NHS

Screening Programmes

Fetal Anomaly

Lethal skeletal dysplasia information for health professionals

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Aim

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Definition, causes and risk factors

Skeletal dysplasias are a group of over 200 conditions which are caused by mutations in the genetic make up of a person, most of which are associated with control of bone development and growth. The majority of skeletal dysplasias occur where there is no family history of similar problems. Parents who themselves have a skeletal dysplasia may be at higher risk of having a child with a skeletal dysplasia. The risk may be as high as 1 in 2.

There are a small number of skeletal dysplasias that are inherited in an autosomal recessive manner, where there may be a risk to siblings of an affected child. Skeletal dysplasias are associated with restricted growth and are characterised by disproportionate short stature and abnormalities of the size and shape of arms, legs, and/or of the trunk.

Some of the skeletal dysplasias fall within the group called lethal skeletal dysplasias. These conditions are associated with very severe abnormalities of bone growth including rib growth. As a result, the chest and lungs of babies with lethal skeletal dysplasias do not fully develop. These conditions are associated with intrauterine death or death soon after delivery in most cases. The most common of the lethal skeletal dysplasias include thanatophoric dysplasia, and Type II osteogenesis imperfecta.

Screening and diagnosis

Lethal skeletal dysplasias are usually detected at the 18^{+0} – 20^{+6} weeks' fetal anomaly ultrasound scan stage as a result of the measurements of the long bones. The bones most often affected in these conditions are the bones of the limbs, and the ribs. There is usually a reduction in bone length and there may also be bowing of the bones. Careful assessment of bone growth using a serial scans may be necessary. Assessment of rib and lung growth is vital in differentiating lethal and non-lethal skeletal dysplasias.

A molecular diagnosis, looking for mutations in a specific DNA gene to confirm the exact condition affecting the baby, is usually impossible unless there is a family history where a molecular diagnosis has been made in a previously affected baby, or where the baby has features of one of the common sketelal dysplasias e.g thanatophoric dysplasia.

A diagnosis of achondroplasia, a type of skeletal dysplasia also known as dwarfism, is often considered when long bone shortening is identified on ultrasound scan. In general it is very unusual to see significant reductions in bone growth in babies with achondroplasia until around 24 weeks of gestation.

The NHS Fetal Anomaly Screening Programme will be audited against a 60% detection rate of all cases of lethal skeletal dysplasia at the 18^{+0} – 20^{+6} weeks' fetal anomaly ultrasound scan.

Prevalence

These skeletal dysplasias as a group affect around 0.95 per 10,000 deliveries. The prevalence of individual conditions varies enormously with the most common affecting around 1 in 25,000 babies and the least common occurring in less that 1 in 100,000 pregnancies.

The majority of the skeletal dysplasias affects all populations equally. There are some which are more common in particular ethnic groups e.g. diastrophic dysplasia is more common in Finland.

Treatment

There is usually no treatment for the lethal skeletal dysplasias. These conditions are almost always fatal, resulting either in intrauterine death or death shortly after birth.

Prognosis

The baby will not survive long after it is born. These conditions are almost always fatal.

Prevention

Where a previous baby has been affected by a lethal skeletal dysplasia there may be a risk to subsequent children depending on the exact diagnosis. In some cases prenatal diagnosis may be possible in a future pregnancy.

There are no environmental factors that predispose to a skeletal dysplasias.

Referral pathway

Following the diagnosis of a lethal skeletal dysplasia at the 18^{+0} – 20^{+6} weeks' fetal anomaly ultrasound scan, a second medical opinion should be sought. A termination of pregnancy should be offered to allow women the option of not continuing with their pregnancy. Paediatric referral in the antenatal period will be appropriate for women who elect to continue their pregnancy.

A formal diagnosis of the condition after delivery is extremely important. This is most commonly achieved by postmortem examination, which may include genetic testing. If a postmortem is refused then obtaining full body x-rays in AP and lateral view of the baby after delivery is vital. Ideally a sample of cord blood should be collected and sent for DNA extraction and storage to the appropriate regional genetics service. These may allow a definitive diagnosis to be made. This is essential for provision of adequate prenatal counselling and, if necessary, diagnosis in future pregnancies. Parents should be referred for genetic counselling.

Further information, charities and support groups

Antenatal Results and Choices (ARC)
73 Charlotte Street
London
W1T 4PN

Tel: 0207 631 0285
Email: info@arc-uk.org
Website: www.arc-uk.org

Antenatal Results and Choices (ARC) provides impartial information and individual support to parents whether they are going through antenatal screening or whose unborn baby has been diagnosed with an abnormality.

Child Growth Foundation
2 Mayfield Avenue
Chiswick
London
W4 1PW

Tel: 020 8995 0257
Email: info@childgrowthfoundation.org
Website: www.childgrowthfoundation.org

The Child Growth Foundation is the UK's leading charity relating to children's growth.

Contact a Family (CAFAMILY)
209 to 211 City Road
London
EC1V 1JN

Tel: 0808 808 3555
Email: info@cafamily.org.uk
Website: www.cafamily.org.uk

Contact a Family is a charity which provides support, advice and information for families with disabled children, no matter what their condition or disability.

DIPEx
PO Box 428
Witney
Oxon
OX28 9EU

Email: info@healthtalkonline.org
Website: www.healthtalkonline.org &
www.youthhealthtalk.org

DIPEx has created a unique database of personal and patient experiences through in-depth qualitative research into over 40 different illnesses and health conditions. The results of their research are published on two websites which are aimed at patients, their carers, family and friends, doctors, nurses and other health professionals. Their target is to complete at least 100 conditions within the next 5 – 10 years.

The Miscarriage Association
c/o Clayton Hospital
Northgate
Wakefield
West Yorkshire
WF1 3JS

Tel: 01924 200799
Email: info@miscarriageassociation.org.uk
Website: www.miscarriageassociation.org.uk

Miscarriage can be a very distressing, frightening and lonely experience. The Miscarriage Association provides support and information.

Further information, charities and support groups continued...

Restricted Growth Association (RGA)
RGA Office
PO BOX 1024
Peterborough
PE1 9GX

Tel: 01733 759 458
Email: office@restrictedgrowth.co.uk
Website: www.restrictedgrowth.co.uk

The RGA is a UK based charity that provides vital information and support to improve the quality of life for persons of restricted growth and their families.

Stillbirth and Neonatal Death Society (SANDS)
28 Portland Place
London
W1B 1LY

Tel: 0207 436 588[illegible]
Email: helpline@uk-sands.org
Website: www.uk-sands.org

SANDS support anyone affected by the death of a baby and promote research to reduce the loss of babies' lives.

This information has been produced on behalf of the NHS Fetal Anomaly Screening Programme. The leaflets have 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

If you have any comments on this booklet or enquiries for the Programme please contact us at the address below:

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