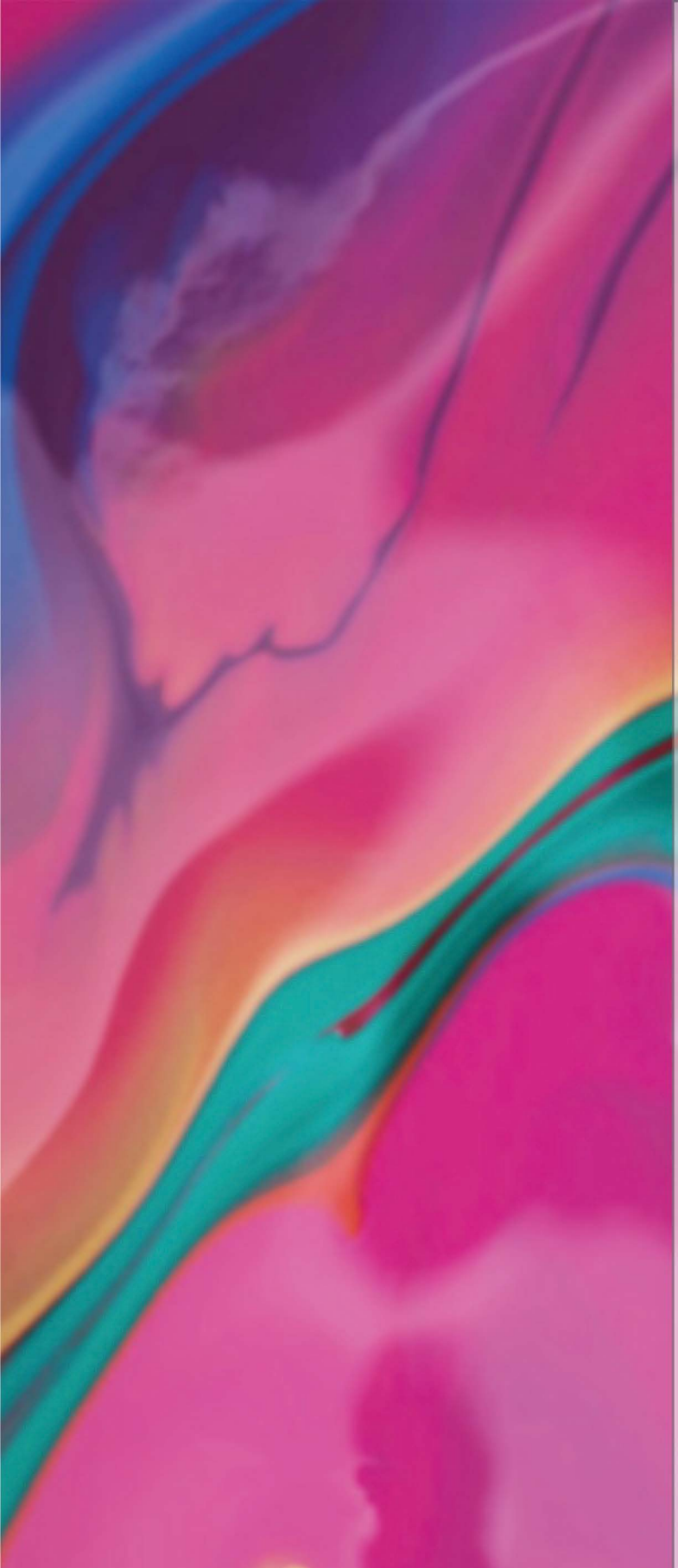

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FAKULTET ZA SPECIJALNU
EDUKACIJU I REHABILITACIJU

UNIVERSITY OF BELGRADE
FACULTY OF SPECIAL EDUCATION
AND REHABILITATION

11.

MEĐUNARODNI NAUČNI SKUP „SPECIJALNA EDUKACIJA I REHABILITACIJA DANAS”

11th

INTERNATIONAL SCIENTIFIC CONFERENCE “SPECIAL EDUCATION AND REHABILITATION TODAY”

ZBORNIK RADOVA

PROCEEDINGS

Beograd, Srbija
29-30. oktobar 2021.

Belgrade, Serbia
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MAPPING HAND FUNCTION IN RARE NEURODEVELOPMENTAL DISORDERS

Andrea Berencsi[**1], Enikő Botló[1], Dóra Fáczán[1], Ilona Kovács[2], Tibor Vámos[1,3]

[1]*Eötvös Loránd University Bárczi Gusztáv, Faculty of Special Needs Education, Hungary*
[2]*Pázmány Péter Catholic University, Laboratory for Psychological Research, Hungary*
[3]*National Institute for Medical Rehabilitation, Hungary*

Introduction: *Appropriate level of hand function is a key to participation in daily living activities, education and social life. Mapping functional abilities related to hand may be challenging but important for ensuring participation in rare neurodevelopmental disorders. We previously showed specific patterns of motor control and learning capacity of the hand in Williams syndrome, a genetically originated neurodevelopmental disorder that involves intellectual disability and motor deficits.*

Aim: *Our aim in the present study was to further map the functional motor skills related to daily living activities and possible sensory dysfunction related to the hand in this rare neurodevelopmental disorder.*

Method: *Participation in activities related to hand function was assessed by the Jebsen-Taylor Hand Function Test. Maximum motor speed in terms of index finger tapping and somatosensory function in terms of two-point discrimination and position sense were assessed.*

Results and conclusion: *Descriptive data analysis revealed that participation in the daily living activities shows difficulties for individuals with Williams syndrome in all domains. Moreover, somatosensory deficits and limitations in motor speed may accompany functional challenges. We also found that the Jebsen-Taylor Hand Function Test was appropriate to use and is a promising tool for daily living activity assessment in the case of mild and moderate intellectual disability with the exception of the "writing" subtest. Regarding somatosensory testing, two-point discrimination test was not applicable for all participants position sense. Our results support the need for further establishment of the relationship between neurophysiological, sensory and motor functional characteristics related to hand.*

Keywords: *fine motor function, hand, somatosensory, Williams syndrome, rare disease*

** berencsi.andrea@barczi.elte.hu

INTRODUCTION

About half of the persons living with rare diseases are schooled without any special support (Linertová et al., 2019). According to the European definition, a rare disease affects of 5 out of 10,000 persons or less and is life-threatening or chronic conditions. The high cumulative number of affected individuals causes recent research to urge the need for mapping special needs of persons with disability due to rare diseases to ensure participation and quality of life (Paz-Lourido et al., 2020). Appropriate hand function is fundamental for cognitive and sensory development and also for participation in education, activities of daily living and quality of life (McLean et al., 2018). Therefore, a major issue in rehabilitation is the promotion of hand function recovery (Knutson et al., 2007).

We have previously shown that in a rare neurodevelopmental disease, namely in Williams syndrome (WS), motor function and learning capacity show differential pattern compared to typical development. In these studies we looked at specific traits in hand function and learning related to hand function. Our results showed that participants with this neurodevelopmental disorder have impaired fine motor function both in terms of speed and accuracy. Prolonged learning was able to improve accuracy but not speed in a fine motor task (Berencsi et al., 2016). We also found that motor learning capacity is related to a specific sleep parameter suggesting distinct neurophysiological processes in the background of motor learning dysfunction (Berencsi et al., 2016).

The co-occurrence of somatosensory and motor impairments is broadly acknowledged in the common neurodevelopmental disorders such as cerebral palsy (Curry & Exner, 1988), autism spectrum disorders (Oldehinkel et al., 2019), attention deficit hyperactivity disorder (Mulligan, 1996) and Down syndrome (Bruni et al., 2010). While mapping fine motor function, the sensory side of movement is considered to be an important but rarely assessed contributing factor in rare neurodevelopmental disorders. Furthermore, assessing participation in daily living activities (ADL) involving hand function in rare diseases also need to be addressed.

AIM

Our aim in the present study was to further map the functional motor abilities and possible sensory dysfunction related to the hand in the rare neurodevelopmental disorder of WS.

METHOD

Participants were 15 individuals diagnosed with WS (9 males and 6 females, age 13-39 years, 2 left-handed).

Variables

Participation in ADL activities was approached by the *Jebsen-Taylor Hand Function Test* a standardised upper limb function test package consisting of seven subtests in everyday activities (n=14). The test measures the time required to perform tasks in the non-dominant and dominant hand. The standard test battery was translated into Hungarian by our research team at ELTE. Here we used a "gestural" version of its translation in WS due to the accompanying intellectual disability. The test was previously reported to be appropriate in intellectual disability (Zikl et al., 2012).

Somatosensory function (n=13) was assessed by the means of *position sense* in the wrist and fingers and by examining *static and dynamic two-point discrimination threshold(mm)* on the palmar surface of the distal phalanx of the index finger.

Maximum motor speed (n=13) in terms of index finger tapping speed (taps/s) was assessed by a self-developed data glove connected to a laptop computer (Berencsi et al., 2016).

Data was collected in the summer camp of the Hungarian Williams Syndrome Association.

ANALYSIS

Due to a low number of participants descriptive data analysis was performed in each domain and compared to typically developing population (TD) data where available.

RESULTS

Jebsen-Taylor Hand Function Test

Out of the seven subtests of Jebsen-Taylor Hand Function Test six were performed by all participants. The 7th "Writing" subtest was presented as an optional task, as handwriting does not always develop in WS. For all subtests tested, lower performance was observed in compared to age and gender standard times (included in the test battery) for both hands (Figure 1). For the dominant hand, the highest performance was in the subtest "Turning over cards" and the lowest in the subtest of "Simulated feeding". Great individual variability was present between performance of WS individuals (Figure 2) Task causing problems and being invincible with the highest prevalence is writing, a fundamental task in educational settings.

Figure 1

Participation in ADL functions assessed by the Jebsen-Taylor Hand Function Test. Standard age and gender matched values in TD provided by the test battery are shown in yellow and orange colour for dominant and non-dominant hand respectively. In WS, performance in both the dominant (grey) and non-dominant (blue) hand were below TD performance.

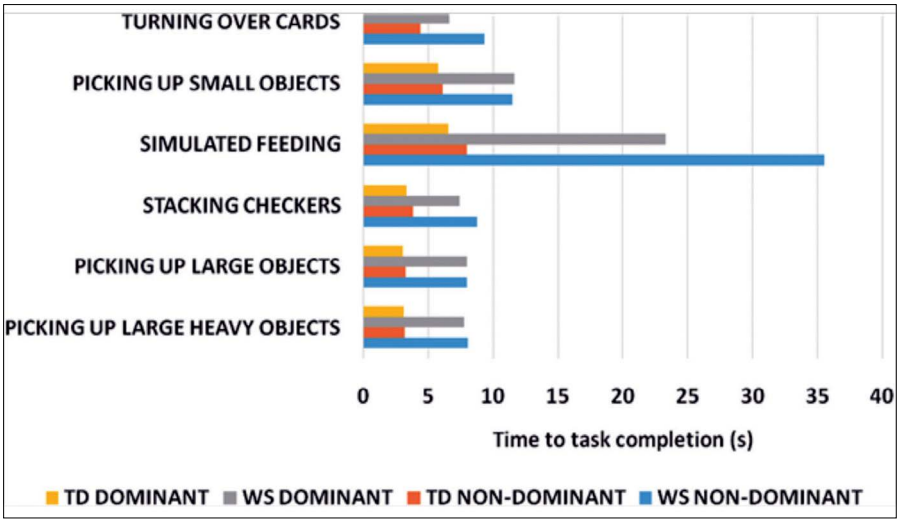
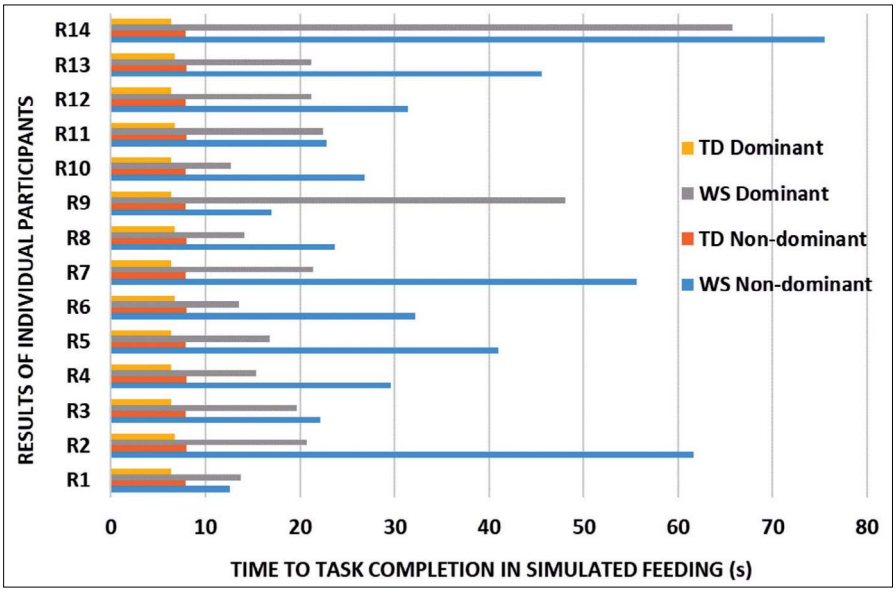


Figure 2

Performance in the simulated feeding subtest of Jebsen-Taylor Hand Function Test. Standard age and gender matched values in TD provided by the test battery are shown in yellow and orange colour for dominant and non-dominant hand respectively. In WS, performance in both the dominant (grey) and non-dominant (blue) hand were below TD performance

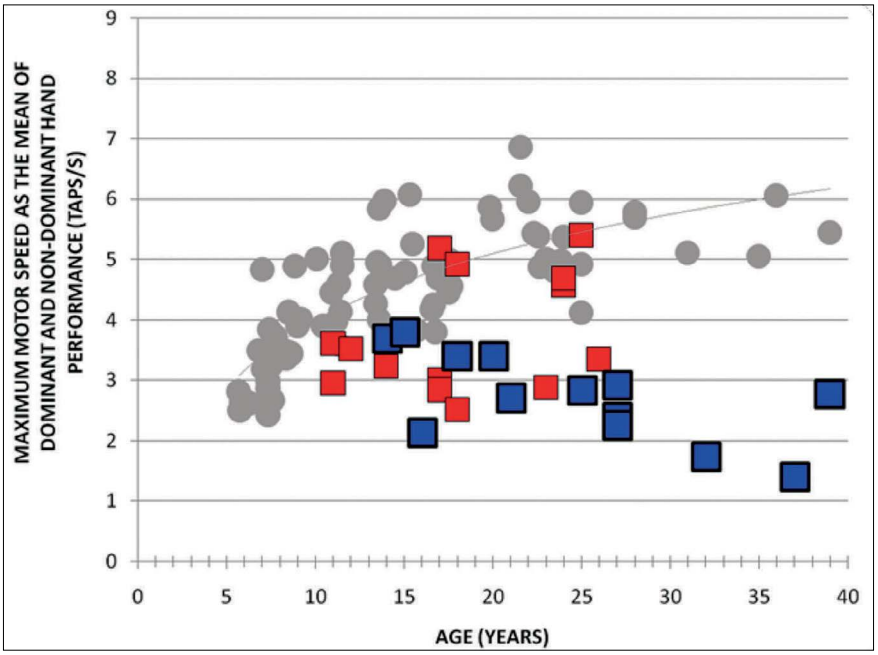


Maximum motor speed

In individuals with WS, the decline in fine motor speed occurs at an earlier age than in typically developing individuals, particularly over the age of 30 (Figure 3, in blue). This result is in line with our previous result from 2009/2010 (Figure 3, in red). Finger tapping speed increases about the age of 40 years in TD than declines with aging (Bartzokis et al., 2010). Early decrease of finger tapping speed may support previous neurophysiological findings suggesting early aging in WS (Gombos et al., 2017).

Figure 3

Maximum motor speed in typical development (grey dots) and in WS (2009/2010- red squares and 2019- blue squares).



Somatosensory function of the hand

When testing the position sense, eleven of the thirteen participants were able to match the position of the wrist, including nine who were able to match the position of the fingers. Due to the low number of somatosensory tests completed successfully, no correlation was found with fine motor function at the trend level.

In the tactile domain, static and dynamic two-point discrimination testing was applicable only to a subgroup of the participants. In the static two-point discrimination threshold test, seven out of nine individuals fall into the 1-5 mm normal zone and one individual falls into the more favourable group. The responses of the remaining participants could not be assessed due to inconsistent responses. The dynamic two-point discrimination threshold tasks yielded results with a similar distribution. The remaining participants could not answer consequently to testing questions. Our results suggests that in neurodevelopmental disorders

with intellectual disability, the replacement of two-point discrimination test needs to be considered by another valid tool for tactile function examination. On the other hand, proprioceptive function in terms of position matching and reporting movement direction with eyes closed was an applicable measure.

CONCLUSION

In our recent pilot study, we demonstrated that impaired fine motor function in WS is accompanied with problems in activities of daily living related to hand function.

Results showed that dynamic proprioceptive cues and processing have no deficit in our cohort. On the contrary, static proprioceptive cues and processing as assessed by position matching may be more difficult to approach, and suggests somatosensory impairment in WS. While co-occurrence of motor and sensory dysfunction is clearly a case in these finding, further research is needed to map the interrelation of somatosensory and motor function and dysfunction of the hand in rare neurodevelopmental disorders to promote participation.

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REFERENCES

Bartzokis, G., Lu, P. H., Tingus, K., Mendez, M. F., Richard, A., Peters, D. G., Oluwadara, B., Barrall, K. A., Finn, J. P., Villablanca, P., Thompson, P. M., & Mintz, J. (2010). Lifespan trajectory of myelin integrity and maximum motor speed. *Neurobiology of Aging*, *31*(9), 1554-1562. https://doi.org/10.1016/j.neurobiolaging.2008.08.015

Berencsi, A., Gombos, F., & Kovács, I. (2016). Capacity to improve fine motor skills in Williams syndrome. *Journal of Intellectual Disability Research*, *60*(10), 956-968. https://doi.org/10.1111/jir.12317

Bruni, M., Cameron, D., Dua, S., & Noy, S. (2010). Reported sensory processing of children with Down syndrome. *Physical & Occupational Therapy in Pediatrics*, *30*(4), 280-293. https://doi.org/10.3109/01942638.2010.486962

Curry, J., & Exner, C. (1988). Comparison of tactile preferences in children with and without cerebral palsy. *American Journal of Occupational Therapy, 42*(6), 371-377. https://doi.org/10.5014/ajot.42.6.371

Gombos, F., Bódizs, R., & Kovács, I. (2017). Increased overall cortical connectivity with syndrome specific local decreases suggested by atypical sleep-EEG synchronization in Williams syndrome. *Scientific Reports*, *7*(1), 1-13. https://doi.org/10.1038/s41598-017-06280-2

Knutson, J. S., Harley, M. Y., Hisel, T. Z., & Chae, J. (2007). Improving hand function in stroke survivors: A pilot study of contralaterally controlled functional electric

stimulation in chronic hemiplegia. *Archives of Physical Medicine and Rehabilitation*, *88*(4), 513-520. https://doi.org/10.1016/j.apmr.2007.01.003

Linertová, R., González-Guadarrama, J., Serrano-Aguilar, P., Posada-De-la-Paz, M., Péntek, M., Iskrov, G., & Ballester, M. (2019). Schooling of children with rare diseases and disability in Europe. *International Journal of Disability, Development and Education*, *66*(4), 362-373. https://doi.org/10.1080/1034912X.2018.1562159

McLean, B., Blakeman, M., Carey, L., Ward, R., Novak, I., Valentine, J., Blair, E., Taylor, S., Bear, N., Bynevelt, M., Basc, E., Rose, S., Reid, L., Pannek, K., Angeli, J., Harpster, K., & Elliott, C. (2018). Discovering the sense of touch: Protocol for a randomised controlled trial examining the efficacy of a somatosensory discrimination intervention for children with hemiplegic cerebral palsy. *BMC Pediatrics*, *18*(1), 252. https://doi.org/10.1186/s12887-018-1217-5

Mulligan, S. (1996). An analysis of score patterns of children with attention disorders on the Sensory Integration and Praxis Tests. *The American Journal of Occupational Therapy: Official Publication of the American Occupational Therapy Association*, *50*(8), 647-654. https://doi.org/10.5014/ajot.50.8.647

Oldehinkel, M., Mennes, M., Marquand, A., Charman, T., Tillmann, J., Ecker, C., Dell'Acqua, F., Brandeis, D., Banaschewski, T., Baumeister, S., Moessnang, C., Baron-Cohen, S., Holt, R., Bölte, S., Durston, S., Kundu, P., Lombardo, M. V., Spooren, W., Loth, E., … EU-AIMS LEAP group. (2019). Altered connectivity between cerebellum, visual, and sensory-motor networks in autism spectrum disorder: Results from the EU-AIMS longitudinal European autism project. Biological Psychiatry. *Cognitive Neuroscience and Neuroimaging*, *4*(3), 260-270. https://doi.org/10.1016/j.bpsc.2018.11.010

Paz-Lourido, B., Negre, F., de la Iglesia, B., & Verger, S. (2020). Influence of schooling on the health-related quality of life of children with rare diseases. *Health and Quality of Life Outcomes*, *18*(1), 109. https://doi.org/10.1186/s12955-020-01351-x

Zikl, P., Zajickova, B., & Tomaskova, M. (2012). Functional motor abilities of the upper extremities in children with mild intellectual disabilities. *Procedia – Social and Behavioral Sciences*, *69*, 2068-2075. https://doi.org/10.1016/j.sbspro.2012.12.166