Common Atrium Associated with Polydactily and Dwarfism in Middle Age Male Patient

Daut R. Gorani1, Lulzim S. Kamberi1, Nora S. Gorani2
Cardiology department, University Clinical Center, Prishtina, Kosova1
Cytogenetic department, Clinic for Gynecology and Obstetrics, University Clinical Center, Prishtina, Kosova2

Polydactyly associated with dwarfism may serve as a hint for the presence of additional congenital cardiac abnormalities, thus rousing the demand for a detailed cardiac and genetic investigation. In our case, echocardiography findings led to the diagnosis of most likely Ellis-van Creveld syndrome. We may conclude that prenatal diagnosis of the syndrome can be readily achieved by fetoscopy, fetal echocardiography, and molecular genetic testing by amniocentesis or DNA extracted from chorionic villus samples. Prenatal diagnosis can also be established using mutation analysis of EVC gene from fetal DNA. These cases emphasize the importance of fetal examination for accurate diagnosis of rare syndromes. Education of the general public, especially parents, on congenital anomalies as well as improvement of medical and diagnostic facilities is therefore suggested, if not demanded.

Key words: Cardiac abnormalities, polydactyly, dwarfism.

Corresponding author: Daut R. Gorani, MD. Cardiology department. University clinical center of Prishtina, Kosova.

1. INTRODUCTION
A congenital anomaly may be viewed as a physical, metabolic, or anatomic deviation from the normal pattern of development. By definition, congenital anomalies are existent at birth or at least become manifest during the neonatal period.

For one reason or another, however, some remain undetected beyond the early days of life. Usually the factors leading to the late diagnosis are: a) the nature and site of anomaly; b) beliefs, customs, and enlightenment of the society; and c) inadequate medical facilities and expertise.

The late diagnosis invariably culminated in high morbidity and mortality, problems of management, and social problems (1). In our case all three factors are linked-up. Therefore, we have got a patient at the age of 54, with many features of most likely Ellis-van Creveld syndrome.

2. CASE PRESENTATION
We report a 54-year-old Caucasian male that presented in our hospital severely ill suggesting of acute pulmonary edema and heart failure, sitting bolt upright complaining on sudden breathlessness followed by other concomitant disorders: tachypnea, cyanosis, productive cough, marked perspiration, chest pain, evident bilateral lower extremity swelling. After the treatment in Emergency Department patient’s condition improved and he was admitted to Cardiology Department, for further diagnosis and treatment. He was a farmer and a father of six healthy children. He didn’t smoke or use alcohol. He was symptomatic since childhood treated as chronic pulmonary disease. He used medications irregularly. No data about the consanguineous parents, and the family history was unremarkable with regard to congenital defects. The patient was 155 cm height, and weighted 65 kg (Figure 1a, 1b); body surface area was (BSA) 23.7; Blood pressure on admission was 100/60 mmHg; pulse rate 138 beats/min.; respiration rate 26/min; body temperature was 36.5°C. Cylindrical thorax with the excavation in the...
CASE REPORT

Common Atrium Associated with Polydactyly and Dwarfism in Middle Age Male Patient

mid sternal part was noted. Postero-anterior convexity of spine column was present. Heart auscultation revealed irregular heartbeats, and systolic murmur was present on the whole precordium. Wheezing mixed with end-inspiratory crackles was heard on auscultation. Expectoration of pink, frothy and bloody tinged sputum was present. Jugular venous distension, hepatic-jugular reflux, lower extremity edema were noted. Liver enlargement with mild tenderness was present on palpation. Other parts of abdomen soft, active bowel sounds, no tenderness, and no pulsatile mass, good pulses throughout, bilateral post axial hexadactyly on hands (additional finger at the ulnar side), and central hexadactyly on feet (doubled second fingers) (Figure 2a, 2b). On ECG presented: atrial fibrillation, right bundle branch block (RBBB) and left anterior hemi block (LAH) (Figure 3). Chest X-ray survey signified enormous cardio-megaly and pulmonary redistribution, (Figure 2a, 2b). Laboratory findings revealed poliglobulia (RBC=5.88x10³, 1); increased hemoglobin 22.7g/l; high hematocrit =72.1 %), increase in white blood cells (WBC), (11.9x10⁹); elevated fibrinogen level (5.4g/L); elevated lactate dehydrogenate (LDH), (519IU/L); decrease of albumins (29g/L). Other laboratory examinations were normal.

Visualization of the heart by trans-thoracic echocardiography was suboptimal; therefore, transesophageal echocardiography (TEE) was performed. Transthoracic and TEE revealed a complete lack of any atrial septal tissue and enlarged common atrium 10x11mm (Figure 3a i 3b), which was not suspected in the previous medical examinations; hypertrophy of the left ventricle and enlargement of the right ventricle, tricuspid valve regurgitation 3+, and mitral valve regurgitation 2+, (Figure 3a, 3b), elevated pulmonary artery systolic pressure (PASP) of 66 mmHg; Mitral and tricuspid valve attachments to the interventricular septum were in the same anatomic plane (Figure 3a, 3b). Abdominal ultrasonography revealed situs solitus and enlarged liver, and showed no abnormalities of spleen, pancreas or kidneys. On both legs, valgus deformity of the knees was apparent. In Kosovo we were unable to carry out genetic research, therefore we have no DNA confirmation of diagnosis.

During the in hospital treatment with cardio tonics, diuretics, aspirin, aminophylline, antibiotics the condition of patient was improved.

Two months follow up resulted in good condition without relapse of acute distress.

3. DISCUSSION

To our knowledge, as described before in the medical literature, common atrium, polydactyly and dwarfism are often associated in some syndromes more frequent of which in syndrome called Ellis-van Creveld. In 1940, Ellis and van Creveld described the syndrome that they termed chondroectodermal dysplasia (2). Ellis-van Creveld syndrome (EVC) is a chondral and ectodermal dysplasia characterized by short ribs, polydactyly, growth retardation, and ectodermal and heart defects (3). Dysplastic nails and teeth are the salient features of the EVC syndrome (4). The exact prevalence is unknown, but the syndrome seems more common among the Amish community, the incidence is estimated at 5 cases per 1000 live births (2, 3). The birth prevalence in non-Amish is estimated to be 7/1,000,000 of life births (5, 6). Frequency of EVC syndrome is equal in males and females (2). Prenatal abnormalities (that may be detected by ultrasound examination) include narrow thorax, shortening of long bones, hexadactyly and cardiac defects. After birth, cardinal features are short stature, short ribs, polydactyly, and dysplastic fingernails and teeth. Congenital heart malformations are described in a 50 to 60% of patients affected by this syndrome. Defects of the mitral and tricuspid valves, patent ductus arteriosus, ventricular septal defects and atrial septal defects are some of the malforma-

Figure 2. 2a and 2b.Chest roentgenograms in posteroanterior (PA) (4a), and lateral view (4b) revealed generalized cardiac enlargement indicating cardiomyopathy.

Figure 3a and 3b: Surface echocardiography (5a) performed in apical view, and trans esophageal echo-cardiography (5b) showed common atrium (CA), mitral valve regurgitation (MR), tricuspid valve regurgitation (TR), an intact interventricular septum (IVS), pericardial effusion, and a dilated right ventricular chamber. LV: left ventricle, RV: right ventricle, CA: common atrium, MR: mitral valve regurgitation, TR: tricuspid valve regurgitation, PE: pericardial effusion, IVS: interventricular septum.
tions described as the principal cause of decreased life-expectancy in these patients. Cognitive and motor development is normal (3, 6, 9).

Chondrodystrophy is the most common clinical feature, affecting the tubular bones producing a serious ossification defect (7, 8). In consequence, the distal extremities of the limbs are short and the patients small in stature (7). The prognosis for this syndrome is related to the difficulty in breathing during the first months of life which derives from potential heart problems and a narrow thorax (3).

This rare condition is inherited as an autosomal recessive trait with variable expression.

EVC syndrome results from defects in one of two Ellis van Creveld syndrome genes (EVC1 and EVC2). The two genes lie next to each other on chromosome 4p16 (2, 3).

EVC belongs to the short rib–polydactyly group (SRP). These SRPs are all autosomal recessive disorders that have been classified into types (Saldino-Noonan syndrome, type I; Majewski syndrome, type II; Verma-Naumoff syndrome, type III; Beemer-Langer syndrome, type IV; and Jeune Dystrophy). They are characterized by hypoplastic thorax due to short ribs, short limbs, frequent polydactyly and visceral abnormalities, and are discussed paren tally. Radiographically and histologically, SRP III (Verma-Naumoff syndrome, OMIM 263510) most resembles some forms of EVC (3, 10, 11).

Postnatally, the essential differential diagnoses include Jeune dystrophy, Mckusick-Kaufman syndrome and Weyers syndrome (3, 12, 13). There are no specific constant features to confirm the diagnosis of presumptive EVC but some features, including congenital heart disease, supernumerary digits and ectodermal dysplasia will mostly support the diagnosis of EVC syndrome than Jeune dystrophy. EVC and Mckusick-Kaufman syndrome (MKK, MIM 236700), both recessively inherited disorders, share postaxial polydactyly and congenital heart defect. Distinguishing characteristics are the osteochondrodysplasia and ectodermal anomalies in EVC syndrome, and hydrometrocolpos in MKK syndrome. MKK is caused by mutations in a gene on chromosome 20p12, encoding a protein similar to members of the chaperonin family. Mutation in the same gene causes Bardet-Biedl syndrome (12, 13, 14). Weyers acrodental dysostosis (OMIM 193530) is, as mentioned above, the heterozygous manifestation of the EVC gene. Disproportionate dwarfism, heart defect and thoracic dysplasia are not present in this autosomal dominant condition.

We coincide with all published studies in regard to the cardiac, stature, and polydactyly features presented in these patients, therefore, we believe that our patient belongs to a EVC syndrome.

4. CONCLUSION

Polydactyly associated with dwarfism may serve as a hint for the presence of additional congenital cardiac abnormalities, thus arousing the demand for a detailed cardiac and genetic investigation. In our case, echocardiography findings led to the diagnosis of most likely Ellis-van Creveld syndrome. We may conclude that prenatal diagnosis of the syndrome can be readily achieved by fetoscopy, fetal echocardiography, and molecular genetic testing by amniocentesis or DNA extracted from chorionic villus samples. Prenatal diagnosis can also be established using mutation analysis of EVC gene from fetal DNA. These cases elucidate, once again, the importance of fetal examination for accurate diagnosis of rare syndromes. Education of the general public, especially parents, on congenital anomalies as well as improvement of medical and diagnostic facilities is therefore suggested, if not demanded.

Abbreviations
EVC: Ellis van Creveld Syndrome; RBBB: Right Bundle Branch Block; LAH: Left Anterior Hemi block; PASP: pulmonary artery systolic pressure; LDH: lactate dehydrogenate; WBC: white blood cells; RBC: red blood cells; CA: Common Atrium.

REFERENCES