NOONAN'S SYNDROME WITH MENTAL RETARDATION PRESENTING WITH AN AFFECTIVE DISORDER – CASE REPORT

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ABSTRACT

A 30 year old Chinese lady initially thought to have Turner's Syndrome was redlagnosed as Noonan's Syndrome following admission to a psychiatric hospital for treatment of an Affective Disorder (Hypomania). The genetics and morphological features in Noonan's Syndrome are briefly discussed and the patient's psychiatric presentation is described.

Key Words: Noonan's Syndrome, Affective Disorder, Hypomania, Turner's Syndrome.

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INTRODUCTION

Noonan's Syndrome is a rare inherited disorder first described by Ulrich in 1930 in an 8 year old girl. Turner described somewhat similar features in 1938 in elderly female patients. However with the discovery of sex chromatin by Barr and Bertram, the distinction became clearer; Caflisch clearly separated the Ulrich Syndrome (chromatin – positive) from Turner's Syndrome (chromatin – negative). After 1963, the eponym Noonan's came into use when Noonan and Ehmke reported the same non-cardiac malformations in a group of children with congenital heart disease (1).

The morphologic features in both the syndromes are similar. (See Table 1). The other features noted in Noonan's Syndrome are given in Table 2. Cardiac symptoms when present in Noonan's Syndrome are due to lesions in the right side of the heart (pulmonary stenosis is common) (1, 2).

Girls with Noonan's Syndrome are chromosomally 46XX and have ovaries and boys are 46XY and have testes. Individuals with Turner's Syndrome have a chromosomal anomaly, usually 45XO and have dysgenetic gonadal streaks (2).

In Noonan's Syndrome, the genetic defect is transmitted in a Mendelian manner but exhibits variable expressivity and is believed to show incomplete penetrance. The incidence of Noonan's Syndrome in the general population is not known. This is likely to be due to the extreme variability of the somatic stigmata from mild to severe, even within the same family.

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Table 1
Morphological Features in Turner's and Noonan's Syndrome

Short stature	Low posterior hairline
Neck webbing	Abnormal dermatoglyphics
Cubitus Valgus	Epicanthus
Hyperteleorism	Shield chest
Ptosis	Sternal deformity
Dental Malocclusion	Low set ears
High arched palate	Nail dystrophy

Antimongoloid slant of the palpebral fissures
Lymphedema of the dorsum of the hands and feet
Hypoplastic or widely set nipples
Woolly consistency of the hair

Table 2
Other Features seen in Noonan's Syndrome

Cryptorchidism	Spina bifida
Delayed menarche	Nerve deafness
Renal anomalies	

CASE REPORT

AH, a 30 year old Chinese lady, was referred with a two day history of restlessness, insomnia and disturbed and abnormal behaviour. This had begun after she accidentally touched an amulet at work.

AH had 2 brief episodes in the past when she was depressed about her future and expressed suicidal thoughts.

There is no family history of an affective disorder, suicide or alcoholism. Neither is there somatic stigmata of Noonan's Syndrome in other family members. Her birth and developmental milestones were normal. The abnormal physical features were noted early and she was diagnosed as a case of Turner's Syndrome at age 8 years. She had primary nocturnal enuresis which stopped when she was 15. AH failed every year in school and left after Primary 6. She stayed home till she began working as a nursery helper a year ago.

AH was described as an introvert with no mood swings. She had no special interests or hobbies till involvement in church activities 2 years ago. When young, AH was frequently teased about her features and she often wondered about her physical development. Menarche was at 16 years and she was frightened by it. As she had no peer relationships, she was lonely and was upset when her sisters began dating. She now has some male acquaintances among church members. But 2 months earlier, a friend had made sexual advances and she became frightened.

On mental state examination, she was restless and distracted. In conversation, she was playful and her voice was low-pitched and hoarse. She had flight of ideas and pressure of speech with rhyming and clang associations. There were grandiose delusions but no hallucinations. Her mood was elated. The cognitive functions were intact.

Physical examination showed the following: short stature, (height 142.5cm), weight 32.5 kg, marked epicanthic folds, broad nasal bridge, low set ears, short webbed neck, cubitus valgus deformity, low hair-line, small breasts, shield-like appearance of the chest.

Investigations

(a) Psychological Assessment: Verbal IQ 67-72. Full Scale IQ 68-70. Borderline Range.
(b) Chromosomal Studies done at the Paediatric Unit, SGH, during the second admission: 46XX. Rediagnosed as Noonan's Syndrome.
(c) Biochemical Investigations: Results within normal limits.

AH was diagnosed as suffering from an acute psychotic illness. With phenothiazines (Chlorpromazine) her mental state improved within a week and her family requested discharge.

But once home, she became overactive, talkative and extravagant. During the second admission her mental state took almost 4 weeks to settle; she was on Tab Haloperidol 6 mg tds, Tab Chlorpromazine 100 mg tds, Tab Artane 4 mg tds and Tab Valium 10 mg on.

After discharge she remained symptom-free and was able to work. The medication was tailed down and discontinued after 9 months. But a year later, AH relapsed with similar signs and symptoms and needed inpatient treatment.

DISCUSSION

This case fits the criteria, for an Affective Disorder, listed by the Diagnostic and Statistical Manual of Mental Disorders (Third Edition) and the Ninth Revision of the International Classification of Diseases.

Noonan's Syndrome is a well-documented disorder and Mental Retardation is commonly noted in Noonan's Syndrome. There is in the medical literature reports on cognitive abilities and psychosocial aspects of patients with Noonan's Syndrome (2, 3).

But there is no regular association with psychiatric problems and we could find no other reported case of a Noonan's Syndrome with an affective disorder. While the syndrome itself is an additional biological and social burden, it is interesting to note that in Turner's Syndrome (where the morphological features are similar), sexual immaturity and body defects do not seem to result in any emotional disturbance (4). Only isolated instances of psychosis have been reported in sex-chromatin negative women (5).

In the absence of a family history of an affective disorder, it is likely that the occurrence of the two together is coincidental.

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