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RESEARCH ARTICLE

FROM DERMATOMYOSITIS SINE DERMATITIS TO A COMPLETE FORM OF JUVENILE DERMATOMYOSITIS: AN INTRIGUING CASE REPORT!

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Abstract

Although rare, juveniledermatomyositis (JD) is the mostcommoninflammatorymyopathy in children. It is a vasculopathycategorizedintoclassical and non-classicalsubtypes, the latter includingamyopathicdermatomyositis (DM) and dermatomyositis (DM) withoutdermatitis. DM sine dermatitisis a rarelydescribedphenotype, representing an atypicalformwith no classiccutaneous manifestations, complicatingdiagnosis, and isthereforeidentified on the basis of well-defined diagnostic criteria.In this article, we report the case of a twelve-year-oldchildpresentingwith muscle weakness and myalgias one monthprior to admission. Clinicalexaminationrevealed a progressive myogenic syndrome withdysphagia to solids, with no other multi-visceralinvolvement or signs of severity. Mucocutaneousexaminationwaspoor, revealingonlyleukokeratosis of the leftlateralborder of the tongue, with no otherassociated skin signs. Muscle enzymes wereelevated. A myositis-specificautoantibodyassayrevealed a positive anti-NXP2. Electromyographyshowed a diffuse emyogenic syndrome. MRI of the lowerlimbsshowed diffuse hypersignal in the muscle tissue. A diagnosis of dermatomyositis sine dermatitiswas made, and treatmentwasinitiatedwith oral prednisone 2 mg/kg/day, combinedwithsubcutaneous Methotrexate 25 mg/week. In the absenceimprovementafter 5 months of treatment, weswitched to mycophenolatemofetil (MMF) at a dose of 600 mg/m² twicedaily, with good progression.Afterlosinghissight and stoppingtreatment for a long time, the patient returnedaftertwoyearswith a full clinicalpicture of juveniledermatomyositis, withsevere muscle weakness and skin signs. This time, given the severity of the disease, ourtherapeuticapproachwas to switch to third-line treatmentwithinfliximab 5 mg/kg biotherapyadministered at specificintervals, with good improvement.

Introduction:-

Juvenile dermatomyositis (JD) is a rare and heterogeneous autoimmune myopathy affecting children under the age of seventeen. Its annual incidence is estimated at between 2 and 4 cases per million **[1,2,3].** It is a chronic idiopathic

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inflammatory micro-angiopathy affecting mainly striated muscles and skin, and its ethiopathogenesis remains obscure, although the role of immunity and environmental factors on genetically predisposed patients is recognized **[2,3].** DMJ generally manifests itself as a neurological clinical picture characterized by a progressive motor deficit, first proximal then distal, with or without typical skin involvement. Signs of severity may appear when the vasculopathy involves other viscera **[3,4,5].** However, atypical cases without cutaneous manifestations exist, making diagnosis more difficult **[4].** This article presents a rare case of DMJ in a child beginning with pure muscle involvement without cutaneous manifestations, shedding light on the diagnostic course, the treatment administered and its unexpected evolution. Our aim is to inform clinicians about this atypical form of DMJ in order to improve recognition and management of this rare disease.

Case-Report:

A twelve-year-oldchildwith a history of nephroblastoma in his cousin, no previousdrug or toxicintake, no knownpersonal or familyautoimmunepathology, no recent viral infection, no myasthenia gravis or familyneuromuscular pathologies, admitted for management of muscularweaknesswithmyalgiasdating back one monthprior to admission, evolving in a context of asthenia. Clinicalexaminationrevealed a consciousbedriddenchildpresentingwith a myogenic syndrome with progressive bilateral proximal then distal muscle deficit, rated at 3/5ths in the upperlimbs and 4/5ths in the lowerlimbs, osteotendinous reflexes present and symmetrical. A positive stoolsign, with no ocularmuscularinvolvement. The patient presentsdysphagia to solids. No respiratorysigns or othersigns of severity. Mucocutaneousexaminationwaspoor, apartfromleukokeratosis of the leftlateralborder of the tongue**(Figure 1),** no Gottron papules **(Figure 2)**, no pseudo-lupuserythema of the face, no purplisherythema of the eyelids, no periungualtelangiectasias.

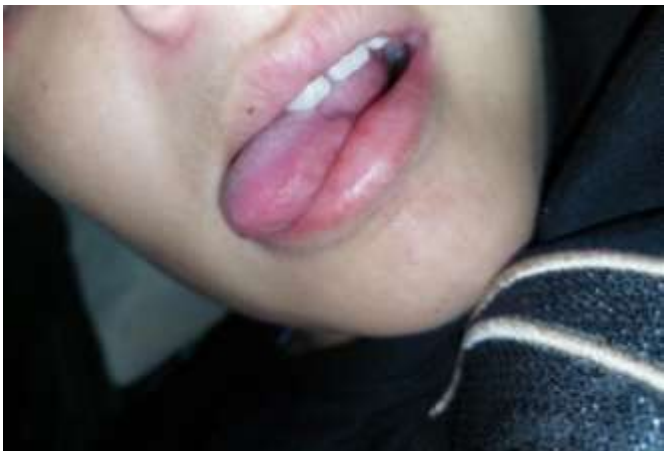

Figure 1:Leukokeratosis of the leftlateral border of the tongue.

Figure 2: Absence of Gottron papules.

Biologically, muscle enzymes showedelevated CPK at 3449, LDH at 912, SGOT at 151.A specificauto-antibodyassayshowed positive anti-NXP2**(Table1).**

Laboratoryparameters	**Initial values**	**Reference ranges**
Hemoglobin(g/dl)	12	13 -15,5
White bloodcell (/ul)	6310	4500 -13500
Platelets (/ul)	273000	150 000 - 450 000
C-reactiveprotein (mg/dl)	20	<10
Erythrocyte sedimentation rate (mm/hr)	25	0-10
Creatinephosphokinase (U/l)	3449	150-499
Lactate dehydrogenase (UI/l)	912	120-300
Serumglutamic-oxaloacetictransaminase(UI/l)	151	10-40
Serumglutamic-pyruvic transaminase (UI/l)	60	7-56
Complement component3 (mg/dl)	1.41	90-180
Complement component4 (mg/dl)	0.28	10-40
Rheumatoidfactor(IU/mL)	42	< 15
Myositis-associatedautoantibodie		
Anti-Mi2	Negatif	Negatif
Anti-MDA5	Negatif	Negatif
Anti-NXP2	Positif	Negatif
Anti- SAE	Negatif	Negatif
Anti-ku	Negatif	Negatif
Anti-TIF1 Gamma	Negatif	Negatif
Anti-Pm100	Negatif	Negatif
Anti-Pm75	Negatif	Negatif
Anti-Jo1	Negatif	Negatif
Anti-SRP	Negatif	Negatif
Anti-PL7	Negatif	Negatif
Anti-PL12	Negatif	Negatif
Anti-EJ	Negatif	Negatif
Anti-OJ	Negatif	Negatif

Table 1:- Table showingourpatient'svariousbiological tests.

Radiologically, an electromyogramshowed a diffusemyogenic syndrome (**Table 2).** A muscle MRI of the thighrevealed diffuse hypersignal in the muscle tissue**(Figure 3 and 4).**

Muscle	**Amplitude MUP**	**Duration MUP**	**Recruitment**
Right deltoid	Diminished	Greatlyreduced	Myogenic
Leftdeltoid	normal	normal	Normal
Right biceps brachii	Normal	normal	Normal
Right anterior tibial	Diminished	Greatlyreduced	Myogenic
Leftanterior tibial	Diminished	Greatlyreduced	Myogenic

Table 2:- Upper and lowerlimbelectromyograms.

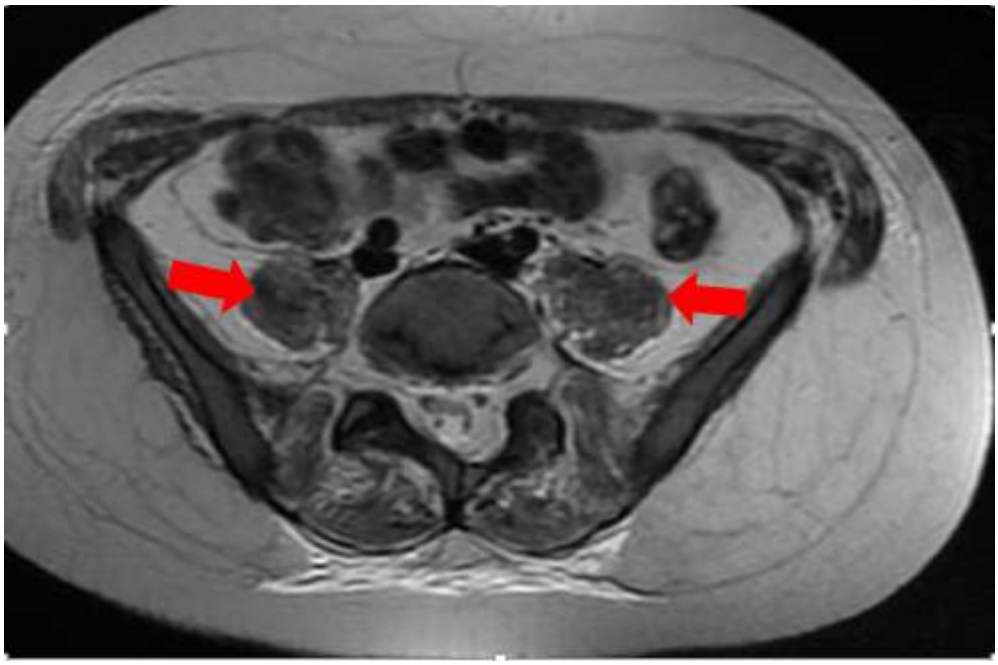

Figure 3:- Axial section of apelvic MRI T2 sequenceshowinghypethrophy of the psoas muscle.

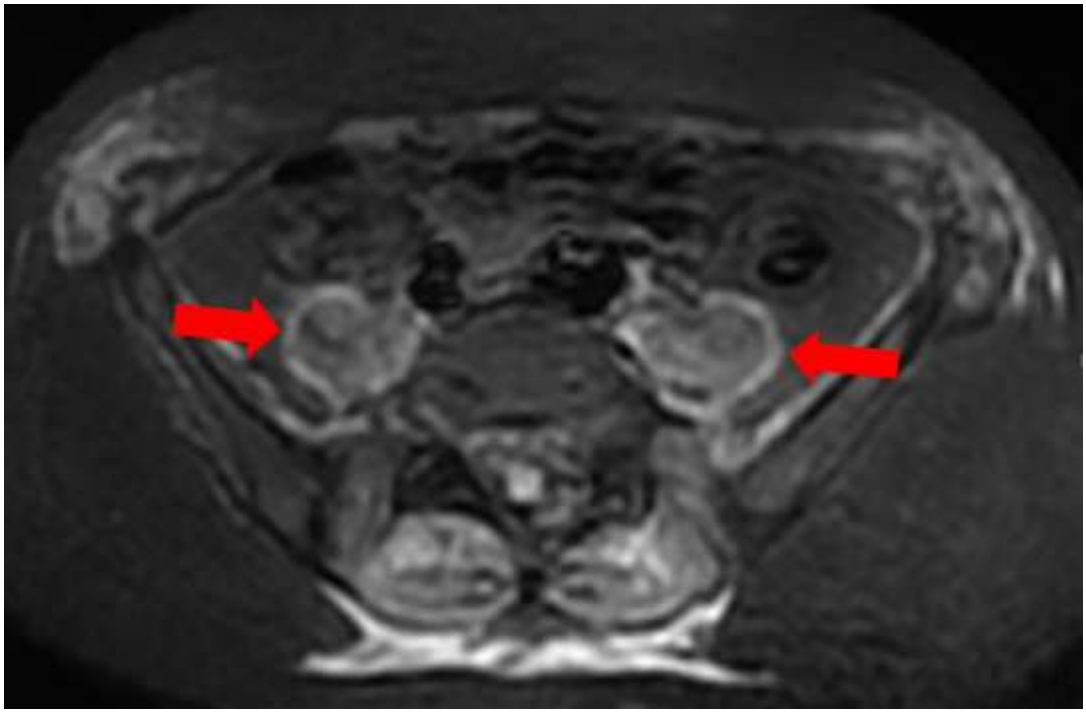

Figure 4:- Axial section of apelvic MRI STIR sequenceshowinghypersignal of the psoas muscle.

On the basis of theseelements, the diagnosis of dermatomyositis sine dermatitiswasaccepted, and the patient was put on prednisone: 02 mg/kg/dr in combinationwith Methotraxate 25 mg/weeksubcutaneously, withfolicacidsupplementation and physiotherapy sessions. In the absence of improvementafter 5 months of first-line treatment, weswitched to mycophenolatemofetil (MMF) at a dose of 600 mg/m² twicedaily, with good progression.

Subsequently, our patient waslost to follow-up, stoppedtreatment and returnedaftertwoyears of relapsing-remitting course with a full clinicalpicture of juveniledermatomyositis, consisting of severe muscle weaknesswithonset of cutaneoussigns**(Figure 5,6) :**

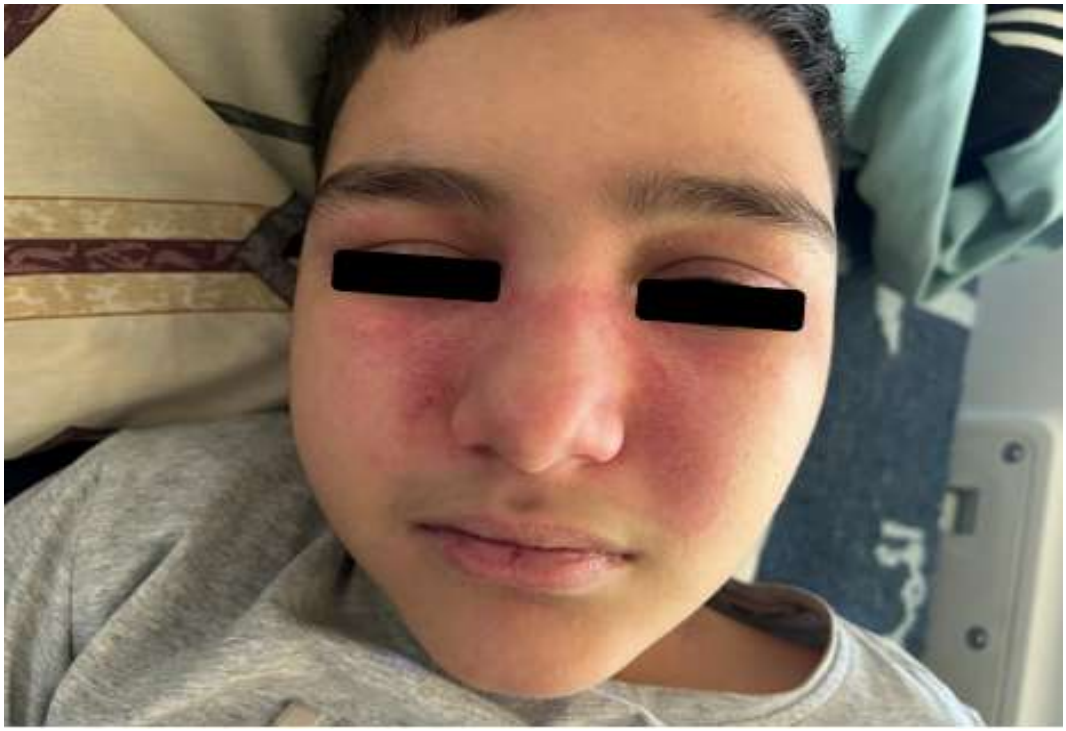

Figure 5:- Image showing installation of heliotropeerythema and pseudo-lupusmask of the face.

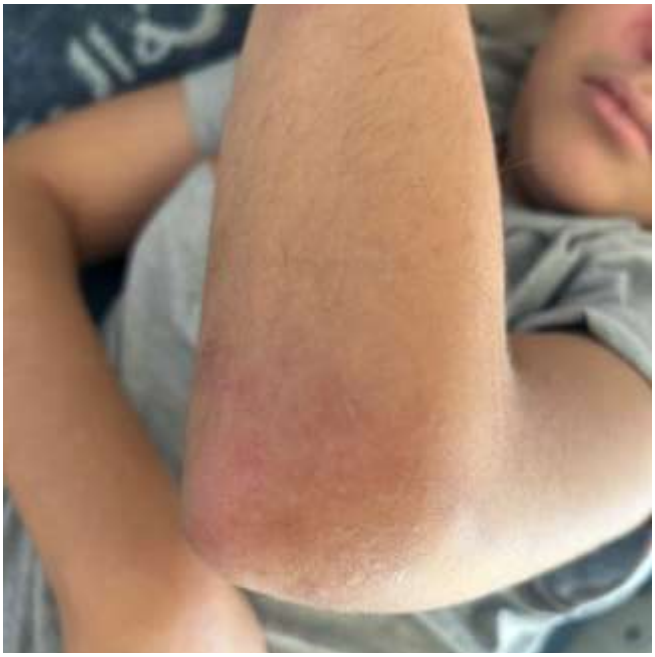

Figure 6:- Gottron papules.

This time, given the severity of the disease, our therapeutic approach was to switch to third-line treatment with infliximab 5 mg/kg biotherapy administered at specific intervals with good improvement.

Discussion:-

Juvenile dermatomyositis (JDM) is a form of myositis that refers to conditions associating muscle deficit with an inflammatory infiltrate of striated muscle. Genetic susceptibility is now indisputable, as predisposing HLA systems have been identified**[5].** This condition affecting children generally appears around the age of 7, with an earlier onset in girls **[1,4,8].** Juvenile dermatomyositis (JDM) without cutaneous signs, also known as dermatomyositis sine dermatitis, represents a rare subtype and atypical form of classic inflammatory myopathy in children **[4,7],** often under-recognized and infrequently described, with an estimated prevalence of 15% **[4].** Unlike DMJ , this variant reveals a purely muscular component, making it particularly difficult to diagnose**[7]** .

The onset of this condition is generally acute or sub-acute, preceded in 64% of cases by an infectious episode, and accompanied by an altered general state of asthenia and anorexia**[1]** .As in the case of our patient, whose clinical symptoms appeared after one month of evolution with a marked alteration in general state.

The European Neuromuscular Center (ENMC) has made it possible to diagnose dermatomyositis sine dermatitis by including all the clinical criteria of classic dermatomyositis, with the exception of the skin rash **[4].**

Historically speaking, the diagnostic criteria for DMJ were established by Bohan and Peter in 1975 **[2,3,8]** , enabling a definitive diagnosis to be made. The diagnosis is definite when four out of five criteria are met, whereas it is considered probable if only three are present. These criteria correspond to the combination of a characteristic cutaneous rash consisting of Gottron's papules, purplish erythema of the eyelids, pseudolupus erythema of the face, periungual erythema and photosensitivity**[1 ,3]** , as well as at least three of the four features indicating muscular involvement. Even in the absence of typical cutaneous manifestations, considered pathognomonic signs of DM, it is crucial not to rule out the possibility of DM sine dermatitis**[4].** And as the cutaneous component was absent in our patient, the other criteria for retaining the diganostic of dermatomyositis without cutaneous signs were represented by: symmetrical and progressive muscular weakness, proximal then distal, predominantly in the girdle and cervical muscles, often accompanied by myalgias. In 30% of cases, the deficit may involve the pharyngeal and oesophageal musculature, and be responsible for dysphagia and swallowing disorders. Our patient presented the same clinical picture of muscle deficit and myalgia, with dysphagia to solids.In 4 to 8% of cases, it also affects the respiratory muscles, causing pneumopathy**[1,7] .** Complications may also affect the heart, causing conduction disorders, and may lead to ophthalmoplegia, ptosis, chemosis or exophthalmos. In the brain, it can lead to bulbar damage, seizures or aphasia, with a high risk of death. However, renal involvement is rarely found **[1].**

The second criterion is biological, corresponding to an elevation in serum muscle enzymes, the most commonly used paraclinical criterion in DMJ. Creatine phosphokinase (CPK) is a good marker for diagnosis and assessment of treatment efficacy. Other enzymes include lactate dehydrogenase (LDH), aminotransferases and aldolase. Myositis-specific autoantibodies (MSA), now frequently used, are found in 50% to 70% of patients with juvenile dermatomyositis, but may also be present in patients with other autoimmune diseases. They provide valuable additional information on response to treatment, underlying mechanisms and prognosis [2], including anti-Mi-2, anti-TIF1, anti-NXP2 and anti-SRP **[5].**

The third criterion corresponds to abnormalities detected by electromyography, although this test is rarely used at present, but can show myogenic damage with short, polyphasic motor potentials, and fibrillation. Muscle MRI, on the other hand, can help with early diagnosis where clinical criteria alone are insufficient, by showing multifocal or diffuse hypersignals in muscle tissue, inflammation without fatty infiltration or subcutaneous calcifications and thus enables good detection of relapses and determination of remission **[1,3].** Our patient benefited from these two examinations, which pointed to the presence of dermatomyositis.

The final criterion is a muscle biopsy, which is the gold standard for diagnosing DMJ. It is not systematically performed in typical cases, to rule out polymyositis or another myopathy or connective tissue disease **[5].** It should be guided by EMG and/or MRI, although its use has diminished due to its invasive nature. It shows perifascicular atrophy with compensatory rarefaction and dilatation of capillaries **[2 ,5].**

We therefore concluded that our patient presented with an unusual form of dermatomyositis, dermatomyositis sine dermatitis, and he was put on oral corticosteroids: prednisone: 02 mg/kg/day in combination with subcutaneous methotraxate 25 mg/week with folic acid supplementation. In the absence of improvement, we switched to mycophenolate mofetil (MMF) at a dose of 600 mg/m² twice daily, with good progression. The treatment of

dermatomyositis sine dermatitis generally follows the same principles as the treatment of classic juvenile dermatomyositis, focusing mainly on managing muscle inflammation and preventing complications.The first-line treatment is oral corticosteroids with prednisone 1 to 2 mg/kg/day for 4 to 6 weeks, then reduced to 10% every 2 to 4 weeks, combined with subcutaneous or intravenous methotrexate 15 to 20 mg/m²/week, monitored monthly by blood count and liver function tests. Bolus corticosteroids are indicated in severe forms with risk of digestive perforation **[1,3]** . Corticosteroids should be withdrawn gradually to avoid relapses. Mycophenolate mofetil (MMF) is indicated as a second-line treatment at a dose of 40-60 µg/h. In refractory forms with severe dysphagia, especially in cases of cortico-dependence or cortico-resistance, IV immunoglobulins are used, with proven efficacy. The combination of corticosteroids/ciclosporin A or tacrolimus, which is indicated above all in cases of associated pulmonary damage, represents 3rd-line treatment. Biotherapy is beginning to have a place in the treatment of DMJ, based on infliximab (anti TNFx) and rituximab at a dose of 5 mg/kg administered at specific intervals. European recommendations suggest that immunosuppressive therapy should be discontinued once a remission of at least one year has been achieved, without recourse to corticosteroids**[2,5].**physiotherapy is regaining its place in DMJ, and is a fundamental pillar of management, particularly during the initial inflammatory phase, to prevent vicious postures**[3].** Concerningthe progression and long-term prospects of dermatomyositis sine dermatitis, there is a lack of documentation. Our clinical case showed a progression over two years, from an atypical presentation withoutcutaneous manifestations to a complete classical form of the disease.

Conclusion:-

Juvenile dermatomyositis is a rare, complex and chronic pathology requiring a specialized, multidisciplinary approach to its management. The absence of typical cutaneous manifestations does not rule out the possibility of DM sine dermatitis. The mortality rate in children has fallen since the 1960s, thanks to improved management.

Diagnostic criteria are currently being redefined, and are likely to be extended to other clinical manifestations and to sensitive, non-invasive tests such as MRI. The impact of juvenile dermatomyositis on health and quality of life remains considerable, despite systemic corticosteroid therapy and immunosuppressants, which have significantly improved prognosis.

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