

Case Report: Job's Syndrome

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Introduction

First described in 1966, the hyperimmunoglobulin E (hyper-IgE or HIE) syndrome is a rare immunodeficiency disorder with autosomal dominant inheritance pattern. HIE syndrome has variable expressivity and is associated with multiple abnormalities. The most common findings are recurrent skin abscesses (hence the name "Job's Syndrome", that comes from the Old Testament Book of Job. Job was tormented with plagues of boils etc.), pneumonia with pneumatocele development, and high serum levels of IgE. Facial, dental, and skeletal features are also associated with this syndrome [1].

Patient history

37-year-old female patient with lumbar and pelvic pain, multiple hyper- and atrophic cicatrices due to surgical interventions and spontaneous suppuration presents for MR postoperative evaluation. The onset of the disease was shortly after birth with apparition of abscesses of the capilitium at the age of 7 months and eczematous skin of the body and the extremities with different aspect and density, accompanied by pus formation. Erithemo-papulous itchy rash with tendency to lichenification had unsuccessfully been treated as eczema. Job's Syndrome was diagnosed at the age of 3 years by histological and micro-

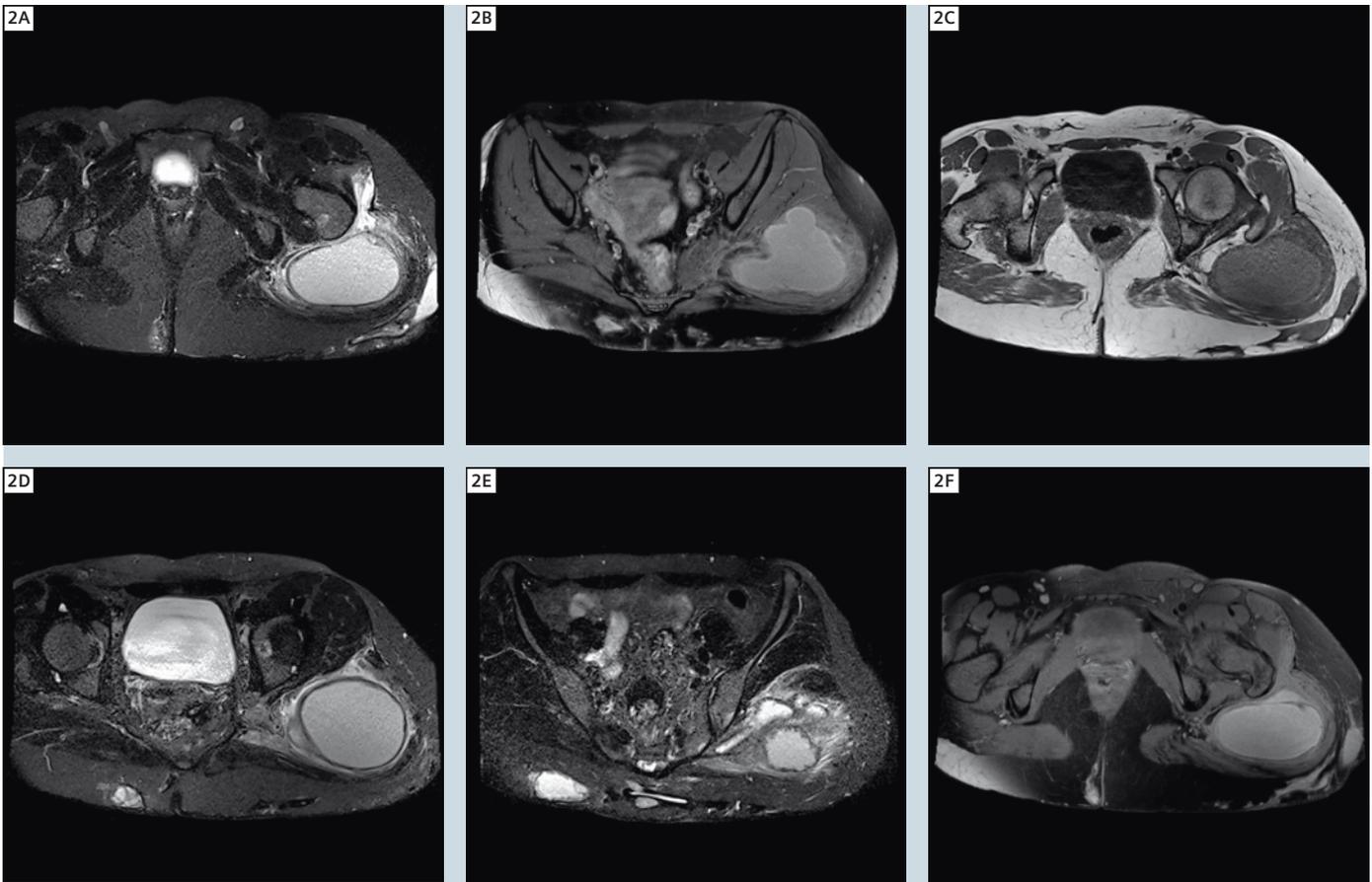
biological examination, and several skin resections, including incisions and necrectomies, were performed. Erythrocyte sedimentation rate (ESR) was 67 mm, C-reactive protein (CRP) 88, Hb 97 and total IgE > 4000 IU/ml (with normal values up to 120 IU/ml).

Sequence details

The following images were acquired on our 1.5T MAGNETOM ESSENZA using a 4-channel body matrix coil. The T2 sequence in axial and sagittal plains is using the following parameters: TR 2600 ms, TE 83 ms, matrix (300 x 448) px², FOV 380 mm, phase



1 Presenting an ovoid, capsulated formation at the level of the left m. gluteus maximus, passing between m. adductor magnus and m. vastus lateralis. (1A) coronal T1, (1B) sagittal T2, (1C) axial T2.



2 (2A) Shows a small amount of liquid close to the iliotibial tract; (2B) edematous m. gluteus maximus and m. piriformis; (2C) subcutaneous liquid collection in communication with the skin. (2D) shows the collection at the level of coccygeal bone; (2E) collection with fistula at the level of sacroiliac joint; (2F) collection behind the rectum with skin fistula formation. Axial TIRM (2A, F), axial PD fat sat (2B, D, E), Axial T1 (2C) images.

FOV 100%, bandwidth 260 Hz/px, flip angle 150°, base resolution 448, phase resolution 100%.

The T1 sequence in axial, sagittal and coronal plains is using the following parameters:

TR 680 ms, TE 8,5 ms, matrix (300 x 448) px², FOV 380 mm, phase FOV 100,0%, bandwidth 260 Hz/px, flip angle 150°, base resolution 448, phase resolution 100%.

The T2 TIRM sequence in the axial plain is using the following parameters:

TR 5560 ms, TE 80 ms, matrix (258 x 384) px², FOV 360 mm, phase FOV 67,2%, bandwidth 260 Hz/px, flip angle 150°, base resolution 384, phase resolution 100%.

The T1 VIBE sequence with fat suppression is using pre-and post contrast injection

with the following parameters:

TR 5.2 ms, TE 2.4 ms, matrix (319 x 384) px², FOV 340 mm, phase FOV 100%, bandwidth 410 Hz/px, base resolution 384, phase resolution 83%, slice resolution 67%.

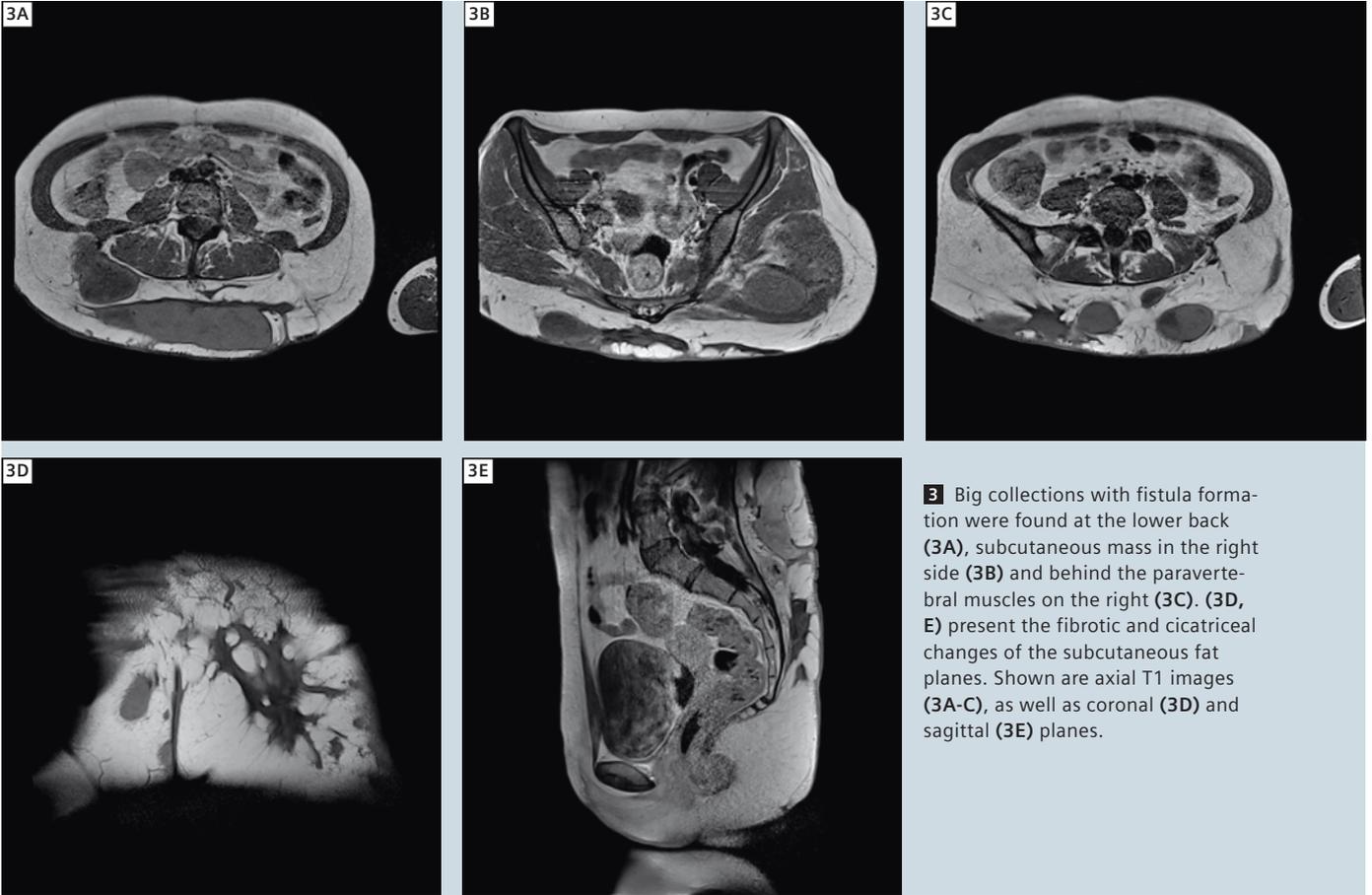
The PD sequence with fat suppression in axial plain was acquired using the following parameters:

TR 2200 ms, TE 12 ms, matrix (304 x 512) px², FOV 380 mm, phase FOV 50%, bandwidth 165 Hz/px, base resolution 512, phase resolution 100%.

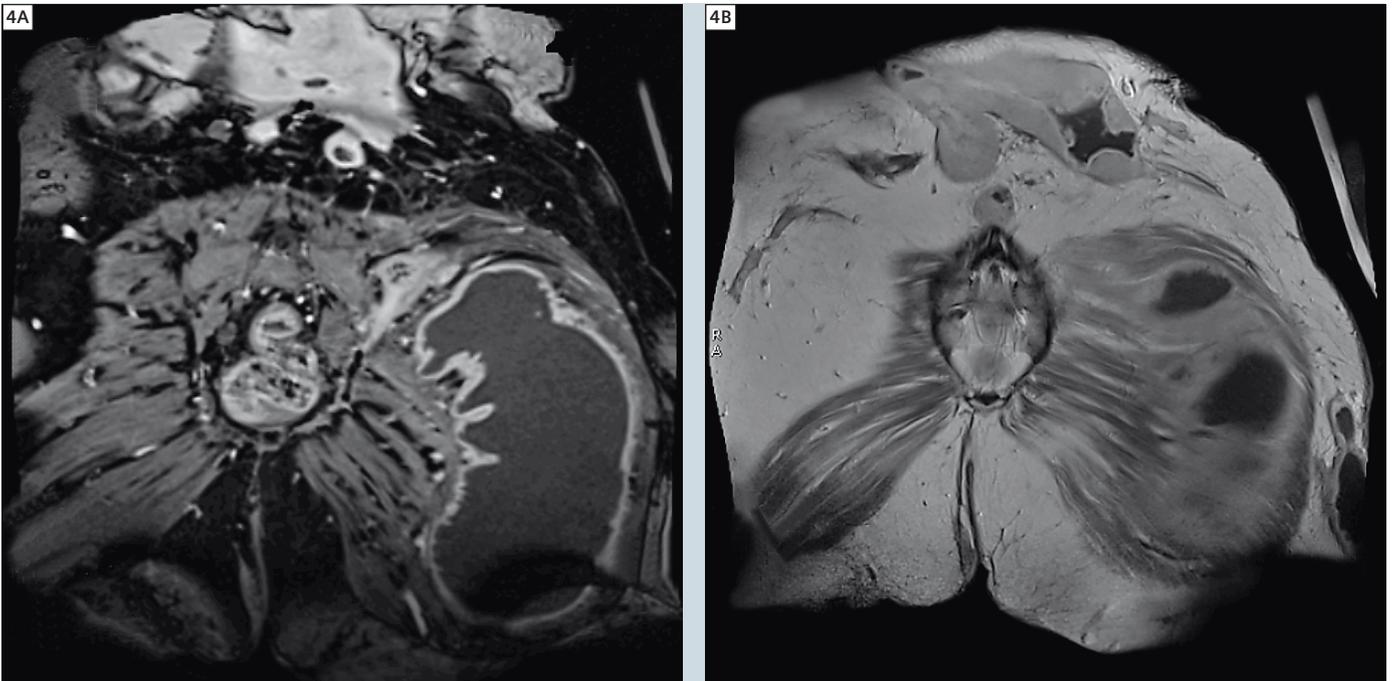
Imaging findings

MRI examination (1.5T Siemens MAGNETOM ESSENZA) at the level of the lower back and pelvis revealed a huge, ovoid, capsulated formation at the level of the left musculus (m.) gluteus maxi-

mus measuring 18/10/5 cm. The mass propagates in caudal direction and is passing through the lateral femoral intermuscular septum, between m. adductor magnus and m. vastus lateralis and is located approximately 10 cm distal to the trochanter major. The content of the lesion presents with hyperintensity on T2 and hypointense on T1 images and corresponds to pus (Fig. 1A-C). There is also a small amount of liquid close to the iliotibial tract (Fig. 2A), accompanied by edematous m. gluteus maximus and m. piriformis. Figure 2B at the level of the trochanter major on the left side reveals two subcutaneous liquid collections with the same signal intensity, both in communication with the skin (Fig. 2C). Additional liquid collections are found on the right side, subcu-



3 Big collections with fistula formation were found at the lower back (3A), subcutaneous mass in the right side (3B) and behind the paravertebral muscles on the right (3C). (3D, E) present the fibrotic and cicatricial changes of the subcutaneous fat planes. Shown are axial T1 images (3A-C), as well as coronal (3D) and sagittal (3E) planes.



4 Following gadolinium injection there was significant contrast enhancement from the periphery of the lesions on T1 VIBE sequence with fat suppression (4A) and on the T1 sequence (4B).

taneously, at the level of the coccygeal bone (Fig. 2D), the sacroiliac articulation with fistula formation (Fig. 2E) and close to the rectum with again pararectal skin fistula formation (Fig. 2F).

Another distinctive collection with fistula formation was found at the lower back, at the level of the biiliac line subcutaneously at the left measuring 12/8/4 cm in the left and medially (Fig. 3A) and a smaller one measuring 4/3 cm in the right side (Fig.3B). A mass measuring 6/3 cm has been found behind the paravertebral muscles in the fat tissues at the right side of the back (Fig. 3C). A massive fibrosis of the subcutaneous fatty tissues in addition to hyper- and atrophic cicatrices resulting from surgical interventions and spontaneous suppuration has been noted (Fig. 3D, E).

Following gadolinium injection there was significant contrast uptake from the capsules of the described lesions as well as from the inflammatory changes of the fatty planes (Figs. 4A, B).

Besides the described soft tissue lesions a small amount of liquid has been detected in the left sacroiliac joint with associated high signal intensity zones in T2 fat sat, corresponding to bone edema in the context of left side sacroileitis (Fig. 8).

Discussion

Autosomal dominant hyper IgE syndrome (AD-HIES), formerly known as 'Job's Syndrome', is a genetic disease affecting several body systems including the immune system. AD-HIES is characterized by abnormally high levels of an immune system protein called immunoglobulin E (IgE) in the blood. Some cases

of Job's Syndrome have been found to be due to a genetic defect in a protein called STAT3 and mutations in the STAT3 gene, but in many others, the cause is unknown [2, 3]. Job's Syndrome's STAT3 gene resides on chromosome 17 on the longer arm. HIES often appears early in life with recurrent staphylococcal and candidal infections, pneumonias, lymphadenitis, conjunctivitis and eczematoid skin. Characteristic facial, dental, and skeletal abnormalities have also been described. Patients with HIES have either delay of or failure in shedding of primary teeth. The characteristic coarse facial features are usually set by age 16. These include facial asymmetry, a prominent forehead, deep-set eyes, a broad nasal bridge, a wide, fleshy nasal tip, and mild prognathism. Additionally, facial skin could be rough with prominent pores. Finally, some patients have scoliosis, as well as bones that fracture easily [4]. A review of reported cases of lymphoma in Job's Syndrome indicates an increase in relative risk [7].

Our patient presented with recurrent suppurative skin infections, chronic eczematoid rash and large skin abscesses. Increased number of eosinophils in blood and high levels of total IgE was also present. People with Job's Syndrome tend also to have high levels of anxiety, depression, and guilt. Recurring pneumonias and fungal infections like infections of the mouth or of the nails were not present in our patient as well as bone fractures. As skin manifestations were dominant, we found the diagnostic value of MRI of utmost importance regarding the precise determination, localization and extent of the lesions,

the effect from previous resections and the fibrotic and cicatricial soft-tissue changes. Due to the excellent contrast resolution, MRI was also useful in our case in considering further surgical treatment, since it provides optimum contrast and high spatial resolution to visualize soft-tissue details demanded by the dermatologists. Fat suppression is often essential in musculoskeletal imaging to obtain superb contrast. We consider MRI a superior imaging modality to computer tomography (CT) in delineating the extent of the disease [5]. Necrotizing fasciitis and gas gangrene are more common necrotizing infections that also cause necrosis of skin, subcutaneous tissue, and muscle [6]. They present with different aspects and are easily differentiated by the cutaneous abscesses within the dermis and deeper skin and soft tissues in our patient.

Conclusion

Contrast-enhanced MRI is a particularly sensitive method for detecting soft-tissue masses, as well as their precise localization and extent. For this purpose different sequences are available nowadays. Our case is an example of the usefulness of MRI as a powerful non-invasive technique in medical diagnosis, applied here to analyze skin disorders and deeper soft-tissue masses with various localizations.

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