MEDDOCS Open Access Publisher

ISSN: 2639-9237

Journal of Case Reports and Medical Images

Open Access | Case Report

Lange's cornelia syndrome: About two cases

Kh Mouaddine*; G Ismaili; M Kmari; A Ourrai; A Hassani; R Abilkassem; A Agadr

Department of Pediatrics, Mohammed V Military Hospital Rabat. Faculty of Medicine and Pharmacy Rabat, Morocco.

***Corresponding Author(s): Khadija Mouaddine**
Department of Pediatrics, Mohammed V Military Hospital Rabat. Faculty of Medicine and Pharmacy Rabat, Riad Loudaya 12020 Temara, Morocco.
Tel: +212635306318;
Email: khadijamouaddine7@gmail.com

Received: Sep 29, 2021
Accepted: Nov 02, 2021
Published Online: Nov 04, 2021
Journal: Journal of Case Reports and Medical Images
Publisher: MedDocs Publishers LLC
Online edition: http://meddocsonline.org/

Case I

M, N a 3-year-old M boy, term born in hospital, the third of 3 siblings, from a consanguineous marriage, admitted for dysmorphic syndrome with mental retardation. The clinical examination showed severe failure to thrive (-4DS), facial dysmorphic included hirsutism, prominent and sloping forehead, dental abnormalities, retrognatism, well-defined, arched and confluent eyebrows, long eyelashes, ant everted nostrils associated with generalized hypotonia without involvement of the extremities (Figure 1).

Cerebral MRI revealed a delay in myelination of the peri-ventricular white matter, ophthalmologic examination, Otorhinology, skeletal X-ray, trans-thoracic ultrasound and abdominopelvic ultrasound are normal.

Hormonal exploration (TSH, 17 OHP, 8 h cortisol) as well as the phosphocalcic balance without abnormalities, the karyotype is normal: 46 XX.

The diagnosis of Cornelia de Lange is retained in our patient due to the association of the typical dysmorphic syndrome with a failure to thrive.

Case II

K, J is a 14 month old male infant, from a non-consanguineous marriage, the youngest of three siblings, without similar cases in the family, birth weight and height were unspecified. Having as antecedents: Neonatal suffering, retardation of psychomotor development, repeated respiratory infections on gastroesophageal reflux disease. The onset was six months old when the parents noticed that the head was missing.

Cite this article: Mouaddine KH, Ismaili G, Kmari M, Ourrai A, Hassani A, Abilkassem R, Agadr A. Lange's Cornelia Syndrome: about two Cases. J Case Rep Med Images. 2021; 4 (2): 1086.

Clinical examination found mucocutaneous paleness, failure to thrive (weight and height less than minus 4 SD). The facial dysmorphia included hirsutism, prominent and sloping forehead, dental abnormalities, retrognatism, well defined, arched and flowing eyebrows (synophrys), long eyelashes, anteverted nostrils. He also had a funnel-shaped chest deformity, without limb abnormalities. Neurological examination revealed hypotonia (Figure 2).

The malformation work-up included ophthalmologic, otorhinologic, brain MRI, abdominal ultrasound, echocardiography, and skeletal x-rays were all normal.

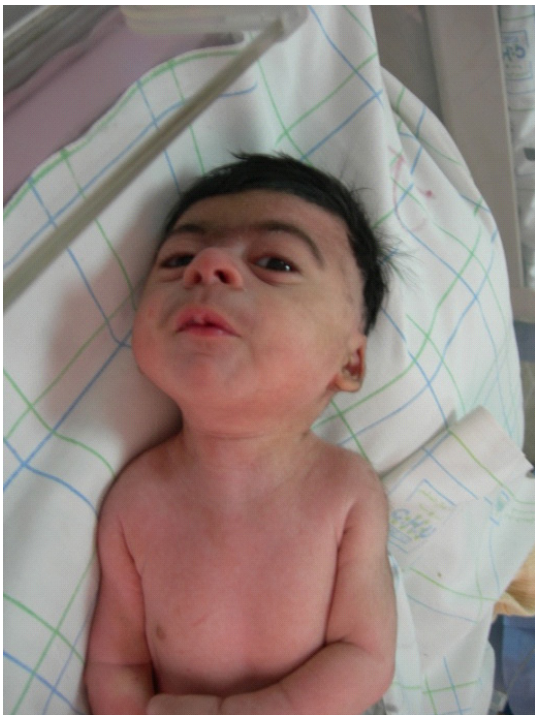

Figure 1: Characteristic aspect of Cornelia de Lange syndrome in patient M.N.

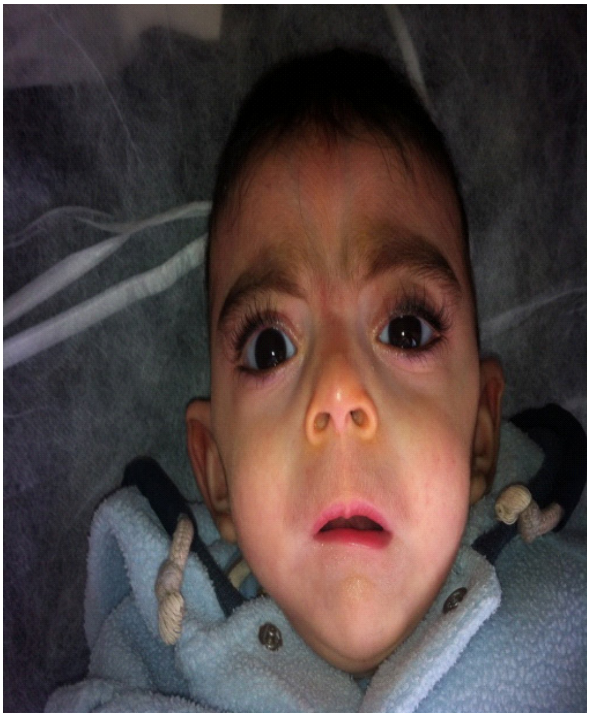

Figure 2: Characteristic aspect of Cornelia de Lange syndrome in patient K.J.