Congenital heart disease is found in 40 - 50% of patients with Down syndrome. Early diagnosis is important because some cases of severe congenital heart disease may remain occult in the first months of life because of a delay in the fall of pulmonary vascular resistance. Late diagnosis may then result in an increased risk from surgical repair, or even inoperability.

The Paediatric and Congenital Council, therefore, recommends that all infants with Down syndrome be seen before two months of age by a physician with experience in the examination of the cardiovascular system of the newborn. The assessment should include echocardiography in all cases. The echocardiogram should be performed by a paediatric cardiologist or paediatric sonographer (under the guidance of a paediatric cardiologist) as per the Standards of Practice for Paediatric Echocardiography (CSANZ/ASUM)*.

In selected well infants, if their family / home is isolated and quick access to a paediatric cardiology service is impractical, a screening ECG and echocardiogram by a non-paediatric cardiologist / sonographer would be acceptable. The ECG and echocardiogram should be sent or transmitted to a paediatric cardiology service for review and, if it is thought to be adequate to exclude a significant problem, then a formal paediatric cardiology review could be organised at the convenience of the family and paediatric cardiologist.

In any unwell neonate with Down syndrome early discussion with a paediatric cardiologist or neonatal specialist is recommended.