Antenatal Screening for Down syndrome and other conditions

Summary January – December 2011

Background

Screening for Down syndrome and other conditions is available to all pregnant women who are less than 20 weeks pregnant. This screening provides a chance estimate for T21 (Down syndrome), T18 (Edwards syndrome), T13 (Patau syndrome) and some other rare genetic disorders.

The two options for screening are first trimester combined screening (serum and ultrasound nuchal translucency) and second trimester serum screening (quadruple test). Ultrasound scanning is performed by radiology practices around New Zealand and the nuchal translucency measurement is forwarded to the laboratories. Serum testing and generation of the risk report is performed by LabPLUS at Auckland District Health Board and Canterbury Health Laboratories at Canterbury District Health Board.

Detection of fetal anomalies offers women information that may help them prepare for the birth of their child, the option of delivery in a setting that has access to specialist surgical or medical care, and the possibility of considering termination or palliative care in the newborn period.

Antenatal screening relies on the diligence and dedication of many health professionals including radiology staff, Lead Maternity Carers, General Practitioners (GPs) and laboratory personnel. Screening relies on all of these groups performing to the highest quality - from informed consent and ultrasound scanning through to testing, issuing reports and follow-up as required. The information provided to the laboratories is key to informing high quality results including details of the pregnancy and nuchal translucency measurements. These details have a significant impact on the risk calculation and report that is issued.

Statistics

For the year 2011, 37276 women had screening in the first trimester and 4545 women had screening in the second trimester. This is approximately 55% of women have screening in comparison to pregnancy estimates. Through the screening, 32624 first trimester and 4545 second trimester risk reports were issued.

Table 1 Screened women and risks issued

First trimester screens	First trimester increased risks	Second trimester screens	Second trimester increased risks
32624	885 (2.7%)	4545	260 (5.7%)

Screening tests are ordered by a variety of practitioners. Approximately 17 % are referred by GPs, 74 % by midwives and 9 % by obstetric specialists.

The mean age of screened women is 30.9, compared to the mean age of women giving birth of 29.6 years. The mean age of screened women is tending to decrease over time.

Responsibilities

The laboratories performing serum testing are IANZ accredited and a peer review audit was undertaken in 2011.

Practitioners performing nuchal translucency are required to have FMF certification (either through the United Kingdom or Australia). The National Screening Unit has a Memorandum of Understanding with IANZ and audits of radiology practices will commence in 2012.

It is recommended that health practitioners involved in this screening should complete e-learning modules provided by the National Screening Unit and found at http://www.learnonline.health.nz/.

Progress throughout 2011

The National Screening Unit has contracted the University of Otago to provide six monthly monitoring reports and to explore future monitoring requirements of this screening.

Best practice reporting templates have been developed for NT ultrasound reporting (http://www.nsu.govt.nz/health-professionals/3814.aspx) and genetic referrals (http://www.nsu.govt.nz/health-professionals/3816.aspx).

A technical advisory group has been established to advise the National Screening Unit on technical and operational matters.

Work is underway on web-based questions and answers, consumer resources, updating the 2010 practitioner guidelines, key messages and education of health practitioners offering this screening.