



Social Functioning of Women with Turner Syndrome

Research results & discussion

ABSTRACT: Zadrożna Ilona, *Social Functioning of Women with Turner Syndrome. Research results & discussion* [Społeczne funkcjonowanie kobiet z zespołem Turnera. Wyniki badań i dyskusja]. Interdyscyplinarne Konteksty Pedagogiki Specjalnej, nr 1, Poznań 2013. Pp. 121-137. Adam Mickiewicz University Press. ISBN 978-83-232-2539-3.

The article concerns social functioning of women with Turner syndrome, focusing particularly on their family life (relations with parents, siblings, partner and children), and relations with others (friends, acquaintances, workmates and members of TS support organizations). The author also tries to find correlations between growth hormone treatment, sex hormones treatment, age of TS diagnosis, karyotype and social functioning of TS women. Turner Syndrome is a quite common (1 in every 2500 live female births) human genetic disorder which affects only females. Females with TS lack all or part of one of two sex X chromosomes. The phenotype of TS women includes short stature and ovarian failure (which usually causes infertility) specific anatomic abnormalities (such as a short neck with a webbed appearance, a low hairline at the back of the neck, and low-set ears) and characteristic neurocognitive profile, which usually does not include mental retardation. The treatment of TS girls includes growth hormone and sex hormones therapy. 71 women took part in the study (30 from Poland and 41 from other countries - USA, Australia and United Kingdom). Researches indicated that generally women with TS showed relatively good functioning in relations in family of origin (with parents and siblings) and in social environment (quite high professional activity, good functioning in relations with friends and acquaintances). Relations with parents, especially with mothers, were usually very close, but often revealed overprotective parents' attitude

towards TS women (even in their adult life). Relations with fathers were slightly worse, more often revealing lack of fathers' acceptance towards TS women. Relations with siblings were rather close, but again revealed overprotective siblings' attitude towards TS sisters. Researches also revealed relatively weak functioning of TS women in relations in family of procreation (with partner and children).

KEY WORDS: social functioning, women, Turner syndrome

Turner Syndrome (TS) is a common, human genetic disorder (one in every 2500 live female births) that affects only females. Females with TS lack all or part of one or both X chromosomes. The phenotype of TS women includes short stature, lack of sexual development and ovarian failure (which usually causes infertility), specific anatomic abnormalities (such as a short neck with a webbed appearance, a low hairline at the back of the neck, and low-set ears) and a characteristic neurocognitive profile, which usually does not include mental retardation.

Turner Syndrome occurs when one sex chromosome is deleted, so that instead of the normal 46 chromosomes, of which two are sex chromosomes (XX in females and XY in males), the chromosomal complement is 45,X. In genetic terms, these patients are neither male nor female because the second, sex-determining chromosome is absent. However, phenotypically, affected individuals develop as females because there is no Y chromosome to direct the fetal gonads to the male configuration. Clinically, patients with Turner Syndrome are short and they have a small chin, prominent folds of skin at the inner corners of the eyes (epicanthal folds), low-set ears, a webbed neck, and a shield-like chest. Individuals with Turner Syndrome also have an increased incidence of anomalies of the heart and large blood vessels. Both the internal and the external genitalia are infantile, and the ovaries are only "streaks" of connective tissue.¹

¹ Robert D. Utiger, *Encycloaepedia Brittanica online*, <<http://www.britannica.com/EBchecked/topic/610330/Turner-syndrome>>.

The survey concerned social functioning of women with Turner Syndrome, focusing particularly on their family life (relationships with parents, siblings, partner and children) and relations with others (friends, acquaintances, workmates and members of TS support organizations). The author also tries to find correlations between growth hormone treatment, sex hormone treatment, age at TS diagnosis, karyotype and social functioning of TS women.

Seventy-one women with Turner Syndrome participated in the study; 30 from Poland [Polish group], and 41 from western countries including the USA, Australia and the United Kingdom [western group]). Most respondents had the characteristic physical TS features: a typical respondent was shorter than the average-height woman from the general population, had higher BMI (body mass index - particularly in the group of women from the western group), and had fewer secondary sex characteristics. Most of them were treated with growth hormone and hormone-replacement therapy (HRT). Most common karyotypes, as in other research, were 45,X (monosomic) and 45,X/46,XX (mosaic).² In general, the survey revealed good functioning of women with TS in relationships in the family of origin, which consists of parents and siblings (women from the Polish group had slightly better relationships), and quite weak functioning in the family of procreation, which consists of partner and children (if there were any). The survey revealed that most women were working (particularly women from the western group) and had few or no problems engaging in contacts with others and maintaining them.

The survey was fielded from May to September 2009, predominantly via an Internet-based³ questionnaire consisting of 42 ques-

² Monosomic or monosomy - denotes the presence of only one chromosome X (instead of the typical two in women) from a pair - 45,X; mosaic or mosaicism denotes the presence of two or more populations of cells with different genotypes i.e. the most common mosaic karyotype 45,X/46,XX denotes the presence of two populations of cells: 45,X and 46,XX in one individual.

³ Some questionnaires were sent by post and some were filled in personally at the Turner Syndrome Support Society meeting.

tions (mostly multiple choice questions) and four attachments (Attachment Styles Questionnaire by M. Plopa, Questionnaire of Retrospective Evaluation of Parental Attitudes of Mothers and Fathers by M. Plopa, and Intimacy, Passion and Commitment Scales on the basis of B. Wojciszke's triangular typology of love). The survey was conducted in Polish and English (back translation was used to translate the questionnaire into English). A pre-test survey was conducted to verify that all survey functionality worked correctly.

Results

Relations in the family of origin

Relations with parents

Most of the women had grown up in families where both mother and father accepted, or accepted and overprotected the TS daughter. Most respondents accepted their parents and felt accepted by them. Additionally, about 50% of women felt overprotected by their mothers/fathers. In general, TS women had slightly better relations with their mothers than with their fathers (fathers were more often rejected and less often accepted by TS daughters).

The survey revealed lack of significant difference in the relationships of women who had been treated with GH and those who had not been treated with GH⁴ with their mothers, and better relationships with fathers in the GH-untreated women (they were more often accepted and less often rejected by their fathers). The women who were the tallest and had a normal (or slightly lower) Body Mass Index had the best relationships with parents (especially with fathers). The shortest and the most obese (those of the highest BMI) women had the worst relationships with parents.

⁴ For the purpose of this study I will use the terms: "GH-treated women" and "GH-untreated women".

The survey also revealed lack of significant difference in relationships of HRT⁵-treated and HRT-untreated women with their fathers and better relationships with mothers in the HRT-untreated women from other countries. Women with the most visible sex characteristics had the best relationships with both mothers and fathers. In comparison, women with no visible sex characters had the worst relationships.

Parents tended to be most protective of girls diagnosed earliest (before they turned 7) but those girls also had the best relations with both mothers and fathers. The women diagnosed relatively late (as teenagers or adults) definitely had the worst relationships with fathers.

Relations with siblings

Most TS women had good relationships with their siblings and were accepted by them. Twenty percent of TS women felt rejected by their siblings. Research revealed a few interesting facts, such as:

- GH-treated women were much more competitive with their siblings, but were also less often rejected by their siblings than the GH-untreated women.
- Women who were diagnosed earliest were much more likely to compete with their siblings in comparison to women who were diagnosed later in life.
- In the Polish group, the shortest women had the worst relationships with their siblings, a correlation not noticed in the western group. In both groups, the shortest women were most often overprotected by their siblings.

Relations in the family of procreation

Relations with partners

The survey revealed that less than 1/3 of respondents had partners. The majority of TS women never had a sexual partner (60% of

⁵ HRT = hormone replacement therapy; for the purpose of this study I will use the terms: "HRT-treated women" and "HRT-untreated women".

the Polish group and 45% of the western group). The criteria for choosing the partner were the same as in the general population of women (love, fascination, partner's reliability) but – in contrast to the general population of women – most of the TS women mentioned the most important criteria, which was their partner's acceptance of them.

Most of the existing relationships with partners revealed a low level of passion and, at the same time, a very high level of commitment,⁶ inadequate to the length of the relationship with the partner (so, in general, respondents were functioning poorly in sexual relations with partners, but put a lot of effort into maintaining the relationship itself, even if it was only a short-term relationship).

The other interesting thing was that the anxious/ambivalent attachment style occurred much more often than in the general population of women (as adults, people having anxious/ambivalent attachment style are worried that they cannot trust their partner and that they may be rejected, and tend to think that they are not accepted by them (Hazan & Shaver, 1987, in: Wojciszke, 2006). The research revealed the need to support girls and women with TS concerning self-acceptance and “healthy” functioning in relationships with partners.

Both GH treatment and hormone replacement therapy revealed a positive correlation with functioning in relationships with partners: the GH/HTZ-treated women trusted their partners and felt accepted by them more often than the GH/HTZ-untreated women; moreover, TS women lacking visible sex characteristics (such as properly developed breasts or a “feminine” silhouette) looking more support in their partners and felt unaccepted by them more often. They also showed anxious/ambivalent and avoidant attach-

⁶ In Sternberg's Triangular theory of love, there are three main aspects of love: passion (sexual excitement, infatuation), intimacy (closeness, support and sharing) and commitment (the decision to take steps to maintain the relationship). One can have none, any one, any two, or all three of the aspects in a relationship. Although there are actually eight types of relationships that the theory assumes, the most desirable kind of love – consummate love – is the combination of all three aspects.

ment style in relationships with their partners much more frequently, and tended to function poorly in sexual relationships in comparison to TS women with one or two visible sex characteristics.

HRT-untreated women lacking visible sex characters tend to build relations characterized by a low level of passion and intimacy and, at the same time, a very high and often inadequate level of commitment.

Age at which the women had been diagnosed with TS seemed to have some influence on their functioning in relationships: the women diagnosed earliest (before they turned 7) had the best relationships, while those diagnosed as adolescents and adults definitely had the worst (women diagnosed as adolescents also functioned the worst in sexual life).

The research also revealed slightly better functioning in relations with partners in women with mosaic karyotype 45,X/46,XX (with normal line) compared to women with classic monosomy 45,X.

Relations with children

Only three respondents had children (due to typical for TS problems with fertility and/or lack of partner), and none of them were biological mothers (children had been adopted).

Relations beyond family

Relations in TS support groups

Only slightly more than 40% of the respondents were members of TS support groups – most often the women diagnosed earliest (in early childhood), and latest (in adulthood). For most of them, this was a place to initiate informal relationships with other members. Neither GH therapy nor HRT showed a significant correlation with belonging to TS support group.

Work and relations with co-workers

The survey revealed a very optimistic picture of TS women as employees, as opposed to what had been thought in previous publications: more than 60% of TS women had been working (slightly more in western than in the Polish group). The HTZ-untreated women and those with less visible sex characteristics (such as average size breasts or more feminine silhouette) were more often unemployed than the HTZ-treated and more “feminine” women (those of more visible sex characters). The shortest, HTZ-untreated women and those of the higher BMI tended to have closer, more friendly and less formal relationships with co-workers.

Relations with friends and acquaintances

Although the majority of previous publications revealed weak and quite limited functioning of women with TS in their “social life” (this term will be used to describe the relationships with friends and acquaintances) (compare with Siegel, Clopper, Stabler, 1998; Suzigan et al., 2004; Carel et al., 2005; Łącka, Ławniczak, w: Twardowski (red.), 2006), the survey seemed to give very optimistic results in this issue:

- 75% of respondents admitted to having frequent contact with their friends/acquaintances;
- although a majority of previous publications described women with TS as avoiding interpersonal relations (e.g. Bondy, 2008), about 50% of respondents declared being extrovert (being energized by being around other people, enjoying social situations, and being surrounded by other people);
- 75% of respondents were satisfied with their social life;
- the vast majority of respondents claimed to have a friend.

In turn, respondents very rarely admitted initiating contacts with others (25% of respondents initiated them rarely or occasionally). Moreover, in the Polish group half of respondents (!) based the choice of their friends on the level at which they were accepted by

them (this pattern was not so obvious in the western group, where only 20% of respondents took this criterion into consideration when choosing friends).

The women diagnosed earliest functioned best in contacts with others (acquaintances). In turn, those diagnosed in adolescence showed the weakest functioning in this domain.

Discussion

*Girls with TS should be diagnosed⁷ as soon as possible.*⁸ Most women had been diagnosed relatively late – over 1/3 of them had been diagnosed between the age of 12 and 18 (which is considered as the usual appearance of puberty) or even later! The need of early diagnosis is explained not only by medical reasons (such as detection and early treatment of some congenital disorders, or early and adequate management of GH and HRT treatment in young TS girls), but also by psychological and social issues: women diagnosed earliest (before they turned 7) functioned best in all tested areas (in relations with parents, siblings, their partners and friends) in comparison with women diagnosed later in life (in late childhood, adolescence and adulthood).

Growth Hormone therapy should be implemented in the optimal and sensitive period in life in order to achieve the best effects (proper adult height⁹). The validity of GH therapy is obvious and confirmed by many medical researches. This study revealed that it is also effective from the social point of view: GH-treated women proved to function

⁷ Some characteristic physical features allow to identify Turner Syndrome as early as in utero or shortly after birth.

⁸ Statistics show that although the age of TS diagnosis constantly lowers, some girls/women are still diagnosed too late – in 1982 about 33% of TS girls had been diagnosed as late as in adolescence (Hall, Sybert, Williamson et al., 1982), in 1991 – about 45% of TS girls had been diagnosed after they had turned 12, and in 2003 – 48% of them had been diagnosed aged 1–12 (Massa, Verlinde, De Schepper et al., 2005).

⁹ The average woman with TS who has not been treated with GH is about 23 cm shorter than the average woman from the general population.

better in close relationships with others – partners and friends, than women who had not been treated with GH. Relationships with mothers and siblings, and functioning in support groups, sexual and social life were similar in both groups (GH-treated and GH-untreated). At the same time, the tallest women functioned best in relations with parents, siblings, and co-workers and in sexual life in comparison to shorter and especially to the shortest women. There are some discussions concerning side effects of GH-treatment but most doctors are inclined to think that the benefits of such treatment (both medical and psychological) significantly outweigh those undesirable effects.

*Hormone replacement therapy should be implemented at the optimal age*¹⁰ in order to achieve induction of puberty and development of sex characteristics at an appropriate age. This study revealed that HRT-treated women functioned better in relationships with partners (also in sexual life), in contacts in support groups, in working life, and in relationships with friends than women who had not been treated with the HRT. Moreover, the women with more visible sex characteristics¹¹ (in most cases the ones treated with HRT) functioned better in relationships within the family of origin (with parents and siblings), relationships (including sexual life), contacts in support groups and in working life in comparison to women lacking in visible sex characteristics. Thus, Hormone Replacement Therapy seems to have an unequivocally positive influence on quality of life for women with TS.

Body Mass Index (BMI)¹² has been proved to be higher than usual in women with TS in many studies. Thus, *it is very important that all possible factors (including growth hormone therapy) which help to*

¹⁰ Newest research (Bondy et al., 2007) recommend implementing hormone replacement therapy at the age of 12.

¹¹ Two secondary sex characteristics have been taken into consideration in this study: breast development and feminine silhouette; development of pubic hair has not been considered as the factor potentially affecting social functioning as: (1) it is not “socially visible”, and (2) some research indicates that development of pubic hair is normal and does not differ from the general pattern.

¹² *Body mass index*, a standardized estimate of an individual’s relative body fat calculated from height and weight; the formula for calculating BMI is weight in kilograms (kg) divided by height in meters (m) squared.

*maintain accurate BMI, are implemented, i.e. relevant dose of physical activity, relevant diet, "healthy" lifestyle (dietician's advice may be helpful).*¹³ The study revealed that women of normal (or lower) BMI functioned better in many aspects including relationships in family of origin (with parents and siblings), contacts in support groups and with acquaintances. Additionally, women of normal/lower BMI from the Polish group admitted to having more friends and better relationships with them than did women of higher BMI (overweight or obese).

In relationships with fathers, *there is a great need for more obvious paternal participation in the bringing up/ treatment process of their daughters with TS, especially for those diagnosed in adolescence.* The women diagnosed in adolescence functioned definitely worst in relationships with partners and in sexual life in adulthood, which could be caused/affected by (mostly) undesirable relations with fathers in adolescence. A father's "presence" and the quality of his relationship with his daughter is very meaningful, especially in her adolescence. The relationship with her father simply affects a daughter's future functioning as a partner (wife) and mother. It also affects a daughter's self-esteem, feeling feminine and sexually attractive to the opposite sex, and the main factor in all of this is the father's acceptance of the daughter. Experiencing this acceptance is most important in adolescence, as this period of life has a lot to do with forming self-esteem and self-acceptance. Providing psychological support for fathers and daughters, especially for those diagnosed in adolescence, might provide desirable improvement in father-daughter relationships.

Hormone Replacement Therapy as well as the Growth Hormone Therapy (the need for daily intake of hormones (both oral and injected)¹⁴) *might be a psychological problem not only for TS girls but also for their parents/siblings.* Women who took HRT seemed to function much worse in their relationships with mothers (in the Western

¹³ Additionally, it helps prevent osteoporosis and diabetes (type II); as the TS women are much more likely than the women from the general population to suffer from these at any stage of their lives.

¹⁴ Daily GH injections may be considered as unfair and leading to pain and fear, especially in young girls who usually have the jabs done by their parents.

group) and with siblings (both groups). In turn, those treated with GH seemed to function worse in relations with fathers (both groups). Therefore, the parents of TS girls should be trained to do GH injections as well as provided with optimal psychological/educational support (the information on how to explain the need of daily injections to the youngest TS girls should be also given).

TS girls as well as their parents should be provided with relevant psychological support, especially at the time of the girl's diagnosis and shortly thereafter, no matter the age at which she was diagnosed. Diagnosis of TS usually modifies the relationships within the family and the way it normally functions; thus, it is vital to communicate it in the appropriate way¹⁵: having found out that their daughter suffers from the genetic disorder, parents are often confused and shocked, as they expected a healthy child which takes after them. In turn, they get the baby who doesn't take after any of them and usually has a lot of health issues. So the parents need to go through a specific kind of "bereavement" – they must part with the picture of the ideal daughter they dreamed of, and accept her as she is. The survey revealed that the shortest women, those lacking sex characteristics, and overweight or obese, had much worse relationships with parents than the women of more "socially accepted" appearance. Surprisingly, the karyotype 45,X (theoretically the one involving most characteristic physical TS features) didn't seem to have any impact on relationships with parents.

TS girls should be supported in learning self-acceptance and the ability to develop "healthy" functioning in relationships. Most existing relationships with partners revealed a very high level of commitment, disproportionate to the length of the relationship (so, in general, respondents functioned poorly in sexual relationships with partners, but put a lot of effort into maintaining the relationship itself,

¹⁵ "Model informing procedure" (Cunningham, Morgan and McGucken) says that parents should (1) be informed about the child's diagnosis as soon as possible, (2) both of them should be informed, and (3) the efforts should be made to give them as detailed information as possible (including information about different support possibilities [psychological/educational/medical support]).

even if it was only a short-term relationship). Also the anxious/ambivalent attachment style occurred much more often than in the general population of women.

Efforts should be made to help¹⁶ *women with TS realize themselves in motherhood as one of the developmental tasks of early adulthood* (Havighurst R., 1971) and an important factor affecting the quality of their lives (compare with the “Integrated Support for TS Affected Persons” diagram created by M. Ławniczak).¹⁷ This particularly refers to Poland (Polish group of respondents) where the adoption procedures are sometimes very long and oocyte donation (which is the most common way of having a baby for infertile couples) is not legally sanctioned.

*Efforts should be made to increase the number of TS girls/women becoming members of support groups. Those groups are an important element of an integrated support system for women with TS, supporting them in improving the quality of their lives.*¹⁸ It should particularly refer to the older girls with TS (in their late childhood and adolescence), as the survey revealed that they were least often the members of support groups and at the same time need support the most. The survey revealed that the support group members were mostly women diagnosed earliest (undoubtedly thanks to their parents) and diagnosed latest – often in their adulthood when they felt the need of support themselves – in that case, it might have been too late for relevant educational, emotional or psychological support). Thus effort should be made to spread information about Turner Syndrome itself, as well as about support groups for those affected by TS in many ways. A good example would be two Polish guidebooks (one edited by Prof K. Łącka, the other – by A. Wiśniewski, M.D.), issued in 2009, in-

¹⁶ Research show that only 2-5% of TS women are able to get pregnant naturally.

¹⁷ This can be found in: M. Ławniczak. *Zintegrowane wspomaganie harmonijnego rozwoju osób dotkniętych zespołem Turnera w Wielkopolsce (aktualny stan, potrzeby)*, <www.turnersyndrom.org.pl>.

¹⁸ Compare with “Integrated Support for TS Affected Persons” diagram created by M. Ławniczak.

tended not only for TS women and their families, but also for GP's (first contact doctors), teachers and other specialists.

Efforts should be made to increase the number of women with TS having active working lives, especially among Polish women with TS (one third of them has not been working). The first step in this process should be implementation, upon entering school, of relevant psychological and educational supports aimed at identification and stimulation of girls' interests/abilities, and support for them in choosing optimal professional careers (professional orientation). Getting education, an occupation and a job is vital in achieving a satisfying quality of life in every adult woman, including those with TS.

Worse functioning in social life and at the same time much better relationships with co-workers has been noticed in women who had not been treated with GH therapy nor took the HRT (thus the shortest, of higher BMI and lacking sex characters), and who had been diagnosed latest. This may lead to the conclusion that maintaining very close contacts at work (similar to relationships) was a kind of compensation for not having a satisfying social life and experiencing difficulties establishing and maintaining relations in the wider social environment. *Thus, supporting TS women in social adaptation and integration (sessions with psychologist/ pedagogue, inculcating self-confidence and high self-esteem from the earliest years) may positively affect their social functioning in adulthood and lead to improvement of their overall quality of life.*

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Spółeczne funkcjonowanie kobiet z zespołem Turnera

Streszczenie

Artykuł dotyczy społecznego funkcjonowania kobiet z ZT, zwłaszcza ich życia rodzinnego (relacji z rodzicami/rodzeństwem/partnerem/dziećmi) oraz relacji społecznych (z przyjaciółmi, znajomymi, współpracownikami).

kami i członkiniami organizacji wsparcia dla kobiet z ZT). Autorka próbuje znaleźć korelacje pomiędzy leczeniem hormonem wzrostu i hormonami płciowymi, wiekiem diagnozy ZT i kariotypem a funkcjonowaniem społecznym kobiet z ZT.

Zespół Turnera (ZT) jest dość często występującym zaburzeniem genetycznym (1/2500 żywych noworodków płci żeńskiej), występującym tylko u kobiet. Kobiety z ZT wykazują całkowity lub częściowy brak jednego z pary chromosomów płciowych X. Fenotyp kobiet z ZT charakteryzują: niski wzrost, zaburzenia rozwoju seksualnego i pierwotna niewydolność jajników (zwykle powodująca bezpłodność), specyficzne wady anatomiczne oraz charakterystyczny profil neuropsychiczny (zazwyczaj nieobejmujący niepełnosprawności intelektualnej). Terapia dziewcząt z ZT polega na leczeniu hormonem wzrostu i hormonami płciowymi.

W badaniach udział wzięło 71 kobiet (30 z Polski oraz 41 z innych krajów – z USA, Australii i Wielkiej Brytanii). Wynika z nich, że generalnie kobiety z ZT funkcjonują stosunkowo dobrze w relacjach w rodzinie pochodzenia (z rodzicami/rodzeństwem) oraz w środowisku społecznym (dość wysoka aktywność zawodowa, dobre funkcjonowanie w kontaktach towarzyskich i relacjach przyjaźni). Relacje z rodzicami/rodzeństwem zwykle były bardzo bliskie, ale często ukazywały nadmiernie ochraniającą postawę rodziców/rodzeństwa wobec kobiety z ZT (nawet w jej dorosłym życiu). Z badań wynika też słabe funkcjonowanie w relacjach w rodzinie prokreacji (z partnerem/dziećmi): więcej kobiet niż w ogólnej populacji miało problemy ze znalezieniem partnera i założeniem rodziny.

Słowa kluczowe: funkcjonowanie społeczne, kobiety, zespół Turnera