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On the Role of Delays to Diagnosis of Facioscapulohumeral Muscular Dystrophy

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Abstract

Facioscapulohumeral muscular dystrophy (FSHD) is a rare genetic muscle disease that predominantly affects the facial, scapular, and humeral musculature. Scapular winging is the most common initial finding in patients with FSHD. FSHD can substantially impact a patient's daily life, ranging from discomfort during work to athletic disadvantages. Another aspect of FSHD that is essential for obtaining successful patient outcomes is timely diagnosis, which can be challenging, possibly due to the heterogeneity of presenting features as well as the rarity of the condition. Some of the methods for increasing the ability to diagnose FSHD should include increased education regarding the disease, as well as other muscular dystrophies, specifically in underserved communities that may have less access to healthcare. Although there is currently no effective pharmacologic intervention for FSHD, recommendations of the American Academy of Neurology and the Practice Issues Review Panel of the American Association of Neuromuscular & Electrodiagnostic Medicine (2015) provide helpful guidance. (**International Journal of Biomedicine. 2022;12(3):474-475.**).

Keywords: facioscapulohumeral muscular dystrophy • scapular winging • diagnosis

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Facioscapulohumeral muscular dystrophy (FSHD) is a rare genetic muscle disease that predominantly affects the facial, scapular, and humeral musculature.[1,2] FSHD is the third most common type of muscular dystrophy. The estimated prevalence of FSHD is between 4 and 10 per 100,000 population.[3] It is an autosomal dominant disorder; however, up to 30% of cases are sporadic, arising from de novo mutations.[1] Patients with FSHD are generally young, and more than 50% of individuals with FSHD show symptoms by age 20.[3] The development of the disease is characterized by progressive muscle weakness involving the face, scapular stabilizers, upper arm, and lower leg. Although there is currently no effective pharmacologic intervention for FSHD, recommendations of the American Academy of Neurology and the Practice Issues Review Panel of the American Association of Neuromuscular & Electrodiagnostic Medicine provide helpful guidance.[1]

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Scapular Winging and Muscular Weakness

Scapular winging is the most common initial finding in patients with FSHD. Preferential weakness of the lower trapezius muscle results in the characteristic upward movement of the scapula during flexion or abduction of the arms.[4]

Through our anonymous surveying of patients with FSHD, we found that all but one of them (23 patients) had scapular winging. These patients also reported difficulty engaging in exercise, i.e., throwing balls or swinging a bat, as well as pain, both expected symptoms of scapular winging. Scapular winging also causes difficulty in professional aspects of a patient's life. This progressive condition results in significant difficulties in maintaining occupations such as carpentry, driving, manual labor, or jobs that require extended periods of sitting at a desk.[4] The significant pain and progressive shoulder weakness are major contributing factors to difficulty maintaining a professional occupation. Rotator cuff and scapular muscular degeneration can cause immense difficulty maintaining speed and agility with common tasks (typing, sitting with arms extended over a keyboard, or handwriting).[2] It is important to propose

vocational rehabilitation to patients affected by FSHD as the condition progresses. Some potential options for vocational rehabilitation should include job retraining, repurposing, redefining occupational expectations, and routine occupational therapy assessments. This would make a meaningful transition to gainful employment.

Delays to Diagnosis of FSHD

Another aspect of FSHD that is essential for obtaining successful patient outcomes is timely diagnosis, which can be challenging, possibly due to the heterogeneity of presenting features as well as the rarity of the condition. FSHD is caused by the deletion of a subset of the D4Z4 macrosatellite repeat units on chromosome 4.[5] At least 11 D4Z4 repeats, yielding a DNA fragment of >38kb, are found in healthy individuals during standard genetic testing. FSHD individuals, in contrast, possess 1–10 repeats, yielding DNA fragments of 10–38 kb in size.[6] Measurement of the size of the residual D4Z4 sequence on 4q35 forms the basis for genetic testing in FSHD.[1] A much smaller number of cases, designated FSHD type 2 (FSHD2), have been ascribed to mutations in the SMCHD1 gene on chromosome 18. It is appropriate to offer genetic counseling (including discussion of potential risks to offspring and reproductive options) to young adults who are affected or at risk.

Prolonged delays to diagnosis are common. Children with very early-onset FSHD type 1 (FSHD1) may require nutritional and/or respiratory support, surgery, and orthopedic procedures. Timely care with use of assistive devices may improve mobility and prevent falls.[7]

We found a strong positive correlation between age of diagnosis and severity of FSHD ($r=0.85$, $P<0.01$). Many of the patients surveyed in the clinic reported difficulty keeping up with their peers athletically as children. Educating teachers and coaches about muscular dystrophy, its symptoms and how to recognize it, could become a tool to recognize students who may be showing symptoms of FSHD. Some of the methods for increasing the ability to diagnose FSHD should include increased education regarding the disease, as well as other muscular dystrophies, specifically in underserved communities that may have less access to healthcare.

Conclusion

FSHD can substantially impact a patient's daily life, ranging from discomfort during work to athletic disadvantages. A focus of medical professionals pursuing a reduced impact of FSHD on patients must be advocating for and increasing access to treatment in underserved communities. Those in underserved communities may be at most risk of delayed diagnosis, as well as those working in blue-collar jobs that require strenuous manual labor and are most impacted by muscular degeneration and scapular winging. Overall, through increased vocational assistance, retraining, and redirecting, as well as a focus on earlier diagnosis of FSHD, patient outcomes may be improved.

Competing Interests

The authors declare that they have no competing interests.

References

1. Tawil R, Kissel JT, Heatwole C, Pandya S, Gronseth G, Benatar M; Guideline Development, Dissemination, and Implementation Subcommittee of the American Academy of Neurology; Practice Issues Review Panel of the American Association of Neuromuscular & Electrodiagnostic Medicine. Evidence-based guideline summary: Evaluation, diagnosis, and management of facioscapulohumeral muscular dystrophy: Report of the Guideline Development, Dissemination, and Implementation Subcommittee of the American Academy of Neurology and the Practice Issues Review Panel of the American Association of Neuromuscular & Electrodiagnostic Medicine. Neurology. 2015 Jul 28;85(4):357-64. doi: 10.1212/WNL.0000000000001783

2. Statland JM, Tawil R. Facioscapulohumeral Muscular Dystrophy. Continuum (Minneap Minn). 2016 Dec;22(6, Muscle and Neuromuscular Junction Disorders):1916-1931. doi: 10.1212/CON.0000000000000399.

3. Preston MK, Tawil R, Wang LH. Facioscapulohumeral Muscular Dystrophy. 1999 Mar 8 [updated 2020 Feb 6]. In: Adam MP, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2022. PMID: 20301616.

4. Martin RM, Fish DE. Scapular winging: anatomical review, diagnosis, and treatments. Curr Rev Musculoskelet Med. 2008 Mar;1(1):1-11. doi: 10.1007/s12178-007-9000-5.

5. Snider L, Geng LN, Lemmers RJ, Kyba M, Ware CB, Nelson AM, Tawil R, Filippova GN, van der Maarel SM, Tapscott SJ, Miller DG. Facioscapulohumeral dystrophy: incomplete suppression of a retrotransposed gene. PLoS Genet. 2010 Oct 28;6(10):e1001181. doi: 10.1371/journal.pgen.1001181.

6. van Deutekom JC, Wijmenga C, van Tienhoven EA, Gruter AM, Hewitt JE, Padberg GW, van Ommen GJ, Hofker MH, Frants RR. FSHD associated DNA rearrangements are due to deletions of integral copies of a 3.2 kb tandemly repeated unit. Hum Mol Genet. 1993 Dec;2(12):2037-42. doi: 10.1093/hmg/2.12.2037.

7. Steel D, Main M, Manzur A, Muntoni F, Munot P. Clinical features of facioscapulohumeral muscular dystrophy 1 in childhood. Dev Med Child Neurol. 2019 Aug;61(8):964-971. doi: 10.1111/dmcn.14142.