

Cayler Cardio-Facial Syndrome: A Rare Case Report

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Abstract

Case Report

Background: Cayler cardio-facial syndrome is a rare syndrome characterised by asymmetric crying facies with congenital heart defects. Syndrome may be associated with other dysmorphic features and systemic anomalies. **Case Report:** We report the clinical observation of a 04 years old boy presenting with Cayler cardio-facial syndrome.

Keywords: Cayler cardio-facial syndrome, asymmetric crying facies, congenital heart defects.

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INTRODUCTION

The Cayler's cardiofacial syndrome is a rare pediatric condition that combines characteristic facial asymmetry with conotruncal cardiovascular malformations. It can be associated with cardiac, ENT, renal, digestive, and osteoarticular malformations with a dysfunction of cellular immunity and hypocalcemia, as well as cognitive and behavioral disorders that appear over the years [1].

In this paper, we provide a case report of a 04 year old boy presenting with the cayler cardio-facial syndrome.

CASE REPORT

We report the clinical observation of 04 years old boy, stemming from a non-consanguineous marriage, born from a poorly followed pregnancy.

His parent's report, since the first week of his birth, a notion of cyanosis of the mouth and extremities and a facial asymmetry during the cries, evolving in a context of psychomotor delay as well as repeated boncho alveolitis.

Physical examination revealed perioral and extremity cyanosis with desaturation at 75%. The Weight and height was less than two standard deviations. Body temperature was normal and blood pressure 110/60 mmhg.

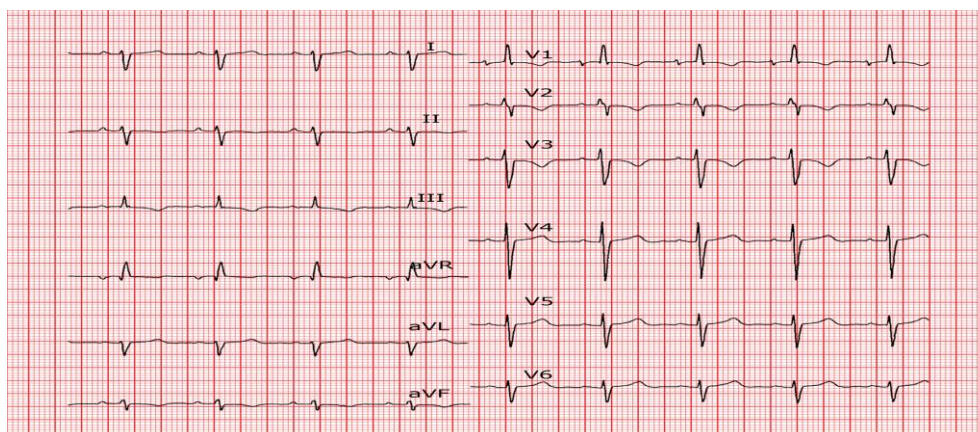


Fig-1: EKG showing a right axial deviation in relation to right ventricular hypertrophy.

Auscultation found a maximal systolic ejectional murmur at the left sternal border and at the pulmonary area radiating towards the back and the left armpit. Peripheral pulses are present and symmetrical. Absence of signs of right heart failure. Abdominal examination doesn't reveal any palpable mass, hepatomegaly, or ascites. Electrocardiogram at admission showed a right axial deviation in relation to

right ventricular hypertrophy. Chest radiograph found a clogged heart appearance.

The echocardiographic evaluation demonstrated a tetralogy of Fallot, associating a wide non-restrictive ventricular septal defect, an aorta straddling the two ventricles, pulmonary stenosis, right ventricular hypertrophy and isosystemic pulmonary hypertension.

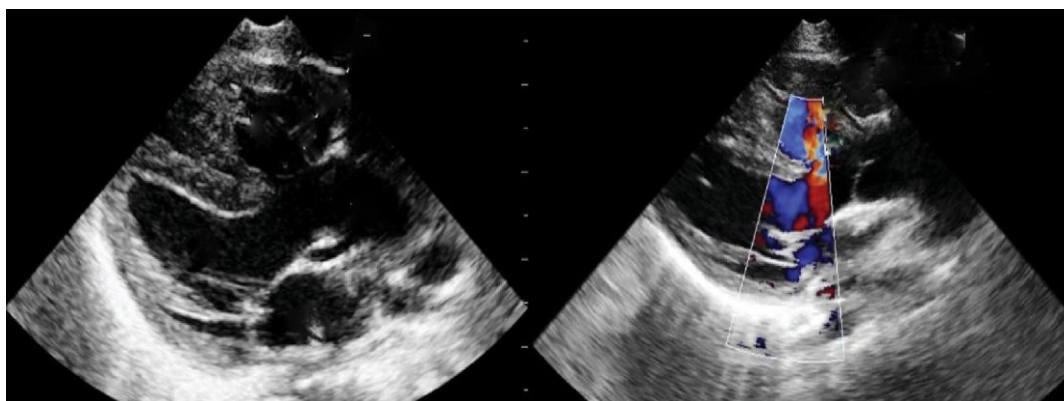


Fig-2: Echocardiographic view showing the ventricular septal defect and the aorta straddling both the two ventricles.

DISCUSSION

The Cayler cardio-facial syndrome is a combination of asymmetric crying facies, caused by the hypoplasia or agenesis of depressor angularis muscle (DAOM) of the lip, with congenital heart disease. This syndrome may be sporadic or associated with 22q11.2 deletion (29%) [2].

Cayler (1967, 1969) reported 5 babies with unilateral partial facial palsy and ventricular septal defect. Nelson and Eng (1972) and Pape and Pickering (1972) noted that asymmetric crying facies was due to congenital hypoplasia of the depressor anguli oris muscle and reported other associated anomalies. Papadatos *et al.* (1974) described apparently autosomal dominant inheritance of congenital hypoplasia of the depressor anguli oris muscle. The effect results in asymmetry of the lower lip, especially evident in smiling or crying [3].

DAOM hypoplasia has almost 50% association with congenital heart diseases [4]. Other common heart defects associated with Cayler cardio facial syndrome may include ventricular septal defect, tetralogy of Fallot and patent ductus arteriosus. The associated malformations which can be seen are microcephaly, micrognathia, microphthalmos, mental retardation, genitourinary abnormalities and limb anomalies like syndactyly [5, 6].

The diagnosis of 22q11.2DS is suspected when clinical symptoms are present. The diagnosis is confirmed by a blood test that can detect a microscopic chromosomal deletion on chromosome 22. There are many new tests to detect this deletion including whole

genome array, SNP array, comparative genomic hybridization, and MLPA. FISH studies have also been useful in finding the deletion in most patients. Furthermore, routine chromosome (cytogenetic) testing is also performed because a small number of affected individuals have a chromosome rearrangement involving chromosome 22q which may change the recurrence risk counseling for the parents [7, 8].

A team approach is often useful and generally recommended when making decisions about the treatment of symptoms in patients with 22q11.2DS. A geneticist, pediatrician, cardiologist, immunologist, endocrinologist, gastroenterologist, otolaryngologist, plastic surgeon, speech pathologist, audiologist, orthodontist, dentist, urologist/nephrologist, orthopedist, child development specialist, neurologist and psychologist may all be called in to consult with the parents, depending on the clinical presentation of the child. Genetic counseling is recommended for families with an affected child [9-11].

CONCLUSION

Cayler's syndrome is a rare pediatric condition that must be suspected in babies with asymmetrical cry pattern and normal facies while sleeping. Echocardiographic assessment is required to diagnose the associated congenital heart diseases. Its management is multidisciplinary including medical and surgical approach.

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