

Exotropia in a case of Ellis Van Creveld syndrome: A rare case report

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Key words: Ellis Van Creveld syndrome, exotropia, nystagmus

A 4-year-old boy presented with a deviation of the right eye along with involuntary movements of both eyes. An ocular examination revealed right exotropia of 90 prism diopters (D) deviation, bilateral nystagmus with both eyes not able to maintain steady fixation, and right face turn [Fig. 1]. Nystagmus was spontaneously present in the primary position—constant, pendular, conjugate, horizontal, increased with distance fixation attempt and dampened with convergence. Atropine refraction revealed a -9.0 D sphere in the right eye and a -7.0 D sphere in the left eye. The anterior segment examination was normal and the fundus examination revealed a tessellated fundus with a temporal myopic crescent. On physical examination, he had coarse facies with short stature (achondroplasia) and long thorax; ectodermal dysplasia and cryptorchidism [Fig. 2]. His weight was 11.5 kg, height was 81 cm, and head circumference was 46.6 cm (all below 2 SD). The chest circumference was 43 cm, the upper segment: lower segment ratio was 1.36:1 and the arm span was 75.5 cm. The upper and lower extremities were short, clubby, and shortened out of proportion to the trunk. The hands were wider than normal and the feet were square-shaped with wide space between the hallux and the rest of the toes. As compared to his hands, the digits were proportionately short. He had thin and sparse hair and eczematous skin reaction over his forehead [Fig. 2].

An oral examination revealed delayed dentition with multiple carious teeth, hypoplastic enamel, and multiple hyperplastic labial freni attached to the gingiva [Fig. 3]. A cardiological evaluation revealed a large atrial septal defect with mild valvular pulmonary stenosis. Radiological investigations of the wrist showed only two carpal bones; indicating bone age to be 2 years while the chronological age was 4 years [Fig. 4]. The distal and middle phalanges were short compared to the proximal ones. In our case, the clinical

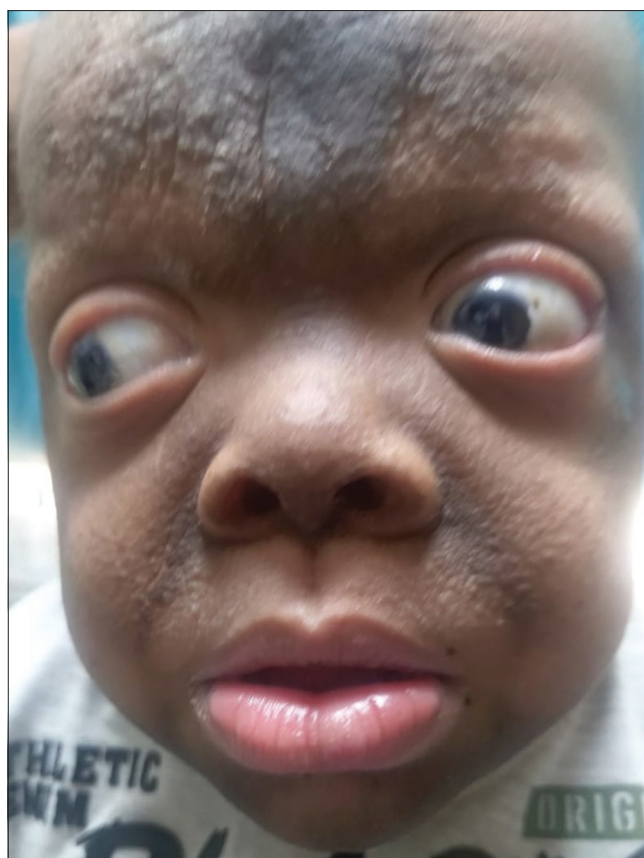


Figure 1: Right eye exotropia with right face turn

and radiological features were suggestive of Ellis Van Creveld syndrome (EVC) with right exotropia and bilateral nystagmus.

Discussion

EVC is a rare genetic disorder with autosomal recessive inheritance affecting bone growth.^[1] The prevalence rate is as low as 0.7/100,000 live births with the maximum cases reported in the Amish population in the Lancaster country, PA, USA.^[2] In India, the disorder is itself rare and only a few cases have been documented with strabismus.^[2,3] The mutation of genes EVC and EVC2/LIMBIN lying in juxtaposition on human chromosome 4p16 leads to the disorders of the primary cilium and results in a syndromic disorder called ciliopathies.^[4] This case describes a case of EVC with classic manifestations of ectodermal dysplasia (thin and sparse hair, eczematous skin, delayed dentition, multiple carious teeth, hypoplastic enamel); chondrodysplasia (short stature); and congenital heart disease (large atrial septal defect with mild valvular pulmonary stenosis).^[5] Though

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Figure 2: Short stature and long thorax



Figure 3: Multiple carious teeth with hyperplastic labial freni



Figure 4: Radiograph showing two carpal bones and short distal and middle phalanges as compared to the proximal ones

polydactyly was absent; distal and middle phalanges were short as compared to the proximal ones and radiological investigation showed only two carpal bones. However, what was interesting in this case of EVC was the presence of right eye exotropia along with nystagmus. The embryonic origin of extraocular muscles involved with the development of exotropia requires an interaction between the mesoderm and migrating neural crest cells, thereby, signifying an extended role of EVC2/LIMBIN in the craniofacial region.^[4] In this case, the nystagmus associated with strabismus also had a myopic refractive error. Hence, correcting refractive errors improves visual acuity and is important at an early age to prevent amblyopia.

Informed consent

Informed consent was taken from the patient's father.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

References

1. Valencia M, Tabet L, Yazbeck N, Araj A, Ruiz-Perez V, Charaffedine K, *et al.* Ellis-Van Creveld syndrome: Mutations uncovered in Lebanese families. *Case Rep Genet* 2015;2015:528481.
2. Chanemougananda S, Jesija JS, Bojan A, Mithra R, Swathy D. Ellis-Van Creveld syndrome: Report of two cases. *Int J Med Dent Case Report* 2014;1:1-5.
3. Das D, Das G, Mahapatra TK, Biswas J. Ellis van Creveld syndrome with unusual association of essential infantile esotropia. *Oman J Ophthalmol* 2010;3:23-5.
4. Louie K, Mishina Y, Zhang H. Molecular and cellular pathogenesis of Ellis-van Creveld syndrome: Lessons from targeted and natural mutations in animal models. *J Dev Biol* 2020;8:25.
5. Cahuana A, Palma C, Gonzales W, Gean E. Oral manifestations in Ellis-van Creveld syndrome: Report of five cases. *Pediatr Dent* 2004;26:277-82.