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Psychological Aspects of Androgen Insensitivity Syndrome- A Case Report

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Abstract

Introduction: Androgen insensitivity syndrome (AIS) is a condition that results in the partial or complete inability of the cell to respond to androgens. A person with androgen insensitivity syndrome can experience significant psychological distress secondary to the ambiguity concerning their sexual anatomy. Psychological evaluation and management of a case of Androgen insensitivity syndrome brought up as a female experiencing such distress is reported here.

Case history: A 25 years aged female patient working as a police constable, was referred for psychiatric evaluation for repeated attempts to commit suicide secondary to depression and anxiety arising out of her sexual ambiguity. She had consulted a gynaecologist for not having attained menarche. Ultrasonography of abdomen showed aplastic uterus with streak ovaries. According to chromosome analysis, she was a phenotypic female with male karyotype-46, XY (sex reversal). Her testosterone levels were increased. She had undergone prophylactic laparoscopic gonadectomy. The impact of test results made her anxious and sad. Her sexual ambiguity resulted in worry about getting transformed into a hijda (Eunuch) which led to repeated attempts to commit suicide. Psychiatric intervention consisted of antidepressants and counselling sessions aimed at coming to terms with ambiguity about her gender.

Conclusion: Androgen insensitivity syndrome, although very rare, can result in considerable psychiatric morbidity. Apart from gynaecological and endocrinal aspects, addressing associated psychological issues is essential to reduce psychiatric morbidity and increase quality of life.

Keywords: Androgen insensitivity syndrome; Depression; Psychiatric morbidity; Psychosocial interventions; Sex assignment

Introduction

Androgen insensitivity syndrome (AIS) is partial or complete inability of the cell to respond to androgens [1,2]. CAIS occurs in 1 out of every 10,000 to 1,30,000 depending on the criteria of severity used [3,4]. Due to its subtle presentation, AIS is divided into three categories that are differentiated by the degree of genital masculinisation. In complete androgen insensitivity syndrome (CAIS) , the external genitalia are that of a normal female, in mild androgen insensitivity syndrome (MAIS) ,the external genitalia are that of a normal male and in partial androgen insensitivity syndrome (PAIS), the external genitalia are partially, but not fully, masculinized [5,6]. The unresponsiveness of the cell to the presence of androgenic hormones can impair or prevent the masculinization of male genitalia in the developing foetus, as well as the development of male secondary sexual characteristics at puberty. But it does not significantly impair female genital or sexual development. As such, the insensitivity to androgens is clinically significant only when it occurs in genetic males (i.e. individuals with a Y-chromosome, or more specifically, an SRY gene) [7]. Persons with androgen insensitivity syndrome can experience significant psychological distress secondary to the ambiguity concerning their sexual anatomy. A case of complete androgen insensitivity syndrome brought up as a female experiencing such distress is presented here. Psychological evaluation and management of such a case is discussed.

Case History

The case is reported as per the sequence of events which unfolded from the time of first consultation till the last follow up. A twenty five year female police constable was referred for psychiatric evaluation for attempting suicide repeatedly. She was worried about having not attained menarche, poor development of breasts, and loss of hair over scalp and persistent thoughts and experiences of getting transformed into a hijda. She was born out of full term normal delivery. She was brought up as female in the family with respect to dress and hair style, as her external genitalia appeared to be that of a female. She used to urinate in squatting position like a female. She used to socialise more with girls and indulged in feminine games. As a child she had normal physical growth and development along with normal intellectual development. During this period she felt that she was a female.

She started masturbating at the age of fifteen years by stimulating her external genitalia with fingers. The stimuli for and the fantasy accompanying masturbatory activity was heterosexual couple or males. She had an encounter with a male cousin and felt pleasure by his stimulation with fingers. She also reported erotic attraction towards male colleague around the age of twenty.

At the age of 18, she consulted a gynaecologist for not having attained menarche. Gynaecological examination revealed labial folds with less of subcutaneous fat, hypo plastic clitoris, scanty pubic hair and poorly developed breasts and buttocks. Ultrasonography of abdomen showed aplastic uterus with streak ovaries (wrongly identified as ovaries which was later found to be testes). They were told that she cannot menstruate in future. At the age of 23 they again approached another gynaecologist seeking help for not attaining menarche.

Endocrinological assessments revealed normal levels of oestradiol as that for males, luteinising hormone, follicle stimulating hormone and thyroid hormones. Testosterone levels were markedly increased (741.9ng/dl). Sample for karyotyping was sent.

She was told that she cannot menstruate and bear children as her uterus is absent. Then she started feeling sad because of her inability to fulfil the personal and societal roles of a woman. Because of intense sadness and anxiety she started making repeated attempts of suicide in a span of about six months. This led to her referral to a psychiatrist. On physical examination, her height was 168 cm, weight was 59kg and BMI was twenty. She had wheatish complexion with proportionate facial features. Other findings were similar to those of earlier gynaecological examination.

On psychological evaluation, she reported intense feelings of sadness and anxiety. After seeing a hijda, she started getting anxious that she also might turn into a hijda. Hijda according to her was a person who was anatomically neither a male nor a female. She started to worry and experienced that her voice and body hair were changing like that of a hijda. They appeared like fleeting incomplete delusions which raised the possibility of a psychotic disorder. She reported erotic feelings towards another male colleague for past 2-3 months and wished to have a complete marital life in a role of woman with him. Awareness of her inability to do so contributed to her distress. She had sleep and appetite disturbances, crying spells, difficulty in experiencing pleasure in previously pleasurable activities and repeated ideas about committing suicide. She started abstaining from work. These features met the criteria for major depressive episode. She did not go for work for almost two months during this depressive episode. Her mood disturbances were treated with antidepressant, Tab. Escitalopram 10mg and Tab. Clonazepam 0.5mg which was titrated according to her clinical response.

Availability of chromosome analysis report which showed that she was a phenotypic female with male karyotype-46, XY (sex reversal), led to increase in her depression. She underwent prophylactic laparoscopic gonadectomy. Histopathological examination showed that they were rudimentary testes.

During counselling sessions she was explained that actually she is genetically a male, but since her body is not responding to male hormones she has not developed male characteristics. She doesn't have complete female physical characteristics as she is not a female. She has female characteristics only externally but not internally. But she has been brought up as a female and hence psychologically she is a female. She was allowed to ventilate her feelings in the sessions. She was helped to come to terms with the confusion existing between her genetic and physical sex and her psychological sexual orientation. Her concerns about further anatomical changes in her body were allayed. She expressed her sexual desires and interest in marriage. She was told that she can have sexual intercourse but she cannot bear children.

With above intervention, her depression improved, fleeting delusions disappeared and she felt better. During follow up she was contemplating marriage with a male with whom she is in a nonphysical relationship at present.

Discussion

Sex determination and differentiation depend on a cascade of events that begin with the establishment of chromosomal sex at fertilization and end with sexual maturation at puberty, subsequently leading to fertility. The case reported here is consistent with complete androgen insensitivity syndrome. As a child, she was brought up as a female. This is the standard practice as per literature [8]. However, some are brought up as males. In them sex re assignment surgeries and hormonal substitution therapy have been tried [9].

Our patient was tall, had proportionate facial features and scanty facial and pubic hair. She felt her libido as adequate and had capacity to experience orgasm. She perceived herself as a female and reported erotic attractions towards male. These findings are consistent with existing literature [10].

In our patient the realisation that she is actually genetically male but not having any male physical characteristics and having only partial female characteristics resulted in significant psychiatric morbidity in the form of a major depressive episode. Depression to the point of breakdown, shock and denial has been observed in other studies [11]. This patient had specific persisting thoughts and experiences that she might undergo anatomical transformation into a Hijda (Eunuch). This was based on her understanding of her sexual anatomy and information she had about hijdas, before she underwent counselling. She started to worry and experienced that her voice and body hair were changing like that of a hijda. They appeared like fleeting incomplete delusions which raised the possibility of a psychotic disorder. Phenomenologically they appeared like incomplete fleeting delusions as she believed that there were anatomical changes in form of her changed voice and also she felt that her body hair might grow like that of a hijda. Thus a diagnosis of psychosis was considered when results of karyotyping were unavailable. But surprisingly they disappeared with antidepressants, anxiolytics and counselling after results of karyotyping were available. There is lack of literature about these issues.

Psychological distress is more common in adults with partial androgen insensitivity syndrome than in those with complete androgen insensitivity syndrome, irrespective of whether they were brought up as male or female [12]. But a controlled study comparing psychopathology between CAIS and controls reported no statistically significant differences in psychological outcomes between two groups. Intensity of psychiatric morbidity can be related to acceptance by the society which in turn is determined by rigidity with which gender roles are assigned and differentiated. Severe psychiatric morbidity in this patient could be secondary to the rigid views prevailing in our society currently about these issues. The type of conflicts faced by a person having CAIS depends upon the time period when the condition is diagnosed. Post marital diagnosis is often reported in literature which results in distress about being infertile [13]. But this patient differed from such patients as she was diagnosed pre martially as advised in literature [14]. But she faced a different conflict which was about finding a suitable match for marriage.

This patient achieved satisfactory level of adjustment to assignment of feminine role after psycho education and counselling. But some

doubt persisted about fulfilling all feminine roles as she was told that she was genetically a male and didn't have child bearing capacities. Many investigators have reported satisfactory adjustment to assignment of feminine roles in persons having CAIS [15], but some have observed that a significant number of women have expressed reservations about such an assignment [16].

Conclusion

Androgen insensitivity syndrome, although very rare, can be extremely distressing to the concerned individual. Psychiatric morbidity in them can have cultural connotations. Apart from physical treatments like gonadectomy, surgical correction and oestrogen replacement, comprehensive psychiatric assessment and intervention go a long way in alleviating distress and enhancing quality of life in a person having CAIS.

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