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EFFECTIVENESS OF A PHYSICAL EXERCISE REHABILITATION PROGRAM ON A PATIENT WITH MOWAT-WILSON SYNDROME

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ABSTRACT

This brief report describes the effects of a rehabilitation program on a patient with confirmed MWS who underwent a psychomotor activity program. The second edition of the Test of Gross Motor Development (TGMD-2) was used to assess movement skills development. The Childhoold Health Assessment Questionnaire (CHAQ) was used to assess the impact of the program on the girl's quality of life. Motor competence could not be measured, since the girl did not understand the tasks proposed in the TGMD-2. The comparison of the pre- and post-intervention results obtained from the CHAQ indicated that the program hardly had any relevance in most of the domains which were assessed. These findings suggest that physical exercise seems to be a feasible tool in the management of MWS although its effects do not have a great impact on the patient's physical and intellectual condition.
Keywords: *Exercise, Mowat-Wilson syndrome, psychomotricity, rehabilitation.*

1. INTRODUCTION

Mowat-Wilson syndrome (MWS) (OMIM #235730) is a rare multiple congenital anomaly syndrome, first clinically delineated in 1998 (Mowat, Croaker, Cass, Kerr, Chaitow, Ades, & Wilson, 1998). MWS is characterized by a distinct facial phenotype, severe mental retardation and variable congenital malformations including Hirschsprung disease (HSCR), congenital heart defects, agenesis of the corpus callosum and genitourinary and eye anomalies (Mowat, Wilson, & Goossens, 2003). MWS is considered a genetic disease caused by heterozygous

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mutations or deletions in the Zinc finger E-box-binding homeobox 2 gene, (*ZEB2*) and to date, about 200 molecularly proven MWS cases with over 100 different *ZEB2* mutations have been reported (Mundhofir, Yntema, van der Burgt, Hamel, Faradz, & van Bon, 2012). There is no specific treatment for MWS, as the neural defect and also other malformations resulting from the mutation occur in the early stage of embryonal development (Bassez, Camand, Cacheux, Kobetz, Dastot-Le Moal, Marchant, & Goossens, 2004). Psychomotor development is retarded in all patients, therefore rehabilitation strategies, including physical and psychomotor therapies are needed (Garavelli & Mainardi, 2007). This short report describes the effects of a physical exercise rehabilitation program on a patient with confirmed MWS.

2. METHODS AND MATERIALS

2.1 Patient Description

A five-year-old girl, clinically diagnosed with MWS, volunteered to take part in a psychomotor activity program, after obtaining informed consent of her parents. She was the second daughter of non-consanguineous parents with no family history of mental retardation, heart disease, epilepsy or Hirschsprung disease. The girl showed moderate mental retardation, hypotonia, gait abnormality (crawling) and difficulty in grabbing objects. Moreover, she also showed repetitive hand movements. She communicated through sign language and displayed a cheerful and carefree behaviour, smiling frequently. She weighed 18 kg and she was 102 cm tall. She also showed a patent and characteristic facial dysmorphism, which included narrow chin, cup ears, wide nose bridge, short philtrum, small mouth, prognathism and thick and separated eyebrows. Her vision was normal; her hands had long interphalangeal joints; and she had flat feet.

2.2 Psychomotor Activity Program

A psychomotor activity program was carried out for five months with a 1-hour session per week. The sessions, which took place in a 20x40 multipurpose room, focused on the development of the general dynamic coordination (locomotion, transportation, balance), as well as on more specific tasks which required fine motor skills (gripping, precision throwing, etcetera). The first 10 sessions were individualized, whereas in the following 15 sessions the girl exercised with her sister. In another five sessions both girls were accompanied by a girl with Down's syndrome. Thus, some cooperation tasks were proposed (cooperative material transportation, throwing and reception, etcetera). All sessions were monitored by

a specialist in physical exercise and motor development. The procedures of this research followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000 (Saif, 2000).

2.3 Measurements

The second edition of the Test of Gross Motor Development (TGMD-2) (Ulrich, 2000) was used to assess movement skills development. The validated Spanish version of the Childhood Health Assessment Questionnaire (CHAQ) was used to assess the impact of the program on the patient's quality of life (DeInocencio, García-Consuegra, Merino, Calvo, García, & Ruperto, 2001).

3. RESULTS

Table 1: Effects of the physical exercise intervention on the patient's quality of life

Domains	Pre-Intervention	Post-Intervention	Improvement (%)
Dressing	2.75	2.2	20
Arasing	0.33	0.30	9.1
Eating	2.66	2.66	0
Walking	2	1	50
Hygiene	2.8	2.8	0
Reach	2	1.5	25
Grip	3	3	0
Activities	2.8	2.8	0
Disability Index	2.29	2.03	11.36%
Evaluation of pain	None applicable	None applicable	None applicable
Evaluation of overall well-being	Very badly	Very badly	0

The girl attended 16 sessions out of 20. Absence was due to transportation problems (1 session) and the presence of the flu (3 sessions). It was not possible to assess the progress experienced in motor competence because the girl had

difficulties in understanding the tasks proposed in the TGMD-2 and she was not able to carry out any of the tasks in accordance with the established protocol. The comparison of the pre- and post-intervention results obtained from the questionnaire indicates that the program hardly had any relevance in most of the domains which were assessed (disability index decreased from 2.28 to 2.02). However, some improvements were found in the ability to walk (50%) and in the dressing and reaching domains (25% and 20% respectively), as it can be observed in Table 1. Apart from the changes observed in the questionnaire, a greater motor independence was seen throughout the sessions. The girl became more autonomous to carry out the tasks proposed, that is, needing less help and understanding the orders more easily. Travel speed and grip firmness were not substantially enhanced, although static and dynamic balance experienced some improvement.

4. DISCUSSION

To the authors' knowledge, this is the first research aimed at identifying the potential effects of a psychomotor program on a person diagnosed with SMW. The results indicate that although the therapy may be easily carried out, the effects are limited to some improvement in the ability to walk and, to a lesser extent, the capacity to stand up. Along these lines, some improvement in gait was obtained by other interventions based on the performance of physical activity by patients with genetic disorders characterized by muscular hypotonia, proving the efficacy of physical rehabilitation in this variable (Vismara, Cimolin, Grugni, Galli, Parisio, Sibilia, & Capodaglio, 2010). However, observation of the girl's progress showed other improvements which the CHAQ could not detect, namely, a greater motor autonomy and an improved intellectual understanding. In this regard, although the CHAQ was chosen because it is considered a valid tool to apply to patients who have motor limitations that can affect functionality and the abilities for activities of daily living (Morales, Funayama, Rangel, Frontarolli, Araújo, Pinto, & Silva, 2008), the results may not show the relevance of the effects of the program. It is important to highlight the fact that the girl exercised better in company, which confirms the idea that when exercising with normally-developed people, individuals with ID take a greater advantage of the program, which has been observed in other studies (Temple & Stanish, 2011). This strategy also contributed to reduce the frequency of erratic behaviour episodes, typically observed in people with MWS when they are in pediatric therapy (Uesugi, Naruse, Inoue, Koeda, Gotou, Nanba, & Tokuhisa, 2010). Although some improvement was observed as regards balance, it was not of great relevance, which is in line with the findings reported by other authors when proposing physical rehabilitation for diseases with similar motor characteristics (Capodaglio,

Cimolin, Vismara, Grugni, Parisio, Sibilia, & Galli, 2011). In spite of the obvious limitations of this brief research, it should be borne in mind that MWS is a very rare genetic condition. Therefore it is important to communicate its features within the pediatric community as well as to provide counseling to the parents. The findings showed here could help to this aim.

5. CONCLUSIONS

The results of this case study show that physical exercise seems to be a feasible tool in the management of MWS although its effects do not have a great impact on the patient's physical condition.

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