Congenital Cardiology Solutions

VARIABLE INTERMEDIATE OUTCOME IN TOF IS EXPLAINED IN PART BY 22Q11.2 DELETION STATUS

Poster Contributions
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Background: Patients with tetralogy of Fallot (TOF) experience variable outcome though the determinants are incompletely understood. We sought to investigate the mechanisms underlying such variability by studying the clinical status of children and adolescents with TOF and the contribution of 22q11.2 deletion (22q11.2DS) to outcome.

Methods: Cross sectional study of TOF patients 8 to 18 years who underwent research-based genetic testing, cardiac magnetic resonance (CMR), exercise stress test (EST) and review of medical history.

Results: We studied 165 patients (12.3 3.1 years), of which 30 had 22q11.2DS (18%), 65% were male, 84% Caucasian and 73% (120) received a transannular patch. For the whole group, by CMR the right ventricular (RV) ejection fraction was 608% (36-82), pulmonary regurgitant fraction 3417% (0-63) and RV end-diastolic volume 11439 (43-222) cc/m2. On EST, predicted maximum oxygen consumption (VO2) for age and gender was 7616 % (28-121). The 22q11.2DS and ND groups were comparable in age, gender, size, pulmonary valve anatomy, age at surgical repair and operative approach. Despite comparable RV function and degree of pulmonary regurgitation, on EST the 22q11.2DS had lower % predicted: Forced Vital Capacity (61.5 16 vs. 80.5 14, p< 0.0001); VO2 (6117 vs. 8012, p<0.0001); and Work (6418 vs. 8622, p=0.0002). Compared to the ND, the 22q11.2DS had more hospitalizations (cardiac + non-cardiac) (6.6 [5; 10] vs. 3 [2; 5], p<0.0001), saw more specialists (3.5 [2; 9] vs. 0 [0; 12], p<0.0001) and used >1 medication (67 vs. 42%, p <0.001).

Conclusions: We demonstrate that genotype may explain some of the clinical variability seen in TOF, in particular with respect to resource utilization and significant differences in exercise performance. This study also provides data on an intermediate aged TOF population with relatively preserved ventricular function despite significant pulmonary insufficiency in the years preceding adult disease.