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SHORT REPORT

Juvenile Hyaline Fibromatosis: Case Presentation (The Rehabilitation of Three Siblings)

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Juvenile hyaline fibromatosis (JHF) is a rare, autosomal recessive hereditary disease. It usually affects one or more siblings, initially presenting in children at 2 to 5 years of age (1). The main clinical appearance includes papular and nodular skin lesions, gingival hyperplasia, joint contractures and bone involvement in variable degrees (2). The multiple papulonodular lesions, whose diameters are variable, are usually painless and grow slowly. They are mostly seen at the nose, ears, scalp, back and knees. Other clinical symptoms and findings of the disease are flexion contractures of several joints, radiolucent osteolytic lesions in bones and indefinable muscle complaints (1). Joint stiffness can also be observed together with the flexion contractures (3).

In this article, the rehabilitation of 3 siblings with JHF, who had functional loss because of the contractures, is presented and the related literature is reviewed.

CASES

First Child

The first child was a 10-year-old male, who presented with limitation in joint movements and difficulty in walking. According to the patient's history, painful joints, especially the knee and elbow, were first observed when he was 5-6 months of age. Flexion contractures of the knee and elbow joints started to develop at 1 year of age, and swellings on the body surfaces, especially around the ears and on the scalp emerged after 2 years of age. Stretching exercises had been applied to both knee joints in several other centers and no response had been observed. The patient, who had remained undiagnosed until 2 years before, was diagnosed as having JHF by the Plastic and Reconstructive Surgery Clinic after a pathologic examination of masses obtained from occipital, frontal, mental areas, both ear helixes, the back and sternum. He was operated on by an orthopedic surgeon for flexion contractures in knees. The patient, who was also operated on for left elbow flexion contracture 4 months before, was admitted to our clinic for a rehabilitation program. The family history revealed that the parents were consanguineous and 2 of his 5 siblings were also diagnosed with JHF. On physical examination, growth retardation, gingival hyperplasia and operation scars behind the ears, in occipital-mental areas, the back and sternum were observed. There were soft tissue masses 1-3 cm in diameter in the lumbar region, and over the ribs and sternum (Figure 1). Examination of other systems was normal. The patient had normal bed activities and sitting balance and was able to stand up with support, but could not walk on neuromusculoskeletal system examination. On examination of the upper extremities, flexion, abduction and external rotation of the shoulder joint were limited to 80º and 10º, respectively. The elbow range of motion was 90-120º. Dorsal flexion and ulnar deviation of the wrist were limited to 20º and palmar flexion and radial deviation were not possible. On examination of the lower extremities,hip flexion and internal rotation were limited

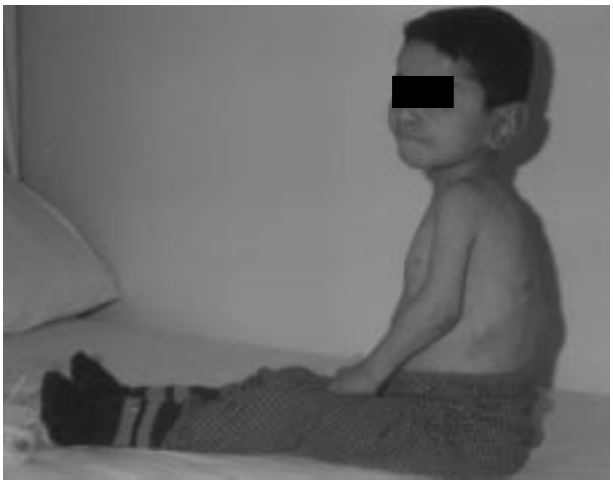

Figure 1. Soft tissue masses in the first child.

after 90º, and 10º, respectively. The knee joint was fixed at extension. The ankle joint was in a neutral position and plantar flexion was not possible. The patient did not have any neurological deficits. According to direct radiological evaluation, although the joint spaces were normal and there was no indication of bone ankyloses, osteoporosis was prevalent.

Second Child

The second child was a 4-year-old male who presented with difficulty in walking. He had pain and limitation in his joints when he was 1 year old and soft tissue masses were observed when he was 2 leading to the diagnosis of JHF. On physical examination, growth retardation, gingival hyperplasia, and pearly hyperemic papular lesions 1-5 mm in diameter on the mouth, chin, ears, occipital, genital and gluteal regions and sternum were determined. The other systems were normal. Bed activities, head and sitting balance were also normal on neuromusculoskeletal examination. He was unable to crawl, kneel or walk, but was able to stand up with continuous support from someone else. The examination of the cranial system was normal. The active range of motion of the bilateral upper extremities could not be evaluated because of cooperation problems. The shoulder joint movements, flexion and abduction were limited to 90º, and internal and external rotations were limited to 20º. Flexion contracture of 30º was noted in the elbow joint. The wrist was on dorsal flexion and ulnar deviation at 10º. Palmar flexion and radial deviation were also limited. All finger joints had flexion contractures. On bilateral lower extremity examination, the internal and external rotations of the hip were limited after 10º. Flexion contractures were observed in both hip joints at 20º and in both knees at 30º. Dorsiflexion was normal and plantar flexion was limited at 10º in the ankles. The laboratory findings were normal. Radiological evaluation indicated normal joint spaces, osteopenic bones and no sign of bone ankylosis.

Third Child

The third child was a 2-year-old girl who presented with limitations in her joint movements. Her parents mentioned that these complaints started in her first year and that skin lesions developed after 1.5 years. Gingival hyperplasia and soft-tissue masses 1-3 mm in diameter on the forehead, ears and chin were found on physical examination. While the patient had head and sitting balance, she did not have standing balance. The other systems were normal. On neuromusculoskeletal system examination, shoulder flexion and abduction were limited to 100º, and internal and external rotations were limited to 10º. There were flexion contractures on her elbows and ulnar deviation on her wrists. Her palmar flexion was limited. There were flexion contractures on both hips at 10º, and on both knees at 45º. Radiological evaluation showed osteopenia.

JHF is a very rare, mesenchimal dysplasia. It was first described in 1969 by Drescher et al. Subsequently, only 30 cases (4) were reported around the world until 1985, and 40 cases (1) were reported in medical articles in English by 1998. It is characterized by the deposition of amorphous hyaline material in the dermis and extracellular spaces of soft tissues (5). A mesenchymal perivascular cellular defect is also evident (6). Its major pathological feature is disseminated hyalinosis in many organs and tissues (7). Mayerda-Silva et al. have described this disease as progressive abnormal differentiation of connective tissue to chondroid tissue (1).

Increased synthesis and degradation of type I collagen in fibroblasts have been shown histologically (1). A loss of hyaline matrix formation, tumoriform lesions, chondroosseous metaplasia and calcium salts are observed (8). All of these pathological findings are caused by joint contructures.

In immunohistochemical studies, type I and type III collagen were observed in hyaline material, while type II and type IV collagen were not. According to quantitative biochemical evaluation, the speed of type I and III collagens was normal (3).

Clinically JHF is an autosomal recessive disorder, which can be diagnosed from the neonatal period to 4 years of age at the latest (7). Since our first case was diagnosed when he was 8 years old, his joint contractures were quite progressed.

The gene related to JHF was identified as a region of homozygosity on chromosome 4q21(9).

The major diagnostic criteria for JHF were a) pearl-like skin papules, translucent plaques and/or nodules, b) gingival hyperplasia, c) osteolytic lesions, d) joint contractures, and e) the histological deposition of amorphous hyaline material (2). There were skin nodules, gingival hyperplasia and joint contractures in our patients. The pathologic evaluation was in accordance with JHF.

The skin lesions may be either multiple or single. These lesions can be small, solid papules, translucent nodules or large subcutaneous masses in several densities. The largest masses tend to localize at the scalp (1).

The hyaline fibromatosis syndromes have an important place among the genetic fibromatosis groups of unknown etiology. The accumulation of hyaline material in different tissues has been described as 2 distinctive clinical syndromes: infantile systemic hyalinosis (ISH) and JHF. ISH differs from JHF as it has visceral involvement (intestinal, cardiac, hepatic and splenic) and a fast fatal ending. Motor development is slow and mental development is normal in both syndromes. Their microscopic findings are not distinctive (2). The clinical course is variable. No articular involvement or only one joint limitation has been described (7). The clinical features of the disease, such as joint contractures, osteolytic lesions and gingival hyperplasia, progress with age (10). The family history is positive for JHF and mental retardation is possible (11).

The differential diagnosis of JHF includes neurofibromatosis, gingival fibromatosis, nodular amyloidosis, infantile systemic hyalinosis, congenital generalized fibromatosis, multicentric infantile myofibromatosis, lipoid proteinosis and Winchester's syndrome.

The essential pathology in JHF making rehabilitation necessary is the contractures and following functional loss. The aim of the rehabilitation of these subjects is to straighten the contractures and prevent their progression or the occurrence of new contractures resulting in functional loss, and to make the patient free to continue his/her daily life.

One can arrange the procedure of the contracture rehabilitation program from simple to aggressive as follows: proper positioning, active range of motion exercise, active assisted range of motion exercise, passive range of motion exercise, static and dynamic splinting, and motor nerve or point blocking. Our patients were also trained in appropriate positioning. A range of motion the exercises was applied to the joints without contracture and stretching exercises were applied to the joints with contracture. The stretch was minimal because of osteoporosis. To reduce the pain, analgesic treatment was given and a hot pack was applied to the patients before stretching. Since the patients were children and securing cooperation from them was difficult, rather than an active range of motion exercises, a passive range of motion was applied 2 times a day. Static splints to the elbow, hand, wrist and lower extremity were given to all 3 children. Vitamin D was recommended for osteopenia. Shoulder, elbow, hip and knee contractures were reduced in the first child. The hip-knee-ankle-foot orthosis was given because of the lower extremity's tendency to flexion, and the patient was ambulated with a walker (Figure 2). All contractures of the second child were reduced by 20º. The patient with improving trunk control was able to stand up. He was ambulated with knee-ankle-foot orthosis and a walker (Figure 3). The reduction in all contractures of the third child was around 10 to 20º. Crawling and kneeling equilibrium responses were provided. The upper extremity contractures had not prevented our patients' daily activities. The third patient was discharged and recommended to return for later walking training because she was not yet old enough to walk (Figure 4).

The family were informed about JHF and given genetic information.

In conclusion, rehabilitation is of a great value in preventing contractures and their progressions, and in stopping the functional loss caused by these contractures.

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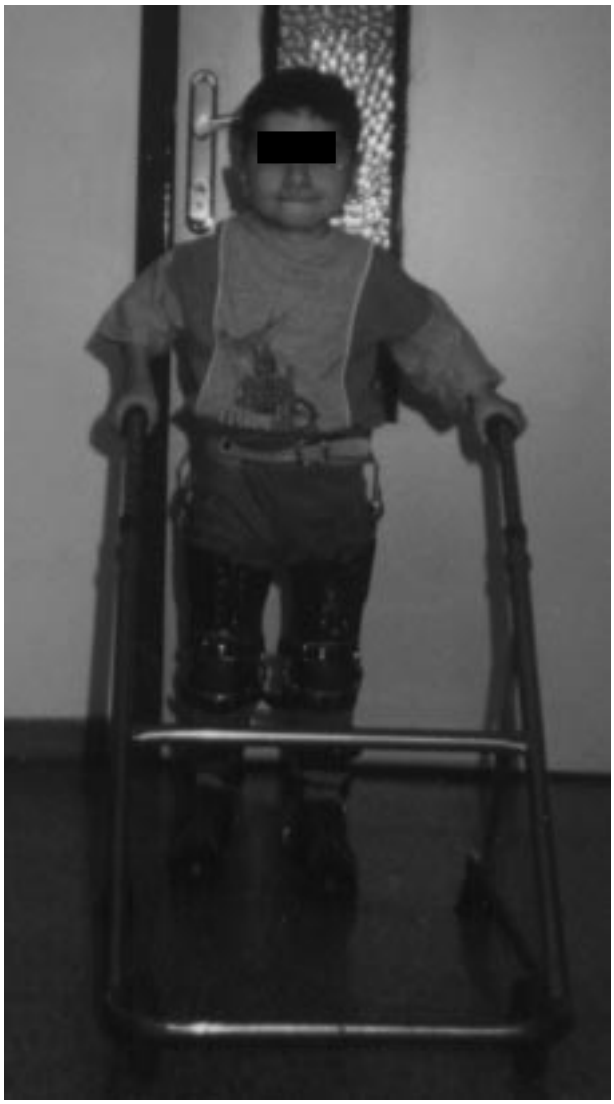

Figure 2. The first child after the rehabilitation program.

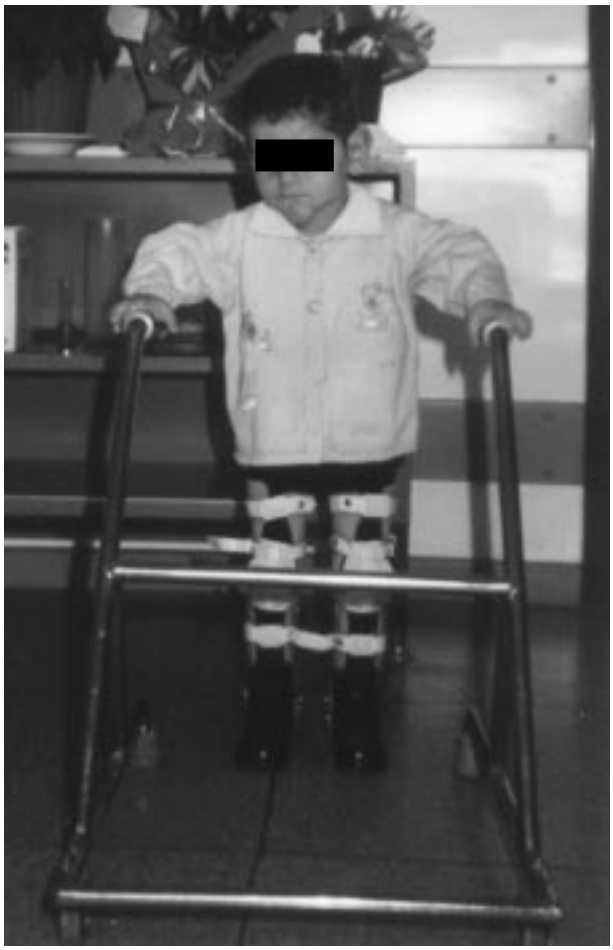

Figure 3. The second child after the rehabilitation program.

Figure 4. Upper and lower extremity static splints in the third child.

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