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Uncommon cardiac malformation in a rare genetic disease original case report

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Objective: The main reason for the presentation of this clinical case was to discover the hidden diagnose of a young patient, who has presented a lipothymia during the sports class, while participating in the running trial.

Material and Method: I am presenting the clinical case of a young man aged 24, who during the sports class in the university while he was participating in the running trial; suddenly he gets dizziness, accelerated heart rate, chest pain and a lipothymia, without losing consciousness. His colleagues intend to call the emergency service, but the young man suddenly gets better and refuses to be committed to the hospital. After this incident he presents other two episodes of dizziness and accelerated heart rate, during the sports class, he goes to the family doctor and this send him to a specialist. During the consultation, I founded at the objective examination rhythmic heart sounds HR=82/min, mid systolic click in the mitral area, proto systolic murmur without irradiation, with character of vapor saltation, BP=120/80mmHg, normal vesicular sound. At the general objective examination the following are determined: the thorax shape-pectus excavatum and scoliosis, longline aspect, arachnodactylia, ligamentous hyper laxity, ogival palatal arch, dental malformations, blue sclera and wearing glasses. The EKG shows a sinus rhythm HR=78/min and a minor right branch block, the cardiac Doppler echography shows a prolapse mitral valve, second degree mitral failure and unexpectedly a large interatrial septum aneurism and an interventricular septum aneurism. The thoracoabdominal CT that has been made was within normal limits. The postoperative evolution of the patient was favorable after the interatrial and interventricular septums have recasted.

Result and Discussion: The mitral valve prolapse and the mitral regurgitation are diseases that are frequently observed in the Marfan syndrome, but the association with a interatrial and interventricular septums aneurisms is very uncommon and rare, actually.

Conclusion: 1.The Marfan syndrome represents known but a very rare a genetic disease. 2. The mitral valve prolapse diagnose associated or not with a mitral failure is also usually. 3. The interatrial and interventricular septums aneurisms are very uncommon. 4. The diagnose is possible if the genetic sub layer of the disease is taken into account and specially the presence of the lax connective tissue in a large amount and ligamentous hyperlaxity, which could be valid in the case of interatrial sand interventricular septums, which are usually more lax and with an exaggerated mobility and can produce these kind of changes. 5. The thoracoabdominal CT has not shown any other aneurism in the aorta artery level. 6. The postoperative evolution has be favorable with the recast of the interatrial and interventricular septums.

Biography

Manuela Stoicescu was Assistant Researcher at University of Cluj Napoca and now she is consultant internal medicine physician, Ph.D., Assistant Professor of University of Oradea, Faculty of Medicine and Pharmacy, Medical Disciplines Department, Romania. Also work at Emergency Hospital Internal Medicine Department and Internal Medicine Office. She has published two books, one monograph and papers in reputed journals. She was invited as a speaker at 9 national and 15 International conferences. She is Member of Romanian Society of Internal Medicine, Cardiology, Medical Chemistry, Biochemistry and Member of the Balkan Society of Medicine.

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