Journal of Pediatric Sciences

Ellis-Van Creveld's Syndrome with Common Atrium

Amar M Taksande, Gautami V

Journal of Pediatric Sciences 2012;4(2):e128

How to cite this article:

Taksande AM, Gautami V. Ellis-Van Creveld's syndrome with common atrium. Journal of Pediatric Sciences. 2012;4(2):e128

CASE REPORT

Ellis-Van Creveld's Syndrome with Common Atrium

Amar M Taksande, Gautami V

Department of Pediatrics, Jawaharlal Nehru Medical College, India

Abstract:
Ellis-van Creveld syndrome (EVCS) is a rare autosomal recessive disorder characterized by a variable spectrum of clinical findings. Classical EVCS comprises a tetrad of clinical manifestations of chondrodystrophy, polydactyly, ectodermal dysplasia, and cardiac defects. An eight years old female child presented with history of breathlessness on exertion has disproportionate short stature, polydactyly, hydratic ectodermal dysplasia suggestive of EVCS. Her echocardiographic examination revealed common atrium (CA) with mild pulmonary hypertension.
Keywords: *Ellis-Van Creveld syndrome, polydactyly, common atrium*

Accepted: 04/14/2012 Published: 06/01/2012

Corresponding author: *Amar M. Taksande. Professor, Department of PediatricsJawaharlal Nehru Medical College, Sawangi Meghe, Wardha, Maharashtra 442102Mail: amar.taksande@gmail.com Phone: 07152-260310*

Introduction

EVCS or chondroectodermal dysplasia is a complex genetic syndrome first described in 1940 by Drs Richard Ellis and Simon van Creveld. Reported incidence is one in 1,500,000 live births. Incidence in India is very rare. Classical EVCS comprises a tetrad of clinical manifestations, chondrodysplasia, ectodermal dysplasia, polydactyly and congenital heart disease [1]. Popli et al had reported only single case of ECVS in India [2]. Here, we present a rare case of EVCS with characteristic findings, including narrow thorax, short stature, bilateral polydactyly of the hands, dystropic nails and congential heart disease.

Case Report

An 8 year old female weighing 13.5kg, presented with history of breathlessness on exertion. She was a product of non-consanguineous marriage and born by preterm vaginal delivery at hospital. Birth weight was 1000gm. Antenatal, natal and postnatal histories were noncontributory. There was no family history of neonatal death. On general examination, she showed stunted growth (height 102cm, expected 127.8 cm) with UpperLimb:LowerLimb ratio was 1.35, narrow chest with widely spaced nipple and polydactyl in both hands (Figure 1) with valgus deformity of the knee. There was hypoplastic tooth with dental caries, multiple frenula (Figure 2), dysplastic nails and sparse hair. She was afebrile, no cyanosis, no clubbing and all peripheral pulses were present. Vitals were Heart Rate: 90/m, Respiratory Rate: 22/m, Blood Pressure: 108/60mmHg and SpO2: 92%. Cardiovascular examination revealed wide & fixed split, 2/6 ejection systolic murmur which was best heard in the upper left parasternal area and 2/6 middiastolic murmur was heard at left lower parasternal area. Other systemic examination was normal. Her psychomotor and mental development was normal. Chest X-ray showed an enlarged cardiac silhouette (Cardiothoracic ratio 0.6) and increased pulmonary vasculature. Electrocardiography showed sinus

rhythm, QRS axis +90 degree, PR interval is 160msec, right atrial enlargement, right bundle branch block with right ventricular hypertrophy. Transthoracic 2D-echocardiography (Figure 3) reveals common atrium with mild tricuspid regurgitation with mild mitral regurgitation with mild pulmonary arterial hypertension(PAH).

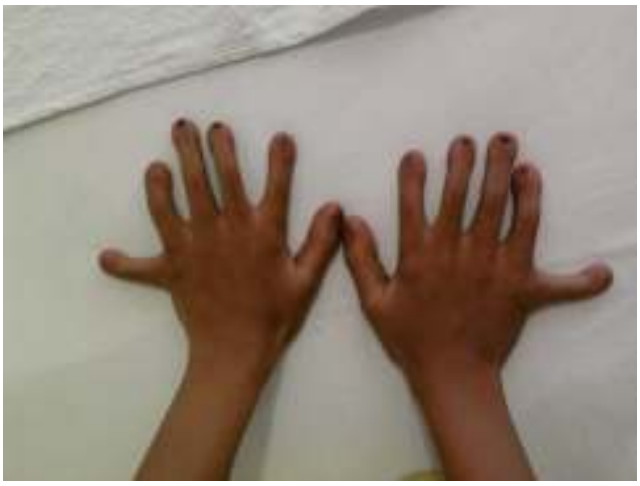

Figure 1. EVCS child shows polydactyly and dystrophic fingernails.

Figure 2: EVCS child shows multiple freleum with dental anomaly with sparse hair

Discussion

EVCS is a rare autosomal recessive disease resulting from a genetic defect located in chromosome 4p16 which was discovered in the year 2000. Mutations of the *EVC1* and *EVC2* genes, located in a head to head configuration on chromosome 4p16, have been identified as causative. Around 150 cases are described in the literature [3-5]. The risk of recurrence for siblings is 1 in 4 (25 %) for each offspring [3-4]. EVC phenotype is variable and affects multiple organs. All embryonic layers appear involved in EVCS. The signs of ectodermal dysplasia are usually limited to nails, teeth and gums, mesodermal involvement indicate abnormalities of the bones, heart and the kidney whereas the endodermal involvement is uncommon, but with lung and liver abnormalities have been reported in some cases [5].

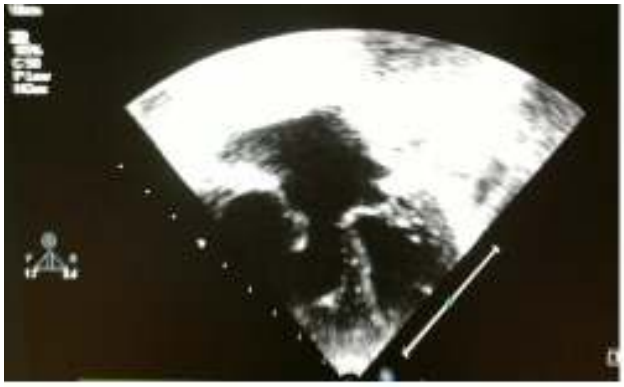

Figure 3. Transthoracic echocardiography demonstrates common atrium

After birth, the cardinal features usually present are [5-9]:

1. Disproportionate small stature and shortening of the middle and distal phalanges as opposed to proximal segments. Valgus deformity of the knee may be present.

2. Post axial polydactyly of hands and occasionally involving feet. It may be just extra soft tissue not adherent to skeleton and devoid of bone, cartilage, joint or tendon or the digit may show duplication.

3. Hidrotic ectodermal dysplasia mainly affecting the nails, hair and teeth. Nails are hypoplastic, dystrophic, and friable. Nails can be completely absent in some cases. Tooth involvement may include neonatal teeth, partial anodontia, small teeth, and delayed eruption. Enamel hypoplasia may result in abnormally shaped teeth with frequent malocclusion.Hair may occasionally be sparse.

4. Congenital heart defects occur in about 50% of case. Single atrium or common atrium is a rare variety of interatrial communication, characterized by absent or virtual absence of atrial septum. Other abnormalities may accompany these lesions, such as aortic atresia, hypoplasia

of the ascending aorta or of the left ventricle. cleft mitral valve, atrial septal defect, ventricular septal defect, transposition of big arteria, mitral valve insufficiency, double orifice mitral valve, and cleft mitral valve [9]. The presence of congenital heart disease may support the diagnosis of the EVCS. In our case, child has disproportionate short stature with valgus deformity of the knee, postaxial polydactyly of hand, dystrophic nails, multiple frenulum, sparse hair and congential heart disease (common atrium) was present. The retrospective review study done by Hill et al [10] found that thirty-two pediatric patients with congenital heart disease (CHD) and EVC syndrome. Twenty-eight (88%) had an endocardial cushion defect, with 15 of these having primary failure of atrial septation resulting in CA. Persistent left superior vena cava (LSVC) and pulmonary venous connection abnormalities were common finding.

Oral lesions includes a fusion of the anterior portion of the upper lip to the maxillary gingival margin, resulting in an absence of mucobuccal fold and the upper lip to present a slight V-notch in the middle. Short upper lip, bound by frenula to alveolar ridge (lip tie). Genitourinary anomalies such as agenesis and renal dysplasia, ureterectasia and nephrocalcinosis usually present in 20% of cases [11]. Musculoskeletal anomalies include low-set shoulders, a narrow thorax frequently leading to respiratory difficulties, knock knees, lumbar lordosis, broad hands and feet, and sausage-shaped fingers. Although, most of the children with EVCS have normal intelligence, but mental retardation and central nervous system abnormalities were also reported in some cases [12]. Our case had normal mental development with no abnormality of urinary and central nervous system.

Radiological features such as curvature of the humerus, enlargement of distal ends of radii and ulnae, supernumerary carpal bone centers with fusion, clinodactyly, synmetacarpalism, synostosis, wedge-shaped tibia epiphyses and genu valgum vary depending on disease severity and age. Infant mortality rate are higher primarily due to cardiorespiratory failure, if they survive, morbidity is significant. The syndrome can be diagnosed during the prenatal period, starting from the 18th week of gestation, by ultrasonography [12]. The clinical diagnosis is based on observation of the symptoms and manifestations and supported by the skeletal survey. The definitive diagnosis is molecular, based on homozygosity for a mutation in the *EVC* and *EVC2* genes by direct sequencing.

EVCS has to be differentiated from asphyxiating thoracic dysplasia, Short-rib polydactyly, Jeune syndrome and orofaciodigital syndromes. Asphyxiating thoracic dysplasia cases have a small chest that appears long and narrow, with death occurring in neonatal period due to respiratory distress. Polydactyly are symptomless and nails are almost normal. Short-rib polydactyly is characterized by hypoplastic thorax due to short ribs, short limbs, frequent polydactyly and visceral abnormalities. Jeune syndrome is a rare, potentially lethal, autosomal recessive disease characterized by thoracic dystrophy, short limbs, small stature, polydactyly and generalized bony dysplasia. Orofaciodigital syndromes result from dominant sex-linked inheritance, they are limited to women and clinically characterized by multiple gingivolabial frenula, hypoplasia of the nasal cartilages, moderate mental retardation, fissured tongue and ankyloglossia. Symptomatic management is mostly required in the neonatal period, including treatment of the respiratory distress due both to narrow chest and heart failure. O'Connor et al [13] mentioned that the surgery for congenital malformations of the heart can be successful in infants with EVCS, but mortality is high and post-operative respiratory morbidity should be expected. Approximately 50% of patients with EVCS die in early infancy as a consequence of cardiorespiratory problems. In conclusion, careful dysmorphological examination should be performed in all patients presenting with dysmorfic features to diagnose as EVCS and should be evaluated for congenital heart defect.

References

1. Dasilva EO, Janovitz D, De Albuquerque SC. Ellis–van Creveld syndrome: report of 15 cases in an inbred kindred. J Med Genet, 1980, 17:349–356.
2. Popli MB, Popli V, Ellis Van Creveld Syndrome. Ind Jour Radiol 2002; 12:549-550.
3. Sanjeet CG, Roy TNS & Venugopal K. Common atrium in a child with Ellis Van Creveld Syndrome Heart 2002; 88: 142.
4. Arya L, Mendiratta V, Sharma RC, Solanki RS. Ellis-van Creveld Syndrome: a report of two cases. Pediatr Dermatol 2001;18:485-9.
5. Varela M, Ramos C. Chondroectodermal dysplasia (Ellis-van Creveld syndrome): a case report. European Journal of Orthodontics 1996; 18: 313-318.
6. Baujat G, Le Merrer M. Ellis-van Creveld syndrome. Orphanet J Rare Dis 2007;2:27.
7. Aldegheri R. Distraction osteogenesis for lengthening of the tibia in patients who have limb-length

discrepancy or short stature. J Bone Joint Surg Am 1999;81:624-34.
8. Ghosh S, Setty S, Sivakumar A, Pai KM. Report of a new syndrome: focus on differential diagnosis and review of Ellis-van Creveld, Curry-Hall, acrofacial dysostosis, and orofacial digital syndromes. Oral Surg Oral Med Oral Pathol Oral Radiol Endod. 2007;103:670-6.
9. Giknis FL:Single atrium and the Ellis-van Creveld syndrome. J Pediatr 1963;62:558-64
10. Hills CB, Kochilas L, Schimmenti LA, Moller JH. Ellis-van Creveld syndrome and congenital heart defects: presentation of an additional 32 cases. Pediatr Cardiol. 2011 Oct;32(7):977-82.
11. Rosemberg S, Cameiro P C, Zerbini M C. Chondroectodermal dysplasia (Ellis-van Creveld) with anomalies of CNS and urinary tract. Am J Med Genet 1983;2:64-71.
12. Zangwill KM, Boal DKB, Ladda RL. Dandy-Walker malformation in Ellis-van Creveld syndrome. Am J Med Genet 1988;31:123-9
13. O'Connor MJ, Rider NL, Thomas Collins R, Hanna BD, Holmes Morton D, Strauss KA. Contemporary management of congenital malformations of the heart in infants with Ellis - van Creveld syndrome: a report of nine cases. Cardiol Young. 2011; 21:145-52.