Cytophagic Histiocytic Panniculitis with Fatal Haemophagocytic Lymphohistiocytosis in a Paediatric Patient with Perforin Gene Mutation

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摘要

Abstract

Cytophagic histiocytic panniculitis (CHP), or histiocytic cytophagic panniculitis, is a rare form of panniculitis associated with haemorrhagic diathesis and histiocytic lymphohistiocytosis (HLH), initially described in 1980 as a benign lymphoproliferative disease.1 In 1991 Gonzalez et al reported a unique entity of subcutaneous T-cell lymphoma with haemophagocytosis, later designated as subcutaneous panniculitis-like T-cell lymphoma (SPTL).2 Marzano et al suggested that CHP and SPTL might span a clinicopathological spectrum in which there is a natural progression from CHP to SPTL.3 HLH is a clinical syndrome of immune deregulation with hypercytokinaemia causing dysfunctions of various organs and a high mortality. Familial HLH (FHL) is associated with several hereditary defects. Stepp et al first showed that the mutation in perforin gene (PRF1) at chromosome 10q21 was responsible for 20 – 40% of FHL patients.4