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Early-onset refractory diarrhea of ¹immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome associated with a novel mutation in the *FOXP3* gene: A case report

Na Su, Cheng Chen, Xia Zhou, Guo-Da Ma, Ri-Ling Chen, Chuan Tian

Abstract

BACKGROUND

Immune dysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome is a rare X-linked recessive disease caused by mutations in the *forkhead box protein 3 (FOXP3)* gene, which is a master transcriptional

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Introduction. The **immune dysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome** is a rare disorder characterized by dysfunction of the regulatory T H 1 lymphocytes (Treg) that causes **immune dysregulation** leading to autoimmune diseases. It is an x-linked recessive disorder caused by mutations of the forkhead box protein 3 (**FOXP3**) **gene** that is located in the short arm of ...

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The best characterized of these is **Immune Dysregulation, Polyendocrinopathy, enteropathy, X-linked (IPEX)**, resulting from mutations affecting **FOXP3**. A number of other **gene** defects that affect T R cell function also give rise to IPEX-related phenotypes, including loss of function mutations in CD25 and STAT5b and ITCH.

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Immune Dysregulation, Polyendocrinopathy, Enteropathy, X ...

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The **immune dysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome** is a rare, x-linked, recessive disorder characterized by dysfunction of the T regulatory (Treg) lymphocytes leading to autoimmune diseases. Herein we **report** a male patient with IPEX **syndrome** who presented with severe **diarrhea**, eczema, and malabsorption leading to failure to thrive and necessitating total ...

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Immune dysregulation

Immune dysregulation is any proposed or confirmed breakdown or maladaptive change in molecular control of immune system processes. For example, dysregulation is a component in the pathogeneses of autoimmune diseases and some cancers, to the extent that the pathophysiology is understood to date. Immune system dysfunction, as seen in IPEX syndrome leads to immune dysfunction, polyendocrinopathy, enteropathy, X-linked. IPEX typically presents during the first few months of life with diabetes mellitus, intractable diarrhea, failure to thrive, eczema, and hemolytic anemia. unrestrained or unregulated immune response.

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