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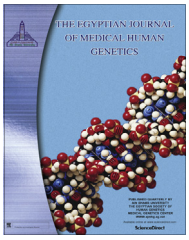

CASE REPORT

Christ-Siemens-Touraine syndrome with cleft palate, absent nipples, gallstones and mild mental retardation in an Egyptian child

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KEYWORDS

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Abstract We report a 6 year old child, second in order of birth of non consanguineous Egyptian parents with typical characteristics of Christ-Siemens-Touraine syndrome. The patient had sparse light hair over the scalp, scanty eyebrows and eyelashes, a high arched cleft palate, decayed oligodontic teeth, hyperpigmentation all over the body more marked in cheeks, perioral area, chin, neck, axillae and back of both knees, fragile dry skin, hyperextensibility of the metacarpophalangeal joints, dysplastic nails and absent nipples. Our patient had also gallstones which were not reported previously and mild mental retardation. His mother had mild teeth abnormalities.

1. Introduction

Ectodermal dysplasia (ED) syndromes comprise a group of genetic disorders characterized by deficient function of at least 2 ectodermal derivatives such as skin, hair, teeth and sweat glands [1,2]. Although more than 170 different subtypes of ectodermal dysplasia have been identified, these disorders are considered to be relatively rare with an estimated incidence of 1 case per 100,000 [3,4].

Ectodermal dysplasia is divided into two types based on the number and function of sweat glands: hidrotic ectodermal dysplasia (Clouston syndrome) and hypohidrotic (anhidrotic) ectodermal dysplasia (HED) (Christ-Siemens-Touraine syndrome) [5].

The Christ-Siemens-Touraine syndrome (CST syndrome) is the rare an- or hypohidrotic form of the ectodermal dysplasia [6]. It was first described in 1848 by Thurnam [7] and later by Darwin [8]. In 1913, Christ characterized it as a congenital ectodermal defect, Siemens confirmed the X-linked nature of inheritance in 1921 and in 1936, and Touraine published works on the wide range of features. It is characterized by sparse hair, heat intolerance, excessively dry skin due to the absence of sweat glands and abnormal spiky or absent teeth [9].

We report a case with the typical features of the CST syndrome who had in addition some unreported features after taking the consent of parents.

2. Case report

We report a 6 year old male child, second in order of birth of healthy non consanguineous Egyptian parents. The patient was delivered at full term by cesarean section. His birth weight was 3 kg. No problems were noted by the mother during

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pregnancy. The patient was referred to the Genetics Clinic, Pediatric Hospital, Ain Shams University complaining of abnormal features since birth.

The patient was admitted to neonatal intensive care unit (NICU) since birth for meconium aspiration. He had scaling of the skin during the neonatal period which was treated with moisturizing creams. The mother noticed that her son had scanty hair, cleft palate, tie tongue, abnormalities of nails and teeth. His mother also noticed that her child had recurrent episodes of unexplained hyperpyrexia; he was not able to sweat, and she had to protect him from overheating during warm weather.

At the age of 3 months, he had tongue-tie surgery. At the age of 8 months he was admitted to hospital for gastroenteritis. He had an attack of gastroenteritis every month for 8 months. He had cleft palate repair operation at the age of 1 year. At the age of 2 years he had gastroenteritis and dehydration which necessitated admission to hospital. During admission, multiple gall bladder stones were discovered on abdominal ultrasound and he had a cholecystectomy operation. Then he was referred to our genetics clinic.

The patient had mild mental retardation. Family history was unremarkable. He had two healthy sibs.

On examination, his weight was 20 kg (50th percentiles), his height was 114 cm (50th percentiles), and his skull circumference was 52 cm (75th percentiles).

He had sparse, thin hair over the scalp, scanty eyebrows and eyelashes, frontal bossing, prominent supraorbital ridges, depressed nasal bridge, saddle nose, dry lips, angular stomatitis, high arched palate and decayed oligodontic teeth (Figs. 1 and 2).

He had absent nipples and hyperpigmentation all over the body mainly in cheeks, perioral area, chin, neck, axillae and the back of both knees (Figs. 1, 3 and 4). He also had fragile skin.

He had hyperextensibility of the metacarpophalangeal joints, dysplastic nails, left partial simian crease and partial syndactyly between 2nd and 3rd toes (Fig. 5).

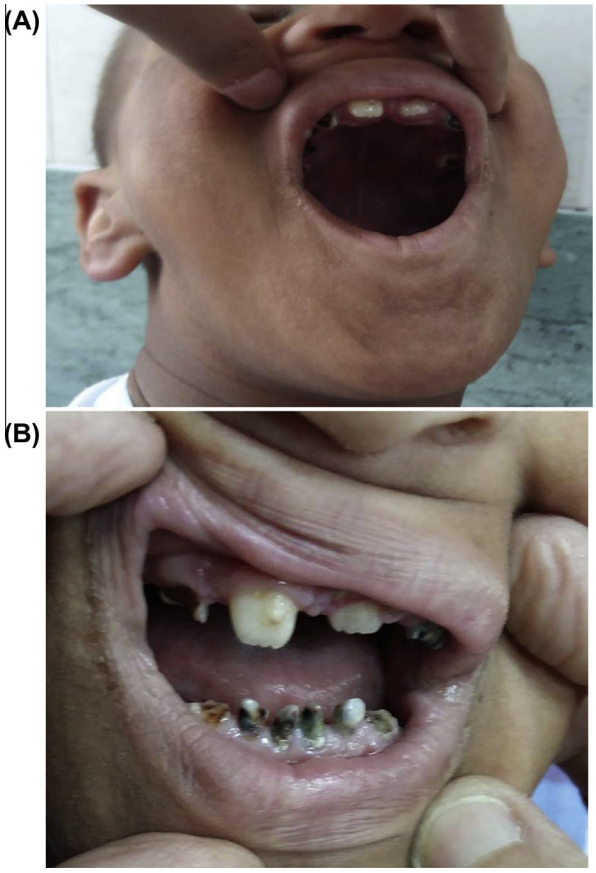

Figure 2 (A) High arched palate, dry lips and angular stomatitis. (B) Decayed oligodontic teeth.

Abdominal examination revealed the scar of the cholecystectomy operation. The back, cardiac and neurological examinations were apparently normal. The genitalia were also normal.

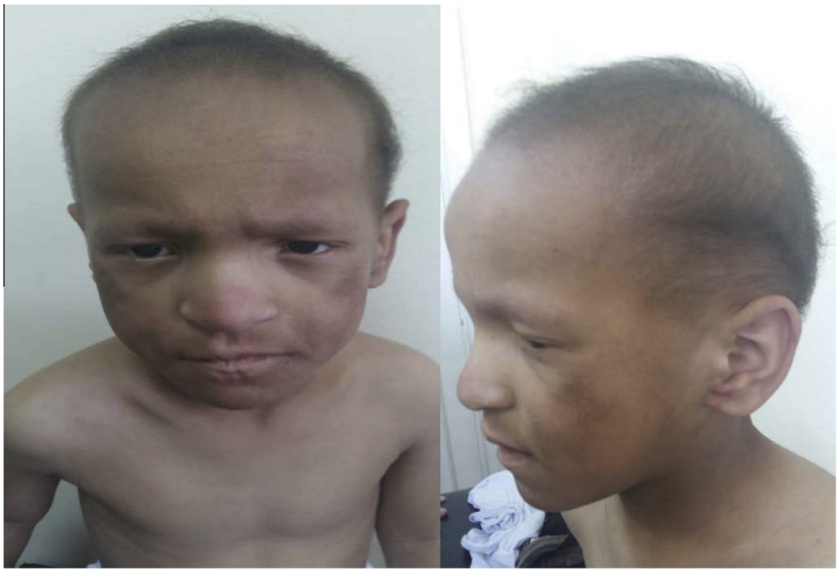

Figure 1 Sparse, thin dry hair over the scalp, scanty eyebrows and eyelashes, frontal bossing, prominent supraorbital ridges, depressed nasal bridge, saddle nose, dry lips and marked hyperpigmentation over the cheeks, perioral area, chin and neck.