Down's Syndrome Association
A Registered Charity No. 1061474

Annual Health Check Information for GPs

Adult cardiac disease in Down's syndrome

Atrioventricular septal defects (Endocardial cushion defects)

Congenital cardiac defects are common in Down's Syndrome (DS), around half of all people with DS have a cardiac defect at birth, in around 1 in 5 these are complex, serious abnormalities. These defects are readily missed unless an echocardiogram has been performed and it is recommended that all people with DS should have had at least one echocardiogram in their life, ideally within days of birth.

The classical cardiac problem in DS is an atrioventricular septal defect (AVSD), which is a congenital abnormality associated with abnormal development of the middle portion of the heart. Commonly there is a large ventricular septal defect and unrepaired this can lead to Eisenmenger's syndrome. Though entirely preventable through appropriate intervention, this was not always practiced 20 years ago, thus patients are still encountered with deep cyanosis, clubbed fingers and effort intolerance. The resultant pulmonary hypertension is now treatable with appropriate medicines.

Eisenmenger's is a complex systemic disorder, including polycythaemia, renal impairment, hyperuricaemia as well as elevated pulmonary pressures leading to effort intolerance, right heart failure and death. The gradual evolution of this condition means that without considerable attention to detail, even severe impairment can easily be overlooked. All patients with unrepaired congenital cardiac defects should be under the care of an adult congenital heart disease specialist, and any loss of interest in usual activities should be explored as possible evidence in a reduction in ability to perform the required physical tasks.

Repaired AVSDs, or those with no septal defects, may be associated with atrioventricular valve defects including regurgitation and more uncommonly stenosis, mainly on the left side. This may lead to heart failure with impaired effort tolerance, which again will only occasionally be reported as frank breathlessness by the patient. Because of the high prevalence of cardiac pathology and altered immune competence, infective endocarditis needs consideration during any severe febrile illness, and tachyarrhythmias are more common. In addition conduction defects leading to syncope or faintness need to be considered. Further lung disorders can result from peri-operative ventilation or recurrent chest infections in earlier life.

Although AVSD is a common congenital diagnosis in Down's syndrome, patients may also have any other form of congenital heart defect which should be followed up by a congenital cardiac specialist. As with AVSDs, patients with simple VSDs may not have been operated on historically and are prone to the development of Eisenmenger's syndrome.

Other cardiac conditions

Though uncommonly reported to date, coronary artery disease needs consideration in patients with Downs syndrome, diabetes and hypertension (metabolic syndrome). The relatively low

prevalence to date probably represents a combination of the previous short life span of individuals with Downs and difficulty diagnosing and investigating ‘older’ patients with Downs syndrome who may have other general health problems.

Sleep apnea is common in Down’s syndrome, but only with comprehensive prospective registries can we determine whether associated disorders such as hypertension are common as this population ages.

There is no reason to believe that people with Down’s syndrome are any less susceptible to any cardiac disease, and good reason to believe that such diagnoses may be under-reported.

Management

Eisenmenger’s is a complex condition requiring pulmonary vasodilator therapy, assiduous dental hygiene, comprehensive vaccination (including influenza) , avoidance of dehydration and great care when any general surgical procedures are performed. In patients with Eisenmenger’s even atrial tachy-arrhythmias can be extremely serious, and endocarditis should be considered whenever febrile illnesses occur.

Patients with repaired cardiac defects remain at risk of endocarditis, heart failure and arrhythmias.

Falling exercise tolerance or loss of interest in usual activities are non specific symptoms, but should lead to consideration of cardiac causes in individuals with Down’s syndrome. The widespread availability of natriuretic peptide testing should help identify patients in need of more extensive evaluation.

The advent of CT coronary angiography should facilitate the diagnosis of coronary disease even in patients with limited ability to co-operate with investigation and where diagnosed standard risk factor management should be instituted. Where appropriate consideration to medical and interventional management of symptoms should be undertaken.

Hypertension where identified should be managed according to standard NICE/JCHB guidance.

Publication date: August 2013

Publication reviewed & updated February 2016