CASE REPORT

Journal of Pre-Clinical and Clinical Research
www.jpccr.eu

Diagnostic difficulties of dermatomyositis – case report

Kinga Wróblewska[1,A-E]✉, Monika Żybowska[1,C-E], Zofia Piaszczyk[1,B-E], Klaudia Miller[1,B-D], Adrianna Wieleba[1,B-D], Klaudia Szukała[2,F], Magdalena Chrościńska-Krawczyk[2,F]

[1] *Student Research Group, Department of Child Neurology, Medical University, Lublin, Poland*
[2] *Department of Child Neurology, Medical University, Lublin, Poland*
A – Research concept and design, B – Collection and/or assembly of data, C – Data analysis and interpretation, D – Writing the article, E – Critical revision of the article, F – Final approval of the article

Wróblewska K, Żybowska M, Piaszczyk Z, Miller K, Wieleba A, Szukała K, Chrościńska-Krawczyk M. Diagnostic difficulties of dermatomyositis – case report. J Pre-Clin Clin Res. doi: 10.26444/jpccr/192305

Abstract

Neuromuscular disorders are a group of diseases which can result in severe development disorders. They can be demonstrated by many different symptoms. The case report presents the history of a patient who reported pain in the lower limbs and muscle weakness that made it difficult to move. Result of the EMG examination revealed neurogenic changes, suggesting the possibility of suffering from a motor variant of chronic inflammatory demyelinating polyradiculoneuropathy (CIDP), although not all the symptoms presented by the patient were characteristic. During subsequent hospitalizations, skin changes were noticed in the form of heliotropic erythema, which, based on a number of additional tests, finally led to the diagnosis of juvenile dermatomyositis. The occurrence of a neurological mask in the course of juvenile dermatomyositis indicates the need for a wide differential diagnosis.

Key words

CIDP, juvenile dermatomyositis, neuromuscular disorders

INTRODUCTION

Neuromuscular disorders are a broad group of different diseases with a prevalence of approximately 160/100 000 people. The diseases have a negative impact on the functioning of the nervous system and a consequent limitation of motor skills. The disorders can include primary muscle pathologies (myopathies) and neuromuscular conduction disorders (e.g. myasthenia gravis), as well as peripheral neuropathies and motor neuron diseases. In the paediatric population, these conditions can lead to severe impairment of psychomotor development. Children with neuromuscular disorders may experience delayed and impaired growth and sexual maturation [1]. Because neuromuscular symptoms may not be pathognomonic of a specific condition and may accompany a variety of diseases, establishing a diagnosis is often difficult and requires a long-term follow-up of the patient, as well as the exclusion of other potential conditions [2].

CASE REPORT

A girl aged 3 years and 11 months was admitted to the Paediatric Neurology Department due to gait disturbances that had been present for 3 months, reluctance to move, and lack of interest in playing. The patient periodically experienced pain in her lower limbs which required analgesics. The girl did not have any chronic illness and was not taking any medication. She was born at term by caesarean section due to the detection of SARS-CoV-2 infection in her mother. From about 3 months of age, the girl was under the care of a physiotherapist due to the weakness in muscle tone observed at that time. The child's development was normal until the age of 2 years.

One month after the onset of the first symptoms, the patient was hospitalised for 14 days in the Paediatric Ward of the district hospital. Initially, polyneuropathy or dermatomyositis was suspected, and steroids and antibiotics were administered. A few days after the child was discharged home, the muscle symptoms returned with increased severity, and follow-up blood tests showed a renewed increase in creatine kinase (CK) levels.

The girl was referred to the University Children's Hospital. On admission, she was in good general condition and no abnormalities were found on general examination. Laboratory tests were performed which revealed high levels of CK and lactate dehydrogenase (LDH) – (611 U/l, normal: <850), aspartate aminotransferase (AST) (83 U/l, normal: <56). The CK level reached 759.0 U/l (normal: <288.0).

During the neurological consultation, the child's gait was very cautious, requiring assistance from the mother. There was involuntary slight contracture of the knees, slightly weakened muscle strength, and reduced distal muscle tone. A positive Gowers sign was found – the girl supported herself and 'climbed' on herself or a nearby object when standing up and sitting down. There were no signs of cranial nerve damage, deep reflexes were symmetrical and vivid, and Babinski's sign was negative.

In the following days, the patient's blood continued to show high levels of CK (700–800 U/l) and AST (71 U/l), as well as D-Dimers (1,300). Blood morphology, electrolytes and coagulation parameters were normal. Virological and bacteriological tests were negative for Lyme disease, CMV, EBV, HSV and chickenpox; however, IgM antibodies against enteroviruses were present, indicating active infection. Tests for rheumatological markers (ANCA profile – anti-neutrophil

✉ Address for correspondence: Kinga Wróblewska, Student Research Group, Department of Child Neurology, Medical University, Lublin, Poland
E-mail: kinga.wroblewska.kw@gmail.com

Received: 25.06.2024; accepted: 13.08.2024; first published: 05.09.2024

cytoplasmic antibodies, RF – rheumatoid factor, CCP – cyclic citrullinated peptide, anticardiolipin and antinuclear antibodies) were negative. In addition, no onco-neuronal antibodies and organic acids were found in the urine.

An EMG study (electromyography) was performed which revealed features of neurogenic damage – the PJR (motor unit potential) had high amplitude and shortened duration, indicating features of demyelinating motor polyneuropathy. An MLPA (Multiplex Ligation-dependent Probe Amplification) genetic test was recommended, using probes targeting the MPZ gene, GJB1, the 17p12 region including the PMP22 gene and the X chromosome. The results ruled out both genomic imbalance and genomic imbalance in critical regions of the microdeletion/microduplication syndromes represented in the probe set used.

A head CT scan was performed which showed no abnormalities. Cerebrospinal fluid (CSF) was collected in which cell counts above the upper limit of normal, lymphocytosis and low protein levels were present. However, due to insufficient collection, it was not possible to perform a panel of 14 pathogens from the CSF.

Due to the suspicion of CIDP (chronic inflammatory demyelinating polyneuropathy), treatment with immunoglobulin pulses for 5 days was implemented. During the treatment, the girl's condition was stable, although her symptoms remained unchanged. A few days after completion of IgG treatment, the patient showed a slow return of motor function and gradually began to walk more, and even run. After 14 days of hospitalisation, the girl was discharged home in good general condition.

After 5 months, the patient was seen for a follow-up visit which showed CK, LDH, AST and ALT levels were still elevated. Renewed immunoglobulin therapy of 5 days duration was administered. The CK level decreased slightly, but the observed effects were not as significant as with the previous doses. A neurological examination was also performed and no changes were observed compared to the previous examination. In addition, an MRI (Magnetic Resonance Imaging) of the head was performed, which showed hyperdense areas without features of restriction or enhancement after contrast administration in the lateral globus pallidus, visual pathway (especially on the right side), and in the white matter of the occipital lobe, which was unrelated to the symptoms presented by the patient.

During hospitalisation, the girl was noted to have skin lesions – erythema of the eyelids (erythema heliotrope), single red nodules on the elbow and interphalangeal joints of the upper limbs. A rheumatology consultation was performed during which symptoms of dermatomyositis were found.

Magnetic resonance imaging of the soft tissues of the thighs was also performed, which showed diffuse, poorly circumscribed areas of increased signal intensity on T2-weighted images with fat saturation in the thigh muscles, most intensely in the vastus lateralis muscles, especially of the left lower limb. Small lesions of a similar nature were also found bilaterally in the gluteal muscles, with predominance on the left side. A decision was made to transfer the girl to the Rheumatology Department for further diagnosis and treatment (Tab 1, Tab 2).

Table 1. The most important symptoms of CIDP

Categories	Type	Description	Result in patient
Clinical (neurological)	Sensory and motor neuropathy, muscle weakness	Symmetrical, proximal and distal neuropathy, which may be monophasic, recurrent and/or progressive; muscle weakness, often with initial difficulty in walking [3]	Partially yes, (muscle weakness occurred, but sensory conduction reserved)
Clinical (neurological)	Reflex	Absence or weakening of tendon reflexes (deep) in all limbs [3]	NO
Electrodiagnostic	EMG examination	Partial conduction block, slow conduction velocity, prolonged distal motor delays, F wave delay or disappearance [4]	Yes
Pictorial	MRI examination	Increased signal intensity on T2-weighted imaging [5]	Yes
Laboratory	Cerebrospinal fluid (CSF)	Increased amount of proteins in cerebrospinal fluid [3,6]	NO
Response to treatment	Treatment with immunoglobulins, corticosteroids, plasmapheresis	positive response to treatment with corticosteroids and/or immunoglobulins and/or plasmapheresis [6]	Yes

Table 2. The most important symptoms of dermatomyositis [7–9]

Category	Type	Description	Occurrence in patient
Cutaneous	Gottron's papules	Erythematous to violaceous papules with hypertrophy of epidermis over extensor surfaces of joints	NO
	Gottron's sign	Erythematous to violaceous macules over extensor surfaces of joints, which are not palpable	Yes
	Erythema	Heliotrope erythema - glasses-shaped, violet in colour, may be accompanied by swelling	Yes
		Covering neckline, 'V-sign'	NO
		Covering neck and shoulders (scarf sign)	NO
	"Mechanic's hands"	Thickening, cracking and peeling of skin on fingertips and palm of hand	NO
Muscular	Symmetrical muscle weakness	Mainly affects proximal muscles (shoulder girdle, pelvic girdle, neck and back); makes everyday functioning difficult, may be accompanied by muscle tenderness and pain	Yes
	Weakness of other muscles of throat, oesophagus and larynx	May lead to dysphagia and dysphonia	NO
	Weakness of respiratory muscles	May cause respiratory failure	NO
General	Fatigue	feeling of chronic fatigue, reduced activity reported by the patient	Yes
	Weight loss	Weight loss not related to caloric deficit	NO

DISCUSSION

In the clinical case described, the patient's symptoms gradually worsened. Initially, pain and weakness occurred in the muscles of the lower limbs, which made movement very difficult. In addition, there were elevated creatine kinase levels and a lack of response to treatment with glucocorticosteroids, which are used for inflammatory myopathies. EMG examination revealed changes indicative of damage to motor fibres with preserved normal conduction in sensory fibres, suggesting the presence of features of demyelinating polyneuropathy. This led to the diagnosis of the most common autoimmune polyneuropathy, i.e. chronic inflammatory demyelinating polyradiculoneuropathy, which is a heterogeneous disease with many types and variants [10].

The main symptoms characterising CIDP include symmetrical motor and sensory neuropathy, which can present as a monophasic, recurrent or progressive disorder [11]. The diagnosis is made on the basis of clinical symptoms, electrodiagnostic, and laboratory criteria which, in the patient described, did not clearly indicate the above disease entity [10]. The girl only showed some features associated with the motor variant of CIDP, including weakness of lower limb muscle strength with normal sensory conduction, a rare F-wave frequency, a characteristic EMG picture, and increased intensity and/or contrast enhancement of nerves on MRI [12].

In contrast, reduced protein levels in the cerebrospinal fluid and preserved tendon reflexes argued against this diagnosis. It should be emphasised that the symptoms presented are not pathognomonic for CIDP, they also occur in other diseases;,it is therefore important to consider alternative diagnoses, hence other inflammatory myopathies were included in the differential diagnosis. Due to the difficulty in diagnosing this disease, treatment was proving to be complicated [11–13].

The motor form of CIDP shows a better response to treatment with immunoglobulins than with steroids, and the cessation of the patient's symptoms after infusion may suggest the presence of this variant of the disease [14].

In addition, due to the young age of the patient and the suspicion of an autoimmune disease with features of demyelinating polyneuropathy, genetic testing was performed using the MLPA technique to exclude mutations in genes responsible for hereditary neuropathies. Clinical signs of classic CMT include: loss of sensation, distal muscle weakness/atrophy and characteristic high foot arches [15].

One month later, the patient's symptoms recurred, revealing previously unreported skin symptoms which indicated the diagnosis of juvenile dermatomyositis, which is the most common form of myositis in children [16].

JDM is a rare immune-mediated connective tissue disease with characteristic changes including cutaneous (heliotropic rash, Gottron papules) and muscle-related manifestations, including: symmetrical proximal muscle weakness, high serum muscle enzyme activity and electromyography (EMG) abnormalities. Elevated LDH, ALT and AST levels may result from muscle breakdown during inflammation [17–19]. The appearance of antibodies to enteroviruses probably suggests another cause of the inflammation present in the young patient.

CONCLUSIONS

The clinical case presented suggests the possibility of a neurological mask in the course of dermatomyositis. In addition to the initially confusing symptoms, it is also distinguished by the co-occurrence of enteroviral infection, which may have had an as yet unexplored effect on the clinical picture. Only detailed diagnostics, including laboratory tests and muscle biopsy, revealed the presence of characteristic inflammatory changes typical of dermatomyositis.

REFERENCES

1. Deenen JC, Horlings CG, Verschuuren JJ, et al. The Epidemiology of Neuromuscular Disorders: A Comprehensive Overview of the Literature. J Neuromuscul Dis. 2015;2(1):73–85. https://doi.org/10.3233/JND-140045
2. McCann LJ, Livermore P, Wilkinson MGL, et al. Juvenile dermatomyositis. Where are we now? Clin Exp Rheumatol. 2022 Feb;40(2):394–403. https://doi.org/10.55563/clinexprheumatol/56ilob
3. Van den Bergh PYK, van Doorn PA, Hadden RDM, et al. European Academy of Neurology/Peripheral Nerve Society guideline on diagnosis and treatment of chronic inflammatory demyelinating polyradiculoneuropathy: Report of a joint Task Force-Second revision. Eur J Neurol. 2021 Nov;28(11):3556–3583. https://doi.org/10.1111/ene.14959
4. Ponnala M, Mullen B, Nawab K, et al. Chronic Inflammatory Demyelinating Polyneuropathy (CIDP): Overview, Treatment, and a Case Study. Cureus. 2023 Oct 22;15(10):e47475. https://doi.org/10.7759/cureus.47475
5. Lewis RA, van Doorn PA, Sommer C. Tips in navigating the diagnostic complexities of chronic inflammatory demyelinating polyradiculoneuropathy. J Neurol Sci. 2022 Dec 15;443:120478. https://doi.org/10.1016/j.jns.2022.120478
6. Ryan M, Ryan SJ. Chronic inflammatory demyelinating polyneuropathy: considerations for diagnosis, management, and population health. Am J Manag Care. 2018 Sep;24(17 Suppl):S371–S379. PMID: 30312032, 24. S371–S379.
7. Rhim JW. Juvenile Dermatomyositis. J Rheum Dis. 2022;29(1):14–21. https://doi.org/10.4078/jrd.2022.29.1.14
8. Li D, Tansley SL. Juvenile Dermatomyositis-Clinical Phenotypes. Curr Rheumatol Rep. 2019 Dec 11;21(12):74. https://doi.org/10.1007/s11926-019-0871-4
9. Mamyrova G, Kishi T, Targoff IN, et al. Features distinguishing clinically amyopathic juvenile dermatomyositis from juvenile dermatomyositis. Rheumatology (Oxford). 2018;57(11):1956–1963. https://doi.org/10.1093/rheumatology/key190
10. Bunschoten C, Jacobs BC, Van den Bergh PYK, et al. Progress in diagnosis and treatment of chronic inflammatory demyelinating polyradiculoneuropathy. Lancet Neurol. 2019 Aug;18(8):784–794. https://doi.org/10.1016/S1474-4422(19)30144-9
11. Vallat JM, Sommer C, Magy L. Chronic inflammatory demyelinating polyradiculoneuropathy: diagnostic and therapeutic challenges for a treatable condition. Lancet Neurol. 2010 Apr;9(4):402–12. https://doi.org/10.1016/S1474-4422(10)70041-7
12. Chen H, Huang X, Bao Y, et al. The diagnostic value of quantitative assessment of MR neurography in chronic inflammatory demyelinating polyradiculoneuropathy: a systematic review and meta-analysis. Br J Radiol. 2023 Nov;96(1151):20221037. https://doi.org/10.1259/bjr.20221037
13. Van den Bergh PYK, van Doorn PA, Hadden RDM, et al. European Academy of Neurology/Peripheral Nerve Society guideline on diagnosis and treatment of chronic inflammatory demyelinating polyradiculoneuropathy: Report of a joint Task Force-Second revision. Eur J Neurol. 2021 Nov;28(11):3556–3583. https://doi.org/10.1111/ene.14959
14. Van den Bergh PYK, van Doorn PA, Hadden RDM, et al. European Academy of Neurology/Peripheral Nerve Society guideline on diagnosis and treatment of chronic inflammatory demyelinating polyradiculoneuropathy: Report of a joint Task Force—Second revision. Journal of the Peripheral Nervous System. 26(3):242–268. https://doi.org/10.1111/jns.12455

15. Barreto LC, Oliveira FS, Nunes PS, de França Costa IM, et al. Epidemiologic Study of Charcot-Marie-Tooth Disease: A Systematic Review. Neuroepidemiology. 2016;46(3):157–65. https://doi.org/10.1159/000443706
16. Quartier P, Gherardi RK. Juvenile dermatomyositis. Handb Clin Neurol. 2013;113:1457–63. https://doi.org/10.1016/B978-0-444-59565-2.00014-9
17. Wu JQ, Lu MP, Reed AM. Juvenile dermatomyositis: advances in clinical presentation, myositis-specific antibodies and treatment. World J Pediatr. 2020 Feb;16(1):31–43. https://doi.org/10.1007/s12519-019-00313-8
18. Patil A, Lu J, Kassir M, Babaei M, Goldust M. Adult and juvenile dermatomyositis treatment. J Cosmet Dermatol. 2023 Feb;22(2):395–401. doi:https://doi.org/10.1111/jocd.15363
19. Swafford C, Roach ES. Juvenile Dermatomyositis and the Inflammatory Myopathies. Semin Neurol. 2020 Jun;40(3):342–348. doi:https://doi.org/10.1055/s-0040-1705120