

## “increased risk of Trisomy 18 (Edwards syndrome)” What does it mean?

*Information from the South Australian Maternal Serum Antenatal Screening (SAMSAS) Program,  
SA Pathology at Women’s and Children’s Hospital, North Adelaide 5006*

### ***Is there something wrong with my baby?***

Everybody who receives a maternal serum screening report saying *Trisomy 18: increased risk* straight away asks “is there something wrong with my baby?” At this stage the only answer that can be given is “probably not, but we should consider checking”.

**96% 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 is designed to tell if there is a greater than expected chance (*an increased risk*) your baby *might* have Trisomy 18.

Your results indicate there is a small chance that your baby *might* have Trisomy 18. The calculated risk is given on your report.

### ***What does “increased risk” mean?***

A report saying *Trisomy 18: increased risk* does *not* mean that your baby *definitely* has Trisomy 18. It is only a *guide* saying that the baby *might* have Trisomy 18. The chance of this being so, that is the *risk*, is shown on your report.

For example, if your report says *Trisomy 18: increased risk (1:100)*, it means that in **99 out of every 100 pregnancies** reported like this, **the baby will not have Trisomy 18**. However, in **1 pregnancy out of every 100** reported as *Trisomy 18: increased risk (1:100)*, **the baby will actually have Trisomy 18** or some other chromosomal problem.

All risk figures reported by SAMSAS are interpreted in this way.

### ***What is Trisomy 18?***

Trisomy 18 is a chromosome abnormality. Babies with Trisomy 18 have an extra chromosome 18. This happens in about 1 in 6000 babies. It is not known why it happens.

Children with Trisomy 18 have severe intellectual, developmental and physical disabilities.

They may have medical problems involving their heart, lungs and kidneys. Abnormalities in the bones, head and face development also occur.

Trisomy 18 is usually fatal - 90% of affected pregnancies fail before term. Of the 10% reaching term, half will not survive the first week and most will not survive the first year.

### ***Must I have further testing?***

No, you do not have to. It is your choice.

In making your choice you should consider the risk figure given on your report. SAMSAS takes a risk of 1:250 at time of screen as the cut off point for deciding what is or is not an increased risk. This figure is a guide only, and **you should decide for yourself** what is an acceptable risk for you and your family.

Discuss this with the doctor looking after your pregnancy, or contact SAMSAS on (08) 8161 7285, which will be able to refer you to counselling and support services. You can choose whether or not to have further testing of your pregnancy.

Only further testing will provide a definite yes/no answer to the question “Does my baby have Trisomy 18?”

### ***What is the further testing?***

#### ***If you are between 10 and 14 weeks pregnant:***

the test which can give you a definite answer to the question “Does my baby have Trisomy 18?” is **chorion villus sampling**. Chorion villus sampling (or CVS) involves inserting a needle into your womb and taking a very small piece of your baby’s placenta. The needle is guided by ultrasound so as not to damage your baby. Testing the piece of placenta will show whether or not your baby has Trisomy 18 or other chromosome abnormality. It takes around 2 weeks to get the result.

There is a small risk of miscarriage as a result of CVS (about 1 in 100-200 tests performed).

***If you are between 14 and 20 weeks pregnant:***

the first test which follows a report which says *Trisomy 18: increased risk* is an **ultrasound scan**. This is best done when you are around 18 weeks pregnant. Ultrasound scanning is a way of seeing your unborn baby using sound waves. There are no known dangers to either you or your baby from this type of ultrasound scan.

The ultrasound scan will check how many weeks pregnant you are (the calculation of the risk of Trisomy 18 in your baby depends on knowing this *exactly*) and will look for any abnormalities in your baby. Unborn babies with Trisomy 18 have characteristics which may be seen on detailed ultrasound scanning. If seen, these features strengthen the *increased risk* report and you will need to decide whether you want further testing. A normal ultrasound scan does *not* mean your baby *does not have* Trisomy 18.

The test which can give you a definite yes/no answer to the question "Does my baby have Trisomy 18?" is **amniocentesis**. Amniocentesis is a test which involves inserting a needle into your womb to take a sample of the fluid around your baby. The needle is guided by ultrasound so as not to damage your baby. Testing the fluid will show definitely whether or not your baby has Trisomy 18. This testing takes around 2 weeks to get the result.

There is a small risk of miscarriage as a result of the amniocentesis, (about 1 in 200 amniocenteses performed).

***Could my baby have something else?***

Occasionally, the ultrasound scan or the CVS/amniocentesis testing may show your baby to have some other abnormality. If your baby does have another abnormality your doctor will explain what it is and tell you what it means.

***What happens if my baby is shown to have Trisomy 18?***

If your unborn baby is shown to have Trisomy 18, 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 know that 90% of affected pregnancies fail before the baby is due to be born. You should ask advice about what medical problems your baby might have if it is born, 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 antenatal screening, and about what an *increased risk* result means, can be obtained from

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

or

the SAMSAS Program  
*telephone* (08) 8161 7285  
*email* [samsas.program@health.sa.gov.au](mailto:samsas.program@health.sa.gov.au)  
*website* [www.wch.sa.gov.au/samsas.html](http://www.wch.sa.gov.au/samsas.html)

*The figures quoted here are from the South Australian Maternal Serum Antenatal Screening (SAMSAS) Program, SA Pathology, operating at the Women's & Children's Hospital, Adelaide, South Australia. They do not apply to other maternal serum testing centres. (Revised Aug 2009)*