The Role of Neurohabilitation in Achieving Maximal Outcomes in Individuals with Rett Syndrome

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Recent advances in pharmaceutical and genetic research are providing great hope for individuals living with Rett syndrome and their families. There are several studies being conducted worldwide investigating drug therapy to improve clinical symptoms, or to correct or replace the mutated gene. As rehabilitation professionals and teachers, who also have family members living with Rett syndrome, we follow these developments with great hope for our loved ones as well as for the entire Rett syndrome community.

We concur with a recent post by Drs. Alan Percy and Walter Kaufman, where they emphasize that no single therapy is likely to yield the same result in all individuals with Rett syndrome. This can be attributed to a number of factors, including age, medical status, type of mutation, and clinical presentation. Individuals with Rett syndrome vary widely in their skill levels. Some individuals walk, while others are nonambulatory. Some have preserved hand function and can participate in feeding or dressing, while others require assistance in all activities of daily living. Some individuals produce words, many use augmentative communication systems of varying complexity, while others have great difficulty in communicating basic needs. Currently, multiple therapies such as physical, occupational, or speech therapy, help the neurological system function at maximum capacity and can result in improved quality of life. These and other related therapies have anecdotally been shown to be effective in promoting improved motor control and communication. What is missing is knowledge about the exact combination of sensory experiences and extent of practice required to promote skill development. This is a focus of neurohabilitative research.

Neurohabilitation trains the individual to acquire skills that were not previously within their repertoire and to recover/minimize further loss of motor skills. It is a necessary part of research in Rett syndrome and is actively being studied in multiple settings across the US and internationally. This type of research will help substantiate and further define the therapies for best practice so that real evidence, rather than just anecdotal stories, will define treatment protocols.

Despite the variability of skill sets in those with Rett syndrome, research is showing that, although at different paces, each individual can learn if we identify the specific motivators and learning styles

needed to aid their successful development. Evidence is being gathered on the effects of various therapies on individuals with Rett syndrome, and early results are promising (for examples, see Downs et al., 2018; Fabio et al., 2018, Mraz et al., 2016). Results from these studies suggest that components of best practice are being clarified and with continued research can be used to guide the development of neurohabilitation therapeutic treatment plans for individuals with Rett syndrome.

Neurohabilitation results are encouraging and indicate that we can help individuals with Rett maximize their capacities now. We are so excited about the outcomes of recent publications and the potential for blending neurohabilitation with other types of genetic and pharmaceutical treatments. We will continue to advocate for continued funding of research studies, across the lifespan, and across multiple functional domains, to improve learning and the quality for life of individuals with Rett syndrome.

References

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