PEDIATRIC RHEUMATOLOGY

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PReS-FINAL-2284: SLE and complement deficiencies: a French multicentric retrospective study

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Introduction

Systemic lupus erythematosus (SLE) is a multifactorial disease. Rare causes of monogenic SLE have been described, including the complement deficiencies.

Objectives

Our objectives were to collect clinical data and outcome of SLE patients associated to complement deficiency in a multicenter retrospective study.

Methods

We conducted a retrospective study within the French paediatric rheumatology society (SOFREMIP) in 2012-2013.to identify patients, with a confirmed deficiency of complement fraction.

Results

Ten cases of SLE with complement deficiency were identified: 2 C1 deficiencies, 2 C2 deficiencies and 6 partial C4 deficiencies. The sex ratio (M/F) is 0/10. The disease onset occurred in childhood in 8 patients with 6 before the age of 10. The first symptoms were cutaneous in 7 children, articular for 2 children and psychiatric for 1 patient. All patients were positive for antinuclear antibodies whereas only half of them were positive for anti-dsDNA antibodies. Anti-Ro (SS-A) antibodies were strongly positive in 8 patients. Anti-phospholipidantibodies were presentin 6 patients. Over time, 5 patients developed a severe disease associated to renal failure (n = 2) or neurolupus (n = 3). Associated autoimmune diseases were found in 4 patients: hypothyroidism (n = 1), autoimmune hepatitis (n = 2), Sjögren syndrome (n = 1). Two children with C2 and C4 deficiency had severe bacterial infections.

Conclusion

Cutaneous or joint manifestations are the most common symptoms but life-threatening complications can occur in the context of C1 deficiency. Anti-SSA antibodies were frequent while anti-DNA are only found in half of the cases. Genetic characterization of complement deficiencies remains challenging. Next generation sequencing may be helpful to better diagnose these monogenic forms of lupus.

Disclosure of interest

None declared.

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