OP0079  LIMB GIRDLE MUSCULAR DYSTROPHY TYPE 2B - A RARE MYOSITIS MIMIC

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Background: Proximal muscle weakness with associated raised creatin 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:

Disclosure of Interests: None declared

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OP0080  OSTEOPOIKILOSIS 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). 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:

Disclosure of Interests: None declared

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OP0081  A CASE OF A TYPICAL MYCOBACTERIUM INFECTION COMPLICATING EXTRA-NOdal ROSAI-DORFMAN DISEASE IN A PATIENT WITH SYSTEMIC LUPUS ERYTHEMATOSUS (SLE)

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Background: 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:

Disclosure of Interests: None declared

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Figure 1. OPK spots in phalanges and wrists.
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 ~2x4x7cm. There was no preceding trauma or skin infection. Histology from a needle biopsy revealed diffuse histiocytosis with positive immunohistochemistry (ICH) 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, 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 complicate 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 (3).

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:


Table 1.

<table>
<thead>
<tr>
<th>SLE Clinical History</th>
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<tbody>
<tr>
<td>Diagnosed 2006 (97 ACR Classification Criteria)</td>
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<tr>
<td>Clinical</td>
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<tr>
<td>Polyarthritis</td>
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<tr>
<td>Glomular (lacrimal swelling)</td>
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<tr>
<td>Pericardial effusion/Pleural Effusion</td>
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<tr>
<td>Lupus</td>
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<td>Scleroderma</td>
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<td>Rheumatoid arthritis</td>
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<td>SLE</td>
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<tr>
<td>Lupus anticoagulant</td>
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<tr>
<td>Anti-cardiolipin (aCL)</td>
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<td>Anti-beta 2-glycoprotein (B2GPI)</td>
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<td>Anti-SS-A/Ro</td>
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<td>Anti-SS-B/La</td>
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Challenges for Patients and Patients’ Organisations in Times of the Pandemic

OP0082-PARE

THE EFFECT COVID-19 HAS ON THE MENTAL HEALTH OF PEOPLE LIVING WITH RHEUMATIC DISEASES. FROM DATA TO INTERVENTIONS.

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Background: Covid-19 has had an important impact on the mental health conditions of over 5 million Italians suffering from one of the over 150 rheumatic diseases. In order to understand the psychological impact of the Covid-19 emergency and the restrictions imposed to counter it, the Italian National Association of People with Rheumatic and Rare Diseases – APARR M全国 E launched the research “Living with a rheumatic pathology”.

Objectives: Gather data directly from Italian patients about the impact of the COVID-19 and consequent restrictions on their mental health and feelings; evaluate the most effective intervention to be implemented to face the pandemic by Patients organization.

Methods: A qualitative-quantitative survey was carried out through a questionnaire administered throughout the national territory to a sample of N = 1,001 people. The people invited to complete the questionnaire were women (55.9%) and men (44.1%), aged 18-85 years (age 18-41 = 26.7%; age 42-65 = 64%; age >65 = 9.3%) with at least one rheumatic pathology. The questionnaire was made up of 39 questions, of which 29 were closed and 10 were open. For the administration of the questionnaires, the CAWI (Computer Aided Web Interview) methodology of on-line survey was used. The 1,001 interviews were carried out from 7 to 14 August 2020.

Results: More than 4 out of 10 people (total sample 44.2%; male 60%, female 35.7%; age 18-41 = 39.1%; age 42-65 = 45.9%; age >65 = 50%) declared that the emergency period has somehow caused a worsening of their health condition. People declared that the deterioration of their health is due to the emergency period for the following reasons: 1) Psychological: such as stress and anxiety; “Too much stress and anxiety made the symptoms worse.”; 2) Inability to perform physiotherapy and motor activities due to the lockdown 3) Postponement of examinations, visits and checks 4) Remote working, in some cases described as harmful for people’s mental and physical health: “Due to Covid19 I had to do remote working and I worked even 12 hours a day including holidays to the detriment of my family life.”

Furthermore, from January 31, 2020 a significant increase emerged in communicable problems with rheumatology specialist compared to the period before the emergency due to Covid-19. The sharp increase may be due to the situation of severe psychological stress to which also the doctors were subjected in the emergency phase: people could not find the comfort of being empathically listened to.