

## Research Article

# Occupational Performance of Children and Adolescents with Mucopolysaccharidosis Using Assistive Technologies

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## Abstract

Mucopolysaccharidoses (MPS) are a specific group of genetic diseases in which due to the accumulation of glycosaminoglycans (GAGs) in different organs and tissues, causes multisystemic changes that compromise the functionality and occupational performance of individuals. Occupational performance, understood as the participation and execution of activities of daily living, may be favoured using Assistive Technology (AT). Since there are no studies reporting the influence of AT on the occupational performance of children and adolescents with MPS, the objective of this study was to evaluate the occupational performance in self-care activities, based on the use of low-cost AT in children and adolescents with Mucopolysaccharidosis. Six individuals with MPS types I, IV-A and VI, aged 9 to 16 years participated. The instruments used for data collection were the Pediatric Disability Assessment Inventory (PEDI) - self-care area only, and the Canadian Occupational Performance Measure (COPM). The results showed that the tasks that presented the greatest disabilities in the performance are the areas of dressing, personal hygiene and bath. Thus, TA resources were made for five activities related to dressing and one for personal hygiene. After the use of AT, there was a positive and significant change in occupational performance and satisfaction of these individuals. Thus, the use of AT can significantly improve the occupational performance of this population.

**Keywords:** Adolescent, Assistive Technology, Child, Mucopolysaccharidosis, Occupational Performance, Self-Care Activities

## Introduction

Mucopolysaccharidoses (MPS) are rare diseases, characterized by genetically determined metabolic errors, which are part of the Lysosomal Deposit Disease group. In these diseases there is accumulation of substrates that are normally degraded in lysosomes, and in MPS, deficiencies of specific enzymes lead to the accumulation of glycosaminoglycans (GAGs), resulting in a series of signs and symptoms, which together bring systemic impairment [1–3]. There is no cure for this group of diseases, and the current treatment is aimed at delaying its progress. Even with treatments, progression is nonetheless long-term, and changes in body structures and functions (joint stiffness, decreased range of motion, joint laxity, claw hand) result in limited functionality in the areas of occupational performance, especially in self-care tasks - related to dressing, personal hygiene and food [4].

Occupational performance is understood as the ability to perform routines and perform roles and tasks, involving the areas of self-care, productivity and leisure, being influenced by the factors of the individual, their skills and the context in which they are inserted [5]. Thus, for individuals with some form of physical limitation, occupational therapists may use Assistive Technology (AT) as an effort

to enable improved independence and occupational performance, to the extent that limitations can be overcome through adaptations and use of ATs.

Assistive Technology allows a person with a limitation to perform activities and tasks more independently, and can be characterized as technology of high complexity (high cost - with electronic components) or low complexity (low cost), the latter being designed from everyday easily accessible materials that can often be made from materials available at home, in the office, at school or in the hospital. This type of AT is something that can be done right away to meet the needs of those who need it, with the resources at hand [7–9]. However, there are no studies linking the use of AT and MPS. Thus, this study aims to evaluate occupational performance in self-care activities, based on the use of low-cost assistive technology in children and adolescents with Mucopolysaccharidoses.

## Methods

This is a prospective and descriptive longitudinal quantitative research, conducted at the outpatient infusion and enzyme replacement therapy center of a reference Hospital for the treatment of rare diseases, located in Rio de Janeiro - Brazil. Participated in the

study: Six children and adolescents of both sexes between 9 years and 6 months and 16 years and 4 months of age, with type I, IV-A and VI MPS, with biochemical diagnosis of MPS that are treated with enzyme replacement in the institution's medical genetics department. Were excluded from this research: Individuals with type III MPS because of neurological impairment; children and adolescents who had severe cognitive and / or motor impairment that prevented them from responding to assessments; and children and adolescents who reached the maximum PEDI score. For data collection, the Pediatric Disability Assessment Inventory - PEDI was used, only Part I - Child Abilities, which reports on the child's functional abilities to perform daily activities and tasks and on the self-care scale [10] and, then, the Canadian Occupational Performance Measure - COPM was applied.

The PEDI was applied through a structured interview with children and adolescents, lasting on average 30 to 40 minutes, where it was identified if individuals can perform certain activities. The COPM was administered in around 10–15 minutes, with participants identifying issues related to their occupational performance related to the activities contained in PEDI. They chose the activities that were meaningful to them, quantifying the degree of satisfaction and importance they attributed to each of the activities. At the end of the application of the instruments, it was made a survey from the chosen activities (the activity that obtained the highest importance score in the COPM) and the possible assistive technology resources to be incorporated in the intervention process of the activity that gained the most quantification, by the participants, including from creating and building a low-cost TA resource to providing guidance to follow during activities performance. With the AT done, its use was trained with the participants and the responsible person accompanying them by the main researcher and after the participant's minimum 2 weeks of AT use, the COPM was reapplied to assess if there were changes in

occupational performance with the aid of the AT. This reapplication was made by a blinded evaluator who had no prior knowledge of previous results.

The COPM was created as an outcome measure, therefore, the total scores of the initial moment and the moment of re-evaluation were used with the objective of comparing the occurrence or not of changes in occupational performance and satisfaction, so it could be proved the effectiveness of an approach or intervention – in this case, the use of Assistive Technology. These changes were calculated by subtracting the evaluation values from the re-evaluation values, both for performance and satisfaction. The participants' scores were not compared with each other, as COPM is an individual measure. With the completion of research data collection, the assistive technology resource made and/or adapted for each participant remained the same for continuous use. This study is part of a project approved by the Research Ethics Committee of the research site, under the number 1.827.932, valid until 31/10/2021, complying with the ethical principles in accordance with resolution 466/2012, and all participants were informed about the study, objectives, benefits and risks.

## Results

From PEDI results we observed impacts on occupational performance, which consequently affects the ability to perform self-care tasks, especially in dressing, personal hygiene and bathing activities, as can be seen in Table 1. The changes in self-care activities observed from PEDI, participants chose the activities that were most significant through COPM, adding a value about it, to quantify its importance in performing it on a daily basis or wanting to execute it. Table 2 shows the chosen activities, the degree of importance and the AT made. It is noted that the activities varied, related to dressing or personal hygiene.

**Table 1.** Affected items grouped by tasks performed in PEDI self-care.

Participant	ITEMS AFFECTED					
	Feeding (14)*	Personal hygiene (14)*	Bathing (10)*	Dressing (20)*	Toilet use (5)*	Sphincter control (10)*
1	-	-	-	12	-	-
2	-	2	2	5	2	-
3	2	2	1	12	1	-
4	-	5	-	11	-	-
5	-	4	-	9	-	-
6	2	4	3	12	2	-

\*: Number of items contained in each self-care task according to PEDI.

After making and training the ATs, Table 3 presents the changes in occupational performance and satisfaction in performing the selected tasks. The improvement of these two parameters was observed throughout the sample. However, it was observed that it was not possible to infer changes in two cases (participant 4 and participant 6), because they did not use the AT after training: participant 4

started training at home, but didn't feel willing to keep using the AT, preferring that his mother did the activity for him; and participant 6, did not use, because he did not wear clothes that have button or zipper at home, only using to go out and preferring that his mother performed the activity.

**Table 2.** Description of activities, importance given by participants - COPM and AT made

Participant	MPS	Activities chosen at COPM	Grau de importância	AT
1	II	Put on socks	9	Sock on Applicator
2	IV-A	Brush hair	9	Hair brush with L-form
3	IV-A	Remove socks	8	Stretch cable to remove socks
4	VI	Put on socks	10	Sock on Applicator
5	VI	Wear lower end (buttoning and zipper handling)	9	Buttoning
6	VI	Dress upper and lower extremity (buttoning and zipper handling)	8	Buttoning

**Table 3.** Importance / Performance / Satisfaction Relationship - Before and after the application of AT and observed changes

Participant/ MPS	Activity	Importance	Initial Evaluation		Revaluation		Change	
			Performance 1	Satisfaction 1	Performance 2	Satisfaction 2	Performance	Satisfaction
1 (type II)	Put on socks	9	2	2	5	8	3	6
2 (type IV)	Brush hair	9	5	3	10	10	5	7
3 (type IV)	Remove socks	8	2	4	4	7	2	3
4 (type VI)	Put on socks	10	1	5	*	*	*	*
5 (type VI)	buttoning and zipper handling	9	2	5	10	10	8	5
6 (type VI)	buttoning and zipper handling	8	3	5	*	*	*	*

Note: \*: Data were not obtained as the participant reported not using the AT

## Discussion

Children and adolescents with MPS, the limitation of mobility caused by the accumulation of glycosaminoglycans in tissues and joints, causes a loss in the ability to perform occupational activities, especially related to activities of daily living (ADLs), especially those requiring fine movements (eg: buttoning) or of large amplitudes (brush hair) [11–15]. It is widely discussed in the literature that progressive musculoskeletal impairment, found regardless of the type of MPS, impacts occupational performance. Studies show that joint stiffness, common in MPS, and even MPS IV-A-specific ligament laxity and muscle weakness, as well as carpal tunnel syndrome and Dupuytren's contractures, all contribute to important limitation in self-care activities such as eating, dressing and personal hygiene [14, 16,17,18]. From the knowledge of body structure and function deficiencies related to self-care activities, it is possible to establish intervention priorities and select better strategies to be used, in order to enhance occupational performance. Among the intervention strategies, AT is a possibility of occupational therapist resource for the promotion of functionality [10].

Although the entire sample showed impairment in the area of dressing, the choices of tasks for making the AT were diverse and did not show a pattern by MPS type. This is because each individual sees itself in a way, and different activities may be a priority for one but not to the other. The activities that a person chooses to engage in are full

of meaning and purpose and are related to their roles and how they relate to the world/environment [19], and therefore each individual attaches meaning and importance to each task of your day to day, like doing one activity is more important than performing another. With the application of COPM, besides allowing the choices of self-care activities that are significant for individuals, it was possible to measure the importance of the activity and quantify its performance and satisfaction. This is because according to the COPM theory was developed occupational performance is viewed as a subjective individual experience [20].

As much as it is not possible to make inferences between participants and their scores, it is possible to say that in the initial assessment of occupational performance, the average among participants was 2.5 points and in the revaluation, an improvement of the results was observed with an average of 7,25 points (minimum value of 4 and maximum of 10) There was also some improvement in the performance rate of activities, in the initial rating the group average was 4 (minimum 2 and maximum of 5) and in the revaluation the average value was 8.75 points (minimum 7 and maximum of 10). According to Carswell (2004), the variation found from 2 or more points in the COPM can be considered a clinically significant intervention [21]. That said, there was an improvement in the occupational performance of individuals with MPS, based on assistive technology, thus seeking to increase the independence of these individuals.

With these changes presented in a significant way, it is possible to suggest that the higher the performance in performing self-care activities, the better the satisfaction in performing it, as seen in the work of Mildner et al., 2017, where the use of AT was described as significant in another health condition [22]. According to Persson et al. (2014) changes in occupational performance are associated with changes in psychosocial functioning and psychological well-being of individuals [23]. Regarding the non-use or abandonment of AT devices by users (occurred with two participants), Costa and collaborators (2015) conducted a literature review on the reasons that led individuals to abandon their resources. The most quoted factors were: problems with the user's physical state; lack of information and training from both professionals and users; pain; functional limitations; preference for another resource or use of remaining capacities [24]. Among the factors mentioned, only the "preference of using remaining capacities" was found in this paper. In addition to this factor it was also quoted "lack of user motivation" and "lack of device functionality".

Regarding AT, social acceptance is an important variable that permeates the decision of the user or his family to use the resource, because even if a certain resource improves the quality of life and occupational performance, but represents a negative social connotation and stigmatizing, the user tends to abandon it. If there is no support or encouragement from family members or if the device is viewed as a validation of being sick/being different (by the individual or family members) the chances of abandonment may be high [24–26].

## Conclusion

AT has become an important occupation therapeutic resource for children and adolescents with problems in performing activities of daily living, such as MPS, increasing their autonomy and personal satisfaction. Thus, we highlight the importance of investing in future research in AT field focusing on occupational performance, especially self-care of individuals with MPS to then guide the intervention and occupational therapeutic care.

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