

Case Report

## A child with Polyglandular autoimmune syndrome Type-I and immune thrombocytopenic purpura

Mohamad Pedram<sup>1</sup>, Korush Riahi<sup>2</sup>, Kaveh Jaseb<sup>3</sup>,  
Mohammad Hasan Alemzadeh Ansari<sup>4</sup>, Mohammad Javad Alemzadeh Ansari<sup>5</sup>

### ABSTRACT

Polyglandular autoimmune syndrome type I (PGA I) is a rare disease. Its hallmarks are chronic mucocutaneous candidiasis, hypoparathyroidism and adrenal insufficiency. Immune thrombocytopenic purpura (ITP) is one of the most common autoimmune disease in children. Association of PGA I with ITP was not found in some previous studies, but, we report a child with PGA I and ITP.

**KEY WORDS:** ITP, Polyglandular autoimmune syndrome.

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### INTRODUCTION

Polyglandular autoimmune syndrome type I (PGA I) is a monogenic autosomal disease with a recessive inheritance pattern. It is characterized by a widely variable combination of autoimmune destruction of tissues (predominantly endocrine glands), chronic superficial candidiasis, and ectodermal dystrophy complex including hypoplasia of the dental enamel, alopecia and nail dystrophy.<sup>1,2</sup> Although, immune thrombocytopenic purpura (ITP) had been reported in PGA II and III,<sup>3</sup> but in previous studies we did not find any article reporting PGA I with ITP. Here, we report a 5 years old boy who presented with a clinical picture of PGA I and ITP.

### CASE REPORT

A five years old boy was admitted to our hospital with a four months history of multiple ecchymosis on the lower limbs. This ecchymosis occurred after mild injuries. On physical examination he had alopecia of head and neck and chronic mucocutaneous candidiasis. (Figure-1,2) Other physical examination were unremarkable.

Routin laboratory examination revealed platelets 7 4000 /mm<sup>3</sup> (normal range 150 000-450 000 /mm<sup>3</sup>), Partial thromboplastin time 41 sec (normal range 30-45 sec), Prothrombin time 13.7 sec (normal range

1. Mohamad Pedram,  
Professor Pediatric Oncologist,  
Research Center of Thalassemia & Hemoglobinopathy,  
Shafa Hospital,  
Jundishapour University of Medical Sciences,  
Ahvaz, Iran.
  2. Korush Riahi,  
Pediatric Endocrinologist,  
Golestan Hospital.
  3. Kaveh Jaseb,  
Pediatric Hematology/Oncology Fellowship,  
Shafa Hospital.
  4. Mohammad Hasan Alemzadeh Ansari,  
Medical Student,  
Faculty of Medicine.
  5. Mohammad Javad Alemzadeh Ansari,  
Resident Cardiology, Tehran Heart Center Hospital,  
Tehran University of Medical Sciences, Tehran - Iran.
- 1-4: Jundishapour University of Medical Sciences,  
Ahvaz, Iran.

Correspondence:

Mohamad Pedram,  
Professor Pediatric Oncologist,  
Research Center of Thalassemia & Hemoglobinopathy,  
Shafa Hospital,  
Jundishapour University of Medical Sciences,  
Ahvaz, Iran.  
E-mail: m\_pedram\_2007@yahoo.com

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Figure-1: Muccocuranus Candidiasis Hyperpigmantation.

11-13 sec), and international normalized ratio 1.4. Other examination indicated serum calcium 5 mg/dl (normal range 8.5-10.5 mg/dl), phosphate 12 mg/dl (normal range 3-6 mg/dl), serum albumin 5 (normal range 4-6 g/dl). Serum cratinin, protein and electrolytes was normal. Parathormone hormone was 3.1 pg/dl (normal range 15-65 pg/dl). T3, TSH, T4, FTI and T3RU were in normal range but anti-TPO was 167.8 Iu/dl (normal range <34 Iu/dl). Serology examinations was negative for anti-nuclear antibodies and LE cell.

Electrocardiogram indicated high corrected QT interval (0.52 sec). He was diagnosed to have PGA I based on presence of hypoparathyroidism and chronic mucocutaneous candidiasis. For approach to thrombocytopenia, bone marrow aspiration was done and its finding was compatible with ITP. After diagnosis of ITP, he was treated by Methylprednisolone 350 mg for five days. After five days the platelets reached to normal range (375 000 /mm<sup>3</sup>).

### DISCUSSION

PGA I has been described under other names, such as Whitaker's syndrome, polyglandular autoimmune disease type 1, or autoimmune polyendocrinopathy, candidosis, ectodermal dystrophy.<sup>4</sup>

PGA I is an autoimmune syndrome with characteristic disease associations that often appear early in life, typically in infants with persistent chronic mucocutaneous candidiasis without the systemic infection generally associated with severe immunodeficiency. The diagnosis of autoimmune polyendocrine syndrome type I is usually made later, when hypocalcemia due to hypoparathyroidism



Figure-2: Alopecia

develops or Addison's disease is recognized in a young child. The syndrome is rare but has an increased prevalence in certain populations (e.g., inhabitants of Finland<sup>5</sup> and Sardinia and Iranian Jews).<sup>6</sup> Mutations in an autoimmune-suppressor gene (AIRE, for autoimmune regulator), which encodes a transcription factor, cause the syndrome. Persons with any two of several specific conditions - mucocutaneous candidiasis, hypoparathyroidism, and Addison's disease- almost always have AIRE mutations.<sup>7,8</sup>

After diagnosis, patients with autoimmune polyendocrine syndrome type I require close monitoring. Monitoring can help prevent illness associated with delayed diagnosis of additional autoimmune diseases such as Addison's disease and hypoparathyroidism, as well as oral cancer, which may develop if candidiasis is not treated aggressively, and infection due to asplenicism, which is present in a subgroup of patients.<sup>9</sup> Our patient was diagnosed PGA I because this manifestations: hypoparathyroid, chronic mucocutaneous candidiasis.

Chronic mucocutaneous candidiasis generally presents earliest in life and is the most frequent of the three main diseases of PGA I. It can appear as early as at the first month after birth up to 21 years of age, with a peak of occurrence in early childhood. Chronic mucocutaneous candidiasis is present in 73-100% of all patients.<sup>4</sup>

Chronic hypoparathyroidism is the first endocrine disease to occur during the time course of PGA I,

usually after chronic mucocutaneous candidiasis and before Addison's disease, and can present between three months to 44 years of age (mean, 7.5 yr). Chronic hypoparathyroidism has been reported in 73–90% of the cases of PGA I.<sup>4</sup>

ITP is one of the most common autoimmune disease in children. Acute ITP occurs in 90% of the children. It is generally seen in children from the ages of one to nine years, though the peak manifestation is between two to five years of age. The diagnosis of ITP was demonstrated by bone marrow aspiration. A treatment option is corticosteroids<sup>5</sup>. Because our patient had multiple echemosis and thrombocytopenia, bone marrow aspiration was done. The result of bone marrow aspiration confirmed the ITP. After treatment with Methylprednisolone 350 mg, the platelets increased to normal range.

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