Immune Dysregulation, Polyendocrinopathy, and Enteropathy X-Linked Syndrome

National Cancer Institute

Source

National Cancer Institute. Immune Dysregulation, Polyendocrinopathy, and Enteropathy X-Linked Syndrome. NCI Thesaurus. Code C131009.

An X-linked recessive autoimmune condition caused by mutation(s) in the FOXP3 gene, encoding the forkhead box P3 transcription factor. The condition is characterized by infantile onset of severe diarrhea due to enteropathy, type 1 diabetes mellitus, and dermatitis. Associated features may include hypothyroidism, autoimmune hemolytic anemia, thrombocytopenia, lymphadenopathy, hepatitis, and nephritis. The condition is usually fatal before age 2 years if not treated with bone marrow transplantation.