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Sub-phenotyping of juvenile dermatomyositis: can it assist clinical decisions?

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Introduction

Juvenile Dermatomyositis (JDM) is a rare serious disease (affecting 2-3 million children/year) presenting with rash and proximal muscle weakness. Serious complications can include calcinosis, GI ulceration, interstitial lung disease (ILD) and even death. It is becoming clear that JDM is a heterogeneous condition. Dividing JDM into sub-phenotypes would allow better prediction of disease severity and more targeted treatments. We have identified novel auto-antibodies in subtypes of JDM that may correlate with specific phenotypes.

Objectives

To define clinical & pathological phenotypes of patients with JDM who have antibodies to Melanoma Differentiation-Associated protein 5 (MDA-5).

Methods

Patients

Patients were included from the Juvenile Dermatomyositis Cohort and Biomarker Study, a multi-centre study including 13 centres from across the UK. The study collects longitudinal clinical and serological data from patients with idiopathic inflammatory myopathies (IIM) of which 85% are diagnosed with JDM or JDM overlap features (currently n=446 patients). Clinical data collected included presence of clinical features, treatment, physicians global assessment and muscle strength assessments including the Childhood Muscle Assessment Score (CMAS).

Autoantibodies

Plasma or serum, available for 285 patients, were screened for the presence of autoantibodies by immunoprecipitation and confirmed by ELISA using recombinant MDA-5 protein.

Muscle biopsies

Muscle biopsies were stained and scored using the JDM Muscle biopsy score tool as described (1,2) .The validated muscle biopsy score tool measures severity of muscle pathology across 4 domains and with a separate visual assessment score (0-10).

Results

Autoantibody screening identified the presence of MDA-5 antibodies in 7.4% of patients (21/285 cases). MDA-5 positive patients had significantly increased incidence of ulceration ($p=0.03$), arthritis ($p<0.01$) and lung disease, yet had less severe muscle involvement, measured by CMAS score ($p=0.03$), than MDA-5 negative patients. In addition, median muscle biopsy scores for the MDA-5+ve patients were significantly lower than the MDA-5-ve patients ($p<0.005$) suggesting a less severe muscle pathology.

Conclusion

JDM is a heterogeneous condition with sub-phenotypes defined by autoantibody status, clinical features and muscle pathology. Identification and classification of sub-phenotypes could be used to predict disease course and severity. In the future, JDM specific autoantibodies could be used as biomarkers allowing for stratified approaches to treatment.

Disclosure of interest

None declared.

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