

# Bilateral Sacroiliitis in a Patient with H Syndrome

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## Abstract

H syndrome is an autosomal recessive form of histiocytosis. The hallmark of this rare syndrome is the presence of cutaneous hyperpigmentation with hypertrichosis overlying an area of indurated skin. Other characteristic findings include short stature, hearing loss, hyperglycemia, heart anomalies and flexion contractures. We report a young girl with H syndrome presenting with bilateral sacroiliitis and bilateral uveitis and we ask the question "Are these features coincidental or part of the syndrome?".

**Keywords:** H syndrome; Sacroiliitis; Histiocytosis; Interphalangeal joints; Melanocytic nevus

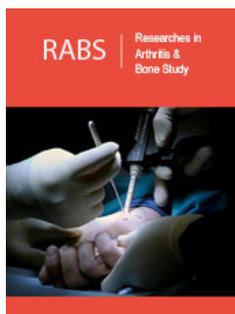
## Introduction

H syndrome (OMIM: 602782) is a rare inherited form of histiocytosis. It is an autosomal recessive disorder caused by a mutation in SLC29A3 gene resulting in histiocytic infiltration of numerous organs [1,2]. It becomes clinically apparent mostly during childhood with characteristic hyperpigmented hypertrichotic indurated skin lesions that mainly involve the lower limbs. Other reported features include Sensorineural hearing loss, heart anomalies, hepatosplenomegaly, lymphadenopathy, insulin dependent diabetes mellitus and flexion contractions of interphalangeal joints [3,4]. we report sacroiliitis, a possible new feature, in a young girl diagnosed with H syndrome.

## Case Description

### A 16-year-old Syrian girl

In 2016 (13 y old), she presented complaining of pain and inability to move her fingers. Upon examination, she was found to have tender Boutonniere like deformity of both litter fingers with clinical evidence of synovitis (Figure 1). Her past medical history was significant for type one diabetes mellitus and bilateral hyperpigmented hypertrichotic skin lesions in lower limbs (Figure 2) diagnosed as epidermal melanocytic nevus based on skin biopsy. She was born to consanguineous parents, and she is up to her age regarding growth and development. Her laboratory workup revealed raised inflammatory markers and positive rheumatoid factor of 40IU/ml. All other autoimmune tests were negative. She was diagnosed to have juvenile idiopathic arthritis and started on NSAIDs with methotrexate. Her pain improved but she continued to have new deformities of her fingers and toes. Her skin lesions developed at 11 years of age and with time they became more indurated. She was reviewed by multiple dermatologists and biopsy was taken 3 times with almost the same description but with different impressions before H syndrome was suspected by dermatologist (Dr. Mariam Baqi). Genetic studies revealed the presence of the pathologic variant c.1279G>A in the SLC29A3 gene in homozygous state confirming the diagnosis of H syndrome. Sadly, other manifestations of H syndrome continue to appear as she developed Sensorineural hearing loss in her right ear and is currently being evaluated by cardiology to rule out cardiac anomalies. After she was diagnosed to have H syndrome, her joints deformities were attributed to the syndrome and methotrexate was stopped. With follow up in rheumatology clinic, she developed new joints symptoms, inflammatory back pain, and bilateral anterior uveitis. MRI of the sacroiliac joint revealed bilateral sacroiliitis (Figure 3). HLA B27 was negative. Accordingly, she was started



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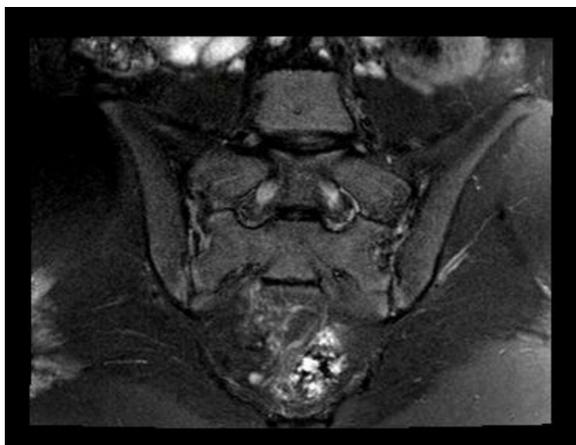
on adalimumab and methotrexate with significant improvement in her symptoms.



**Figure 1:** Hands of the patient showing Boutonniere like deformity of both little fingers.



**Figure 2:** Bilateral hyperpigmented hypertrichotic skin lesions in lower limbs of the patient.



**Figure 3:** Subchondral edema are noted at the anterior inferior aspects of the sacroiliac joints bilaterally.

## Discussion

Since its first description as an entity by Molho-Pessach V et al. [3], H syndrome is increasingly being reported worldwide. To date, around 130 patients have been described in world literature [5]. The pathophysiology of this syndrome is centered around a genetic mutation in the SLC29A3 gene leading to multiorgan histiocytic infiltration causing what is currently recognized as monogenic autoinflammatory disease [6]. The hallmark of H syndrome is the appearance of pigmented thickened skin patches with overlying hypertrichosis typically affecting the lower limbs. A wide variety of other systemic features have been described including sensorineural hearing loss, hypogonadism, heart anomalies, hepatosplenomegaly, insulin-dependent diabetes, short stature, and lymphadenopathy [3,4,7-10]. We are interested in the rheumatologic manifestation of this syndrome. The presence of recurrent episodes of inflammatory polyarthritis with Boutonniere like deformity of both little fingers along with positive rheumatoid factor and raised inflammatory markers led to the misdiagnosis of juvenile idiopathic arthritis in our patient. Several case reports have demonstrated the presence of arthritis as part of the systemic manifestation of H syndrome [7,9,11]. However, our patient developed another manifestation that we did not come across in our literature review. She developed bilateral sacroiliitis along with bilateral anterior uveitis. These findings in our patient raise the question whether she truly has a rheumatic illness beside H syndrome, or merely represent newly recognized manifestation of the H syndrome. Similar to other reported cases, our patient showed positive response to methotrexate and adalimumab treatment with improvement in her joints and eyes related manifestations along with decrements in inflammatory markers [9].

## Conclusion

H syndrome is a recently recognized autosomal recessive condition with characteristic dermatologic and systemic manifestation. Along with arthritis, sacroiliitis and uveitis could represent part of the phenotypic description of this rare entity.

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