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Hypomorphic RAG1 Deficiency

National Cancer Institute

Source

National Cancer Institute. *Hypomorphic RAG1 Deficiency*. NCI Thesaurus. Code C126338.

A condition of decreased or absent presence or activity of V(D)J recombination-activating protein 1. Deficiency of this protein is associated with Omenn syndrome, severe combined immunodeficiency, b cell-negative, combined cellular and humoral immune defects with granulomas, and alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity.