

## Juvenile dermatomyositis with rare cutaneous manifestation: generalised hypertrichosis: A case report

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### Abstract

Juvenile dermatomyositis (JDM) is a rare autoimmune disease characterised by inflammation of muscles and skin with extra muscular involvement of joints, heart, intestine, and liver. Pathogenesis of JDM is believed to be due to vasculopathy. Along with classic cutaneous features of JDM, rare findings include hypertrichosis, lipoatrophy, photosensitivity, bullous lesions, and hyperhidrosis. We present, here, a case of JDM with hypertrichosis as very few cases have been reported previously.

**Keywords:** Juvenile dermatomyositis, Hypertrichosis, Autoimmune disease.

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### Introduction

Juvenile dermatomyositis (JDM) is a rare, childhood, inflammatory myopathy presenting with proximal muscle weakness and cutaneous manifestation, having a range of skin rashes, ulcers, and pigmental and hair problems.<sup>1</sup> The approximate prevalence of JDM is 2 to 4 per 1,000,000 children per annum in the United States.<sup>2</sup> Moreover, hair issues related to JDM can vary from hair loss to hypertrichosis. Hypertrichosis, is a disorder of hair growth characterised by excessive hair and if involving the whole body, termed as generalised hypertrichosis.<sup>3</sup> Keeping in mind the rarity of such a co-finding,<sup>4</sup> this case report will focus on generalised hypertrichosis in a child with JDM.

### Case Report

A two-year-old girl, from Kohat, came to the dermatology unit, Lady Reading Hospital, Peshawar, on November 15, 2021, presenting with a three-month history of generalised dusky, erythematous to violaceous plaques with noticeable swelling over eyes, face, knees, and acral body parts, along with intermittent low-grade fever. In addition, generalised hypertrichosis with hyperpigmentation and inability to walk during the past two months was reported. Twenty days back, multiple ulcers appeared on her neck, axillary,

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**Figure:** Generalised Hypertrichosis with Juvenile Dermatomyositis-Heliotrope rash, Poikilodermatous skin changes, Periungual erythema and Gottron's papules.

and groin region (Figure). She had a past medical history significant for measles and pneumonia in the previous year. As for her birth, it was an uneventful normal vaginal delivery with all developmental milestones achieved till the age of 18 months. There is no significant family history of delayed walking, myopathies, or similar rash. Her previous medical records showed use of topical steroids for three months. She had no complaint of cough, wheeze, central or peripheral cyanosis, joint pain, or any difficulty in swallowing and breathing.

Upon general physical examination, she was running a high-grade fever of 101 Fahrenheit (F) with the rest of the examination being unremarkable. As for cutaneous examination, there was significant periorbital erythema with excessive hair growth and hyperpigmentation on the face and body. Periungual erythema and poikilodermatous changes, such as hypo/hyperpigmentation, telangiectasias, and atrophy, were observed in the upper truncal area. Ulcerative lesions were also noted around the neck, axillary, and groin region due to vasculopathy.

Musculoskeletal examination revealed that power in all four limbs was reduced to 3/5, muscle tenderness to touch was appreciable but deep tendon reflexes were normal and no visible joint deformity, muscle atrophy, or calcification could be seen.

**Table:** Complete Blood Count and Serum Enzymes.

Investigations	Results	Normal ranges
White blood cell count	16.3x10.e3/ $\mu$ l	4-11x10.e3/ $\mu$ l
Haemoglobin level	8.75 g/dL	12-16g/dl
Haematocrit	28.7%	42%-54%
MCHC	30.5g/dL	33.4–35.5g/dL
MCV	53.9 fl	80–100 fl
Platelet count	574x 10.e3/ $\mu$ l	150-450 x 10.e3/ $\mu$ l
Serum lactate dehydrogenase	549U/L.	240-480U/L
Serum creatinine phosphokinase	189U/L	26-170U/L

MCHC: mean corpuscular haemoglobin concentration, MCV: mean corpuscular volume,  $\mu$ l: microlitre, dL: decilitre fl: femtolitre, U/L: units per litre.

Blood Investigations showed raised total leukocyte count (TLC) and muscle enzymes (Serum creatinine kinase and Lactate dehydrogenase) with low haemoglobin levels, whereas urine routine examination, serum albumin, calcium, potassium, magnesium, zinc, vitamin-D, liver, and kidney function tests were normal (Table).

Autoimmune screening was positive for anti-nuclear antibodies, whereas specific autoantibodies, including anti-Mi-2, anti-ARS, anti-NXP2, anti-SAE anti-TIF1-gamma, anti-MDA5, were not done due to unavailability of tests at the hospital and poor socioeconomic status of the patient's family. Furthermore, electromyography (EMG) was suggestive of myopathy. Imaging tests such as muscle MRI was also supportive of the diagnosis.

Based on Bohan and Peter criteria,<sup>5</sup> she was diagnosed with juvenile dermatomyositis, as she met four out of the five components, such as proximal muscle weakness, elevation of muscle enzymes, typical cutaneous changes, muscle MRI and EMG findings. It also met the updated criteria of the European League Against Rheumatism/American College of Rheumatology (EULAR/ ACR).<sup>6</sup>

After the confirmation of diagnosis, she was advised bed rest, sun protection, and hydration. Meanwhile, she was prescribed intravenous antibiotics and antipyretics followed by pulse steroid therapy for three days that was later changed to oral Prednisolone and intravenous DMARD (Ciclosporin).<sup>1</sup> She responded very well to the treatment and showed visible improvement in overall mood, and ability to walk with the reduction in body swelling. Furthermore, the parents were very pleased with their child's response to treatment and provided us with a written consent to share this rare co-finding to help future clinicians.

## Discussion

The term Dermatomyositis was first used in 1891 by Unverricht<sup>7</sup> for a condition that represented features of polymyositis with cutaneous manifestation. Juvenile dermatomyositis (JDM) is a subcategory of

dermatomyositis that presents in childhood involving inflammatory changes in the skin, muscle, heart, joints, and gastrointestinal tract. The age group for this condition falls under 16 years, with the female population having a higher risk.<sup>8</sup>

JDM can present with a range of cutaneous features among which heliotrope rash, periorbital oedema, and periungual telangiectasias are considered characteristic findings.<sup>9</sup> An identical violet rash with periorbital oedema and telangiectasias was seen in our patient. Skin rash appears in the early course of the disease in roughly two-thirds of the patients followed by muscle involvement. In addition to other diagnostic features, erythematous, and poikilodermatous lesions appear in the "shawl" or "V" pattern.<sup>10</sup> Vasculitic ulcers seen in JDM can involve flexural areas such as the axilla, groin, elbow, and knee.<sup>11</sup> Similar findings were obtained on examination of our patient.

Her hands had violaceous plaques over extensor surfaces of the metacarpophalangeal and distal interphalangeal joints with periungual erythema and prominent swelling, consistent with the pathognomonic Gottron's papules that occur in 70% of dermatomyositis.<sup>1</sup> Proximal muscle weakness often presents itself in JDM with a positive Gower's sign that requires pushing oneself up from prone position. Our patient had difficulty in standing and independent sitting.

JDM can be diagnosed clinically based on muscle weakness and characteristic rash in a child, but our patient was subjected to Bohan and Peter's criteria 1975<sup>5</sup> for which she had four out of five features that labelled her as a definitive case of JDM.

Keeping in consideration the unusual cutaneous findings of this rare disease, hypertrichosis is the least reported one with no literature available up to 1994.<sup>4</sup> Hypertrichosis is defined as excessive body hair that is further from the normal variations and affects parts that are androgen-independent. It is often confused for hirsutism which is a term used to describe excessive hair growth with male pattern distribution in patients, mainly females due to hyperandrogenism.<sup>12,13</sup>

It is classified into categories based on the age of appearance (congenital or acquired), degree of skin involvement (generalised or localised), site affected, follicle type (lanugo, vellous, and terminal) and whether it is an isolated disorder or associated with other conditions.

Congenital hypertrichosis, is a rare condition noted at birth that can present as an isolated skin anomaly or a feature of another disorder, whereas acquired hypertrichosis is more common, presenting after birth secondary to a range of

causes, such as malnutrition, drugs, iatrogenic, prepubertal, endocrine, and metabolic disorders, malignancies, autoimmune-cutaneous, and infectious conditions.<sup>14</sup>

In our case, the patient had dark pigmented vellus hair covering her entire body, predominately her forehead, cheeks, arms, and legs that became noticeable in two months' time putting her under the category of generalised acquired hypertrichosis.

As mentioned earlier, generalised acquired hypertrichosis has a variety of causes that needs to be excluded to identify the causative factor to plan its management. Patients may have to deal with difficulty in assessment as the characteristics are diverse and can lead to delay in the establishment of diagnosis.<sup>15</sup> Therefore, timely identification of such conditions helps to determine the need for early interventions, screening for malignancies, cutaneous diseases, endocrine and metabolic disorders along with counselling of patients and their family members.

This case report highlights the rare finding of generalised acquired hypertrichosis in a patient of JDM to encourage development of protocols and investigations to be done by future clinicians in making the most appropriate decisions when encountering such a case.

## Conclusion

This study shows that generalised acquired hypertrichosis, despite being a rare manifestation of JDM, may be more frequent than reported. Most importantly, such a co-finding can be seen and does not require excessive investigations to rule out other causes of hypertrichosis in a patient of JDM. This case report reinforces a need for further studies, reporting rare findings, establishing protocols, good decision making, and training for the identification and management of hypertrichosis and its associated disorders.

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## References

1. Rook A, Barker J, Bleiker T, Chalmers R, Creamer D, Griffiths C. Rook's textbook of dermatology 8th ed. Chichester, West Sussex: Wiley Blackwell, 2010; pp-53.9-53.12.
2. Mendez EP, Lipton R, Ramsey-Goldman R, Roettcher P, Bowyer S, Dyer A, et al. NIAMS Juvenile DM Registry Physician Referral Group. US incidence of juvenile dermatomyositis, 1995-1998: results from the National Institute of Arthritis and Musculoskeletal and Skin Diseases Registry. *Arthritis Rheum.* 2003; 49:300-5. doi: 10.1002/art.11122.
3. Wendelin DS, Pope DN, Mallory SB. Hypertrichosis. *J Am Acad Dermatol.* 2003; 48:161-79. doi: 10.1067/mjd.2003.100.
4. Pope DN, Strimling RB, Mallory SB. Hypertrichosis in juvenile dermatomyositis. *J Am Acad Dermatol.* 1994; 31:383-7. doi: 10.1016/s0190-9622(08)80361-1.
5. Bohan A, Peter JB. Polymyositis and dermatomyositis. *N Engl J Med.* 1975; 292:344-7. doi: 10.1056/NEJM197502132920706.
6. Oldroyd A, Chinoy H. Recent developments in classification criteria and diagnosis guidelines for idiopathic inflammatory myopathies. *Curr Opin Rheumatol.* 2018; 30:606-13. doi: 10.1097/BOR.0000000000000549.
7. Bronner IM, Linssen WH, van der Meulen MF, Hoogendijk JE, de Visser M. Polymyositis: an ongoing discussion about a disease entity. *Arch Neurol.* 2004; 61:132-5. doi: 10.1001/archneur.61.1.132.
8. Ishaque S, Ahmed S, Ali R, Minhas K. Juvenile dermatomyositis. *J Coll Physicians Surg Pak.* 2011; 21:434-6.
9. Kovacs SO, Kovacs SC. Dermatomyositis. *J Am Acad Dermatol.* 1998; 39:899-920. doi: 10.1016/s0190-9622(98)70263-4.
10. Symmons DP, Sills JA, Davis SM. The incidence of juvenile dermatomyositis: results from a nation-wide study. *Br J Rheumatol.* 1995; 34:732-6. doi: 10.1093/rheumatology/34.8.732.
11. Gupta A, Paliana RK, Prasad VD, Guleria S. Extensive skin ulcers in a child with juvenile dermatomyositis. *BMJ Case Rep.* 2018; 2018:bcr2017222915. doi: 10.1136/bcr-2017-222915.
12. Saleh D, Yarrarapu SNS, Cook C. Hypertrichosis. In: Saleh D, Yarrarapu SNS, Cook C. Treasure Island: Stat Pearls Publishing, 2022.
13. Buch J, Ranganath P. Approach to inherited hypertrichosis: A brief review. *Indian J Dermatol Venereol Leprol.* 2021; 88:11-21. doi: 10.25259/IJDVL\_629\_20.
14. Pavone P, Praticò AD, Falsaperla R, Ruggieri M, Zollino M, Corsello G, Neri G, et al. Congenital generalized hypertrichosis: the skin as a clue to complex malformation syndromes. *Ital J Paediatr.* 2015; 41:55. doi: 10.1186/s13052-015-0161-3.
15. Carneiro VF, Barbosa MC, Martelli DRB, Bonan PR, Aguiar MJB, Martelli Júnior H. A review of genetic syndromes associated with hypertrichosis. *Rev Assoc Med Bras.* 2021; 67:1508-14. doi: 10.1590/1806-9282.20210666.