

BASIC MEDICAL SURVEILLANCE ESSENTIALS FOR PEOPLE WITH DOWN'S SYNDROME.

CARDIAC DISEASE: CONGENITAL AND ACQUIRED (Revised 2007)

(One of a set of guidelines drawn up by the Down's Syndrome Medical Interest Group)

1. Between 40 and 60% of babies with Down's syndrome have congenital heart defects. Of these 30 - 40% are complete atrioventricular septal defects (AVSD)^{1,2,3}. Most AVSD can be successfully treated if the diagnosis is made early and the baby referred for full corrective surgery before irreversible pulmonary vascular disease (PVD) is established^{4,5,6,7,8}. Other lesions can usually be approached with less surgical urgency.
2. There must be a high level of clinical suspicion of congenital heart disease (CHD) for all newborns with the syndrome. Despite overall awareness of the risk of serious CHD in children with Down's syndrome some with important and sometimes severe CHD continue to present too late for the best chance of an optimum cardiac outcome (personal communications. Archer, Dennis, Tulloh)
3. Irreversible PVD is more likely to develop quickly in children with Down's syndrome and AVSD^{7,8,9}. Ideally surgery is desirable by 6 months^{5,8} and there is some evidence that surgery before 4 months may achieve best possible outcome⁶.
4. We suggest that the surveillance goal should be to establish by age 6 weeks at the latest whether or not there is a significant cardiac problem. This is because from a practical point of view this should be achievable over a wide range of clinical settings in the UK and Republic of Ireland and it is sufficiently early to ensure that by the time surgery can take place very few babies will already have irreversible PVD. We suggest also that for babies potentially at high risk for PVD it is prudent to attempt to achieve this by age 2 weeks (see 5.1)
5. Diagnostic methods
Clinical examination alone is insufficient to detect cardiac disease in the newborn period. Even the most serious abnormalities can be missed¹⁰. It is very unlikely however that a serious abnormality (AVSD or other major shunt lesion) requiring early intervention will be missed if the following course of action is taken.^{1,3,11,12}

5.1 Babies diagnosed with Down's syndrome in the early neonatal period

Shortly after diagnosis a careful clinical examination and ECG should be carried out. On the basis of this the degree of urgency for echocardiogram and expert cardiological assessment can be established as follows¹²:

- Those with abnormal clinical signs or ECG abnormality (in particular a superior QRS axis¹³) are potentially at high risk for PVD and it is desirable that they are referred and seen within 2 weeks of birth for expert clinical assessment and echocardiogram by someone with appropriate paediatric cardiological training.

- Those with no abnormal clinical signs or ECG abnormality on initial examination may nevertheless have cardiac disease^{5,12,13}. These babies should all be referred and seen within 6 weeks of birth by someone with appropriate paediatric cardiological training for further clinical assessment and echocardiogram

5.2 Babies diagnosed later in the neonatal period

These should have immediate ECG and clinical examination and accelerated referral to someone with appropriate paediatric cardiological training with the aim, wherever possible, of achieving the 6 week deadline given above.

5.3 Babies with a prenatal diagnosis of Down's syndrome

In the absence of evidence about the sensitivity of fetal echocardiography we suggest that those who had a fetal echocardiogram should still follow the above neonatal pathway.

5.4 Older children who have never had an echocardiogram should be dealt with as follows:

- Those with no symptoms or clinical signs and normal ECG should be referred routinely for further clinical assessment by someone with appropriate paediatric cardiological training
 - Those who are symptomatic and/or have abnormal clinical signs or ECG should be referred urgently.
6. People with heart lesions are at increased risk of infective endocarditis. They and their parents and carers should be given verbal and written advice about endocarditis prevention. Red cards from the British Heart Foundation¹⁴ are useful, and local paediatric cardiac centres will have their own preferred literature.
 7. It must always be remembered that those with Down's syndrome and a normal heart at birth can, like other children, develop pulmonary vascular disease and right heart failure secondary to airway/respiratory problems¹⁵.
 8. It must be noted that occasionally, even in expert hands, echocardiography, particularly in the first few days after birth, may fail to diagnose AVSD and other major shunt lesions. Hence there should be a low threshold for repeating this investigation if symptoms or signs of cardiac disease are detected at any age even in the presence of 'normal' early echocardiogram. (Personal communications. Archer, Dennis, Ward)
 9. From late adolescence onwards there is evidence of an increased incidence of asymptomatic mitral valve prolapse (MVP) with no clinical signs and of aortic regurgitation (AR)^{16,17,18,19}.

There is however insufficient evidence of benefit to make detailed recommendations about cardiac surveillance in adult life. MVP and AR are usually considered benign conditions but there may be implications for infective endocarditis prevention, particularly because of the high incidence of periodontal disease among this population²⁰. Hence careful cardiac assessment may be indicated before some dental procedures¹⁶.

There will be some whose MVP progresses to regurgitation (MVR). In order to identify these we recommend that auscultation of the heart should be included as part of routine medical monitoring on discharge from paediatric care and throughout adult life²¹. Those with MVR

should be monitored for signs of atrial fibrillation and/or left ventricular failure^{17,22}. For some of these restriction of competitive sporting activities may be advised.^{22,23}

10. Even if the above guidelines are effectively used there will still for some time to come be individuals for whom the difficult issues raised by the availability of heart lung transplant will need to be considered.²⁴.

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