CASE REPORT: ELLIS VAN CREVELD SYNDROME WITH A NOVEL MUTATION

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Background

Ellis Van Creveld syndrome (EVC) is a rare condition which is characterized with disproportionate short stature, postaxial polydactyly, and dysplastic nails and teeth. It is a rare autosomal recessive disorder due to mutations of EVC 1 and 2 genes located on chromosome 4p16. EVC syndrome is a chondroectodermal dysplasia. Congenital heart defects; especially atrial septal defect and single atrium occurs in 60% of affected individuals. Here we report a 5 year-old female patient admitted to our clinic with short stature. A novel homozygous mutation of EVC2 gene is detected.

Case

A 5-year-old female patient admitted to pediatric endocrinology department with the complaint of short stature. She was born at term via cesarean section. Birth height was unknown but family described short stature was prominent at birth.. She had the history of natal mandibular anterior teeth, they exfoliated spontaneously within the first month of life and non-eruption of anterior maxillary and mandibular teeth. She had typically dysplastic finger and toenails, which never needed to be cut. She was operated for postaxial polydactyly. In physical examination height 93.5 cm (- 4.57 SD), weight 16 kg (-1.72 SD), head circumference 52 cm (0.69 SD), arm length 17 cm, forearm 14 cm measured. Despite most of the affected individuals had congenital cardiac defects her echocardiography was normal. With these clinical features Ellis Van Creveld syndrome was suspected. The whole exome sequencing of EVC2 gene in this patient revealed homozygous p.Gln1074* (c.3220 C>T) mutation. This mutation has not been described in the human gene database previously.

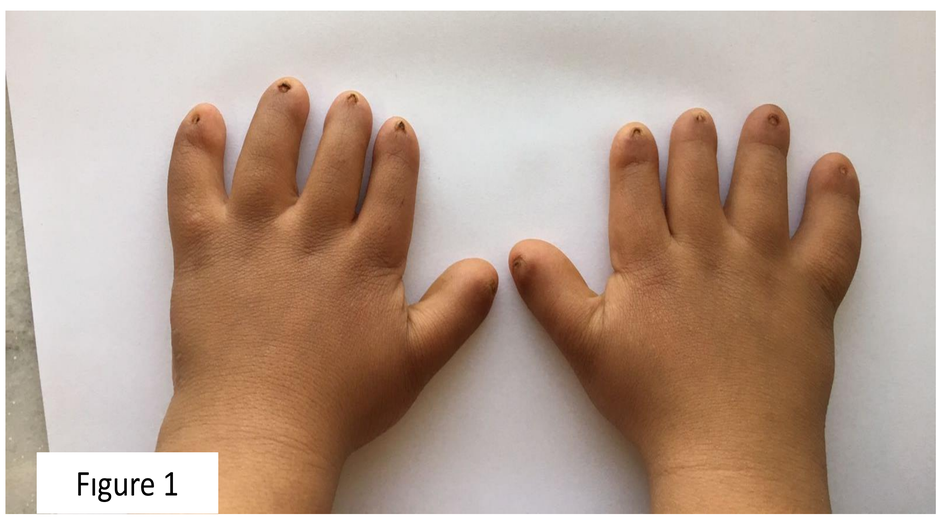

Figure 1

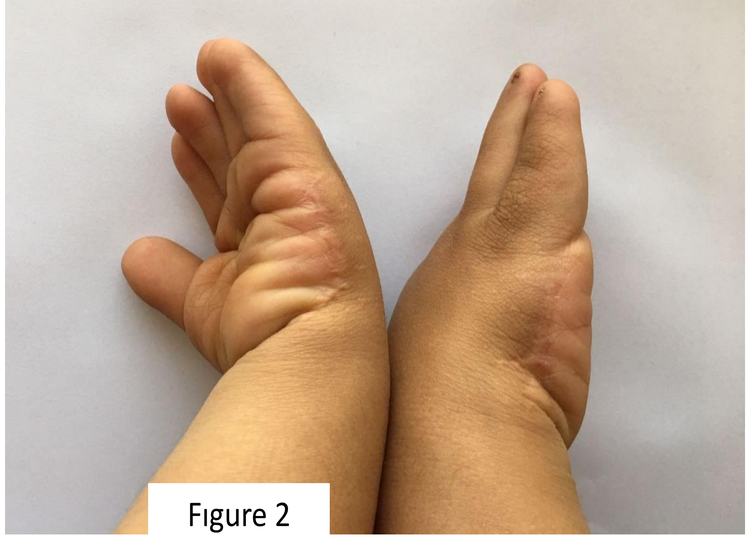

Figure 2

Figure 1. Dysplastic finger nails
Figure 2. Operation scar of polidactyly

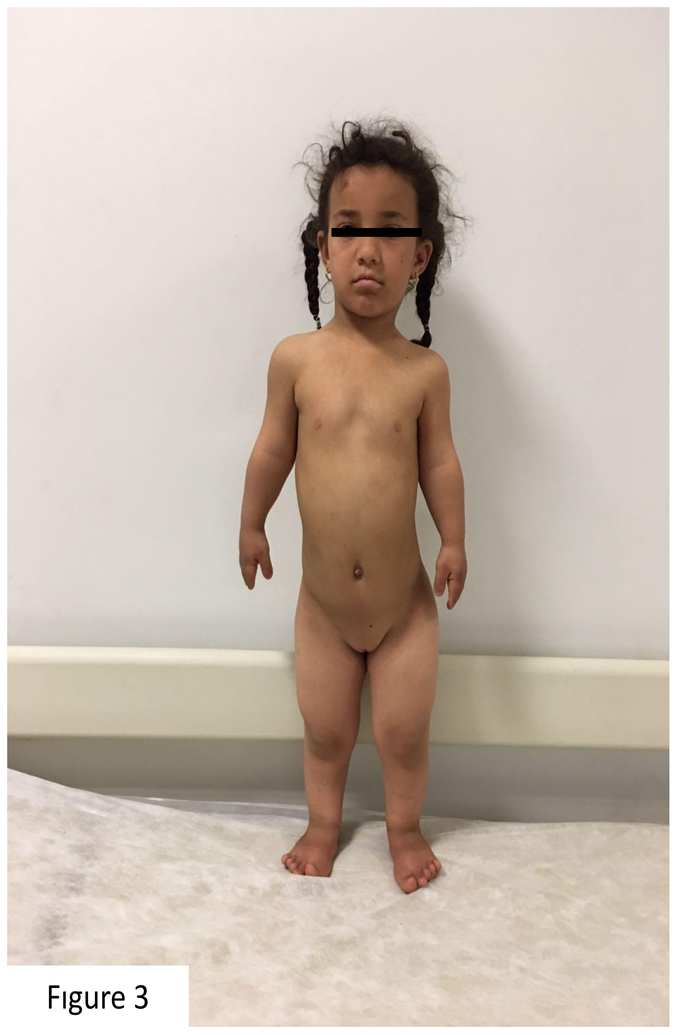

Figure 3

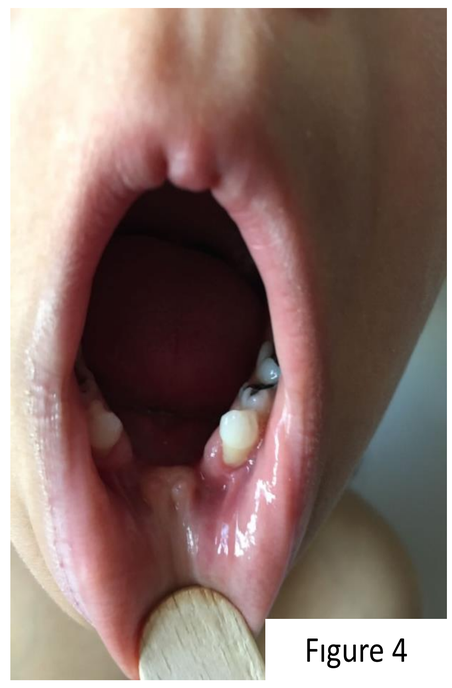

Figure 4

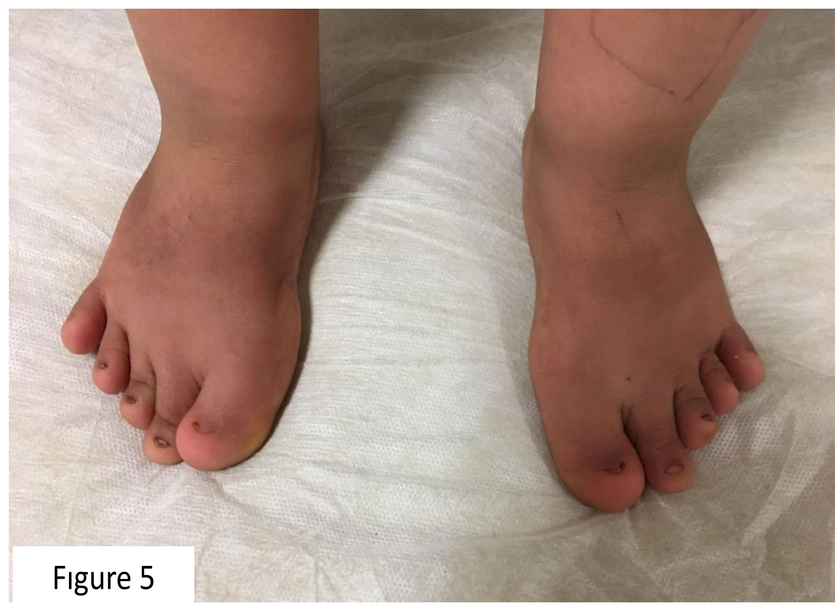

Figure 5

Figure 3. Disproportionate short stature
Figure 4. non-eruption of anterior maxillary and mandibular teeth
Figure 5. Dysplastic toenails

Results

The EVC syndrome (OMIM #225500) is a rare autosomal recessive disorder. Despite it is a rare disorder of chondroectodermal tissue,it is more common in communities where consanguineous marriages are frequent. The diagnosis of the EVC syndrome is based on the clinical and radiographic findings of skeleton. Disproportionate short stature, postaxial polydactyly, displastic nails and teeth, congenital heart defects are main features.. The direct sequencing of the EVC syndrome 1 and 2 genes may also be performed. We detected a novel homozygous mutation in EVC2 gene. EVC syndrome must be kept in mind in the patients who has ectodermal dysplasia associated to disproportionate short stature.

References

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2. Tompson, S. W. J., Ruiz-Perez, V. L., Blair, H. J., Barton, S., Navarro, V., Robson, J. L., Wright, M. J., Goodship, J. A. **Sequencing EVC and EVC2 identifies mutations in two-thirds of Ellis-van Creveld syndrome patients.** Hum. Genet. 120: 663-670, 2007.

57ESPE 269--P2
Growth and syndromes (to include Turner syndrome)
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Poster presented at: 57th ESPE 2018 Meeting ATHENS GREECE 27-29 September 2018
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