

Lethal skeletal dysplasias

Information for parents

With draft



Information to help you understand more about lethal skeletal dysplasias.

1. What is lethal skeletal dysplasia?

Skeletal dysplasias are a group of over 350 different conditions associated with abnormalities in the bones of the arms, legs, chest or, sometimes, the skull. Babies with these conditions have short limbs or a short trunk (chest and tummy) which are out of proportion with the rest of the body.

Some skeletal dysplasias cause very abnormal bone growth. In these cases, the chest and lungs do not fully develop and the baby cannot survive. These conditions are called lethal skeletal dysplasias.

2. How common is it?

Lethal skeletal dysplasias occur in about 1 of every 10,000 births.

3. How is it diagnosed and confirmed?

A lethal skeletal dysplasia is usually noticed at the Fetal Anomaly ultrasound scan carried out between 18 weeks and 21 weeks of pregnancy.

A second scan, carried out by a specialist, will be needed to confirm the diagnosis. Doctors may also offer you further tests to confirm the exact type of skeletal dysplasia that your baby has. Even after all of these tests, a precise diagnosis may not be made before birth.

4. Is there any treatment?

Unfortunately, there is no treatment for lethal skeletal dysplasias.

5. What is the outlook for the baby?

Sadly, babies with these conditions die during pregnancy or shortly after birth.

6. What happens next?

You will be given the chance to talk to specialists about your options. You will have the opportunity to discuss the possible implications of ending or continuing your pregnancy.

You will be offered a termination to end the pregnancy. If you choose to have a termination your health professional will talk to you about the procedure and support you through the process.

If you choose to continue your pregnancy, your healthcare team will help you plan how your care, including delivery, is managed. As there is no cure for lethal skeletal dysplasias, doctors will discuss palliative care with you. Palliative care is care aimed at relieving the symptoms of a condition rather than treating or providing a cure. You may be referred to the children's palliative care team and the local bereavement service.

Whatever you decide, your decision will be respected and you will be supported by your midwife and doctor.

7. How likely is it to happen in a future pregnancy?

You are much more likely to have a normal, healthy baby in your next pregnancy than to have another baby with a lethal skeletal dysplasia.

Anyone can have a baby with a lethal skeletal dysplasia. There is no way to prevent this condition. It is not due to anything you have or have not done.

In most cases, lethal skeletal dysplasias occur where there is no family history of the condition. In rare cases, other family members may have had another skeletal dysplasia (either lethal or non-lethal).

You will be given the opportunity to speak with a genetic counsellor, who should be able to talk through your chances of having another pregnancy affected by this condition. The chance of you having another baby with similar problems will depend on the exact diagnosis of your baby. The exact diagnosis can usually only be made after X-rays of the bones and a detailed examination of the bones after death. This is called a post-mortem examination.

8. Where can I get more information and support?

You may feel you only want to talk to your family and friends, or a particular doctor or midwife from the hospital. However, there are other people and organisations that can provide information, help you make your decisions and support you in your pregnancy and afterwards. You may also want to talk things through with the hospital chaplain or your own minister or faith leader.

9. Further information, charities and support organisations

The following organisation can offer you support. There are details of other support organisations on our website at www.fetalanomaly.screening.nhs.uk. If you have any questions about the information in this leaflet or where the information came from, email us at enquiries@ansnsc.co.uk.

Antenatal Results and Choices (ARC)

Email: info@arc-uk.org

Helpline: 0845 077 2290

Website: www.arc-uk.org

Antenatal Results and Choices (ARC) provides information and support to parents before, during and after antenatal screening and diagnostic tests, especially those parents making difficult decisions about testing, or about continuing or ending a pregnancy after a diagnosis. ARC offers ongoing support whatever decisions are made.

This information has been produced on behalf of the NHS Fetal Anomaly Screening Programme for the NHS in England. In other countries, check with a health professional to find out whether there are any differences in approaches to screening.

This leaflet has been developed through consultation with the NHS Fetal Anomaly Screening Programme expert groups.

All of our publications can be found online at www.fetalanomaly.screening.nhs.uk.

NHS staff can reproduce any information in this booklet. Please make sure you have permission to reuse images. You must discuss any amendments with the original author.

© NHS Fetal Anomaly Screening Programme 2012

If you have any comments or feedback, email them to us at enquiries@ansnsc.co.uk.