INTRODUCTION

We describe a family in which parents had consanguinity, being children of real sisters. They had given birth to five children. In their family, children remained healthy from birth to pre-school age and then started having symptoms around the age of 5 years and two of them succumbed to this illness. Polyglandular autoimmune syndrome Type-1 is a rare sporadic autosomal recessive disease. It is characterized by the existence of two or more endocrinial disorders. Patients may require lifelong hormone replacement therapy for survival.

Key words: Autoimmune, hypoparathyroidism, moniliasis, polyglandular, primary adrenal insufficiency

ABSTRACT

We describe a family in which parents had consanguinity, being children of real sisters. They had given birth to five children. In their family, children remained healthy from birth to pre-school age and then started having symptoms around the age of 5 years and two of them succumbed to this illness. Polyglandular autoimmune syndrome Type-1 is a rare sporadic autosomal recessive disease. It is characterized by the existence of two or more endocrinial disorders. Patients may require lifelong hormone replacement therapy for survival.

Key words: Autoimmune, hypoparathyroidism, moniliasis, polyglandular, primary adrenal insufficiency

Failure of siblings to thrive beyond 5 years of age

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until date. Fifth child, a daughter of around 3 months is also healthy until date.

**INVESTIGATIONS**

**Child 1 - Other Investigations**
- Bilirubin - 2.5 mg/dl, serum glutamic oxaloacetic transaminase - 109 IU/L, serum glutamic-pyruvic transaminase - 40.1 IU/L.
- **Urea** - 133.8 mg/dl, Serum creatinine - 1.43 mg/dl
- **Hb%** - 9.2 g/dl, mean corpuscular volume - 89.3 fl, platelets - 49,000, C-reactive protein - 1.2 mg/L
- Contrast enhanced computed tomography head - Basal ganglia calcification seen.

**Child 2 - Index case**
- Thyroid profile - normal
- Hb% - 11.6 gram%
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- MCV - 73.2 fl
- Scraping of the tongue on potassium hydroxide mount showed fungal filaments [Figure 3]
- Blood sugar levels were normal
- Spleen normal, ear examination normal and no evidence of Otosclerosis
- Fundus was normal.

**DISCUSSION**

Polyglandular autoimmune syndrome Type-1 (PGA 1)\(^\text{[1]}\) is a rare autosomal recessive disorder, also known as Type I autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy or Whitaker syndrome is also referred as HAM (Hypoparathyroidism, Addison's disease, and Monalisis) syndrome. A single gene mutation of autoimmune regulator,\(^\text{[2]}\) which codes for a transcription factor featuring two zinc motifs, is believed to be likely pathogenic for PGA-I. PGA\(^{[1]}\) has found increased correlation with human leukocyte antigen (HLA)-A28 and HLA-A3. People with PGA-1 have at least two of the disease’s main symptoms: Fungal infections of the skin and mucous membranes, decreased function in the parathyroid glands and decreased function in the adrenal glands (primary adrenal insufficiency). Many people with the disease have all three main symptoms. Our index case of PGA \(^{[1]}\) also presented with oral moniliasis, Features of hypoparathyroidism in the form of muscle spasm, tetany, weakness, with features of hypoadrenalism as salt craving, lethargy (hypoandrogen-metabolic syndrome). On treatment case improved and is living a fruitful life.

From this experience, we have learnt that, 1) In diagnosing PGA-1 history of consanguity is important. 2) In young patients presenting with convulsions, hypocalcemia should be kept as differential (hypoparathyroidism). 3) In patients of seizures, CT finding of Basal Ganglion calcification should alert us to look for Hypoparathyroidism. 4) In hypocalcaemic seizures using Dilantin/phenobarbitone may actually worsen the seizures because they have Antivitamin D effect.

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