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Zaburzenia narządu ruchu u kobiet w zespole Turnera – studium przypadku

Musculoskeletal disturbances in women with Turner's syndrome – case report

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Streszczenie:
Mianem zespołu Turnera określa się najczęściej spotykaną aberrację genetyczną związaną z chromosomami płciowymi. U dziewcząt i kobiet z zespołem Turnera występują charakterystyczne cechy związane z wyglądem zewnętrznym, które określa się mianem fenotypu turnerowskiego. Ponadto, u pacjentek występują także zaburzenia, które bezpośrednio dotyczą narządu ruchu.
Niniejszy opis przypadku oparty jest na wynikach oceny funkcjonalnej 18 letniej – wybranej losowo – pacjentki Oddziału Pediatrii, Endokrynologii i Diabetologii Dziecięcej, gdzie na podstawie badania cytogenetycznego stwierdzono wysokie stężenie gonadotropin, typowe dla zespołu Turnera. W ramach przeprowadzonej oceny zostały wykonane wybrane badania funkcjonalne, które wykazały liczne zaburzenia dotyczące nieprawidłowości postawy ciała, zaburzeń orientacji miednicy w przestrzeni, asymetrii rozkładu masy ciała na płaszczyznę podparcia oraz zaburzeń wzorca chodu. Nie zaobserwowano natomiast zaburzeń w zakresie wyników badań spirometrycznych, a także podstawowych miar stabilograficznych.
Uzyskane wyniki mogą wskazywać na występowanie innych, niż wykazane w literaturze przedmiotu, zaburzeń narządu ruchu u kobiet z zespołem Turnera.
Potwierdzenie zaburzeń funkcjonalnych wynikających z zaburzeń narządu ruchu u kobiet z zespołem Turnera może wskazywać na potrzebę przeprowadzania w tej grupie pacjentek diagnostyki funkcjonalnej oraz konieczność podejmowania wczesnej interwencji fizjoterapeutycznej.

Słowa kluczowe:
fotogrametryczna ocena postawy ciała, pomiary spirometryczne, komputerowa ocena wysklepienia stóp, ocena rozkładu masy ciała, trójpłaszczyznowa analiza chodu

Abstract
Turner's syndrome term defines the most commonly encountered genetic aberration, connect-ed with genital chromosomes. In girls and women with Turner's syndrome, characteristic ex-ternal appearance features (called Turner's phenotype) occur. Moreover, patients also exhibit disturbances directly related to musculosceletal system. Presented case study is based on the results of the functional assessment of 18 years old – randomly selected – patient from Paediatric, Endocrinology and Children's Diabetology, where on the basis of cytogenetic examination high concentration of gonadotropins typical for Turner's syndrome was found. As part of the research, following exams have been made: ob-jective photogrammetric assessment of body posture, spirometry measurement, computerized evaluation of feet arches, assessment of body weight distribution and three dimensional gait analysis. The results of this study revealed various functional disturbances in: postural control, pelvic space orientation, weight bearing distribution and in gait pattern. However, there were no spirometry and basic stabilography measures disturbances. Received results may suggest the presence of other disturbances in Turner's syndrome than those described in literature. Confirmation of musculosceletal functional disorders would indicate the need for early functional diagnostic procedures and early physiotherapy intervention in patients with Turner's syndrome starting in infancy.

Key words:
photogrammetric assessment of body posture, spirometric measurements, computer assessment of feet arches, assessment of body-weight distribution, three-dimensional gait analysis

Background

The term Turner's syndrome (TS) refers to the most common genetic aberration associated with the sex chromosomes, which occurs with an average frequency of 1 for 2500 cases [1]. At present, it is estimated that for 2000-3000 births, one child with TS is born, worldwide live 1.5 million of people with TS, while in Poland live 8-9 thousands. The basis of this syndrome is monosomy X (the total absence of an entire X chromosome), the lack of a part of a chromosome X or when the one of two chromosomes is damaged. This defect may occur in all or in some of the cells of the body [2, 3, 4].

Diagnosing TS is already possible during perinatal period by ultrasonography, which allows to detect such abnormalities as: fetal edema, coarctation of the aorta, increased nuchal translucency, renal anomalies, brachycephaly, cystic hygroma, polyhydramnios, oligohydramnios or growth retardation. Since none of these abnormalities can be perceived as a certain diagnosis, it is essential to confirm it with karyotype of a baby [1, 2, 4]. Despite the fact that it is possible to diagnose TS during pregnancy, this syndrome is rarely diagnosed during perinatal period, shortly after birth or in the first weeks of living. The most common symptom, which distinguishes girls among peers and which simultaneously might be a cause of detailed diagnostics, is the low growth. In girls and women with TS, characteristic external appearance features (called Turner's phenotype) occur. Among these features stand out: short stature, lack of or incomplete sexual development (gonadal dysgenesis, sexual infantilism), defects in the construction of the body shells (Webbed neck), the drawbacks of some internal organs, significantly reduced hairline, malocclusion (most often: short, reversed lower jaw), or the occurrence of pigmented moles. Among concomitant diseases stand out: hypertension, heart diseases and aortic deformities (coarctation of the aorta, widening of the ascending aorta, aortic arch hypoplasia, bicuspid aortic valve), thyroid diseases (hypothyroidism, Hashimoto's disease), diabetes, celiac disease, chronic inflammatory bowel disease. Moreover, patients also exhibit disturbances directly related to musculosceletal system, such as: scoliosis, hyperkyphosis, pectus excavatum, congenital hip dislocation, flat feet and knee valgus which might be associated with patella dislocation or chronic pain [5, 6, 7, 8, 9].

The above mentioned dysfunctions related to TS, may result in functional deficits, which adversely affect the quality of life of patients. In most cases, girls and women with TS, do not have intellectual disorders and identify themselves with female gender, while 10% of patients exhibit significant developmental delays, regardless of the karyotype. Many patients have difficulty with hand-eye coordination, orientation in space, the visual focus of attention, effective action planning, troubleshooting, clear speech, and remembering words [10]. In girls and women with TS may also occur self-esteem issues, which may result from immaturity, social isolation or anxiety in adolescence [5, 11]. In literature most attention is paid to intellectual and

neuropsychological aspects of TS, while reflections on motor functioning of patients are scarce, except for such exceptions as: Lewin (1926), Hartgenbusch (1927) or Koffka (1935) [12]. Physiotherapy plays an important role in the whole treatment process, however full restoration of motor skills is not possible due to genetic factors. Striving for improving patient's quality of life, by delaying the occurrence and mitigating the consequences of physical dysfunction, is the goal, which is faced against the physiotherapist and the patient.

The purpose of this study is to present the results of a functional comprehensive assessment of the patient with TS. Through appropriate interpretation and taking them into account in treatment program, these results may help to improve the comfort and the quality of life of girls and women with TS. Epidemiological data and the diversity of functional disorders clearly indicate how important is, from the sociological point of view, early identification, as well as early implementation of the particular treatment program in this group of patients.

Case description

Presented case study is based on the results of functional assessment of 18 years old – randomly selected – patient from Paediatric, Endocrinology and Children's Diabetology Department at Upper Silesia Health Center for Children and Mother in Katowice, where on the basis of cytogenetic examination high concentration of gonadotropins typical for TS was found.

In the patient, three cardiac surgeries have been carried out: aortic arch surgery, coarctation of the aorta, recoarctation of the aorta and percutaneous aortic stent (AS-30XXL) implantation. Since 2010 until today the patient is under the care of Paediatric, Endocrinology and Children's Diabetology Department at (GCZD) in Katowice, where she is treated with recombinant growth hormone treatment. Since 2012 the patient has been diagnosed with aortic aneurysm, focal fatty liver, hypertransaminasemia, decreased bone mineral density and low body mass and it was decided to implement estrogen therapy treatment. The preliminary examination of the patient revealed characteristic phenotype features: webbed neck, arched palate, valgus elbows, widely spaced nipples. Moreover, height and weight measurements (using the body composition analyzer TANITA) showed short stature (154 cm) and underweight (BMI 17.8).

Examination

As part of the patient's functional assessment following tests have been carried out:

1) Photogrammetric assessment of body posture during self-confident freely standing position, on the basis of body surface topography method through the use of CQ Electronic System [13].

2) Computed assessment of feet arches using CQ-ST Electronic System.

3) Assessment of body weight distribution on a support plane and the measurement of the basic stabilographic parameters using PDM platform (Zebris Medizintechnik GmbH).
4) Three – dimensional gait analysis (3DGA), carried out on the basis of the Compact Measuring System (CMS-HS3D) with software WinGait (Zebris Medizintechnik GmbH).
5) Spirometric measurements – using spirometer MicroLabMK8 Viasys [14].
6) Measurement of body composition using the TANITA body composition analyzer.
Additionally, to determine the gait pattern, the indicator of deviations of gait pattern from the normal gait named in the literature as Gillette Gait Index (GGI) was evaluated. The GGI (formerly the Normalcy Index, NI) is a single number which is used to indicate the deviation of a patient's gait pattern from the average gait pattern without any pathology. The higher value of GGI, the more abnormal gait pattern is [15, 16].
The abovementioned patient's functional assessments were designed to: evaluate the efficiency of the locomotor system and its dysfunctions during physiological activities in a specific section of musculoskeletal system, to assess the compensation and the possibility of its eventual correction in the case of fixed dysfunction and to analyze biomechanical dysfunction phenomena, which allows the selection of appropriate methods and treatment techniques.

Results

Obtained photogrammetric results of the patient with TS revealed numerous anomalies. This concerned especially the occurrence of thoracolumbar double curve scoliosis (Fig. 1a), with the primary curvature of 24° in the lumbar segment curvature (secondary curvature of 17° in thoracic segment) with the rotation of the lumbar spine (10° at the top of the lumbar segment) and pelvic anteversion (Tab. 1).
The results of computed assessment of feet arches indicate bilateral transverse flat feet associated with increased longitudinal arch in both feet, which indicates a significant antigravitational insufficiency in both feet (Fig. 1b, Tab. 1.).
Furthermore, computer assessment of the feet arches revealed a significant (13.8%) difference in feet prints surfaces, which in comparison with the results of the body weight distribution may indicate a distinct asymmetry of the body weight distribution – with a tendency to overload the right side (Fig. 2).
Although both spatio-temporal parameters (Fig. 2) and the GGI (GGIr = 32.6; GGIl= 29.8) ranged within a wide range of normal values, this analysis revealed disturbances in the gait patter demonstrate that the walking pattern in the stance phase as well as in the swing phase. It concerned mainly limitation of the range of motion of the pelvis in the frontal and transverse plane during to the middle phase of stance (midstance) and the middle phase of swing (midswing). Static pelvic orientation disorder, which involves excessive

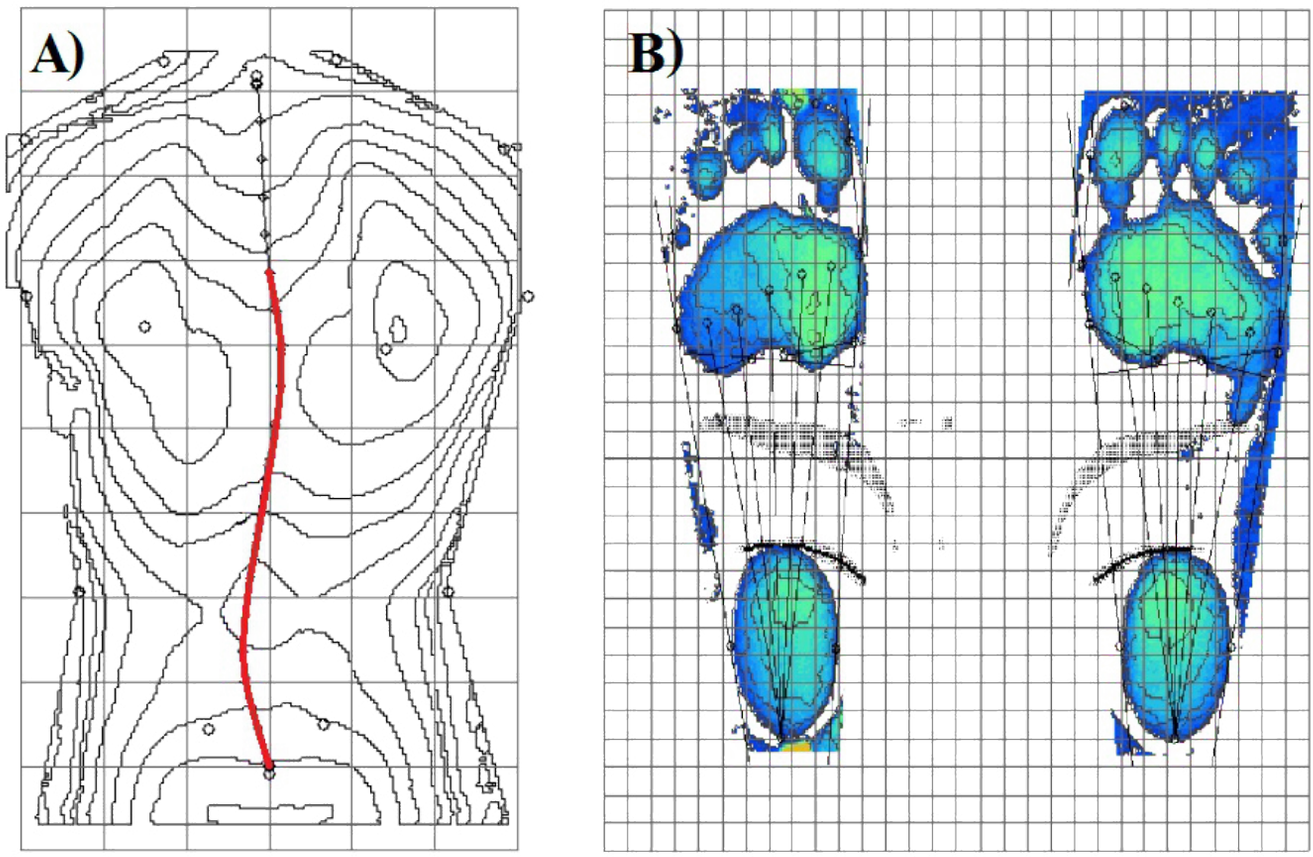
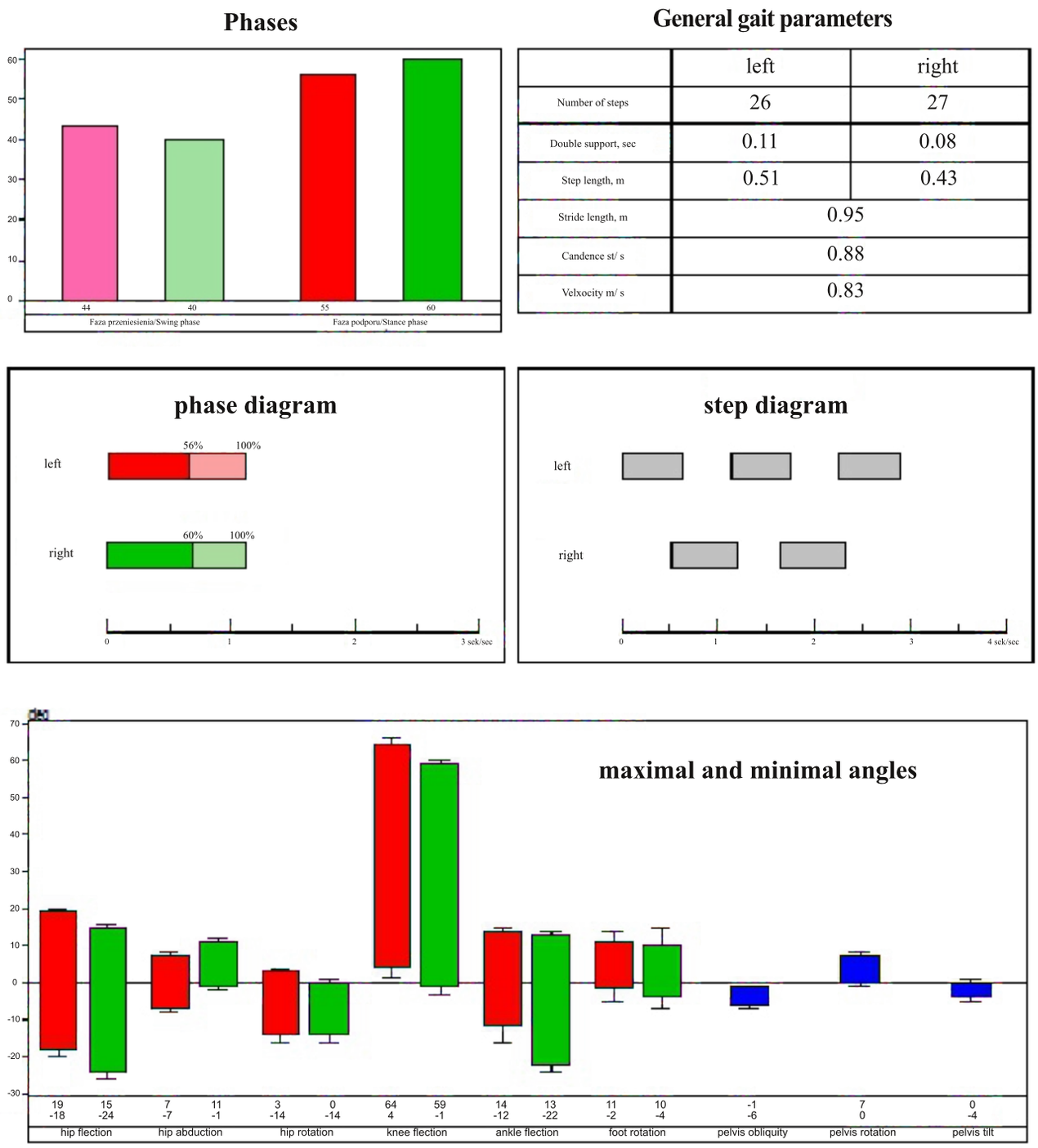


Fig. 1. Graphic interpretation of the simultaneous assessment using the method of surface topography (A) and computed assessment of feet arches (B) in TS patient

Table 1. The results of the simultaneous assessment using the method of surface topography and computed assessment of feet arches in TS patient.

	FOPC Surface topography			KOWS Computed Assessment of feet arches	
	Kąt Angle [°]	Długość Length [mm]	Strzałka Sagittal [mm]	KD Lewa LL Left	KD Prawa LL Right
Thoracic kyphosis	155.1				
Lumbar lordosis	149.6				
Pelvic tilt	9.8				
Pelvic torsion	-10.0				
Spinal curvature – right	164.6	198.0	24.0		
Spinal curvature – left	174.8	198.0	-14.0		
Wejsflog's index				2.73	2.73
Sztriter-Godunow index				0.37	0.37
Clarke angle (°)				77.20	77.20

Figure 3. Spatio-temporal parameters from the 3DGA gait analysis of the patient with TS



elevation of the concave side of the curvature, and as a consequence limitation of the hip abduction range of movement, probably are the basic cause of the disturbances of gait pattern. Both spirometric (Tab. 2) and basic stabilographic measurements did not show any abnormalities (Fig. 2). Results of the TANITA measurements revealed short stature (154 cm) and underweight (BMI 17.8) (Tab.2).

Table 2. Spirometric and body composition measurements in TS patient

	Spirometer		Tanita		
	Value	%	kg	%	
FVC (l)	2.93	93			
FEV1 (l)	2.92	107			
EVC (l)	3.43	110			
VC (l)	3.43	110			
FEV1/VC (%)	85				
FEV1/FVC (%)	100	118			
FEF25 (l/s)	5.59	96			
FEF50 (l/s)	4.26	101			
FEF75 (l/s)	3.11	150			
FEF25-75 (l/s)	4.15	105			
Weight			42.3		
Fat tissue				3.7	
Fat tissue weight			1.6		
Muscle weight			38.6		
PPM			40.7		
Water weight			29.8		
Water content				70.4	
BMI					17.8

Discussion

The reasons for raising this issue were: surprisingly small amount of research in the field of one of the most common female genetically determined diseases, accompanied with vertebrae and limb bones developmental defects and the second reason was impoverished literature. So far, results of barely three studies on the incidence of body posture defects in TS have been published (Kim et al., Elder et al., Olszewska

et al.) [8, 9, 17]. In the literature, there is a lack of studies concerning functional diagnosis of other disorders, such as feet arches disturbances, stabliographic and posturometric disorders, gait pattern deviations or impaired cardiopulmonary efficiency.

The results of the studies performed in one, randomly selected patient with TS may indicate the presence of others, than reported in the literature, musculoskeletal random – a patient with ZK may indicate the presence of others, than reported in the literature for musculoskeletal disturbances in women with TS.

The results of the assessment of patient's body posture are supported by a modest literature. In 1998 Nijhuis-van der Sanden et al. elaborated a literature review of the years 1962 to 1995 in terms of neuropsychological and locomotor dysfunctions in TS. Their study shows that girls with TS had significantly more problems with motor tasks, which require space orientation, rather than children in the control group [18]. The group of girls also showed reduced perception of their motor skills. Another studies on the posture of girls with TS have reported the presence of increased thoracic kyphosis (Deborah A. Elder et al., 2012) [9], as well as scoliosis [8, 12, 19] in this group of patients. Ricotti et al. have conducted clinical and radiological examination on 49 patients with TS to investigate the incidence of scoliosis. Their test results showed a higher incidence of scoliosis in patients which was correlated with age, growth and growth hormone therapy [19]. Kim JY et al. have developed a retrospective analysis of the incidence of scoliosis in patients with TS. Based on the results, they described the incidence of 11.6% (higher than in the control group – 2.4%) [12]. Studies conducted by Olszewska et al., concerning the assessment of body posture in women with TS conducted in a large population of patients with TS (63 women) unambiguously confirmed the presence of scoliosis and deepened thoracic kyphosis in more than half of patients [17]. All of the reported cases of scoliosis concerned curvatures above 34°, which according to the guidelines of SOSORT (Society on Scoliosis Orthopedic and Rehabilitation Treatment) is a second degree scoliosis, that is those which besides physiotherapy require corseting.

According to revealing numerous and diverse functional disorders associated with body posture abnormalities, disorders of pelvic spatial orientation, asymmetrical distribution of body weight on a support plane and gait pattern disorders in a random young woman with TS, routine functional assessment of each patient with TS, preferable immediately just after diagnosis, should be considered as advisable.

To identify the range of musculoskeletal disorders and the frequency of occurrence of these tests should be carried out on a larger population of women with TS. Disclosed musculoskeletal abnormalities can significantly affect the quality of life and functioning of patients with TS. Therefore, prevention of movement disorders from the moment of

detection of the disease in young girls should be taken into consideration.

As a result, improving the quality of life in both physical and mental sphere of life of patients with TS, would be more effective. Confirmation of functional disorders resulting from musculoskeletal disorders in women with TS will indicate the need for early physical therapy intervention in girls with TS already during infancy.

From the clinical point of view it seems advisable to recommend a comprehensive functional assessment of patients with TS, proposed as the part of this study.

It may help to facilitate the comparison of the results of the assessment of locomotor posture and a better understanding of the prevalence of various disorders in patients with TS. This will require better cooperation between orthopaedists, endocrinologists and physiotherapists, because of the fact that serious worsening of faulty posture in patients with TS treated with growth hormone threatens the aim of the therapy [20].

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