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MAYER-ROKITANSKY-KUSTER-HAUSER (MRKH) SYNDROME WITH BEHAVIOURAL DISTURBANCE AND MENTAL RETARDATION: A CASE REPORT

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ABSTRACT

The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome affects 1 out of 4500 female in which there is underdeveloped or absence of female genitourinary system, vagina or uterus. It is transmitted as an autosomal dominant trait with an incomplete degree of penetrance and variable expressivity. The phenotypic expression of this syndrome involve anomaly of the reproductive system. The case " MIss X" presented with episodes of disorganised behaviour (self injurious). Miss X was provisionally diagnosed as seizure disorder with mental retardation. Detailed evaluation marked the abesnce of reproductive system. This possiblility of dual association of a major physical/anatomical abnormality and psychological disorder in young adolescent girl, is being presented here after taking valid consent.

KEYWORD: MRKH syndrome, seizure, disorganised behaviour, Mental Retardation

INTRODUCTION

A 26 year old Unmarried female, presented with complaints of withdrawn behavior since past 6 years, and episodes of excessive fearfulness, suspiciousness, muttering to self and episodes of self harm e.g. biting herself and scratching her body parts since past 3 years. The frequency of such episodes would be 7-8 episodes per week and each episode would last for around 10-15mins. Being from lower social-economic strata where cultural norms advocate the alternative form of treatments which are easily accessible, and hence had been she was treated by various faith healers in the past without significant improvement. Physical examination revealed multiple scratch marks on various parts of body including face. There was also bilateral chronic otitis media since age of 10 years. Patient had also broad and wide nasal bridge with frontal bossing. Mental state examination revealed a thinly build women with well developed secondary sexual characters. During coversation, she would make silly and inappropriate smile with restricted range of facial expressions and most of her answers would be in mono syllables, yes/no or short sentences. Because of her paucity of speech, no formal thought disorder, delusional disorder and perceptual disorder could be elicited. She was conscious and followed commands. Her attention could be easily drawn but concentration was sustained with difficulty. Memory was impaired and insight was absent. With complaints of primary amenorrhoea, ultrasound report revealed absence of uterus along with upper two-third part of vagina. Diagnosis of MRKH was entertained by the presence of primary amenorrhea, normal development of secondary sexual characteristics including external genitalia, and ultrasonography finding of absence of the uterine structure between the bladder and rectum. Her CT scan head and EEG came out to be within normal limits. Psychological assessment revealed I.Q. level of 65. Negative history revealed absence of episodic tonic clonic movements with loss of consciousness, persistent sadness of mood, grandiose ideas/big plans etc. The patient provisionally diagnosed as Mayer Rokitansky Kuster Hauser Syndrome with boderline mental retardation with behavioral disturbance (?? Psychosis). The patient was put on Risperidone 0.5mg-1.5mg in gradually increasing dose and there was significant decrease in the Self harm episodes. She became more emotionally stable with improvement in sleep, self care and reduced self muttering behaviour. After regular follow up for 6 months, the patient is significantly improved without symptoms suggestive of muttering to self. Her level of interaction and maintaing eye contact have been improved significantly.

Discussion:

MRKH syndrome affects reproductive system of females[1]. It is congenital abnormality characterized by the absence of vagina, cervix and the uterus. Due to incomplete development of the mullerian duct. Mullerian agenesis occurs in every 1 out of 4000-10,000 females[2-3]. The diagnosis of the syndrome is usually made at puberty. Patients usually present with

primary amenorrhea in adolescence age groups with normal physical growth and secondary sexual development. Affected individuals have a female chromosome pattern (46,XX) and normal functioning ovaries[4]. Although women with this condition are usually unable to carry a pregnancy, they may be able to have children through assisted reproduction. This diagnosis of mullerian agenesis has been an incidental finding in our patient who actually presented with borderline intelligence with behavioral abnormalities. Often a specific type of physical abnormality or deficiency may have chromosomal or neuro-developmental etiology which itself may play as a common etiological factor for the psychological disorder as well as the physical deformity. Therefore, keeping in mind this possible dual association of a major physical/anatomical abnormality and psychological disorder in young adolescent girl, the above case has been presented here after taking valid consent from the family members of the patient. The psychiatric co-morbidities in female population with mullerian agenesis is an area with limited research. A study from AIIMS, revealed a rare association of mental retardation and psychosis along with the MRKH syndrome. Whenever there is a physical deformity, there is stress which may lead to the psychological symptoms. Due to inability to bear child affected individuals may be more prone to neuropsychiatric manifestations such as distortions of body image, anxiety, depression, suicide, inter-personal sensitivity, and high psychological distress[5]. Also, physical deformity can have common genetic linkage with the psychological disturbance. Numerous studies reveal that 17q12 micro deletions in patients of MRKH associated with psychosis [6-7]. Our is first case report showing association of MRKH syndrome with psychosis and borderline intelligence. Owing to all these problems there is constant need to monitor psychological disturbance in patients of MRKH syndrome.

Hence, on the basis of this case report, authors emphasize need of awareness among psychiatrist and the Gynaecologist about the MRKH and its premature psychiatric manifestations.

Conclusion

Hence, with this case report we want to urge that, both psychiatrist and the Gynaecologist should be aware about the MRKH. A proper physical examination, personal history to be done of each psychiatric case. Both, psychiatrists and gynecologists need to be aware of this association of MRKH with psychological disturbance and should seek psychiatrist advice if needed. Proper psychoeducation of patient and the family members about the illness, so that they can cope up well.

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