Ovarian endometriotic cyst in a patient with Ellis-van Creveld's syndrome: case report

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Summary

Ellis-van Creveld (EvC) syndrome, or chondroectodermal dysplasia, is a rare genetic disorder associated with chondrodysplasia, ectodermal dysplasia, polydactyly, and congenital cardiac malformations. The disorder is due to an autosomal-recessive mutation mapped to chromosome 4p16. It may occur with different phenotypes. The case of an ovarian endometriotic cyst in a patient suffering from EvC syndrome is reported.

Key words: Ovarian cyst; Ellis-van Creveld's syndrome.

Introduction

Ellis-van Creveld (EvC) syndrome is a rare autosomal recessive disease also known as chondroectodermal dysplasia [1]. EvC is characterized by a tetrad of manifestations including disproportionate dwarfism, ectodermal dysplasia, cardiac malformations, and polydactyly [2]. The physical features of patients suffering from this disease are typical and characteristic: long trunk with short limbs, dystrophic nails, extra fingers/toes, teeth malformations (taurodontism, missing incisors), harelip, narrow rib cage (barrel chest with short ribs), single atrium (interatrial and interventricular defects), delayed endochondral ossification, and nephrotic syndrome. The chromosome alteration appears as a mutation of the short arm (p) of chromosome 4 (cr4p16). The heterozygous condition involves polydactyly and cardiac malformations, but not necessarily short stature [3]. It can be diagnosed echographically from the 27th week of pregnancy and is not a life threatening disorder. Prenatal diagnosis is important both for family pathology and for early surgical correction of cardiac defects and osteochondrodysplastic abnormalities. As with all diseases, there are variants of the classic symptomatology. In the literature, variants have been described which were associated with kidney and liver abnormalities, changes in the central nervous system, and mucometrocolpos [4]. We report a case of a 19-year-old patient suffering from an ovarian cyst associated with EvC syndrome, both being dysontogenetic disorders.

Case Report

A 19-year-old hexadactyly patient with a history of cardiac disorders and suffering from EvC syndrome was admitted to our hospital to undergo a laparoscopic intervention for removal of an ovarian cyst. Anamnestic data indicated a familial propensity for heart disease. Surgical correction of the cardiopathy (partial AV channel, single atrium, unroofed superior vena cava, no venous innominate trunk) had been performed when the patient was 12 years old. Subsequently, her bilateral hexadactyly with dystrophic toenails was also corrected. The patient was 1.60 m in height as her case of heterozygous EvC syndrome was not associated with disproportionate dwarfism. She presented the typical craniofacial morphology: small posterior cranial base, small jaw, and large lower jaw with an increased angle (malocclusion). Dental anomalies could be observed: shovel-shaped incisors, claw pointed teeth, reduced size of the crown and supernumerary teeth.

When admitted to the hospital the patient underwent electrocardiography and hematochemical tests which showed normal parameters and negative tumor markers. Pelvic echography showed a 58 x 47 cm right adnexal ovular nonhomogeneous mass consisting of both non-echogenic and echogenic areas with irregular but clear margins. There was fluid in the pouch of Douglas. After receiving preventive antibiotic therapy, the patient underwent laparoscopic removal of the endometriotic cyst in the right ovary. No complications were observed during the surgical intervention or postoperatively. She was discharged after 24 hours. Pathological examination confirmed the diagnosis of ovarian endometriosis. Cytological and bacteriological examinations of the peritoneal fluid were negative.

Discussion

Given that the etiology of endometriosis is considered by some to be a genetic immune disorder, it is interesting to note that in this case this etiology was associated with an autosomal recessive disorder [5]. It is important that researchers describe cases associated with rare syn-

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dromes that even through today, given the advances in surgery, particularly heart surgery, are no longer life threatening. Patients with pathologies, once described as very rare, are today becoming more common. Our patient, in addition to the esthetic damage due to the median thoracotomy and polydactyly, could in the future have had more problems related to endometriosis: infertility, chronic pelvic pain and, as the disease progresses, dysmenorrhea, dysuria, dyschezia, and pelvic adhesion syndrome.

In this case the endometriosis was still in an early stage (Ib), the patient's pelvis had no tenacious adhesions and thus stripping the cystic wall was easy. We believe that the publication of this unusual case will be useful as we found no reports in the literature describing any association of these diseases [6-9].

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