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Anna Zwierzyńska[1,A,D,F], **Grzegorz Świercz**[2,D-F], **Mariola Głowacka**[3,D-F], **Jakub Gruszka**[4,A,D,F], **Olga Adamczyk-Gruszka**[5,A,D-F]

THE ELLIS VAN CREVELD SYNDROME

Zespół Ellisa van Crevelda

[1] Collegium Medicum, Uniwersytet Jana Kochanowskiego w Kielcach, Polska
[2] Klinika Położnictwa i Ginekologii, Wojewódzki Szpital Zespolony, Kielce, Polska
[3] Collegium Medicum, Wydział Nauk o Zdrowiu. Katedra Zintegrowanej Opieki Medycznej, Zakład Pielęgniarstwa,, Akademia Mazowiecka w Płocku, Polska, Polska
[4] II Katedra i Klinika Położnictwa i Ginekologii, Wydział Lekarski, Warszawski Uniwersytet Medyczny, Polska
[5] Katedra Ginekologii i Położnictwa, Collegium Medicum, Uniwersytet Jana Kochanowskiego w Kielcach, Polska

A - Koncepcja i projekt badania, B - Gromadzenie i/lub zestawianie danych, C - Analiza i interpretacja danych, D - Napisanie artykułu, E - Krytyczne zrecenzowanie artykułu, F - Zatwierdzenie ostatecznej wersji artykułu

Abstract (in Polish):
Wstęp: Zespół Ellisa van Crevelda to rzadka choroba genetyczna. Przybiera postać dysplazji chondroektodermalnej z charakterystycznym obrazem klinicznym. Do charakterystycznych objawów należy nieproporcjonalnie niski wzrost, heksadaktylia w dłoniach, rzadziej w stopach; dysplazja ektodermalna wpływająca na paznokcie, włosy, zęby; wrodzone wady serca (wady zastawki mitralnej i trójdzielnej; przetrwały przewód tętniczy, ubytek w przegrodzie międzykomorowej/przedsionkowej); ubytki w jamie ustnej (nieprawidłowy zgryz, zrosty i przerost dziąseł, przerost wędzidełka, hipoplazja szkliwa). Zdecydowana większość przypadków choroby dotyczy osób z mutacjami w obrębie genu EVC lub EVC 2, zlokalizowanego w locus 4p16.2. Jest to znacznie bardziej powszechne w społeczności Amiszów.

Cel pracy: W artykule przedstawiono przypadek pacjenta z EVC.
Opis przypadku: W diagnostyce prenatalnej wykonanej na 22 1/7 hbd u dziecka stwierdzono polidaktylię pozaosiową obu stóp. Rozpoznano u niego wrodzoną wadę serca - przetrwały otwór owalny. W opisywanym przypadku liczne zmiany w jamie ustnej zostały znalezione.
Dyskusja i wnioski: Zespół Elisa van Crevelda (EVC) to rzadka choroba genetyczna dziedziczona autosomalnie recesywnie. Pierwszy raz opisana przez Richard W.B. Ellis of Edinburgh and Simon van Creveld of Amsterdam w 1940 roku.]. Częstość występowania choroby szacuje się 7 / 1 000 000. Jest znacznie częstsza w społeczności Amiszów, co związane jest z pokrewieństwem rodziców. Opisany pacjent również należy do typowej manifestacji klinicznej choroby, jednak niektóre objawy mogą pojawić się w późniejszych latach. Konieczne jest ścisłe monitorowanie rozwoju tego dziecka.

Abstract (in English):
Introduction: Ellis van Creveld Syndrome is a rare genetic disorder. It takes the form of chondroectodermal dysplasia with a characteristic clinical manifestation. Incidence of the disease is 7 / 1,000,000. The characteristic symptoms include disproportionately shortness of stature, hexadactyly in the hands, less often in the feet ; ectodermal dysplasia affecting nails, hair, teeth; congenital heart defects (mitral and tricuspid valve defects; patent ductus arteriosus, ventricular/atrial septal defect); cavities in the oral cavity (mal-occlusion, adhesions and gingival hypertrophy, frenulum hypertrophy, enamel hypoplasia). The vast majority of cases of the disease concern people with mutations within the EVC or EVC 2 gene, located at the 4p 16.2 locus. It is much more common in the Amish community.
Aim: The article presents a case of a patient wEVC.
Clinical case: During the prenatal diagnosis, performed on the 22 1/7 hbd, the child was diagnosed with extra – axial polydactyly on both feet. He was diagnosed with a congenital heart defect - a patent foramen ovale. In the case described, numerous changes in the oral cavity were found.
Discussion and conclusions: The patient described is also part of the typical clinical manifestation of the disease, but some symptoms may appear in later years. It is necessary to closely monitor the development of this child.

Keywords (in Polish): badania prenatalne, wady wrodzone, choroby genetyczne, EVC.

Keywords (in English): birth defects, genetic diseases, EVC, prenatal tests.

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Short title
Zespół Ellisa van Crevelda

Corresponding author
Anna Zwierzyńska
Collegium Medicum, Uniwersytet Jana Kochanowskiego w Kielcach, Polska;
email: anna.zwierzynska@o2.pl
Phone: +48666047755

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Introduction

The Ellis van Creveld syndrome was firstly described by Richard W. B. Ellis of Edinburgh and Simon van Creveld of Amsterdam in 1940. EVC is a rare genetic disorder inherited autosomal recessively. It takes the form of chondroectodermal dysplasia with a characteristic clinical manifestation [1]. The EVC phenotype affects many organs. The main symptoms are shortness of stature, short ribs, polydactyly and a congenital structural heart defect. Apart from symptoms related to growth whilst developing, facial dysmorphia and dental anomalies are also observed. The vast majority of cases of the disease concern people with mutations within the EVC or EVC2 gene, located at the 4p16.2 locus [2]. These genes code the ciliary proteins of the basic body [3]. The prevalence of the disease is estimated at 7/1,000,000. It is much more common in the Amish community due to the relationship of parents [4,5].

Aim

The article presents a case of a patient with EVC.

Clinical case

A patient in the second pregnancy, gave birth to 1 child in 2019 by Caesarean section for obstetric indications. In the second pregnancy, during the prenatal diagnosis, performed on the 22 1/7 hbd, the child was diagnosed with extra – axial polydactyly on both feet (Fig.1.) – that is, an extra sixth toe, alongside the fifth toe, on each foot; no other organs showed changes in ultrasound diagnosis, nor was non-axial polydactyly diagnosed, in either hand.

In the 39th week of pregnancy, the pregnancy was terminated by Caesarean section, due to the condition of the mother, *post*-Caesarean section.

One live, male foetus was born in good general condition with a birth weight of 3810 g and an Apgar rating of 10.

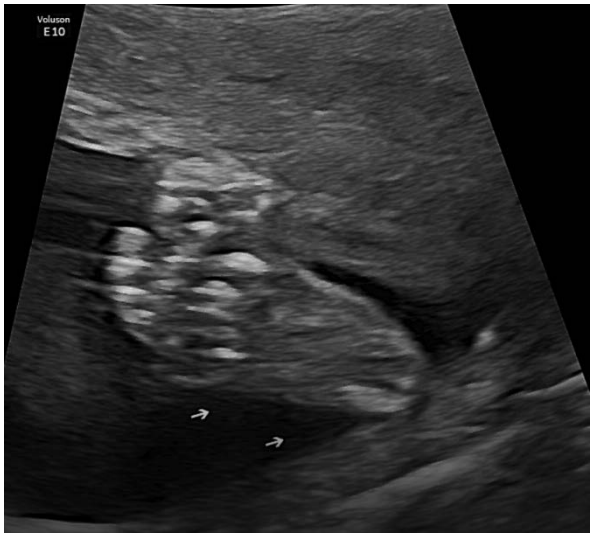

Fig.1. Ultrasound photo of the patient

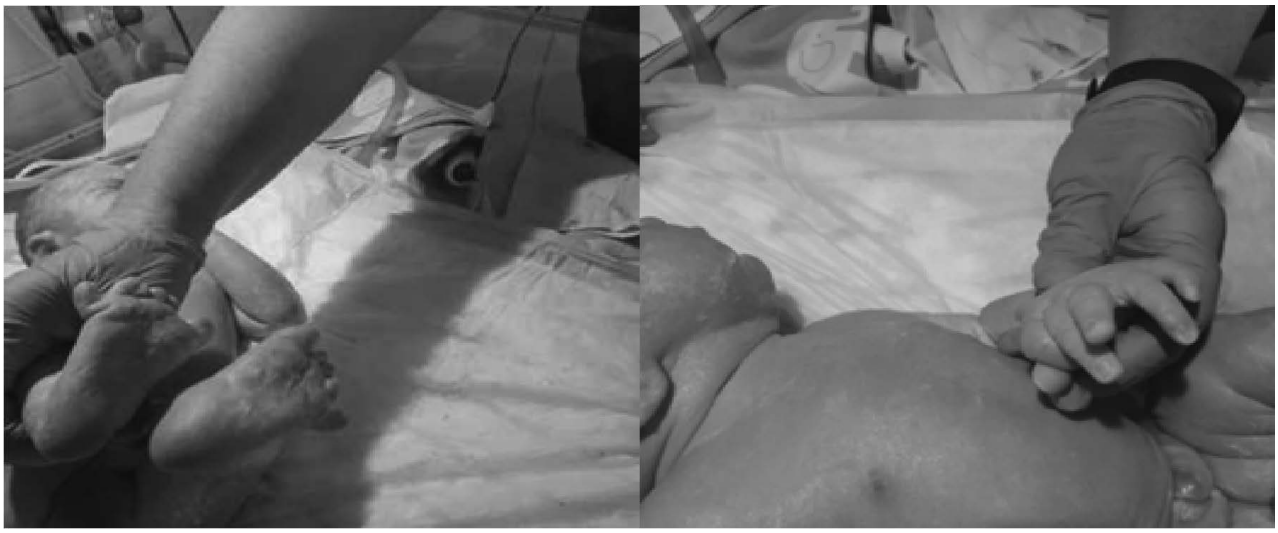

Fig. 2. The lower limbs and the hand of the patient

After birth, additional fingers and toes were found on both sides of the feet and hands (Fig.2.). The adaptation processes in the ward went well. After delivery, Crede's Procedure and hearing tests were performed using the otoacoustic emission method; vaccinations against hepatitis B and BCG were also administered, in accordance with the vaccination schedule. Screening for phenylketonuria, cystic fibrosis, hypothyroidism and metabolic diseases, as well as a screening echo of the heart, ultrasonography and ultrasound of the abdomen and retroperitoneal space were also performed. The echocardiographic examination revealed an abnormality in the form of an oval hole with a width of about 3mm with left-right bleed. An unlocked FO was identified. Other parameters of the heart were without the characteristics of pathology. Further care in a cardiac clinic was recommended.

In the ultrasound examination of the abdomen and retroperitoneal space, among the pathological changes, there was a partially post-renal, renal pelvis with a thickness of about 4.5 mm; chalices were not extended; other organs of the abdominal cavity did not show pathological deviations. In the trans-gland ultrasound examination, the images of the ventricular system, brain, choroidal plexuses and lateral ventricles were correct.

The newborn, with Ellis van Creveld syndrome, was discharged home on the fourth day of life in good general condition, with recommendations to be followed up at the Nephrology and Cardiology Clinic.

Discussion and conclusions

Ellis van Creveld's disease is a syndrome first described in 1940. Most cases are described in the Amish Society [6]. The characteristic symptoms include disproportionately shortness of stature, hexadactyly in the hands, less often in the feet; ectodermal dysplasia affecting nails, hair, teeth; congenital heart defects (mitral and tricuspid valve defects; patent ductus arteriosus, ventricular/atrial septal defect); cavities in the oral cavity (mal-occlusion, adhesions and gingival hypertrophy, frenulum hypertrophy, enamel hypoplasia) [7].

The patient described shows characteristic features in the upper and lower extremities *viz.,* hexadactyly. He was diagnosed with a congenital heart defect - a patent foramen ovale. Polydactyly was also found in the patient described by Pena-Cardelles *et al.* [8]. In the case described, numerous changes in the oral cavity were found.

The child's prenatal examination showed only hexadactyly in the feet. IUGR features were not found. The patient was described by Alessa *et al.* [9]. The ultrasound examination in 34 hbd showed that the lengths of the femur and humerus were too short for the gestational age. The child showed the typical tetrad of symptoms mentioned in the literature. Intrauterine diagnosis of EVC is in most cases limited to USG in the second and third trimesters. The examination reveals features that may indicate the presence of Ellis van Creveld syndrome in the child, i.e., shortness of limbs, hexadactyly, congenital heart defects, narrow chest or IUGR. In rare cases, this is detected in the first trimester. Due to the genetic basis of the disease, it is recommended to extend diagnostics to include molecular testing [10].

Ellis van Creveld's syndrome can be characterised by a variety of clinical manifestations. The vast majority of cases show features falling within the group of the four main changes, characteristic of this disease and have a genetic basis. The patient described is also part of the typical clinical manifestation of the disease, but some symptoms may appear in later years. It is necessary to closely monitor the development of this child.

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