STUDENT ARTICLE

What Can Be Learned from an Adolescent with Marfan Syndrome and Mental Health

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ABSTRACT

This case describes an adolescent with Marfan Syndrome (MFS), focusing on the psychologic aspects of the case. While cardiology, ophthalmology, and orthopedics are crucial to the care of the MFS patient, it is vital to consider psychologic manifestations of living with the syndrome, especially in younger populations. This report will explore strategies for effective psychologic assistance as supported by the case and the literature.

INTRODUCTION

Marfan Syndrome (MFS) is an inherited connective tissue disorder that affects 1 in 3000 to 5000. Mutations involve fibrillin-1, which is a crucial component of elastic and non-elastic connective tissues.[1] Signs and symptoms of MFS are highly variable, but are known to include aortic disease, mitral valve prolapse, ectopia lentis, dural ectasia, excess linear growth in long bones, arachnodactyly, pectus deformity, hindfoot valgus, scoliosis, and protrusio acetabuli.[1] These features are characterized in the Ghent scoring system to establish the diagnosis.[2] Workup after diagnosis may include genetic testing, echocardiogram, eye examination, and electrocardiogram.[3] Cardiology, ophthalmology, and orthopedic surgery consults are frequently placed, and specialists continue to follow patients throughout their lifetime. Treatment is largely supportive, including bracing for scoliosis, glasses for ocular concerns, and pain management, and may also include cardiovascular or other surgeries.[3] MFS patients have lifespans reaching the 70s in modern day, even with the high rate of heart involvement.[1]

CASE REPORT

A 16-year-old African American female with an established diagnosis of MFS presented with her mother to her primary care office for a well-child visit. She had a history of prior aortic root surgery and knee surgery. She was followed by ophthalmology and cardiology. Physical examination revealed severe arthrogryposis in the joints, scoliosis, exotropia especially in the right eye with an inability to converge, ectopia lentis, hyperlaxity of joints, and club foot malformation.

The patient reported that, over the last few years, she had experienced increased bullying in school due to her condition. Her mother noted a sad affect, social isolation, decreased appetite, quiet demeanor, and falling grades. The patient had not had suicidal ideation. In October 2020, the patient's PHQ-9 score was 15, indicating moderate-severe depressive symptoms.

Due to the integrated care model of the primary care physician's office, a prompt behavioral health assessment by a psychologist was completed. Assessment and follow-up revealed adjustment disorder with mixed emotional features, major depressive disorder, and generalized anxiety. She began individual therapy sessions with the behavioral health staff addressing coping strategies for bullying, self-esteem, and academic achievement. In January 2021, her PHQ-9 score decreased to 4. Her overall demeanor was brighter, and her grades had started to improve. Continued monitoring by behavioral health was a priority in the care of this patient. The patient did not receive psychotropic medications.

DISCUSSION

MFS can be disfiguring, isolating, stressful, and worrisome for all age groups affected, especially the pediatric population. School bullies can be relentless, and a medical condition with various obvious physical manifestations make for an easy target. This case illustrated the profound impact that managing MFS can have on the psyche of an adolescent. An important aspect of care described in this case study was the integrated health model with easy access to behavioral health services at the patient's primary care office, including assessment of conditions, therapy, and medical treatment, if necessary. Focusing on coping mechanisms proved to be an effective strategy engaged by this behavioral health team.

The psychiatric aspect of the management of MFS has not taken appropriate precedence previously. Recent studies have attempted to further explore this topic.

The Pediatric Heart Network Marfan Trial compared Pediatric Quality of Life Inventory Generic Core Scales between MFS patients and healthy population norms. Those ages 5-18 years with MFS had lower mean scores for physical and psychosocial domains, and those ages 19-25 years had higher psychosocial scores than healthy norms.[4] Curiously, the severity of MFS-related physical findings were not associated with lower quality of life scores.[4] These data were significant as they noted that perhaps the adults in the study had already developed coping skills to manage their challenges and allow for higher levels of psychosocial functioning despite their physical manifestations of the syndrome. These lower quality of life scores in the children and adolescents should alert pediatricians and other specialists involved in the care of MFS patients to increase monitoring of psychological and neurodevelopmental issues and provide earlier opportunities for appropriate treatment. Furthermore, Nielson's article, "A Review of Psychosocial Factors of Marfan Syndrome: Adolescents, Adults, Families, and Providers," analyzed various reports to craft treatment recommendations including encouraging social activity involvement, development of coping strategies, provider discussion of concerns with the patient and family, and a multidisciplinary team approach to best care for MFS patents.[5] These strategies can foster resilience and a strong understanding of each patient's unique challenges.

As strides are being made to include psychologic evaluation as an element of first line MFS management, this case report illustrated the importance of screening tools such as the PHQ-9, regular visits with a primary care physician who can engage appropriate mental health care providers, and family involvement as part of the treatment plan. The literature echoes these notions. In future research, it would be worthy to explore the effects of different therapeutic methodologies on psychosocial concerns in MFS as well as other strategies to help those with MFS cope with their unique challenges.

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