

第151回 北大小児科 特別集談会

2022年5月18日(水)

『My journey from being a fresh graduated medical student to an assistant professor: what I did and will do』

北大小児科 助教

Shimaa Said Mohamed Ali Abdrabou

Summary

【Research part】

My research focuses on the diagnosis and analyzing the pathophysiology of the inborn errors of immunity including XIAP deficiency, SAP deficiency, HFM, WASP, NEMO, STAT3.etc.

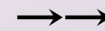
X-linked lymphoproliferative disease (XLP) is a rare primary immunodeficiency disorder characterized by its distinctive susceptibility to hemophagocytic lymphohistiocytosis (HLH) that is often triggered by EBV infection. Mutations in *SH2DIA* gene encoding *SLAM* associated protein (SAP) form XLP1 while mutations in X-linked inhibitor of apoptosis protein (XIAP) form XLP2. In this study, four unrelated Japanese patients were diagnosed with XIAP deficiency. In addition, two brothers were identified with SAP deficiency based on absent XIAP and SAP expression in T-cell blasts. All of the patients eventually underwent hematopoietic stem cell transplantation, leading to a complete or partial remission of the disease.

【Teaching part】

We will soon launch Medical English club to exchange our knowledge and improve our English language skills, so please stay alert and try to join us!

18:00~19:00 (予定) Web (Cisco Webex) にて開催

どなたでもお気軽にご参加下さい！



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