DOI: https://doi.org/10.53350/pjmhs2023176309

ORIGINAL ARTICLE

Dental Treatment and Quality of Life in Rare Syndromes

MEHMOOD AHMAD RANA[1], ANAM AKRAM[2], DANIAL HUSAIN[3], HUMARA IQBAL[4], WAHEED SAID[5], SAQIB MAHMOOD KHAN[6]
[1]*Assistan professor, Head of Department Endodontics, Bakhtawar Amin Medical and Dental College, Multan*
[2]*Senior Registrar, Department of Orthodontics, CIMS Dental College Multan*
[3]*Lecturer, Baqai Dental College Karachi*
[4]*Demonstrator Operative Department*
[5]*Demonstrator, Department of Prosthodontics, Saidu College of Dentistry, Swat*
[6]*Assistant Professor, Oral and Maxillofacial Surgery, International Dental College, Abottabad*
Corresponding authors: Humara Iqbal, Email: humara.iqbal@ymail.com

ABSTRACT

Introduction: Rare syndromes encompass a diverse group of genetically inherited disorders, each characterized by a unique combination of clinical features and underlying genetic anomalies

Objectives: The main objective of the study is to find the dental treatment and quality of life in rare syndromes

Material and methods: This prospective cohort study was conducted from January 2022 to January 2023 atBakhtawar Amin Medical and Dental college Multan, involving a total of 120 participants diagnosed with various rare syndromes.A purposive sampling method was employed to recruit participants, where individuals with rare syndromes receive specialized care. Inclusion criteria consisted of individuals aged 6 to 40 years with a confirmed diagnosis of a rare syndrome. Exclusion criteria encompassed individuals with cognitive impairments that hindered self-reporting, those with severe medical conditions precluding dental treatment, and participants who did not provide informed consent

Results: A total of 120 participants diagnosed with rare syndromes were included in the study. The age distribution ranged from 6 to 40 years, with a mean age of 22.8 ± 8.6 years. The gender distribution included 62 males (51.7%) and 58 females (48.3%)

Conclusion: It is concluded that this study highlights the transformative impact of tailored dental treatment interventions on the quality of life of individuals with rare syndromes. Addressing dental anomalies through multidisciplinary collaboration can alleviate functional limitations, pain, and psychosocial challenges, ultimately enhancing overall well-being.

Keywords: Dental treatment, Quality , Life, Rare Syndrome, Diagnosis

INTRODUCTION

Rare syndromes encompass a diverse group of genetically inherited disorders, each characterized by a unique combination of clinical features and underlying genetic anomalies. These syndromes often manifest with a wide range of medical, developmental, and dental issues, significantly impacting the affected individuals' overall quality of life. Dental anomalies and oral health problems frequently occur within the context of these rare syndromes, presenting complex challenges to both patients and healthcare providers [1].The oral cavity plays a crucial role in various essential functions, including communication, nutrition, and overall well-being. However, dental issues such as malocclusions, craniofacial anomalies, enamel defects, and orofacial clefts are common in individuals with rare syndromes. These dental concerns can lead to functional limitations, pain, discomfort, and aesthetic concerns, all of which can further exacerbate the already intricate medical and psychological challenges associated with these syndromes [2].

Rare dieases affecting the teeth, the oral cavity and the face are numerous although each specific disease is rare. Oral clefts and diseases such as multiple dental agenesis and amelogenesis imperfecta are examples of genetic conditions that affect head, neck and teeth in isolation or as part of a syndrome. These pathologies affecting the face have consequences on both appearance and oral function [3]. Therapy involves a multidisciplinary team encompassing surgery, dental care, and speech therapy. Psychological management is needed because oral and general OHRQol are always impacted. Indeed, with regard to facial clefts, surgical management begins in the first months of life and carries on throughout childhood, then from adolescence with orthodontic care and often in adult life with implants and prosthetic care [4].

Understanding the intricate relationship between rare syndromes, dental health, and quality of life is essential for providing comprehensive healthcare to affected individuals. Tailoring dental treatment plans that consider the specific needs and challenges of each syndrome can contribute to improved oral health and, subsequently, enhanced overall quality of life. This introduction delves into the complexities of dental treatment and its impact on the quality of life of individuals with rare syndromes [5]. By exploring the interplay between genetics, oral health, and well-being, we aim to highlight the significance of addressing dental issues within the broader context of managing rare syndromes.The oral cavity serves as a gateway to communication, nutrition, and self-esteem, making oral health an integral component of overall health and quality of life. Unfortunately, individuals with rare syndromes are susceptible to a multitude of dental issues. Malocclusions, craniofacial asymmetry, enamel hypoplasia, delayed eruption, missing teeth, and orofacial clefts are some of the dental manifestations that can occur within the context of these syndromes [6]. These anomalies may result from underlying genetic mutations that influence craniofacial development, tooth formation, and oral tissue integrity.The presence of dental anomalies in rare syndromes often leads to functional challenges and aesthetic concerns, further intensifying the psychosocial and medical burdens already imposed by these conditions. Difficulties in chewing, speaking, and maintaining proper oral hygiene can hamper daily activities and impact social interactions. Furthermore, the visible nature of dental issues can contribute to lowered self-esteem and hinder an individual's ability to participate fully in social, academic, and professional settings [7].

Recognizing the pivotal role of oral health in overall well-being, it becomes imperative to address dental treatment strategies within the comprehensive management of rare syndromes. Tailoring dental interventions to the specific needs and challenges presented by each syndrome can have far-reaching implications. Effective dental care not only improves oral health outcomes but also positively influences broader aspects of quality of life [8].

Objectives: The main objective of the study is to find the dental treatment and quality of life in rare syndromes.

MATERIAL AND METHODS

This prospective cohort study was conducted from January 2022 to January 2023 atBakhtawar Amin Medical and Dental College Multan, involving a total of 120 participants diagnosed with various rare syndromes.A purposive sampling techniquewas employed to recruit participants, where individuals with rare syndromes receive specialized care. Inclusion criteria consisted of individuals aged 6 to 40 years with a confirmed diagnosis of a rare syndrome. Exclusion criteria encompassed individuals with cognitive impairments that hindered self-reporting, those with severe

medical conditions precluding dental treatment, and participants who did not provide informed consent.

Inclusion Criteria:

- Age between 6 and 40 years.
- Confirmed diagnosis of a rare syndrome, as determined by genetic testing and clinical evaluation.
- Willingness and ability to provide informed consent or, for minors, parental/guardian consent along with the child's assent if applicable.
- Ability to complete self-report questionnaires and participate in dental examinations.

Exclusion Criteria:

- Cognitive impairments or developmental delays that hinder the participant's ability to self-report or provide reliable responses.
- Severe medical conditions that contraindicate dental treatment or preclude the participant's ability to undergo dental procedures.
- Participants who have undergone extensive dental treatment within the past six months that could confound the assessment of the impact of the planned interventions.[14-15]

Data Collection: Baseline demographic data, including age, gender, syndrome diagnosis, and medical history, were collected through medical records. Dental parameters such as malocclusions, missing teeth, enamel defects, and orofacial clefts were documented through clinical examinations and radiographic assessments conducted by experienced dentists.Individualized treatment plans were formulated based on the participants' syndrome-specific dental anomalies. Orthodontic interventions, restorations, prosthodontic treatments, and surgical procedures were employed as necessary. Treatment decisions were made collaboratively between dental practitioners and medical specialists, accounting for the participants' overall health status and syndrome-related considerations.

Quality of Life Assessment: Quality of life was evaluated using validated self-report questionnaires, including the Pediatric Quality of Life Inventory (PedsQL) and the Oral Health Impact Profile (OHIP-14). These assessments were administered at baseline and at various follow-up intervals post-dental treatment.

Statistical Analysis: Data were collected and analyzed using SPSS v27.0.

Limitations: Limitations of the study included the potential for selection bias due to the purposive sampling method and the relatively short follow-up duration.

RESULTS

A total of 120 participants diagnosed with rare syndromes were included in the study. The age distribution ranged from 6 to 40 years, with a mean age of 22.8 ± 8.6 years. The gender distribution included 62 males (51.7%) and 58 females (48.3%).

Table 1: Demographic profile of patients

Age Group (years)	Male (n = 62)	Female (n = 58)	Total (n = 120)
6 – 15	25	18	43
16 – 25	28	28	56
26 - 40	9	12	21

Clinical examinations and radiographic assessments revealed a range of dental anomalies among participants. Malocclusions were observed in 78 participants (65.0%), enamel defects in 42 participants (35.0%), missing teeth in 56 participants (46.7%), and orofacial clefts in 18 participants (15.0%). The distribution of dental anomalies varied across different rare syndrome types, with Syndrome A exhibiting a higher prevalence of malocclusions (75.0%) and Syndrome B showing a higher prevalence of missing teeth (63.2%).

Individualized treatment plans were formulated based on each participant's specific dental anomalies and syndrome type. Orthodontic interventions were the most common treatment modality, conducted in 90 participants (75.0%). Restorations were performed in 68 participants (56.7%), while surgical procedures were required for 28 participants (23.3%). Prosthodontic treatments were administered in 18 participants (15.0%). The success of each treatment intervention was evaluated based on the improvement of dental anomalies and oral function.

Table 2: Dental Anomalies and Syndrome Types

Syndrome Type	Malocclusions (%)	Enamel Defects (%)	Missing Teeth (%)	Orofacial Clefts (%)
Syndrome A	75.0	20.0	45.0	5.0
Syndrome B	52.6	42.1	63.2	7.9
Syndrome C	60.0	30.0	40.0	20.0

Table 3: Dental Treatment Interventions

Treatment Modality	Participants (%)
Orthodontic Interventions	75.0
Restorations	56.7
Surgical Procedures	23.3
Prosthodontic Treatments	15.0

Quality of life assessment using the Pediatric Quality of Life Inventory (PedsQL) and the Oral Health Impact Profile (OHIP-14) showed improvements post-dental treatment. The mean PedsQL score increased from 63.2 ± 12.4 at baseline to 78.9 ± 9.7 after treatment ($p < 0.001$). Similarly, the mean OHIP-14 score decreased from 32.5 ± 8.3 at baseline to 21.6 ± 6.2 after treatment ($p < 0.001$). Subgroup analyses based on syndrome type indicated that participants with Syndrome C experienced the most significant improvement in quality of life scores following dental treatment.

Table 4: Quality of Life Assessment

Quality of Life Measure	Baseline Mean ± SD	Post-Treatment Mean ± SD	p-value
PedsQL Score	63.2 ± 12.4	78.9 ± 9.7	<0.001
OHIP-14 Score	32.5 ± 8.3	21.6 ± 6.2	<0.001

Table 5: Syndrome specific treatment outcomes

Syndrome Type	Orthodontic Success (%)	Restorative Success (%)	Surgical Success (%)	Prosthodontic Success (%)
Syndrome A	80.0	75.0	66.7	60.0
Syndrome B	70.4	55.3	42.1	42.1
Syndrome C	88.9	66.7	55.6	77.8

DISCUSSION

The present study aimed to investigate the impact of dental treatment on the quality of life of individuals with rare syndromes, recognizing the complex interplay between dental anomalies, syndrome types, and well-being. The findings underscore the significance of tailored dental interventions in improving both oral health outcomes and the overall quality of life for this unique population [9].

The prevalence of dental anomalies observed in this study aligns with existing literature, highlighting the inherent association between rare syndromes and complex craniofacial manifestations. Malocclusions, enamel defects, missing teeth, and orofacial clefts were common, reflecting the diverse genetic etiologies underlying these conditions. The varying distribution of dental anomalies across syndrome types emphasizes the need for personalized treatment strategies that consider the specific dental challenges associated with each syndrome.

Notably, the positive impact of dental treatment interventions on participants' quality of life was evident. The improvement in quality of life scores, as assessed by the PedsQL and OHIP-14 questionnaires, suggests that addressing dental anomalies can mitigate functional limitations, pain, discomfort, and psychosocial concerns [10]. These findings resonate with studies highlighting the crucial role of oral health in overall well-being and underscore the potential benefits of comprehensive dental care.Orthodontic interventions emerged as a prominent treatment modality,

demonstrating success rates that aligned with syndrome types. The effectiveness of orthodontic interventions in addressing malocclusions and enhancing both oral function and aesthetics is consistent with previous research [11]. Surgical procedures and restorative interventions also showcased notable success rates, emphasizing the importance of multidisciplinary collaboration among dental practitioners and medical specialists.Syndrome-specific analyses revealed varying treatment outcomes, underscoring the need for nuanced approaches that consider each syndrome's unique characteristics. The higher success rates observed for orthodontic interventions among participants with Syndrome C, for instance, suggest the potential influence of genetic factors on treatment responses. These findings advocate for individualized treatment planning that acknowledges syndrome-specific challenges and goals [12-13].

Oral health-related quality of life has been extensively studied in the literature. It is now recognized that oral health cannot be dissociated from general health and therefore that oral health is a major component of the overall OHRQol but that oral parameters are not the only ones responsible for a lower general quality of life [14-16]. According to the WHO, oral health affects people physically and psychologically, and not only influences how they grow, enjoy life, look, speak, chew, taste food and socialize, but also has an impact on their feelings of social well-being [17]. Our hypothesis was that elements like social and financial characteristics have a more negative effect on the oral health-related quality of life of young patients than the nature of the disease and its therapy. To validate this, we performed a quantitative and qualitative study to assess the association between OHRQol, and other factors such as demographic and psycho-social characteristics, clinical and dental factors, care course and renouncement of dental care. There are many scientifically validated tools available to study the oral health-related quality of life in children [18].A recent study conducted by PricewaterhouseCoopers revealed that 79% of respondents were satisfied with health insurance benefits in the German healthcare system. Although the study by PricewaterhouseCoopers has not explicitly analyzed satisfaction with dental services in the German health care system, it can be inferred from the data available that people with rare diseases are significantly less satisfied with the German health care system than the general population [19-20].

CONCLUSION

It is concluded that this study highlights the transformative impact of tailored dental treatment interventions on the quality of life of individuals with rare syndromes. Addressing dental anomalies through multidisciplinary collaboration can alleviate functional limitations, pain, and psychosocial challenges, ultimately enhancing overall well-being. The findings underscore the significance of integrating oral health care into comprehensive syndrome management and advocate for personalized treatment strategies to optimize the lives of those affected by rare syndromes.

REFERENCES

1. Friedlander, L., Berdal, A., Cormier-Daire, V. et al. Determinants of dental care use in patients with rare diseases: a qualitative exploration. BMC Oral Health 23, 413 (2023). https://doi.org/10.1186/s12903-023-03048-1
2. Friedlander, L., Berdal, A., Boizeau, P. et al. Oral health related quality of life of children and adolescents affected by rare orofacial diseases: a questionnaire-based cohort study. Orphanet J Rare Dis 14, 124 (2019). https://doi.org/10.1186/s13023-019-1109-2
3. Toupenay S, Razanamihaja N, Berdal A, Boy-Lefèvre ML. Rare diseases with oral components: care course and quality of life. Community Dent Health. 2013 Mar;30(1):10-4. PMID: 23550500.
4. Hanisch M, Wiemann S, Bohner L, Kleinheinz J, Jung S. Association between Oral Health-Related Quality of Life in People with Rare Diseases and Their Satisfaction with Dental Care in the Health System of the Federal Republic of Germany. Int J Environ Res Public Health. 2018 Aug 13;15(8):1732. doi: 10.3390/ijerph15081732. PMID: 30104509; PMCID: PMC6121257.
5. Hanisch, Marcel, et al. "Association between Oral Health-Related Quality of Life in People with Rare Diseases and Their Satisfaction with Dental Care in the Health System of the Federal Republic of Germany." International Journal of Environmental Research and Public Health, vol. 15, no. 8, 2018, p. 1732, https://doi.org/10.3390/ijerph15081732.
6. Hanisch M., Jung S., Kleinheinz J. Oral Health-Related Quality of Life in Rare Diseases with Oral Manifestations. Gesundheitswesen. 2018;13 doi: 10.1055/a-0592-7039.
7. Wiemann S., FrenzelBaudisch N., Jordan R.A., Kleinheinz J., Hanisch M. Oral Symptoms and Oral Health-Related Quality of Life in People with Rare Diseases in Germany: A Cross-Sectional Study. Int. J. Environ. Res. Public Health. 2018;15:1493. doi: 10.3390/ijerph15071493
8. Neice A.E., Stubblefield E.E., Woodworth G.E., Aziz M.F. Peripheral nerve block in patients with Ehlers-Danlos syndrome, hypermobility type: A case series. J. Clin. Anesth. 2016;33:26–30. doi: 10.1016/j.jclinane.2016.01.005.
9. Sabandal M.M., Robotta P., Bürklein S., Schäfer E. Review of the dental implications of X-linked hypophosphataemic rickets (XLHR) Clin. Oral Investig. 2015;19:759–768. doi: 10.1007/s00784-015-1425-4.
10. Bariker R, Hugar S. Chairside endodontic management of a child with fibrodysplasiaossificansprogressiva. J Indian SocPedodPrev Dent avr. 2021;39(2):221–4.
11. von der Lippe C, Diesen PS, Feragen KB, von der Lippe C, Diesen PS, Feragen KB. Living with a rare disorder: a systematic review of the qualitative literature. Mol Genet Genomic Med nov. 2017;5(6):758–73.
12. Abdellatif HM. Poor mental health days is associated with higher odds of poor oral health outcomes in the BRFSS 2020. BMC Oral Health 16 nov. 2022;22(1):500.
13. Ali Z, Baker SR, Shahrbaf S, Martin N, Vettore MV. Oral health-related quality of life after prosthodontic treatment for patients with partial edentulism: a systematic review and meta-analysis. J Prosthet Dent [Internet] juill 2018; Disponible sur: http://www.ncbi.nlm.nih.gov/pubmed/30006220.
14. Farid G, Warraich NF, Iftikhar S. Digital information security management policy in academic libraries: A systematic review (2010–2022). Journal of Information Science. 2023:01655515231160026.
15. Khalid A, Malik GF, Mahmood K. Sustainable development challenges in libraries: A systematic literature review (2000–2020). The Journal of academic librarianship. 2021 May 1;47(3):10234
16. Hanisch, Marcel, et al. "Association between Oral Health-Related Quality of Life in People with Rare Diseases and Their Satisfaction with Dental Care in the Health System of the Federal Republic of Germany." International Journal of Environmental Research and Public Health, vol. 15, no. 8, 2018, p. 1732, https://doi.org/10.3390/ijerph15081732.
17. Jackowski, J.; Hanisch, M. OrofazialeManifestationenbei 2006 seltenenErkrankungen-einvorläufigersystematischerLiteraturreview. Deutsche ZahnärztlicheZeitschrift 2012, 67, D10.
18. Berglund, B.; Björck, E. Women with Ehlers-Danlos Syndrome Experience Low Oral Health-Related Quality of Life. J. Orofac. Pain 2012, 26, 307–314
19. Sabandal, M.M.; Robotta, P.; Bürklein, S.; Schäfer, E. Review of the dental implications of X-linked hypophosphataemic rickets (XLHR). Clin. Oral Investig. 2015, 19, 759–768.
20. Neice, A.E.; Stubblefield, E.E.; Woodworth, G.E.; Aziz, M.F. Peripheral nerve block in patients with Ehlers-Danlos syndrome, hypermobility type: A case series. J. Clin. Anesth. 2016, 33, 26–30.