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Autoimmune Polyglandular Syndrome Type 1

National Cancer Institute

Source

National Cancer Institute. *Autoimmune Polyglandular Syndrome Type 1*. NCI Thesaurus. Code C129727.

Autoimmune polyglandular syndrome caused by homozygous, compound heterozygous, or heterozygous mutation(s) in the AIRE gene, encoding autoimmune regulator protein. Diagnosis requires at least 2 of the 3 major clinical features: chronic mucocutaneous candidiasis, primary adrenal insufficiency, or primary hypoparathyroidism. Antibodies against type 1 interferons and interleukin 17 cytokines are almost always present. Heterozygous AIRE mutation(s) typically result in a narrower disease spectrum.