

**Cardiovascular Features of Marfan Syndrome at Children in North-East Region of the Romania - 10 years study**

Luca A.C.(1,2), Iordache C.(2), Holoc A.S.(2), Toma C(2), Braha E.(1,2)

(1) University of Medicine and Pharmacy "Gr. T.Popa" Iasi, Romania(2) Emergency Hospital for Children "St. Mary", Iasi, Romania

*Introduction:* Marfan Syndrome is a spectrum disorder caused by a heritable genetic defect of connective tissue that has an autosomal dominant mode of transmission. The defect itself has been isolated to the FBN1 gene on chromosome 15, which codes for the connective tissue protein fibrillin-1. Marfan syndrome affects most organs and tissues, especially the skeleton, lungs, eyes, heart, and large blood vessel. The prevalence of Marfan syndrome is estimated at 1/5000.

*Methods:* Prospective clinical and echocardiographic evaluation of consecutively recruited children at North-Eastern Region of Romania diagnosed with Marfan syndrome.

*Results:* We have analysed the prevalence of cardiovascular abnormality in 58 children with Marfan syndrome recorded in the files of Medical Genetic Center Iasi, Romania in the last 10 years (January 2006 –December 2015). There were 30 girls and 28 boys diagnosed at median age of 12,4 years (range from 4 years to 18 years old).

The diagnosis was based on anamnesis, the presence of the characteristic features, EKG, thoracic X-Ray, echocardiography, established Z score, ophthalmological and orthopedic exam.

Cardiac abnormalities were present in 82.75% of cases and they are represented by: mitral valve prolaps (65,51%), aortic route dilation (46.55%), mitral regurgitation (63,8%), tricuspid valve prolaps (3,4%), dilatation of pulmonary artery (1,7%). Aortic dissection did not appear. Aortic route surgery was needed for 5.17% cases.

*Conclusions:* Cardiovascular manifestation of Marfan syndrome remain among the central issues in diagnosis and management. Regular monitoring of valvular function and aortic diameter, early initiation of long-term  $\beta$ -adrenergic blockade and elective repair of a moderately regurgitant mitral valve or of a moderately aortic root dilatation represent the standards of care.