ABSTRACT

Ellis-van Creveld syndrome is a rare autosomal recessive disorder characterized by a narrow thorax with short ribs, short extremities with polydactyly, and heart defects.

CASE REPORT

A 29-year-old woman, gravida 2, para 1, was referred for ultrasound level 2 at 32 gestational weeks because of suspected fetal skeletal dysplasia. Prenatal ultrasound revealed shortening of all the long bones (-7SD), bilateral postaxial polydactyly, a narrow thorax, primum atrial septal defect, persistent left super vena cava and hypospadias.

Due to a chest-to-abdominal circumference ratio <0.6 strongly suggesting a perinatal lethal condition, the couple was counseled and opted for termination of pregnancy. At the delivery, a 1300-g male fetus with all prenatal US aspects and dysplastic teeth were confirmed.

Postmortem X-ray exploration showed micromelia associated with narrowing bell-shaped thorax, trident-shaped pelvis, an acetabular roof with medial spurs, handlebar clavicles and iliac bones of square shape.

The molecular analysis by clinical exome sequencing revealed a compound heterozygosity of two novel mutations in the EVC gene:

(NM_153717.2(EVC):c.758C>G(p.Ser253Ter) and NM_153717.2(EVC):c.2376del(p.Tyr793IlefsTer20)).

This genetic finding confirmed the clinical diagnosis of Ellis-van Creveld syndrome. The trio exome sequencing after that confirmed the parental origin of these mutation.

CONCLUSION

Prenatal sonographic identification of atrial septal defect in association with shortening of the long bones should alert clinicians to the possibility of EVC syndrome and prompt a careful research of polydactyly of the hands. These novel mutations of the EVC gene warrant an appropriate genetic counseling for next pregnancy.

dr.votason@gmail.com