8° GIORNATA DELLO SPECIALIZZANDO IN NEUROLOGIA
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Necrotizing autoimmune myopathy revealing an undetected mitochondrial myopathy

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Case report

C.S., 13 y.o. boy

Referred to our center for severe muscle weakness (MRC 1-2), during a Mycoplasma Pneumoniae infection

No family history of neurological disease.

Medical history: mild fatigue from 6 months , respiratory tract infection

Blood Exams revealed extremely high CK levels with peack value 15 800 U/L (4 653 at the dimission).

Our neurological examination showed mild bilateral ptosis, proximal muscles weakness (3/5), scapular winging, hypotonia, waddling gait, absence of deep tendon reflexes and positive Gower's maneuver.

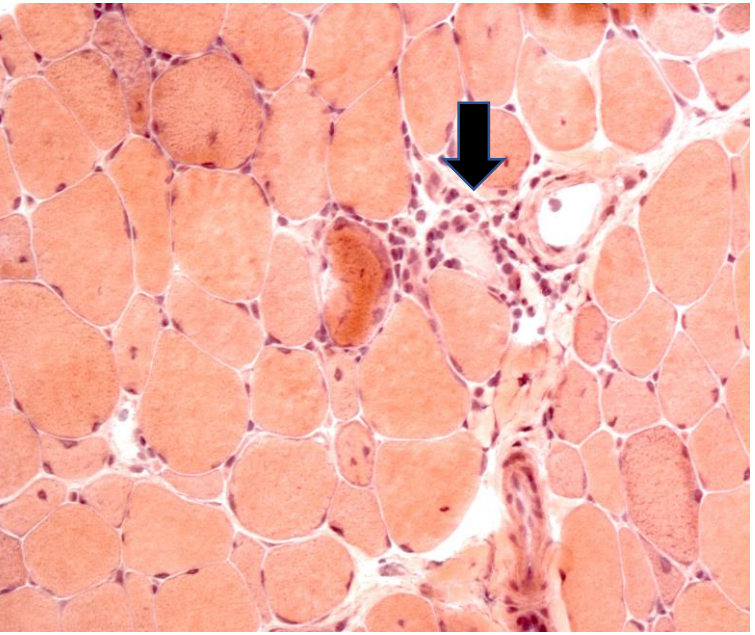
Investigation

EMG examination evidenced severe myopathic changes of proximal and distal muscles (deltoid, brachial biceps, quadriceps): fibrillation at rest and polyphasic potentials

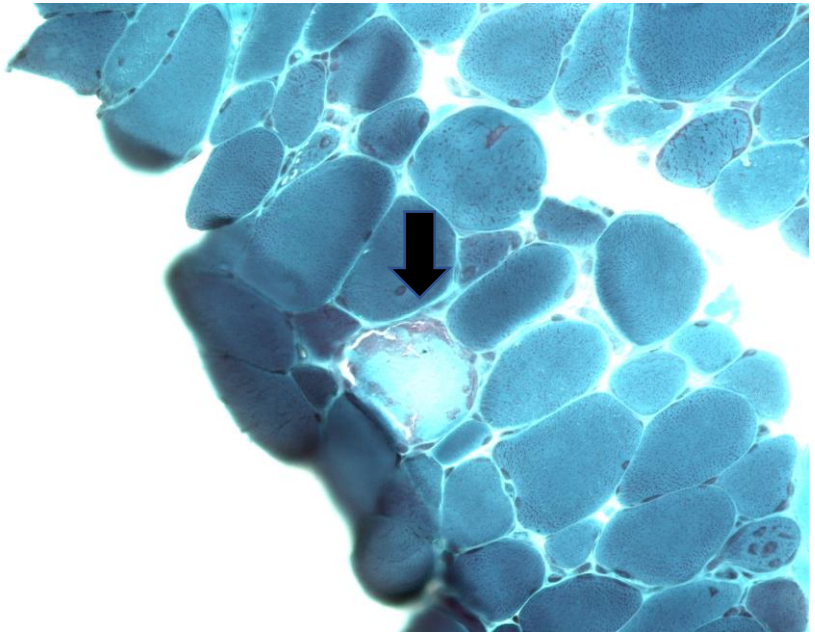
Myositis-Specific Antibodies (Jo-1, PL-7, PI-12, EJ, SRP, Mi-2, MDA-5, TIF1-gamma, SSA, SAE1, SAE2, NXP-2): negative

Screening for rheumatologic disease: negative

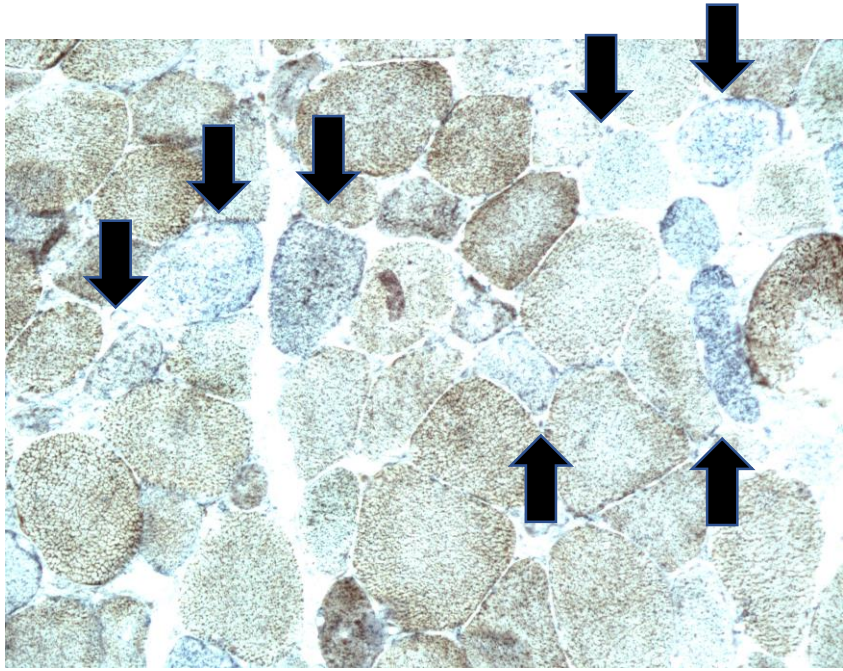
Muscle biopsy showed necrotic fibers and some inflammatory infiltrates, several ragged-red fibers and 20% COX-negative fibers.



H&E stain



Trichrome staining



Cox activity

Serum positivity for anti-3-hydroxy-3-methylglutaryl-coenzyme A reductase (HMGCR) antibodies

Immune-mediated-necrotizing-myopathy (IMNM)

Form of idiopathic inflammatory myopathy (IIM)

Association with Anti-HMGCR and Anti-SRP

Anti-HMGCR: typical of adult with a history of statin exposure

Other RF: malignancy, connective tissue disease , HIV. 50% idiopathic.

Young adults and children without statin exposure may also develope IMNM (phenotype like LGMD)

Typical Clinical Features of anti-HMGCR Myopathy	
Pattern of Weakness	Progressive, proximal weakness, especially posterior thigh, medial thigh, and gluteal compartments
Creatine Kinase	Usually >1,000-10,000 Units/L
Electromyography	Myopathic, usually with spontaneous activity in the form of fibrillations and sharp waves
Muscle Biopsy	Necrotizing myopathy; Increased MHC-1 or MAC staining
Muscle Imaging	T1 hyperintensity especially in posterior thigh; STIR signal is increased and may be asymmetric
Risk Factors	Statin drugs or supplements; HLA-DRB1*11:01 and 07:01 alleles

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Review

Anti-HMGCR Myopathy

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Rheumatology key messages

- Paediatric necrotizing myopathy with anti-3-hydroxy-3-methylglutaryl-coenzyme A reductase antibodies may not be rare.
- Autoantibody testing should be considered for paediatric patients suspected to have undiagnosed muscular dystrophy.
- Early diagnosis of paediatric necrotizing myopathy with anti-3-hydroxy-3-methylglutaryl-coenzyme A reductase antibodies is crucial for outcome.

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Original article

Pediatric necrotizing myopathy associated with anti-3-hydroxy-3-methylglutaryl-coenzyme A reductase antibodies

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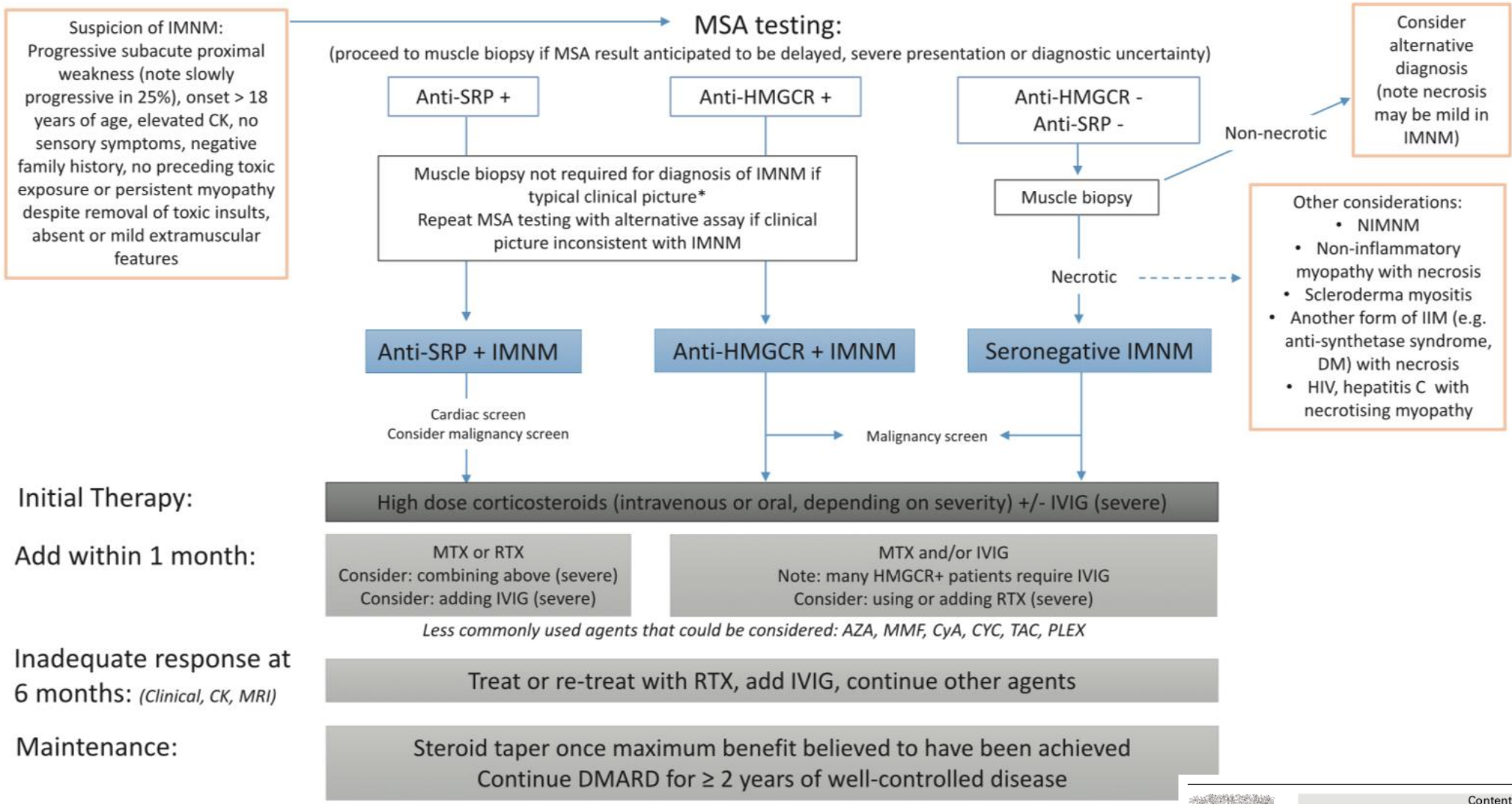

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Immune-mediated necrotising myopathy: A critical review of current concepts

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- High dose corticosteroid therapy (1mg/kg) with improvement at one month follow up:
    - MRC 4/5
    - CK levels 3 000 U/L
- Only IMNM??


THANKS FOR
WATCHING