

From “Atopic Eczema” to Polymyositis-Systemic Sclerosis Overlap Syndrome requiring mechanical ventilation: A Diagnostic Odyssey

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Background

Polymyositis-Systemic Sclerosis (PM-SSc) overlap syndrome is a rare autoimmune disease with multisystem involvement. **Early recognition is essential for timely and effective treatment.**

A 14-year-old female presented to the University Hospital Leipzig (UKL) with respiratory failure of unknown cause, requiring mechanical ventilation. Her symptoms began six months earlier with skin lesions, initially diagnosed as atopic eczema.

This report highlights the clinical course of a pediatric patient initially misdiagnosed, **emphasizing key diagnostic milestones.**

Conclusion

This case highlights **the diagnostic challenges of PM-SSc overlap syndrome** particularly when initial presentations resemble common conditions like atopic eczema. The progression of muscle weakness, refractory skin findings and systemic symptoms underlines **the importance of early referral and detailed investigation** in a center specialized on treating rare rheumatological diseases.

In pediatric myositis syndromes, **accurate diagnosis and timely intervention are crucial for preventing severe complications** such as the necessity of mechanical ventilation or other long-term sequelae to optimize outcomes in such rare pediatric autoimmune diseases.

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Case Report

- **Initial Symptoms** (6 months prior): Skin lesions diagnosed as atopic eczema, worsening despite treatment with topical corticosteroids and Dupilumab
- **Progressive Symptoms:** Muscle weakness, joint pain, fatigue, unintentional weight loss (5 kg over four weeks), difficulty with daily activities (e.g. climbing stairs, opening bottles)
- **Acute Worsening** (1 week before admission): Severe dyspnea and proximal muscle weakness, hospitalized for hypoxemia and respiratory distress
- **Initial Investigations** (Local Hospital): Pulmonary embolism excluded, elevated Troponin T and creatine kinase CK - suggestive of cardiac involvement
- **Further Evaluation:** Intubated and transferred to Leipzig Heart Center, myocardial pump function disorder excluded, transferred to Pediatric Intensive Care Unit (UKL)
- **Key Dermatological Findings:** Mild heliotrope rash, minor Gottron's papules, hyperpigmented “mechanic's hands”



Fig. 1: „Mechanic's hands”: Dry, erythematous, rough skin and hyperkeratosis as well as scaling

Capillaroscopic pattern

- **Pathological findings of the nailfold capillaroscopy**
- Density: Borderline **lowered density** (approx. 6/mm) **with focal capillary loss**
- Morphology: Hardly any classic hairpin shape (<20%), mostly convex capillary tip, few torquings (<18%), **significantly increased ramification** (>20%), few elongations (1.6%), transition to bushy capillaries, hardly any caliber fluctuations
- Dimension: isolated ectasia up to 32µm, no giant capillary
- Microhemorrhages: none

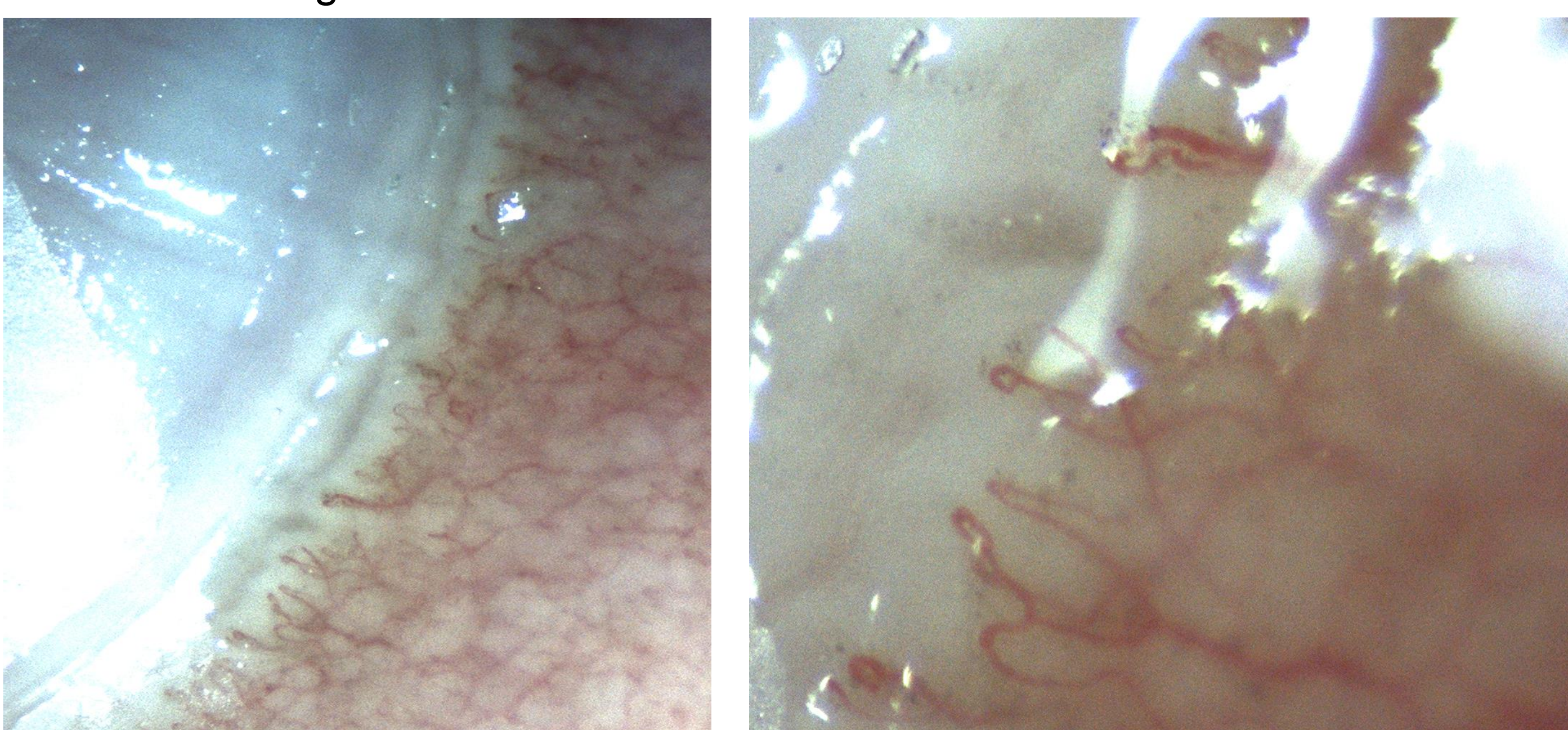


Fig. 2: Chronic changes in the capillaroscopy

Diagnosis & Histopathology

- **Suspected Autoimmune Process:** dermatological findings, markedly elevated ANA (1:7100) and CK (max. 33µkat/l, nv <2,05)
- **Confirmed Diagnosis of PM-SSc overlap syndrome:** Muscle biopsy and high anti-PM/Sc170 antibody titers (associated with a favorable prognosis)

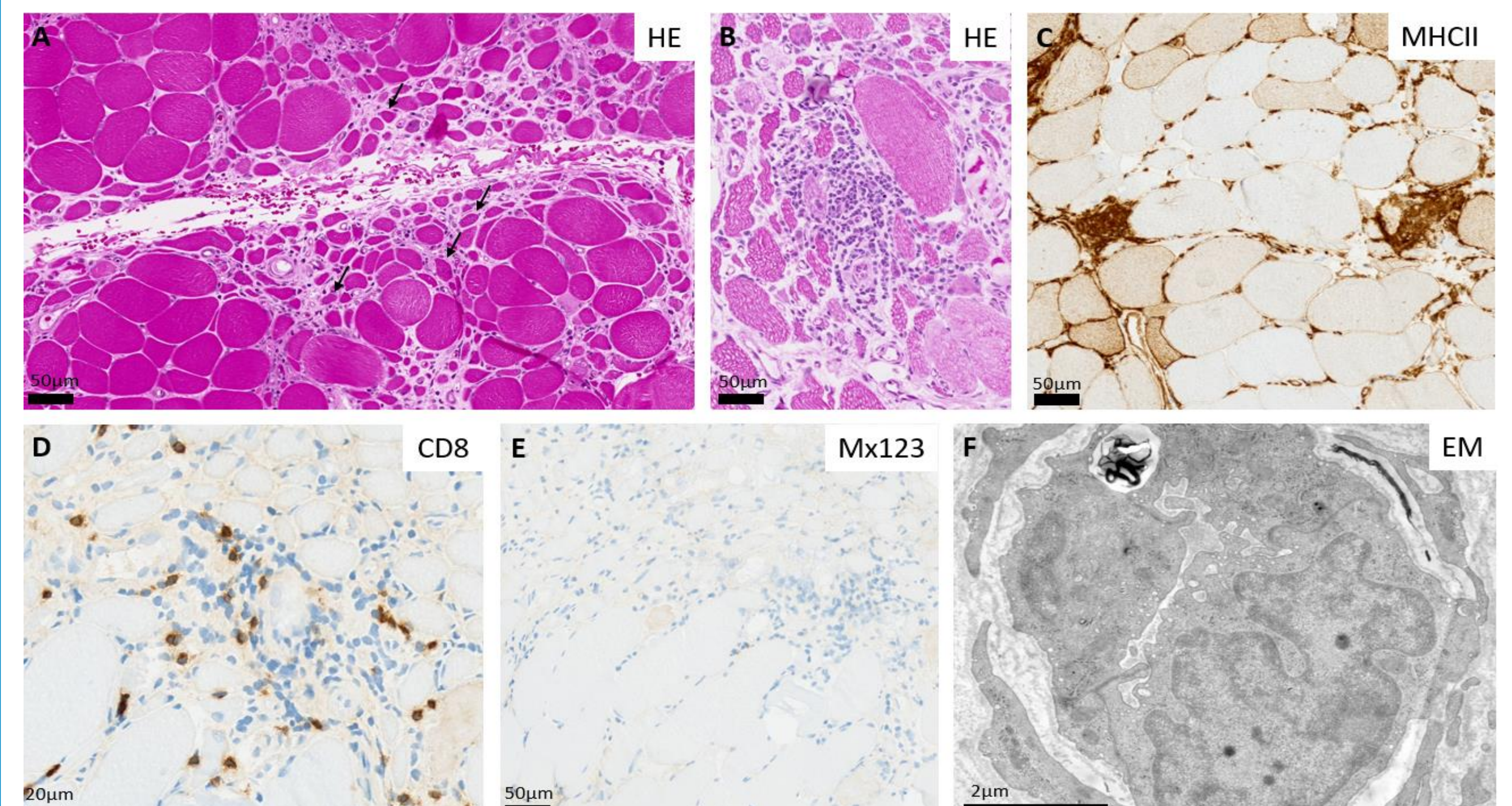


Fig. 3: HE staining shows severe perifascicular atrophy and muscle fibers with internal nuclei (arrows, A) next to groups of endomysial lymphocytic infiltrates (B). MHCII labels interspersed myophagocytosis (C). Lymphocytic infiltrates are partially positive for CD8 (D). There is upregulation of Mx123 (E). Electron microscopy (EM) shows endothelial cell activation and basal membrane thickening (F).

Therapy and Clinical course

- High-dose **methylprednisolone pulses** and **Immunglobulin** in conjunction with oral **prednisolone**, **Methotrexate** and **Mycophenolate-Mofetil**
- Despite transiently marked B-cell expansion, Rituximab was withheld due to clinical improvement, with **B-cell numbers normalizing spontaneously**
- **Respiratory Recovery:** Gradual weaning from invasive ventilation to CPAP via tracheostomy over several weeks
- **Functional Rehabilitation** and **CMAS Score Progression** (11 to 27/52 within one month, 44/52 six months later, currently 50/52)
- **Full Recovery:** Resumed daily activities (even dancing classes) without overt residual sequelae

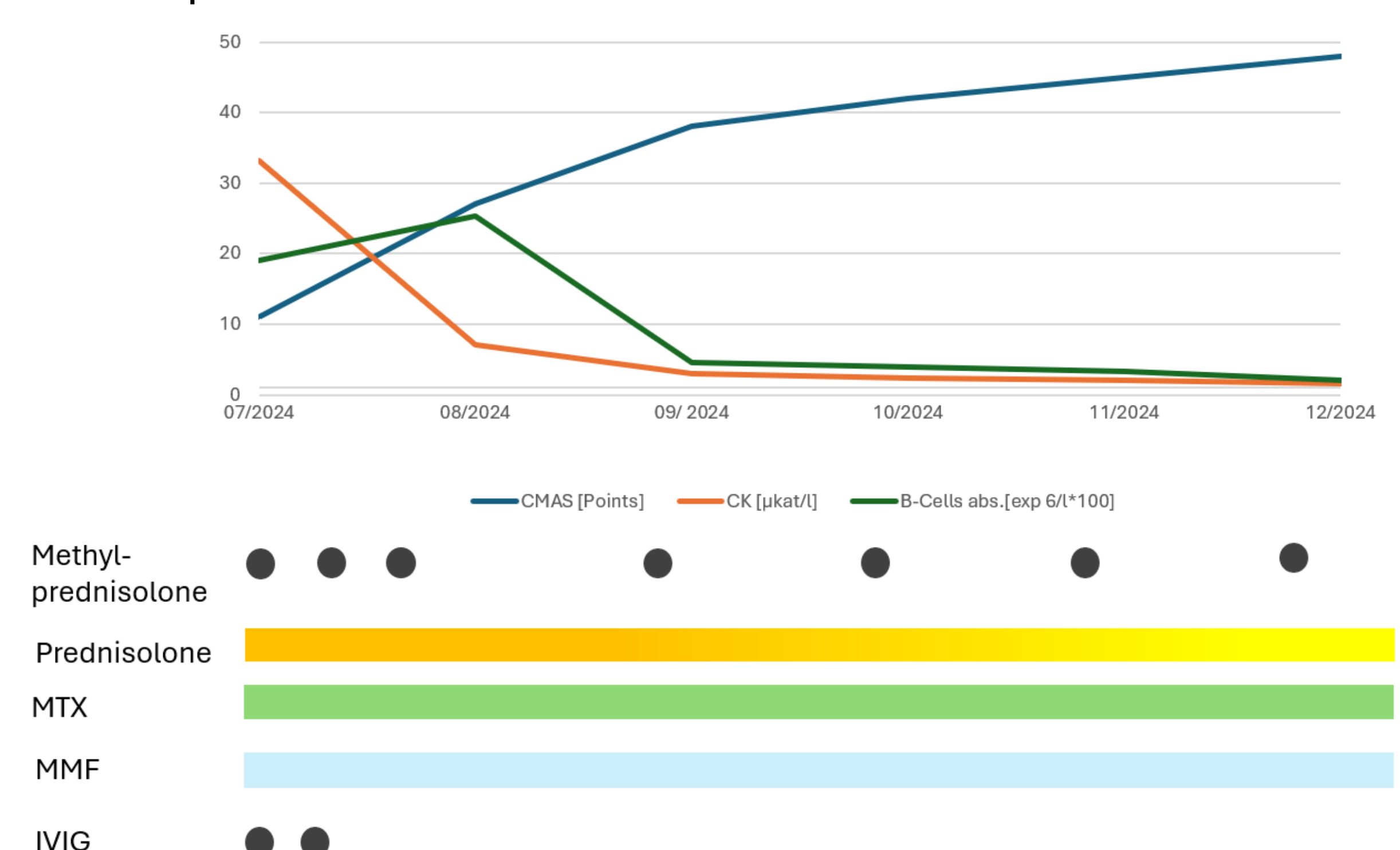


Fig. 4: Progression of CK, B-Cells and CMAS (Childhood Myositis Assessment Scale)