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Genetic variant spectrum in 265 Chinese patients with hemophagocytic lymphohistiocytosis: Molecular analysis of *PRF1*, *UNC13D*, *STX11*, *STXBP2*, *SH2D1A* and *XIAP*

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Hemophagocytic lymphohistiocytosis (HLH) is a rare life-threatening hyperinflammatory disease. This study aimed to investigate the frequencies and distributions of inherited variants in *PRF1*, *UNC13D*, *STX11*, *STXBP2*, *SH2D1A* and *XIAP* genes in Chinese patients with HLH. A total of 265 patients diagnosed with HLH from January 2010 to December 2016 were recruited and analyzed for the six genes. Genetic variants were observed in 87 (32.83%) patients. 36 (13.58%) exhibited variants in *UNC13D*, 18 (6.79%) exhibited *PRF1* variants, 10 (3.77%) had variants in *XIAP*, 9 (3.40%) exhibited variants in *STXBP2*, 6 (2.26%) carried variants in *SH2D1A*, 1 (0.38%) had *STX11* variant, and 7 (2.64%) exhibited digenic variants. Monoallelic variants were the most common, which accounted for 49.43% of all cases with variants. All variants were confirmed to be germline-derived. The present study describes a distinct variant spectrum in Chinese patients with HLH, whereby *UNC13D* is the most frequently mutated gene with missense variants that are the most common molecular defects. The variant profile of Chinese HLH patients is quite different from that of Western cohorts but similar to that of Korean patients, yet showing its own uniqueness. This racial difference shows the role of genetic background in the occurrence of HLH.

Biography

Dongchu Wang, having received a BSc Hons Biomedical Science by doing human genome project, became interested in the producing, characterizing, and tracking recombinant protein for pharmaceutical purpose as part of a Master's degree at the University of Greenwich. During his PhD, he focused on anti-sense drug intracellular trafficking via endocytosis at Greenwich. Since 2017, he has been pursuing his interests combining molecular biology and immunology with the intention of developing new and more efficient immunotherapy for leukemia in Lu Daopei Hospital.

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