CASE REPORT

CHONDROECTODERMAL SYNDROME

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Chondroectodermal syndrome or Ellis Van Creveld (EVC) is a rare autosomal recessive congenital disorder. It was first described by Richard W.B.Ellis and Simon Van Creveld in 1940. Parental consanguinity is present in about 30% of the cases. A large number of cases were reported in Amish population of Lancaster County, Pennsylvania USA and also in Aboriginal community of Australia in 1964. The incidence in Amish population is 1/5000 live births and in general population 7/1,000,000. There are only 150 cases reported worldwide. The principal feature of this syndrome is a tetrad of disproportionate dwarfism, ectodermal dysplasia, bilateral postaxial polydactyly and congenital heart defects.

Keywords: Chondroectodermal dysplasia; Ellis Van Creveld syndrome; Postaxial Polydactyly

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INTRODUCTION

Chondroectodermal syndrome or Ellis Van Creveld (EVC) is a rare congenital disorder. It was first described by Richard W.B.Ellis and Simon Van Creveld in 1940. The inheritance pattern is autosomal recessive and is thought to be a genetic defect in chromosome 4p16. Parental consanguinity is present in about 30% of the cases.[1–5] A large number of cases were reported in Amish population of Lancaster county, Pennsylvania USA and also in Aboriginal community of Australia in 1964.[6] The incidence in Amish population is 1/5000live births and in general population 7/1,000,000.[4,5] There are only 150 cases reported worldwide. The principal feature of this syndrome is a tetrad of disproportionate dwarfism, ectodermal dysplasia, bilateral postaxial polydactyly and congenital heart defects.[3–5]

CASE REPORT

Two siblings, a sister 8 yrs. old and brother 6 yrs. old presented with their mother on 23 Sep. 2017 to outpatient department of National Orthopaedic Hospital Bahawalpur. Mother was giving history of walking difficulty in both of them since they started walking. Family history was positive for consanguinity and there were other family members affected by the same disorder. These included 4 of their cousins of different age groups. But due to social constrains they were unwilling to provide any pictures except these two kids. Antenatal, natal and neonatal histories were non-contributory. IQ was within normal limits. GPE was remarkable for short stature, post axial polydactyly in all the limbs, cleft palate and lip, hypoplasia of nails and generalized ligamentous laxity. They both had severe genu valgum deformities with more than 10 cm of inter-malleolar distance. X-rays were done which excluded other differentials. They were referred to cardiologist, paediatrician and dentist to detect other manifestation of this disease. Both cardiologist and paediatrician opinions were normal and dentist advised prosthetic surgery after their skeletal maturity.

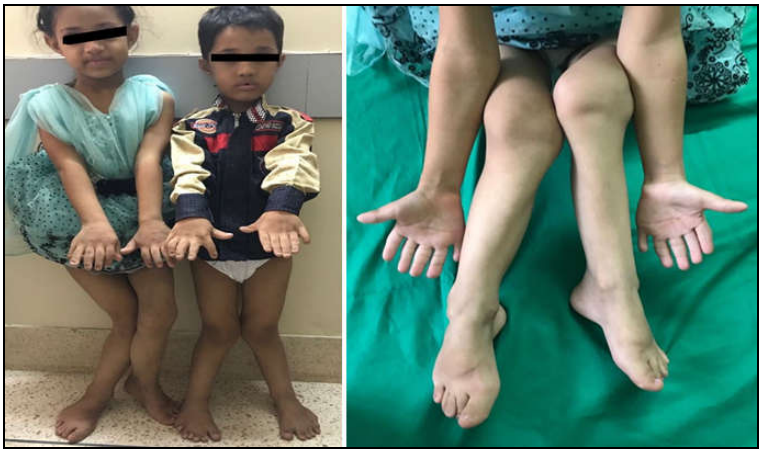

Figure-1: Polydectaly and genu valgum

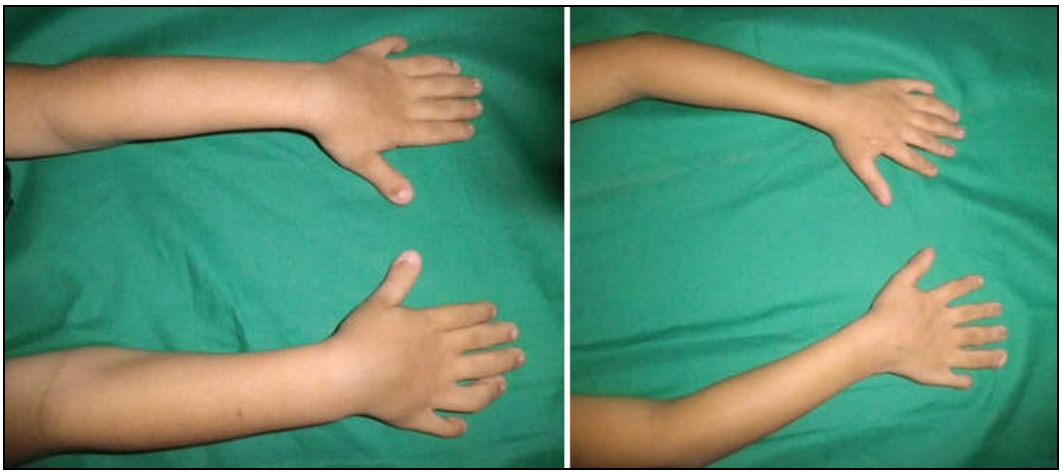

Figure-2: Note the post-axial polydactyly and nail changes in both patients' hands

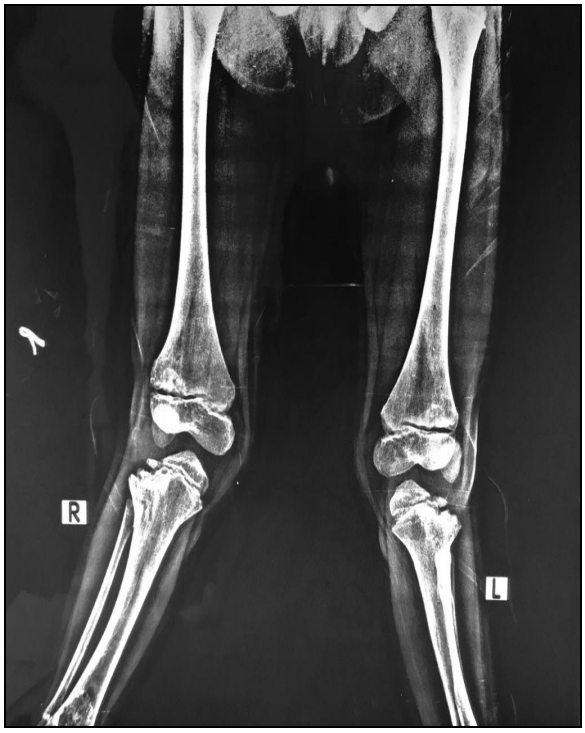

Figure-3: X-ray showing deformity of knees

DISCUSSION

Chondro-ectodermal dysplasia or Ellis Van Creveld Syndrome is a rare genetic disorder. While it is autosomal recessive disorder, 70% patients had no parental consanguinity.[2,3] It is more common in Amish community of Pennsylvania USA with a birth prevalence of 1/5000 live births.[2] Although, antenatal diagnosis can be made by detail ultrasound as early as 18th week of gestation, clinical diagnosis is more certain after birth.[4,7,8] The most striking clinical feature is chondrodysplasia affecting the ossification of long bones. This result in dystrophic dwarfism affecting the middle and distal part of limbs called acromesomelic dwarfism.[3,4] Orthopaedic features include short stubby hands and feet, dysplastic nails and post axial polydactly commonly affecting upper limb but can occur in lower limb in 10% of the cases. Other features include knee genu valgum, bowing of humerus or femur, talipes equinovarus, talipes calcaneovalgus and pectus carinatum with a long narrow chest.[2,4,8]

Congenital heart defects are present in 50-60% of the cases. These include patent ductus arteriosis, atrial septal defects and ventricular septal defects, mitral and tricuspid valve defects. Many of

these patients die young due to these malformations.[8,9] The oral manifestations include upper lip fusion to the gingival margin causing mucobuccal fold absence, cleft lip or palate, multiple small accessory frenula, ankyloglossia, mal-occlusion, microdontia teeth, delayed eruption and supernumerary teeth.[5,10] Urogenital abnormalities are present in about 25% of the cases and include mega-ureter, renal stones, renal agenesis, uteral and vulvar atresia. Cryptorchidism and hypospadias has been reported occasionally.[3,8] Infrequently, thoracic wall abnormalities resulting in decreased pulmonary functions are seen. Also, strabismus and hematopoietic disorders like aplastic anemia and leukemia are rarely found. The cognitive and motor developments of patients affected by EVC syndrome are normal.[3,8,9]

Differential diagnosis of EVC is asphyxiating thoracic dystrophy (Jeune syndrome), achondroplasia and Morquio's syndrome. Asphyxiating thoracic dystrophy or Jeune syndrome is autosomal recessive condition and is lethal in nature. The characteristic features include diastrophic dwarfism which is rhizomelic (Humerus shorter than forearm and femur shorter than tibia) rather than mesomelic, thoracic dystrophy, polydactyly and generalized bony dysplasia. Additionally, abnormal retinal pigmentation, renal defects and lung hypoplasia is seen.[2–4]

Achondroplasia is characterized by rhizomelic dwarfism, normal trunk adult height, depressed nose, frontal bossing, trident hands (fingers same length with divergent ring and middle fingers), genu varum, radial head subluxation, muscular hypotonia, thoracolumbar kyphosis and Morquio's syndrome presents with proportionate dwarfism, normal intelligence, waddling gait, genu valgum, thoracic kyphosis and corneal clouding. Radiographs show thickened skull, wide ribs, vertebral beaking, coxa vara with non-ossified femoral head, bullet-shaped metacarpals and odontoid hypoplasia leading to cervical instability.[4]

In conclusion, a multidisciplinary approach is always needed. These include orthopaedic surgeon, maxillo-facial surgeon, cardiologist, pulmonologist, urologist and paediatrician for suitable diagnosis, management and rehabilitation of such patients. Orthopaedic surgeon has an important role in early diagnosis and recommending treatment protocols that improves the quality of life of patients and establishing a differential diagnosis with other congenital pathologies.

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