



1st and 2nd trimester screening for chromosomal abnormality

Dr Glenn Gardener

Director Maternal Fetal Medicine

Mater Mothers Hospital



Q1 Down syndrome (DS) is a hereditary genetic abnormality.

True or False?

95% of Down syndrome pregnancies have arisen from a spontaneous event - uneven division of the maternal ovum prior to fertilisation

'Non-disjunction' frequency increases as the age of the maternal ovum increases, hence the relationship with DS and maternal age

Only 5% of pregnancies with Down syndrome arise from an unbalanced translocation which may be inherited

Hence DS risk usually does not relate to family history



Q2 How common is Down syndrome?

- a) 1 in 450 births
- b) 1 in 550 births
- c) 1 in 800 births
- d) 1 in 700 births

Approximately 1 in 550 babies are born with Down syndrome

Birth prevalence of Down syndrome increases as average maternal age increases

Prevalence of DS is approx 30% higher at 12 weeks than at term

Mater estimated no. DS at 12 wks 25 and 17 at term.



Q3. Down syndrome screening is only recommended for women over 35 years

True or False?

Most babies with DS are born to women <35 years

Every pregnancy is at risk of Down syndrome

35 yrs of age was an arbitrary cut-off that defined 5% of the population that would benefit from invasive prenatal diagnosis

Maternal age was the first screening test for Down syndrome

Age is incorporated in current screening algorithms but is not of sufficient accuracy to be used alone

Detection rate using maternal age alone = 30% for 5% false positive rate, 40% for a 15% FPR (% current population >35yrs)



Q4. Every pregnant woman should have Down syndrome screening.

True or False?

Testing for Down syndrome in pregnancy should be offered with appropriate counselling

Testing is entirely optional - it's the mother's choice

Assumptions should not be made based on religious background or other perceived factors – it should be offered to all

There should be no presumption that testing for Down syndrome implies termination of pregnancy if it is discovered

Down syndrome pregnancies are high risk for in-utero demise and monitoring is necessary for the timing of delivery

Preparation for the birth of a baby with DS is a valid reason for testing



Q5. The 1st trimester combined screen is ...

- a) Nuchal translucency + AFP + PAPP-A
- b) Nuchal translucency + PAPP-A + BHCG
- c) Nuchal translucency + AFP + UE3
- d) Nuchal translucency + PAPP-A + inhibin

PAPP-A = Pregnancy associated plasma protein A

BHCG = Beta subunit human chorionic gonadotrophin

Detection rate 85-90% for screen positive rate of 5%

Dating, viability, multiple pregnancy, some abnormalities

Preference for earlier testing



Nuchal translucency



Q6 The triple test is a recommended screening test for Down syndrome

True or False?

- Triple test (BhCG, unconjugated oestriol UE3, alphafetoprotein AFP)
- Detection rate 77% (Suress 2003)
- 1 in 20 women screened with triple test will be screen positive ie 'high risk' (same FPR as 1st TM screen)
- Can be performed from 15-20 weeks gestation



Increased AFP in 2nd trimester



Open neural tube defects



Gastroschisis



Q7. A 1st trimester combined screen followed by a 2nd trimester triple test is better than either one alone

True or False?

Only one or the other test should be performed but not both in a sequential fashion

Doing two tests doubles the false positive rate (>10%)

Dilemma of conflicting results – one high risk and the other low risk ???

Creates confusion and anxiety for both doctor and patient

1st trimester combined test is preferred

2nd trimester triple test is a satisfactory option



Q8. The 18-20 week morphology is a good option if no other screening tests have been performed

True or False?

- Detection rate of DS from morphology scan is poor
- False positives with 'soft markers' is high >10%
- No pretest counselling or consent regarding DS screening
- Anxiety ++ from soft markers
- Association between 'soft markers' and DS poor
- 'Soft markers' have a high prevalence in normal fetuses
- 'Tertiary scan' cannot diagnose Down syndrome



Q9. Women over 35yrs should have prenatal diagnosis ie CVS or amniocentesis and not screening tests

True or False?

- The 1st trimester and 2nd trimester screening tests are just as valid in older women at determining who would benefit from invasive tests
- Significantly fewer CVS and amniocentesis would be needed if all women >35yrs had screening in place of invasive testing
- Detection rates of DS would be similar and reduced loss of normal pregnancy from invasive testing



Q10. 3D and 4D ultrasound is helpful in the diagnosis of Down syndrome

True or False?

- 3D and 4D ultrasound has not to date added any improvement over 2D imaging for Down syndrome screening or diagnosis
- The facial features of children with Down syndrome are not distinguishable in-utero with 3D/4D ultrasound



Q11. Medicare does provide a rebate for nuchal translucency for a low risk woman

True or False?

There is a new Medicare item for nuchal translucency when it is performed with CRL of 45-84mm

1st and 2nd trimester biochemistry MBS rebate - only one rebate allowed per pregnancy

Many Radiology practices not accredited for nuchal translucency and those that are charge a gap (\$70-200)



Q12. A 1st trimester combined test risk has reported a risk of DS as 1 in 367 (age risk in 940). Do you..

- a) Counsel regarding the increased risk and refer for invasive testing
- b) Counsel regarding the low risk result
- c) Recommend a triple test in 2nd trimester
- d) Advise that a morphology scan at 18-20 weeks will provide reassurance

This a low risk result (<1 in 300) and is a better indicator of risk than her age, a triple test or a morphology scan

Woman of all risks have the option of diagnostic testing if they accept the risk of pregnancy loss



Q13. A transabdominal CVS has a higher risk of pregnancy loss than amniocentesis

True or False?

- The risk of pregnancy loss from CVS and amniocentesis is the same and up to 1% based on randomised studies
- CVS provides an earlier result and is available <15 weeks
- Amniocentesis is performed >15 weeks
- Women should not have to wait for amniocentesis if they are high risk on a 1st trimester screen



Q16. GP/hospital are at risk medico-legally if baby born with Down syndrome and no screening or testing offered to mother

True or False?

- There have been claims in Australia regarding birth with DS
- No current Qld Health screening/testing policy for Down syndrome screening
- RANZCOG/HGSA guidelines are applicable to care provided by O&G Specialists



What about multiples?

- Nuchal translucency alone (some labs will adjust for twins) recommended and 1st trimester is the best time to assess chorionicity



Mater CPD Women's Health Conference 2010



Mater Maternal Fetal Medicine

- Rapid access for prenatal genetic counselling and diagnostic testing
- Tertiary ultrasound and consultation for fetal abnormality
- Consultation for complex maternal and/or fetal conditions
- Referrals usually via Obstetrician or if urgent direct

- Contact MFM doctors line 3163 1597

