Omenn Syndrome—Review of Several Phenotypes of Omenn Syndrome and RAG1/RAG2 Mutations in Japan

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Omenn syndrome (OS) is a form of severe combined immunodeficiency (SCID) characterized by erythrodermia, hepatosplenomegaly, lymphadenopathy, and alopecia. In patients with OS, B cells are mostly absent, T-cell counts are normal to elevated, and T cells are frequently activated and express a restricted T-cell receptor (TCR) repertoire. Thus far, inherited hypomorphic mutations of the recombination activating genes either 1 or 2 (RAG1/2) have been detected in most OS patients. We have recently experienced a rare case of OS showing the revertant mosaicism due to multiple second-site mutations leading to typical OS clinical features with RAG1-deficient SCID. In this review, we will focus on the variation of several phenotypes of OS.

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