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CASE REPORT

C syndrome with skeletal anomalies, mental retardation, eyelid chalazion, Bitot's spots and agenesis of the corpus callosum in an Egyptian child

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KEYWORDS

C syndrome;
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Agenesis of corpus callosum

Abstract We report a 2.5 year old female child, third in order of birth of healthy non consanguineous Egyptian parents with C syndrome. The patient had moderate mental retardation, trigonocephaly, protruding forehead, low anterior hair line, wide upslanted palpebral fissures, depressed nasal bridge, broad nose, high arched palate, microretrognathia, low set ears, short neck, scoliosis, hypertrichosis over the back, talipes equinovarus as well as interatrial septal defect. The patient had in addition chalazion in left lower eyelid as well as bilateral Bitot's spots most probably due to vitamin A deficiency. MRI brain revealed agenesis of the corpus callosum.

1. Introduction

C syndrome, also known as Opitz trigonocephaly syndrome (OTCS), is a rare and heterogeneous malformation syndrome characterized by trigonocephaly, variable mental retardation, hypotonia, variable cardiac defects, redundant skin, and dysmorphic facial features, including upslanted palpebral fissures, epicanthal folds, depressed nasal bridge, and low-set, posteriorly rotated ears [1]. The premature closure of the metopic suture results in a growth restriction of the frontal bones and trigonocephaly [2]. Morbidity and mortality are very high in this syndrome [3]. OTCS is a heterogeneous genetic disorder which occurs sporadically, although familial cases have also been reported [4].

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

2. Case report

A 2.5 year old female child, third in order of birth of healthy non consanguineous Egyptian parents. The patient was delivered at full term by vaginal delivery after uncomplicated pregnancy. The patient was referred to the Genetics Clinic, Pediatric Hospital, Ain Shams University complaining of developmental delay and abnormal skull shape.

The condition started since birth when her mother noticed that her baby had abnormal skull shape. At the age of 2 months, she developed focal convulsions (twitches in the eyes) and was given Tiratam for 6 months. The convulsions stopped at the age of 1 year and the patient stopped having

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anticonvulsant drug. At the age of 1 year, she had repeated chest infections and was admitted to the hospital twice. At the age of 2 years, she developed chalazion in the right eye then in the left eye. Then she sought medical advice at our Genetics Clinic.

Family history was unremarkable. Both parents were normal.

On examination, the patient had moderate mental retardation, her weight was 6 kg (below 3rd percentile), her length was 80 cm (below 3rd percentile), and her skull circumference was 46.5 cm (15th percentile).

The patient had trigonocephaly, protruding forehead, low anterior hair line, wide upslanted palpebral fissures, chalazion in the left lower lid, bilateral Bitot's spots, depressed nasal bridge, broad nose, high arched palate, microretrognathia, low set ears, short neck, scoliosis and hypertrichosis over the back (Figs. 1–3). She also had talipes equinovarus (Fig. 4).

Cardiac examination was apparently normal. Abdominal examination revealed small umbilical hernia (Fig. 5). Genital and neurologic examinations were normal.

Abdomino-pelvic ultrasonography was normal. ECHO cardiography revealed interatrial septal defect. Extended metabolic screen, serum lactate and serum ammonium were normal. Karyotype was also normal. MRI brain revealed complete agenesis corpus callosum (Fig. 6), premature closure of metopic suture. Ophthalmology examination revealed lower eyelid chalazion, dryness and Bitot's spots in both eyes (Fig. 2). Fundus examination was normal.

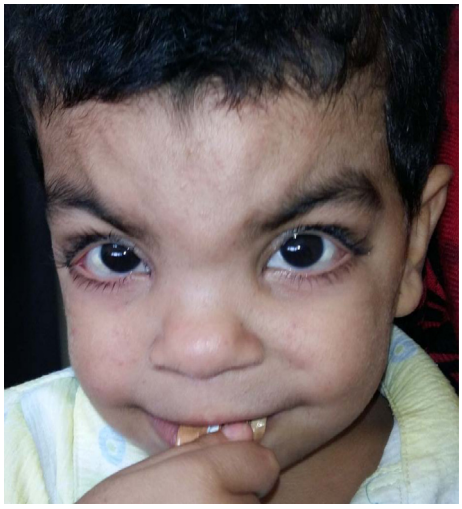

Figure 2 Facial features after surgery for craniosynostosis and bilateral Bitot's spots.

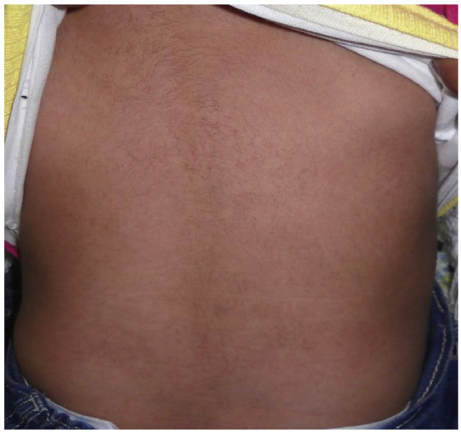

Figure 3 Hypertrichosis over the back.

3. Discussion

We report a 2.5 year old female child with Opitz trigonocephaly C syndrome (OTCS) with trigonocephaly, protruding forehead, low anterior hair line, wide palpebral fissures, chalazion in the left lower lid, depressed nasal bridge, broad nose, high arched palate, microretrognathia, low set ears, short neck, scoliosis, hypertrichosis over the back and talipes equinovarus.

Opitz trigonocephaly C syndrome (OTCS) is a rare and heterogeneous genetic disorder characterized by synostosis of metopic suture, dysmorphic facial features, variable mental

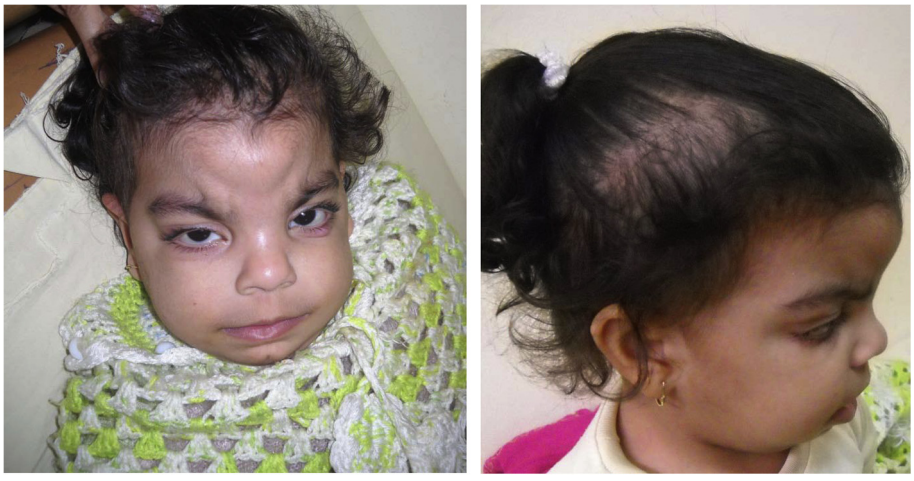

Figure 1 Trigonocephaly, protruding forehead, low anterior hair line, wide upslanted palpebral fissures, chalazion in the left lower eye lid, depressed nasal bridge, broad nose, microretrognathia, low set ears and short neck.