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A research study on Quality of life of people living with marfan syndrome

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ABSTRACT:

OBJECTIVE: Genetically determined multiorgan illness Marfan syndrome (MFS) causes significant physiological and psychosocial deficits in adults. There is limited consensus about QOL in those with MFS. The present study looked at the quality-of-life QOL of a sample of MFS patients.

METHODS: At the Mayo hospital, Lahore patient-reported results measures from 102 individuals with MFS having mean age of 39.3 years and standard deviation of 13.1 years being 40.2% female were retrospectively examined and compared to those people who had different congenital cardiac abnormalities. The improved five-level EQ-5D was used to evaluate QOL.

RESULTS: The variations between the two populations were investigated. Individuals with MFS reported a general decline in QOL. Those with MFS rated considerably lower on the aspects of pain, anxiety, mobility, and normal activities compared to CHD patients (P 0.05).

CONCLUSIONS: MFS patients are more prone to have poorer QOL, particularly in the physical and mental dimensions. MFS's psychosocial effects drain resources from both patients' and professionals' budgets. The necessity of extra psychological assistance in coping with obstacles caused by sickness is highlighted by recent research. Their subjective wellbeing should get more focus in order to possibly enhance their QOL and enduring health consequences.

KEYWORDS: quality of life, marfan syndrome, psychology.

INTRODUCTION: The tissue all across the body, including the core neurological systems, skeletal, ophthalmic, cardiovascular, and pulmonary is affected by Marfan syndrome (MFS), a multiorgan illness that is genetically determined. MFS is thought to impact between 0.002% and 0.017% of the population (1). The development of aneurysms and aortic dissection are among the most serious side effects from a medical standpoint. As a result, the majority of individuals are continuously watched, beginning with early childhood cardiovascular exams (2). Moreover, MFS sufferers often deal with skeletal or visual anomalies that give them a distinct look that is both subjective to them and evident to others (3). Recent research has shown that MFS sufferers may experience a reduction in QOL due to the interaction of psychological and physiological symptoms (4).

Research on the specific psychological issues faced by people with MFS is still lacking, despite the increased interest in the psychosocial effects of congenital heart defects (CHD) patients (5). While "hard" outcome measures like death, morbidity, and physical function have historically dominated clinical research, the idea of QOL is becoming more widely acknowledged as a crucial patient-reported outcome measure in the assessment of care and therapy (6). Individuals with MFS are more likely to have negative psychosocial consequences, such as lower QOL, especially in the psychological area (7). Some MFS patients also exhibit signs of mental discomfort, such as anxiety and sadness (8). In addition to having an adverse effect on a patient's general health, emotional anguish may also raise cardiovascular morbidity and hasten death (9).

The current study's objectives were to (I) comprehensively evaluate QOL in a sizable sample of MFS patients, and (II) evaluate the findings with those from patients with other kinds of CHD. The results of this research should highlight how critical it is to understand the psychosocial effects of Marfan syndrome (MFS) and the need of assessing patients' psychological wellbeing. This knowledge may help medical practitioners create suitable support systems and treatment plans to improve the overall quality of life for MFS patients. In the end, this may result in better results and a greater understanding of the many problems MFS patients experience.

METHODS: The Mayo Hospital in Lahore was the one who started and carried out the questionnaire-based survey. Between December 2019 and January 2023, data were gathered. Before beginning the documentation, all participating patients provided their written informed consent. Data privacy laws and best practices for pharmacoepidemiology were adhered to.

To conduct a sub-analysis, a specific group of patients (1) having a confirmed diagnosis of Marfan syndrome (MFS); (2) being at least 18 years old; and (III) having the physical, mental, and linguistic ability to complete self-report questionnaires, was selected, which were part of the data collection process. These criteria were essential to ensure that the participants could provide accurate and reliable information for the study. If a participant was too young or had serious cognitive impairment, they were excluded from the study. Patients were not initially chosen; instead, they were sequentially added in the sequence that they arrived at the facility.

The survey may be answered by mail, online or in person. The modified five-level version of the EQ-5D (EQ-5D-5L), which offers a straightforward, general assessment of a patient's subjective health state, was used to measure QOL. In comparison to the original EQ-5D, this version exhibits a much greater dependability, discriminating power, and reduction of ceiling effects. Two elements make up the paper-based, self-administered EQ-5D-5L questionnaire: a descriptive system questionnaire and a visual analogue scale (VAS). The five characteristics that make up the descriptive system are mobility, anxiety, self-care, pain, and regular activities. Using a 5-point Likert scale, the patient is asked to rank their perceived limitations, from "no issues" to "serious problems/unable," according to how severe they are. The degree of the impairment on each aspect is expressed in the responses as single-digit figures. It is possible to aggregate replies into a single weighted index score using population preference ratings (EQ- 5D index). The EQ-VAS uses a vertical scale from 0 ("The worst health you can imagine") to 100 ("The greatest health you can imagine") to assess a patient's total health. As a result, it gives a patient's perceived health a numerical measurement.

With the help of SPSS version 26, statistical analysis was carried out. The patient demographics, cardiovascular and non-cardiac diagnoses, and medical records were examined. Statistics were evaluated anonymously and without tying them to specific individuals. For the features of the sociodemographic sample, descriptive metrics were generated. The effect of Marfan syndrome (MFS) on the QOL dimensions was examined using logistic regression models with the EQ-5D-5L as the dependent variable. T-tests were performed to compare average values, and chi-squared tests were done to see if there were any demographic differences. For categorical or interval scaled variables, the research reported continuous data as mean, standard deviation, absolute values, and percentages.

RESULTS: A final sample of 102 people with Marfan syndrome (MFS) were chosen for the study from the original pool of 3,885 patients, with 40.2% of them being female (see Table 1). The patients with MFS varied in age from 20 to 85, with a mean age of 39.3 and an SD of 13.1. To provide a point of comparison made up of non-MFS ACHD, the remaining 3,783 ACHD, whose mean age was 42.0 and standard deviation was 17.3, were consulted. Their age ranged from 18 to 97 years, and 46.6% of them were female. In terms of age distribution, residence area, and insurance status, both populations were similar.

Table 1: Demographic information of study population

	Marfan		**ACHD**	
Demographics	**N=102**	**%**	**N=3,783**	**%**
Age	39.3±13.1	20-85	42.0±17.3	18-97
Gender (Female)	41	40.20%	1,763	46.60%
Age group				
65+	6	5.9	501	13.2
35–64	55	53.9	1,678	44.4
18–34	41	40.2	1,604	42.4
Missing	0		111	
Residence				
Rural	61	60.4	2,418	64.7
Town	14	13.9	571	15.3
City	26	25.7	747	20
Missing (n)	1		158	
Insurance				
No insurance	0	0	7	0.2
Private	7	7	210	5.5
Public	93	93	3,570	94.3
Missing	2		107	

Table 2: Effect of the Marfan diagnosis on the EQ-5D dimensions

Dimensions	**Value**	**Range**
Anxiety	0.598	0.40–0.90
Pain	0.37	0.25–0.55
Usual activities	0.487	0.32–0.75
Self-Care	1.07	0.41–2.77
Mobility	0.6	0.37–0.96

Table 3: EQ5D Dimensions distribution

Dimensions	**Marfan, n (%)**	**ACHD, n (%)**	**P value**

Anxiety/depression			
Extremely anxious/depressed	0(0%)	31(0.8%)	0.022
Severe	3(3.2%)	148(4%)	
Moderate	14(15.1%)	346(9.4%)	
Slight	33(35.5%)	981(26.6%)	
No	43(46.2%)	2181(59.2%)	
Missing			
Pain/discomfort			
Extreme pain	0(0%)	15(0.4%)	<0.001
Severe	7(7.6%)	130(3.5%)	
Moderate	16(17.4%)	450(12.3%)	
Slight	39(42.4%)	922(25.1%)	
No	30(32.6%)	2154(58.7%)	
Missing			
Usual activities			
Unable to do	0(0%)	44(1.2%)	0.002
Severe	6(6.5%)	119(3.2%)	
Moderate	13(14%)	320(8.6%)	
Slight	23(24.7%)	619(16.7%)	
No	51(54.8%)	2599(70.2%)	
Missing			
Self-care			
Unable to wash/dress	0(0%)	25(0.7%)	0.483
Severe	1(1.1%)	33(0.9%)	
Moderate	1(1.1%)	70(1.9%)	
Slight	3(3.2%)	141(3.8%)	
No	88(94.6%)	3442(92.8%)	
Missing			
Mobility			
Unable to walk	1(1.1%)	17(0.4%)	0.059
Severe	2(2.2%)	117(3.2%)	
Moderate	10(10.8%)	309(8.3%)	
Slight	18(19.4%)	449(12.1%)	
No	62(66.7%)	2820(76%)	
Missing			

Table 4: VAS and cross walk values

		VAS	**Crosswalk**
ACHD	**Mean**	76.23	90.67
	SD	19.04	15.51
Marfan	**Mean**	72.58	86.97

	SD	15.95	15.12
P value		0.073	0.025

Table 2 displays how MFS has affected each of the five QOL dimensions. With the exception of self-care, having MFS substantially reduced the likelihood of having no issues on each corresponding dimension (P 0.05). Similar findings are seen in Table 3, which contrasts the EQ-5D-5L subscales for the MFS sample with the selected reference group of ACHD.

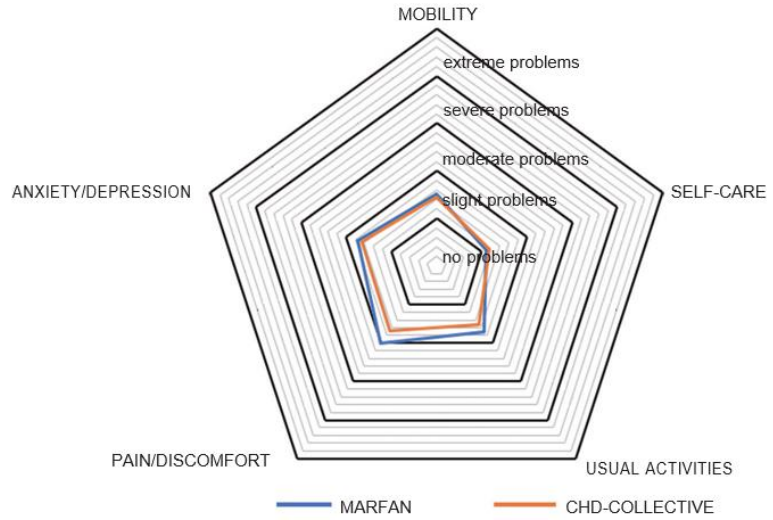

Figure 1: EQ-5D-5L scores of people with Marfan syndrome compared to a control group of those with ACHD.

Patients with MFS reported substantially lower QOL in daily activities compared to the larger ACHD group, with p-values of 0.002, 0.001, and 0.022 respectively for pain and anxiety. The pain/discomfort measure showed the most dramatic observed variances. The mobility (P=0.059) and self-care (P=0.483) categories showed no evidence of significant differences. The comparison of the EQ-5D-5L's five dimensions for the two samples is shown graphically in Figure 1.

Patients with MFS exhibited significantly lower VAS scores (P-value=0.073) and lower descriptive index values (P-value=0.025) compared to patients with non-MFS ACHD on the combined QOL measures (Table 4). The VAS ratings, however, showed fewer dramatic variations across the groups. The observed variances in QOL were also influenced by the kind of measurement used. As a result, the descriptive index value for QOL was much higher than the mean VAS score.

DISCUSSIONS: The lives of those with MFS have been significantly improved in recent decades by significant medical improvements. As a result, many doctors consider MFS to be one of the hereditary disorders that is now easiest to control (10). Yet, a large number of afflicted individuals disagree with this viewpoint and report significant QOL deficits (4). There haven't been many studies done to assess MFS's psychological consequences. In this research, the EQ-5D-5L was used to evaluate QOL in people with MFS and expand on prior results. The EQ-5D-5L is a very accurate and trustworthy measure of outcome in the cardiac sector (11). It offers a more complete picture of quality of life in terms of total life satisfaction by using two separate assessment techniques—a short descriptive system questionnaire and a VAS. The findings of this study indicate that people with MFS have significant mental and physical impairments and are at an especially high risk of having a worse quality of life.

According to recent research, individuals with MFS have considerably worse quality of life (QOL) in the physical domain than their peers with other types of CHD. Moreover, prior studies have shown that a prominent and ongoing issue in MFS is the prevalence of chronic pain, which varies from 47% to 92% in people with MFS (12). Recent research on pain in people with MFS indicates that pain significantly lowers quality of life (13). The most painful condition is back pain, which is followed by migraines and cervical discomfort (14). Clinical signs of MFS include degenerative disc degeneration, dural ectasia, early arthritis,

and kyphosis. These conditions often cause severe pain (15). Unfortunately, there are currently few choices for managing chronic pain, and research efforts in this field seem disjointed. Just a tiny fraction of MFS patients get medical therapy for pain, and less than half of them are satisfied with their current pain management, according to a research (16). These results are particularly concerning since persistent pain is associated with severe impairment and heavy psychological strain (17). To describe the direction of pain, mental health, and QOL in individuals with MFS in more detail, further study is required. Improved management may increase a person's happiness with life by encouraging engagement in daily activities and at work.

The current data suggest that having MFS enhanced the likelihood of reporting issues with routine tasks, such as housework, study, work, leisure activities). In general, it has been shown that there is a substantial correlation between physical inactivity and labor engagement (18). Research show that MFS sufferers' employment rates are much lower than those of the general public (19). Discrimination and stigma at work are additional elements that people with MFS face on a daily basis (20). According to a research, 20% of patients with MFS reported experiencing incidents of occupational discrimination, and 32% of people with MFS reported feeling socially prejudiced against. Because of this, some patients choose to avoid social events or conceal their disease (21). 80 percent of patients also said that their MFS caused them to work less hours or miss, on average, 6.5 to 7 months of work owing to their therapy (22). Mental illness, a gloomy attitude on MFS, and poor self-esteem are all highly connected with social stigma (23). These results show that providing MFS patients with psychological support is essential to facilitating their coping mechanisms and enhancing long-term outcomes.

Given the results about individuals with MFS and their experiences with pain, it is not unexpected that MFS was substantially linked to the incidence of mobility limitations. Chronic pain and exhaustion, two symptoms of MFS that are often present, may limit movement and reduce physical quality of life (24). Age and the existence of scoliosis have been shown to be significantly associated with physical QOL problems. Moreover, persistent pain was linked to extreme physical and mental exhaustion, which is one of the most common complaints among MFS patients (25). It is yet unknown if tiredness is a symptom of MFS or a side effect of pharmaceutical usage (26). In order to identify modifiable characteristics of MFS that might be taken into account in treating patients with MFS to enhance their QOL, further study is required to determine the precise cause of tiredness in patients with MFS.

Patients with MFS had significant emotional deficits, which was in line with past results. Depression and anxiety have been identified as the bio- behavioral variables that, of all disease-related factors, have the greatest and most direct impact on quality of life (QOL) (27). Another research revealed that substantial levels of depression were present in more than 40% of the study sample (28). Poor psychosocial outcomes, such as body image concerns, unfavorable illness perceptions, and lower self-esteem, as well as poor cardiovascular outcomes, such as increased morbidity and early death, are known to be significantly influenced by emotional distress (29). Patients often complain about experiencing unseen in the healthcare system and having trouble getting enough treatment since they may not exhibit obvious symptoms (30). Based on these results, physicians should take into account the possibility that psychological characteristics that go beyond the core characteristics of MFS may have an equivalent impact on a patient's quality of life.

CONCLUSIONS: Very few research have been conducted to thoroughly evaluate the psychosocial components of MFS. According to the most recent research, those who have MFS are more likely to have poorer QOL in both the physical and mental health categories. Comparing the research group to those with other types of CHD, the significant QOL limitations are significantly more evident. Hence, it is crucial to take into account the psychological effects of MFS, which need resources from both patients and professionals, including time, money, and energy. By integrating psychosocial and mental health support into cardiac treatment for MFS, clinicians may assist patients with MFS in lowering this expense.

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