International Journal Dental and Medical Sciences Research
Volume 4, Issue 1, Jan-Feb 2022 pp 576-579 www.ijdmsrjournal.com ISSN: 2582-6018

Juvenile dermatomyositis, a diagnostic challenge:Experience from a general hospital in Eastern India

DrAbhishek Kumar[1*], DrParthaSarathi Bal[2], Dr Alok kumar[3], Dr V K Singh[4]

1 Senior Specialist, Department of Pediatrics,CentralHospitalRamgarh, Jharkhand
2 Senior Specialist, Department of Medicine, Central Hospital Ramgarh, Jharkhand
3 Senior Specialist,Department of Pathology, Central Hospital Ramgarh, Jharkhand
4 CMS Incharge, Central Hospital Ramgarh, Jharkhand
Correspondenceto :Dr Abhishek Kumar, Senior specialist, Central Hospital Ramgarh, Jharkhand, India 892122.

Submitted: 01-02-2022 Revised: 11-02-2022 Accepted: 14-02-2022

ABSTRACT
Juvenile Dermatomyositis is a rare multisystemic autoimmune vasculopathy,characterised by pathognomic skin manifestations (heliotrope rash over eyelid and Gottron papule) and proximal muscle weakness.It can also involve any other organ system and can cause constitutional symptoms.Genetic factors and environmental triggers are likely to play important roles in aetiology, although the exact mechanism is largely elusive.There is a higher incidence of retrospective parent reported infective illnesses (mainly gastrointestinal and upper respiratory tract) in the months prior to onset of JDM. A 10 year old child exhibited rare initial presentation (loose stools,generalised edema,weight loss) before developing characteristic cutaneous manisfestations which caused delay in diagnosis and initiation of treatment.This case report highlights that it is a rare but life threatening condition that needs to be diagnosed early and treated aggressively for a fruitful outcome.

I. INTRODUCTION

JDM is a childhood form of myositis primarily affecting striated muscles and skin. Though most common manifestation of JDM is skin rash (often consisting of a heliotrope eyelid discoloration or gottron's papule) and proximal muscle weakness, other organ systems including gastrointestinal tract, lungs, heart, joints and nervous system can be involved.It differs from adult dermatomyositis in involving a greater degree of vascular inflammation and thrombosis . The pathologic feature of JDM is widespread capillary lesion in which endothelium swells and occludes the lumen and the intracellular structures are in disarray.[1] A genetic predisposition exists, but studies suggest an environmental infectious pathogen trigger, especially in children who report a history of gastrointestinal or respiratory infections in the previous three months e.g. coxsackie B virus or entero virus [2].

JDM may be difficult to diagnose because of variations in clinical presentation. Only 50% children present with rash as their initial symptom and 25% with weakness as their first symptom [3]. Therefore, high index of suspicion is required to diagnose it early and to initiate appropriate treatment as soon as possible to improve outcome.

A Case description

A 10 yr old female presented to us with complaints of loose stools and vomiting since last 3 months. She was born out of a non-consanguinous marriage belonging to lower middle socioeconomic status with uneventful antenatal, natal and post natal period, developmentally normal and immunized for age with normal premorbid diet and health.

Child had swelling all over body, progressive weight loss and generalized weakness for last 3 months . She had rashes over body since last 15 days. Child had two previous hospital admissions in last 1 month for complaints of loose stools and swelling over body.

Workupfornephrotic syndrome, tuberculosis, celiac disease and HIV was negative. Child was thin builtwith BMI of 11.6 kg/m2 (<-3 SD) with bilateral pitting pedal edemaand periorbitalpuffiness. Child had non-scarring alopecia and angular cheilitis withsmooth tongue. She alsohad erythematous papular rashes over posterior surface of neck and back and perifollicular

haemorrhage over buttocks and legs. In due course of time erythematous papular non-blanchable rashes appeared over bilateral elbows,bilateral knees and interphalangeal joints (Gottron's papule) (**Fig 1 &2)**. Systemic examination was normal except for proximal

International Journal Dental and Medical Sciences Research
Volume 4, Issue 1, Jan-Feb 2022 pp 576-579 www.ijdmsrjournal.com ISSN: 2582-6018

muscle weakness. Power in hip, shoulder, neck and truncal muscle groups was less in comparison to distal muscles of wrist, ankle and elbow. Child also had a waddling gait. Initial possibilities kept were: Juvenile dermatomyositis, celiac disease, SLE and HIV infection for which the child was investigated.Complete blood count, renal function test, liver function tests,serum electrolytes, CPK NAC, CPK MB, TTG, ANA profile, Vit B12 levels were normal.

Celiac disease and HIV was ruled out by above investigations and SLE was ruled out by ACR 1997 criteria. Further investigations were done to establish the diagnosis of JDM. Electromyography showed increased insertional activity, small polyphasic motor unit potentials and early recruitment suggestive of myositis. Muscle biopsy was not done however skin biopsy from lesion and histopathological examination showed upper dermal

edema and perivascular inflammation .MRI thigh was planned to look for edema however this facility was not available in hospital and child could not be shifted to appropriate centre for investigation due to her sick condition. As our patient had typical skin rash (gottron papule) plus two other findings, proximal muscle weakness and EMG findings suggestive of myositis, a diagnosis of probable JDM was made based on Bohan Peter diagnostic criteria [4].Intravenous methylprednisolone was given at a dose of30 mg/kg/day for three consecutive days followed by oral prednisolone at 2mg/kg/day.Afterfew days of treatment child showed symptomatic improvement. Pain abdomen and loosestools subsided, oedema and rashes decreased, oral acceptance improved.However, there wasno significant improvement in muscle power.Child was discharged on oralprednisolone with plan to add methotrexate on follow up. Child presented 3 days later inemergency with pain and mild tenderness in abdomen.X- ray abdomen was normal.Withintwo hours of arrival child developed profuse hematemesis, went into shock and couldn't berevived.

II. DISCUSSION

Though JDM is the most common idiopathic inflammatory myopathy, incidence is very lowapproximately3 cases per million children per year without racial predilection[5]. JDMvasculopathy principally affects muscle and skin but may involve joints, lung, heart and other internal organs. Constitutional symptoms like fever,fatigue,anorexia,malaise,weight loss may predate the classical rash and muscle weakness by months, delaying the diagnosis.

The five diagnostic criteria for Dermatomyositis established by Bohan and Peter in 1975 predates the use of MRI and have not been validated in children. Definite diagnosis requires – (1)Typical skin rash (gottron papule and heliotrope rash) and at least three of the following four criteria: (2) Proximal muscle weakness (3) Elevation of muscle enzymes (4) Abnormal Electromyography suggestive of inflammatory myopathy (5) Abnormal muscle biopsy suggestive of inflammatory myopathy. If only two of the above four are met diagnosis of probable JDM is made, as in the present case.In general, the first two criteria i.e. classic rash and proximal muscle weakness are almost always present. Criteria 3,4 and 5 provide additional support for diagnosis.Current practice reveals necessity of broadening diagnostic criteria by incorporating new techniques such as MRI and USG and the significance of skin disease in JDM. Diagnostic criteria are under revision and will need further adjustment as new outcome tools especially autoantibodies and biomarkers are being developed. Hence, at present diagnosis of JDM should not necessarily be excluded by failure to meet one or more of the criteria proposed by Bohan and Peter except that related to dermatitis. A European initiative called Single Hub and Access point for pediatric Rheumatology in Europe (SHARE), has formulated evidence based guidelines for diagnosis and treatment of JDM [13] It suggests a list of investigations in every suspected case of JDM which is available online.

Early and aggressive therapy prevents mortality and significant morbidity associated with disease complications like calcinosis.Treatment is largely based on the experience of treating pediatric rheumatologist. Mainstay of therapy is high dose corticosteroid initially in combination with disease modifying drugs like methotrexate and ciclosporin A . Only randomized controlled trial for newly diagnosed patients was perfomed by Pediatric Rheumatology INternational Trial Organisation (PRINTO) from 2006 to 2011 comparing three commonly used protocols prednisolone alone vs combination of prednisolone with either methotrexate or ciclosporin A [10]. Combination of steroid and methotrexate had the best outcome for efficacy and safety.Immediately after diagnosis high dose corticosteroid (preferably methylprednisolone pulse 15-30 mg/kg/dose for three consecutive days) followed by oral prednisolone 1-2 mg/kg /day should be started. Methotrexate at a dose of 15-20 mg/m^2 weekly

subcutaneously should be added. Steroids have to be slowly tapered over a period of 12-24 months after indicators of inflammation normalize and muscle strength improves. Stopping Methotrexate should be considered when disease is in remission for a minimum period of one year off steroids[13].Treatment of refractory cases is still a challenge, options that have been tried include intravenous immunoglobulin, cyclophosphamide, ciclosporin A, azathioprine, mycophenolate mofetil, hydroxychloroquine, tacrolimus, rituximab and autologous stem cell transplantation. Survival has improved considerably in JDM pts in developed world where mortality rates are now in the range of 1-3% [6,7]. However mortality continues to be high in developing countries. In one Indian study it was found to be 11.1% [8].Common causes of mortality include gastrointestinal vasculitis, bleeding and intestinal perforation,interstitial pneumonitis, myocarditis and aspiration pneumonia[8.9.11,12]

Major risk factors for poor prognosis includes delay in treatment or inadequate treatment .Other risk factors for poor prognosis include severe disease activity,cutaneous ulceration,extensive calcinosis,dysphagia,dysphonia,advanced nail fold capillary abnormality, a high serum creatine kinase, a non-inflammatory vasculopathy on muscle biopsy and presence of certain myositis specific antigens (MSA) such as Antisynthetase antibody, signal recognition particle (SRP) autoantibodies[6].

To conclude,early diagnosis and initiation of aggressive and adequate treatment as soon as possible is the key to survival in these patients. High index of suspicion is required to diagnose the cases early and diagnosis should not be refuted or treatment delayed even if few diagnostic criteria are not met.

REFERENCES

[1]. Ramanan AV, Feldman BM: Clinical feature and outcomes of Juvenile dermatomyositis and other childhood onset myositis syndromes. Rheumatic Disease Clinics of North America 2002 Nov;28 (4):833-857

[2]. Pachman L.M., Lipton R., Ramsay Goldman R, Shamiyeh E, Abott K, Mendez E & Borzy M. History of infection before the onset of Juvenile Dermatomyositis: results from the national institute of arthritis and musculoskeletal and skin diseases research registry. arthritis& rheumatism. 2005 Apr15; 53(2):166-172

[3]. Feldmen B M, Rider L G, Reed A M & Pachman L M. Juvenile Dermatomyositis and other idiopathic inflammatory myopathies of childhood. Lancet. 2008. Jun 28; 371 (9631):2201-2212

[4]. Bohan A & Peter JB. Polymyositis and Dermatomyositis (first of two parts). New England Journal of Medicine.1975 Feb13;292(7) :344-347

[5]. Robinson AB ,Reed AM. Juvenile Dermatomyositis.in: Kliegman RM , Stanton BF, St. Geme JW , Schor NF (eds) Nelson Textbook of Pediatrics, 20th ed. Elsevier Philadelphia 2016 p 1181-1185

[6]. Rider LG, Lindsey CB, Cassidy JT. Juvenile dermatomyositis. In: Cassidy JT, Petty RE, Laxer RM, Lindsey CB (eds). Textbook of pediatric rheumatology, 6th ed. WB Saunders, Philadelphia ; 2011. P. 375-413

[7]. Ravelli A, Trail L, Ferrari C, Ruperto N, Pistorio A, Pilkington C et al .Long term outcome and prognostic factors of juvenile dermatomyositis: a multinational, multicentric study of 490 patients. Arthritis Care Res.(Hoboken) 2010 Jan15; 62 (1): 63–72

[8]. Singh S, Suri D, AulakhR,Gupta A, Rawat A, Kumar R M. Mortality in children with Juvenile Dermatomyositis: Two decades experience from a single tertiary care centre in North India . Clin Rheumatology .2014 Nov ; 33(11):1675-1679

[9]. Chowdhary V, Wakhlu A, AgarwalA,Misra R. Outcome in juvenile dermatomyositis. Indian Pediatr . 2002; 39 (11) 931–935

[10]. Ruperto N, Pistorio A, Oliveira S, ZulianF, Cutting R, Ravelli A,Fischbach M et al Prednisone versus predisone plus ciclosporin versus prednisone plus methotrexate in new onset juvenile dermatomyositis: a randomized trial. Lancet 2016 Feb 13;387(10019):671-78

[11]. Spencer CH, Hanson V, Singsen BH, Bernstein BH,Kornreich HK,King KK (1984) Course of treated juvenile dermatomyositis. J Pediatr 1984 Sept; 105(3):399–408

[12]. Ponyi A, Constantin T, Balogh Z, Szalai Z, Borgulya G, Molnar K (et al). Disease course, frequency of relapses and survival of 73 patients with juvenile or adult dermatomyositis. Clin Exp Rheumatol. 2005 Jan-Feb; 23 (1) 50–56

[13]. Enders F B, Bader-Meunier B, Baildam E, Constantin T, Dolezalova P, Feldman B M et al. Consensus based recommendations for

the management of Juvenile Dermatomyositis: Ann Rheum Dis. 2017 Feb;76 (2)329-340

Fig 1 Bilateral erythematous patch over the knees

Fig 2 Erythematous rashes over metacrpophalangeal and interphalangeal joints