

Ellis-van Creveld Syndrome associated with Atlantoaxial Dislocation

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ABSTRACT

Ellis-van Creveld (EVC) syndrome is an autosomal recessive syndrome with features of chondroectodermal dysplasia. Involvement of craniovertebral (CV) junction in association with this syndrome is extremely rare. We present a case of 9-year-old boy who presented with myelopathy and the management options related to this anomaly. The atlantoaxial instability was reducible in radiological examination so posterior occipito cervical fusion was performed. Considering the fact that patients with EVC syndrome may have spinal instability especially CV junction, they should also undergo a screening X-ray of the craniovertebral junction to rule out any instability.

Keywords: Atlantoaxial dislocation, Chondroectodermal dysplasia, Ellis-van creveld syndrome, Genu valgum, Polydactyly.

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INTRODUCTION

Ellis-van Creveld (EVC) syndrome is an autosomal recessive syndrome with features of chondroectodermal dysplasia. In 1933, McIntosh first described a girl with hereditary ectodermal dysplasia, associated with polydactylism and chondrodystrophy leading to short limb dwarfism, rudimentary teeth, and nails.¹ Then it was in 1940 when Richard WB Ellis and Simon van Creveld described this syndrome in two more patients. The complete syndrome was defined as consisting of a tetrad of chondrodysplasia, ectodermal dysplasia, polydactyly, and congenital heart disease.¹ Since then about 100 cases have been reported in the literature with

variable presentation. The syndrome has been classically reported to be predominant in the Amish population of Pennsylvania in US² with a reported incidence of 1 in 5,000 births. The syndrome affects multiple organs and has a highly variable phenotype. The chondrodysplasia gives rise to multiple skeletal abnormalities which may be detected even prenatally by as early as 13th week of gestation in the form of increased fetal nuchal translucency in the first-trimester.³ After birth, the chondrodysplasia manifests as short limbed dwarfism with polydactyly of the hands as well as feet.⁴ Here, we present a case of EVC syndrome presenting with myelopathy due to atlantoaxial dislocation (AAD) which has not been previously reported in the literature and discuss its management issues.

CASE REPORT

A 9-year-old boy was brought to our outpatient clinic by the parents with a spectrum of progressive complaints. The child was born to non-consanguineous parents and none of the family member had history of similar deformities in the past. At birth, the child had bilateral upper limb postaxial (ulnar) polydactyly, left foot preaxial polysyndactyly, and right foot syndactyly. This child had delayed motor milestones and attained standing with support at 18 months and walking without support only at 24 months. At the age of 1 year, he underwent surgical correction of right undescended testis (cryptorchidism). The child had bilateral discharging preauricular sinuses which were surgically managed 4 years back. The child was reported to be shorter as compared to his peers by the parents. As he grew, it was evident that the child had short limbed dwarfism with distal limbs involved more than proximal. He had genu valgum (knock-knee) with gait unsteadiness (Figs 1A to C). The teeth were abnormally spaced and peg-like with a high arched palate. For the last 6 months, the boy had developed progressive spastic quadriparesis with Ashworth score 3. There was no associated neck pain and no restriction of neck movements. Imaging revealed a reducible AAD with severe compression of the cervicomedullary junction with fused posterior elements of C2-C3 vertebrae. The odontoid process was short and below the level of C1 arch. On dynamic imaging, the compromise of the

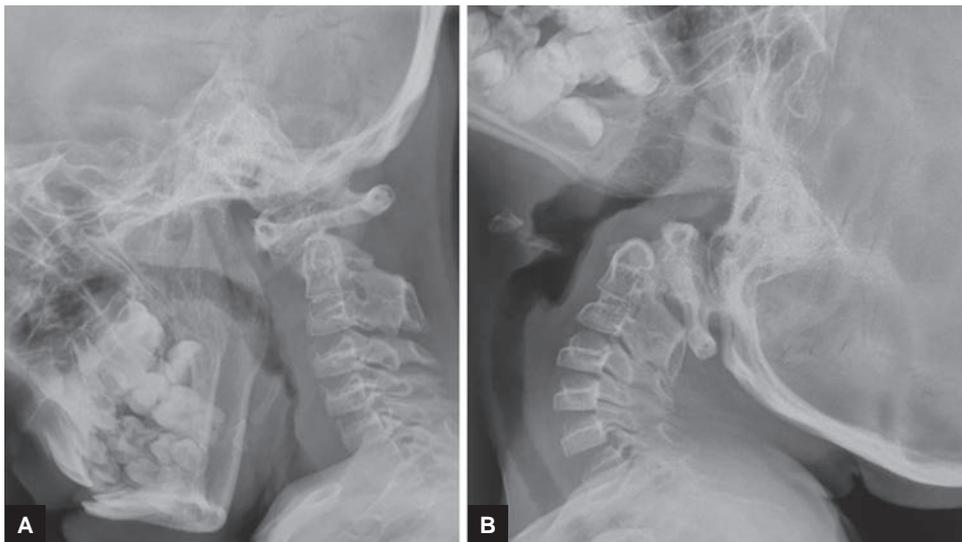
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Figs 1A to C: External morphological features of Ellis-van Creveld syndrome



Figs 2A and B: Dynamic X-rays of CV junction showing AAD

cervicomedullary region was evident. He was planned for a posterior bony decompression at the craniovertebral (CV) junction and an occipitoaxial fusion (Figs 2A and B). Considering the softness of his bones, laminar hooks were used over the C2-C3 fused laminae rather than sublaminar wires. They were then fixed with contoured rod plate assembly to the subocciput. Postoperative imaging showed satisfactory reduction and his lowerlimb tone and gait improved within the initial few days. On the last follow-up, after 1 year, the child has significant reduction in limb spasticity.

DISCUSSION

Atlantoaxial dislocation is a potentially life threatening condition which can result in sudden death due to respiratory arrest, if the cervicomedullary region is acutely compressed.⁵ Though many chondroplastic conditions have previously been described in association with EVC syndrome, AAD has not been described before in the literature. The published data on associated skeletal deformities includes short limbed dwarfism due to mesomelic shortening primarily affecting the

lower limbs.⁶ The hands are also deformed with stout appearance and post axial polydactyly. Hand polydactyly is seen in 100% cases of EVC syndrome whereas feet involvement is reported in only 10 to 25% of patients.⁷ Other associated limb anomalies include short broad middle phalanges, hypoplastic distal phalanges, malformed carpals and hypoplasia of upper lateral tibia with knock-knees.⁶ None of these are life endangering and barring the cosmetic and functional limitations, an individual with EVC syndrome can survive. None of the previously reported osteochondral deformities require an emergent treatment as is needed in a case of AAD to prevent sudden death.

Since the AAD was of reducible type, a posterior occipitoaxial fusion as a standalone procedure was enough for the child. Had it been a case of irreducible AAD, he would have required an anterior decompression as well.

CONCLUSION

This case report is unique since it is the first time that AAD has been reported in association with EVC syndrome.

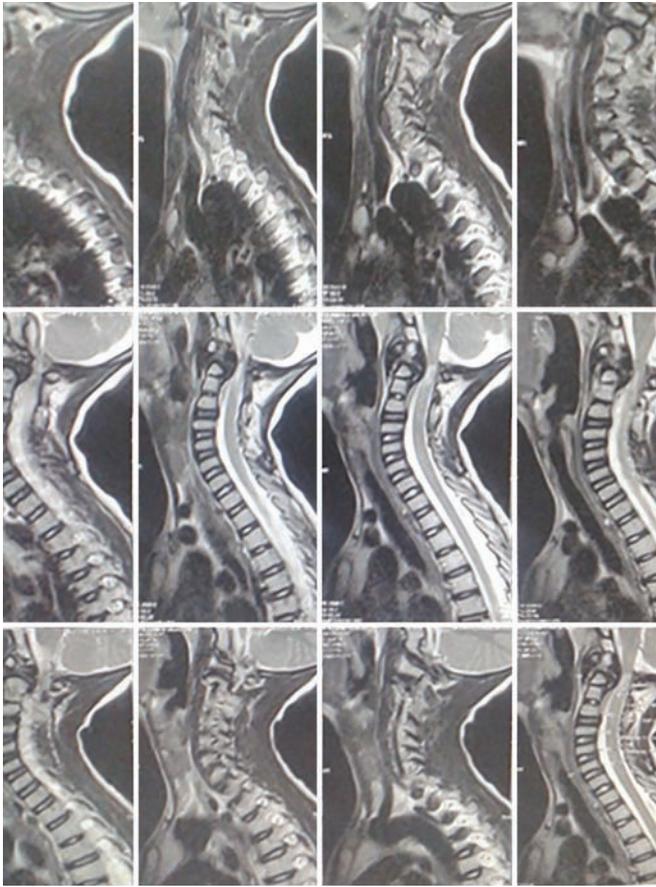
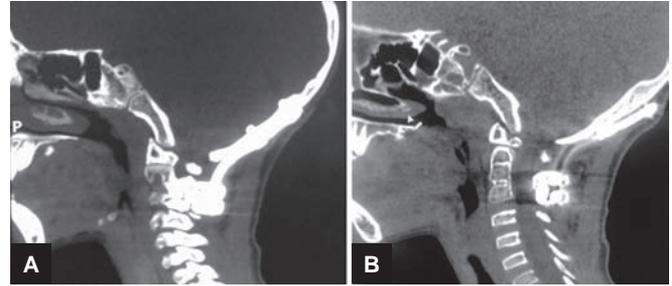


Fig. 3: Magnetic resonance imaging of the CV junction showing compression at level of CV junction

Hence, patients of EVC syndrome should also undergo a screening X-ray of the CV junction to rule out any instability (Figs 3; 4A and B).



Figs 4A and B: Post C1-C2 reduction CT scan of CV junction

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