Quality of Life in individuals with Syndromic Autism Spectrum Disorders

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BACKGROUND

Quality of life (QOL) survey 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 (PMD), Rett syndrome (RTT), and *SYNGAP1* related intellectual disability (*SYNGAP1*-ID). PMD is one of the most common monogenic forms of ASD, due to genomic deletions of the *SHANK3* gene. RTT is an X-linked dominantly inherited postnatal neurodevelopmental disorder caused by mutations in the gene *MECP2*, most commonly in girls. *SYNGAP1*-ID is a neurological disorder caused by mutations in the *SYNGAP1* gene, which provides instructions for making a protein called SynGAP that plays an important role in nerve cells in the brain.

PURPOSE

To determine the quality of life in individuals with genetically defined ASDs or caregivers of those with ASDs using online anonymous surveys.

METHODS

- A cross-sectional study design was implemented and the survey was administrated as an online questionnaire.
- Standardized PedsQL survey was answered by guardians of children age 2-18 with Phelan-McDermid Syndrome (n=213), Rett Syndrome (n=148), and *SYNGAP1* related intellectual disability (n=30).
- PedsQL segment assessed children's HRQOL while Family Quality of Life Scale (FQOL) assessed parents' life satisfaction within their nuclear family.

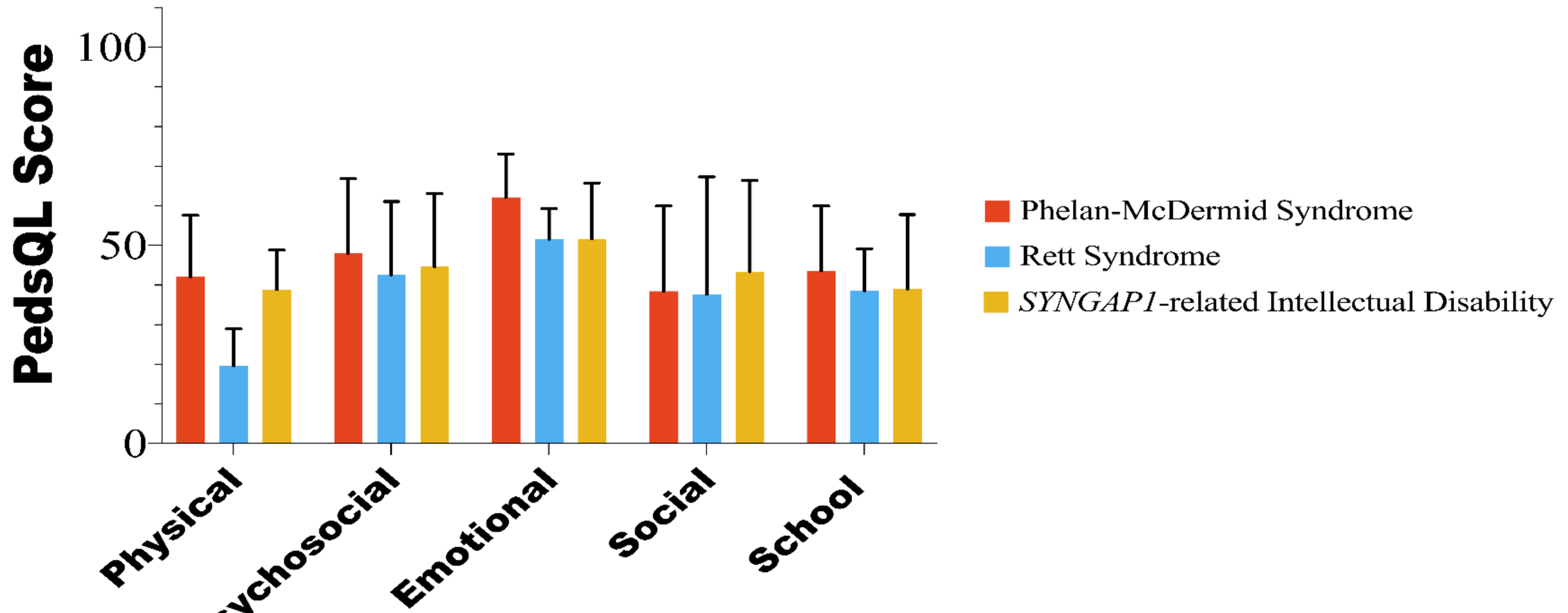

Fig 1: Cross comparison analysis of functioning domains between PMD and RTT groups revealed greatest disparity in social functioning domain for PMD group (T-Test p<0.05) and in the physical functioning domain for the RTT group (Test-T p<0.01).

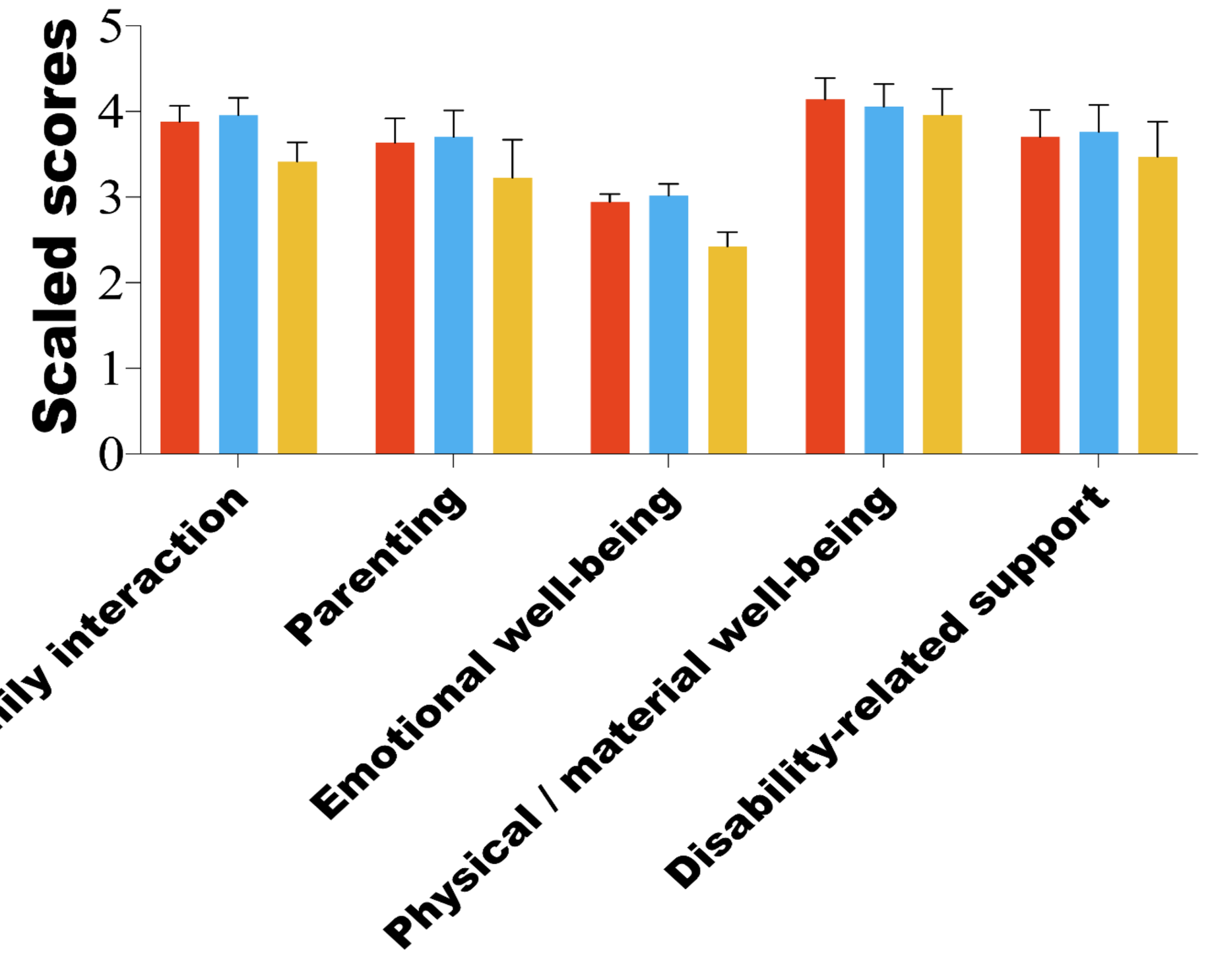

Fig 3: Family Quality of Life scale. No significant differences identified between PMD and RTT syndrome, except for receiving dental care needed (T-Test p<0.05) and family member with disability receiving support to accomplish goals at school and workplace, showing higher satisfaction in Phelan-McDermid syndrome. (T-Test p<0.5).

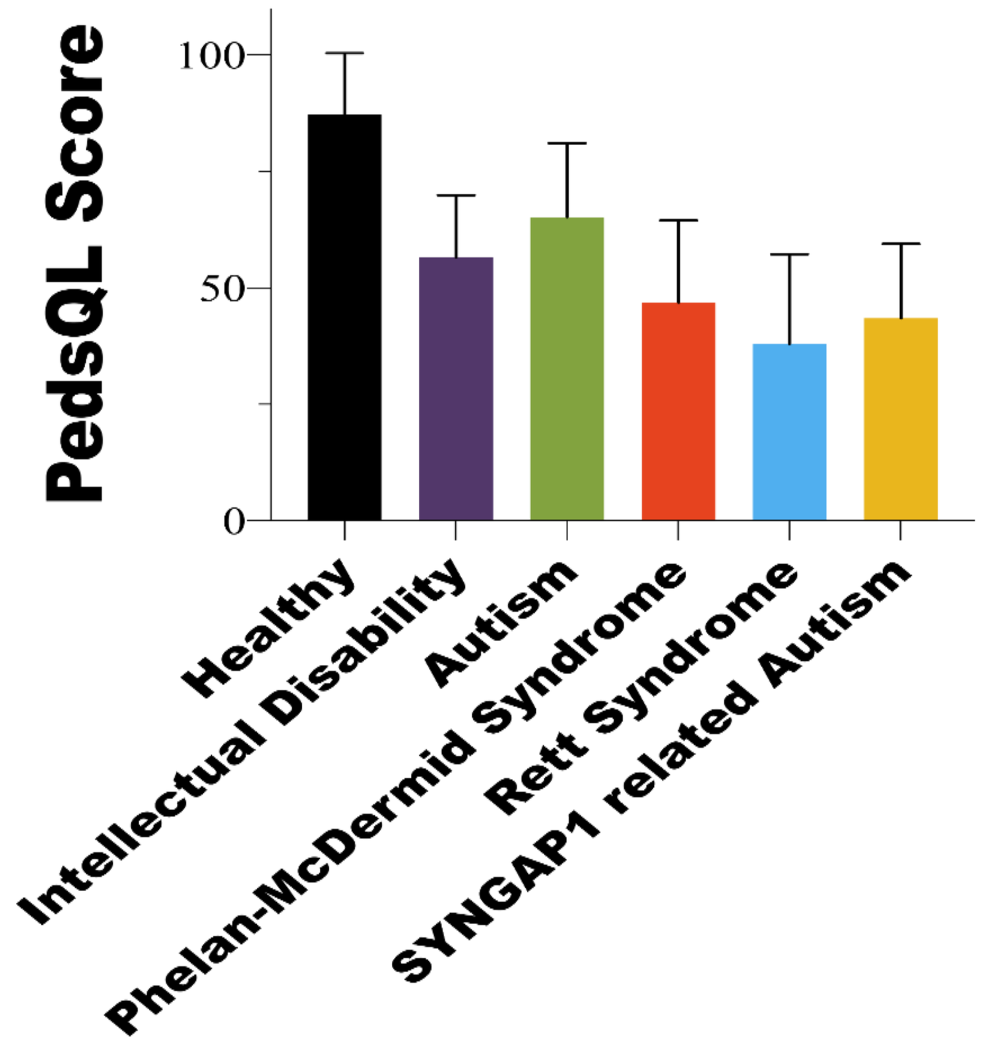

Fig 2: Total quality of life score in syndromic autisms compared with other chronic disorders in children. Children with RTT syndrome score the lowest.

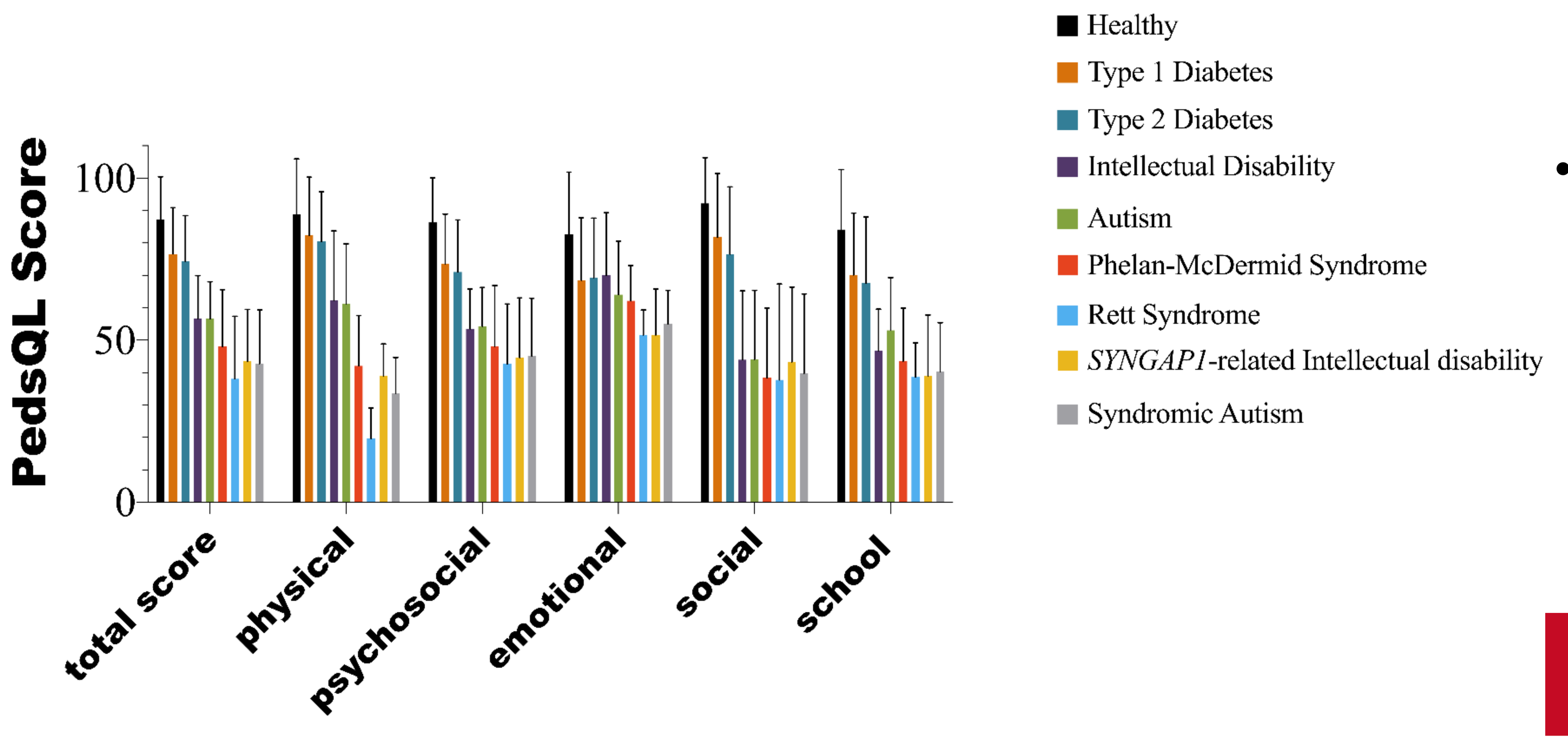

Fig 4: Subscale scores of PedsQL across disorders show that syndromic ASDs score significantly lower than other chronic childhood illnesses such as Diabetes type 1 and type 2.

Table 1 Peds QL items administered to the Phelan-McDermid and Rett syndrome groups

PedsQL variables		Total Phelan-McDermid group (n = 213)		Total Rett syndrome group (n = 148)		Group differences
Scores converted to 0 – 100 from (min = 1 ; max = 5) Likert scale		M	(SD)	M	(SD)	P-value (for two-tailed t test)
Physical functioning (problems with...)						
1	Walking more than one block	61.03	15.53	23.64	4.43	<0.0001
2	Running	47.64	8.40	15.20	3.39	<0.0001
3	Participating in sports activity or exercise	34.43	3.55	15.54	2.93	<0.0001
4	Lifting something heavy	37.32	4.15	13.17	4.16	<0.0001
5	Taking a bath or shower by him or herself	19.95	4.86	12.24	4.80	<0.05
6	Doing chores around the house	24.06	1.79	10.54	4.19	<0.005
7	Having hurts or aches	54.97	7.05	33.90	6.05	<0.0001
8	Low energy level	57.34	7.05	32.93	6.39	<0.0001
Emotional functioning (problems with...)						
9	Feeling afraid or scared	67.18	11.19	52.73	8.54	<0.0001
10	Feeling sad or blue	64.33	9.75	51.19	9.80	<0.0001
11	Feeling angry	59.38	8.29	55.27	10.38	NS
12	Trouble sleeping	45.18	5.14	39.11	5.52	NS
13	Worrying about what will happen to him or her	74.63	22.09	59.64	9.68	<0.0001
Social functioning (problems with...)						
14	Getting along with other children	50	5.92	60.95	11.16	<0.01
15	Other kids not wanting to be his or her friend	41.99	4.31	42.41	4.42	NS
16	Getting teased by other children	65.70	11.61	70.48	13.21	NS
17	Not able to do things that other children his or her age can do	11.49	1.03	4.28	0.40	<0.005
18	Keeping up when playing with other children	23.09	1.76	10.44	1.96	<0.0005
School functioning (problems with...)						
19	Paying attention in class	28.29	3.16	30.29	4.98	NS
20	Forgetting things	33.58	1.96	37.69	5.74	NS
21	Keeping up with schoolwork	33.89	4.56	26.51	3.53	NS
22	Missing school because of not feeling well	64.65	11.76	51.47	6.30	<0.005
23	Missing school to go to the doctor or hospital	57.59	7.15	46.92	6.33	<0.005

Table 1: PedsQL Health-Related Quality of Life revealed that across all quality of life categories, the greatest impairment for PMS was found in social functioning (M = 38.45), followed by physical functioning (M = 42.09), school functioning (M = 43.60), and emotional functioning (M = 62.14). The greatest challenges faced by children with Rett syndrome appear to lie in the domain of physical functioning (M = 19.65), followed by social functioning (M = 37.71), school functioning (M = 38.58), and emotional functioning (M = 51.59).

Table 2 Family QL items administered to the Phelan-McDermid and Rett Syndrome groups

PedsQL variables		Total Phelan-McDermid group (n = 213)		Total Rett group (n = 148)		Group differences
Likert scale range for analysis (min = 1 ; max = 5)		M	(SD)	M	(SD)	P-value (for two-tailed t test)
Physical functioning (problems with...)						
1	My family enjoys spending time together	3.91	0.18	4.02	0.17	NS
2	My family members talk openly with each other	3.59	0.16	3.51	0.14	NS
3	Our family solves problems together	2.86	0.09	2.93	0.10	NS
4	My family members support each other to accomplish goals	3.05	0.09	3.20	0.14	NS
5	My family members show that they love and care for each other	3.22	0.09	3.25	0.12	NS
6	My family is able to handle life's ups and downs	4.06	0.18	3.83	0.15	NS
7	My family members help the children be independent	3.83	0.16	3.76	0.17	NS
8	My family members help the children with school work and activities	3.81	0.17	3.90	0.18	NS
9	My family members teach the children how to get along with each other	2.98	0.07	3.03	0.09	NS
10	Adults in our family teach the children to make good decisions	3.79	0.18	3.84	0.17	NS
11	Adults in my family know other people in my children's lives (friends, teachers, etc.)	3.79	0.18	3.95	0.18	NS
12	Adults in my family have time to take care of the individual needs of every child	4.23	0.22	4.33	0.09	NS
13	My family has the support we need to relieve stress	2.88	0.05	2.89	0.17	NS
14	My family members have friends or others who provide support	3.97	0.21	4.06	0.19	NS
15	My family members have some time to pursue our own interests	4.27	0.22	4.21	0.23	NS
16	My family has outside help available to us to take care of special needs of all family members	3.75	0.13	3.79	0.05	NS
17	My family members have transportation to get to the places they need to be	3.79	0.16	3.92	0.20	NS
18	My family gets medical care when needed	3.70	0.17	3.82	0.20	NS
19	My family has a way to take care of our expenses	3.40	0.12	3.56	0.15	NS
20	My family has dental care when needed	4.25	0.2	4.02	0.17	<0.05
21	My family feels safe at home, work, school, and in our neighborhood	4.37	0.23	4.42	0.24	NS
22	My family member with a disability has support to accomplish goals at school or at workplace	3.86	0.16	3.74	0.18	<0.05
23	My family member with a disability has support to accomplish goals at home	3.87	0.17	3.89	0.18	NS
24	My family member with a disability has support to make friends	3.22	0.09	3.33	0.09	NS
25	My family has good relationships with the service providers who provide services and support to our family members with a disability	3.83	0.14	4.07	0.19	NS

Table 2: PedsQL Family Quality of Life revealed that no significant differences were found across groups in regards to family satisfaction with the exception of dental care received (T-Test = p<0.05), and perceived support received by family member with disability to accomplish goals at school or workplace (T-Test = p<0.05), showing higher satisfaction amongst the PMS group.

RESULTS

- The survey revealed that across all categories for QOL, the greatest impairments for syndromic ASDs was in social and physical functioning.
- When parsing the data based on genetic diagnosis, the greatest impairment for PMD was in social functioning while the greatest impairment for RTT was physical functioning.
- When directly comparing each domain between PMD 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 PMD group (PMD M = 3.85; RTT M = 3.55), and "perceived support received by family member with disability" (T-Test p<0.05) also showing higher satisfaction within PMD (PMD M = 3.39; RTT M= 3.11).

CONCLUSION

Impairments in cognitive and language development, along with deficits in adaptive behavior, make social functioning challenging for PMD 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.

ACKNOWLEDGEMENTS

- We are indebted to the individuals who participated in the study
- Funding through the Robbins Foundation