QUALITY OF LIFE IN INDIVIDUALS WITH SYNDROMIC AUTISM SPECTRUM DISORDERS

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Background: Quality of life (QOL) instruments can provide insight into caregiver preferences in neurodevelopmental disorders. While such studies have been conducted for idiopathic autism, few have focused on syndromic Autism Spectrum Disorders (ASDs). Here we evaluate the QOL in children with three syndromic ASDs: Phelan-McDermid syndrome (PMS), Rett syndrome (RTT), and SYNGAP1 related intellectual disability (SYNGAP1-ID).

Materials/Methods: A cross-sectional study design was implemented, and a standardized survey, Pediatric Quality of Life Inventory (PedsQL), was administrated as an online questionnaire to guardians of children age 2-18 with PMS (n=213), RTT (n=148), and SYNGAP1-ID (n=30). PedsQL segment assessed children's HRQOL while Family Quality of Life Scale (FQOL) component assessed parents' life satisfaction with their nuclear family.

Results: The greatest impairments for syndromic ASDs across all categories was in social and physical functioning. The greatest impairment for PMS was in social functioning while the greatest impairment for RTT was physical functioning. When directly comparing each domain between PMS and RTT, there was greater reported dysfunction in physical functioning of girls with RTT (t-test $p<0.01$) and for all other domains there was no significant difference. The only significant differences found between groups in FQOL survey were in "dental care received" (t-test $p<0.05$) showing higher satisfaction amongst the PMS group (PMS M = 3.85; Rett M = 3.55), and "perceived support received by family member with disability" (t-test $p<0.05$) also showing higher satisfaction amongst the PMS group (PMS M = 3.39; Rett M = 3.11).

Conclusions: Impairments in cognitive and language development, along with deficits in adaptive behavior, make social functioning challenging for PMS patients. Diminishing mobility, scoliosis, osteoporosis at a young age, and hypotonia are symptoms of RTT which impair physical functioning, thus physical activity programs tailored to the individual may improve present and future health conditions. Due to the difficulty in obtaining adequate compliancy as well as the possible presence of gastroesophageal reflux, ASD patients are at a higher risk of developing caries than the rest of the pediatric population. Further studies addressing areas of improvement for families of children with syndromic ASDs regarding access to medical care, along with support received by ASD affected may provide insight regarding QOL improvement.