A Psychotic Reaction in a Sex-Chromatin Negative Female*

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SUMMARY

Neuropsychiatric disorders found in association with Turner's syndrome (gonadal agenesis) are unusual. The case report describes a patient with Turner's syndrome who suffered an acute psychotic reaction.

A brief review of the literature concerning this association is surveyed.

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A considerable variety of karyotypes with abnormalities involving the sex chromosomes have been described, either possessing varying numbers of both X and Y chromosomes, or showing evidence of more than one stem-line of cells—so-called mosaicism.[1] From among these anomalies, several have emerged with sufficient degree of constancy and frequency of their clinical features for them to be regarded as specific syndromes. In anatomical males these include XXY (Klinefelter's syndrome) and XYY, while in anatomical females XO (Turner's syndrome) and XXX are found.

Turner described a syndrome in women comprising the triad: infantilism (short stature and underdeveloped secondary sexual characteristics), webbing of the neck, and deformity of the elbow (cubitus valgus).[2,3] These individuals were later shown to be chromatin-negative, and to have the XO sex chromosome pattern.[4] This particular configuration is the most common pattern demonstrated, but variants do occur, usually as mosaics with a stem-line of normal cells.[1]

Turner's syndrome itself is a rare condition occurring in only 0,3 - 0,5% of a 'world-pool of new-born girls'.[5,6] The anomaly is common among spontaneous abortions, and girls with the XO configuration are more apt to die in childhood than other girls.[5] Consequently the condition is still more rare among adult females.

Since the original report by Turner, a great variety of associated physical anomalies have been described, but there are only few scattered reports on neuropsychiatric disorders in association with the syndrome of gonadal agenesis (Turner's syndrome), whether it occurs in the pure form or as a mosaic pattern. Therefore, a psychotic reaction in an individual with the XO configuration seems worth reporting.

It appears that many of the psychological characteristics observed in Turner's syndrome are secondary, being reactions to oddness of appearance or physiological function. This is borne out by the psychiatric study carried out by Sabbath *et al.*[7] on 7 adolescent girls with ovarian agenesis. The degree of dwarfing of stature appeared to play a considerable part in their attitudes and behaviour patterns. Characteristic of the group was the vagueness and blandness of their personalities.

Similar findings of passivity of personality, vagueness and lack of involvement were made by Hampson *et al.*[8] They studied 19 women with Turner's syndrome from a psychological point of view. None of the women was considered neurotic or psychotic. Their personality make-ups showed marked lack of aggressiveness, drive and initiative.

As regards the actual association of Turner's syndrome and psychosis, there are only a few scattered reports. Chromosomal investigations of already diagnosed schizophrenics have not shown a greater or lesser incidence of the association with Turner's syndrome. Kaplan and Cotton,[9] in a chromosomal investigation of 986 institutionalized schizophrenics, were able to find only 4 who had an associated Turner's syndrome. Similar large studies did not show any association whatsoever.[10-13]

Slater and Zilkha[14] described a case of Turner's syndrome showing an unusual form of myopathy, and a brief psychotic episode diagnosed as schizophrenia. Milcu *et al.*[15] described another case in which Turner's syndrome was found in association with a psychosis diagnosed as schizophrenia. Mellbin[15] studied a small select group of 4 women with Turner's syndrome who showed a common abnormal EEG pattern, but varying psychiatric symptomatology. Of these, 2 women showed psychotic reactions, one of the chronic variety and the other a single acute episode.

CASE REPORT

The patient, aged 33 years on admission, is the youngest of 6 siblings. She is 1,49 m tall and weighs 69,7 kg. She looks old for her age with low-set protruding ears and a broad, short neck. Her wrists are broad and mis-shapen, and she displays a marked cubitus valgus. Her breasts are hypoplastic with retracted nipples in the anterior axillary line; she has a small portio vaginalis, but no palpable uterus or ovaries. She has never menstruated.

Chromosomal analysis showed that the patient had the karyotype 45/XO (Fig. 1).

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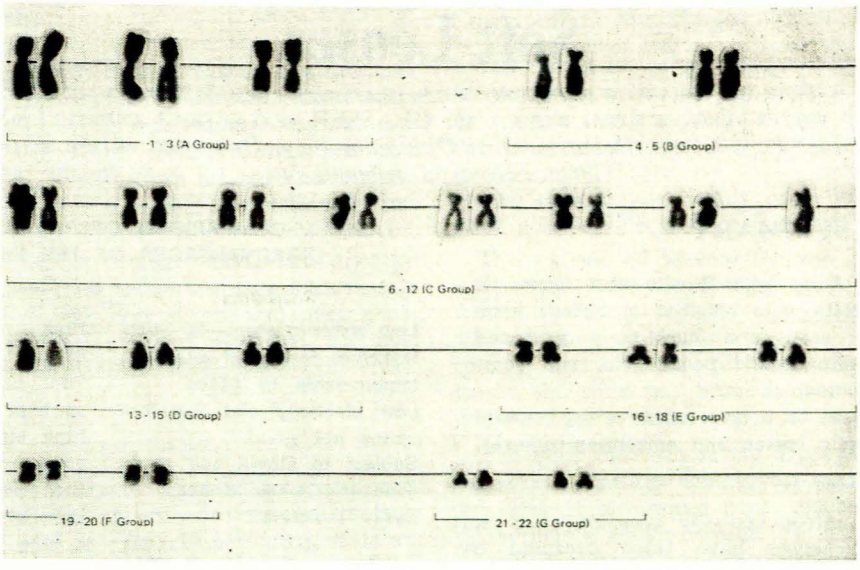

Fig. 1. Chromosomal analysis showing 45/XO karyotype.

Her birth was normal and followed an uneventful pregnancy. She developed normally as a child, and managed well at school, passing Std VIII before leaving school. Since that time she has worked consistently as a machine-operator for the same firm.

Always a quiet, retiring person, the patient suddenly became withdrawn, despondent and emotionally labile 3 months before her admission to hospital. Her attitude towards her mother changed, and she would lock herself in her room for long periods, refusing to see anyone. During this period she became acutely anxious and complained of auditory hallucinations. Impulsively she tied a cord around her neck in an attempt to strangle herself, and she was found unconscious with a suffused face and the burn marks of the cord on her neck.

She was admitted urgently to hospital where she responded well to 6 sessions of electroconvulsive therapy and a combination of perphenazine and amitriptyline. At follow-up 12 months later, she was apsychotic and coping well.

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