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Narrative Review

Genetic counseling in atypical genitalia: Challenges and proposed communication approach

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ABSTRACT

Atypical genitalia (AG) is a rare condition in which a child's external genitals are not properly distinguished. The genitals can be seen as incompletely developed or have both male and female internal and/or external genital structures. Identification of genetic causes in such children is important for appropriate genetic counseling. Genetic counseling in AG is very crucial and sensitive. Also, there are lots of challenges involved in offering and communicating the information to parents or individuals. The article highlights the challenges in counseling and proposes a simple communication approach in AG.

Keywords: Atypical genitalia, Genetic counseling, Genetic testing, Genetic counselor, Disorder of sex development

INTRODUCTION

Atypical Genitalia (AG) and Differences of Sex Development (DSD) are conditions that are congenital and may be secondary to abnormalities of chromosomes or genes, anatomical, gonadal, and hormonal profiles and attributes.^[1] Atypical genitalia is characterized as a birth defect wherein a child's external genitals are not congruent with their internal genital organs and/or the genetic sex. Differences of sex development often manifest with atypical genitalia. There are various types of DSDs, and the causes may vary from case to case.^[2] It can range from very subtle physical alterations to severe outcomes, which can have long-term consequences. The prevalence of AG is 1 in 1000 to 4500 live births. Hypospadias is the presentation in the majority of the cases. Chromosomal variants are one of the major contributory factors for AG. In neonatal cases and in genetic practice, AG is a case of medical and social emergency as some forms of DSDs, like congenital adrenal hyperplasia and certain malformation syndromes, can be potentially life threatening. In the past, identification of genetic etiology by cytogenetic techniques was limited to sex chromosomal variations. However, the availability of advanced genetic testing has drastically changed the understanding of genetic etiology at molecular level, as well as enhanced the genetic counseling aspect.^[3] Genetic counseling for the parents and conveying the test results is very challenging considering the complexity and sensitivity of the information to be shared. The stigma, confusion, anxiety, and shame associated with

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the condition are the major challenges while counseling the individual and parents. In the last decade, changes have been seen in the acceptability of the condition among parents and society with the availability of different treatment modalities. Improved genetic testing services, increased attention toward patients, understanding AG in a broader aspect, education of parents, and the formation of parent support groups have significantly improved care of such children. The article highlights the importance of genetic counseling, challenges in genetic counseling, and proposes a simple but effective communication approach in AG.

CHALLENGES IN COUNSELING

The limited number and availability of medical geneticists and genetic counselors is a major challenge in developing countries like India. The majority of the time individuals are referred to a medical geneticist or genetic counselor after the test is done by a medical professional. Usually, the test reports are disseminated by clinicians who are not trained enough to interpret the genetic reports. Although acceptance by parents and patients has increased in recent years, effective disclosing about the complex medical condition has not been significantly resolved. Stigma associated with sexual ambiguity and differences in sexual development leads to psychological, social, emotional, and cultural concerns. In developing countries like India, an effective drive for sex education is not in place, and hence there is limited understanding of sexual development in individuals and parents. Additionally, there is an acute need to create awareness and educate the medical professionals regarding the availability of genetic tests and the interpretation of test results. In a country like India, the sex of the fetus is not revealed to the parents during the antenatal period. Post-delivery; the first question asked is the sex of the child whether the newborn is a “boy” or a “girl”. This causes psychological trauma to the parents and counseling such a couple is very challenging as diagnosing the underlying etiology takes time. Time taken for chromosomal karyotype report is also one of the concerns as it takes around weeks’ time. As it is the gold standard test even in the era of genomics, which gives accurate genotypic information in this condition. Revealing accurate diagnostic information to the individual or parents needs a perfect balance of communicating the complex condition and simultaneously maintaining a positive sense of self. Educating them about genetic evaluation and confirmation of the cause will help them in the management of the child, and to make them understand the importance of preventing the same in the next pregnancy is also at times challenging.^[4]

COMMUNICATION APPROACH FOR THE GENETIC COUNSELORS

Effective communication with the individual or parents is very important, as it’s a very complex process and lots of concerns are involved in the process.^[5] Based on our experience of counseling individuals born with DSD, the following approach can be followed.

Building confidence

The first thing to do in a counseling session is to build their confidence so that they can share their concerns openly. This can be done by letting them know that they are not the only parents or individuals born with DSD, and doctors see such cases regularly. This will help to open the discussion.

Understanding the concerns

It is very important to know the understanding of the individual or parents about the condition and their biggest concerns, and for this, sufficient time needs to be given by the counselor. Minute and seemingly unrelated queries, which generally stem from social beliefs, customs, etc., need to be addressed. This discussion will help the team of physicians understand the overall situation of the family and the challenges they are experiencing. Additionally, the team can unearth other medical problems running in the family, including those having potential risks for future offsprings developing DSD.

Educating them about the condition

To bring a sense of comfort, one should educate them about the disease condition and the process of sexual development. This can be best achieved by showing pictures, videos, or atlases. At the same time, it is necessary to educate the family regarding the risk to the individual and in the next pregnancy. It is also important to update them about the newer technologies and treatment modalities available. Drawing pictures can help for better understanding and should be practiced. The individual or parents may get confused in a single meeting, and hence, ideally, counseling should be done in more than one session. These follow-up sessions help them understand the condition in a better way, and important aspects are not overlooked. The preferences of the parents need to be taken into consideration, and counseling should be done accordingly.

Issue counseling letter

The DSD and associated genetic findings are very complex conditions and difficult to understand. It has been observed that the awareness about genetic conditions and the interpretation of genetic test reports is often a difficult task for clinicians. So, as a routine practice, we avoid the technical

terms and counsel the patients in simple language, using their own words. At the end of the counseling session, we summarize the whole discussion and ask the patients to repeat the discussion in their own words to check whether they have understood and interpreted it properly. A pamphlet or an information booklet with brief counseling details can be very helpful to them. This will help the patient forever, especially those who don't have access to trained genetic counselors or medical geneticists.

Information about support groups

Once the disease condition is confirmed, it is always beneficial to connect the family with the established support groups. This will also give a chance to interact with the parents of children with similar conditions. Nowadays, various established support groups conduct informative programs to improve knowledge and understanding among the patient or parents.

CONCLUSION

DSD is a complex congenital condition. Laboratory genetic diagnosis is very important for counseling parents or patient. DSD is a very sensitive condition associated with psychological, social, and emotional components. The communication process in individuals born with DSD is very crucial and should be followed with caution. Since genetic reports are complex and not applicable to the patient, only genetic counseling should be carried out by a trained genetic counselor or a medical geneticist. All clinics for individuals born with DSD should have genetic counselors and geneticists who are adept at explaining the clinical condition of the patient.

Author contributions

SP did the conceptualization. SP did patient recruitment. SP did genetic counseling, and SB arranged the sessions. HG did processing of the sample. HG and SP did the karyotyping. Data analysis was done by SP, SB, and HG. Editing and writing of the manuscript was done by SP and SB. SB did the documentation. SP, SB, and HG reviewed the manuscript. All authors have read and agreed to the published version of the manuscript.

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Data availability

The related data are made available in the manuscript.

Ethical approval

Institutional Review Board approval is not required.

Declaration of patient consent

Patient's consent not required as there are no patients in this study.

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Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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