Documents

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Update on hereditary C1q deficiency: pathophysiology, clinical presentation, genotype and management
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Abstract
Purpose of review C1q deficiency is a rare inborn error of immunity characterized by susceptibility to severe infections and profound immune dysregulation, with a systemic lupus erythematosus-like phenotype. The management of patients with C1q deficiency is challenged by the rarity of this condition and the wide clinical variability. This review aims to emphasize the importance of a thorough immunological and clinical characterization to help guide a personalized and comprehensive approach to patients.Recent findingsWe focus on the concept of C1q deficiency as a bridge between the monogenic form of systemic lupus erythematosus and the Mendelian type I interferonopathies. Moreover, we explore the role of new treatment strategies such as Janus-associated kinase (JAK) inhibitors and allogeneic stem cell transplantation.SummaryIn this narrative review, we provide a systematic overview of C1q deficiency, starting with the description of the pathophysiological background and the variable clinical phenotype, and then exploring the different prognoses, the consequent treatment strategies and future directions.

Author Keywords
allogenic hematopoietic stem cell transplantation; C1q deficiency; Inborn Errors of Immunity; Mendelian type I interferonopathies; monogenic systemic lupus erythematosus

Index Keywords
antibiotic agent, antinuclear antibody, complement component C1q, complement component C1q antibody, disease modifying antirheumatic drug, double stranded DNA antibody, fresh frozen plasma, Haemophilus influenzae vaccine, immunosuppressive agent, Janus kinase inhibitor, meningitis vaccine, Pneumococcus vaccine, ribonucleoprotein antibody, Ro antibody, Sm antibody, complement component C1q, Janus kinase inhibitor; adaptive immunity, allogeneic hematopoietic stem cell transplantation, antibiotic prophylaxis, complement deficiency, disease severity, genetic disorder, Haemophilus influenzae infection, human, immunosuppressive treatment, infection risk, innate immunity, interferonopathy, loss of function mutation, Neisseriaceae infection, pneumococcal infection, Review, systemic lupus erythematosus, vaccination, animal, diagnosis, genetics, genotype, immunology, phenotype, therapy; Animals, Complement C1q, Genotype, Humans, Janus Kinase Inhibitors, Lupus Erythematosus, Systemic, Phenotype

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