A case study of young patients affected by Turner syndrome: competency and psychopathology

Studio casistico di giovani pazienti affette da sindrome di Turner: competenze e psicopatologia

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Summary

Aim

This study's purpose was to evaluate psychological and affective-relational characteristics in a group of pre adolescents and adolescents affected by Turner syndrome, with the aim to focus on the importance of a multidisciplinary intervention.

Materials and methods

A group of 47 girls affected by Turner syndrome and a group of 47 healthy girls were enrolled. The age of the subjects was between 10 and 19 years. In order to evaluate emotional and social aspects in Turner girls compared to healthy girls, a semi-structured interview was conducted to collect personal data, family, psycho-social, extra-school habits and therapeutic intervention. Furthermore, in order to evaluate psychopathological dimensions, T. Achenbach's Youth Self Report was administered to patients and controls.

Results

This study highlighted some critical aspects regarding Turner girls, like dissatisfaction with their appearance, the importance of the relationship with parents which influences both the acceptance of the illness and several other psycho-social aspects, difficulties with their peer groups, and the presence of psychopathological symptoms (internalizing problems rather than externalizing ones).

Conclusions

Patients undergoing a psychiatric/psychological consultation received benefits, particularly regarding social integration, self-fulfilment, and satisfaction with one's own appearance. A multidisciplinary intervention is strongly suggested with patients affected by Turner syndrome, particularly in adolescent age.

Key words

Turner syndrome • Adolescence • Psycho-social functioning • Psychopathology • Multidisciplinary interventions.

Riassunto

Scopo

Il presente studio si propone di valutare gli aspetti psicologici e affettivo-relazionali in un gruppo di preadolescenti e adolescenti affette da sindrome di Turner, allo scopo di focalizzare l'importanza dell'intervento multidisciplinare.

Materiali e metodi

Sono stati arruolati un gruppo di 47 ragazze affette da sindrome di Turner e un gruppo di 47 ragazze sane di età compresa fra i 10 e i 19 anni. Al fine di valutare gli aspetti emotivi e socio-relazionali delle ragazze affette da Turner confrontate con quelle sane, è stata condotta un'intervista semistrutturata per raccogliere dati personali e familiari, relativi ad abitudini psicosociali ed extrascolastiche, e all'intervento terapeutico. Inoltre, per valutare elementi psicopatologici è stato somministrato a pazienti e controlli lo YSR di T. Achembach.

Risultati

Questo studio evidenzia alcuni aspetti critici riguardanti le ragazze affette da sindrome di Turner, quali l'insoddisfazione per l'aspetto fisico, l'importanza della relazioni con i genitori che influenza sia l'accettazione della malattia sia diversi altri aspetti psico-sociali, le difficoltà con il gruppo dei pari, e la presenza di sintomi psicopatologici (problemi internalizzanti piuttosto che esternalizzanti).

Conclusioni

Le pazienti sottoposte a intervento psicologico-psichiatrico ricevono dei benefici, in particolare circa l'integrazione sociale, l'autostima e la soddisfazione circa il proprio aspetto. Un intervento multidisciplinare è fortemente raccomandato con pazienti affette da sindrome di Turner, specie in età adolescenziale.

Parole chiave

Sindrome di Turner • Adolescenza • Funzionamento psico-sociale • Psicopatologia • Interventi multidisciplinari

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Aim

This study evaluated the affective-relational and psycho-social characteristics (with particular attention paid to familiar and peer relationships) and possible psychopathological aspects of adolescent girls affected by Turner syndrome compared to those of the case control group, with the aim to focalize the importance of a multidisciplinary intervention that contemplates psychosocial aspects of these patients close to the somatic ones. According to that, we wanted also to analyze how the psychological intervention, when used as treatment associated with the pediatric one in the studied sample, has influenced the above mentioned characteristic. The initial hypothesis was that Turner syndrome girls have more psychological diseases and psychopathologies then controls, and for this reason a psychological-psychiatric treatment should be applied as part of the protocol rather then being considered an optional intervention.

The sample

Of all the 55 patients (age between 10 and 19), affected by the Turner syndrome followed up by the Pediatric Unit of Padua Hospital at the moment of this study, two (both 14 years old) were excluded from the case study, because they showed comorbility with another pathology (X-fragile Syndrome and Cardiopathy). Three other girls (two of 14 and one of 18 years of age) decided not to be included in the study. Besides, three of the girls who had initially decided to take part in the study, they changed their mind and did not fill in the questionnaire as their parents thought they were too young to answer same of the questions (their age was: 10, 11, 12 years).
So the final sample includes 47 girls with a median age of 15 ± 2.9 years, respectively 23 individuals aged between 10 and 15 and 24 individuals aged between 16 and 19 years.

The control group

The group consists of 47 girls with a median age 15.2 ± 2.8 and includes 21 girls aged between 10 and 15 and 26 girls aged between 16 and 19 years.
As reported in Table I, the two groups are of similar age, ethnicity, familiar cultural level (evaluated on single parent's education), parents' marital status.

Materials and methods

The research has been conducted in full accordance with ethical principles, including the World Medical Association Declaration of Helsinki. Parents received information on the study and gave written consent for themselves and the adolescents. The adolescents were also informed and gave their consent.

Some individual, familiar and clinical data reported in table 1 were partly collected from case history, and partly asked during a meeting focused on talking about the research's aim, to collect the informed consensus and to conduct a semistructured interview (*Valutazione di Abilità Definizione di Obiettivi*, VADO) [1] where aspects related to individual characteristics (self perception, acceptance and satisfaction, projects for future), relationships within family and extra family environment, school (attitudes, performance, classmates) and activities were investigated. Besides, the Youth Self Report by Thomas M. Achenbach (YRS, 1991 version) [2] for the evaluation of competences and psycho-behavioural problems, was given to the girls. Raven Test was performed to exclude mental retardation [3].

Statistical analysis

The database, for both cases and controls, was developed and kept up-to-date using SPSS® software (SPSS Inc., Chicago, IL, USA). Descriptive and bivariate analysis was carried out using SPSS® software, instead Multivariate analysis was performed with the SAS® package, rel. 9.1.3 (SAS Institute Inc., Cary, NC, USA).
The descriptive analysis included the observed frequencies calculation with the respective percentages for each variable collected in the questionnaires, separately for girls with Turner Syndrome and for the control group. Moreover the crude odds ratios (ORs) estimate with confidence intervals and p-values was carried out. Categorical data are given as numbers with percentages and continuous data as medians with ranges. Then a bivariate analysis was performed to study the possible relations between couples of variables, for both cases and controls. Since data are independent, the Chi-Square Test was used to compare proportions in m*n tables (without the Yates correction) and the Fisher exact Test was selected for 2*2 tables. Only the statistically significant ($p < 0.05$) relations resulted for the group of cases and not for the control group are reported. Multivariate analysis was performed using a stepwise logistic regression analysis (significance level for entering = 0.15 and significance level for removing = 0.10) to identify those psychological, affective-relational, psychosocial factors that had a statistically significant correlation with Turner Syndrome. The considered variables, together with those about YSR (competences and problems), can be checked in Tables 1-V.

Results

Tables I and III show the distribution of cases and controls according to individual, familiar and clinical variables.
Table II indicates observed frequencies and the respective percentages for cases and controls, according to scholas-

TABLE I.
Distribution of cases and controls by ethnicity, cultural level, parental status, parents-child relationship, having brothers and/or sisters, relationship with brother and/or sisters and age interval [observed frequencies (%)], height and weight [median (range)] and crude odds ratio (OR) for case-control data with corresponding CI and p-value. *Distribuzione dei casi e dei controlli per etnia, livello culturale, stato genitoriale, rapporto genitori-bambino, fratria, rapporto con fratelli e sorelle, intervallo di età [frequenze osservate in percentuale], altezza e peso [mediana (intervallo)] e odds ratio grezzo (OR) per dati caso-controllo con rispettivi intervalli di confidenza e valori di p.*

		Cases (n = 47)	Controls (n = 47)	OR	CI	p-value
Ethnicity	Caucasian	47 (100.0)	47 (100.0)			
Cultural level	Low	12 (25.5)	14 (29.8)	1		
	Medium	19 (40.4)	20 (42.5)	0.925	0.755-1.462	0.9834
	Hight	16 (34.1)	13 (27.7)	0.764	0.431-1.368	0.8552
Parental status	Married	45 (95.7)	43 (91.5)	1		
	Separated/divorced	2 (4.3)	4 (8.5)	0.812	0.582-1.505	0.8630
Parents-child relationship	Good and serene (ref.)	30 (63.8)	28 (59.6)	1		
	Difficult	17 (36.2)	19 (40.4)	0.835	0.363-1.921	0.6717
Brothers and/or sisters	Brothers or sisters (ref.)	23 (48.9)	41 (87.2)	1		
	Only child	24 (51.1)	6 (12.8)	7.13	2.545-19.973	0.0002
Relationship with brothers and/or sisters	Negative	3 (13.0)	9 (22.0)	1		
	Quite good	16 (69.6)	18 (43.9)	2.667	0.613-11.598	0.6682
	Good	4 (17.4)	14 (34.1)	0.857	0.154-4.764	0.0401
Age interval	Yrs 15-19 (ref.)	31 (66.0)	35 (74.5)	1		
	Yrs 10-14	16 (34.0)	12 (25.5)	1.505	0.618-3.669	0.3680
Height (cm)		148 (125-163)	161 (145-180)	0.811*	0.744*-1.884*	< 0.0001
Weight (kg)		50 (30-75)	50 (60-69)	0.98*	0.937*-1.025*	0.3776

Ref.: reference category; CI: confidence intervals; *Crude odds ratios and 95% CIs for 1-unit increase.

tic and extra scholastic variables (VADO and YSR) and some personal characteristics for both cases and controls (VADO).

Table IV shows the comparison between the observed frequencies with the corresponding percentages of eight syndrome scales by Youth Self Report for cases and controls.

A B*ivariate Analysis* was performed through a Chi-Square Test (or a Fisher's exact Test for the relationship between two dichotomous variables) to study the possible relations between couples of variables, for both cases and controls. Given the aim of the research, they are below reported the relations of "parent-child relationships" (a) and of "psychological-psychiatric consultation" (b) with other variables. Only the statistically significant ($p < 0.05$) relations resulted for the group of cases and not for the control group are reported.

a. *Parents-child relationship* * look satisfaction
parents-child relationship * desire for change appearance
parents-child relationship * psychological/psychiatric consultations
parents-child relationship * feelings towards disease
parents-child relationship * anxious-depressive problems (YSR)
parents-child relationship * social problems (YSR)
parents-child relationship * problems of attention (YSR)
parents-child relationship * delinquent behaviour (YSR).

Girls presenting a bad parents-child relationship have a higher probability to present problems of attention, anxious-depressive aspects, difficulties to socialise and a delinquent behaviour. On the other hand girls with a serene relationship with their parents present a higher look satisfaction and consequently they don't express the desire to change physical appearance; moreover they present positive feelings towards disease and they don't need psychological/psychiatric consultations.

b. *Psychological/psychiatric consultations* * integration with the classmates
psychological/psychiatric consultations * self-fulfilment
psychological/psychiatric consultations * look satisfaction
psychological/psychiatric consultations * desire for change appearance
psychological/psychiatric consultations * feeling of difference compared with others
psychological/psychiatric consultations * anxious-depressive problems (YSR)
psychological/psychiatric consultations * social problems (YSR)

TABLE II.
Distribution of cases and controls according to school attended, favourite subject at school, school result, school motivation, integration with the classmates, extra-school relationships, feeling of difference compared with others and desire for change appearance [observed frequencies (%)] and crude odds ratio (OR) for case-control data with corresponding CI and p-value. *Distribuzione di casi e di controlli secondo la scuola, materia preferita, risultato scolastico, motivazione a scuola, integrazione in classe, rapporti extra-scolastici, sentimenti di diversità rispetto agli altri, desiderio di cambiare aspetto [frequenze osservate in percentuale], altezza e peso [mediana (intervallo)] e odds ratio grezzo (OR) per dati caso-controllo con rispettivi intervalli di confidenza e valori di p.*

		Cases (n = 47)	Controls (n = 47)	OR	CI	p-value
School attended	Primary school (ref.)	8 (17.0)	7 (14.9)	1		
	Middle school	9 (19.1)	3 (6.3)	2.625	0.502-13.725	0.1172
	High school	30 (63.9)	37 (78.8)	0.709	0.231-2.181	0.0908
Favourite subject	History and geography (ref.)	6 (12.8)	11 (23.5)	1		
	Italian	26 (55.3)	13 (27.6)	3.666	1.108-12.135	0.0075
	Foreign languages	8 (17.0)	10 (21.3)	1.467	0.376-5.723	0.9323
	Biological and scientific sciences	7 (14.9)	13 (27.6)	0.987	0.255-3.824	0.2837
Scholastic result	Not sufficient	0 (0.0)	5 (10.6)			
	Sufficient (ref.)	18 (38.3)	15 (31.9)	1		
	Good	21 (44.7)	21 (44.7)	1.111	0.334-2.079	0.6598
	Very good	8 (17.0)	6 (12.8)	0.833	0.315-3.921	0.8699
Scholastic motivation	Low (ref.)	6 (12.8)	8 (17.0)	1		
	Quite good	23 (48.9)	33 (70.2)	0.929	0.284-3.039	0.0883
	Excellent	18 (38.3)	6 (12.8)	4.000	0.981-16.302	0.0111
Integration with the classmates	Low (ref.)	9 (19.1)	5 (10.6)	1		
	Quite good	18 (38.3)	23 (48.9)	0.435	0.124-1.526	0.2095
	Excellent	20 (42.6)	19 (40.5)	0.585	0.166-2.063	0.7910
Extra scholastic relationships	No none (ref.)	11 (23.4)	3 (6.3)	1		
	Only females	30 (63.8)	28 (59.6)	0.292	0.074-1.158	0.8515
	Females and males	6 (12.8)	16 (34.1)	0.102	0.021-0.499	0.0050
Extra scholastic activities	Any activities (ref.)	11 (23.5)	7 (14.9)	1		
	Parish activities	3 (6.3)	6 (12.8)	0.318	0.059-1.705	0.9768
	Sport	28 (59.6)	25 (53.2)	1.713	0.240-2.121	0.9666
	Music	5 (10.6)	7 (14.9)	0.455	0.103-2.013	0.9722
	Other activities	0 (0.00)	2 (4.2)			
Sentimental relationship	Yes (ref.)	15 (31.9)	30 (63.8)	1		
	No	32 (68.1)	17 (36.2)	3.764	1.602-8.846	0.0024
Plan to have a family	Yes (ref.)	37 (78.7)	40 (85.1)	1		
	No	10 (21.3)	7 (14.9)	1.544	0.533-4.477	0.4235
Plan to have children	Yes (ref.)	31 (66.0)	32 (68.1)	1		
	No	16 (34.0)	15 (31.9)	1.101	0.466-2.603	0.8264
Self-fulfilment	Low (ref.)	18 (38.3)	19 (40.4)	1		
	Quite good	24 (51.1)	23 (49.0)	1.101	0.465-2.608	0.8799
	Good	5 (10.6)	5 (10.6)	1.056	0.261-4.268	0.9931
Look satisfaction	Low (ref.)	11 (23.5)	7 (14.9)1			
	Quite good	3 (6.3)	6 (12.8)	0.278	0.110-0.704	0.3896
	Good	28 (59.6)	25 (53.2)	0.179	0.038-1.833	0.1380
Feeling of difference	Yes (ref.)	20 (42.6)	22 (46.8)	1		
	No	27 (57.4)	25 (53.2)	1.188	0.526-2.681	0.6785
Desire for change	Yes (ref.)	39 (83.0)	29 (61.7)	1		
	No	8 (17.0)	18 (38.3)	0.331	0.126-0.865	0.0241

Ref.: reference category; CI: confidence intervals.

TABLE III.
Distribution of cases and controls by type of support, presence of NPI consultations, NPI effect, usefulness of services and pharmacological treatment [observed frequencies (%)]. *Distribuzione dei casi e dei controlli per tipo di sostegno, presenza di consultazioni NPI, effetto del NPI, utilità dei servizi e del trattamento farmacologico [frequenze osservate in percentuale].*

		Cases (n = 47)	Control (n = 47)
Support	Paediatric	37 (78.7)	12 (25.5)
	Familiar	7 (14.9)	2 (4.3)
	Psychological/psychiatric	3 (6.4)	1 (2.1)
	No support	0 (0.00)	32 (68.1)
Psychological/psychiatric consultations	Yes	29 (61.7)	20 (42.5)
	No	18 (38.3)	27 (57.5)
		Cases (n = 29)	Controls (n = 20)
NPI effect	Useful	17 (58.6)	12 (60.0)
	Useless	12 (41.4)	8 (40.0)
		Cases (n = 47)	Controls (n = 19
			28 missing values
Usefulness of service	Useful	30 (63.8)	10 (52.6)
	Useless	17 (36.2)	9 (47.4)
		Cases (n = 47)	Controls (n = 28)
			19 missing values
Pharmacological treatment	Yes	39 (83.0)	9 (32.1)
	No	8 (17.0)	19 (67.9)

Girls affected by Turner syndrome following psychological/psychiatric consultations seem to have a higher self-fulfilment and look satisfaction (consequently they don't express the desire to change physical appearance), to present a better integration with the classmates, to feel higher difference compared with others, to present borderline anxious-depressive aspects and difficulties to socialise compared to girls who are not following any psychological/psychiatric consultation.
Table V shows the results of the *Logistic Regression Analysis* which was adopted to compare patients with Turner syndrome and controls, using Sas System: the seven variables entered in the model are height, weight, favorite scholastic subject, integration with the classmates, brothers and/or sisters, social withdrawal and aggressive behavior.

Discussion

School performances and competences

About scholastic performance there are discordant data in literature. Some studies underlined how Turner syndrome patients have worse performance with respect to the "healthy" girls [4] and a frequency of mental retardation in Turner patients significantly higher than that observed in general population, even if it is not affected by statural impairment [5].
On the other side other studies underlined that girls affected by Turner syndrome obtain good results both at school and at work, have a higher education compared to the national average, attending University more frequently then the general population [6 7]. This data is confirmed by those studies that state there is no increased incidence of either severe or moderate mental retardation; intelligence is similar regardless of karyotype or somatic stigmata, and the characteristic cognitive deficit is similar among both children and young adults affected by Turner syndrome [8 9].
In our case study the pre-adolescent show an adequate overall performance, which ends to be reduced in mid-late adolescence [10-14 years: 13/16 (81%) referred good and very good results; 15-19 years: 16/31 (51%) referred good and very good results; p-value = 0.015]. As far as the scholastic result, without considering age, Table II shows there are no significant differences between cases and controls, more then half of girls referring a good and very good scholastic results. On the other hand what has been detached about school motivation is different, resulting Turner syndrome more motivated then controls; this data suggest a Turner girls' bigger investment on school.
According to literature which shows that girls affected by Turner syndrome have more difficulties in mathematical and arithmetical subjects [10-12], our data, as reported in Table II, confirm that only 10.6% attend a scientific High school; the majority attend a humanistic, artistic or social pedagogical High school. These girls do not like scientific subjects but prefer literature; instead in the control group

the subject preference is more homogenous and the frequencies are more uniformly distributed (Table II).

About neuropsychological performances, a study of Lagrou et al. [13] revealed elevated mean score at the subscales of attention problems in women affected by Turner syndrome compared to a healthy control group. Another recent study by Russel et al. [14] emphasized a high prevalence of ADHD in girls affected by Turner syndrome compared to the general population. On the contrary, in our study girls affected by Turner syndrome have less problems of attention than controls (Table IV). Considering that among those Turner girls affected by attentive disorder there is a bigger percentage of difficulties to socialize and bad relationships with parents ($p < 0.05$), we can suppose that the attentive disorder is correlated to an emotional problem rather then being a specific cognitive deficit.

Results reported in Table II show that there are no differences in extra scholastic activities in Turner compared with controls. In our sample 59.6% of Turner syndrome girls practice a sport outside the school; this could suggests that girls affected by Turner syndrome are not significantly limited by physical abnormalities and differences with healthy girls (height and weight f.i.) in activities. However, a study by Ross et al. in 1998 [15], which evaluated the athletic capacity of Turner syndrome patients, showed how these girls have problems in performing gym exercises and in coordinating their movements.

Self image, psycho-emotional and relational functioning

With regards to self image and perception, some Authors had emphasized the fact that, because of their height, these girls are often treated as if they were younger than their age, this causing problems of self esteem [16]; moreover recent study findings [17] show that girls with Turner

TABLE IV.
Distribution of cases and controls by social withdrawal, somatic complaint, anxious-depressive symptoms, difficulties to socialize, thought problems, problems of attention, delinquent behaviour and aggressive behaviour [observed frequencies (%)] and crude odds ratio (OR) for case-control data with corresponding CI and p-value. *Distribuzione dei casi e dei controlli per ritiro sociale, lamentele somatiche, sintomi ansioso-depressivi, problemi di socializzare, problemi ideativi, problemi attenzionali, comportamento delinquenziale, comportamento aggressivo [frequenze osservate in percentuale], altezza e peso [mediana (intervallo)] e odds ratio grezzo (OR) per dati caso-controllo con rispettivi intervalli di confidenza e valori di p.*

		Cases (n = 47)	Controls (n = 47)	OR	CI	p-value
Social withdrawal	Normal (ref.)	36 (76.6)	43 (91.5)	1		
	Borderline	3 (6.4)	3 (6.4)	1.194	0.227-6.285	0.3320
	Pathological	8 (17.0)	1 (2.1)	9.556	1.141-80.046	0.0576
Somatic complaint	Normal (ref.)	38 (80.8)	37 (78.7)	1		
	Borderline	6 (12.8)	6 (12.8)	0.974	0.288-3.294	0.8525
	Pathological	3 (6.4)	4 (8.5)	0.730	0.153-3.489	0.7151
Anxious-depressive symptoms	Normal (ref.)	28 (59.6)	33 (70.2)	1		
	Borderline	12 (25.3)	9 (19.2)	1.571	0.578-4.273	0.7113
	Pathological	7 (14.9)	5 (10.6)	1.650	0.471-5.778	0.6670
Social problems	Normal (ref.)	25 (53.2)	41 (87.2)	1		
	Borderline	11 (23.4)	4 (8.5)	4.510	1.295-15.706	0.5670
	Pathological	11 (23.4)	2 (4.3)	9.017	1.845-44.061	0.0822
Thought problems	Normal (ref.)	42 (89.4)	39 (83.0)	1		
	Borderline	1 (2.1)	5 (10.6)	0.186	0.021-1.661	0.1245
	Pathological	4 (8.5)	3 (6.4)	1.238	0.260-5.887	0.2648
Problems of attention	Normal (ref.)	36 (74.5)	29 (61.7)	1		
	Borderline	7 (14.9)	16 (34.0)	0.363	0.131-1.001	0.0285
	Pathological	5 (10.6)	2 (4.3)	2.071	0.374-11.477	0.1583
Delinquent behaviour	Normal (ref.)	36 (76.6)	34 (72.3)	1		
	Borderline	9 (19.1)	10 (21.3)	0.850	0.308-2.346	0.9168
	Pathological	2 (4.3)	3 (6.4)	0.630	0.099-4.003	0.6878
Aggressive behaviour	Normal (ref.)	36 (76.6)	28 (59.6)	1		
	Borderline	6 (12.8)	10 (21.3)	0.467	0.151-1.439	0.5682
	Pathological	5 (10.6)	9 (19.1)	0.432	0.130-1.434	0.4654

Ref.: reference category; CI: confidence intervals.

syndrome have lower self esteem and higher state anxiety level than healthy controls. Our sample does not differ significantly from the controls about self fulfillment, look satisfaction and filling of difference compared with others; besides there is a significant difference about what concerns the desire of change appearance (which means that satisfaction respect to self image is not conflict free) (Table II). As said in literature, Turner syndrome girls tend to be overweight [18] and this data is also confirmed in our sample: 21 patients (44.3%) are over the 51-70 percentile and 4 patients (8.5%) are over the 75 percentile of the growth weight scale. The BMI in 19.1% of cases is between 25 and 29.9 and in 6.4% of the cases is between 30 and 34.9. About that, it is to be underlined the result reported in table 5, which shows that the desire for change appearance is more connected with weight than with height. In fact it seems that once variable weight entered in the logistic regression model, the difference between cases and controls about the covariate "desire for change appearance" is already explained. This means girls affected by Turner syndrome would like to change appearance because they feel different in weight rather than in height. With regards to this result, a study by Lagrou [13] about psychosocial functioning and self perception in young Turner adults, concludes about the importance of a follow up of Turner adults which should not neglect the problem of overweight and associated psychosocial consequences.

Observing the logistic regression analysis outputs (Table V), obtained from Sas System, we can notice that "height" is the first variable entering in the model: it already predicts 92.5% of the whole model which aim is to find out the main covariates associated with the dependent variable "to be Turner or not". Once variable "height" entered in the first step of the analysis, the Chi-Square significances were recalculated for the remaining covariates. In the second step variables "weight", "integration with the classmates" and "aggressive behaviour" find a new and higher discriminating power; whereas variables as "scholastic motivation" and "extra scholastic relationships" loose significance because they are connected with height. The discriminating power of these last two variables, evident only if they are taken alone, decreases a lot when height is already entered in the model.

This factor can be explained considering the variable age: in a standard population of girls with age 10-19 (as it could be the control group) height is very associated with age: following a simple general logic, older you are, more likely you are higher than a younger girl. This reasoning can't be totally applied to girls affected by Turner syndrome, because they grow less evidently than normal girls. From data in Table II we observe Turner girls are more motivated at school and they less easily form a friendship (if they make friends with someone, it's more likely that she is a female). This aspect is evident also in the control group, but only in the age interval 10-14 years. Instead for girls belonging to the age interval 15-19 years, the scholastic motivation decreases and the attention is also addressed to males. This underlines a difference between cases and controls only in the age interval 15-19, which also represents the group of higher normal girls. Taken the two groups after-height (and after-age) it seems there is an overall difference between Turner girls and controls about the variables "scholastic motivation" and "extra scholastic relationships", but this association disappears once "height" enters in the logistic regression model as explicative variable, because it explains also the difference between cases and controls concerning "scholastic motivation" and "extra scholastic relationship". Table V shows that, on the whole, girls affected by Turner

TABLE V.
Results of Logistic Regression Analysis for girls affected by Turner syndrome (cases) compared to controls. *Risultati del "Logist Regression Analysis" per ragazze affette da sindrome di Turner (casi), paragonate ai controlli.*

	Maximum likelihood estimate	Standard error	p-value	OR	95% CI
Height (cm)	-0,517	0,146	0,0004	0,596	0,448-0,794
Weight (kg)	0,304	0,102	0,0028	1,356	1,110-1,655
Favourite subject (ref. Italian)	2,677	1,320	0,0425	14,548	1,094-193,397
Integration with the classmates (ref. low integration)	4,243	2,012	0,035	69,597	1,349-> 999,999
Brothers and/or sisters (ref. only child)	3,434	1,278	0,0072	30,996	2,531-379,552
Social withdrawal (ref. normal)	-5,519	2,608	0,0343	0,004	< 0,001-0,665
Aggressive behaviour (ref. normal)	3,332	1,562	0,033	27,985	1,310-598,071

Likelihood ratio test: $p < 0,0001$; Ref.: reference category; OR: odds ratio; CI: confidence intervals.

syndrome compared with controls have a higher weight and a lower height, prefer human subjects at school, have a lower integration with the classmates, are more likely only child, have more social and withdrawal problems and less aggressive behaviour. Moreover, Turner girls are more motivated at school and they have less extra-scholastic relations too, especially in age interval 15-19 years. Our results seem to suggest that Turner girls feel a sense of inferiority with respect to healthy girls of the same age group. All of them would like to be physically different although they believe that appearance is not as important as the way they are and feel. Results reported in Table II suggest that on one hand Turner girls think of themselves as controls do (the both of the groups state they like themselves, they are satisfied with themselves and they don't feel different from the others), but on the other they feel about themselves differently, being the percentage of Turner girls who would change their appearance statistically bigger then the relative percentage of controls (Table II).

With regards to self image and perception there is also to mention the pharmacological therapy (the growth hormone and the use of estroprogestinic) which is a is consolidated intervention, positively influencing the concept of self-esteem and behavior of girls affected by the Turner syndrome [19 20] In our study this data is confirmed by the scholastic motivation ($p = 0.02$) and by a better integration with school friends in girls following the therapy ($p = 0.03$).

Studies about emotional and relational functioning of Turner syndrome girls evidenced that the social integration is low [5 6 21]. On the contrary, in our sample almost of girls are well integrated with school friends, they attend parish activities, have many friends and practice an extra scholastic activity (Table II). About the desire too meet other Turner girls of the 36 girls (76.6%) who are interested in, the majority (15 = 41.6%) is under 14 years of age. This data confirm the literature which reported that the desire to meet other girls affected by Turner syndrome is greater in preadolescent girls with respect to older girls, who refused the contact and the confrontation with other girls sharing their same condition [22].

At the question "who would you like to live with", all the girls answered that they wished to live alone or with friends. Another time it is suggested a gap between desire and reality: in fact, our sample confirms the poor self-government of Turner girls: almost all the girls of the control group work (17% between 10 an 14 years and 46% between 15 and 19 years), while only few of those affected by Turner syndrome do the same (6% between 10 an 14 years and 32% between 15 and 19 years).

It is confirmed by literature that Turner syndrome girls show wish to have a family and children with a higher incidence compared to healthy girls and this also reflect choices related to the type of school and job [21]. Wishes of these girls seem in contradiction with reality as it resulted from literature which evidenced that these girls tend to stay with their original family also as adults, only few get married or live on their own, and from this study where, even if Turner desire to have a family and children like controls, in fact there is a significant minor percentage of Turner, compared to controls, that have an actual sentimental relationship (31.9% *vs.* 63.8%) and mixed gender friends rather then only females (12.8% *vs.* 34.1%) (Table II). This data reflect the fact that the sense of identity and the image of being sexually mature are fundamental at this age and sexuality is one of the most important steps of evolution; in this respect girls affected by Turner syndrome have to deal with problems of sterility and therefore this dimension is difficult to accept.

In this study a particular attention was paid to the Turner girls' relationship with parents. More than half of the girls reported they had a difficult relationship with their parents (Table I). This conflict not only expressed a research of independency typical of this age, but can also be seen as a reaction to the over protective attitude of parents caused by the disease. In literature this parents' behavior is seen on one hand as a depressive and disappointed attitude and on the other as aggressiveness [16]. In fact, it appears extremely difficult to elaborate the feeling of loss and the feeling of guilt linked to acceptance about having an imperfect daughter. Some parents are too overprotective [21 22], and this prevents a psychological growth in the girls. Besides, this can cause a behavioral regression which affects the development of autonomy in their daughters [23 24].

Parents' fear, shame and guilt about illness could be a partial interpretation of our result reported in Tables I and V which shows as, compared to controls, Turner girls are more frequently only daughter. In our sample the relationship with parents influence different aspects: the girls presenting a bad parents-child relationship have a higher probability to present problems of attention, anxious-depressive aspects, difficulties to socialise and a delinquent behaviour. On the other hand girls with a serene relationship with their parents present a higher look satisfaction and consequently they don't express the desire to change physical appearance; moreover they present positive feelings towards disease and they don't need psychological/psychiatric consultations (see results from bivariate analysis in results section).

Psychopathology and psychological-psychiatric intervention

In literature there are different studies that deeply investigated the psychopathological comorbility in Turner syndrome patients: some Authors emphasized an association

between Turner syndrome and autism [25 26]; instead others emphasized an association with schizophrenia [27 28], with depression [29], with anorexia [30] and with anxiety and social withdrawal [31].

The evaluation of the YSR questionnaire of our patients (Table IV) confirms the most of related literature according to which girls affected by Turner syndrome seems to be more socially withdrawn, anxious and depressed and show social problems (internalized discomfort) with respect to normal population; they have less frequently attention problems and an aggressive behavior (externalized discomfort) [16-18 32 35]. Specifically, bivariate analysis regarding YSR, indicates that girls presenting anxious-depressive problems have a higher probability to have a bad parents-child relationship.

As regards to therapeutic approach to the psychological and psychopathological aspects, some Authors advice Turner syndrome patients to talk about their problems and discomfort during the meetings: actually counseling individuals with Turner syndrome and their families about the need to carefully develop and nurture social skills and relationships may prove useful in advancing the social adaptation of these young women [36 37].

Results from bivariate analysis confirm that psychological/psychiatric consultation is overall because of internalising problems (anxious-depressive aspects and difficulties to socialise) and that psychotherapy implies benefits: actually girls affected by Turner syndrome following psychological/psychiatric consultations seem to have a higher self-fulfilment and look satisfaction (consequently they don't express the desire to change physical appearance), to present a better integration with the classmates compared to girls who are not following any psychological/psychiatric consultation. This data support the idea that a comprehensive treatment of girls with Turner syndrome should included psycho-behavioural interventions in addition to traditional medical therapies.

Conclusions

According to the aims of the study and considering its results, we can state that it is relevant to assure to girls affected by Turner syndrome a multidisciplinary intervention which contemplates a psychological/psychiatric support, because of the fragile self perception, the difficulty in affective-relational area and the risk of psychopathology (Turner syndrome girls have a higher risk to develop a psychopathological morbidity, internalising problems overall). It is relevant to assure to these girls a multidisciplinary intervention which seems particularly to favour social integration, better self-fulfilment and look satisfaction. This support should take into account the whole family given that the relation of dependency which links girls affected by Turner syndrome to their parents conditions different psychosocial aspects of their life. In this sense a psychological space where daughter and parents can elaborate the problems due to the disease is always important, also considering the adolescent phase of the patients, during which the process of separation and individuation from the parental figures is one of the developmental target to be reached.

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