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Original article

SINGLE CASE STUDY: EFFECTS OF THE PHYSICAL ACTIVITY AND EXERCISE IN A SEDENTARY SUBJECT SUFFERING FROM METABOLIC TURNER'S SYNDROME

ANNUNZIATA PASSALIA¹, IZZO RICCARDO¹, MAURO ANDREANI², FEDERICA TURCHI², MAURIZIO SUDANO², VINCENZO BIANCALANA¹

Abstract

Turner's Syndrome (TS) is a genetic disorder associated with abnormalities of the X chromosome. TS, is usually related with reduced adult height, gonad dysgenesis and thus insufficient circulation levels of female sex steroids leading to premature ovarian failure and infertility. Often show high levels of cholesterol and increased triglycerides, determined by obesity and hyperinsulinemia. In the women with Turner's Syndrome is impaired glucose tolerance (10-34%). In addition, adult women tend to develop insulin resistance and propensity for type 2 Diabetes Mellitus. Physical activity is important in the management of type 2 diabetes mellitus

The aim of this study was to examine the effects of the physical activity and exercise in sedentary subject suffering from metabolic TS.

The patient with TS, was recruited during the counseling for exercise in the diabetes clinic. For counseling has used the transtheoretical model in accordance with the guidelines of the American College of Sports Medicine. She was monitored for one year, and before and after the start of the educational exercise has undergone a series of tests: blood, and anthropometric. The level of physical activity was estimated by the 7 day physical activity recall (7-DR). The exercise prescription is in accordance with the guidelines of the American Diabetes Association for the improvement of metabolic fitness.

Results: From the literature, it is clear that young women with TS have a low level of physical activity. Our intervention allowed the patient to improve her level of physical activity going from low to a moderate level. Improvement of the level of physical activity was assessed by 7-DR, and through the armband. Therefore, analysis of the data, it appears a significant improvement in BMI, abdominal girth, blood pressure and blood glucose, compared to baseline.

Conclusion: We believe that the results obtained after one year to serve as a stimulus not only to reduce morbidity and mortality, then, but also to improve the lifestyle.

Key words: Turner's Syndrome, Physical Activity, Fitness Metabolic.

Introduction

Since the description of Turner syndrome (TS) by Henry H. Turner in 1938, a wealth of information has been added and our current understanding of the syndrome is continuously being broadened. The syndrome affects only females and care must include the close collaboration of several specialties such as genetics, embryology, pediatrics, gynecology and obstetrics, endocrinology, cardiology, gastroenterology, oto-rhinology, ophthalmology and others.

The genetic background of the TS phenotype is highly variable, but includes complete or partial absence of the sex chromosomes (the X and/or Y chromosomes). In addition, mosaicism with two or more cell lines may be present. The first described cases were with the 'classical' karyotype 45,X. In more recent series the classical karyotype only accounts for 50% of cases; the remaining cases comprise mosaic karyotypes (i.e. has cells with 45,X and cells with 46,XX), karyotypes with an isochromosome of X—for

example i(Xq) or i(Xp)—or karyotypes with an entire or part of an Y chromosome. The genetic basis for the findings in TS is being further unraveled as the functions of the SHOX gene become clearer. Haploinsufficiency of SHOX explains the reduction in final height, changes in bone morphology, sensorineural deafness and other features.

Prenatal prevalence of the syndrome is much higher than the postnatal prevalence, for there is a well-described increased intrauterine mortality. Prenatal diagnosis of TS may not always be correct; therefore a more precise diagnosis rests on inclusion of high-resolution ultrasound scan or foetal echocardiography and other modern investigations (Baena, et al., 2004; Nielsen, Wohler, 1991)

Most postnatal diagnoses are made at birth (15%), during teenage years (26%), and in adulthood (38%), with the remainder being diagnosed during childhood (Savendahl, Davenport, 2000) and therefore there is a considerable delay in diagnosing girls and adolescents. Interestingly, the key to diagnosis was lymphedema in

¹ Department Of Biomolecular Sciences, Division Of Movement And Health Sciences; University Of Urbino "Carlo Bo" – Urbino (Pu), ITALY

² Integrated Territorial Service of Diabetology and Metabolic Diseases, Asur Marche Zone N², Hospital "Santa Maria Della Misericordia"– Urbino (Pu), ITALY.

E-mail address: vincenzo.biancalana@uniurb.it

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97% during infancy, and short stature in 82% during childhood and adolescence.

Morbidity is considerably increased in TS. In a study of all females diagnosed with TS compared with the general population of women, we compared the incidence rates of specific diseases we suspected might occur with increased frequency. The relative risk (RR) of an endocrine diagnosis in TS patients is significantly increased to 4.9 overall, with a significantly increased risk of hypothyroidism (RR 5.8), type 1 diabetes (RR 11.6) and type 2 diabetes (T2DM) (RR 4.4). The risk of ischemic heart disease and arteriosclerosis (RR 2.1), hypertension (RR 2.9) and vascular disease of the brain (RR 2.7) were also significantly increased. The risk of other conditions such as cirrhosis of the liver (RR 5.7), osteoporosis (RR 10.1) and fractures (RR 2.16) was also significantly increased, as were the risks for congenital malformations of the heart, of the urinary system, of the face, ears and neck. The risk for all cancers was comparable to other women, with only the risk of colonic and rectal cancers being significantly elevated (RR 4.94). Congenital malformations are most frequent among women with the 45,X karyotype, whereas endocrine diseases, heart disease, hypertension and arteriosclerosis are more frequent in women with other TS karyotypes.

Mortality is also increased in TS. In a British cohort study the RR of premature death was increased to 4.2 with increases due to diseases in the nervous, digestive, cardiovascular (CV), respiratory and genitourinary systems. Death due to cancer was lower than expected, corroborating the morbidity studies (Swerdlow et al., 2001). We found a comparable increase in mortality among Danish patients, with important differences between patients with 45,X or an isochromosome, who had a four-fold increase in mortality, while patients with other karyotypes only had a two-fold increase in mortality.

In summary, TS is a clinical description without firm guidelines for the diagnosis, but the cardinal stigmata include growth retardation with reduced adult height and, except in rare cases, gonadal insufficiency and infertility.

Typical features in Turner's syndrome such as growth deficit and abnormal body proportions may result in perceiving Turner girls as being physically weaker. Hence, physical activity systematically undertaken by those girls would be of utmost importance for keeping healthy and for an adequate relation of body fat to muscle mass, sustaining or increasing physical capacity and for prevention of osteoporosis and hypertension (Pasquino et al., 1997).

Physical activity not only contributes to well-being, but is also essential for good health. People who are physically active reduce their risk of developing major chronic diseases – such as coronary heart disease, stroke and type 2 diabetes – by up to 50%, and the risk of premature death by about 20-30%. The evidence also clearly demonstrates that achieving the weekly recommendation is not the preserve of the sports

enthusiast. We all can and should be more active. The evidence of the potential health gains from active lifestyles is clear. Changing inactive lifestyles and levels of inactivity presents a tremendous public health challenge – a challenge we must rise to if we are to improve health.

Physical activity and exercise are often used interchangeably, but these terms are not synonymous. Physical activity is defined as any bodily movement produced by the contraction of skeletal muscles that result in a substantial increase over resting energy expenditure. Exercise is a type of physical activity consisting of planned, structured and repetitive bodily movement done to improve or maintain one or more components of physical fitness (ACSM's, 2000).

The aim of this study was to evaluate the effects of physical activity and physical exercise within the activity counseling in a patient suffering Turner syndrome dysmetabolic.

Materials and Methods

Dysmetabolic patient suffering from Turner syndrome was recruited through the counseling exercise in the diabetes clinic of the hospital of S. Maria Misericordia Urbino. The diagnosis of Turner syndrome aberrant to 60% have been diagnosed by the age of 5 years. At the beginning of the study, the patient was 24 years old. The study lasted 12 months. Before the study anthropometric and metabolic characteristics were as follows: Height: 1,60, Weight: 73,5, BMI: 28,7, Waist Circumference: 85 , Fasting Glucose: 104, systolic blood pressure: 140, diastolic blood pressure: 85 (American Diabetes Association. Standards of Medical Care in Diabetes-2013).

The level of Physical Activity was estimated by the questionnaire 7 DR, and it resulted sedentary since the patient took an exercise of less than 150 minutes/week.

The 7 DR, is a semi-structure interview that estimates an individual's time spent in physical activity, strength, and flexibility activities for the 7 days prior to the interview. The general interview format is as follows: an interviewer asks the participant to recall time spent sleeping and doing physical activities for the past 7 days. The interviewer guides the participant through the recall process, day-by-day, to determine duration and intensity of the physical activities. From hours spent in moderate, hard, and very hard intensity physical activities, total Kilocalories/day can be estimated. The role of the interviewer is very important as it must collect accurate information from the participant. This is not always easy. If a standard format is not followed, an interviewer might gather information that is biased by subjective thoughts and feelings he/she may have about what physical activity a participant actually does or does not perform (Sallis, 1985)

The primary outcome was to educate dysmetabolic patient, which was highly sedentary, to increase

physical activity in daily hours and more to perform 150 minutes of exercise a week at least. The motivational tool to encourage the patient towards a more active lifestyle has been counseling exercise. In fact, they are significant studies (Di Loreto et al., 2003; Moyer, 2012; Peterson, 2007) that confirm the importance of counseling to promote physical activity both in healthy population and in the population affected by metabolic disorders.

The counseling technique used was of type transtheoretic (transtheoretical model of behavior change) in accordance with the guidelines of the American College of Sports Medicine (Battistini D., Piana N., De Feo, 2007). In the transtheoretical model change is a "process involving progress through a series of stages: precontemplation, Contemplation, Preparation, Action, Maintenance, Termination. In addition, the researchers conceptualized "relapse" (recycling) which is not a stage in itself but rather the "return from Action or Maintenance to an earlier stage".

Results

The dependent variables under study were measured at time 0 (baseline) and at time1 (after 12 months).

As illustrated in the following graphs (Fig. 1, 2, 3) shows a significant improvement in the variables considered: fasting blood glucose, weight, BMI, blood pressure.

Fig.1 (Fasting Glucose) denotes a significant improvement in fasting plasma glucose, from a value of 104 (borderline diabetes) to a value of 71 condition of perfect normality.

Fig.2 (Weight / BMI), indicates a decrease in weight and therefore in BMI. The reduction of these two variables leads to a general improvement in the patient, which passes from a state of overweight with a BMI = 28.7 to a condition of normal with BMI = 26.

Figure 3 (Blood Pressor), outlines a reduction of pressure: systolic 140 to 120; diastolic blood pressure from 85 to 80.

Moreover, in the summary table is also outlined to improve of physical activity level (Physical Activity Recall, PAL) estimated by the 7 DR. Therefore, we observe a significant improvement as the sedentary patient moves from a sedentary PAL <150 min / week to an active PAL > 150 min. / week.

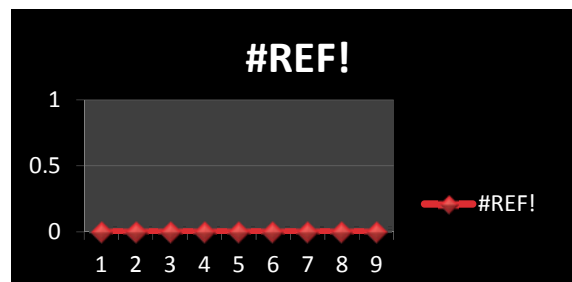


Figura 1 Fasting Glucose

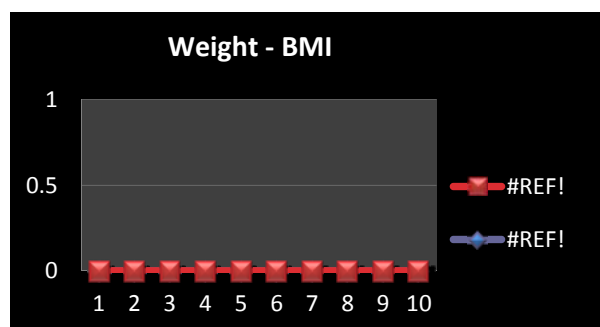


Figura 2 Weight / BMI

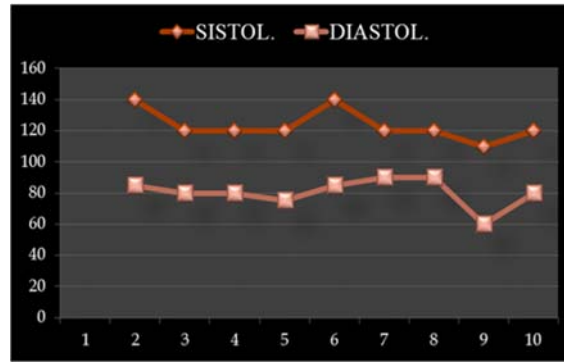


Figura 3 Blood Pressor

VARIABLE	BASELINE		12 MONTH	
Fasting Glucose	104		71	
Weight	73,5		68	
BMI	28,7		26	
Blood Pressure	S	D	S	D
❖ PAL	140	85	120	80
	Sedentary		Active	
	< 150 min/week		> 150 min/week	

Table 1 – Change in Fasting Glucose, Weight Body Mass Index (BMI), Blood Pressor, and Physical Activity Level (PAL), after counseling exercise.

Conclusion

Patients with TS need comprehensive care preferably from a multidisciplinary team. Knowledge concerning TS is still very limited. Glucose metabolism, weight, thyroid function, bone metabolism, blood pressure, liver function and CV status should be assessed This study suggests that physicians' efforts in physical activity counseling may have the best impact when provided in the context of a health problem. Our counseling strategy was designed based on the conclusions of the report released in 1996 by the U.S. Department of Health and Human Services (U.S. Department of Health and Human Services, 1996), regarding the efficacy of the interventions to promote physical activity in adults. In effect an increase in physical activity improves insulin action in obesity, with or without a concomitant reduction in body weight and fat stores (Felton, Parsons, 1994). Moreover, for some individuals, improvement may occur within 1 week of the intervention. This is an important and often overlooked salutary effect of regular exercise, suggesting that physical activity is as efficacious in preventing insulin resistance as losing body weight. The level of physical activity of girls with Turner's

syndrome, is relatively low, low interest in practicing sports ;

The results obtained in this study show that the counseling exercise and physical activity lead to an improvement in the health of the patient, resulting in a decrease and then an optimization of the primary outcomes and consequently the lifestyle.

Discussion

(Turner Syndrome: Updating the Paradigm of Clinical Care)

Turner syndrome (TS), in which there is loss of all or part of one sex chromosome, occurs in one in 2500 live-born females and is associated with characteristic findings, such as growth failure, pubertal delay, and cardiac anomalies (1). Recent studies show that evaluation of the clinical findings in TS cannot occur in isolation because all findings relate to the underlying pathophysiology of this genetic disorder. Consequently, clinicians cannot solely rely on knowledge gleaned from previous experience treating other isolated conditions when making diagnostic and treatment decisions for women with TS. For example, historically young girls with TS diagnosed for reasons unrelated to poor growth were treated the same as any child



with short stature, with delay of initiation of GH therapy until height velocity began to noticeably decrease. Some studies suggest, however, that girls with TS may benefit from significantly earlier treatment because their growth rates often decrease in the first few years of life (4).

In addition, mounting evidence suggests that hypogonadism in TS leads either directly or indirectly to a reduced quality of life: Haploinsufficiency of genes on the X chromosome has been implicated in the presence of an increased risk of congenital malformations: cardiovascular features, hormones levels, feature related to sex hormones, metabolic features.

Metabolic diseases involve an alteration body composition, as decreased muscle mass, increased total fat mass and visceral fat mass, a more sedentary lifestyle and decreased VO_{2max} and muscle strength. These factors contribute to the risk of developing reduced insulin sensitivity and type 2 diabetes. Therefore, regular physical activity is an important part of the TS management plan regular exercise may prevent type 2 diabetes, as regular exercise has been shown to improve blood glucose control, reduce cardiovascular risk factors, contribute to weight loss, and improve well-being.

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