



CHECKING FOR PROBLEMS WITH THE BABY IN EARLY PREGNANCY

**It's Your Choice to Test for
Down Syndrome & Neural Tube Defects**



Presented by:

Copyright:

Menzies School of Health Research 2010. All rights reserved.

ISBN: 978-1-921576-26-3



<http://www.menzies.edu.au>

Authors:

Kayli Wild, Alice Rumbold, Lawurrpa Maypilama, Jacqui Boyle, Sue Kildea, Lesley Barclay.

Suggested citation:

Wild K, Rumbold A, Maypilama L, Boyle J, Kildea S & Barclay L (2010). Checking for Problems with the Baby in Early Pregnancy: It's Your Choice to Test for Down Syndrome and Neural Tube Defects. Menzies School of Health Research, Darwin.

Design, layout and illustrations:

Mary Kate Wright,



<http://www.mkillustrations.com>

Background art:

Anne Hanning

<http://www.anehanning.com>

This resource is available online at:

<http://www.menzies.edu.au/childhealthresources>

Purpose of the resource:

This resource was developed as part of a research project investigating Aboriginal women's views of prenatal screening in the Northern Territory. The resource is designed to assist healthcare providers in discussing fetal anomaly screening tests with women, their partners and their families. It may also help educators to facilitate discussion about these tests with women (and men) in the community.

Acknowledgements:

We would like to acknowledge preliminary work undertaken by Maria Nickels and Jillian Richards who previously worked on this project. The resource was developed based on fieldwork with Yolngu women in Galiwin'ku, Northern Territory and a workshop held in Darwin with women's health educators and midwives. We would like to thank the Top End Strong Women Coordinators for their input throughout the process of developing this resource. We also acknowledge that feedback from interviews with Aboriginal parents (women and men), Aboriginal health workers, midwives, general practitioners and obstetricians from both Central Australia and the Top End helped to shape subsequent drafts.

The research and resource development was funded by the National Health and Medical Research Council of Australia (project grant #490314).

Most women will have a healthy baby.

It doesn't happen very often, but sometimes there can be a problem with the baby (where the baby is not put together properly).

There are two main problems the doctors can test for in early pregnancy – Down syndrome¹ and neural tube defects.

It is important to know that these tests can't pick up all possible problems like Fetal Alcohol Spectrum Disorder, Cerebral Palsy or other problems that happen later in pregnancy.



¹Down syndrome (Trisomy 21) is the most common chromosomal abnormality. Other less common problems are also tested for including Trisomy 18 (Edward syndrome), Trisomy 13 (Patau syndrome) and sex chromosome abnormalities.

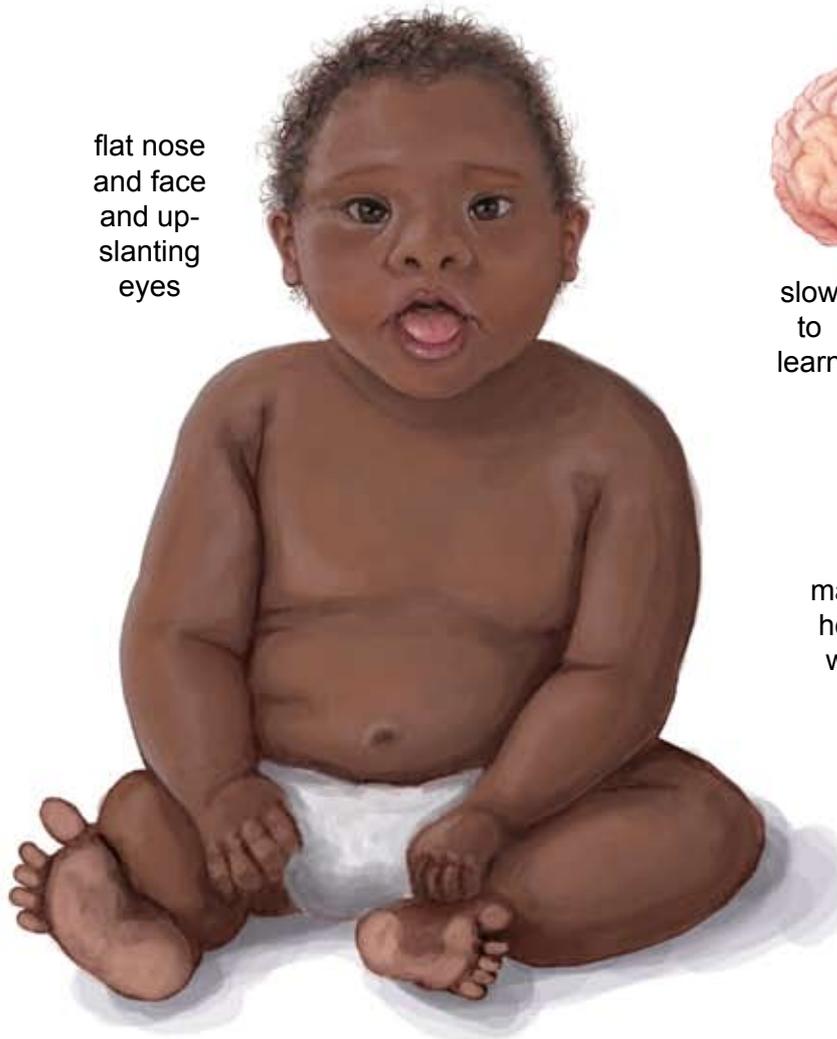
Information about these tests should be given to all women in early pregnancy. It's up to women, their partners and their family to decide whether they want these tests.



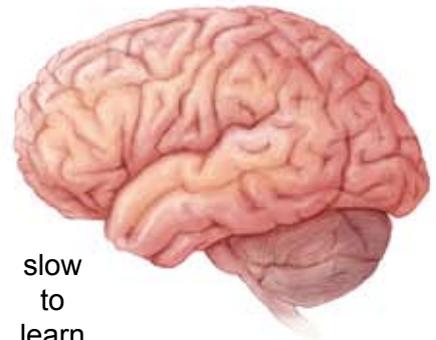
DOWN SYNDROME

Down syndrome is where the baby looks different and is also slow to learn things. This will affect the baby for the rest of its life.

The baby may have other health problems to do with the heart, lungs and stomach.

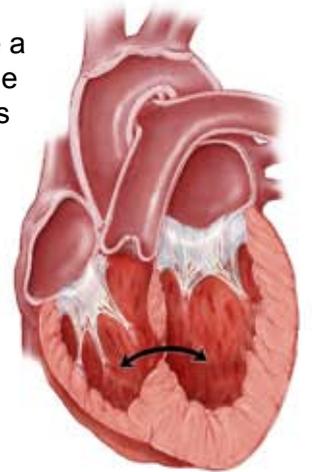


flat nose and face and up-slanting eyes

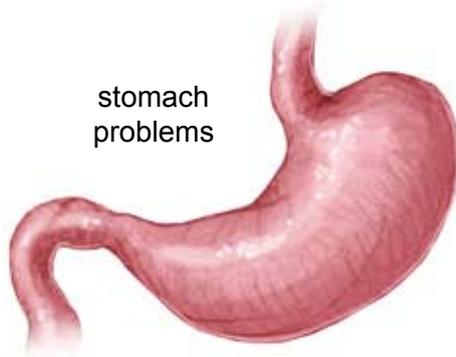


slow to learn

may have a hole in the wall of its heart



stomach problems



problems with breathing

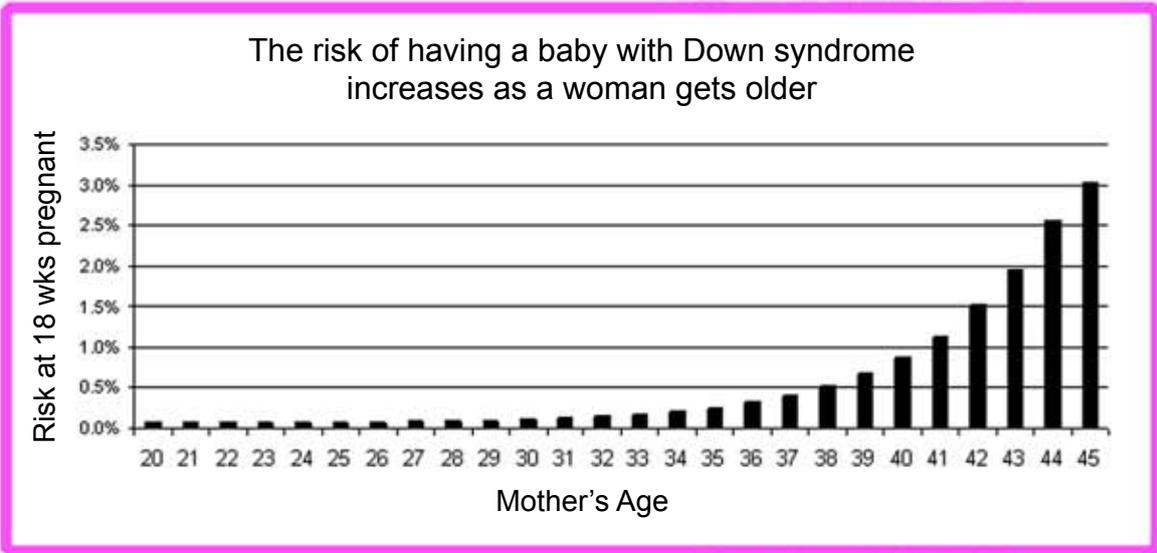


The chance of having a baby with Down syndrome for all Australian women is about 1/400.¹

Many babies with Down syndrome won't survive pregnancy and will miscarry (lose the baby) on their own.²

When they are born, some babies with Down syndrome are only a little bit affected. With medical help and extra support they can grow up healthy and take part in community life. Other babies with Down syndrome can be severely affected and need to spend lots of time in hospital.

We don't know how to prevent Down syndrome but we do know that the chance of having a baby with this problem increases as a woman gets older, or if there is a history of it in the family.



¹This figure includes births plus terminations of pregnancy. The risk of having a baby with Down syndrome is the same for Indigenous and non-Indigenous women (Abeywardana & Sullivan 2008).

²Many babies with Down syndrome will miscarry on their own (Morris, Wald & Watt 1999).

NEURAL TUBE DEFECTS

The most common type of neural tube defect is called spina bifida. This is when there is a hole at the back of the baby's spine.

When the baby grows up he or she might have trouble walking and problems controlling their bowel and bladder (might need to wear a nappy). These babies can often have an operation at the hospital to help fix the hole.

There are other types of neural tube defects and it can be more serious if the hole happens close to the head.¹

The chance of having a baby with a neural tube defect is small² and the risk becomes higher when women don't have enough of the vitamin called folate.

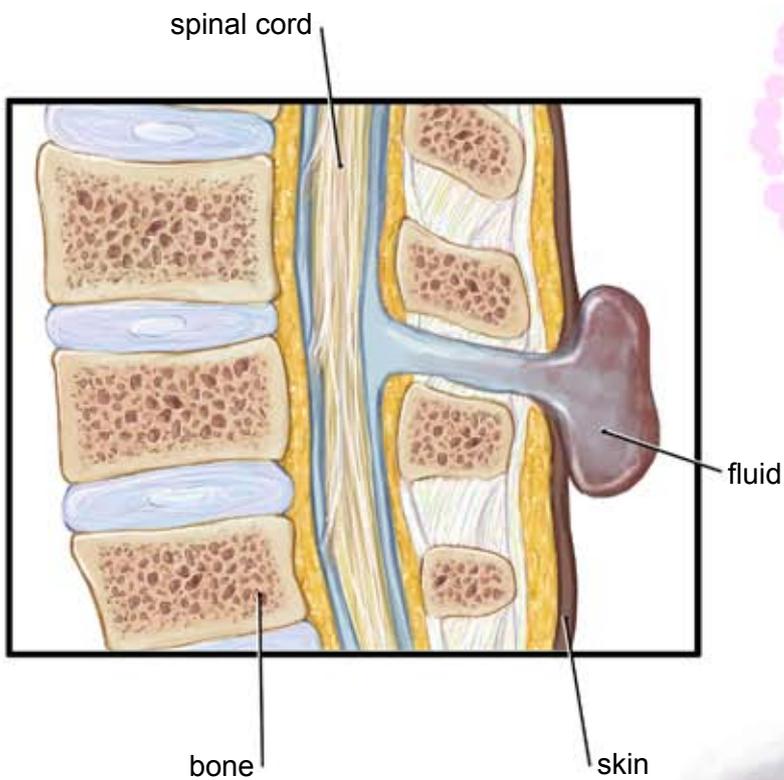


¹ Anencephaly is when the baby's brain and skull don't form properly. Babies with this condition usually don't survive.

² The risk of having a baby with a neural tube defect is about 1/1000, including births and terminations of pregnancy. Neural tube defects are twice as common amongst Indigenous women as non-Indigenous women (Abeywardana & Sullivan 2008).



The hole can be big or small, and can be high or low on the back.

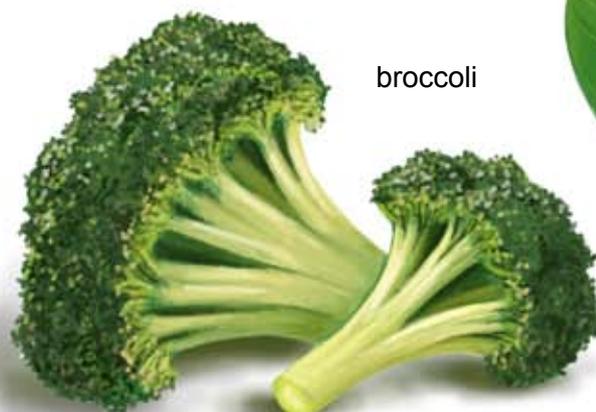
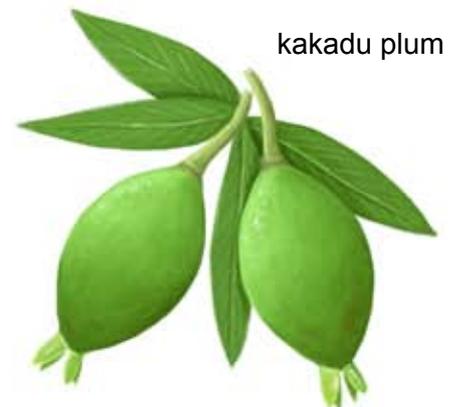
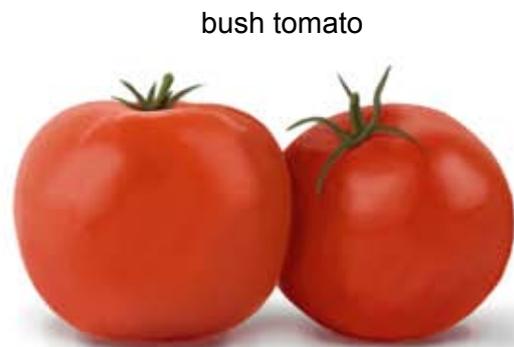
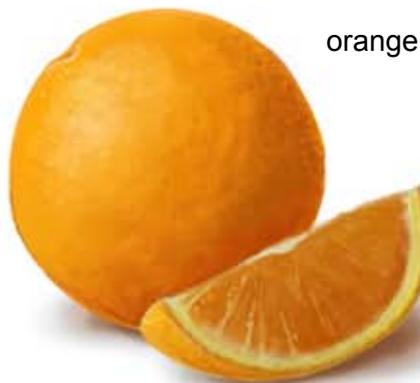


The child may have trouble walking when it's older, and may have to wear a nappy.

FOLATE

Women can help prevent neural tube defects by having enough folate in their diet.

- Women need to have extra folate before they even get pregnant as neural tube problems happen in the few first weeks of pregnancy.
- It's hard to get enough folate needed for pregnancy from food alone, so folic acid vitamins should also be taken.¹
- Folate can be found in bread, cereal, green leafy vegetables and other fresh food like oranges and tomatoes.



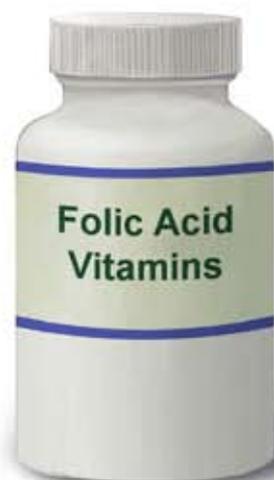
wholemeal
bread



bran cereal



beans



folic acid
vitamin
tablets

If a woman thinks she could get pregnant she should take folic acid vitamin tablets (available from the health centre or supermarket) because folate helps the baby's brain and spinal cord develop normally and helps prevent neural tube defects.

¹Women should take a vitamin supplement containing 400µg of folic acid daily. Much higher levels (5000µg) of folic acid supplementation are recommended for women on epilepsy medication or with a family history of neural tube defects. Folic acid supplements should be taken one month before and three months after conception (NHMRC 2006).

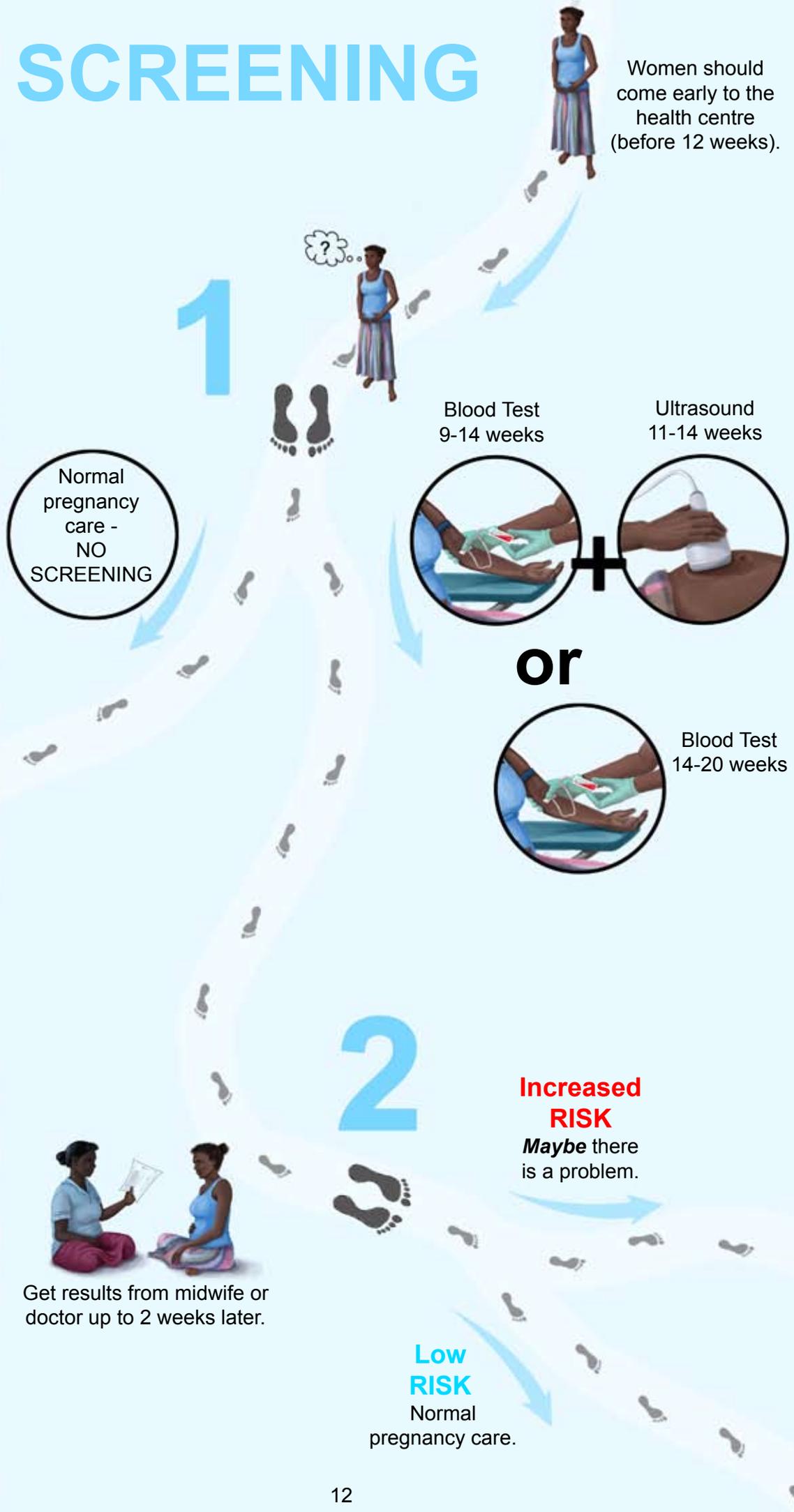
SCREENING

There is a two-step process to check for these problems.

The first step is called **SCREENING** because it sorts out which women have a lower chance and which women have an increased chance of having a baby with one of these problems.

This first step tells a woman about her individual chances, her own 'risk' result.

Women should come early to the health centre (before 12 weeks).



DIAGNOSIS

For women who get an increased risk result, they can decide to find out for sure whether the baby has a problem. Checking for sure is the second step, the **DIAGNOSIS**, and is a bit more involved.

5



Prepare for a baby with a disability.



End pregnancy.



Down Syndrome or Neural Tube Defect Found



No Problems Found

Continue with normal pregnancy care.



4



Get results from midwife or doctor up to 2 weeks later.

Amniocentesis-checking for sure. 16-20 weeks.



Normal pregnancy care - **NO DIAGNOSIS**

No further tests for these problems. Women won't find out for sure until birth.

3



SCREENING



Reasons why women might want to have the tests:

- To find out more about the pregnancy
- To prepare for a baby with a disability
- To terminate the pregnancy if a disability is found

Talk with your midwife or doctor about the tests.

You can think about whether you want these tests and talk with your partner or family. If you want you can make a decision together.

1

You can decide not to check for Down syndrome or neural tube defects.

This means you will have regular pregnancy care, but won't find out in advance your chances of having a baby with one of these problems.



or



You can decide to have the screening tests. It's important that you have the tests at the right time in pregnancy. When you are about 3 months (9 to 14 weeks) pregnant you can have an ultrasound AND have blood taken to test for Down syndrome.¹ The ultrasound is usually done at a hospital in town.²

or When you are 4 to 5 months (14 to 20 weeks) pregnant you can have just the blood test to check for Down syndrome AND neural tube defects. This test is not as accurate³ as having the ultrasound as well, but you can get this blood test done in the local health centre or at the hospital.



2

It takes up to 2 weeks to get the results back, and the midwife or doctor will discuss your test results with you.



**Low
RISK**

or

**Increased
RISK⁴**

If you have a 'low risk' result then it is very unlikely that the baby will have a problem like Down syndrome (but there are no guarantees).

If you have an 'increased risk' result it means maybe there is a problem. It is important to remember that most women (96%) who get this result will go on to have a normal healthy baby.⁵

¹The blood test and the ultrasound do not need to be done on the same day as long as they are done within the correct weeks of pregnancy. Ideally blood should be taken the week before having the ultrasound. Even though this tests only for Down syndrome, neural tube defects can usually be identified at the routine 'morphology' ultrasound done at 18 to 20 weeks in pregnancy.

²Sometimes women will need to pay for the ultrasound, ask your midwife or doctor about costs. The first trimester blood test plus nuchal translucency ultrasound will pick up about 90% of babies with Down syndrome (SAMSAS 2007).

³The second trimester blood test picks up about 60% of babies with Down syndrome, and neural tube defects can often be found on the regular 'morphology' ultrasound women have at 4 to 5 months in their pregnancy (SAMSAS 2007).

⁴The result will come back with a number reporting your individual risk. A result of 1/250 or above is considered an 'increased risk' result. See examples of interpreting different risk results at the back of this resource.

⁵(SAMSAS 2009a).

DIAGNOSIS



If you get an increased risk result, you can decide whether you want further tests to find out for sure if the baby has a problem. You might want to talk with your partner, family or friend before deciding.

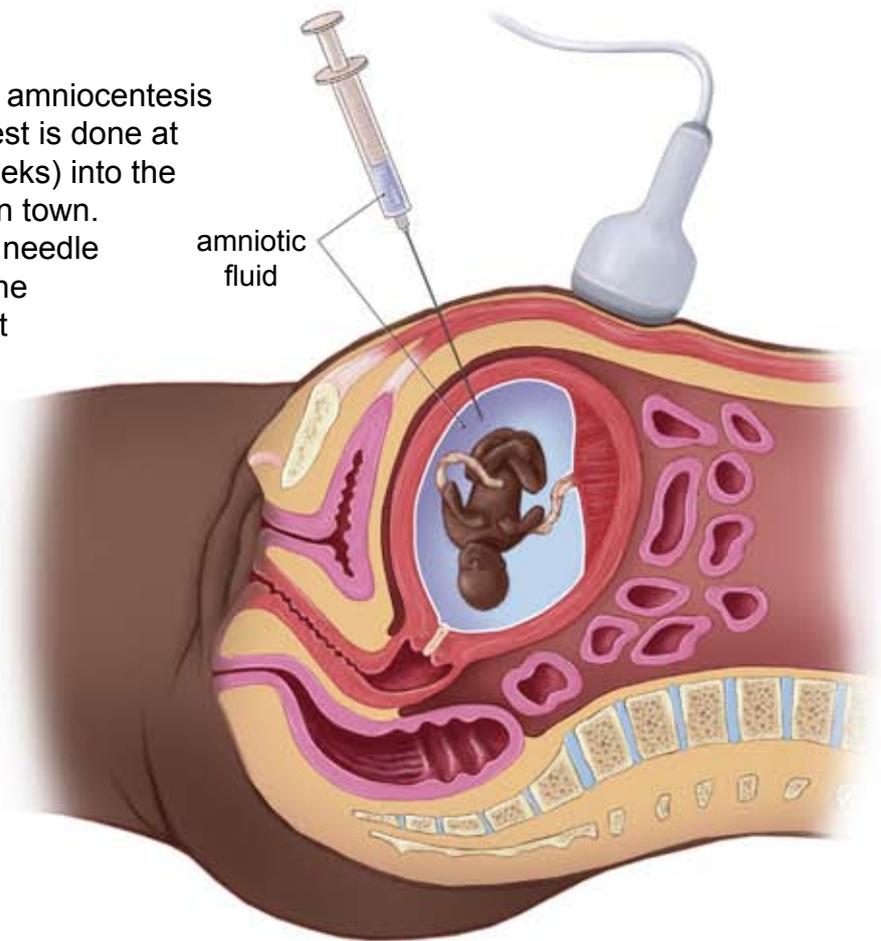
3

If you get an increased risk result, you can choose not to have further tests. This means you won't know for sure if the baby has a problem until it's born.



or

You can decide to have an amniocentesis to find out for sure.¹ This test is done at 4 to 5 months (16 to 20 weeks) into the pregnancy at the hospital in town. The test involves putting a needle through the stomach into the uterus (baby bag) to collect a little bit of liquid from around the baby. The doctor uses an ultrasound to look and make sure they don't touch the baby with the needle. But there is a small chance that this test could cause a miscarriage.



The risk of miscarriage after having an amniocentesis is about 1/200.

¹There is a similar test called chorionic villus sampling (CVS) which can be done earlier, in the first 3 months (10 to 12 weeks) of pregnancy. If you would like access to this earlier test talk to your doctor about where it is available as you may need to travel outside of the Northern Territory.

4

It takes up to 2 weeks to get the results back, and the midwife or doctor will discuss your test results with you.



No Problem Found

or

Down Syndrome or Neural Tube Defect Found



If no problems are found then continue with normal pregnancy care at the health centre.



If the baby is found to have Down syndrome or a neural tube defect your doctor can give you more information about the problems the baby might have.

DIAGNOSIS



If an abnormality is found, you can make a further decision about whether to keep the baby or to have a termination. You can discuss your options with your doctor and partner, family or close friend before making any decisions.

5

Prepare for a baby with Down syndrome or a neural tube defect.



Where to get support:

Down Syndrome Association of the Northern Territory

Shop 16, Rapid Creek Shopping Centre
48 Trower Rd
Rapid Creek NT 0810
Phone: (08) 8985 6222
Email: dsant@octa4.net

Department of Health and Families Office of Disability Access Point

Phone: FREECALL 1 800 139 656

Spina Bifida and Hydrocephalus Association of South Australia

Phone: (08) 8366 5900

<http://www.spinabifida.asn.au>

Trisomy Oz Prenatal Support

Phone: 0414 494 853

<http://www.trisomyoz.bounce.com.au>

or End the pregnancy early.¹



Where to get support:

Danila Dilba Emotional and Social Wellbeing Centre

Unit 1/3 Malak Place
Malak NT 0812
Phone: (08) 8927 9335

Stillbirth and Neonatal Death Support (SANDS) South Australia

Phone: (08) 8277 0304

Birthline Pregnancy Support South Australia

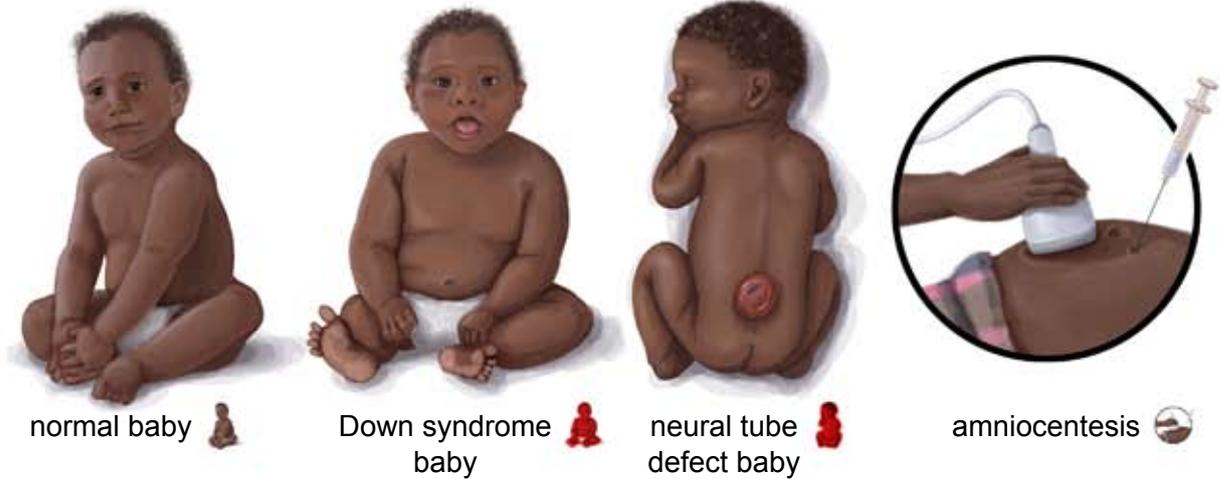
After Abortion Support

Phone: 1300 655 156
<http://www.pregnancysupport.com.au>

¹Late termination means that labour is induced.

INTERPRETING RISK

Use the following charts to help you to understand the different risks.



1/10 This is an example of an 'increased risk' result.
There is a 1 in 10 chance that the baby will have a problem.
There is a 9 in 10 chance that the baby will be normal.



1/20

This is an example of an 'increased risk' result.

There is a 1 in 20 chance that the baby will have a problem.

There is a 19 in 20 chance that the baby will be normal.



1/100

This is an example of an 'increased risk' result.

There is a 1 in 100 chance that the baby will have a problem.

There is a 99 in 100 chance that the baby will be normal.

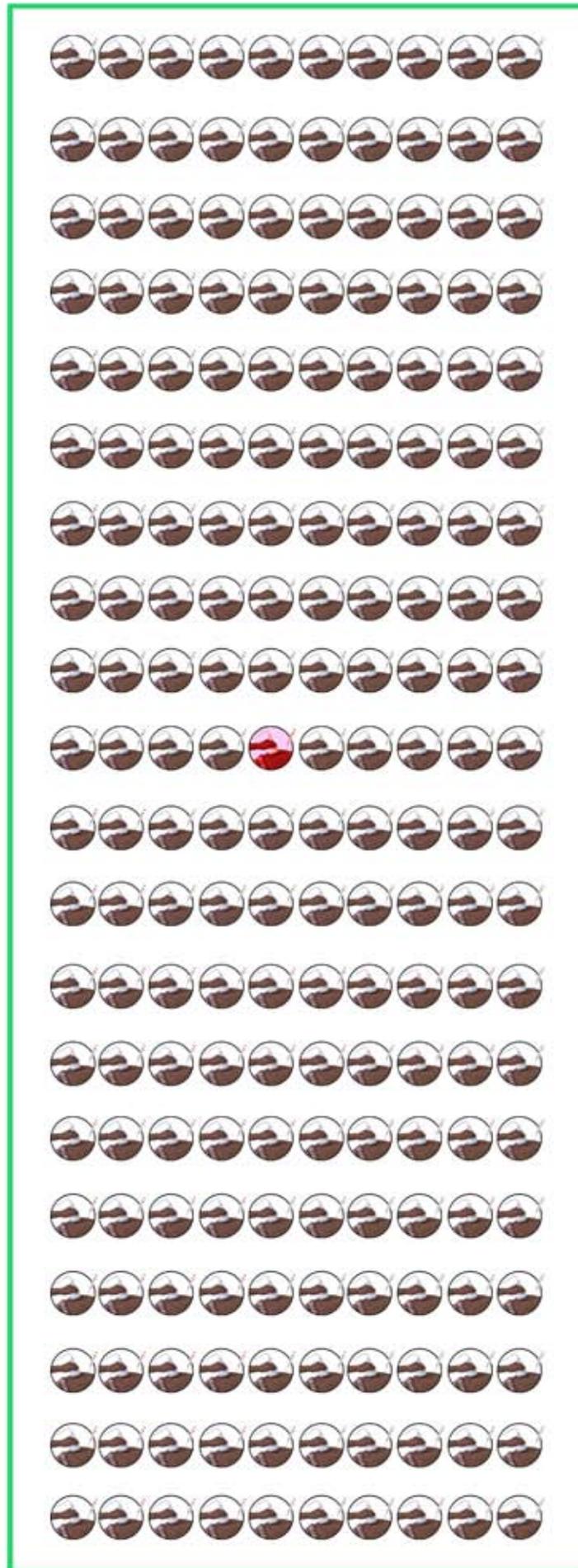


1/200

This is the risk of miscarriage after having an amniocentesis.

If 200 women had an amniocentesis, 1 might have a miscarriage.

If 200 women had an amniocentesis, 199 would not miscarry.



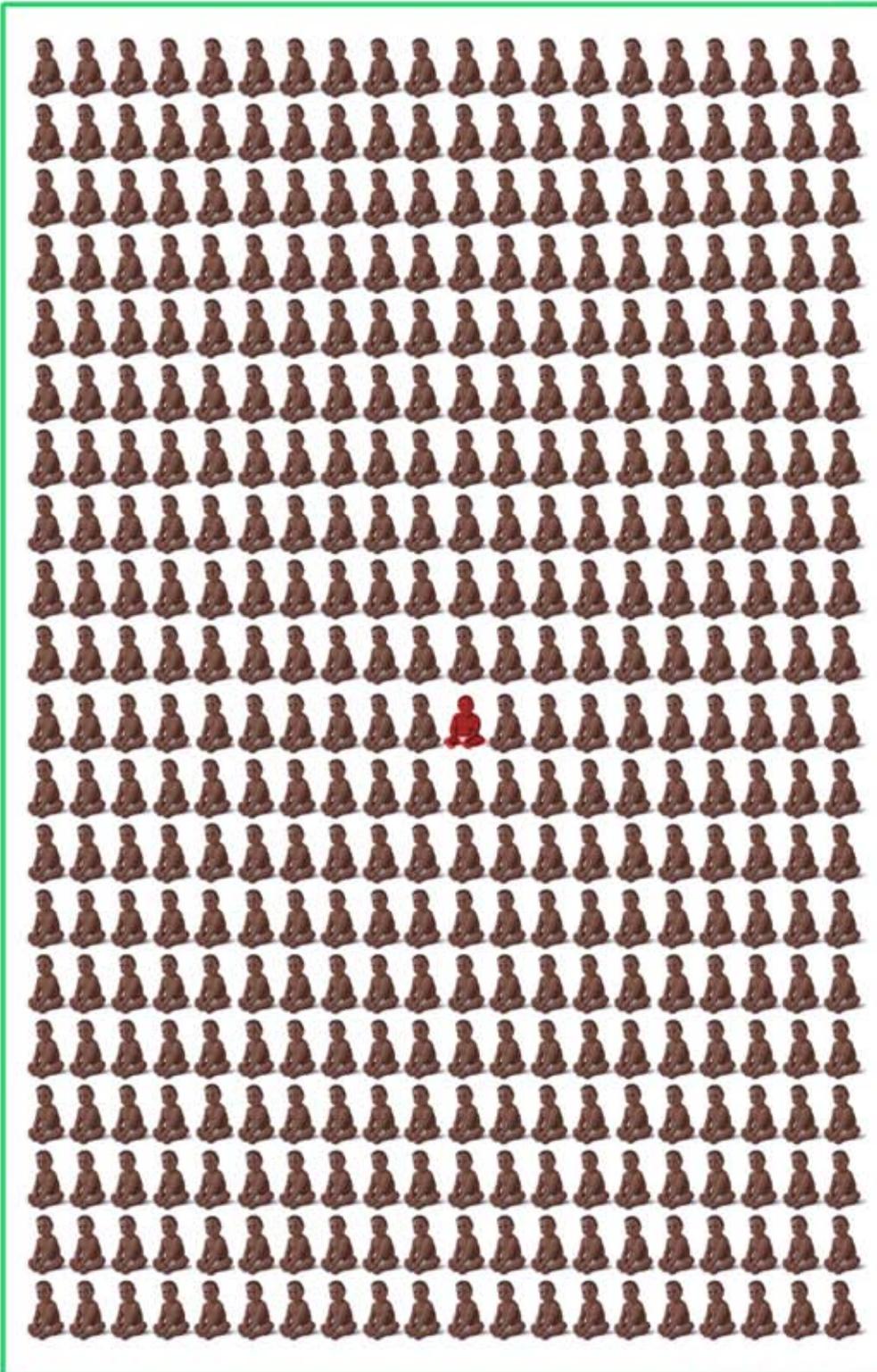
1/400

This is the risk of having a baby with Down syndrome for all women in Australia.

This is also a 'low risk' result.

If 400 women got pregnant, 1 of those babies might have Down syndrome.

If 400 women got pregnant, 399 of those babies would be normal.



The risk of having a baby with Down syndrome increases as a woman gets older¹

Mother's Age	Risk of Down Syndrome Pregnancy
20	1/1330
21	1/1316
22	1/1297
23	1/1273
24	1/1243
25	1/1203
26	1/1155
27	1/1096
28	1/1026
29	1/945
30	1/855
31	1/759
32	1/659
33	1/561
34	1/468
35	1/383
36	1/308
37	1/245
38	1/191
39	1/149
40	1/114
41	1/88
42	1/66
43	1/51
44	1/39
45	1/33

¹Chance of having a Down syndrome pregnancy at 18 weeks gestation (SAMSAS 2009b).

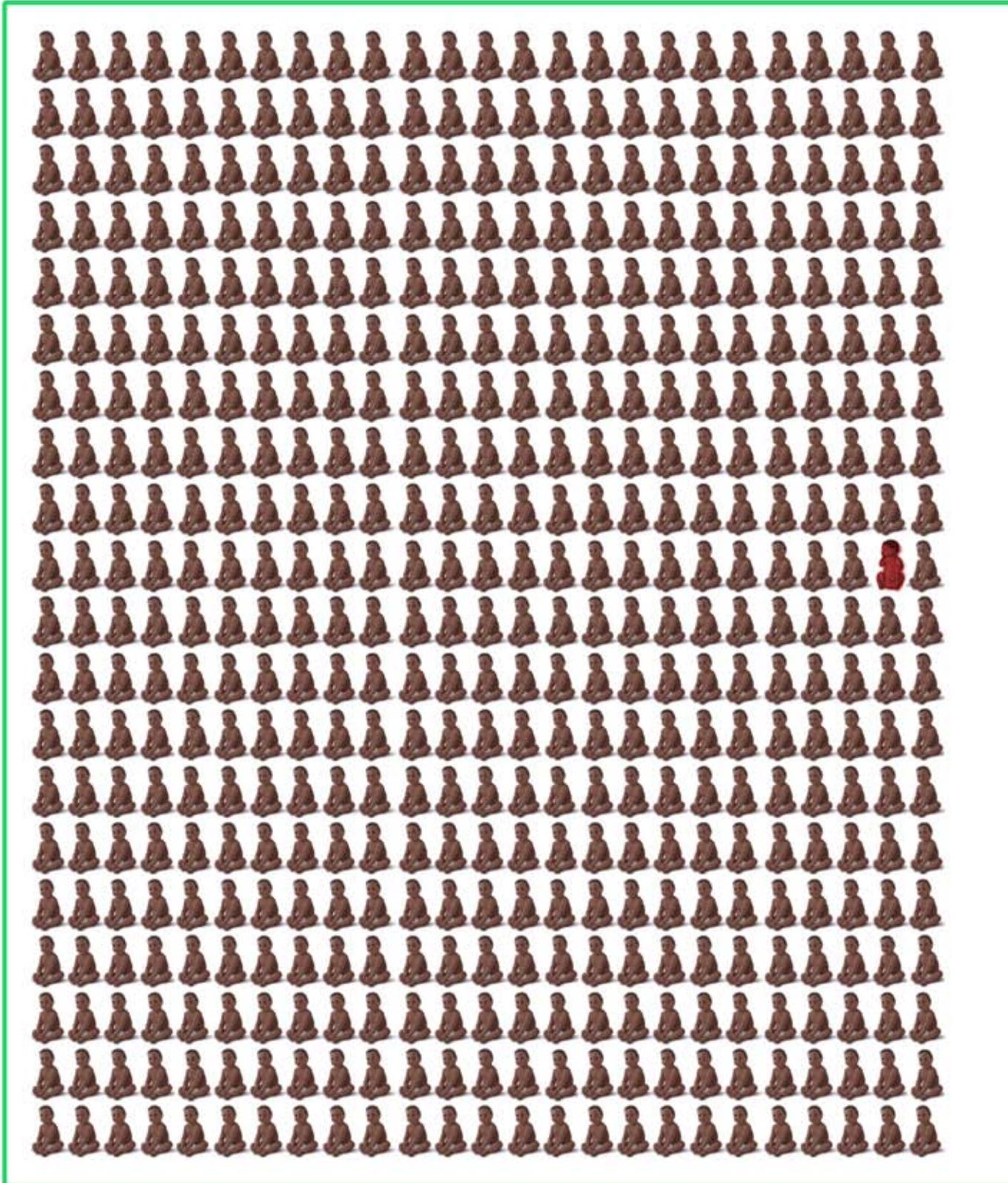
1/1000

This is the risk of having a baby with a neural tube defect for all Australian women.

If 1000 women got pregnant, 1 of those babies might have a neural tube defect.

If 1000 women got pregnant, 999 of those babies would be normal.

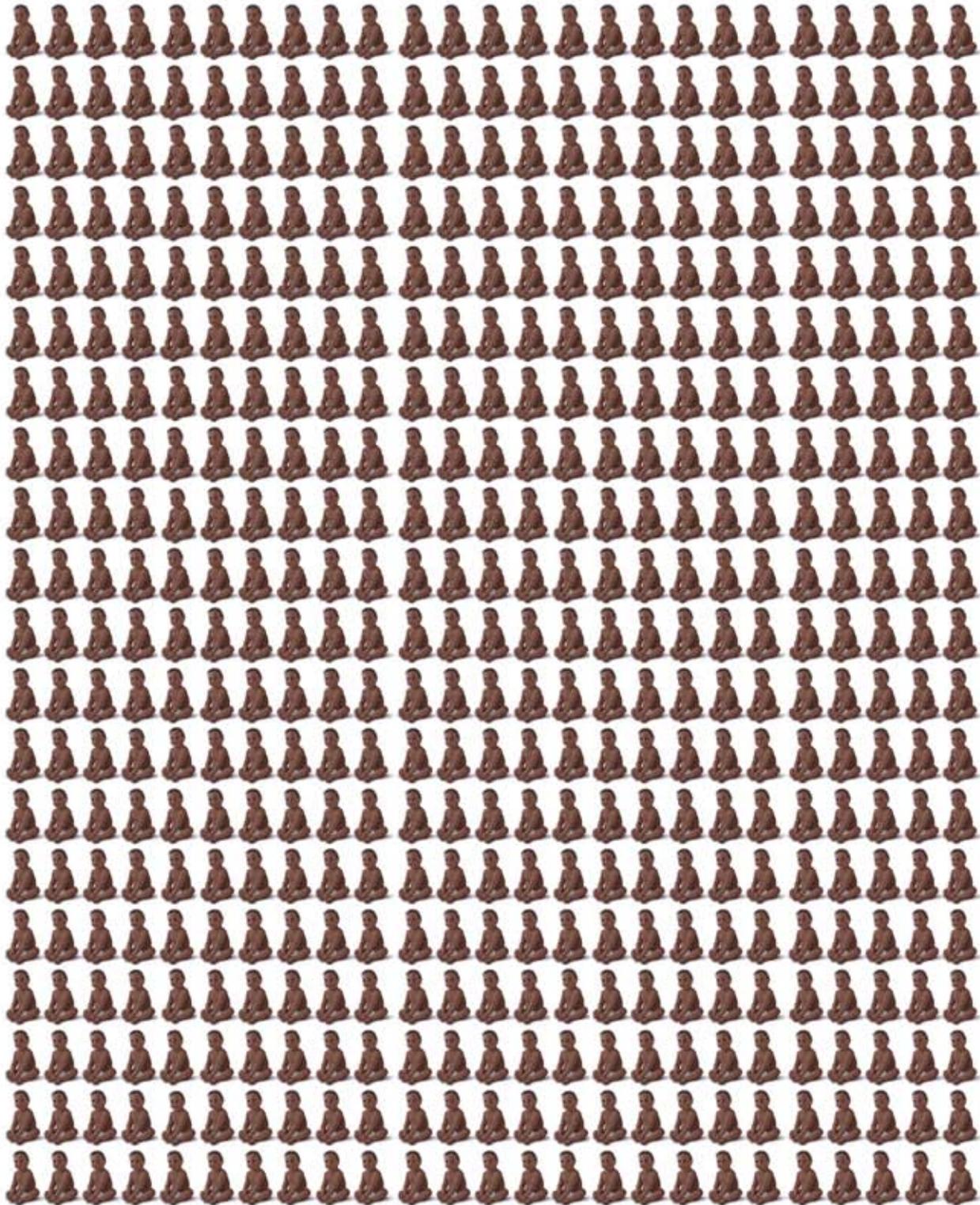
The risk of having a baby with a neural tube defect is twice as high for Indigenous women than non-Indigenous women.



This is also a 'low risk' result.

If 1000 women got pregnant, 1 of those babies might have a problem.

If 1000 women got pregnant, 999 of those babies would be normal.



More Information for Parents

About the screening tests:

Pre-test information for parents

http://www.wch.sa.gov.au/services/az/divisions/labs/geneticmed/documents/SAPathology-1PreTestInfo_SAMSAS2009_000.pdf

A decision aid to help parents decide whether they want screening for abnormalities like Down syndrome

<http://www.mcri.edu.au/downloads/prenataltestingdecisionaid.pdf>

Centre for Genetics Education (has lots of information and factsheets on genetic conditions, what causes them, and the screening and diagnostic tests)

<http://www.genetics.com.au/factsheet/index.asp>

An overview of prenatal testing

<http://www.genetics.com.au/pdf/factsheets/fs17.pdf>

About an increased risk result:

Increased risk of Down syndrome: What does it mean?

http://www.wch.sa.gov.au/services/az/divisions/labs/geneticmed/documents/SAPathologyD-SRisk_Website_001.pdf

Increased risk of Neural Tube Defect: What does it mean?

http://www.wch.sa.gov.au/services/az/divisions/labs/geneticmed/documents/SAPathologyNT-DRisk_Website_000.pdf

Increased risk of Trisomy 18 (Edwards syndrome): What does it mean?

http://www.wch.sa.gov.au/services/az/divisions/labs/geneticmed/documents/SAPathologyT18Risk_Website_001.pdf

About chromosomes and genes:

In relation to Machado Joseph Disease but tells a story about chromosomes and genetic disease

English: http://www.mjd.org.au/cms/file_library/Other/Other_281.pdf

Anindilyakwa: http://www.mjd.org.au/cms/file_library/Other/Other_282.pdf

Guidelines

Women's Business Manual (pages 96-100). Available through:

Centre for Remote Health

Phone: (08) 8951 4700

Email: crh.reception@flinders.edu.au

RANZCOG (Royal Australian and New Zealand College of Obstetricians and Gynaecologists)
College Statement on Prenatal Screening

<http://www.ranzcog.edu.au/publications/statements/C-obs4.pdf>

Three Centre Consensus Guidelines on Antenatal Care (Prenatal Screening for Down Syndrome, pages 19-22)

<http://www.health.vic.gov.au/maternitycare/anteguide.pdf>

More Information for Health Providers

SAMSAS (South Australian Maternal Serum Antenatal Screening) Program

Phone: (08) 8161 7285

Email: samsas.program@health.sa.gov.au

<http://www.wch.sa.gov.au/samsas.html>

SAMSAS Request Form (request form pads can be ordered directly from SAMSAS (08) 8161 7285. In the interim a form can be downloaded using the below link. In the absence of a SAMSAS request form a standard pathology request form may be used but must include patient's DOB, weight in kg, gestation and the name of the imaging group for a 1st trimester screen)

http://www.wch.sa.gov.au/services/az/divisions/labs/geneticmed/documents/SAPathologySAMSASForm3247Av2_000.pdf

Genetics in Family Medicine: The Australian Handbook for General Practitioners (see Section 3: Testing and pregnancy)

http://www.nhmrc.gov.au/your_health/egenetics/practitioners/gems.htm

Genetic Counselling

Visiting genetic services are available in some areas of the Northern Territory. Clinical geneticists can provide information to families who have a baby that has been diagnosed with a chromosomal abnormality and the risks associated with subsequent pregnancies.

Speak to your local doctor about organising these services.

References

Abeywardana S & Sullivan EA (2008). *Congenital anomalies in Australia 2002–2003*. Birth Anomalies Series No. 3, Cat. No. PER 41. Sydney: AIHW National Perinatal Statistics Unit. Available online:

[http://www.preru.unsw.edu.au/PRERUWeb.nsf/resources/CA+2/\\$file/ca3a.pdf](http://www.preru.unsw.edu.au/PRERUWeb.nsf/resources/CA+2/$file/ca3a.pdf) (accessed: 18/08/10)

Morris JK, Wald NJ & Watt HC (1999). *Fetal Loss in Down Syndrome Pregnancies*. *Prenatal Diagnosis*, 19:142–145. Available online:

[http://onlinelibrary.wiley.com/doi/10.1002/\(SICI\)1097-0223\(199902\)19:2%3C142::AID-PD486%3E3.0.CO;2-7/pdf](http://onlinelibrary.wiley.com/doi/10.1002/(SICI)1097-0223(199902)19:2%3C142::AID-PD486%3E3.0.CO;2-7/pdf) (accessed 18/08/10)

NHMRC (2006). *Nutrient Reference Values for Australia and New Zealand: Including Recommended Dietary Intakes*. Available online:

http://www.nhmrc.gov.au/_files_nhmrc/file/publications/synopses/n35.pdf (accessed: 15/09/10)

SAMSAS (2007). *First Trimester Screening NT Provider Progress Report 6*. Available online:

<http://whs.sa.gov.au/services/az/divisions/labs/geneticmed/documents/NTProvidersProgressReport6.doc> (accessed: 18/08/10)

SAMSAS (2009a). *Increased risk of Down syndrome: What does it mean?* Available online:

http://www.wch.sa.gov.au/services/az/divisions/labs/geneticmed/documents/SAPathologyD-SRisk_Website_001.pdf (accessed: 18/08/10)

SAMSAS (2009b). *Update 15: SAMSAS Prenatal Screening for Down syndrome, revised age specific performance*. Available online:

<http://www.wch.sa.gov.au/services/az/divisions/labs/geneticmed/documents/Update15T21andT-18Supplement.pdf> (accessed: 18/08/10)



© Menzie's School of Health Research 2010.
All rights reserved.

<http://www.menzies.edu.au>

Design, layout & illustrations by Mary Kate Wright

<http://www.mkillustrations.com>