

Open Peer Review on Qeios

Autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

<u>Autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections</u>

<u>syndrome</u>. ORPHA:391487

Autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome is an extremely rare, autosomal dominant immunological disorder characterized by variable enteropathy, endocrine disorders (e.g. type 1 diabetes mellitus, hypothyroidism), immune dysregulation with pulmonary and blood-borne bacterial infections, and fungal infections (chronic mucocutaneous candidiasis) developing in infancy. Other manifestations include short stature, eczema, hepatosplenomegaly, delayed puberty, and osteoporosis/osteopenia.

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