ACC.11
60th Annual Scientific Session & Expo

E440
JACC April 5, 2011
Volume 57, Issue 14

CONGENITAL CARDIOLOGY SOLUTIONS
(ADULT CONGENITAL AND PEDIATRIC CARDIOLOGY)

CRITICAL APPRAISAL OF THE REVISED MARFAN NOSOLOGY IN CARDIOLOGICAL PRACTICE

ACC Poster Contributions
Ernest N. Morial Convention Center, Hall F
Sunday, April 03, 2011, 3:30 p.m.-4:45 p.m.

Session Title: Adult Congenital Heart Disease
Abstract Category: 43. Adult Congenital Heart Disease
Session-Poster Board Number: 1065-441

Authors: *Piet De Witte, Teodora Radonic, Maarten Groenink, Rianne A. de Bruin - Bon, Janneke Timmermans, Arthur J. Scholte, Maarten P. van den Berg, Marieke J. Baars, Peter J. van Tintelen, Marlies Kempers, Aeilko H. Zwinderman, Barbara J. Mulder, Academic Medical Center, Amsterdam, The Netherlands*

Background: Marfan syndrome (MFS) is a connective tissue disorder with features in cardiovascular, ocular and skeletal systems. Different diagnostic criteria have been proposed in order to standardize the diagnosis of MFS. Recently, revised Marfan nosology was proposed where aortic root measurements were based on the Z-score.

Objective: We applied the revised Marfan nosology in an established adult Marfan population in order to define practical repercussions of novel criteria for clinical practice and individual patients.

Results: Out of 180 MFS patients, in 90.6% (n=163) the diagnosis of MFS remained. Out of 17 patients with rejected diagnosis, four patients were diagnosed as MASS phenotype, four as ectopia lentis syndrome and in nine patients no alternative diagnosis could be established. In 14 patients the diagnosis was rejected because the Z-score of the aortic root was <2, even though the aortic diameter was larger than 40 mm in seven of them. In three other patients the diagnosis of MFS was rejected because dural ectasia was given less weight in the revised nosology.

Conclusions: Following the revised Marfan nosology, the diagnosis of MFS would be rejected in 9.4% of patients, mostly due to no aortic root dilatation defined as Z-score ≥2. Currently used Z-scores seem to underestimate aortic root dilatation, especially in patients with a large Body Surface Area (BSA). We recommend re-evaluation of the criteria for aortic root involvement in adult patients with a suspected diagnosis of MFS.