J Orthop Spine Trauma. 2023 September; 9(3): 149-51. DOI: 10.18502/jost.v9i3.13038

Case Report

Familial Synovial Hypertrophy: A Case Report

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Received: 21 January 2023; **Revised:** 17 March 2023; **Accepted:** 24 May 2023

Abstract

Background: Familial synovial hypertrophy is a rare condition presenting as the flexion contracture of the small joints of the hands and feet, in addition to the large joint effusion and range of motion (ROM) restriction.
Case Report: Herein, we introduce a 14-year-old boy with a long history of deformities in the hand and foot joints. Painless swelling and limited motion of big joints were present since birth. There were no similar diseases in paternal or maternal relatives, while his 9-year-old brother also had a lifelong history of same deformities.
Conclusion: Involvement of the hands and feet can severely restrict young patients' function in daily tasks, raising the importance of early diagnosis and treatment in familial synovial hypertrophy.

Keywords: Familial Synovial Hypertrophy; Joint Contracture; Familial Hypertrophic Synovitis; Synovitis

Citation: Shayan-Moghadam R, Sharafi M, Mahmoudi E, Nabian MH. **Familial Synovial Hypertrophy: A Case Report.** *J Orthop Spine Trauma* 2023; 9(3): 149-51.

Background

Familial hypertrophic synovitis is a rare condition associated with flexion contracture of the hand and feet small joints since early infancy, in addition to the large joints effusion and range of motion (ROM) restriction (1). Despite the rarity of familial hypertrophic synovitis, it should be kept in mind as a differential diagnosis of polyarthritis associated with contracture. Familial hypertrophic synovitis should be considered in the differential diagnosis of juvenile rheumatoid arthritis (JRA), since this disease could be mistaken as a symmetric polyarticular form (2). Only a few studies reported the familial hypertrophic synovitis. Since a correct diagnosis allows for the early commencement of therapy and rehabilitation for patients, allowing for proper growth and development, we report a rare case of familial hypertrophic synovitis that was previously misdiagnosed as JRA.

Case Report

A 14-year-old boy that was born to consanguineous parents was referred to the orthopedic clinic with chief complaints of the hand and feet deformity, difficulty writing and walking.

In the past medical history, we found that he had swollen hand joints since birth; gradually in the first year of life, he developed flexion contracture of swollen joints of the finger especially metacarpophalangeal (MCP) and proximal interphalangeal (PIP) contracture of thumbs, and then some subcutaneous bulging in upper limbs, then bilateral large joint (knees, elbows, wrists, and ankles) swelling happened which was associated with elbow limited motion, and clawing of the toes with the big toe preference (Figure 1). Occasionally, the patient complained of joint pain, especially in the wrists. As the deformity of fingers worsened, doing daily tasks and school writing became difficult for the patient. Besides, due to hip limited motion, sitting crossed legs was impossible for him.

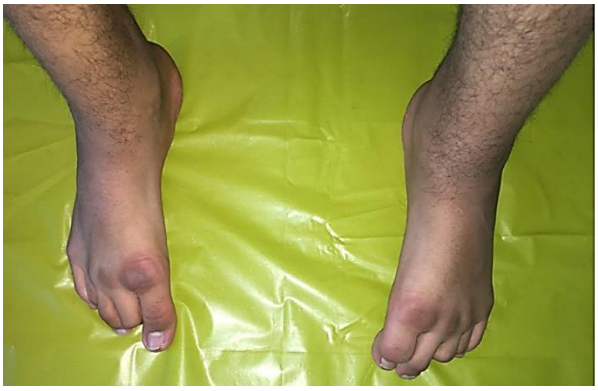

Figure 1. Clawing of the toes with the big toe preference

In his drug history, the patient has been taking methotrexate (MTX) and prednisolone, which did not respond to these drugs. In family history, there have been no reports of identical symptoms in paternal or maternal relatives. The patient in good health with normal weight and height was examined physically. Chest, cardiovascular, and neurological examinations were normal. The patient could independently walk with some difficulty, and the hip's ROM was as follows: 90° of flexion, 15° of internal rotation, 20° of external rotation, and 40° of abduction.

Both knees showed swelling without local warmth and a normal ROM, both ankles revealed a limited and painful ROM, both feet exhibited cavovarus abnormalities, and flexion contracture of the second and third toes was observed. The PIP and MCP of both thumbs exhibited severe flexion contracture in both hands, and the ROM of both wrists was normal but painful. Both elbows were

determined to have flexion contractures (30 degrees).

Laboratory results looked at typical complete blood count (CBC), erythrocyte sedimentation rate (ESR), and C-reactive protein (CRP). Rheumatoid factor (RF) and antinuclear antibody (ANA) returned negative results. The liver function test (LFT) and kidney function test (KFT) results were normal. The knee synovial fluid analysis revealed 450/mm^3 white blood cells (WBCs) (neutrophils: 85%, lymphocytes: 8%).

X-rays of the large joints revealed soft tissue edema, genu valgum, normal bone density, and no signs of a sclerotic or lytic lesion. PIP and cavovarus deformities were visible on X-rays of the hands and feet, respectively (Figure 2).

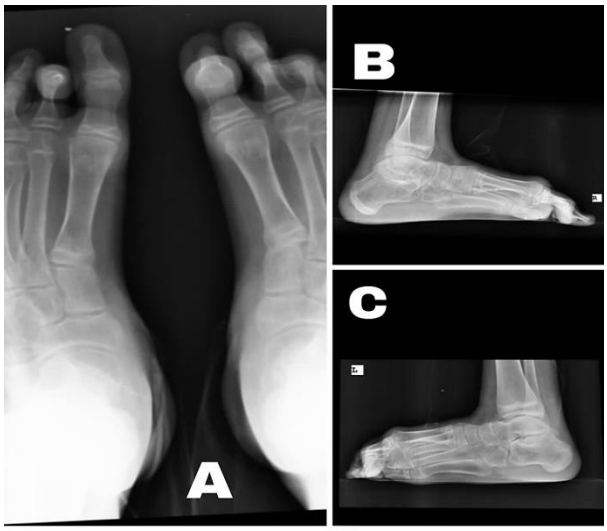

Figure 2. X-rays of the feet showing flexion contracture of the proximal interphalangeal (PIP) joints of the toes
A: Bilateral anteroposterior view; B: Right lateral view; C: Left lateral view

At the age of 4, patient was diagnosed and treated as JRA, and a corticosteroid was injected into the hand's subcutaneous bulge and both knees. His 9-year-old sibling has had swollen and deformed hand and foot joints, as well as limited ROM in his knees and hips since the first month of his birth. Between the two of them, the family has a healthy son and a healthy daughter.

Finally, due to the diagnosis of familial hypertrophic synovitis, we performed plantar fascia release, midfoot and calcaneal translational osteotomy, thumb interphalangeal (IP) fusion, and second finger PIP fusion for our case (Figure 3).

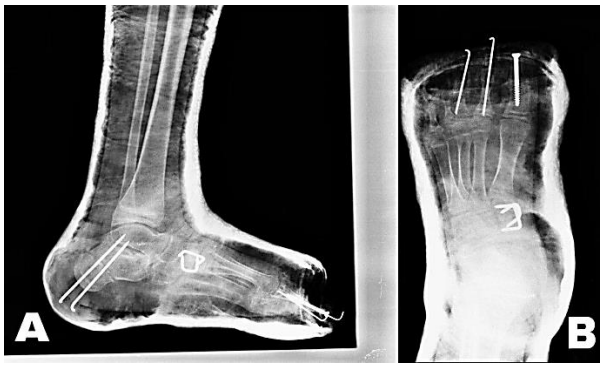

Figure 3. The postoperative X-ray of the patient
A: Post-operative lateral view; B: post-operative anteroposterior view

In 6 months, 1 year, and 2 years of follow-up, our case showed a well condition of feet and walking.

This research was not funded by any individual or organization.

Discussion

In this study, we present a 14-year-old boy with chief complaints of the hand and feet deformity and difficulty writing and walking. The patient's laboratory tests were normal and no evidence of inflammation was observed in the tests. However, X-ray showed PIP and cavovarus deformities of the hands and feet, respectively. At the age of 4, patient was diagnosed and treated as JRA and a corticosteroid was injected into the hand's subcutaneous bulge and both knees. However, the disease progressed and did not response to prednisolone and MTX. Finally, our patient that underwent plantar fascia release, midfoot and calcaneal translational osteotomy, thumb IP fusion, and second finger PIP fusion revealed good foot and walking conditions at 6 months, 1 year, and 2 years.

Familial hypertrophic synovitis clinical manifestation was first described by Jacobs and Downey as a rare condition of familial origin that presents with swelling of the hand and feet small joints in early infancy associated with flexion deformities of the finger and toes and symmetrical large joint involvement (3).

In studies conducted by Jacobs and Downey (3) and Athreya and Schumacher (4), the first diagnostic sign of the disease is a bent thumb which is often noted soon after birth. Later on, the patient develops symmetric joint effusion mainly in the large joints (knees, ankles, hips, and wrists). Pain is not a prominent feature of this disease and the systemic manifestation of fever and inflammation is absent. In their patients, radiography demonstrated soft tissue swelling and in the hips, there was some proximal flattening of the femoral ossification center. The synovial biopsy showed synovial hyperplasia, necrotic villi, deposition of eosinophilic and periodic acid-Schiff (PAS)-positive material, and a large number of multinucleated giant cells. The sedimentation rate is usually normal and the synovial fluid is non-inflammatory. Hammoudeh and Siam reported a 14-year-old male patient that had flexion contracture of the 3rd, 4th, and 5th fingers in addition to the bent thumb (5). This was not seen in patients reported by previous reports. In this study, the synovial biopsy obtained from the knee of the patient showed focal proliferation of the synovial cells, hyalinization of the stroma, and a few giant cells. Moreover, in another study, Rezende et al. reported a case of a 7-year-old boy with flexion finger deformities and painless joint effusion of the large joints since birth, without systemic involvement. The patient had a cousin with similar manifestations (2).

The point of the above studies and our study in terms of diagnosis of familial hypertrophic synovitis is the absence of systemic manifestations (fever, involvement of internal organs) in the studied patients. Thus, fever, rash, or other constitutional symptoms suggest a different diagnosis. CBC, ESR, and CRP showed normal values, and synovial analysis showed a non-inflammatory condition. Moreover, synovial biopsy showed synovial hypertrophy, necrotic villi, PAS-positive material, and giant cells. Unlike previous reports, our case developed cavovarus deformity of both feet which made walking difficult and severe hand deformity deteriorated writing skills making it difficult to continue his education.

Familial hypertrophic synovitis should be considered in rare cases where there are atypical polyarticular symptoms with contractures that did not get better with standard treatment. In these cases, treatments with stronger anti-inflammatory drugs that are not good for this condition should be avoided. A correct diagnosis allows for the early commencement of therapy and rehabilitation for patients, allowing for proper growth and development.

Conclusion

Familial hypertrophic synovitis should be considered in the differential diagnosis of JRA, since this disease could be mistaken as a symmetric polyarticular form. Pain and functional limitation are not completely absent. Orthopedic reconstruction surgeries can be helpful to decrease the difficulties of these patients and elevate their condition.

Conflict of Interest

The authors declare no conflict of interest in this study.

Acknowledgements

We thank the patient and his family for their participation in the study and signing the informed consent.

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