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Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 59597

Manuscript Type: CASE REPORT

Adult onset type 2 familial hemophagocytic lymphohistiocytosis with *PRF1*
 c.65delC/c.163C>T compound heterozygous mutations: A case report

Liu XY *et al.* Adult onset FHL2

Xin-Yi Liu, Yan-Bo Nie, Xue-Jing Chen, Xiao-Hui Gao, Li-Jia Zhai, Feng-Ling Min

Abstract

BACKGROUND

Familial hemophagocytic lymphohistiocytosis (FHL) ¹ is a primary immunodeficiency disease caused by gene defects. The onset of FHL in adolescents and adults may lead clinicians to ignore or even misdiagnose the disease. To our knowledge, we are the first to describe in detail the clinical features of type 2 FHL (FHL2) with compound



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Genetic testing identified compound heterozygous mutations in PRF1 (Patients 1 and 2) and UNC13D (Patient 3), with no evidence of systemic disease except decreased NK-cell function.

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