

Psychological and Psychiatric Aspects of Turner Syndrome: Genotypic or Environmental Effects?

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The objective of this study is to assess the psychological aspects of Turner syndrome and to identify their possible etiological relation to either chromosomal or environmental factors. Ten girls with Turner syndrome (Karyotype) and eleven Turner - Like girls (without chromosomal aberration) were properly matched and investigated using the psychiatric interview, WICS, BSRI, CBCL and SCL-90 and results were compared. Results revealed that both groups had cognitive deficit characterized by discrepancy between performance IQ and verbal IQ. Performance IQ scores were much less than verbal ones. No substantial gender identity disturbance was detected in both groups. Except for an equally high tendency for 'Interpersonal sensitivity' on the SCL-90 in both groups, results on all measures of behavior and psychiatric status were nearly within normal for the two groups. However, girls with Turner syndrome were astonishingly less symptomatizing than Turner-Like ones on the SCL-90. To conclude, the so called characteristic style of Turner syndrome on intelligence tests may not be characteristic as it was equally identified in Turner - Like girls. Our findings do not support the hypothesis that the chromosomal anomaly is considered responsible for such characteristic style, but suggest the possible effective role of environmental factors, arising secondary to the physical characteristics of the syndrome.

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Introduction

Turner syndrome occurs at least in one every 5000 female live birth (Lippe et al, 1988; Thompson et al, 1991). Classical clinical features of the syndrome include small stature and sexual infantilism. Webbing of the neck, elbow deformity and congenital heart diseases are common features. (Nyhan and Sakati, 1979). However, the clinical picture depends, to a great extent on the karyotype variants. (Temtamy et al, 1992). Almost 50% of the females with Turner syndrome have 45X karyotype, 25% have mosaic karyotype and the remaining 25% show structural abnormalities of

the X chromosome. The latter variant might produce partial monosomy. (Thompson et al. 1991).

Pseudo-Turner syndrome or Noonan syndrome occurs in one every 1,000-2,500 female live births. (Gronchy and Turleau, 1984). It is called pseudo because its clinical features are similar to the Turner syndrome but in the absence of chromosomal abnormalities i.e. normal female karyotype is 46 XX.

Turner syndrome has been claimed to be associated with cognitive and behavioral disorders. Some studies reported poor performance of females with Turner syndrome on intelligence tests. Money (1963) reported significant difference between verbal IQ and performance IQ. Garron and Vander - Stoep (1969) added evidence for the presence of poor visuospatial abilities of intelligence and confirmed the normality of verbal abilities. However, in contrast to these earlier studies, recent studies state that the overall IQ distribution of females with Turner syndrome appear to be normal. (Thapar et al, 1994).

Greater attention should be directed to behavioral and psychological disturbances in these females. The concept of "mental problems" had been used by many authors investigating Turner syndrome in a very broad sense, ranging from very mild to manifest mental illness. Some studied personality and found traits of passivity, vagueness and lack of initiative and mastery (Christodoresen et al, 1970); while others described children with Turner syndrome to be friendly and extremely easy to manage (Nielsen and Silesan, 1981).

To study cognitive and behavioral characteristics of Turner syndrome, we need to clarify the following controversy: Are these cognitive and behavioral characteristics of girls with Turner syndrome secondary to the chromosomal abnormality,

with its direct endocrinal impact on the brain? or are they related to environmental factors and the obvious phenotypic characters of these girls: Many authors (Stafford, 1961; Garron and Vander - Stoep, 1969; Goodenough et al, 1977 and Weber, 1979) adopted the first opinion i.e. a deficit in the brain function, while others (Nielsen et al, 1977 and Nielsen, 1985) believed more in the influential effect of the environmental factors, especially the role of the parents in managing these girls.

Aim of the Work

1- Cognitive and behavioral assessment of Egyptian girls with Turner syndrome.

2- A trial to answer the question of whether the cognitive and behavioral abnormalities - if present - are due to chromosomal or environmental factors.

Subjects and Method

Twenty-one consecutive female patients aged between 8-16 years were recruited in this study. They constituted all the whole clinical cases referred for suspected Turner syndrome to the outpatient clinic of the Human Genetic Department of the National Research Centre - Egypt. The duration of the study was one and half years.

The cytogenic study was done by chromosomal analysis of G-banded chromosome from peripheral blood lymphocytes cultures. This analysis showed that ten cases have the karyotype of Turner syndrome and eleven cases - in spite of having the phenotype of Turner syndrome - had normal karyotype (46 XX). Analysis of the Turner group (10 cases) revealed two types: A) six cases were of the classical type (45 X b.) four cases were not classical (three with karyotype 45, x/46, Xi (Xq) and one case with karyotype 46, XX (q).

Therefore, we have two main groups: Turner group (n= 10) and Turner - Like female group (n= 11).

Both groups were subjected to:

1- Full medical assessment recording dysmorphic features of Turner syndrome and anthropometry.

2- Full psychobiogram.

3- Psychometric assessment:

a) Wechsler Intelligence Scale for Children (WISC) - Arabic version to assess IQ.

b) Bem - Sex Role Inventory (BSRI) - Arabic version (Demerdash and El - Hady - unpublished translation). This inventory was used to assess the masculinity - femininity dimension.

c) Child Behavior Checklist is given to the parents or teachers of the child to assess several syndromes and symptoms in children.

d) The Symptoms Checklist-Arabic version (El Defrawi et al, 1992). This check list (SCL-90) - Arabic version (Ibrahim, 1985). This is a selfreport clinically rating scale oriented to measure the symptomatic behavior of psychiatric outpatients.

The results were subjected to statistical analysis, namely means, standard deviations, t-test and chi-square.

Results

1-Demographic data: (Table 1)

demographic data show that the two groups are matched for age, level of education as well as occupation.

2-Anthropometry and Turner syndrome physical stigma: (table 2,3) Table (2) shows that all patients of both groups were of short stature. Both groups were matched for body weight and height. Table (3) shows that the percentage of face, neck, limbs, chest, and heart anomalies in both groups are simi-

lar. This means that the clinical features of both groups are alike, i.e. Turner features.

3-Intelligence: (Table 4,5)

Table (4) shows the means of subscales of WISC in both groups. There were no statistical differences between both groups on all subscales. Table (5) shows that only two patients from every group have IQ less than 65 and one from every group can be diagnosed as borderline. It is also noticed that performance IQ, the difference being around 33.

4-Bem-Sex Role Inventory: (Table 6)

The mean femininity scores for both groups were similar. On masculinity scores, the mean for Turner - Like group was just significantly higher than the Turner group ($p= 0.050$). All cases are considered feminine. None of the cases showed any deviance for sex role identity.

5-Child Behavior Checklist (CBCL): (table 7)

Both groups showed no significant differences on all scores of SCLC. When our results were compared with norms of CBCL according to Achenbach and Edelbrock (1983) our total scores of child behavior profile exceeded the upper limits of normal ranges in only two cases (20%) in Turner group and three cases (27%) in Turner - Like group.

6-The Symptoms Checklist (SCI-90): (Table 8)

Our results revealed that the Turner - Like group showed statistically significant higher scores than Turner group on all scales of the check list except for one item "the interpersonal sensitivity" in which both groups were similar. This means that the Turner group girls show less self reported symptoms than Turner - Like girls.

Table 1
Comparison of the Demographic Data of the Two Groups (TS and TL)

Demographic Data	Turner N = 10		Turner - Like N = 11		P*
	No	%	No	%	
Age					
8-11	3	30%	4	36.4%	
12-16	7	70%	7	36.6%	
Mean	13.00		12.82		
SD	2.79		1.83		0.43
Occupation					
Student	9	90%	9	81.8%	
Farmer	1	10%			
No work	0		2	18.2%	
Level of Education					
Illiterate	1	10%	2	18.2%	0.9289
Primary School Student	3	30%	2	18.2%	0.9028
Preparatory School Student	5	50%	7	63.6%	0.7498
special School Student (remedial school)	1	10%	0		

* Chi - square test.

Ts : Turner - Syndrome

TL: Turner - Like group

Table 2
Weight and Height at the Age of Physical Examinations in the
Two Groups Compared

	Turner		Turner - Like		P*
Weight					0.3720
Mean	28.85	Kg 12.0	25.45	Kg 8.27	
SD					
Height	120.95	Cm 17.05	120.76	Cm 12.06	
Mean					0.4885
SD					

* T test

Table 3
The Frequency of Somatic Features Known to Occur in the Syndrome in the Turner Group (Karyotypically Turner) and the Turner - Like Group (Karyotypically 46, XX) Compared

Physical Findings	Turner N = 10		Turner - Like N = 11		P*
	No	%	No	%	
A. Face					
1- Wide epicanthal fold	2	20%	4	36.4%	0.7298
2- Slanting eyes	3	30%	3	27.3%	0.7398
3- Hypoplastic maxilla	1	10%	3	27.3%	0.6524
B. Neck					
1- Short	3	30%	3	27.3%	0.7298
2- Webbing	2	20%	1	9.09%	0.9289
C. Limbs					
1- Cubitus valgus	7	70%	5	45.5%	0.4879
2- Short 4th & 5th metacarpals	7	70%	6	54.6%	0.7806
D. Chest					
1- Shield chest	1	10%	2	18.18%	0.9289
2- Widely spaced nipple	3	30%	2	18.18%	0.9089
E. Pigmentation	4	40%	2	18.18%	
F. Puberty					0.5341
1- Perpubertal	9	90%	11	100%	
2- Reached	1	10%	0		
G. Heart					
1- Free	10	100%	10	90.91%	
2- Functional - murmur	0		1	9.09%	

* Chi - square test.

Table 4
Mean IQ (WISC) of the Two Groups Compared

	Turner	Turner-Like	T	P
Total				
Mean	88.50	84.72		
SD	21.70	23.37	0.39	0.35
U.L.-M.S.	105.40	102.45		
SD	23.71	25.62	0.27	0.39
Performance IQ				
Mean	72.80	68.63		

Table 5
IQ Distribution Among the Two Groups

Total IQ Distribution	Turner N = 10	Turner - Like N = 11
110	2	2
100-110	1	5
75-90	1	1
65-75		

Table 6
Results of the Bem Sex-Role Inventory in the Two Groups Compared

Bem Sex - Role Inventory	Turner No. = 10	Turner - Like No. = 11	T	P
Masculinity "b"				
Mean	1.08	1.32		
SD	0.28	0.34	1.92	0.05
Femininity "a"				
Mean	2.55	2.59		
SD	0.34	0.40	0.27	0.39

Table 7
Results of the Child Behaviour Checklist in the Turner and Turner - Like Compared

Child Behaviour Checklist	Turner		Turner - Like		T	P
	Mean	SD	Mean	SD		
Total Score	42.4	23.3	43.18	21.12	0.08	0.46
Internalizing Syndromes						
1- Total	17.9	9.8	21.9	13.9	0.75	0.23
2- Schizoid	3.3	2.11	3.36	2.29	0.06	0.47
3- Depressed	7.9	5.04	8.36	5.75	0.19	0.42
4- Uncommunicative	4.2	2.29	4.09	3.01	0.09	0.46
5- Obsessive Compulsive	5.5	3.44	4.81	3.21	0.46	0.32
6- Somatic Complaints	3.8	2.65	6.46	5.46	1.43	0.08
Externalizing Syndromes						
1- Total	18.0	9.82	14.9	7.5	0.81	21
2- Hyperactive	6.40	3.43	4.76	2.99	1.35	0.09
3- Aggressive	11.20	7.67	11.54	6.53	0.11	0.45
4- Delinquent	1.60	2.11	1.18	1.25	0.55	0.29
Mixed Syndrome (Social withdrawal)	4.2	2.89	3.54	2.24	0.56	0.28

Table 8
Mean Scores on Different Symptom Dimensions of the SCL-90
in the Two Group Compared

The Symptoms Dimensions	Turner No. = 10	Turner - Like No. = 11	T	P
1- Somatization***				
Mean	2.00	10.36		
SD	2.21	7.10	-3.56	0.001
2- Obsessive-Compulsive **				
Mean	5.80	11.72		
SD	4.96	3.38	-2.35	0.01
3- Interpersonal ~ Sensitivity				
Mean	5.80	9.00		
SD	8.09	7.77	-0.92	0.18
4- Depression *				
Mean	5.30	10.72		
SD	6.16	7.07	-1.86	0.03
5- Anxiety *				
Mean	4.00	8.45		
SD	3.59	5.92	-2.05	0.02
6- Hostility **				
Mean	09.90	2.63		
SD	0.99	2.29	-2.20	0.01
7- Phobic Anxiety*				
Mean	3.20	7.72		
SD	4.46	6.08	-1.92	0.03
8- Paranoid Ideation*				
Mean	1.30	2.90		
SD	2.11	2.34	-1.64	0.05
9- Psychoticism *				
Mean	2.10	4.63		
SD	2.96	3.61	-1.74	0.04

Discussion

This study covered cognitive as well as behavioral profiles of two well matched groups of Turner syndrome and Turner - Like groups (46 XX). The mutual application of self report and reports by parents add more weight to the findings.

Our results showed that the cognitive abilities of both groups were disturbed in the same pattern. The main deficit was in the performance IQ as measured by WISC. The differences between the verbal IQ and performance IQ were marked.

Our findings confirm earlier studies on Turner syndrome (Shaffer, 1962; Money, 1963; Garron and Vander - Stoep, 1969 and Christensen and Nielsen, 1981). Moreover, the advantageous use of a control group of girls with Turner - Like syndrome in our study adds more weight to the hypothesis of environmental factors imposing direct effect on the cognitive abilities of those girls. The similar profile of scores on IQ test in both Turner (cases) and Turner-Like (controls) gives no support to the hypothesis of chromosomal effects on the cognitive or cerebral functions, whatever its mechanism.

Our findings, therefore, favour the environmental influence in both groups. It highlights the possible effect of evident physical stigmata on those girls and the possible role of modes of upbringing that might be different from that of normal girls. We therefore agree with Nielson (1985) who stressed the importance of the effect of parents, teachers and ultimately society on the cognitive development of females with Turner syndrome.

A similar percentage of mild mental retardation (20%) in Turner syndrome and Turner - Like group was represented in our sample. A percentage which is clearly higher than that reported for the general population. However, accurate deductions cannot be made from such a small sample. Exact prevalence of mental retardation should only be obtained from bigger samples or best from epidemiological studies. It ought to be mentioned that all cases were referred for evaluation of the cause of short stature and mental retardation is a common association.

The second important finding in our study was the absence of gender role problems in both groups. All participating girls exhibited good scores on femininity questions of BSRI and low scores on masculinity. This finding agrees with Ihrhardt et al (1970), who have indicated that the psychological aspects of sexual development of these girls during this phase - mostly preadolescence - are intact. However, if we are to expect problems in their psychosexual development, it will be during adolescence : a point which is beyond our study.

The third finding in our study is the report of parents on CBCL pointing to the normality of most girls in both groups (80% of Turner group and 78% of Turner - Like group). These results are comparable with reports of parents of normal children (Jones et al, 1988 and

Sandberg et al, (1991). A possible explanation for this finding is the presence of relatively good cognitive abilities of these girls with physical stigmata, that make them seek proper means of communication with others and attempt to gain their acceptance, especially if the attitudes of those others are positive.

The last finding in our study to explain is the presence of significant discrepancy between the two groups on their self report (SCL-90). The Turner group girls were much less significantly symptomatizing than Turner-Like group. The only exception was on the interpersonal sensitivity scores which reflected sensitivity and feelings of inadequacy in interpersonal relationships in both groups; a point which is easily explained by the shared physical stigma (Nielson and Silesen, 1981 and Meyer Bahlburg, 1985).

Results on the other dimensions provoke a question mark. Although both groups show no significant differences in somatic features, anthropometry, intelligence, gender role, parents reports and even the interpersonal sensitivity, why then are the Turner girls less symptomatizing?

From our point of view there are two possible explanations. First is that girls with Turner syndrome are more defensive and are possibly denying their symptoms. This denial might be attributed to early discovery of the disorder by the parents that implements a change in their attitudes towards those girls leading ultimately to such defensiveness on the part of the girls. On the other hand, late diagnosis and consequently less negative attitude of parents renders Turner - like syndrome girls less defensive and less denying to their conflicts. The second possible explanation is the low validity of SCL - 90, being a self reporting tool and possibly exhausting (Lewis, 1989)

or boring to the child. This might even explain the discrepancy between the children's (SC - 90) and the parents' (CBCL) reports where the latter results showed no difference between the two groups. However, and in spite of the suggested explanations, our question needs further investigation and more elaborate tools to be accurately answered.

To conclude, the overall intelligence of girls having Turner syndrome is mostly similar to that of Turner - Like syndrome ones. The so called characteristic profile of discrepancy between Performance IQ and Verbal IQ is not characteristic to Turner syndrome but is possibly present in most short statured girls and is mostly attributed to environmental factors. The behavior of those girls are found to be within the normal ranges.

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Les aspects psychologiques et psychiatriques du syndrome de Turner: Influence du génotype ou de l'environnement ?

L'objet de cette étude est d'évaluer les aspects psychologiques du syndrome de Turner et d'identifier leurs relations étiologiques avec soit des facteurs chromosomiques soit environnementaux. Un nombre de jeunes filles atteintes du syndrome de Turner, avec et sans anomalies chromosomiques, ont fait l'objet d'une évaluation comparative. Nos résultats ne soutiennent pas l'hypothèse que les anomalies chromosomiques sont à l'origine du style psychologique caractéristique de ce syndrome, il nous semble que les facteurs environnementaux jouent un rôle important et ceci comme conséquence aux traits physiques relatifs au syndrome.

الجانب الطبى النفسى لزملة ترنر التأثير الوراثى أم البيئى؟

تناقش هذه الدراسة الجانب النفسى والطبى النفسى لزملة ترنر للتعرف على احتمال وجود علاقة بين العوامل الوراثية والبيئية المسببة والعوامل النفسية والطبى النفسية.

وقد تم فحص وتقييم عشرة فتيات مصابات بزملة ترنر (مع وجود تشخيص وراثى) ، واحدى عشرة فتاة من اللاتى يعانين من نفس الشكوى الجسدية (بدون تأكيد تشخيص وراثى)، وقد تم إجراء المقارنة من حيث الاعراض الطبى النفسية والمشاكل والصعوبات السلوكية - والاختبارات الذكائية المعرفية (مقياس وكسلر لذكاء الأطفال).

وقد أشارت النتائج إلى وجود اختلال فى الوظائف المعرفية الادائية واللغوية بالإضافة إلى وجود فروقات بين المجموعتين - كما ناقشت الدراسة العلاقة بين العوامل الوراثية والنفسية المختلفة وبين الأسلوب المميز لأداء الفتيات المصابات بزملة ترنر - واستبعدت الدراسة أن تكون الاختلالات الوراثية فى الكروموزومات هى السبب الأساسى للمظاهر السلوكية والمعرفية المصاحبة لهذا الاضطراب.