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A case report of severe idiopathic dermatomyositis with NXP-2 positive antibody

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Abstract

Dermatomyositis (DM) is a rare myopathy associated with muscle weakness and inflammation. It often poses diagnostic difficulty for clinicians. In recent times, various antibodies including anti NXP-2 have been identified and may aid diagnosis. The aim of this case report is to increase awareness and knowledge among clinicians through shared experience of severe cases of DM. Here, we present the case of a 22-year-old Caucasian female, with no previous medical history, who was diagnosed with severe DM. She presented to the Emergency Department with proximal weakness of all four limbs and a skin rash. Clinical examination, imaging and laboratory investigations confirmed the diagnosis of NXP-2 positive DM. Treatment was initiated with high dose intravenous corticosteroids then switched to oral steroids and eventually her condition improved over time.

Keywords

dermatomyositis; muscle; oral steroids

Introduction

Dermatomyositis is an idiopathic inflammatory myopathy with characteristic cutaneous findings that may occur in children and adults [1]. It is a systemic disorder that commonly affects the skin and the muscles but can have extra-muscular manifestations [1,2]. Distinct clinical phenotypes of DM have been linked to various circulating auto antibodies [3,4].

In our case, nuclear matrix protein (NXP-2) auto antibody was identified. NXP-2 antibodies have been found in previous studies to be associated with 25% of cases of juvenile DM and recently has been identified in adult cases [4]. Unfortunately, NXP-2 positivity has been described in cases of adult DM strongly associated with malignancy [5]. NXP-2 positivity has also been associated with calcinosis (often a late complication), severe muscle weakness and gastrointestinal tract involvement [4].

Case Presentation

A 22-year-old previously healthy Caucasian female was brought in by ambulance to the Emergency Department with severe upper and lower limb weakness, a rash on the face and dorsum of both hands (Figure 1) and inability to walk. She became unwell initially in March 2018 when she attended her General Practitioner (GP) complaining of pain in both thighs. Subsequently her legs

became weak, followed by her shoulder girdle over a period of four weeks. She was referred to the tertiary care hospital by her GP, at which point she had developed a rash on the dorsum of both hands and mild weakness of the proximal upper limbs. While there, she was evaluated for olymyositis/dermatomyositis and she had a full body Magnetic Resonance Imaging (MRI). The MRI report was consistent with diffuse active muscle inflammation, features that were compatible with dermatomyositis (Figure 2). Computerized Tomography (CT) and MRI scans were negative for any occult malignancy. She was commenced on oral prednisolone 60 milligrams (mg) once daily (qd) and discharged home. After a period of ten days at home, her weakness progressed to the extent where she was unable to walk due to lower limb weakness or carry out any activities of daily living that involved the use of her upper limbs. She also developed swallowing difficulties. At this point, her parents decided to call an ambulance to take her to the nearby hospital.

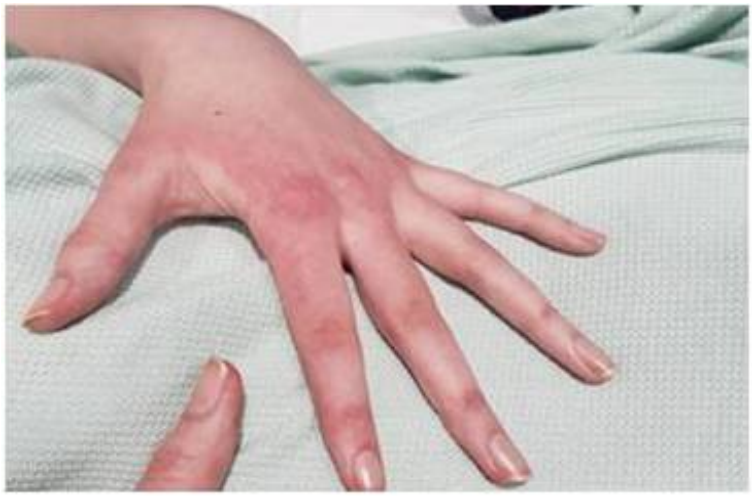

Figure 1: Rash on the dorsum of the both hands

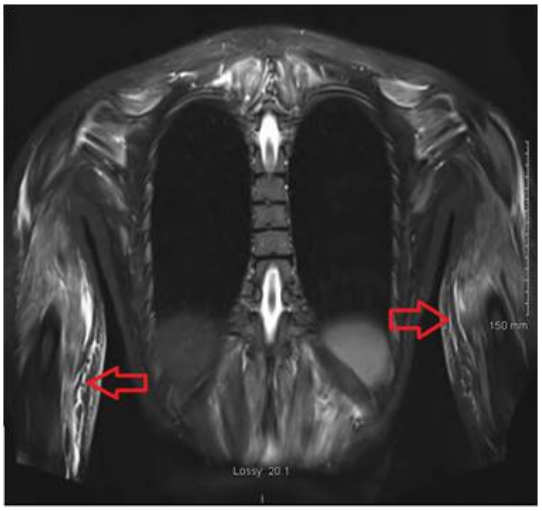

Figure 2: RMRI showing diffuse muscle inflammation and subcutaneous oedema

On examination, she was found to be proximally weak in all four limbs, with power of 2/5. Distal power in all four limbs was 4/5 on initial examination. Reflexes were reduced at the patella, but present at the ankle bilaterally. Plantar reflexes were down-going bilaterally. Biceps and triceps eflexes were absent bilaterally but the brachioradialis reflex was preserved bilaterally. Cranial nerves, sensation and coordination were all intact. On the skin, a macular, violaceous rash was visible on the face and on the dorsum of both hands (Figure 1).

Initial blood results revealed a Creatine Kinase (CK) of 12,415 U/L (29-168), aspartate transaminase (AST) 441 U/L (5-34), Lactate Dehydrogenase (LDH) 710 IU/L (125-220), erythrocyte sedimentation rate (ESR) 31 mm/hr (0-12), Haemoglobin 14.4 g/dl (12-15) normal CRP.

Based on the blood results and clinical history and examination, a provisional diagnosis of severe dermatomyositis was made that was unresponsive to the current dose of oral steroid. She was reviewed by the Rheumatologist, who commenced her on IV methylprednisolone 500mg/d, advised the team to send a muscle biopsy, autoimmune screen and myositis antibody panel. She was also started on physical therapy. Unfortunately on day 3, the patient then deteriorated to the extent that she developed significant truncal weakness and was unable to maintain her posture. She developed significant oedema of both arms. She was unable to swallow at this stage and was reviewed by a Speech and Language therapist. A nasogastric tube was passed for feeding purposes and the patient was transferred to intensive care. Her IV methylprednisolone dose was then increased to 1 gram/d. Her deltoid muscle biopsy showed

frequent perifascicular degenerate/regenerating fibres, occasional necrotic fibres, and mild focal perivascular chronic inflammation. There was also MCH Class 1 sarcolemmal upregulation. These findings were consistent with an immune-mediated myopathy, likely DM. ANA, dsDNA, anti-Ro, anti-La and anti-centromere antibodies were all negative. The myositis antibody panel revealed a positive anti-NXP-2 antibody and was negative for anti-MI2 ALHA, anti-MI2 BETA, anti-MI2 TIF1 GAMMA, anti-MDA5, anti-SAE, anti-KU ABS, anti-PM 100 AB, anti-PM 75 AB, anti-JO1 AB, anti-SRP AB, anti-PL7 AB, anti-PL12 AB, anti-EJ AB, anti-OJ AB, and MYOSITIS MARKER AB.

So, a final diagnosis of Severe Idiopathic NXP-2 positive Dermatomyositis was made. The patient's clinical condition improved significantly after receiving 3 doses of IV methylprednisolone 1 gm. Her medication was then switched to oral prednisolone 60 mg once daily and she was discharged on a tapering dose of 10mg/week after the completion of 4 weeks of the 60 milligram per day dose. Currently, the patient is being followed –up in the Rheumatology out-patient clinic and is undergoing age appropriate cancer screening.

Discussion

Dermatomyositis is a rare systemic disorder that can pose diagnostic challenges early in the disease course [3]. There is no current criteria available to classify disease severity, however it has been observed in recent trials that myositis specific autoantibodies can be linked to various clinical presentations and associated outcomes [6]. Hereby, we present the case of NXP-2 antibody positive DM associated with severe weakness, dysphagia and significant limb oedema.

Anti-NXP-2 was first described in 1997. NXP2 is a 140-kDa protein that comprises RNA- and nuclear matrix-binding domains. It plays a role in RNA metabolism, maintenance of nuclear architecture and induction of cellular senescence [7]. Anti NXP-2 antibody is known to be commonly associated with juvenile DM, with severe muscular weakness and other features [4]. It is also known to cause cutaneous calcinosis and joint contractures in the juvenile population. In the adult population, NXP-2 positivity has a strong association with malignancy [5].

Hereby, we present this case of a 22 year old female with NXP-2 positive DM with features of severity to further expand the existing body of evidence that may point towards NXP-2 autoantibody positivity being a rare, but clinically important phenotype of dermatomyositis. It is our opinion that it is imperative to share clinical experience of such a severe disease so that all clinicians can have a better understanding of this entity and potentially in the future, it may lead to targeted treatments for this disease. Looking to the future, we need to gather further information on these cases so that we may be able to categorize them accurately.

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