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Phenotypic aspects and their emotional impacts on subjects with Turner syndrome and their parents

Abstract

The present article aims to offer Turner Syndrome subjects and their families some orientations related to possible emotional difficulties during their development. Thus familiar constitution is highlighted for its importance. Turner Syndrome (TS) etiology is genetic, associated to partial or total monosomy of X chromosome. TS are prevalent in women and is generally diagnosed during adolescence.[1,2] TS subjects and their parents usually experience challenging times when they are informed about the diagnosis. Parents feel breaking expectations of the ideally imagined child. TS girls, teenagers and women may also feel the painful losses they may be in contact with. Frustration and deception reactions are associated with such moments and are equivalent to mourning feeling. Other feelings may arise needing support to elaboration and emotional overcome. We resort to Winnicott's ideas for presenting a solid psychoanalytic theory that presents a comprehensive reading of situations with the purpose of guidance. Considering the genetic profile, a great phenotypic variability can occur, with the most frequent characteristics of TS being: short stature, obesity, infertility due to the decline of ovarian reserves and comorbidities such as hypothyroidism, osteoporosis. Different cognitive changes may occur.[2] These elements justify the need for specialized work such as that of the psychologist and the psychopedagogue. By relating the emotional impact of these issues, it is suggested the development of a Guide to help families and TS subjects.

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Introduction

The development of a child is closely linked to the family environment, especially the mother's presence. The most important aspect of this relationship, especially in the early stages of life, is the affectivity that will provide the foundation for the child's mental life.

The family environment and the relationships established in it determine the child's human future. However, when the child carries a genetic anomaly, affective relationships and the environment take on specific characteristics. Genetic anomalies are expressed by different traits and tend to vary among subjects of the same syndrome, presenting different phenotypes. This is the case of subjects with Turner Syndrome (TS), as development takes place, they will start to exhibit the characteristics resulting from the anomaly, not always detected at birth. Genetic anomalies in children impose on parents a set of experiences and experiences, establishing a unique and specialized routine for the well-being of their children.

When the child is born, the environment is its mother with its care, attention and protection. According to Winnicott[3](1956/1993) in her relationship with the baby she manifests the "Primary Maternal Concern", which is a close bond with the baby. The mother develops a sensitivity in order to adapt to him and begins to recognize her needs. In turn, he gets in touch with his own needs, which represents the beginning of a mental life. This mother-baby binomial gives the little one the opportunity not to feel deprived and not to be exaggerated, as it is welcomed by the sensitive mother, although sometimes she can have her flaws, however, this is the momentcalled "good enough mother" by Winnicott[3]. This author is a reference in the psychoanalytic field, having provided significant studies on the issues of the parental relationship.

For the mother, her baby is the child imagined during her pregnancy, the idealized child who will one day become an adult and who will conquer the world and fulfill her dreams.

The close bond between the mother and her child can often inhibit the real perception of their development, this can be one of the flaws, as it is difficult for a mother to be "good enough" all the time. However, certain failures can produce effects on the affective development process, especially when the mother, or family members, fail to notice certain difficulties that the baby presents and that may be related to genetic anomalies.

In this work we will make considerations about the development of Turner subjects and the possible relationship with emotional aspects. The basic objective is to be able to implement guidelines for families and the subjects themselves.with Turner syndrome. We highlight families for having a relevant role both in the constitution of the subject and in its individualization. By recognizing the child's needs and dependence, families can offer opportunities for experiences and learning for the purpose of greater emotional integration and adaptation to the world.

Winnicott[4] points out that, for the subject to achieve a healthy mental life, it is important that in the mother-baby relationship she accepts dependence, which is absolute in the beginning, and which gradually becomes a relative dependence, until reaching the autonomy. However, when the family and the Turner girls begin to notice the body changes and difficulties and the diagnosis is completed, the feeling of mourning arises. Couto, bearing in mind these theoretical considerations as a reference for the understanding of emotional relationships, we will present some characteristics of this syndrome, both in terms of the genetic condition and those of a cognitive nature, in order to contextualize our considerations.

Characterization of turner syndrome (TS)

It is a genetic syndrome of the X chromosome, prevalent in women, with an occurrence of 1 for every 2,500 live births,[2] whose etiology is associated with the partial or total absence of an X chromosome.[1]

weaves a long discussion about the different aspects that will be visible in corporeality, here we analyze how phenotypic, as well as present cognitive, emotional and social difficulties.

The diagnosis of TS is genetic, through the examination of the karyotype. It is important to highlight the progressive increase in diagnosis at birth (mainly due to the presence of lymphedema of the hands and feet, other physical characteristics and congenital heart diseases, indicating the performance of a karyotype); and even during pregnancy, for example, the detection of cystic hygroma in a female fetus, by routine ultrasound during prenatal care, which is a strong indication of the diagnosis of TS.[5] In this way, the latter would point to the performance of prenatal testing of fetal DNA through maternal blood.[2]

There may be great phenotypic variability in TS carriers in terms of the severity of clinical, cognitive, emotional and social aspects, and most of these characteristics emerge as the developmental stages occur, many of which are due to a specific karyotype. It is noteworthy that some genetic variations associated with TS are known to be related to differences in the cognitive profile. For example, young women with TS who have mosaicism appear to be at a lower risk of presenting difficulties in visuospatial skills and in learning mathematics, compared to those with the 45,X karyotype. In contrast, girls, young women or women with TS carrying the 45,X/46,X karyotype or X-ring mosaicism [r(X)] present greater likelihood of having significant cognitive impairment with the possibility of increased difficulties in visuospatial reasoning, visuospatial memory, memory, attention, math skills, motor skills and executive functions,[2] in as a result of the relationship of such karyotypes with the commitment to the process of inactivation of the preponderance of X chromosome, a process responsible for the dose compensation of these same genes.[25,23]

It is important to emphasize that TS carriers showed normal performance, both in intelligence assessment through Piagetian tests,[6] and in research regarding social cognition through non-visual tests.[7]

Furthermore, TS carriers seem to present, during their development, cognitive compensation of a reduced visuospatial skills profile.[6] Thus, Ricardi et al.,[1] carried out a psychopedagogical intervention to optimize this mechanism. In this investigation, three pairs of 45, X girls matched for age were evaluated at two moments (pre and post-test) by the Bender test (TB) and tests or Piaget scales (EP). The experimental intervention through the process of Environmental Request,[8] was applied for one year to the girl of each pair with learning difficulties. Therefore, the data from this study suggested that the intervention instruments contributed to the increase in the performance improvement of the experimental TS girls.

Such cognitive characteristics need to be part of specialized psychological follow-up, collaborating with affective states so that TS can react more adequately in the face of cognitive limitations that may be present and that are in conjunction with emotional aspects. Psychological intervention that can strengthen the development of the self, according to Winnicottian ideas.

About the diagnosis

When a baby is born, the concern is about its health conditions, being the expectation of a normal and healthy baby. But when the doctors report the presence of a syndrome, the parents go through a very critical moment of disappointment, frustration and fear. And this is the beginning of mourning the loss of the desired normal child. Grief is the psychic suffering of the parents.

In the case of TS, the diagnosis is usually not made at birth, but at the beginning of puberty. Jung[9], reviewing the medical records of 178 ST patients, found that the diagnosis was given at a mean age of around 12.6 years and that the diagnosis was suspected due to the signs presented. The most frequent signs were short stature, webbed neck and lymphedema of the hands and feet. Other signs that follow are: cubitus valgus, high palate, breast hypertelorism, short pasterns, low hairline, anomalous ears. The same author observed comorbidities that are present in TS: hypothyroidism, osteoporosis and ear diseases (dyacusis) and spontaneous menarche in a few cases.

As a diagnosis determined later in the subject's life, it is considered that the grieving process will be dual, both for the parents and for the subject.

This grieving process is accompanied by a set of regretful feelings that will occupy the minds of the parents. Uncertainties and insecurities are part of the grief of parents and daughter. Because it is an X chromosome abnormality, the mother often feels guilty. Guilt is one of the common feelings in this process. Those who love feel guilty for thinking they have done something bad for the loved object. According to Kübler-Ross[10] feelings resulting from grief go through phases. Acceptance is the feeling of the end of the process. And it starts with the parents' adaptation to parenthood, that is, being a mother or father of this daughter. This daughter who has a genetic abnormality.

It is observed in clinical situations that many parents dedicate themselves with a certain exaggeration to their daughter with TS, some parents even go as far as overprotection, as a way of repairing the feelings of the type "where did I go wrong".

It is added that there is danger in overprotection, when subjects with TS are treated more in relation to their stature than in relation to age, which can bring emotional effects and affect the cognitive.[11]

Parents who dedicate themselves a lot to the care of their daughter with TS and end up not accompanying their other children are also observed. Some parents even express the desire to "cure" their ST daughter to justify their extensive concerns and care. However, there are cases of psychic denial of the problem, when one of the parents does not have enough psychic structure and leaves the family.

The feeling of grief is painful, it involves depressive and disquieting moments until it reaches the point of acceptance of the daughter's condition.

It is understood that these reactions are part of the process of accepting the daughter's condition. Psychological follow-up at this time is useful, according to, Winnicott[12] the mother's emotional bond with her daughter may not be harmed. A good-enough mother is able to meet her daughter's needs. She feels accepted and loved. Parents can carry on with their parenting duties and giving the snuggle. This support avoids the daughter's worries and opens the possibility for her to feel more whole and not emotionally fragmented. In addition, guidance and the necessary psychoeducational processes help to overcome many difficulties.

Impact of diagnosis on girls with TS

Confirmation of the diagnosis for girls usually occurs when they enter puberty. Short stature is common in this syndrome, which they are already noticing, as there is a comparison with other people of the same age.

Obesity is another aspect present in subjects with TS, such subjects will resent their appearance. The anguish of the lived body compared to the body valued by the culture, bring emotional conflicts in their

psyche. Increased morbidity implies lifelong follow-up, preferably by a multidisciplinary, follow-up.[13]

Another important aspect is information about amenorrhea and they will understand that this accompanies infertility.

Aberastury and Knobel[14] studying adolescence state that they go through a period of normal crisis because they are transforming their bodies and developing a new identity. These authors add that there is a mourning for the loss of the child's body, living moments of anguish that will also be present in the elaboration of the new body and the new identity. The sexual characters are accentuated and they start to have the procreative capacity. This may not occur to Turner subjects.

It is believed that the set of this moment brings the girlswith ST distressing emotional states, as they are simultaneous losses. The development process continues to occur, but they need emotional resources to strengthen and constitute their identity.

In these circumstances, the environment needs to be a facilitator and one that gives affective support to stimulate their natural potential to be and to grow, directed towards maturation. These are the necessary conditions for the bereavement to be overcome and for the development of a central and true self.[12]

Added to this is the cultural issue about the body, which proposes standards and ends up giving people a concept that demarcates certain conditions and even limitations, regardless of the person's particular and biological condition. The body in psychoanalysis is the felt body that has been elaborated with significant representations, drives and fantasies.[15] The body pattern is in the imagery of teenagers. Sometimes this idealized body becomes a prison in the minds of teenagers leading them to suffering, which can be of significant importance.to those with TS.

The Turner girls live delicate moments about the information of the diagnosis, the expectations that were broken, the anguish of their parents. There are many losses that you need to understand in order to assimilate in yourpsychological world. They are also already realizing their differences, in relation to their peers as well as those related to cultural issues. Often being discriminated against by social prejudice of what is expected as ideal.

Regarding the prejudices of people with syndrome Silva[16] makes the following contributions:

"Prejudice against people with disabilities is configured as a mechanism of social denial, since their differences are highlighted as a lack, lack or impossibility. Disability inscribes its particular character in the individual's own body. The disabled body is insufficient for a society that demands intensive use that leads to wear and tear (...)" (page 426)

Despite the social debate for inclusion, there are many mechanisms that marginalize people from the denial of the real possibilities of the subjects. The holding is the psychological support that is originally related to the care of a good enough mother for.Winnicott[12] A professional with the attitude of welcoming, or holding, supports these critical emotional moments and walks towards the structuring of feelings that are fragmented.

The professional listens, trying to be continent while providing the moment to experience the differences he perceives, the ideas that pervade his mind and how they can relate to his conditions. This work offers the opportunity to understand your subjective states that have a lot of influence on actions.

This kind of emotional acceptance shows the way to inhabit your body with its peculiarities, its individuality and its potential.

About infertility

Turner subjects have a rapid decline in ovarian reserves, in those without mosaicism the reserves are depleted in the first years of life and in those with mosaicism the reserves can reach post-menarcheal age.

Oktay et al.,[17] describe a series of fertility preservation techniques, most of them in an experimental phase. They recall that in pregnancies of women with TS there is a greater propensity for complications due to heart problems, thyroid dysfunction, obesity, diabetes and hypertensive disorders, including preeclampsia (up to 40%), in addition to obstetric risks.

Costa[18] conducted a review of the literature on fertility, only 30% develop pubertyspontaneously, 10% reach menarche and 2-10% achieve spontaneous pregnancy. It states that spontaneous pregnancy can occur in cases of mosaic karyotype, but is associated with a high rate of miscarriage. She also discusses that there is a need for guidance and counseling for Turners on fertility and pregnancy issues.

The infertility of ST women brings feelings of frustration. Although there is a set of fertilization techniques, the risk of pregnancy is a reality.

However, the desire for parenthood is a project of many couples. Emotional counseling and specialized care are essential, as this is a time of mourning for the loss of the possibility of being a biological mother.

Short stature and tendency to obesity

The most frequent alteration is short stature, which may have effects on neuropsychomotor development.[19] According to the work of Suzigan et al.,[20] the average height is between l42 and 146.8 cm. This group of scholars considers that the final height is related to genetic conditions such as primary amenorrhea, height of the parents, among others. Marini[15] reports that the average height is between 136.4 and 148.7. In the same sense, Baldin[21] complements the issue of short stature by adding another variant, which is the tendency to obesity. This is another important element regarding the perception of the body among Turner subjects.

This item brings information that refers to the concern of generating information for parents and promoting the follow-up of specialized professionals so that TS patients feel encouraged to overcome emotional obstacles.[22-26]

Final considerations

The birth of a child causes a reorganization in the family system, with plans and expectations for the future.

When parents are made aware of having a child with a genetic anomaly, the family configuration changes. The emotional component has an important place. Turner women benefit from information about their developmentand its potential. Guidelines for treatment are necessary, affective life can gain important progress.

There are particularities about what TS carriers and their families should be advised of as we were able to discuss some of them.

Information, guidance and psychological and psychopedagogical interventions are increasingly necessary, aiming at emotional strengthening and valuing the natural potential that all TS carriers have.

We believe that the elaboration of an Orientation Guide for parents and subjectswith ST would be of great value to clarify the most basic questions, some of which are dealt with in this text. The guide could consist of items such as the following:

I. The main clinical features

II. Generic profiles

III. Support team – an example of psychopedagogical intervention

IV. Support Groups and Reflections.

These suggestions are not finalists, they admit the collaboration of everyone who cares about and studies the theme.

Summary

In this work, considerations were presented regarding the development of subjects with Turner syndrome (TS) and the possible relationship with emotional aspects. The basic objective is to be able to provide guidance to families and subjects with TS. We highlight families for their relevant role in the family system. Turner Syndrome is genetic of the X chromosome, prevalent in women, the diagnosis usually occurs in adolescence.[1]

ST subjects and their parents live delicate moments from the information of the diagnosis. Parents feel that they are breaking the expectations of the imagined ideal child. ST subjects will also suffer the painful losses they come in contact with. The reactions of frustration and disappointment are associated with these moments and are equivalent to the feeling of mourning. Other feelings arise, needing support for emotional elaboration and overcoming. We resort to Winnicott's ideas for presenting a solid psychoanalytic theory that presents a comprehensive reading of situations with the purpose of guidance.

As a result of the genetic profile, a great phenotypic variability can occur, with the most frequent characteristics of ST subjects: short stature, obesity, infertility due to the decline of ovarian reserves and comorbidities such as hypothyroidism, osteoporosis. Different cognitive changes may occur.[2] These elements justify the need for specialized work such as that of the psychologist and the psychopedagogue. By relating the emotional impact of these issues, the elaboration of an orientation guide for families and STs is suggested.

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Conflicts of interest

The authors declare that there is no conflict of interest.

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