P-77

FBN1 mutation does not influence handling of paediatric patients with confirmed Marfan syndrome (MFS)

Stark V., Arndt F., Mueller G.C., Steiner K., Weil J., Mir T.S.
University Heart Center Hamburg, Germany

Objectives: There is no single molecular or clinical test to demonstrate or rule out MFS. Clinical diagnosis of MFS is established according to Revised Ghent Criteria (RGC) and molecular diagnosis relies on fibrillin-1 (FBN1) mutation. In fact, although FBN1 mutation is the cause of classical MFS there are cases FBN1 mutation could not be detected. Therefore we evaluate whether there is a difference concerning clinical manifestation between paediatric Marfan patients with FBN1 mutation and those without and whether there is a need to distinguish those patient groups concerning follow-up, prophylaxis and therapy.

Methods: This study includes a cohort of 262 patients (10.9 ± 5.3 y) with confirmed or assumed MFS. They were subjected to standardised diagnostic programme including echocardiography and examination according to RGC. In addition molecular analysis was assessed whenever indicated. Prevalence and age of manifestation of the three cardinal symptoms of RGC were analysed.

Results: After all MFS was diagnosed in 103 patients (genetic analysis n=86). The detection rate for FBN1 was 73.8 %. We did not find any significant differences concerning prevalence or age of manifestation of the three cardinal symptoms of RGC by comparing patients with or without FBN1 mutation. Also analysis of different types of mutation did not show any significant variation.

Table 1: Prevalence and age of manifestation of dilatation of sinus of Valsalva (SV), ectopia lentis (EL), systemic manifestation (SysM) in patients with and without FBN1 mutation

	FBN1 mutation positive Prevalence	*FBN1 mutation negative Prevalence*	*FBN1 mutation positive Age (years±SD)*	*FBN1 mutation negative Age (years±SD)*	*p-value*
SV	*65.6 % (42/64)*	*68.2 % (15/22)*	*8.9 ± 0.8*	*11.1 ± 1.5*	*ns*
EL	*25.0 % (16/64)*	*13.6 % (3/22)*	*7.2 ± 1.2*	*3.9 ± 0.2*	*ns*
SysM	*40.6 % (26/64)*	*77.3 % (17/22)*	*11.3 ± 1.0*	*13.7 ± 0.9*	*ns*

Conclusions: All three cardinal symptoms did not show any significant variation between patients with or without FBN1. Both prevalence and mean age of manifestation were almost similar in the two patient groups. We conclude that there is no need to differentiate between paediatric patients with and without FBN1 mutation concerning follow-up, prophylaxis and therapy.