IPEX Syndrome

PLAY PICMONIC

Characteristic

X-Linked
X-suit
IPEX syndrome is inherited in an X-linked fashion.

Onset First Year of Life
On-switch First-place
The onset of IPEX syndrome occurs in the first year of life. The syndrome presents with the clinical triad of endocrinopathy, enteropathy, and dermatitis

Immune Dysregulation

Regulatory T Cells Dysfunction
Referee Tennis-ball Dysfunctioning
Regulatory T cells potentiate immune suppression, which plays a crucial role in immune homeostasis. IPEX syndrome is characterized by the dysfunction of regulatory T cells resulting in autoimmunity.

Deficiency of FOXP3
Deficient FOX-P (3) Tree
IPEX syndrome is caused by a mutation of the transcription factor FOXP3 which is important for the function of regulatory T cells. FOXP3 plays a role in the differentiation of CD4+ Tregs. The high level of FOXP3 expression results in a suppressive capacity of Tregs. Deficiency of FOXP3 will cause autoimmunity.

Autoimmune
Auto-in-moon
The dysregulation of the immune in IPEX syndrome causes autoimmune disease and allergic inflammation.

Polyendocrinopathy

Endocrinopathy
Endocrine-glands-path
The hallmark of IPEX is the early onset of autoimmune endocrinopathies, typically manifest as diabetes or thyroiditis.

Type I Diabetes Mellitus
(1) Wand Dyed-bead-pancreas
IPEX syndrome is associated with diabetes in male infants. Diabetes mellitus type I is the most common endocrinopathy. Hyperglycemia may be seen at birth due to immune-mediated destruction of islet cells.

Enteropathy

Enteropathy
Intestine-path
Enteropathy is a common manifestation seen in infants with IPEX syndrome. It can cause failure to thrive. Manifestations may include intractable diarrhea, gastritis, vomiting, ulcerative colitis, celiac disease, and ileus.

Severe Diarrhea
Severed Toilet
Enteropathy can be featured as severe diarrhea with mucoid or blood. It can be aggravated by a diet change from breastfeeding to formula. This symptom can be life-threatening in patients.

Other Features

Dermatitis
Dermatologist-examining-rash
Dermatitis in IPEX syndrome manifests as an eczematous rash (mainly atopic dermatitis) that occurs in early infancy. Other manifestations may include alopecia, cheilitis, and onychodystrophy.

Nail Dystrophy
Nail Disc-trophy
An abnormal change in color, texture, shape, and growth of the fingernails or toenails characterizes nail dystrophy. This feature can also be seen in patients with IPEX syndrome.

Treatment

Bone Marrow Transplantation
Bone Train-plant
Patients with IPEX syndrome will need to be treated with supportive care. The standard treatment for this disorder is bone marrow transplantation, and if left untreated, it's often fatal.
