Lange’s cornelia syndrome: About two cases

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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, anteverted 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.

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.