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Nursing diagnoses in Turner syndrome

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Abstract

Turner's syndrome (TS) is a rare genetic disease and has an important impact on the health care of these patients. Although TS is not uncommon, there are still gaps in the literature about nursing care. The objective of the study was to infer nursing diagnoses from the clinical conditions presented by patients with Turner syndrome, according to NANDA-I taxonomy II. This is a cross-sectional and retrospective study, with a quantitative approach. The sample consisted of 59 patients with TS, diagnosed from 1993 to 2019. The data were extracted from their medical records and submitted to diagnostic inference based on the NANDA-I taxonomy II. The clinical judgment occurred with the main dysmorphias described among the patients. Regarding the results, the most common chromosomal constitution was X chromosome monosomy (40.7%) and the mean age of the patients at diagnosis was 15.9 years (ranged from 1 month to 34 years). The main dysmorphias described were: short stature, ulna valgus, pectus excavatum, and ogival palate. The diagnoses inferred for the studied population were social isolation, impaired physical mobility, impaired swallowing, low situational self-esteem, and ineffective breathing patterns. It was concluded that the proposal of nursing diagnoses makes it possible improve the assistance of these patients, based on evidence in the care of patients with rare diseases.

Keywords: Turner syndrome. Genetics. Standardized Nursing Terminology.

INTRODUCTION

Turner's syndrome (TS) or Ullrich-Turner syndrome is clinically characterized by short stature, winged neck, and hypogonadism. It was described in 1938 by Henry Turner, and its genetic basis was discovered in 1959. TS has an incidence of 1/2,500 female births, and the associated chromosomal constitutions involve complete or partial monosomy (mainly involving the short arm) of the X chromosome. These can occur in isolation, or as a mosaic (involving more than one cell line)[1]. It is noteworthy that TS is the only chromosomal monosomy described to be compatible with living[2].

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Dysmorphia or clinical signs in TS are variable, with short stature being one of the most striking features, followed by micrognathia, the ogival palate, the valgus ulcer, delayed bone age, nail hypoplasia, and wide chest (shield chest), and renal and cardiac changes[3,4]. However, the clinical picture can be subtle. In addition, patients with TS have a greater predisposition for the occurrence of several comorbidities, such as obesity, hypothyroidism, arterial hypertension, and deficits in intellectual and neuropsychomotor development[5].

It is worth noting that in TS there can be a great phenotypic variability, and there may be girls with the classic shape evidenced by the main dysmorphias described, as well as those who have few dysmorphias; therefore, the diagnosis of girls and women with the syndrome often occurs late and implies the appearance of complications from various organ systems. Currently, the evidence shows that there is no classic karyotype and phenotype, and that the diagnosis is not always evident or even almost non-existent, requiring clinical and cytogenetic evaluations[5,6].

Thus, it is necessary for nurses to act not only in the care of these patients with TS, but also in the mitigation of possible complications, through the mapping of their evolution, in order to gather strategies for an adequate therapeutic approach. The scope of care for patients with diseases of genetic origin is inserted in the area called "nursing in genetics and genomics", which is of fundamental importance in the nurse's care practice for patients with rare diseases[6]. According to the International Society of Nurses in Genetics (ISONG), this is defined as the protection, promotion, and optimization of health; the prevention of disease and possible injuries; the relief of suffering through the diagnosis of human responses; and proactively defending the rights of individuals, their families, and communities under their care[7].

In Brazil, there are initiatives about the dissemination of nursing knowledge in genetics and genomics, which are legally supported by the opinion no. 032 of 2011, of the Regional Nursing Council of São Paulo, which concludes that nurses can work in the field of genetics, and Resolution No. 468/2014 of the Federal Nursing Council (COFEN), which establishes guidelines for the private performance of nurses within genetic counseling within the scope of the nursing team according to specific training for the area[8]. In a study carried out in Brazil[9], in a specialized center for the follow-up of women with TS, it was found through genetic counseling that psychosocial demands were portrayed as priorities by patients, with the nursing process being a way to plan the execution of improvements in the quality of life of these patients. The applicability of understanding nursing diagnoses (NDs) in the care provided by nurses with knowledge in genetics and genomics comes from the need for them to provide comprehensive care that is guided by health promotion, through communication with the family and the patient, as well as well as supporting therapeutic decisions that involve the family's quality of life within its context, acting in the management of the symptoms presented by patients with genetic conditions[8,9].

As in other genetic diseases, TS is sometimes underemphasized in the curricula and training of nursing assistants, and the information aimed at these professionals is poorly disseminated. They face the challenge of acquiring knowledge in genetics and genomics, in order to provide safe care based on clinical reasoning, so that it may serve as a foundation for the main NDs and, thus, plan the best form of assistance[9]. There are still gaps in the literature about systematized nursing care and in the study of

human responses to the health condition and life process of patients with this condition. Therefore, nursing research aimed at assisting clinical judgment, through determination of nursing diagnoses, is required in order to assist the professional in the practical process of genetic counseling. Therefore, given the gaps, it is necessary to build and disseminate knowledge about the systematization of nursing care for patients with TS, in order to enable comprehensive care.

Thus, the objective of the study was to infer nursing diagnoses from the clinical conditions presented by patients with TS, according to NANDA-I (North American Nursing Diagnosis Association-International) taxonomy II.

METHOD

This was a cross-sectional and retrospective study, with a quantitative approach, carried out at the Clinical Genetics Services of UFCSPA/ Santa Casa de Misericórdia de Porto Alegre (SCMPA) and of the Hospital Materno-Infantil Presidente Vargas (HMIPV), between January 2018 and February 2019. 59 patients with TS diagnosed in the period from 1993 to 2019 were studied. The variables collected were: age of diagnosis, karyotype examination, and dysmorphia described in the clinical evaluation. The exclusion criterion was the lack of description of dysmorphia. However, there were no losses.

The clinical judgment to infer nursing diagnoses took place in two stages: in the first, the main dysmorphias described among the patients were listed and a literature review was carried out in search of studies on the subject. In the second stage, the NDs associated with dysmorphias were identified based on the NANDA-I Taxonomy, verifying the definition of the diagnosis title and defining characteristics, in addition to the related factors and associated conditions. This study was written according to the guidelines for communicating observational studies in epidemiology (STROBE).

This study was approved by the Research Ethics Committees of the Federal University of Health Sciences of Porto Alegre (UFCSPA) with the CAAE report: 69178217.7.0000.5345 by the consubstantiated opinion no. 2.230.086 and the Hospital Materno Infantil Presidente Vargas (HMIPV) with the CAAE report: 09909712.3.1001.5329 according to opinion no. 2.326.171, with respect to all ethical and legal precepts, as recommended by Resolution 466/12 of the National Health Council, assuring anonymity of the participants.

RESULTS

The sample consisted of 59 patients, 25.4% of whom were under 2 years old, 30.5% were between 2 and 13 years old, and 44% were 13 years old or more. Their average age was 15.9 years and the median, 10.6 years (ranged from 1 month to 34 years).

Table 2 shows that in relation to the karyotype exam. 40.7% of the patients had monosomy of the X chromosome (45, X) and the remaining 59.3% had a mosaicism and structural alterations, of which the most prominent were the deletion of the short arm

of the X chromosome [del(Xp)] (n=7) and the isochromosome of the long arm of the X chromosome [i(Xq)] (n=6), according to the order of frequency in the sample.

Regarding the dysmorphias shown in Table 2, the largest dysmorphias consisted of short stature (83%), *cubitus* valgus (45.8%), ogival palate (37.2%), and *pectus excavatum* (15.2%). Other dysmorphisms also present included typical nails (hypoplastic/hyperconvex) (61.2%), low posterior hair implantation (52.1%), epicanthus (16.9%), and breast hypertelorism (22%), characteristics that are part of the clinical spectrum of TS but have a lesser impact on the quality of life and activities of daily living of these patients.

From the main dysmorphias described among the patients: short stature, *cubitus* valgus, ogival palate, and *pectus excavatum*, 5 NDs were listed, which involved the domains of comfort, activity/rest, nutrition, and self-perception, according to NANDA-I Taxonomy II. The NDs listed were: ineffective breathing pattern, impaired physical mobility, impaired swallowing, low situational self-esteem, and social isolation, as can be seen in Table 3.

Table 1– Sample characterization. Porto Alegre - RS, 2019.

Variables	n (%)
Age*	15.9±10.6
Age range	
< 2 years old	15 (25.4)
2 to 13 years old	18 (30.5)
> 13 years old	26 (44)

Data presented in absolute frequency and relative frequency.
* Data presented as mean ± standard deviation.

Table 2– Frequência das dismorfias e alterações cromossômicas apresentadas pelas pacientes com ST. Porto Alegre - RS, 2019.

Chromosomal changes	n (%)
45,X	24 (40.7)
Others	35 (59.3)
Mosaicism	21 (60)
Structural changes	14 (40)
Dysmorphia	
Short stature	49 (83)
Typical nails	36 (61.2)
Low deployment	31 (52.1)
Cubitus valgus	27 (45.8)
Ogival palate	22 (37.2)
Breast hypertelorism	13 (22)
Epicanthus	10 (16.9)
Pectus excavatum	9 (15.2)

Data presented in absolute frequency and relative frequency.
* Data presented as mean ± standard deviation.

Table 3– List of dysmorphia, associated conditions, defining characteristics, and factors related to the priority nursing diagnoses (NDs) identified in the sample. Porto Alegre - RS, 2019.

Dysmorphia	Condições Associadas*	Características definidoras*	Fatores relacionados*	DE*
Short stature	- Changes in physical appearance - Change in well-being	- Disease - Feeling different from others	- Difficulty in establishing relationships	Social isolation
Cubito Valgus	- Alteration in the integrity of bone structures - Prescribed movement restrictions	- Reduced range of motion	- Joint stiffness	Impaired physical mobility
Ogival palate	- Abnormality of the oropharynx	- Ineffective handle - Ineffective suction	- Eating behavior problem	Impaired swallowing
Ogival palate	- Functional impairment	- Situational challenge to own value	- Alteration of body image	Low situational self-esteem
Pectus excavatum	- Deformity in the chest wall - Musculoskeletal damage	- Dyspnea - Altered chest excursion	- Respiratory muscle fatigue	Ineffective breathing pattern

*Source: NANDA-International. Nanda's Nursing Diagnoses: definitions and classifications[10].

DISCUSSION

TS is a genetic condition that affects only women and is considered a rare syndrome. Currently, the evidence demonstrates that there can be a great variability in the clinical manifestations of these patients, making the diagnostic suspicion not so evident. Therefore, its clinical and cytogenetic evaluation is important[11]. In a study carried out in Brazil[12], it was identified that the constitution of 45,X observed in the karyotype exam is present in 40-60% of the patients, followed by mosaicism and structural changes, which is in agreement with the findings of our study.

In Brazil, the average age of diagnosis of patients with TS is around 12 years old, with half of them being identified between 12 and 18 years old[13]. In our study, the mean age at diagnosis was 15.9 years and the median was 10.6 years, showing that patients are being diagnosed late. This average age is above that described in a recent Ukrainian study, which found a value of 9.3 years old[14] which is recommended for patients with TS, according to the clinical protocol and related therapeutic guidelines of the Ministry of Health, which recommends a diagnosis between 2 and 12 years old[15]. Therefore, the late diagnosis has important implications, such as delay in the induction of puberty, since these patients very often need estrogen therapy for the development of secondary sexual characteristics. At the beginning of therapy with somatropin, or growth hormone, which according to the meta-analysis carried

out by Baxter *et al.*[16], has a significant impact on the quality of life and adult height of these patients. It should be noted that the difficulty of early diagnosis appears to stem from socioeconomic determinants, from less marked growth deficits, and the absence of evident physical changes (such as dysmorphias).

Regarding body changes, short stature is the most common clinical sign of TS, observed in 95-100% of patients. It is related to the haploinsufficiency of the SHOX[16] gene. In our sample, its frequency was 83%. In relation to other dysmorphias, the ogival palate and the ulna valgus stand out, which in our study were more commonly described than in another study also performed in Latin America[17]; noting that the pectus excavatum which had a relative frequency of 15.2 % in our sample was not described in this Latin American study[17].

Given the above, it is necessary to understand the impact that this condition has on the lives of these patients. Thus, the congruence of a multiprofessional team focused on long-term monitoring of these individuals is essential, as they have a chronic health condition. With the advent of studies in genomics, there was an improvement in the identification of risks, screening, and diagnosis of diseases, as well as optimization of therapy. Nurses, by incorporating the knowledge of genetics and genomics in their practice, make care based on the patient's quality of life possible and incorporate technological advances in their care[18].

The interaction between nursing with genetics and genomics is born from an approach in which they provide care with a new specialty, a new technology, and a new perspective when caring for patients. In addition, nursing science adds different worldviews and perspectives, dealing with human responses to health conditions and defending patients' rights in an era when genetic information strongly influences health decisions[19]. Thus, based on the nurse's clinical judgment founded upon the dysmorphisms presented by the patients, it was possible to identify possible NDs for the studied sample.

Short stature is an important clinical finding among patients with TS and has an important psychosocial impact on the contexts of life. In the work carried out by American researchers[20], the authors argue that patients with TS have a weaker social support network when compared to other women, as well as have fewer lasting relationships, and portray that short stature has a great impact on interpersonal relationships, resulting in rejection in the social environment and personality changes. Thus, for patients with a short stature, which was the most frequently observed finding in our study, the priority diagnosis of “social isolation” was listed, substantiated by the defining characteristics of illness and feeling different from others, due to difficulties in establishing relationships, based upon changes in well-being and physical appearance as associated conditions.

The ulna valgus is a deviation out of the forearm in extension that may even require surgical treatments due to aesthetic reasons. However, there are cases in the literature of patients with TS who develop functional limitations and secondary ulnar nerve paralysis. Ulnar neuropathy occurs when there is a compression of the nerve of the same name, causing numbness, tingling, and weakness. This is a chronic condition that often requires surgical intervention. In addition, these patients have a decrease in limb mobility and need motor rehabilitation[21]. Thus, the ND “impaired physical mobility” was defined for this dysmorphia, based on its defining characteristic: reduced range of motion, related to joint stiffness and having

as an associated clinical condition, based on medical diagnosis, changes in the integrity of bone structures and the prescribed restriction of movement.

Abnormalities in the oropharynx are common among patients with TS, and the ogival or high-arched palate has an important articulatory function, since the hard palate is an integral part of the stomatognathic system and participates in the functions of swallowing, sucking, chewing, and phonoarticulation, and has a significant impact on the quality of life of these patients[22]. The "impaired swallowing" ND, which is defined as the "abnormal functioning of the swallowing mechanism associated with deficits in oral, pharyngeal, or esophageal structures or function", was substantiated with the defining characteristics: ineffective grip and suction, as related to food behavior problems, which is a medical condition associated with the abnormality of the oropharynx.

Furthermore, these patients, due to their range of possible physical changes, may be predisposed to difficulties in social changes, which have a negative impact on well-being, as demonstrated by the review by British researchers[23]. Thus, the ND "low situational self-esteem" was listed, which is defined as the "development of a negative perception of one's own value in response to a current situation" and is present in the domain of self-perception based upon the defining characteristic of situational challenge to one's own value as pertains to changes in body image, grounded on the associated condition of functional impairment.

For dysmorphia pectus excavatum, the defining characteristics of dyspnea and altered chest excursion were identified along with accessory muscle fatigue as a factor from associated conditions, which are medical evidence, in addition to chest wall deformity and skeletal muscle damage. Pectus excavatum is a change in the chest characterized by depression of the sternum and anterior costal cartilages. Most patients who present this finding are asymptomatic; however, at the beginning of puberty, complaints of dyspnea and fatigue on exertion often arise, which may be related to cardiovascular problems, such as decreased right ventricular volume[24]. The decrease in lung function is still controversial in the literature. However, there is evidence of patients with TS displaying decreased pulmonary function tests[25]. The multicenter study carried out in North America showed that 62.1% of patients with pectus excavatum had shortness of breath[25]. Thus, the priority ND for the defining characteristics and the related factor was "ineffective breathing pattern", since the ventilation process in these patients may, in more severe cases and with progression, be impaired.

As a limitation of the study, the absence of some complementary data present in the patients' medical records stands out, and in these cases secondary information present in the literature about the theme was used. The advances in health technologies, in particular genetics and genomics, allow to glimpse future perspectives of an increasingly individualized and personalized healthcare. As this is a new competence in the nursing practice that is on the rise, studies in this field may assist in the development of care protocols to be applied to patients with rare diseases, as well as providing individualized care.

CONCLUSION

The NDs "social isolation", "impaired physical mobility", "impaired swallowing", "low situational self-esteem", and "ineffective breathing pattern" were listed from the dysmorphias identified in the studied sample, which greatly supports nursing care given to patients with TS.

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