



Rarity Redefined: Anti-Ku Antibody–Associated Myopathy In a Child

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Abstract

Background

Anti-Ku antibody–associated inflammatory myopathy is a rare autoimmune disorder, predominantly described in adults and only rarely reported in pediatric patients.

Case-Presentation

An 8-year-old male presented with a one-month history of high-grade fever followed by progressive proximal muscle weakness, leading to difficulty in walking and performing overhead activities. There was no history of rash, joint symptoms, or trauma. Examination revealed symmetric proximal muscle weakness.

Management & Outcome

Laboratory evaluation showed markedly elevated muscle enzymes (CK, AST, LDH) and inflammatory markers. Autoimmune workup revealed positivity for anti-Ku antibodies. MRI of lower limbs demonstrated T2/STIR hyperintensities suggestive of inflammatory myopathy. The patient was treated with pulse methylprednisolone followed by oral corticosteroids and methotrexate, resulting in significant clinical improvement and restoration of functional mobility.

Conclusion

This case highlights a rare early presentation of anti-Ku myopathy in childhood. Early recognition using serological and radiological correlation and prompt immunosuppressive therapy can significantly improve outcomes.

Keywords

Anti-Ku antibody, Inflammatory myopathy, pediatric myositis, autoimmune myopathy, rare case

1. Introduction

Idiopathic inflammatory myopathies are a heterogeneous group of autoimmune muscle disorders, including dermatomyositis, polymyositis, immune-mediated necrotizing myopathy, and overlap syndromes. Myositis-specific and myositis-associated autoantibodies play an important role in classification and prognosis [1-2].

Anti-Ku antibodies, initially described in overlap syndromes involving polymyositis and systemic sclerosis, are rarely associated with isolated inflammatory myopathy. Pediatric cases are exceedingly uncommon, with most reported cases occurring in adults. Here, we report a rare case of anti-Ku antibody-associated myopathy in a young child, expanding the known clinical spectrum [2-5].

2. Case Description

An 8-year-old male child, born of non-consanguineous marriage with an unremarkable birth and developmental history, presented with complaints of fever and

progressive weakness of all four limbs for one month.

The illness began with high-grade continuous fever associated with chills and rigors, partially relieved with medications. After one week, the child developed insidious onset weakness, initially involving proximal muscles. He had difficulty rising from sitting, climbing stairs, and combing hair, which progressively worsened to inability to walk independently and difficulty in extending upper limbs.

There was no history of trauma, rash, joint pain, joint swelling, or sensory symptoms.

3. Examination

- Conscious, hemodynamically stable
- No rash or organomegaly
- Neurological examination:
 - Symmetric proximal muscle weakness (proximal > distal)
 - MRC score: 42/60 (table I)
 - Reflexes preserved
 - No cranial nerve involvement

Table I : Muscle Power Grading of the Patient

Muscle Group	At Presentation	Post Treatment
Shoulder	3/5	5/5
Elbow	3/5	5/5
Wrist	3/5	5/5
Hip	2/5	4/5
Knee	2/5	5/5
Ankle	3/5	5/5

4. Management And Outcome

Investigations showed elevated muscle enzymes (Aspartate Transaminase- AST, Lactate Dehydrogenase- LDH, Creatinine Kinase- CK), with positive inflammatory markers (elevated white blood cell counts, C-reactive protein- CRP, Erythrocyte Sedimentation Rate- ESR). Blood culture was sterile. With the background of chronic history of fever with quadriparesis (Proximal> Distal, MRC score- 42) and elevated muscle enzymes, child was provisionally diagnosed as Inflammatory Myopathies. Autoimmune workup was sent that showed negative anti-nuclear antigen, normal complement levels. Extended myositis panel was ordered, that showed positivity for Anti-Ku Antibodies (Table

1).

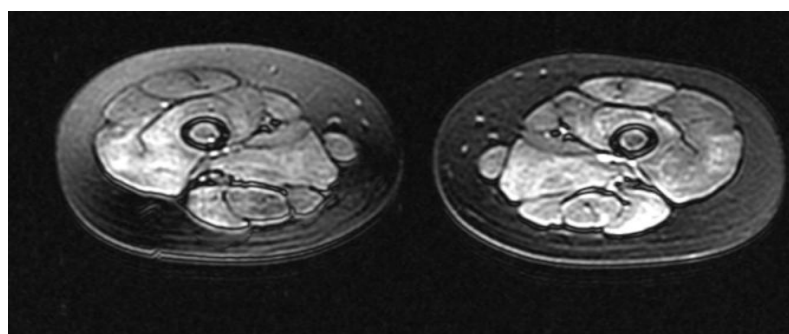
MRI Lower limb STIR sequence was planned to see for any evidence of muscle inflammation, that showed hyperintensities in T2 Axial images and STIR sequence (Figure 1, 2). Muscle biopsy was not done for the child.

The child was screened for any cardiac involvement, cardiac biomarkers were negative and 2d echocardiography showed normal systolic and diastolic function.

Child was treated with pulse dose of steroids and continued maintenance oral steroids for which was gradually tapered with addition of second line agent (Methotrexate). On follow up visits, the child had improved significantly, with child resuming to her previous activities and return to normal life with minimal disability. (Table 1).

Table II : Laboratory Investigation

Investigation	Result
Hemoglobin/Total WBCs	10.6g/dL/ 13020/mm ³
C reactive protein	20mg/L
Erythrocyte sedimentation rate	40 mm at the end of 1st hour
Creatinine Kinase/ AST/ LDH	22969U/L/ 839U/L / 3128U/L
ANA/ANCA/C3/C4	Negative/negative/ 1.2g/L / 0.3g/L
Anti Ku Antibody	Positive
Mi-2a-Ab	Negative
Mi-2B Ab	
TIF1-Y Ab	Negative
MDA-5 Ab	Negative
NXP2 Ab	Negative
SAE-1 Ab	
PM Scl-100 Ab	Negative
PM Scl-75 Ab	
Jo-1 Ab	Negative
SRP Ab	
PL-7 Ab	Negative
PL-12 Ab	
EJ Ab	Negative
OJ Ab	
Ro-52 Ab	Negative

**Figure 1- MRI showing T2/STIR hyperintensities in affected muscles suggestive of inflammatory myopathy****5. DISCUSSION**

The autoimmune myopathies consist of five main conditions, namely dermatomyositis (DM), polymyositis (PM), myositis associated with anti-synthetase syndrome (ASS), immune mediated necrotizing myopathy (IMNM), and inclusion body myositis (IBM). These conditions can occur in isolation or can be associated with systemic malignancies or connective tissue disorders (overlap syndromes) and all are often associated with autoantibodies [1].

The anti-Ku-antibody was first described in 1981 in patients of polymyositis-scleroderma-overlap-syndromes [2]. Thereafter, the Anti-Ku antibodies have been implicated in various connective tissue disorders including SLE, systemic sclerosis, Sjögren's syndrome, idiopathic lung fibrosis, overlap syndromes with systemic sclerosis (Ssc) and myositis. Although prevalence of Anti Ku positive inflammatory myopathies has been described till 23% in some population [5], anti-Ku + myositis remains a rare entity, particularly in children.

Eukaryotic Ku is a heterodimer complex of two polypeptides of 70 and 80 KD (Ku70(XRCC6) and Ku80 (XRCC5) respectively). Ku complex plays a key role in DNA repair, and is implicated in the regulation of many cellular processes such as immunoglobulin gene recombination, telomere protection and regulation of gene transcription [4-5]. The precise pathogenesis of autoimmunity in anti Ku positive disease is rather elusive, but studies suggest a possible interaction between Ku, Ku-antibodies, and autophagy/chaperone systems, resulting in disturbed DNA repair and protein aggregation [6].

The clinical spectrum of anti ku positive disease varies from isolated myositis to multi organ manifestations like cardiac involvement or skin involvement, arthritis, and interstitial lung disease (ILD), which seem to differ among the associated connective tissue diseases [7-11].

Although, classified under the spectrum of idiopathic inflammatory myopathies, the pattern of weakness described is predominantly distal [7], though the literature suggests proximal muscle weakness also. The hallmark histopathological feature of anti-Ku myositis is myofibre necrosis with minimal inflammation, thereby mimicking immune mediated necrotising myopathies [6].

Anti Ku inflammatory myopathy has been predominantly seen in male aged between 20s to 50s [6,7], it has rarely been reported in the pediatric population, primarily in adolescents. Our case stands out as an early presentation in a young child, expanding the understanding of the age spectrum of this condition.

Diagnosing anti-Ku myositis in children requires a comprehensive approach, integrating radiological, serological parameters with clinical background. Our patient presented with symptoms consistent with inflammatory myopathies, which was confirmed with elevated creatinine kinase and MRI that showed altered signal changes, with a positive anti Ku antibody, thus a diagnosis Anti Ku inflammatory myopathy was made.

The treatment of anti-Ku myositis follows general principles applied to other inflammatory myopathies, with glucocorticoids serving as the cornerstone of therapy. The addition of steroid-sparing immunosuppressants such as methotrexate, azathioprine, or mycophenolate mofetil is often required for disease control and to minimize long-term corticosteroid toxicity [1].

6. Conclusion

This case represents one of the youngest reported presentations of anti-Ku myopathy. Despite the rare occurrence of Anti-Ku myositis in Pediatrics patients, the child responded well to immunosuppressive therapy, including pulse steroids and methotrexate, which resulted in significant clinical improvement.

The discussion underscores the importance of recognizing the rare presentation of inflammatory myopathies in children, as well as the role of targeted diagnostic testing, such as the Anti-Ku antibody panel and MRI.

This case expands our understanding of Anti-Ku inflammatory myopathy in the pediatric population and reinforces the critical role of early diagnosis and personalized treatment in achieving positive outcomes for rare conditions. Conflict of interest: None

Reference

1. McGrath ER, Doughty CT, Amato AA. Autoimmune Myopathies: Updates on Evaluation and Treatment. *Neurotherapeutics*. 2018 Oct;15(4):976–94.
2. Mimori T, Akizuki M, Yamagata H, Inada S, Yoshida S, Homma M. Characterization of a high molecular weight acidic nuclear protein recognized by autoantibodies in sera from patients with polymyositis-scleroderma overlap. *J Clin Invest*. 1981 Sep 1;68(3):611–20.
3. Belizna C, Henrion D, Beucher A, Lavigne C, Ghaali A, Lévesque H. Anti-Ku antibodies: Clinical, genetic and diagnostic insights. *Autoimmun Rev*. 2010 Aug;9(10):691–4.
4. Fell VL, Schild-Poulter C. The Ku heterodimer: Function in DNA repair and beyond. *Mutat Res Mutat Res*. 2015 Jan 1;763:15–29.
5. Involvement of the Ku autoantigen in the cellular response to DNA double-strand breaks. [Internet]. [cited 2025 Mar 22]. Available from: <https://www.pnas.org/doi/10.1073/pnas.91.16.7623>
6. Holzer MT, Uruha A, Roos A, Hentschel A, Schänzer A, Weis J, et al. Anti-Ku + myositis: an acquired inflammatory protein-aggregate myopathy. *Acta Neuropathol (Berl)*. 2024;148(1):6.
7. Casal-Dominguez M, Pinal-Fernandez I,

Derfoul A, Graf R, Michelle H, Albayda J, et al. The phenotype of myositis patients with anti-Ku autoantibodies. *Semin Arthritis Rheum*. 2021 Aug;51(4):728–34.

8. Cavazzana I, Ceribelli A, Quinzanini M, Scarsi M, Airò P, Cattaneo R, et al. Prevalence and clinical associations of anti-Ku antibodies in systemic autoimmune diseases. *Lupus*. 2008 Aug;17(8):727–32.

9. Rigolet A, Musset L, Dubourg O, Maisonobe T, Grenier P, Charuel JL, et al. Inflammatory Myopathies With Anti-Ku Antibodies: A Prognosis Dependent on

Associated Lung Disease. *Medicine (Baltimore)*. 2012 Mar;91(2):95.

10. Spielmann L, Séverac F, Meyer A. Response to: 'Anti-Ku syndrome with elevated CK: association with myocardial involvement in systemic sclerosis' by Campochiaro et al. *Ann Rheum Dis*. 2021 Jul 1;80(7):e114.

11. Spielmann L, Nespola B, Séverac F, Andres E, Kessler R, Guffroy A, et al. Anti-Ku syndrome with elevated CK and anti-Ku syndrome with anti-dsDNA are two distinct entities with different outcomes. *Ann Rheum Dis*. 2019 Aug 1;78(8):1101–6.