International Journal of Contemporary Pediatrics
Jain R et al. Int J Contemp Pediatr. 2016 Nov;3(4):1463-1465
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: http://dx.doi.org/10.18203/2349-3291.ijcp20163701

Case Report

Coffin siris syndrome: a rare clinical entity

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Received: 09 July 2016
Revised: 18 July 2016
Accepted: 05 August 2016

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ABSTRACT

Coffin-siris syndrome (CSS) is a rare, clinically heterogeneous disorder considered in the setting of prenatal onset of mild to moderate growth deficiency, facial dysmorphism, cognitive/developmental delay, and speech impairment, moderate to severe hypotonia, seizures and 5th finger/nail hypoplasia. The child had distinctive features of CSS like developmental delay, seizures, and coarse facial features, body hypertrichosis, scalp hypotrichosis and SNHL.

Keywords: Coffin siris syndrome, Global development delay, Seizures, Generalized hirsutism

INTRODUCTION

Coffin-Siris syndrome (CSS) is a rare, clinically heterogeneous disorder considered in the setting of prenatal onset of mild to moderate growth deficiency, facial dysmorphism, cognitive/developmental delay, and speech impairment, moderate to severe hypotonia, seizures and 5th finger/nail hypoplasia.[1] Coffin siris syndrome was first described in 1970. It is a rare syndrome and around 100 cases have been reported so far. It includes features such as developmental delay, seizures, coarse facial features and generalized hirsutism.[2]

CASE REPORT

A 3.5 year old male child of Indian origin born of a non-consanguineous marriage presented to our hospital with chief complaints of global developmental delay and convulsions. Child was a full term normal delivery, birth weight of 2.2 kg, second in birth order.

Parents observed developmental delay after three months of age and subsequently child had recurrent seizures in form of infantile spasm which later converted in to generalized tonic convulsion after the age of nine months. He was on sodium valproate for seizures with response and had no history of receiving phenytoin, steroids or other drugs causing hypertrichosis.

Figure 1: Dysmorphic facial features like low set ears, large pinna, depressed nasal bridge, flat nasal tip, thick eyebrows and long eyelashes.

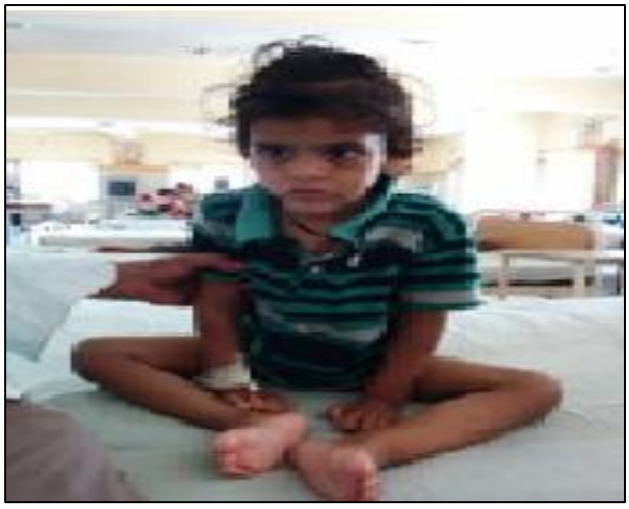

Figure 2: Short stubby hand and generalized hypotonia.

Figure 3: Tanner stage 2 genitalia.

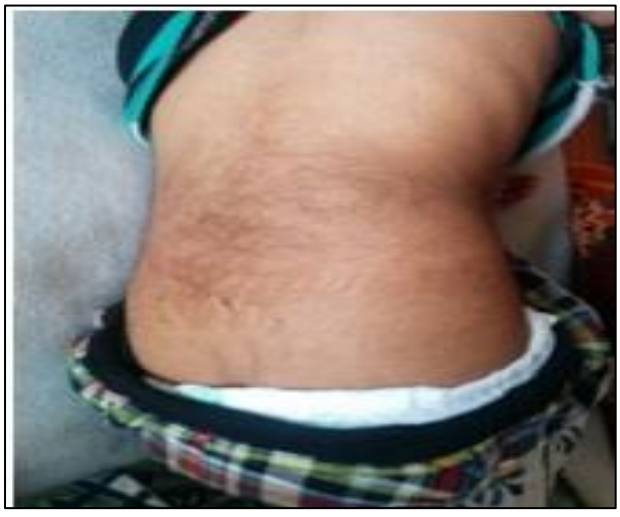

Figure 4: Generalised hypertrichosis.

Family history is unremarkable and both the parents and an elder sibling were healthy.

On physical examinations, height was 97 cm (between median and -1SD), weight was 13.5 kg (between median and -1SD), head circumference was 48 cm (between 15th and 3rd percentile). He had dysmorphic facial features like low set ears, large pinna, depressed nasal bridge, flat nasal tip, generalized hypertrichosis, thick eyebrows, long eyelashes, short stubby hand and generalized hypotonia. Developmental age of child was approximately 1 year, language milestones were more delayed. Rest of the neurological and systemic examination was unremarkable.

Child's routine blood investigations, ECHO, USG abdomen, MRI brain and karyotyping were normal. Bone age was same as chronological age with no other skeletal abnormality present. Vision was normal. BERA suggestive of near normal hearing in right ear and 50 db hearing loss in left ear. EEG shows frequent epileptiform discharges from both hemispheres.

DISCUSSION

Coffin siris syndrome (CSS) is characterized by distinctive facial features, moderate to severe developmental/cognitive delay and aplasia or hypoplasia of the distal phalanx or nail of the fifth digit. Expressive language is more severely affected than receptive language. Most individuals with a clinical diagnosis of CSS have all three of the following major findings and one of each of the three following categories of minor findings.[3]

Major findings

Presence of all three-developmental or cognitive delay (100%); facial features; fifth digit nail/distal phalanx hypoplasia / aplasia; (Some of the patients lacked some typical CSS abnormalities, such as hypoplastic or absent fingernails or toenails).[2]

Minor findings

At least one feature from each of the three following categories: ectodermal- hirsutism/hypertrichosis (93%). Hair growth in atypical areas (e.g. the back) or excessive hair growth on the arms or face, sparse scalp hair, especially in infancy, dental anomalies; constitutional-microcephaly, intrauterine growth retardation, short stature, failure to thrive, frequent infections, organ related-cardiac anomalies, feeding difficulties, gastrointestinal anomalies, genitourinary/renal anomalies, brain/cranial malformations or seizures, vision changes, hearing loss.[4] The differential diagnosis of CSS includes brachymorphism-onychodysplasia-dysphalangism(BOD) syndrome, DOOR syndrome, Nicolaides-Baraitster syndrome (NCBRS), coffin lowry syndrome, fetal hydantoinsyndrome.[5]

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Jain R, Gediya S, Khan A, Goyal S, Poswal L. Coffin siris syndrome: a rare clinical entity. Int J Contemp Pediatr 2016;3:1463-5.