Cardiac outcomes in adolescent and adult patients with Down syndrome and congenital heart defect.

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Background: trisomy 21 is the most common single chromosome abnormality at birth and is frequently associated with congenital heart disease (CHD). Advances in diagnosis and management of patients with CHD have also involved patients with Down’s syndrome and CHD leading them to survive to adulthood. However, few studies have focused on cardiac outcomes in adult patients with Down’s syndrome and CHD.

Aims: to determine cardiac outcomes in adolescent and adult patients with Down’s syndrome and CHD.

Methods: we determined the late survival in all patients (≥15 years old) with Down’s syndrome and CHD. Then, we examined cardiac outcomes in the subsets of patients with trisomy 21 and 1/ with repaired atrioventricular septal defect (AVSD) (n=33) and 2/ with Eisenmenger syndrome (n=71). We evaluated the impact of Down’s syndrome on the results by comparing cardiac outcomes between patients with Down’s syndrome and non-Down patients.

Results: Survival of 136 adult and adolescent patients with Down’s syndrome and CHD was 58.2% at 40 years. In the 33 patients with repaired AVSD, late complications occurred in 12% and were essentially infective endocarditis. Although actuarial survival after AVSD repair was not different between patients with Down’s syndrome and group with normal karyotype (n=50), clinically significant cardiac lesions were more common in no-Down patients (68% vs 27%, p<0.02), including arrhythmia, anomalies of mitral valve, and heart failure. In Eisenmenger patients, survival was significantly impaired in Down’s syndrome compared to patients with normal karyotype (n=118): survival at 40 years was respectively 51.9% vs 67.5% (p<0.0001, logrank test). Eisenmenger patients with complex anatomy had a significant worse prognosis when compared with those with simple anatomy (p<0.0001, logrank test), independently of karyotype, because Down syndrome patients had more complex heart defect, which is AVSD commonly associated with trisomy 21 (86% vs 61%, p<0.001).

Conclusion: adults and adolescents patients with Down syndrome have better long-term outcomes after AVSD repair than patients with normal karyotype, but Eisenmenger patients with Down syndrome have a markedly reduced survival, compared with no-Down patients, due to the complex CHD commonly associated with trisomy 21, which is AVSD.