LIMB GIRDLE MUSCULAR DYSTROPHY TYPE 2B - A RARE MYOSITIS MIMIC
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Background: Proximal muscle weakness with associated raised creatine kinase (CK) commonly leads to referral to Rheumatology for the investigation of Idiopathic Inflammatory Myopathy (IIM). Some genetic myopathies can have a similar presentation with investigations that suggest inflammatory disease, leading to difficulty with accurate diagnosis (Amato & Brown, 2011; Harlan & Mammen, 2019).

Objectives: To describe the case of a patient with Limb Girdle Muscular Dystrophy Type 2B (LGMD2B), whose initial presentation mimicked an inflammatory myopathy.

Methods: Case report.

Results: A 43-year-old patient was reviewed by Rheumatology due to proximal muscle weakness with a raised CK. Muscle biopsy was suggestive of inflammatory myopathy. Therefore, he was started on treatment with corticosteroids. Corticosteroid treatment resulted in no improvement in his weakness or CK. His diagnosis was reviewed, and he was referred to the Neurology and Genetics services. Following molecular genetic analysis, a diagnosis of Limb Girdle Muscular Dystrophy Type 2B was made.

Conclusion: Muscle biopsies can suggest an inflammatory aetiology in some genetic myopathies (Amato & Brown, 2011; Harlan & Mammen, 2019). If a patient with suspected IIM presents with atypical features, or they do not respond as expected to treatment, then consider a genetic myopathy such as LGMD2B as a cause and involve the Neurology and Genetics services in the case.

REFERENCES:

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OSTEOPIOKILOSIS IN PATIENT WITH ANKYLOSING SPONDYLITIS
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Background: Osteopoikilosis (OPK) – a rare congenital genetic-mediated benign sclerosing disease of skeleton. OPK is usually clinically asymptomatic and is most often an incidental finding during an X-ray study [1]. The lesions are from 2 to 10 mm and are located in the area of the ischial, pubic bones, metaepiphyseal regions of the tubular bones. The axial skeleton is usually not affected in OPK. As early described, OPK may be associated with inflammatory rheumatic diseases, such as rheumatoid arthritis, spondyloarthritides, reactive arthritis and scleroderma [2-4]. Ankylosing spondylitis is a chronic rheumatic disease from the group of spondyloarthritis with obligatory involvement of the sacroiliac joints and frequent involvement of entheses and peripheral joints. Although a significant mutual influence of OPK and AS has not been proven, their combination can lead to unreasonable over-examination in real clinical practice.

Objectives: To present a clinical case of rare coexisting of osteopoikilosis and ankylosing spondylitis.

Methods: Case report. The patient was treated and examined based on real clinical practice.

Results: A 33-year-old man was admitted to our rheumatology department with a three-year history of inflammation pain in cervical region of spine and hip, knees and shoulder joints and long morning stiffness duration (up to 3 hours); AS was earlier verified based on modified New York criteria (1984) and Nonsteroidal anti-inflammatory drugs were prescribed promptly. On the outpatient basis he also received sulfasalazine and methotrexate due to arthritis and enthesitis. Nevertheless he had high disease activity (ESR 66 mm/h, CRP 775 mg/l, BADAI = 75), severely limited range of motion in the cervical, thoracic and lumbar spine, arthritis, enthesitis when he entered in our clinic. He had been treated with secukinumab 150 mg for fourteen months and then he was switched to golimumab 50 mg per month because of lack of efficacy. Low disease activity has now been achieved.

Conclusion: We’ve reported rare case of AS and OPK coexisting in young patient. The identification of typical radiological phenomena and correct evaluation of clinical data may allow to avoid unnecessary expensive examinations.

REFERENCES:

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A CASE OF ATYPICAL MYCOBACTERIUM INFECTION COMPLICATING EXTRA-NODAL ROSAI-DORFMAN DISEASE IN A PATIENT WITH SYSTEMIC LUPUS ERYTHEMATHOUS (SLE)
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Background: We present the case of a 28 year old Black-British female with severe SLE requiring treatment with rituximab in 2012 due to persistent low-grade activity and severe episodes of pleuro-pericardial effusions, pancytopaenia, fever and weight loss.

During the assessment of radiographs of the pelvic bones and spine, typical changes of the late stage of ankylosing spondylitis (syndesmophytes, stage 4 sacroiliitis) were revealed at the first visit in our clinic. At the same time, a number of phenomena were discovered that were not related to the course of the AS, which required expanding the differential diagnosis range. The tumor process and endocrine pathologies were excluded. OPK was diagnosed on the basis of characteristic changes in various parts of the skeleton, especially in phalanges, wrists and pelvis bones (fig.1).

Figure 1. OPK spots in phalanges and wrists.

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Her other background includes beta-thalassaemia trait and excision of calcific fibrotic tissue on bilateral anterolateral orbits in 2015. In 2018 she reported an 18-month history of non-tender, non-fluctuant, slow growing left thigh mass with USG revealing a well demarcated subcutaneous complex cystic lesion of ~2x4x7 cm. There was no preceding trauma or skin infection. Histology from a needle biopsy revealed diffuse histiocytosis with positive immunohistochemistry (IHC) for S100, CD68 and CD31, it was negative for CD1a, consistent with Extra-nodal Rosai-Dorfman disease (RDD).

She developed constitutional symptoms after reporting months of gradual weight loss with gradual ESR, CRP rise and leucocytosis. Her SLE symptoms were stable and given lack of SLE-specific symptoms; PET-CT was used to identify systemic RDD; the thigh mass showed strong FDG avidity along with a small focus of uptake in the small bowel, thought to be RDD related with no other areas of uptake. She had ongoing ooze from the enlarging thigh lesion (5 x 26 x 15 cm), this was sent for MCS and AAFB; which isolated Mycobacterium avium. She was treated with rifampicin, ethambutol and clarithromycin resulting in improved thigh lesion, constitutional symptoms and inflammatory markers.

Objectives:
1. To describe a rare associated complication of severe SLE and to educate and inform clinicians regarding possible masquerades of disease.
2. To education and inform about the approach to diagnosis of mycobacterium infection.

Methods: Case report and literature review.

Results: Mycobacterium infections rarely cause RDD; to date, only one case report is published involving an HIV infected patient with RDD confirmed on LN biopsy presenting with splenomegaly and treated with oral corticosteroids (OCS) complicated by Mycobacterium avium complex and Salmonella enterica confirmed on bone marrow biopsy/culture, similar to our patient, he presented with constitutional symptoms and weight loss [2]. Mycobacterium can also mimic RDD, a case report has described a 74 year old with tender lymphadenopathy diagnosed with RDD on LN biopsy. She was treated with IV and OCS, but was unresponsive. A repeat LN biopsy and CT imaging revealed the presence of mycobacterium kansasii; her biopsy was positive for CD68/S100 throughout. Of note, she had high levels of anti-interferon autoantibodies and was diagnosed with adult-onset immunodeficiency syndrome [3].

Conclusion: This case illustrates the need for a MDT approach for multi-system diseases such as SLE and RDD, and the need to consider atypical infections when blood tests are incongruent with clinical state.

REFERENCES:

Image 1.