OLGU SUNUMU CASE REPORT

DOI: 10.31609/jpmrs.2020-78486

Hyaline Fibromatosis Syndrome: Presentation of a Rare Case in Adult and Advanced Stage

Hiyalin Fibromatozis Sendromu: Erişkin ve İleri Evrede, Nadir Görülen Bir Vakanın Sunumu

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ABSTRACTHyaline fibromatosis syndrome (HFS) is a rare hereditary disease that occurs especially in early childhood and patients rarely reach adulthood. It is characterized by abnormal accumulation of hyaline material in body tissues, and this accumulation causes progressive deformity and dysfunction. If they are not treated, most patients will develop severe physical limitations. Early rehabilitation programs that start before development of contractures in the joints can keep patients functionally active and prevent the development of disability. With this case report, an adult patient with diffuse nodules, painfull joint contractures and gingival hyperplasia was discussed. The importance of early diagnosis and treatment of HFS and the necessity of genetic counseling for parents were emphasized.

Keywords: Hyaline fibromatosis syndrome; joint contractures; rehabilitation

ÖZET Hiyalin fibromatozis sendromu (HFS), özellikle erken çocukluk döneminde ortaya çıkan ve hastaların nadiren erişkinliğe ulaşabildiği, ender görülen bir kalıtsal hastalıktır. Vücut dokularında anormal hiyalin birikimi ile karakterize olup, bu birikim giderek artan şekil ve fonksiyon bozukluklarına neden olur. Tedavi edilmezler ise hastaların çoğunda ciddi fiziksel kısıtlamalar gelişir. Eklemlerde, kontraktürler gelişmeden başlayan erken rehabilitasyon programları, hastaları fonksiyonel olarak aktif tutup, sakatlık gelişmesini engelleyebilir. Bu olgu sunumu ile yaygın nodülleri, ağrılı eklem kontraktürleri ve diş eti hiperplazisi olan erişkin bir hasta tartışıldı, HFS'nin erken tanı ve tedavisinin önemi, ebeveynlere genetik danışmanlık verilmesinin gerekliliği vurgulandı.

Anahtar Kelimeler: Hiyalin fibromatozis sendromu; eklem kontraktürleri; rehabilitasyon

Hyaline fibromatosis syndrome (HFS) is a rare, progressive, and autosomal recessive disease.[1] It is characterized by abnormal accumulation of hyaline material in body tissues. This disorder affects many body parts, including the skin, joints, bones, and visceral organs.[2] Nodular lesions, as a result of abnormal growth of hyalinized fibrous tissue, are characteristics of this syndrome and may cause swelling and deformities in the skin, pain and contractures in the joints, deformation and osteoporosis in the bones, gingival hyperplasia, visceral organ dysfunctions and developmental retardation. Malignant transformation is not seen in HFS. This syndrome is commonly diagnosed in early childhood, but there are some cases diagnosed at adulthood.[3,4] Although there are serious physical limitations in individuals with HFS, mental development is usually normal. In most patients, lesions tend to be additive over time from childhood to adulthood.[5] Few patients with HFS can reach adulthood. Most of them have severe physical limitations. We reported a young adult case with HFS who was not referred for rehabilitation program early after diagnosis and consulted to our department with severe deformities and disability.

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Peer review under responsibility of Journal of Physical Medicine and Rehabilitation Science.

Received: 22 Aug 2020 ***Received in revised form:*** 10 Oct 2020 ***Accepted:*** 13 Nov 2020 ***Available online:*** 05 May 2021

CASE REPORT

A 22 years old male patient was admitted to our clinic with complaints of difficulty in walking and joint pain. He was born of related parents and his younger brother has died because of similar symptoms. His skin lesions began to occur at the age of 2-3 years, and he has undergone many surgical interventions since then. His walking difficulties has first started when he was 10 years old, and for the last 5 years he was unable to walk without support. When he applied to our department, he had many nodules in his head and neck, arms and legs. On physical examination, we detected contractures at the elbow, shoulder, hip, knee, wrist, and ankle joints. He was able to stand with support and had difficulty while using his arms. Mild gingival hyperplasia was present. Other systemic examination and cognitive functions were normal. According to the pathology report of the previously excised nodular lesions, the patient was diagnosed with HFS. A medical and physical therapy program (TENS, shortwave diathermy) was organized for musculoskeletal pain and movement limitations. The rehabilitation program included joint range of motion and stretching exercises. At the end of 15 days, the patient's pain score measured by visual analogue scale was reduced 50%. He also underwent surgery for bilateral knee flexion contractures. After all these treatments, the patient was able to ambulate independently at a short distance with the aid of a walker and knee brace. The patient was also consulted to genetic department for future recommendations to him and his parents. A written informed consent was obtained from the patient.

DISCUSSION

Juvenile hyaline fibromatosis (JHF) was first described by Murray in 1873 as molluscum fibrosum.[4] The term HFS was first introduced by Nofal et al. in 2009.[5] HFS is the unifying term for infantile systemic hyalinosis (ISH) and JHF.[6] An early onset, more severe and usually fatal form, which is called as ISH, had long been differentiated from a later onset and less severe form called as HFS. The prognosis for this disease is variable.[3] Usually patients die before adulthood, because of stubborn diarrhea and recurrent infections.[7] Although the etiology of HFS is not known, it is thought that gene mutation may be responsible for it.[8] The literature related with HFS is very limited and publications are mostly in the form of case reports. Since most patients die before adulthood or reach adulthood with severe disabilities, most of the cases in the literature are related with childhood disease. Joints are the one of the most important targets of the disease and joint stiffness and deformities are frequently seen. The skin covering the joints is often hyperpigmented.[8] Villous atrophy and intestinal lymphangiectasia result in severe diarrhea and cachexia.[7] The age of disease onset can vary from birth to late childhood.[5-8] The most common initial symptoms are nodular lesions on the skin and soft tissue, extreme pain at minimal handling and progressive joint contractures as they were in our patients.[3-7] Symptomatic treatment is important especially in patients with severe pain. Regular physiotherapy should be carried out to maintain the joint range of motion and to prevent joint contractures.[9] It is important that these patients should be referred to a physiatrist after they have been diagnosed. Early rehabilitation programs can prevent joint contracture and keep patients functionally active. Our patient came to us during the advanced stage of the disease and with fixed joint contractures. The main aim for rehabilitation of this patients should be to preserve and if possible, to improve functionality. Treatment of skin papules and subcutaneous nodules needs surgical excision. However, local recurrence is common. The use of intralesional systemic steroids in the treatment of subcutaneous nodule has been suggested, however this has not yet been found to be effective.[10] Capsulotomy of joint contractures has a temporary beneficial effect. On the other hand, radiotherapy has been found ineffective.[10] Partial gingivectomy can be done if necessary. Physiotherapy is especially important in the treatment of musculoskeletal disabilities, especially in patients who have not developed joint contractures yet.[2,9] Early diagnosis of the patients and early and active rehabilitation programs, both improve daily life activities and minimize the disabilities that may develop in the future.[9]

CONCLUSION

HFS is a rare disease and requires multidisciplinary care. In this respect, it is important for these patients to have an early diagnosis and treatment. Rehabilitation programs can be effective if it starts with diagnosis and before contracture development.

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