عنوان مقاله:
Chédiak–Higashi syndrome

محل انتشار:
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خلاصه مقاله:
Chédiak-Higashi syndrome is a rare autosomal recessive congenital immunodeficiency mainly characterized by a condition called oculo-cutaneous albinism. The affected subjects have light-colored hair, vision problems, blood clotting (coagulation) abnormalities and in adulthood varying neurologic disorders. Recurrent infections, particularly viral infection with other disorders in childhood are usually life threatening. It has demonstrated mutations throughout the CHS١/LYST gene. The nature of the mutation can be a predictor of the severity of the disease. The current therapeutic options are: Antibiotics, chemotherapy and bone marrow transplantation. This review will discuss the clinical and molecular aspects of this syndrome for better understanding of the factors that may cause abnormalities.

کلمات کلیدی:
Chediak-Higashi syndrome, Albinism, Immunodeficiency