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Analysis of the quality of life in families with individuals diagnosed with williams syndrome in Pakistan

[1]Marwa Aslam, [2]Dr Muhammad Sami Bilal, [3]Dr Muhammad Kamran Tariq, [4]Mobeen Ali, [5]Umar Khan, [6]Kamran Safdar

[1]Mayo Hospital Lahore
[2]Assistant Professor of Psychiatry, CPSP Karachi
[3]SR Medicine, Shahida Islam Medical Complex, Lodhran, Pakistan
[4]PIMS
[5]PIMS
[6]UHS Lahore

ABSTRACT:
OBJECTIVE: A developmental disease called Williams Syndrome is characterized by a varied intellectual impairment. Williams Syndrome patients need the assistance of numerous educational and medical experts throughout their lifetime. The effects of this handicap, particularly on families, are not well understood, but, in the local context. Knowing the degree of quality of life (QOL)as stated by families with Williams Syndrome (WS) was the goal.
METHODS: The kids life Scale was used to examine 33 families that made up the sample. Their young people ranged in age from four to twenty. Emotional health, physical health, material health, personal growth, interpersonal relationships, self-determination, social inclusion, and rights were the eight key quality-of-life areas that were assessed.
RESULTS: The data gained showed that a person's quality of life might be affected by a variety of factors to varying degrees, regardless of the severity and existence of an intellectual handicap. No variations in the quality of life were found that were statistically significant, however, there were disparities in reliance levels in the self-determination subarea (p-value <.05).
CONCLUSIONS: Based on these findings, we examined how families and their surroundings could be affected socially and emotionally.
KEYWORDS: Williams syndrome, quality of life, emotional health

INTRODUCTION: A de novo loss of 26–28 genes from chromosome 7 q11.23 causes Williams Syndrome (WS), a neurodevelopmental condition that affects one in every 7500 live births [1]. Clinically, WS is distinguished by a few recognizable facial features, a mild to moderately severe intellectual disability (ID), with marked deficits in some areas (psychomotricity, visuospatial assimilation, limited attention ability, ability to focus), and relatively preserved abilities in others (language and musicality), a sociable demeanor, sporadic hypercalcemia in infancy, and vascular disease with supravalvular aortic dissection [2]. Their ID denotes an average Intelligence Quotient (IQ) of 55–60 points, with a possible range of 40–100 points [3]. This very large variation necessitates careful and specific adaptation of any provided therapy to each person with WS [4].

In terms of behavioral characteristics, WS patients often display high levels of anxiety, specific phobias, and attention deficit disorder (ADD), which may lower quality of life (QoL), particularly in adults [5]. Parents see them as being vivacious, upbeat, well-balanced, bashful, overly sociable, and energetic. They don't exhibit any gender or age disparities. All of these WS prototype clinical traits may have an impact on the daily activities of WS patients as well as their surroundings, which affects their quality of life [6].

The word "quality of life" refers to a notion that, depending on the era, social group, and culture to which a person belongs, may be seen in considerably different ways [7]. Nonetheless, the majority of methods have highlighted the significance of supporting the biopsychosocial character of people and show how this idea is integrated [8]. Based on the previously described biopsychosocial conception, several studies

have addressed the potential effects that having ID may have on a person's surroundings [9]. As a result, it was said that families highlight several areas where their quality of life is lacking and that they may be able to depend on counseling, professionals, or parent groups to help them arrange appropriate activities for their kids [10]. Hence, it would seem that factors influencing the amount of assistance needed by families [11] include the structure of the family, its members' views, and the environment in which they reside.

In the last ten years, the idea of family quality of life (QoL) has been more important as a way to understand the effects of having a family member with some disabilities and to assess the potential effects of the services and assistance that families get [12]. Also, the majority of studies place more emphasis on the family QoL than it does on the QoL of the individual with a handicap. The QoL of a person with an ID, however, obviously has an impact on the QoL of their family [13]. Despite the potential significance that information on this topic may have on families' QoL, research on the QoL of families with boys or girls with WS is almost nonexistent as of yet. As a result, there is no scientific agreement that relatives of WS patients would prefer to speak about what their loved ones might do with the right help rather than what they are incapable of doing [14].

METHODS: In this research, 33 parents of WS-diagnosed kids between the ages of 4 and 20 took part. 26 moms and 7 dads were among them. The parents were between the ages of 30 and 60. There are 10 households with teenagers between the ages of 13 and 20 and 23 families with children between the ages of 4 and 12.

The KidsLife Scale measures the quality of life for kids and teenagers with ID. It assesses individual QoL outcomes in children and adolescents with ID between the ages of 4 and 21. It consists of a series of inquiries concerning visible characteristics of quality of life that may be responded to by an outside observer who is familiar with the kid and has the opportunity to spend a lot of time seeing him or her in various settings. The scale has 96 questions and gives percentile and standard scores for each of the eight major dimensions of quality of life: emotional health, physical health, material health, personal growth, interpersonal relationships, social inclusion, rights, and self-determination. There are four alternatives for each item, but only one must be selected (always, frequently, sometimes, or never). The size, directionality, and statistical significance of the standardized coefficients (which are always greater than 0.70, positive, and significant with p 0.01) provide evidence of convergent validity. In terms of the scale's reliability, the overall score achieved a Cronbach's alpha value of 0.96, while the domain scores varied from 0.80 (physical health) to 0.90. (personal development).

Three factors from the KidsLife scale were used to examine the data: the degree of dependency (moderate, severe, and high); the degree of need (minimal, intermittent, extensive, and generalized); and the degree of handicap (mild, moderate, severe and profound). Family members answer questions on these three factors in the first section of the questionnaire, and the results match the data on personal traits.

Informed consent forms were sent to the mothers and dads of their children They received the written questionnaire to fill out after they signed this form. Each participant completed this questionnaire on their own during a 20–30-minute session. The Kolmogorov-Smirnoff test was used to confirm the sample's normality, yielding a non-parametric test. The U-Mann Whitney test was then used to assess the data. Spearman's Rho test was used to examine the correlation between the variables.

RESULTS: According to the claim, there were no significant disparities among the scale's different subareas' standard scores. There is no evidence to support the null hypothesis that there are significant differences between the subareas, according to the statistical analysis, which revealed that the p-value was larger than 0.05 for all of the subareas.

The information in Table 1 includes the mean scores and standard deviations for each subarea, which provides insight into the scoring distribution. The findings that there are no significant variations between the subareas are further supported by the fact that the mean scores for each subarea are pretty near to one another.

Self-determination, social inclusion, rights, interpersonal relationships, personal growth, and emotional, physical, and financial well-being are among the subareas on the scale. The scale is intended to gauge a person's total well-being, with each subarea concentrating on a different component of well-being.
According to the findings, there were significant differences between parents whose sons or daughters had WS and exhibited a moderate degree of reliance compared to those whose children had WS and exhibited a high level of dependence ($p < 0.05$).
The individuals' ages, which were split into two groups of 4 to 12 and 13 to 20 years old, were also taken into consideration while analyzing the data. For each of the tested domains, there were no differences between the two age groups.
Table 1: Medium QoL domain scores

		Need Level				**Disability Level**				**Dependence Level**		
		1	2	3	4	1	2	3	4	1	2	3
Interpersonal relations	Mean	36	32.8	32.83	31.12	12.81	37	38.66	0	34.36	37.21	37.4
	SD	5.6	4.3	9.1	6.4	7.8	5	6.2		7.6	6.5	7.2
Personal development	Mean	42	33.33	35.16	40	13.19	38.58	40.5	0	35.63	38.92	41.4
	SD	8.5	4.5	9.2	6.9	8.5	5.7	5.3		8.3	6.8	5
Rights	Mean	47	35	36.83	38.37	10.36	39	39.16	0	37.18	40.64	40.4
	SD	1.4	6.8	10.9	5.8	9	6.9	6.5		9.8	5.6	5.7
Material	Mean	43.5	36.66	42.66	41.75	43.08	41.16	44	0	42.81	42.28	42.4
	SD	4.9	5.5	3.8	4.7	4.4	5.5	3		4.2	5.2	4.1
Physical	Mean	40.5	33.86	39.16	39.5	44.08	37.25	41	0	38.72	39.57	41.4
	SD	0.7	5.1	2.6	6.8	3.5	5	6.4		4.5	5.1	5.9
Emotional	Mean	42.5	32	37.16	42.75	38.5	37.33	38.66	0	37.36	37.64	40.8
	SD	2.1	5.7	4.8	6.8	5.1	4.9	8.4		4.2	5.1	7.1
Self-determination	Mean	33.5	27.66	29	29.5	33.16	30.5	27.16	0	31.64	30.85	29.4
	SD	9.2	6.2	6.3	4.6	6.2	4.8	5		5.5	6.8	2.9
Social inclusión	Mean	38.5	29.66	64.66	34.37	37.25	33.41	32.5	0	35.45	33.71	36.2
	SD	4.9	5.8	5.9	5.4	5.4	5	5.3		5.3	4.7	4.3

DISCUSSIONS: The purpose of the current research was to examine families' perspectives on the quality of life (QoL) of persons with WS. A QoL profile created from this data enables help to be arranged on an individual basis [15]. To achieve this, we first adopted the most current understanding of disability, which was defined as the outcome of a person's interaction with their environment since the provision of assistance would materially improve their level of functioning [16]. The gathered statistics were particularly intriguing since they revealed that the existence of the impairment rather than its severity was what defined the family's quality of life. The degree of dependency and need both follow the same logic. Without taking other factors like the level of impairment, dependency, or need into consideration, the existence of WS predisposes families to a certain profile. According to these characteristics, families do not view their QoL to be different, which suggests a more uniform profile for the consequences associated with having WS. It is also important to note that parents' perceptions of their children's evolutionary

development do not vary with time; rather, an agreement on the most obvious challenges prevails in light of the findings.

The findings for the social inclusion domain of the QoL domains showed that, despite increased awareness, social inclusion for these groups is still a problem [17].

Secondly, let's look at what, based on the statistics, could be the key QoL restriction: mastering self-determination, despite the lack of any discernible distinctions. According to research, the person with ID is not consulted when parents make critical decisions about things like education centers or the style of schooling between infancy and adolescence. [18] Our findings, therefore, show that there are some variations in the level of determination depending on the degree of reliance. In a similar vein, the outcomes supplied by families do not line up with those reported in other research studies, which show a strong correlation between decision-making and level of impairment [19]. As a result, the severity of the handicap does not seem to have an impact on the family. This could be because they don't place as much emphasis on the unique distinctions that people with disabilities might contribute. Families see people with WS as people with disabilities in the broadest sense possible; nonetheless, the severity of the condition is unimportant.

A moderate level of satisfaction in these subareas is indicated by the findings of the emotional health, physical health, and material health domains [20]. These findings conflict with those of other studies, which claim that behavioral issues brought on by emotional distress often have a detrimental effect on both individual and family health [21]. A study emphasizes the increased financial burden of families with relatives with ID, which has a detrimental impact on family QoL, about these groups as well. [22] As a consequence, the findings of our study vary from those of other writers. This can be the result of unrecognized cultural and social inequalities.

Once again, research linking this subarea to the degree of handicap does not support the findings provided by parents for the rights component. Parents often mention that children with ID have limited chances and that there isn't enough assistance for them to exercise their rights when the impairment is mild or severe. As a result, to some degree, families continue to see their children as vulnerable, which may sometimes result in overprotection [23].

Regarding these people's personal development, the collected data emphasize that parents often consider that their children engage in a lifelong process of social skill acquisition, which is crucial for personal development and, therefore, for a higher-than-average QoL [24]. Since persons with ID and their families often have fewer relationships and are more vulnerable to the danger of social isolation, the interpersonal interactions dimension outcomes tend to be poorer than those in other domains [25]. It is also important to note that moms filled out the majority of the scales, compared to just a tiny portion of dads. The "primary carer" is often designated to administer the majority of QoL questionnaires. This suggests that women serve as the primary "carer" or point of reference. According to different research, moms are responsible for providing care since they are in control of their children's daily life. [26] The idea of family quality of life is dynamic and interrelated, and how each family member functions may have an impact on the family's overall well-being [27]. The sample size may be to blame. Relatives believe that the quality of life (QoL) of people with ID may be different from what close professionals to these patients believe [28]. Thus, it would be fascinating to enhance our data with information from experts who often interact with patients who have WS.

CONCLUSIONS: In conclusion, since the idea of QoL is so significantly impacted by the many elements that affect personal well-being, it is challenging to homogenize everyone with ID (in our instance WS) and their families. Yet, it is crucial to take these factors into account while providing the assistance and tools needed to assist families of WS patients throughout their lifespan.

REFERENCES:

- Nascimento, J. A. D., Fagundes Silva, L. A., Samelli, A. G., & Matas, C. G. Peripheral Auditory Pathway and ABR Characterization in Adults with Williams Syndrome. *Available at SSRN 4300145*.

- Aouraghe, H., Ayad, A., Abilkassem, R., & Agadr, A. (2022). Cardiovascular Abnormalities in Williams-Beuren Syndrome. *Asian Journal of Pediatric Research*, *9*(3), 19-23.
- Atkinson, J. (2022). Williams syndrome. In *Oxford Research Encyclopedia of Psychology*.
- Ridley, E., Arnott, B., Riby, D. M., Burt, D. M., Hanley, M., & Leekam, S. R. (2022). The quality of everyday eye contact in Williams Syndrome: insights from cross-syndrome comparisons. *American Journal on Intellectual and Developmental Disabilities*, *127*(4), 293-312.
- Sideropoulos, V., Sokhn, N., Palikara, O., Van Herwegen, J., & Samson, A. (2022). Anxiety and Emotion Regulation in Young People with Williams Syndrome and Down Syndrome During the COVID-19 Outbreak: A Global Study.
- Mervis, C. B., Greiner de Magalhaes, C., & Cardoso-Martins, C. (2022). Concurrent predictors of word reading and reading comprehension for 9-year-olds with Williams syndrome. *Reading and Writing*, *35*(2), 377-397.
- Richter, C. G., Cardoso-Martins, C., & Mervis, C. B. (2022). Longitudinal predictors of word reading for children with Williams syndrome. *Reading and Writing*, 1-27.
- Abuelazm, M., Muhammad, S., Gamal, M., Labieb, F., Amin, M. A., Abdelazeem, B., & Brašić, J. R. (2022). The effect of vitamin D supplementation on the severity of symptoms and the quality of life in irritable bowel syndrome patients: a systematic review and meta-analysis of randomized controlled trials. *Nutrients*, *14*(13), 2618.
- Jacoby, P., Williams, K., Reddihough, D., Leonard, H., Whitehouse, A., & Downs, J. (2022). Modelling quality of life in children with intellectual disability using regression trees. *Developmental Medicine & Child Neurology*, *64*(9), 1145-1155.
- Grad, M., Nir, A., Levy, G., Trangle, S. S., Shapira, G., Shomron, N., ... & Barak, B. (2022). Altered white matter and microRNA expression in a murine model related to Williams Syndrome suggests that miR-34b/c affects brain development via Ptpru and Dcx Modulation. *Cells*, *11*(1), 158.
- Treichel, N., Dukes, D., Barisnikov, K., & Samson, A. C. (2022). How cognitive, social, and emotional profiles impact humor appreciation: sense of humor in autism spectrum disorder and Williams syndrome. *HUMOR*, *35*(1), 113-133.
- Tabacof, L., Tosto-Mancuso, J., Wood, J., Cortes, M., Kontorovich, A., McCarthy, D., ... & Putrino, D. (2022). Post-acute COVID-19 syndrome negatively impacts physical function, cognitive function, health-related quality of life, and participation. *American journal of physical medicine & rehabilitation*, *101*(1), 48.
- Pons, A., Whalley, G., Sneddon, K., Williams, M., & Coffey, S. (2022). Predictors of quality of life after revascularization for ischemic heart disease: A systematic review. *Health Sciences Review*, 100017.
- Lakhani, F., Racette, S. B., Park, L. K., Deych, E., Williams, D., McKenzie, K. M., ... & Peterson, L. R. (2023). Prospective Study of the Impact of Outpatient Intensive Cardiac Rehabilitation on Diet Quality, Health-related Quality of Life, and Cardiovascular Health Indices. *The American Journal of Cardiology*, *192*, 60-66.
- Melchior, C., Algera, J., Colomier, E., Törnblom, H., Simrén, M., & Störsrud, S. (2022). Food avoidance and restriction in irritable bowel syndrome: relevance for symptoms, quality of life and nutrient intake. *Clinical Gastroenterology and Hepatology*, *20*(6), 1290-1298.
- Williams, Z. J., Cascio, C. J., & Woynaroski, T. G. (2023). Measuring subjective quality of life in autistic adults with the PROMIS global–10: Psychometric study and development of an autism-specific scoring method. *Autism*, *27*(1), 145-157.
- Callahan, K. P., Flibotte, J., Skraban, C., Wild, K. T., Joffe, S., Munson, D., & Feudtner, C. (2022). Influence of genetic information on neonatologists' decisions: a psychological experiment. *Pediatrics*, *149*(3).

- Kleberg, J. L., Willfors, C., Björlin Avdic, H., Riby, D., Galazka, M. A., Guath, M., ... & Strannegård, C. (2023). Social feedback enhances learning in Williams syndrome. *Scientific Reports*, *13*(1), 164.
- Dougherty, J. D., Nygaard, K. R., Maloney, S. E., Swift, R. G., McCullough, K. B., Wagner, R. E., ... & Veenstra-VanderWeele, J. (2023). Extensive characterization of a Williams Syndrome murine model shows Gtf2ird1-mediated rescue of select sensorimotor tasks, but no effect on enhanced social behavior. *bioRxiv*, 2023-01.
- Das, D., & Makhija, N. (2023). Williams-Beuren syndrome with pseudoaneurysm of aortic arch and infective vegetations for modified broms procedure: anesthetic concerns & Echocardiographic illustrations. *Annals of Cardiac Anaesthesia*, *26*(1), 94.
- Zinyandu, T., Montero, A. J., Thomas, A. S., Sassis, L., Kefala-Karli, P., Knight, J., & Kochilas, L. K. (2023). Thirty-Year Survival After Cardiac Surgery in Children With Williams–Beuren Syndrome (from the Pediatric Cardiac Care Consortium Study). *The American Journal of Cardiology*, *187*, 48-53.
- Velarde-Acosta, K., Baltodano-Arellano, R., Cachicatari-Beltran, A., Medina-Maguiña, J. M., Cupe-Chacaltaje, K., Rojas, P., & Levano-Pachas, G. (2023). Williams-Beuren syndrome associated with hammock mitral valve: First case report. *Echocardiography*.
- Tsagkas, N., Katsanevakis, E., Karagioti, N., Perdikaris, P., & Billis, M. (2023). Prenatal Diagnosis of Williams-Beuren Syndrome Based on Suspected Fetal Hypotonia in Early Pregnancy. *Cureus*, *15*(2).
- Ghisleni, C., Parma, B., Cianci, P., De Paoli, A., Pangallo, E., Agovino, T., ... & Selicorni, A. (2023). Celiac disease prevalence and predisposing-HLA in a cohort of 93 Williams-Beuren syndrome patients. *American Journal of Medical Genetics Part A*.
- Torres-Pérez, J. V., Anagianni, S., Mech, A. M., Havelange, W., García-González, J., Fraser, S. E., ... & Brennan, C. H. (2023). baz1b loss-of-function in zebrafish produces phenotypic alterations consistent with the domestication syndrome. *Iscience*, *26*(1), 105704.
- Romero, A. A. J., Birlanga, O. M., Inoriza, B., Bernal, F. L., & Jabalquinto, M. D. R. C. (2023). Williams-Campbell Syndrome: A Rare Case of Bronchiectasis. *Open Respiratory Archives*, *5*(1), 100217.
- Heck, R., Fischer-Zirnsak, B., Photiadis, J., Horn, D., & Gehle, P. (2023). Aorto-Aortic Bypass in an infant with Middle Aortic-and Marfan Syndrome: A 15-Year follow-up. *Interdisciplinary CardioVascular and Thoracic Surgery*.
- Martinelli, M., & Staiano, A. (2023). Motility Problems in Developmental Disorders. In *Pediatric Neurogastroenterology: Gastrointestinal Motility Disorders and Disorders of Gut Brain Interaction in Children* (pp. 371-380). Cham: Springer International Publishing.