Information for parents

“increased risk of Down Syndrome”
what does it mean?

Information from the South Australian Maternal Serum Antenatal Screening (SAMSAS) Programme,
Department of Chemical Pathology,
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Is there something wrong with my baby?

Everybody who receives a screening test report saying increased risk of Down syndrome straight away asks “is there something wrong with my baby?” At this stage the only answer is “probably not, but you should consider checking”.

97% of women who receive a report like this go on to have a normal healthy baby.

Why have I got this report?

We have performed a screening test which tells if there is a chance your baby might have Down syndrome. Your results indicate there is a chance (1 in 50) your baby might have Down syndrome.

What is Down syndrome?

Down syndrome arises when a baby has an extra copy of its chromosome 21. This happens in about 1 in 660 babies who are born. It is not known why it happens.

Children with Down syndrome usually have a characteristic appearance. They have varying levels of intellectual disability. They may also have one or more medical problems involving the bones, heart, bowel and thyroid gland. Some may have poor sight and hearing as well. With medical treatment and social support, children with Down syndrome may grow up in good health and with a resonable quality of life.

What does “increased risk” mean?

A screening report saying increased risk of Down syndrome is only a guide saying that your baby might have Down syndrome. It does not mean that your baby definitely has Down syndrome. In 29 out of every 30 pregnancies reported as being at increased risk of Down syndrome, the baby will be born normal and healthy.
In 1 pregnancy out of every 30 reported as being at *increased risk of Down syndrome* the baby will actually have Down syndrome or some other rare chromosomal problem. Only further testing will show whether or not your baby has Down syndrome.

**What is the further testing?**

The first test which usually follows a report saying *increased risk of Down syndrome* is an ultrasound scan. Ultrasound scanning is a way of seeing your baby using sound waves. There are no known dangers to either you or your baby from ultrasound scanning.

The ultrasound scan is done firstly to check on how many weeks pregnant you are. The ultrasound scan may show your baby is younger than you thought. If so, the *increased risk of Down syndrome* found by screening may change to *not at increased risk*.

Unborn babies with Down syndrome do not always look different from normal unborn babies on ultrasound scan. A normal ultrasound scan does not exclude the possibility your baby *might* have Down syndrome. You will be offered a test called *amniocentesis*.

**What is amniocentesis?**

Amniocentesis is a test which involves inserting a needle into your womb to take a sample of the fluid surrounding your baby. The needle is guided under ultrasound scan so as not to damage your baby. Testing the fluid will show definitely whether or not your baby has Down syndrome. This testing takes around 2 weeks to get the result.

There is a small risk that there will be damage to your pregnancy as a result of the amniocentesis, which may cause a miscarriage (about 1 in 200 amniocenteses performed).

**Must I have further testing?**

No, you do not have to. It is your choice. Discuss this with the doctor looking after your pregnancy, or contact the South Australian Maternal Serum Antenatal Screening Programme on (08) 8204 7285, who will be able to refer you to counselling and support services.

**Could my baby have something else?**

Very rarely, a baby might have another type of abnormality which shows up on ultrasound scan or amniocentesis testing. If this happens, your doctor will give you information about the likely effects this will have on the rest of your pregnancy and on the baby which may be born.

**What happens if my baby is shown to have Down syndrome?**

If your baby is shown to have Down syndrome, you will be given information about the likely effects this will have on the rest of your pregnancy and on the baby which may be born. You should ask advice about what medical problems your baby might have, and what support services are available to you and your family.

You will then have to decide whether to continue your pregnancy or to end it early.
Where can I get more information?

More information about maternal serum screening, and about what an increased risk result means, can be obtained from

the doctor who is looking after you during your pregnancy,
or

the SAMSAS Programme,
telephone (08) 8204 7285,
and its website
www.chempathadelaide.com/samsas

More information about Down syndrome is available from

the Down Syndrome Society of South Australia Inc., telephone (08) 8365 3510
and their website www.downssa.mtx.net
or

the Down Syndrome Association of Tasmania Inc., telephone (03) 6224 0490

The figures quoted here are from the South Australian/Tasmanian Maternal Serum Antenatal Screening (SAMSAS/TAMSAS) Programme, operating in the Department of Chemical Pathology of the Women’s and Children’s Hospital, Adelaide, South Australia. They do not apply to other maternal serum testing centres. If consumers have results from another centre for maternal serum screening, they are advised to check with that centre for the interpretations relating to their results. (Published September 2000)