An interesting case report of Psychosis in Turners syndrome.

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Abstract: This is a case report of a 21 years unmarried female presenting with auditory hallucinations, not attained menarche with primary ovarian dysgenesis with subclinical hypothyroidism and mild mental retardation.

Introduction: Psychiatric disturbances are seen in many of the sex chromosomal disorders. The clinical presentation may vary from frank psychosis to diagnostically inconclusive neuropsychiatric disturbances. No fixed pattern of established psychiatric disorder is the rule in such conditions eventhough occasionally some of them may mimic disorders that warrant a specific diagnosis based according to the ICD or DSM classifications. Moreover these conditions also impose restrictions and bring limitations to the use of psychiatric drugs because of their complex etiology. Turner’s syndrome is a sex chromosomal disorder that occurs in 1/2000 live births. Turner’s syndrome is due to the abnormality in the 45 X chromosome. It may be a monosomy in X Chromosome 45 (60%), 45, X/46, XX mosaicism (30%) or any other structural abnormality. Turner’s syndrome presents with predominant skeletal, reproductive, and lymphatic system disturbances or abnormalities. Physical characteristics include short stature, failure or reduction in secondary sexual characters, gonadal dysgenesis, and superficial features including webbed neck, hair loss, and cubitus valgus. Associated with the above, there may be mild mental subnormality or mental retardation, problems in attention, disturbances in memory, difficulties in social cognition and anxiety.

Case History:
An unmarried female aged 21 years, reported to the outpatient department of psychiatry referred by the physician. Reasons for referral were crying spells, headache, sleep disturbances and hearing voices all for period of 3 weeks. She was found withdrawn, communicating with hesitation in a low voice. She told about the voices inside her head (unknown male and female voices with derogatory comments) making her feel tense and giving her trouble. She was tearful most of the times during the interview.

Personal history revealed that she had not yet attained puberty. She stopped going to school after 8th standard. She was said to be far below average in her studies at school by the teachers. She was looking after household chores helping her mother. Physical examination revealed short neck, high arched palate, sparse axillary and pubic hair, breast Tanner’s Stage 2 suggestive of Turner’s syndrome. She had moderate mental retardation with an intelligence quotient of 56 on assessment. Routine haematological investigations revealed no significant abnormalities. Hormonal assays as suggested by the Endocrinologist showed the following: Elevated levels of serum Luteinizing hormone (LH) -16.88 mIU/ml (normal values 1.2-12.5 mIU/ml) and serum Follicle stimulating hormone (FSH)-93.61mIU/ml (normal values 3.2-15 mIU/ml). Serum Prolactin level was within normal range. She also had subclinical hypothyroidism. Total T4 - 10.03 microgram/dl and Thyroid stimulating hormone (TSH) 8.81 IU/ml. Ultrasound study of the abdomen showed hypoplastic uterus and non visualization of bilateral ovaries (streaky ovaries). A diagnosis of ovarian dysgenesis and subclinical hypothyroidism was made by the endocrinologist with the advice to start on Ethinylestradiol and Thyroxine sodium orally regularly. Screening of cardiac status revealed bicuspid aortic valve with moderate aortic regurgitation. She was started on antipsychotic risperidone and she had improvement of her psychological status. Her auditory hallucinations and other symptoms subsided within two weeks.

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Discussion:
Several cases of Turner Syndrome with Psychosis or affective disorders have been reported. Some investigators have hypothesized that mental disorders observed in Turner’s syndrome are a consequence of an X-chromosome defect. However, large population studies have demonstrated that the prevalence of mental illness in Turner Syndrome is not significantly high. Studies attempting to correlate Turner’s syndrome with psychiatric illness statistically have yielded mixed results. McCauley et al. studied 39 individuals with Turner’s syndrome and found high levels of major psychiatric difficulties and low self-esteem. The majority of individuals with Turner’s syndrome are known to have normal intelligence with normal verbal abilities. On the other hand, several studies have revealed that Turner’s syndrome patients may show decreased full scale IQ and have selective impairments with regard to visuo-spatial processing and visuo-perceptual skills. The risk of memory and attention problems and the risk of developing autism and attention deficit hyperactivity disorder in Turner syndrome are higher than in normal population. Turner’s syndrome personality characterized by excessive dependence, immaturity, depressiveness, passivity, distractibility and docility is suggested by Nielsen and Thomsen, although no rigorous scientific study has examined these claims. Downey et al. evaluated 23 Turner’s syndrome women and 23 controls, who were women with constitutional short stature, as well as 10 sisters of the Turner’s syndrome women with several rating scales measuring mood disorders, psychiatric symptoms, measures of adjustment and sexual behavior. They came to conflicting results. On one hand, they found that Turner syndrome women had fewer overt psychiatric symptoms; on the other, a global evaluation of psychological function and development, Turner syndrome women ranked lower than the short stature controls, but similar to their sisters. The XO karyotype of Turner’s syndrome is characterized by primary amenorrhea, with streak ovaries and consequent sexual infantilism, short stature and multiple congenital anomalies. Facial appearance is often characteristic, with a small jaw, fish lime mouth and low set ears. Other systems commonly involved include cardiovascular system (bicuspid aortic valve in 50%, coarctation of the aorta in upto 20% and frequent hypertension), the urinary system (horse shoe and other structural abnormalities of the kidneys) and endocrine system (primary hypothyroidism in upto 50% and glucose tolerance is common).

Conclusion:
Despite the descriptive and observational information, the literature lacks rigorous, statistical examination directed towards identifying the described syndrome in Turner’s syndrome. The fact the syndrome is not widely present in Turner’s syndrome women hints at a shared organic basis vulnerability to this particular psychopathology inherent in a subset of Turner’s syndrome patients, rather than a direct genetic cause for the syndrome. Although familial Turner’s syndrome has been reported, there have been no solid reports of familial cases in which both Turner’s syndrome and Psychosis are co-transmitted. It is true that in genetic disorders any associated chromosomal abnormalities may permit the identification of a disease causing gene, and analysis of a contiguous gene syndrome may clarify a genotype-phenotype correlation. If it is a unique syndrome, it is important to identify it in order to avoid labeling patients with a more severe diagnosis, to determine its frequency and epidemiology, to understand its biological basis, and most importantly to identify optimal treatment strategies.

References: