Immune dysregulation, polyendocrinopathy, enteropathy, x-linked (IPEX) syndrome: A case report

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Background:

IPEX syndrome is a rare, fatal x-linked immune dysregulatory disorder. The key features are refractory diarrhea, eczematous dermatitis, and autoimmune endocrinopathies which usually include early-onset insulin-dependent diabetes mellitus (DM type1), thyroiditis, or both. IPEX syndrome is caused by mutation of the forkhead box protein 3 gene (FOXP3), a key regulator of immune tolerance.

Aims:

To remind the pediatrician about IPEX syndrome which is a rare disease that cause intractable diarrhea and autoimmune endocrinopathies.

Case outline:

The Thai male infant was born with a birth weight of 3,020 grams. At the age of 10 days, he developed acute diarrhea with severe dehydration. The initial blood sugar was 1,064 mg/dl, urine glucose 4+, and positive serum ketone. Diabetic ketoacidosis was diagnosed and he was treated with intravenous regular insulin. He was referred to QSNICH for investigation about the cause of chronic diarrhea and DM type 1. The infant was found to have autoimmune thyroiditis (positive antithyroglobulin antibody 1:346, positive antimicrosomal antibody 1:213) with hypothyroidism. Other findings showed high plasma IgE level with IgG, IgA, IgM within normal limit. ANA and Anti DNA were negative while anti smooth muscle antibody was positive and anti goblet cell IgG were positive (1:100). Upper endoscopy was done and cytomegalovirus duodenitis diagnosed. He was treated with gancyclovir but his symptoms did not improve. IPEX syndrome was diagnosed from classic clinical manifestrations of refractory diarrhea, autoimmune thyroiditis, and DM type 1. DNA testing confirmed IVS6-1G>C mutations in the FOXP3 gene. Insulin therapy, combined with parenteral and amino acid formula were given with no improvement. The infant succumbed to Candida albicans septicemia at 6 months of age.

Conclusion:

Evaluation for IPEX syndrome should be considered in cases of male infants presenting with intractable diarrhea and autoimmune endocrinopathies. The diagnosis is confirmed by genetic analysis of FOXP3 gene.

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