

## GP Information on Down Syndrome Screening

First antenatal clinic visits for public patients and private patients frequently occur well into the 2nd trimester.

GPs are now the main clinicians providing information on and arrangement of Down Syndrome screening for antenatal patients. Here is some important information for GPs who may be unfamiliar with this testing

- RANZCOG recommends **all** pregnant women be made **aware** of the availability of screening tests for Down Syndrome
- eligibility for rebate on radiology only if > 35yrs age or other recognised risk factors listed in Medicare Benefits Schedule book under item number 55707

The **Combined Test** is the most commonly offered screening test as it has the best detection rate.

- It involves **both** a blood test for PAPP-A and free  $\beta$ hCG and a nuchal fold translucency ultrasound
- It is performed between 11 weeks & 13 weeks + 6 days
- The timing of this test is **critical** and as such accurate assessment of gestational age is also vital
- It detects 90% of cases of Down Syndrome and carries a 5% false +ve rate
- It may also lead to diagnosis of other chromosomal abnormalities
- Usually the Nuchal Translucency is booked and pathology arranged to be done on same day or a few days prior to the NT scan.
- blood results / age / race / smoking status are all entered into specialised software – this software then produces **results as an adjusted risk**
- The scan must be performed by accredited personnel who have the relevant training and access to the software
- The total scan cost via private radiology providers if not eligible for a rebate is around \$150. If eligible for a rebate the rebate is currently about \$55. Patients can obtain prices directly from the radiology provider.
- This test is not currently available at Nambour General Hospital. A free public service exists at Royal Women's Hospital and at the Mater Mothers Hospital but referral must be by a public hospital obstetrician.
- If the test shows an adjusted Down Syndrome risk that is greater than 1 in 300 then the woman should be referred as soon as possible to a private obstetrician or the Nambour Antenatal Clinic for consideration of amniocentesis (or CVS if earlier than 14 weeks gestation)

### Summary of Screening Tests for Down syndrome

Remember all of the following tests are screening tests and carry a risk of false positives and false negatives. However screening tests are non invasive and carry no risk of unintentional foetal loss

Screening test	Gestation date performed	Detection Rate	Comments
<b>Maternal Age alone</b> (> 35-37 yrs)		30%	
<b>1<sup>st</sup> trimester biochemistry testing only</b> free βhCG & PAPP-A	10-13 wks	60%	
<b>1<sup>st</sup> trimester nuchal translucency only</b>	11- 13 wks + 6 days gestational age <b>critical</b> for accuracy of test if unsure of dates consider prior US for accurate gest age	80%	NGH refers to Mater Mothers or Royal Womens (no cost public pts) Private referrals must be with an accredited centre
<b>Combined Test</b> (Nuchal Trans & free βhCG & PAPP-A)	11- 13 wks + 6 days	90%	Arrange tests for same time or few days prior (ie two working days)
<b>2<sup>nd</sup> trimester Biochemistry only</b> hCG, Oestriol, alphaFP	14 -18 wks	60-70%	
<b>2<sup>nd</sup> trimester morphology scan</b>	18 -20 wks	20-30%	Low sensitivity and high false +ve rate

Follow up: A positive screening test requires further follow up. The patient should ideally see both an obstetrician and the genetic counsellor.

Diagnostic Test	Gestation	miscarriage rate	Results/sample not obtained
<b>CVS</b> Chorionic Villus Sampling	11-14 wks	0.5-1%	1.4%
<b>Amniocentesis</b>	>15 wks	0.5-1%	2.3%

For Genetic counselling contact the Queensland Clinical Genetic Service: Ph **5441 7167**

Genetic counsellor: **Ms Karen Crowe**

Other enquiries Antenatal Clinic: ph **54706701**

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