# X-linked immunodeficiency, polyendocrinopathy, enteropathy(IPEX)

## GENERAL INFORMATION

### Description:

IPEX, an X-linked syndrome is a rare disorder occurring in boys. The syndrome is caused by a mutation in FOXP3, a putative DNA-binding protein. This protein has significant homology to forkhead/winged-helix transcription factor family. Most mutations of FOXP3 are localized in the carboxyl-terminal forkhead DNA binding domain.

### Alternative names:

- IPEX
- X-linked autoimmunity-allergic dysregulation syndrome, XLAAD
- IDDM-secretory diarrhea syndrome, DMSD
- Autoimmunity-immunodeficiency syndrome
- Diarrhea, polyendocrinopathy, fatal infection syndrome
- Enteropathy, autoimmune, with hemolytic anemia and polyendocrinopathy
- Polyendocrinopathy, immune dysfunction, and diarrhea; XPID
- Diabetes mellitus, congenital insulin-dependent, with fatal secretory diarrhea
- Immunodeficiency, polyendocrinopathy, and enteropathy, formerly absence of islets of Langerhans

### Classification:

- Other well-defined immunodeficiency syndromes
  - Autoimmune disorders

### Inheritance:

X-linked

### OMIM:

- #304790 Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked; IPEX
- *300292 Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked; IPEX

### Incidence:

Incidence is not known yet.

## CLINICAL INFORMATION

### Description:

Patients have protracted diarrhea, ichthyosiform dermatitis, early-onset insulin-dependent miellitus, thyroiditis, hemolytic anemia, variable autoimmune phenomena and infections. It frequently results in death during infancy or childhood.

### Diagnosis:

### Diagnostic laboratories:

**Clinical:**

- Immune dysregulation, polyendocrinopathy, enteropathy, X-linked, ORPHANET
- T-cell disorders, eMedicine

**Genetic:**

- FOXP3, IDdiagnostics
- Centro di Genetica Umana - Ente Ospedaliero Ospedale Galliera (Genova), EDDNAL

## Therapeutic options:

- Supportive therapy with total parenteral nutrition, insulin and blood transfusions is beneficial. Allogenic bone marrow transplantation from an HLA-identical person might be a curative option, even in the case of complications.
- T-cell disorders, eMedicine

## Research programs, clinical trials:

- Italian IPEX study group
- European Initiative for Primary Immunodeficiencies

# GENE INFORMATION

## Names:

**HUGO name:** FOXP3
**Alias(es):** AIID, DIETER, IPEX, PIDX, XPID, SCURFIN, forkhead box P3 1

## Localization:

**Chromosomal Location:**

Xp11.23-q13.3

**Maps:**

FOXP3 (Map View)

## Variations / Mutations:

- FOXP3base: Mutation registry for Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked; IPEX; IDbases

## Other gene-based resources:

Ensembl: ENSG00000049768, GENATLAS: FOXP3, GeneCard: FOXP3, UniGene: 247700, Entrez Gene: 50943, euGenes: 50943, GDB: 10796361

# PROTEIN INFORMATION

## Description:

## Other features:

## Expression pattern for human:

| Tissue | Exp. (%) | Clones |
|---|---|---|
| mixed | 64.64 | 1:60341 |
| stomach | 35.36 | 1:110283 |

# OTHER RESOURCES

## Societies:

**General:**

- International Patient Organization for Primary Immunodeficiencies
- Immune Deficiency Foundation
- European Society for Immunodeficiencies
- NIH/National Institute of Allergy and Infectious Diseases