ISSN (print): 2589-9120
ISSN (online): 2589-9139

International Journal of Medical Science and Applied Research (IJMSAR)

Available Online at: https://www.ijmsar.com

Volume – 3, Issue –5, September – October - 2020, Page No. : 65 – 70

Nxp2 Antibody Dermatomyositis In Children: Case Report Of A Young Girl

[1]Dr. Akhrif Mariam, Children's Hospital of Rabat, CHIS. Faculty of Medicine and Pharmacy of Rabat. MOROCCO

[2]Dr. N. Ahjiej, Department of Pediatric, Mohamed V Military Training Hospital, Rabat, Morocco.

[3]Dr. S. Saghir, Department of Pediatric, Mohamed V Military Training Hospital, Rabat, Morocco.

[4]Dr. M. Kmari, Department of Pediatric, Mohamed V Military Training Hospital, Rabat, Morocco.

[5]Dr. A. Ourrai, Department of Pediatric, Mohamed V Military Training Hospital, Rabat, Morocco.

[6]Dr. A. Hassani, Department of Pediatric, Mohamed V Military Training Hospital, Rabat, Morocco.

[7]Dr. R. Abilkassem, Department of Pediatric, Mohamed V Military Training Hospital, Rabat, Morocco.

[8]Dr. A. Agadr, Department of Pediatric, Mohamed V Military Training Hospital, Rabat, Morocco.

Corresponding Author: Dr. Akhrif Mariam, Children's Hospital of Rabat, CHIS. Faculty of Medicine and Pharmacy of Rabat. MOROCCO

Type of Publication: A Case Report

Conflicts of Interest: Nil

Abstract

Dermatomyositis (DM) is an idiopathic inflammatory myopathy (IIM) characterized by distinct skin lesions and a clinically heterogeneous constellation of systemic manifestations. In the absence of characteristic dermatologic findings or myopathy, DM can be difficult to diagnose. In addition, historical approaches to the diagnosis of DM have adopted the use of "overlap" syndromes to account for clinical heterogeneity, making diagnosis even more difficult. Juvenile dermatomyositis remains a rare condition but the most frequent of chronic inflammatory myopathies in children. It is an affetion characterized by the constancy of characteristic skin involvement which is very often associated with proximal muscle involvement although not obligatory. Its impact on the state of health and on the quality of life remains considerable despite the systemic corticosteroid therapy and immunosuppressant's, which have significantly improved the prognosis because before the advent of corticosteroid therapy, the course of the disease was bleak, but despite this better prognosis, it remains hampered by the particularly harmful side effects of long-term treatment. Given the very heterogeneous expression of this condition, autoantibodies specifically associated with DM appear to define homogeneous subgroups. Among them, the anti-NXP2 antibody is known for its high prevalence in juvenile DM (25%). The objective of this work is to underline the clinical and paraclinical characteristics of this condition as well as the appropriate therapeutic strategy.

Materials and Methods

We report the case of a 14-year-old girl with no pathological history, presented 3 months before hospitalization with skin lesions of the face and trunk, associated with muscular fatigue involving the shoulder girdle, admitted to a tetraparesis table without sphincter disorders or associated sensitives.

Key words: Juvenile dermatomyositis; Dermatomyositis; Myositis; Myositis, anti NXP2 antibodies

Introduction

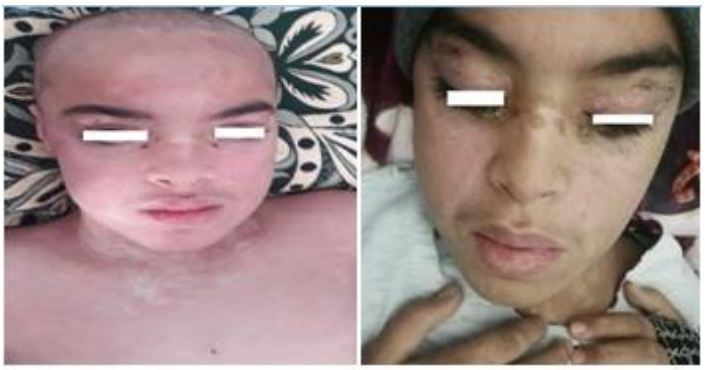
Juvenile dermatomyositis is a rare infantile autoimmune disease characterized by inflammation of small vessels in the skin, muscles, and major organs. The pronosis is

variable, varying from a monocyclic disease to a chronic disease extending into adulthood with serious complications. [1]. There is increasing emphasis on early and aggressive treatment to improve long-term outcomes, but response to medicinal treatments is unpredictable and many medecines have side effects. For a "treat-to-target" approach, it is important to understand the pathogenesis of the underlying disease. It increasingly seems that genotype, the state of autoantibodies and muscle histology may provide biomarkers to guide personalized treatments. [2].

Case report

It's a case of a 14-year-old female child with no significant pathological history, whose symptoms began 3 months before hospitalization with the appearance of erythema and puffiness of the face which were masked by taking corticosteroid therapy (prednisone 20mg 1cp and a half per day for 3 months) associated with muscular fatigue affecting the shoulder girdle with difficulty in raising the arms followed by difficulty in getting up and walking, thus producing a tetraparesis without associated sphincter or sensory disorders.

The evolution was marked by deterioration in general condition and weight loss estimated at 20kg during this 3-month period. Clinical examination revealed flaccid tetraparesis with abolition of osteotendinous reflexes, dysphonia, and predominantly crusty depigmentation patches in the trunk and scarring alopecia of the scalp. However, the patient did not show any clinically detectable sign of digestive, pulmonary or cardiac involvement. At the paraclinical level, we note an inflammatory syndrome revealed by the sedimentation rate at 15 and ferritinemia at 1089, muscle enzymes returning to normal, protein electrophoresis objectified a profile compatible with a moderate inflammatory syndrome, muscle biopsy showed a slight inflammatory myositis on the other hand the skin biopsy was normal, besides the electromyogram was normal too . Chest x-ray, abdominal ultrasound, trans-thoracic ultrasound and spinal cord MRI without special features. The immunoassay detected positivity for anti-NXP2 antibodies. Consequently, the diagnosis of anti NXP2 Ab dermatomyositis was retained. The initial treatment comprised 3 boluses of solumedrol IV (1g / 1.73m2) followed by oral corticosteroid therapy (1mg / Kg / day) associated with methotrexate (20mg SC per week). A decrease in corticosteroid therapy is expected with the continuation of the disease-modifying treatment with methotrexate



The characteristic skin lesions of dermatomyositis present in our patient

Discussion

Juvenile dermatomyositis is a rare and severe autoimmune disease of childhood, which is characterized by systemic vasculopathy of small vessels, which usually affects the skin and muscles, but can also affect the joints, intestine, lungs, heart and other internal organs. Juvenile dermatomyositis is the most common idiopathic inflammatory myopathy (IBD) of childhood. There is growing evidence that early and energetic management of DMJ improves outcomes, while conversely, a long duration of untreated disease is associated with longer time to remission and rates of complications higher, such as ongoing skin disease or osteoporosis [1,2]. Therefore, recognition, prompt referral to specialist care and early treatment of patients with juvenile dermatomyositis is of utmost importance.

Diagnosis

Juvenile dermatomyositis encompasses many very heterogeneous conditions. The reason for this heterogeneity is being investigated and it becomes possible to identify homogeneous groups of patients on the basis of antibodies (Ab) specifically associated with Juvenile dermatomyositis. The only clinical form defined by consensus is "amyopathic dermatomyositis" [3], characterized by a purely cutaneous involvement. There are purely muscular forms called "DM sine dermatitis" whose diagnosis is based on a muscle biopsy and the presence of specific Abs. [4].

Clinical evaluation

Clinical examination is essential to guide the diagnosis and identify signs of muscle damage (severity of the deficit and swallowing disorders) and extramuscular (respiratory or digestive). The skin (especially the hands, face, elbows, knees and back) and oral mucosa should be examined carefully: the dermatological manifestations, which are the key elements of the diagnosis, can be subtle. Growth in height and weight as well as puberty stage should be systematically assessed. The patient's medical history should be taken in order to establish a family tree that can detail personal history and / or family history of inflammatory myositis, autoimmune diseases, and hereditary myopathies; determine if any medication has been ingested in the weeks before the first signs; determine how the muscle damage appeared (the attack is sub acute for a few weeks or months, unlike myopathy, in which the muscle involvement develops much more slowly and is insidious); search all functional signs to guide the diagnosis and identify signs of severity; assess growth and stages of psychomotor development. Muscle involvement usually progresses within a few weeks, with varying severity. Muscular involvement is bilateral, proximal, and symmetrical: it predominates in the scapular and pelvic regions; in the case of our patient, she had muscle fatigability involving the shoulder girdle with difficulty in raising the arms followed by difficulty in getting up and walking, thus producing a picture of tetraparesis without associated sphincter or sensory disorders, without respiratory disorder or swallowing however she had a nasal voice ; but can also affect the axial muscles, especially in children [4]. Muscle involvement should be assessed using the standardized Childhood Myositis Assessment Scale (CMAS) method, which assesses muscle endurance, and Manual Muscle Testing (MMT), which assesses muscle strength. The evaluation should include a search for signs of severity:

- The effect on swallowing: search for signs suggestive of dysphagia (cough during feeding, modification of the voice (nasal), examine the contraction of the palate;
- Respiratory failure;
- Cardiac involvement;
- Digestive disorders (abdominal pain, pseudo-occlusion or gastrointestinal bleeding).). [4].

Skin signs are characteristic of DMJ. They can be of varying intensity, sometimes very inconspicuous, and should be carefully looked for when there are signs of muscle deficit. The characteristic skin lesions are:

- Involvement of the face: erythematous or even purplish coloring of the upper eyelids, frequent erythema of the cheekbones, sometimes of the forehead and temples, with a purplish-red heliotropic appearance; our patient presented the same lesions.
- Hand involvement: Gottron papules;
- Macular lesions, erythematous squamous symmetrical violin-shaped and / or confluent on the tops of the extensions of the metacarpophalangeal and / or interphalangeal joints; for our patient, she reported palpebral and hand edema masked by taking corticosteroid therapy during admission

- Peri-nail telangiectasias, sometimes visible to the naked eye, and gingival telangiectasias.
- There are many other non-specific skin signs: skin and / or mucous membrane ulcerations, digital necrosis, Raynaud's phenomenon, edematous forms of poikiloderma, lip dystrophy, erythroderma, follicular hyperkeratosis, panniculitis, alopecia, vasculitis, our patient had alopecia.
- Mechanic's hands, photosensitivity, and lichenoid or even necrotic erythema. Calcinosis is rare in the initial stage. In some forms, cutaneous involvement is absent or atypical. Therefore, a muscle biopsy is necessary to confirm the diagnosis in these forms,
- Other signs may be present :
- Involvement of the joints in 25 to 50% of cases;
- Cardiac involvement: subclinical cardiac manifestations are present in half of the cases, although clinical failure is rare [5]
- Pulmonary involvement and / or respiratory muscle deficiency: it is very important to test respiratory involvement, because it is a major cause of mortality. The impairment may be due to diaphragmatic and / or intercostal involvement (restrictive ventilatory disorder), inflammation of the parenchymal tissue, or inhalation pneumonia due to difficulty in swallowing. There may be subclinical abnormalities in the functional respiratory test [6];
- Early or late digestive disorders, sometimes very severe [7].
- The fever is usually absent or moderate. A deterioration in the general condition with weight loss or asthenia can be a complication of swallowing disorders or a sign of loss of muscle mass; our patient had very significant weight loss with asthenia;

Paraclinical exploration

paraclinical examinations necessary to establish the diagnoses are:

Determination of muscle enzyme levels:

The elevation of at least one muscle enzyme among CK, the ASAT, the LDH or aldolase is seen in about 90% of patients. in practice, it is recommended to test myself that the Ck. the absence of an elevation of a muscle enzyme does not rule out the diagnosis. [4]. this is the case with our patient who had a normal CK level after taking corticosteroid therapy for 3 months.

Muscle biopsy: this is not routinely performed for typical cases but is essential for atypical cases. it can help confirm the diagnosis (for example, in cases without skin involvement or atypical). four types of lesions can be observed [7]:

- ❖ Inflammatory involvement: mainly perivascular and perivascular inflammation composed of B and T lymphocytes, CD4 + cells, some of which are believed to secrete interferon (INF), and macrophages, in our case, muscle biopsy showed slight inflammatory myositis.
- ❖ Vascular involvement: capillary loss associated with early capillary deposition of complement membranolytic attack complex (MAC) C5b-9, thickening of the endothelium of arterioles.
- ❖ Muscle involvement: necrosis / regeneration, perifascicular atrophy, micro-infractus, myofibrillar loss, re-expression of HLA class I molecules with perifascicular reinforcement.
- ❖ Endo- and peri-mysial fibrosis. a score enabling these lesions to be quantified has been validated in children [4];

skin biopsy: this should be easily performed if the diagnosis remains uncertain. in most cases, it presents

an appearance similar to that of lupus with generally inconspicuous interface dermatitis with basal vacuolation, rare apoptotic keratinocytes and lymphocytic infiltration of the superficial dermis; in our patient she returned to normal.

The electromyogram is not useful for typical cases [4], in our patient the EMG was without abnormality, this result was falsified by taking long-term corticosteroid therapy.

The electromyogram is not useful for typical cases [4], in our patient the EMG was without abnormality, this result was falsified by taking long-term corticosteroid therapy.

The electromyogram is not useful for typical cases [4], in our patient the EMG was without abnormality, this result was falsified by taking long-term corticosteroid therapy.

Magnetic resonance imagery of the muscle: It may be useful to confirm the involvement of the muscle in atypical cases. it is also a diagnostic criterion according to the ENMC. T2 STIR (short tau inversion recovery) sequences are used to detect muscle edema, which is an indirect and non-specific indication of inflammation. the sensitivity of MRI for diagnosing myositis ranges from 70 to 100%, depending on the study. however, normal MRI does not exclude muscle involvement. the usefulness of MRI to guide the biopsy to improve its accuracy is questionable [4]

Capillaroscopy is not necessary for diagnosis, but the presence of significant abnormalities may indicate JDD [8];

The electromyogram is not useful for typical cases [4], in our patient the EMG was without abnormality, this result was falsified by taking long-term corticosteroid therapy.

The paraclinical examinations necessary to classify the DMJ are: [4].

- autoantibody testing:

- Myositis-specific antibodies (MSA) not observed in other autoimmune diseases: anti-Mi2, anti-SAE, anti-TIF1-gamma, anti-NXP2, or anti-MDA5, in our patient the anti- NXP2 came back positive.
- Auto-anticops associated with myositis (MAA) which may be present in other autoimmune diseases: non-specific anti-nucleus, anti-U1RNP, anti-RO52, anti-RO60, anti-SSB, anti-Ku, or anti-PM / Sel.

The paraclinical examinations which are necessary to study the camplications and / or the specific disorders are: [4].

- Blood count (CBC), (ASAT), (ALAT), gamma-GT, creatinine, albuminuria in edema (capillary leak), blood sugar, triglyceridemia, cholesterol, and bone densitometry;
- Respiratory function tests with a CO diffusion study (DLCO) to look for pulmonary involvement; a lung scan should always be done if there is a clinical or radiological abnormality or an abnormality identified by a respiratory function test. in other cases, a chest x-ray should be done.
- Electrocardiographic examination.

All of these examinations were normal in our patient.

Therapeutic Strategies

There are still no pharmacologic therapies specifically approved for JDD. the first-line treatment recommended when diagnosing JDD consists of high doses of corticosteroids (oral or intravenous) and methotrexate (European recommendations [9]). this recommendation was supported by a randomized trial which concluded that the combination therapy with prednisone and ciclosporin or methotrexate was more effective than prednisone alone; this is the treatment prescribed for our patient and the

outcome was favorable given the reduction in the inflammatory syndrome (ferritin at 86 compared to 1089). in the event of resistance to the treatment in a newly diagnosed patient, intensification of therapy should be considered every 12 weeks, after consultation with a specialized center (European recommendations) [4]. in the event of intolerance to methotrexate, another treatment should be prescribed (European recommendation). the decision to introduce second-line treatment should be discussed with the competent centers. the French working group recommends that mycophenolate be considered as a second-line approach (4). Intravenous immunoglobulins may be offered as adjuvant treatment in the event of dependence or resistance to corticosteroids, in particular for persistent skin conditions (European recommendations).

References

1. Mathiesen P, Hegaard H, Herlin T, Zak M, Pedersen FK, Nielsen S. Long-term outcome in patients with juvenile dermatomyositis: a cross-sectional follow-up study. Scand J Rheumatol 2012 Feb;41(1):50e8. PubMed PMID: 22044089. Epub 2011/11/01. eng.
2. Q. Wu et al. Juvenile dermatomyositis: Latest advances. Best Pract Res Clin Rheumatol. 2017 Aug; 31(4):535-557. doi :10.1016/j.berh.2017.12.003. Epub 2018 Jan 10
3. Gerami P, Walling HW, Lewis J, et al. A systematic review of juvenile-onset clinically amyopathic dermatomyositis. Br J Dermatol 2007;157:637–44
4. B. Bader-Meunier et al. French expert opinion for the management of juvenile dermatomyositis / Archives de Pédiatrie 26 (2019) 120–125
5. Schwartz T, Sanner H, Gjesdal O, et al. In juvenile dermatomyositis, cardiac systolic dysfunction is present after long-term follow-up and is predicted by sustained early skin activity. Ann Rheum Dis 2014; 73:1805–10.
6. Pouessel G, Deschildre A, Le Bourgeois M, et al. The lung is involved in juvenile dermatomyositis. Pediatr Pulmonol 2013;48:1016–25.
7. Mamyrova G, Kleiner DE, James-Newton L, et al. Late-onset gastrointestinal pain in juvenile dermatomyositis as a manifestation of ischemic ulceration fromchronic endarteropathy. Arthritis Rheum 2007;57:881–4.
8. Mugii N, Hasegawa M, Matsushita T, et al. Association between nail-fold capillary findings and disease activity in dermatomyositis. Rheumatology (Oxford) 2011;50:1091–8.
9. Bellutti Enders F, Bader-Meunier B, Baildam E, et al. Evidence based recommandations for diagnosis and treatment of juvenile dermatomyositis. Ann Rheum Dis 2017;76:329–40.