vaginoplasty and in some cases, clitoral reduction is neccessay.</td>
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<td>• Need to monitor undescended testes for malignancies.</td>
<td>• Risk of adrenal insufficiency.</td>
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46, XY DSD:

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<td>• Partial Androgen Insensitivity Syndrome (PAIS) - Initial gender assignment should take into account that higher degrees of prenatal androgen exposure may direct the brain to develop in with male inclination. In patients who are raised as girls, the testes will cause some pubertal virilization;</td>
<td>• Repair of hypospadias (child being raised male), testicular biopsy at puberty, removal of intra-abdominal gonad. Intra-areolar mammary reduction surgery around puberty</td>
</tr>
<tr>
<td>• Complete Androgen Insensitivity Syndrome (CAIS)-Infertile with current technologies. Raised as girls. Undescended testicular tissue presents increased risk of malignancy after puberty; counsel patient to consider orchidectomy following puberty. (Delay until puberty allows patient to experience a natural feminizing puberty and to choose orchidectomy.) Vagina lengthening may be request by the patiens as she grow. Secondarily surgical intervention need for that.</td>
<td>• Complete Androgen Insensitivity Syndrome (CAIS) - Raised as girls. Undescended testicular tissue presents increased risk of malignancy after puberty; counsel patient to consider orchidectomy following puberty. (Delay until puberty allows patient to experience a natural feminizing puberty and to choose orchidectomy.) Vagina lengthening may be request by the patiens as she grow. Secondarily surgical intervention need for that.</td>
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<td>• 5-alpha reductase (5-AR) deficiency—there is varied clinical presentation and sex of rearing.</td>
<td>• 5-alpha reductase (5-AR) deficiency—If patient raised as girl, decisions need to be made before puberty about management of masculinizing puberty (e.g., patient may elect orchidectomy). If raised as boy then masculinizing genitoplasty is carried out.</td>
</tr>
<tr>
<td>• 46,XY 3-beta-hydroxysteroid dehydrogenase (HSD) deficiency—Usually lethal; risk of severe adrenal deficiency.</td>
<td>• Endocrine management necessary for maintenance of health and fertility.</td>
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</table>
- Gonadal dysgenesis (partial and complete)—Dysgenetic gonads present substantially elevated risk for malignancies. Infertile. Karyotype varies

- Ovotesticular DSD—ovary and testis, and/or ovotestes (historically called true hermaphroditism)—Testicular tissue presents an increased risk for malignancies.

- Swyer Syndrome (another name for 46,XY gonadal dysgenesis)

- 45,X (Turner Syndrome) - High phenotypic variability. Genitalia typically unambiguously female.

- Sex-chromosome mosaicism (e.g., 45,X/46,XY)—Genotypes and phenotypes vary; may appear ambiguous or may appear unambiguously male or female.

- Generally raised male unless there is no penis.

- Repair of hypospadias, removal of intra-abdominal testis, biopsy of scrotal testis at puberty (child being raised male).

- When reared as females: removal of testicular tissue & phallic reduction procedure.

- Appropriate counseling is provided to assist decision making.

If raised as male:

- Repair of hypospadias, removal of intra-abdominal testis, biopsy of scrotal testis at puberty.

- When reared as females: removal of testicular tissue & phallic reduction procedure.

- Counseling and support from learning disability specialists.

- Appropriate counseling is provided to assist decision making.

If raised as male:

- Repair of hypospadias, removal of intra-abdominal testis, biopsy of scrotal testis at puberty.

- When reared as females: removal of testicular tissue & phallic reduction procedure.

- Monitor for gonadal malignancies.

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**Where external genitalia are not ambiguous and the condition is only detectable when other clinical features are present**

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<td>Complete Androgen Insensitivity Syndrome</td>
<td>Nature of surgery depends upon the cognitive development of the child at the time of presentation. All opportunities would be given to keep the best interest of the child. Parents would be explained and appropriate procedure chosen which would vary <em>from case to case.</em></td>
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<tr>
<td></td>
<td>As androgens have been present around the perinatal life of the child, masculinizing genitoplasty or PHALLOPLASTY may be desired by the child and family and the same would be offered.³</td>
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Removal of intra-abdominal testes that cannot be brought down.

In case the child is being reared as a female:

- Removal of testes is generally recommended after puberty has been completed, as the hormonal metabolism helps in body habitus. Child & parents would be informed about the pros & cons of this approach. The final decision would be only after consulting them.
- Vaginal lengthening procedure would be offered as & when desired by the child when grown up.

- Raise in concordance with chromosomal sex.
- Corrective, masculinizing genitoplasty to be carried out. Reproductive technologies may be used to enhance fertility. Some learning disabilities are associated with 47,XXY; Counseling and support from learning disability specialists is recommended.
- Mullerian structures to be carefully removed without injuring vas deferens.

**Physical anomalies in the external genitalia but these are not regarded as ambiguous genitalia**

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<td>46,XY cloacal extrophy-Complex disorder with variable presentation; long-term survival approximately 70%. In the past many of these children were raised as girls; a notable number of these children have transitioned to boys</td>
<td>Often, multi-system reconstructive surgical procedures are required by experts in tertiary care centres. Removal of healthy testes should not be performed without patient's consent as it eliminates fertility.</td>
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<td>Bladder Exstrophy</td>
<td>Reconstructive surgical procedures are required by experts in tertiary care centres</td>
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<td>46,XY micropenis-These children do well as boys. If assigned as girls, there could be gender dysphoria later in life. Causes are variable; one cause of 46,XY micropenis is congenital pan-hypopituitarism, which in males is commonly associated with potentially lethal hypoglycemia, the result of growth hormone and ACTH deficiencies.</td>
<td>Unless the pan-hypopituitarism is corrected, this hypoglycemia is typically unresponsive to most standard interventions.</td>
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<tr>
<td>Aphallia-these children have normal testes and masculine inclination as they grow. Perinatal exposure to androgens have already influenced the brain.</td>
<td>They may be fertile and that potential should be preserved. As their urinary bladder opens directly into the rectum, this communication should be disconnected and urethroplasty to be carried out, preferably before school going.</td>
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- Kallman Syndrome-Males potentially fertile. May have anosmia (absence of sense of smell).
- 47,XXY (Klinefelter Syndrome)-Genitalia typically unambiguously male, although testes may be small. Gynecomastia common at puberty. Likelihood of azoospermia.
- Persistent Mullerian Duct Syndrome-Risk of cryptorchidism and associated complications. Increased risk of infertility.
- Hypospadias—Hypospadias in conjunction with cryptorchidism increases the likelihood of underlying DSD.
- Needs full clinical, genetic and endocrine workup as for DSD.
- Kallman Syndrome—Males potentially fertile. May have anosmia (absence of sense of smell).
- Raise in concordance with chromosomal sex.
- Mayer, Rokitansky, Kuster, Hauser Syndrome (also known as MRKH, Mullerian agenesis, vaginal agenesis)—Ovaries present with uterus absent, or small; associated with kidney and spine anomalies in a minority of patients.
- Patient may elect pressure dilation or secondarily surgical augmentation if she seeks increased vaginal length. Sometime, there is vaginal agenesis. All efforts should be made to provide vaginal replacement and access to uterine cavity for fertility by an appropriate surgical technique. 
- Progestin-induced virilization—History of virilizing hormone exposure is limited to prenatal life.
- As virilization will not progress, patient should be closely monitored.

Indian mythology has many references to altered sexual states. The name Ardhanarishwara refers to God, who is half man and half woman, an androgynous deity. In various versions of Ramayana, there is reference to King Ila, who spent half his life as man and half as woman. In Mahabharata, Arjuna, one of the fiercest warriors of his time, spent a year of his life in intersexed condition. There is also reference to King Bangasvana, who was changed into a woman by Lord Indra, whom he had offended. Another reference during Mahabharata is to Shikhandini. He was born female, but raised like a man and trained in warfare. After an encounter with a Yaksha, Shikhandini came back as a man, was called Shikhandi and fathered children.

Transgender communities have existed in most parts of the world with their own local identities, customs and rituals. They are called hijras, jogappas, jotgas, shiv-shaktis and aravanis in India. The hijra community in India, which has a recorded history of more than 4,000 years, was considered to have special powers because of its third-gender status. It was part of a well-established ‘eunuch culture’ in many societies.

**Recommendation by the Hon'ble Supreme Court of India**


1. Hijras, Eunuchs, apart from binary gender, be treated as "third gender" for the purpose of safeguarding their rights under Part III of our Constitution and the laws made by the Parliament and the State Legislature.

2. Transgender persons' right to decide their self-identified gender is also upheld and the Centre and State Governments are directed to grant legal recognition of their gender identity such as male, female or as third gender.

3. We direct the Centre and the State Governments to take steps to treat them as socially and educationally backward classes of citizens and extend all kinds of reservation in cases of admission in educational institutions and for public appointments.

4. Centre and State Governments are directed to operate separate HIV Sero-surveillance Centres since Hijras/Transgenders face several sexual health issues.

5. Centre and State Governments should seriously address the problems being faced by Hijras/Transgenders such as fear, shame, gender dysphoria, social pressure, depression, suicidal tendencies, social stigma, etc. and any insistence for SRS for declaring one’s gender is immoral and illegal.

6. Centre and State Governments should take proper measures to provide medical care to TGs in the hospitals and also provide them separate.
Importance of the Report to Handling Transgenders Issues in India.

1. Diagnostic Criteria for Gender Identity Disorder as mentioned in the document should be followed by Hospitals in India.

2. Criteria for assessment and treatment of children, adolescent and adults with gender dysphoria/gender incongruence mentioned in the documents should be met and followed.

3. Criteria for undertaking Gender Affirmation Surgery (GAS) or Sex Reassignment Surgery (SRS) mentioned in the documents should be met and followed Before undertaking a GAS/SRS.

4. Criteria for hormone therapy mentioned in the documents should be followed.

5. Criteria for psychosocial issues in TRANSGENDER health in Children, Adolescents & Adults with Gender Dysphoria/ Gender incongruence mentioned in the document should be followed.

6. Expert Working Groups should be constituted which will provide advice on:
   a). Current clinical practices & areas needing immediate attention

References

1. Intersex Society of North America. 2006

New Delhi
May 1, 2013

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