SOFT UK

Support Organisation for Trisomy 13/18

Your Pregnancy

SOFT UK is a Registered Charity: 1002918 (England and Wales),

Your Pregnancy

SOFT UK

INTRODUCTION

SOFT UK is a Support Organisation for Trisomy 13 (Patau's Syndrome) and Trisomy 18 (Edwards' Syndrome). It was founded in 1991 by two mothers who both had children affected by these conditions. They found that was little information or support available for parents when their children were born.

This information can be used by anyone, but has been written primarily for parents who have been told they have a higher chance of trisomy 13 or trisomy 18 during antenatal screening or those with a confirmed prenatal diagnosis for their baby. Our aim is to help you gain the information, advice and support you may require during your pregnancy and to make the decisions that are right for you and your child.

We have supported many hundreds of families affected by trisomies 13 and 18, whether they have chosen to continue or end a pregnancy, those who are bereaved and those who are caring for surviving babies, children and adults.

We recognise that every family is unique. What may be right for one child or family is not necessarily right for another. SOFT UK does not recommend a particular decision about a pregnancy or treatment for your child. We seek to provide information and support to parents and families along their entire trisomy journey, connecting families to share experiences and enabling parents to make informed decisions that are right for them.

SOFT UK does not represent or endorse any political, religious or campaign groups.

UNDERSTANDING THE GENETICS

WHAT IS A CHROMOSOME?

Chromosomes are thread-like structures located inside the cells in our body. Each chromosome is made of protein and a single molecule of deoxyribonucleic acid (DNA). Passed from parents to their child, DNA contains the specific instructions that make each person unique.

There are 22 pairs of chromosomes, numbered 1-22 in order of size, plus a pair of sex chromosomes, XX in females and XY in males, making a total of 46 chromosomes in each cell.

WHAT CHROMOSOMES DO?

To create a baby, it is important that reproductive cells, such as eggs and sperm, contain the right number of chromosomes and that those chromosomes have the correct structure. If not, the resulting baby may fail to develop properly. This is what happens when the baby has trisomy 13 or trisomy 18 when there are 3 copies of chromosome 13 or 18 instead of the usual two.

When an egg is produced by the mother, the pairs of chromosomes split into two so that only one of each pair remains in the egg. The same happens with the sperm cell. This means that the egg cell and the sperm cell each contain 23 chromosomes. Therefore, when a healthy egg is fertilised by a healthy sperm, the fertilised egg has 46 chromosomes and is the unique blueprint for the baby that grows.

ABOUT TRISOMY

The “tri” in the word trisomy means three. In trisomy there is one set of three identical chromosomes among the normal pairs in each cell. In trisomy 13 there are 3 copies of chromosome 13 instead of the usual two and in trisomy 18 there are 3 copies of chromosome 18 instead of the usual two. This means there are 47 chromosomes in each cell, instead of the usual 46.

The extra chromosome may be in the egg cell produced by the mother, or the sperm cell produced by the father, and the additional genetic material affects every stage of the development of the baby.

HOW COMMON ARE TRISOMY 13 AND TRISOMY 18?

About 1.6 in 10,000 pregnancies are diagnosed with trisomy 13. and about 4 in 10,000 pregnancies are diagnosed with trisomy 18 *(Goel N, Morris J. K. et al., 2019)*. They are the second and third most common autosomal trisomies after Downs syndrome (trisomy 21).

Most cases result from a random change in the egg or sperm in healthy parents. This change is not caused by anything parents did or did not do before or during pregnancy.

Around 80% of babies born with trisomy 13 and 94% of babies born with trisomy 18, will be affected by **Full Trisomy**. *(Cereda A & Carey J., 2012)* This means that the extra chromosome originated at the point of conception during the process called **Meiosis** and is present in every cell. This condition is not inherited from parents.

The remainder of babies will have trisomy as a **Mosaicism** or **Translocation**.

PARTIAL TRISOMY

Partial trisomy is rare. A partial trisomy means the affected person has the usual **two** copies of the affected chromosome **plus** a partial or incomplete extra copy.

Sometimes the smaller copy attaches itself to another chromosome, called an **Unbalanced Translocation**. This can make it more difficult to diagnose.

The partial copy can be of any length, from just a small part to almost the full chromosome.

Some cases of partial trisomy are caused by hereditary factors.

MOSAICISM

Mosaicism can occur when there is a problem with **Mitosis**, which is the process of cell division that occurs as the embryo develops. If the embryo does not divide evenly as it develops, then some cells may have the extra chromosome 13 or 18 and some cells have the usual pair.

Therefore, the affected baby has 47 chromosomes in **some** cells but the typical 46 chromosomes in other cells.

Children with mosaic or partial trisomy 13 or trisomy 18 may have a much larger range of health or developmental outcomes than those with full trisomy. The exact outcome depends upon how many of their cells contain the extra chromosome and where those cells are located in the body. Some children may have much less serious developmental and physical health issues or even have no observable effect. However, some children may seem as affected as a child with full trisomy.

RELATED DISORDERS

Ring chromosomes and deletions affecting chromosomes 13 or 18 are very rare but can occur. Ring chromosomes are when loss of genetic material from both ends of the chromosome causes it to form a ring. Deletions are when a part of the chromosome is missing. The long arm of the chromosome is referred to as q, and the short arm as p. A deletion of the long arm of chromosome 13 would be 13q-, and numbers can be used to specify exact areas of the chromosome.

DOES THE AGE OF THE MOTHER AFFECT THE RISK?

The risk of trisomy increases with maternal age, showing an increase in cases in mothers over 35 years of age. *(A. Cereda, J Carey, 2012)* However, most babies with these conditions are born to mothers in their twenties because more babies are born to women in this age group.

HOW MIGHT THE BABY BE AFFECTED?

Trisomy 13 and 18 will affect each individual child differently and this is difficult to predict.

There are a number of impacts of trisomy 13 and trisomy 18, some of which can be serious. It is unlikely that a baby will have all of the physical health issues identified below.

The following table shows the associated anomalies that were present in 648 babies born with trisomy 18, and 306 babies born with trisomy 13, as recorded in 25 registers of congenital anomalies between 2000 – 2011. *(EUROCAT registries in 2000-2011).* This gives an idea of the type of anomaly and how frequently it may be seen.

Anomaly	Trisomy 18 (%)	Trisomy 13 (%)
Brain, including Microcephaly and Holoprosencephaly	 3 15	 20 31
Eye	2	23
Heart Severe heart anomalies	60 14	45 14
Respiratory	4	6
Digestive system	13	8
Abdominal wall defects	5	10
Urinary	13	12
Genital	4	9
Limb	21	38
Oro-facial clefts	7	36

HOLOPROSENCEPHALY (HPE)

Holoprosencephaly is when the front part of the brain fails to develop into separate right and left halves. Generally, this severely affects the function of the brain, although among children with

this condition there is considerable variability in the degree of malformation and its effects. Babies diagnosed with this condition usually have a small head and excessive fluid in the brain. Although a baby with holoprosencephaly may not have abnormal chromosomes, it is something that occurs in babies primarily with trisomy 13.

HEART DEFECTS

Trisomies 13 and 18 are often associated with congenital heart defects (CHD). In trisomy 13 80 % of babies may be affected with CHD and in trisomy 18 it is 90%. *(Peterson J.K. et al., 2017).* These defects include **atrial septal defects** (ASD) or **atrioventricular septal defects** (AVSD) when there are holes between the chambers of the right and left sides of the heart; **patent ductus arteriosus** (PDA) which is a persistent opening between the two major blood vessels leading from the heart; **atrioventricular septal defect** (AVSD) when there are holes between the chambers of the right and left sides of the heart; **tetralogy of Fallot** (TOF) which affects normal blood flow through the heart; and others. These defects can often be successfully repaired if the child is able to undergo surgery.

DEVELOPMENT

Children with trisomy 13 or 18 will have some level of learning disability and developmental delay, which can be severe.

SURVIVAL RATES

It is not generally realised that up to half of all pregnancies are miscarried, often before a woman realises she is pregnant, and 1 in 5 confirmed pregnancies end naturally within 3 months. *(Cohain J. S. et al., 2017).* It is thought that many of these losses are caused by a chromosome abnormality, but a woman is unlikely to be offered chromosome tests to confirm the reason unless previous problems during pregnancy have indicated that this might be a cause.

It has been estimated that as many as 49% of pregnancies diagnosed with trisomy 13 and 72% diagnosed with trisomy 18 are lost in early pregnancy. This rate reduces as the pregnancy progresses. Between 24 weeks and full term the rate of miscarriage reduces to around 35% for trisomy 13 and 59% for trisomy 18. Male babies with trisomy 18 appear more likely to be lost to miscarriage. *(Morris J. K. & Savva G. M., 2008)*

Between 2015 – 2017 8.7% of babies diagnosed with Patau's syndrome and 10.8% of babies diagnosed with Edwards' syndrome resulted in a live birth in England (*National Congenital Anomaly and Rare Disease Register 2017*)

CAN TRISOMY BE INHERITED ?

Most cases of trisomy 13 and 18 occur as a result of a random event at conception and are not inherited from either parent.

A baby can have **Partial Trisomy** when the parents have normal chromosomes, but in rare cases a parent may be a carrier if he or she has a balanced rearrangement of their chromosomes. A balanced rearrangement is when a piece of one chromosome is attached to another without there being any extra or missing genetic material. The (carrier) parent is healthy and unaware of the different pattern in their chromosomes unless tests are performed. When the parent cell divides to produce an egg or sperm the following can happen:

- The baby inherits the normal chromosome from each pair and has normal chromosomes

and is healthy

- The baby inherits the chromosome with a piece missing and the chromosome with the extra piece attached i.e. both the rearranged chromosome so overall there is not extra or missing genetic material. The baby is healthy but caries the balanced translocation which may affect any children they have.
- The baby inherits a normal chromosome and the one with an extra piece i.e. extra genetic material and has a **Partial Trisomy** of that chromosome.
- The baby inherits the chromosome and the one with a piece missing, ie. there is genetic material missing and the baby has a **deletion** of that chromosome.

Other family members may carry the balanced translocation and a clinical geneticist can explain how a future baby might be affected. Larger hospitals usually have a Genetics Department, and to obtain a referral you can ask your Obstetrician or G.P.

GENETIC INFORMATION

Genetics is the study of human cells, and a Clinical Geneticist will interpret the results of chromosome tests taken from blood, skin, bone marrow, or connective tissue. When tests during pregnancy have shown a chromosome abnormality the clinical geneticist, if consulted, will discuss the results with the family.

Depending on the particular abnormality affecting the unborn child, it may not always be possible to give you the definitive answers you seek, and in some cases the information will be a mixture of the certain, the probable, the possible, and an element of 'wait and see'. Just as all babies have different characteristics, so babies with trisomy 13 or 18 will have individual differences although sharing the same syndrome.

When a **Translocation** is present it can be additionally difficult to predict the effect on the child as this depends on the exact amount of genetic material that is extra or missing. This is also true when **Mosaicism** is diagnosed as it then depends upon how many cells contain the additional chromosome and where they are located in the body.

Blood cells may show a particular percentage of trisomy cells, skin cells can show a higher or lower percentage, and cells in key organs such as the brain cannot be examined.

A detailed ultrasound scan can show abnormalities of organs such as the heart or kidneys, but it is not always possible to predict how severe the effects will be.

PRENATAL SCREENING AND DIAGNOSTIC TESTS

All pregnant women are offered screening tests, such as scans and/or blood tests, to assess the chance of a chromosomal abnormality in the baby. These tests are non-invasive and so do not have any risk to the baby. Screening tests can indicate if a pregnancy is in a low or high chance category, but do not give a diagnosis for the baby.

Screening Tests

You will be offered a number of different screening tests when you are pregnant – blood tests, ultrasound scans or a combination of both. Some of these tests are to check if the baby has a chance of a chromosomal condition such as trisomy 13 (Patau's syndrome), trisomy 18 (Edwards' syndrome) or trisomy 21 (Downs syndrome).

It is your choice whether you undergo any or none of these screening tests. The screening tests themselves do not pose any risk to you or your baby's health.

Screening tests can tell you whether your baby has a **higher chance** of a problem, but they cannot tell you for certain or give you a definite diagnosis of a condition. Results are usually reported as a statistical chance – a higher chance or lower chance of the baby having a chromosomal condition. They are not 100% accurate therefore, sometimes the results of a screening test can make parents worried when their baby does not have a condition. The opposite is also true and sometimes the tests indicate that there is a low risk, when a baby does have a condition. The efficacy of screening tests varies depending on which type of test you have, so it is important to read the information for the specific test you are having.

If you have received a higher chance result of your baby having trisomy 13 or trisomy 18 from the **combined** or **quadruple screening test** you have 3 options:

- You can decide to have no further testing
- You can have a more accurate non-invasive prenatal testing (**NIPT**) which is a simple blood test and can tell you whether your baby is likely to have the condition. This test has a high level of accuracy but is not 100%. From 2021 it is available on the NHS in England, Scotland and Wales.
- You can have an invasive diagnostic test – chorionic villus sampling (**CVS**) or **Amniocentesis**.

DIAGNOSTIC TESTS

Diagnostic tests will be able to tell you if your baby **definitely has** a chromosomal condition such as trisomy 13 or 18. Diagnostic antenatal testing carries a small risk to the pregnancy. The risk incurred with each form of testing varies and is detailed in the **specific** information on that test. Risks can be higher in a multiple pregnancy, so please discuss this specifically with your healthcare team.

CHORIONIC VILLUS SAMPLING

CVS is performed from around 11 weeks of pregnancy.

A small sample of the placenta is obtained, which can give information about the baby because the baby and placenta developed from the same cell.

Using ultrasound scan for guidance, a needle is inserted through the woman's abdomen to obtain a small sample from the placenta.

Occasionally a sample from the placenta can be obtained through the cervix, in a similar manner to having a cervical smear.

The risk of miscarriage is between 1 and 2%, and the test result is slightly more likely to be inconclusive than amniocentesis.

AMNIOCENTESIS

Amniocentesis can be performed from 15 weeks of pregnancy onwards. (Before this time there is not enough amniotic fluid to safely take a sample)

Using ultrasound scan for guidance a needle is inserted through the woman's abdomen to

obtain a small sample of amniotic fluid which contains cells from the baby. The risk of miscarriage is about 1%

CORDOCENTESIS

This test can be carried out after 18 weeks of pregnancy and results take about a week. A needle is inserted into the mother's abdomen and a sample of the baby's blood is taken from the umbilical cord. The risk of miscarriage is between 1 and 4%.

ULTRASOUND

All pregnant women are usually offered two ultrasound scans. Ultrasound scans cannot diagnose a chromosome abnormality but can look for structural abnormalities, (for example, a heart defect or holoprosencephaly), and can look for **chromosomal markers**. These markers, or 'clue signs', are signs that can be seen in normal babies, but are more common in babies with a chromosome problem. Abnormalities seen on a scan, or a cluster of chromosomal markers may alert medical staff to suspect a chromosome problem, but such a diagnosis can only be confirmed by an invasive diagnostic test.

DATING SCAN

The first scan is called an early dating scan and it usually happens after 8 weeks of pregnancy.

The purpose of the dating scan is:

- To find out how many weeks pregnant you are
- To check whether you are expecting more than one baby
- To measure the size of the fluid area at the back of the baby's neck (this area is called the nuchal translucency (NT)
- To check that the baby is growing in the right place
- To check your baby's development

Some abnormalities may also be detected at this scan such as neural tube defects, for example spina bifida.

MID TRIMESTER ANOMALY SCAN

The second ultrasound scan is often called a mid-trimester anomaly scan and it usually happens between 18 - 20 weeks of the pregnancy and screens for major structural anomalies. The objective of this scan is to:

- Offer choice to women and their partners about their screening options.
- Identify serious abnormalities at a time when choices can be made whether to continue or terminate a pregnancy.
- Identify abnormalities which may benefit from or need early treatment following delivery.

The aim of this scan is to check your baby's development and any problems with your baby. It is important to note that not all abnormalities can be seen on an ultrasound scan; just because nothing is picked up on the scan does not mean that your baby will have no problems or anomalies. There are things that the ultrasound scan cannot pick up.

If the sonographer thinks that there may be a problem with your baby, you may be offered a further scan or an invasive diagnostic test such as chorionic villus sampling or amniocentesis.

You can find more detailed information about prenatal screening tests and their outcomes at Antenatal Results and Choices (ARC) www.arc-org.uk

FETAL CARDIOLOGY

Many babies with trisomy 13 or 18 have heart defects and specialist centres can perform a detailed scan of the baby's heart during pregnancy.

MAKING A DECISION ABOUT YOUR PREGNANCY

When trisomy 13 or 18 is identified as a high chance via a screening test, or is diagnosed by a diagnostic test during pregnancy, full information about the condition may not be available immediately. It is not always possible to give full information about how your baby will be affected. These are rare conditions which your medical team may not have much prior experience of. Try to find out as much information that you can, so that you can make an informed decision that is right for you and your family.

'Any one of us may see only a few families of babies with trisomy 13 or 18 and may not have the latest information to hand. This booklet can be used as a basis for discussion between a health professional and parents.' (Una MacFadyen, Paediatric Consultant (Retired) and Professional Adviser to SOFT UK)

You are likely to be given the choice of continuing or ending the pregnancy by medical termination.

In the case of twin or multiple pregnancies when only one of the babies has abnormal chromosomes, you may be given the option of ending the pregnancy of the affected baby. You will be warned there is a risk that the baby with normal chromosomes may be miscarried as a result.

You should not be pressurised into making a quick decision. In most cases a week or two is not going to alter the situation radically. You are likely to be in shock, and your emotions will roller coaster from hope to despair. Give yourself time to accept what has happened, and time to realise this baby may be not the baby you planned before you even think about making a decision.

'The specialist explained trisomy 18 in detail, and he strongly recommended termination. We were mourning the loss of our imagined perfect baby and bereavement is never a good time to make decisions.' (Parent)

You might find it helpful to talk to and listen to your medical team, family and friends to help gather information and gain some perspective. Contact SOFT UK and we can put you in touch with families who have had a similar experience to you – hearing about the lived experience of others can be helpful. Take your time to ensure you are making an informed decision. Ultimately, the decision you make should be the right one for you and non-one else can tell you what that should be.

'We were so grateful for the chance to talk to other families who had faced a prenatal diagnosis, it really helped us not feel so alone, as well as giving us useful facts.' (Parent)

ASK QUESTIONS

When you meet with your medical team prepare a list of every question you want to ask. Don't feel intimidated by your doctors or the medical terms they use. Ask them to explain everything you do not understand. Write down important points as you may not remember them when you get home. But remember that not every question will have an answer.

SEARCH FOR INFORMATION

Talk to your obstetrician, paediatrician and geneticist. There is a wealth of information available via the internet but beware of information that is out of date, is not evidence based or from a reliable source. Written information about the medical problems associated with trisomy 13 and 18, information on caring for a baby with one of these conditions and family stories can be obtained from the SOFT UK website.

DON'T BLAME YOURSELF

There is NOTHING you could have done to prevent this.

'I felt devastated. What had I done to cause this to happen? I asked and asked and was told that it was just something that happened in a few cases.' (Parent)

When you are told the results of screening or diagnostic tests show that your baby is affected by trisomy 13 or 18, it can be like a bereavement. The feelings of grief, loss and guilt can be overwhelming.

IF YOU AND YOUR PARTNER DISAGREE WHAT DECISION TO TAKE

If as a couple, you cannot agree on what to do, counselling may help to explore your own and your partner's fears in a sensitive way.

WHEN YOUR PREGNANCY IS ENDED BY TERMINATION

Making the decision to end the pregnancy by termination can be incredibly difficult and can raise questions of guilt, morality and ethics, and this can make it difficult for you to discuss this option with family and friends. SOFT UK can provide contact with trained Support Volunteers who have been through a similar situation, which can lessen the sense isolation.

If you decide to end your pregnancy by termination, you can discuss how this will happen with your medical team. There are 2 types of termination: medical and surgical. Before 24 weeks, you should be given a choice unless there are medical reasons why one method would be safer for you. You should receive a clear explanation about what your options are so you can make an informed decision.

MEDICALTERMINATION

This involves taking medicine to end the pregnancy and it can be used at any stage of pregnancy. Two doses of medicine are taken 48 hours apart. A medical termination can be managed at home, but if you are more than 10 weeks pregnant, you are more likely to need to stay in hospital.

SEEING AND HOLDING YOUR BABY

Some parents decide that they would like to name or see or hold their baby after a medical termination. There is no right or wrong thing to do – it is about what you feel is right for you. If you do not want to see the baby, you can ask hospital staff to take a photograph or hand and footprints.

'She was only 2lbs, 7oz. Her skin was soft and pink and she had this beautiful dark hair. We kept her with us all night, cuddling her and taking photographs. Then we finally let her go.' (Parent)

SURGICAL TERMINATION

Surgical termination will be performed in one of two ways depending on how far along you are in your pregnancy. In both cases, you will have a general anaesthetic which means you'll be asleep or conscious sedation where you'll be relaxed, but awake.

You will not be able to see or hold your baby or have a photograph taken if you have a surgical termination.

Vacuum or suction aspiration

This can be used up to 15 weeks of pregnancy. The doctor uses a local anaesthetic to numb the entrance to the womb (the cervix). A tube is then inserted through the cervix) and the pregnancy is removed using suction. Vacuum aspiration takes 5 to 10 minutes, and most women go home a few hours later.

Dilatation and evacuation (D&E)

This is used from around 15 – 24 weeks of pregnancy. It involves inserting special instruments – forceps and suction, through the cervix and into the womb to remove the pregnancy. The baby is not removed intact.

It normally takes 10 to 20 minutes, and you might be able to go home the same day.

If you are 23 weeks pregnant for more, it is necessary to administer an injection info the foetus to stop the heartbeat – Feticide, before it is removed.

You can find more detailed information about termination of pregnancy at www.nhs,uk

AFTER ENDING YOUR PREGNANCY

Whatever type of termination you have; a follow-up appointment should be made for you at about six weeks. Your medical team will check how you are doing physically and talk through what happened with you.

Tests can be done after a medical or surgical termination confirm the baby's trisomy 13 or 18 genetic condition.

FUNERAL ARRANGEMENTS AFTER A TERMINATION

Parents may choose to name their baby and have a funeral which some see as an acknowledgement of the baby's arrival. Other people choose to have another way of remembrance and celebration of their child. Should you choose not to have a private funeral, the hospital will ensure your baby is laid to rest with dignity.

If you are having a surgical termination and you want to bury or cremate your baby's remains, you should tell them before your procedure.

You may be asked for permission to have a post-mortem, and it is up to you to agree or refuse.

FEELINGS OF LOSS

Sometimes parents feel they are expected to get on with life after a termination and the loss of their baby is not really acknowledged. Those around you, and even you yourself, may expect to feel better when the termination is over, but this may not be the case and it is quite normal to have feelings of anger, guilt, sadness and even depression. Many parents describe a feeling of emptiness. Remember there is no right way to grieve and no time limit for grief. Acknowledge your feelings, find someone you can talk to about how you feel and get support if you need it.

'Immediately after the termination I felt a great sense of relief. But later I suffered feelings of intense grief and my husband felt so angry.' (Parent)

'Even though we agreed it was the right thing for us and for our baby, my wife suffered a deep sense of guilt that we had killed our baby. She says she sometimes wishes she had carried him for as long as nature intended. But she found it so difficult to feel him develop and know he would not make it.' (Father)

'One thing that really helped us was a song called 'I will always love you', which we heard on the radio just after our termination. It just summed up how we felt. (Parent)'

Whether you tell family or friends that you ended your pregnancy is entirely your decision. You may want to talk about everything that happened or only that you have lost your baby. You are the best judge of what you are comfortable with sharing with others, so take the time you need to think about it and tell people when you feel ready.

'I was grateful for people who asked useful questions and acknowledged our baby in what they said or wrote.' (Parent)

Help from the hospital bereavement support should be available and some parents are offered specific counselling. Counselling involves talking to a trained therapist who will listen to you and help you find ways to cope with your loss. Your GP will be able to refer you to a local counselling service.

The loss of a baby in any circumstance is devastating, and anniversaries can reawaken the sadness. It can help to talk to other parents who have been through a similar situation and SOFT UK can put you in touch with other parents and trained support volunteers.

WHEN YOU CONTINUE YOUR PREGNANCY

Your pregnancy is a precious time to get to know your baby and make as many memories as you can. It can feel a scary time, not knowing what the outcome for your baby might be. Contact SOFT UK if you would find it helpful to talk to others in a similar situation.

'We look back on our pregnancy as a special time because she was with us. She was such a special member of our family.' (Parent)

'It was difficult talking to other expectant mums I knew, as their pregnancies were so different from mine. I felt really lonely, actually.' (Parent)

As you prepare to welcome a very special baby you will want to discuss possible problems and outcomes with your paediatrician. Develop a specific birth plan and discuss in detail what you want to happen with the neonatal team who will care for your baby.

'We were told that babies with Patau's syndrome did not live. Then we met with a paediatrician who discussed with us the medical intervention we would want. We agreed to keep the birth as

natural as possible and do nothing to artificially prolong his life. When he was born, 4 weeks early, he needed special care and oxygen but otherwise did not need any intervention. It was so exciting to see him moving and hear his tiny cry. He lived for 4 months, and we are so grateful we gave him the chance of life.' (Parent)

There are some risks to consider when deciding to continue with the pregnancy of a baby with trisomy 13 or 18:

- There is an increased risk of premature labour
- There is a high risk that the baby may be miscarried, stillborn, or only survive for a short time after birth.

BIRTH PLAN

Your birth plan should not just be about the usual things that parents consider, it should also take account of decisions that may need to be made very quickly during labour and after birth. Do you want the baby to have access to full resuscitation if they require it at birth? Do you want the medical team to actively intervene to enhance the baby's chance of survival, or do you want a palliative care (comfort care) approach, keeping your baby comfortable and letting nature take its course?

Discuss the options for how the baby will be born, for example whether a planned Caesarean section should be considered.

'We opted for a caesarean under spinal anaesthetic. She was beautiful. Her lungs were too fragile to be ventilated for long. We were allowed to hold her. She lived for 5 hours.' (Parent)

A copy of a birth plan can be found on the SOFT UK website, which may be helpful to help you design yours and think about what you might want to consider.

'She was a breech vaginal delivery and needed no resuscitation apart from oxygen. She was looked after in Special Care Unit but didn't need any support other than being tube fed. Then we were able to take her home.' (Parent)

WHAT WILL MY BABY LOOK LIKE?

Many newborn babies with Trisomy 13 or 18 look like any other new infant.

It is common for babies to have a light birth weight. Some babies may have slightly lower than normal ears, clenched fists, unusual palm and fingertip patterns, and slower or absent reflexes. *(EUROCAT registries 2000-2011)*

Some babies have other noticeable problems associated with these conditions such as a cleft lip, club foot, defects of the abdomen or eyes. Detailed ultrasound scans may have detected these before birth and allowed parents to be prepared and make provisional plans with the paediatrician about treatment options, if required, before your baby is born.

CARING FOR A BABY NEEDING INTENSIVE CARE

Some babies require the facilities of the Special Care Baby Unit only until breathing and feeding are established and then may be able to go home when hospital care is no longer required.

'It was scary when we brought him home and we have had a few worrying times along the way,

but he has thrived and is such a happy child.' (Parent)

Where there are more major or life-threatening problems your baby may require longer term intensive care. This should not mean you cannot provide care for your baby. Being involved in your baby's care while they are in hospital can really help you and your baby. It might feel as though your baby isn't aware of you being there. But they will recognise your voice and your smell. Hearing your voice and being near to you will comfort them. Holding your baby, skin to skin contact with your baby, washing, feeding, changing nappies are all things that you can do for your baby and help you develop a relationship with them.

'SCBU were marvellous letting me hold her and spend as much time with her as I wanted. I got really confident in caring for her.' (Parent)

You should be able to have regular discussions with the medical team caring for your baby, so that your wishes in relation to their treatment are understood. Getting as much information as possible will give you more confidence that you are doing what is right for your baby. There may be some difficult decisions to make about the treatment your baby might need.

In some cases, there comes a point when there is no longer any further medical treatment that can be offered for your baby. Making the decision to limit or stop your baby's active treatment is devastating and you may not feel emotionally prepared to deal with this. You may disagree with the medical staff or even your partner about it. You may feel that you are not ready to make that decision. Take your time, talk to your baby's doctors, ask questions and discuss all the options, so you are making informed decisions and can decide what you want to do in your baby's best interests.

'The doctors recommended doing no surgery or heroics, he just wasn't strong enough. We agreed with them at that time.' (Parent)

Some parents report that they feel they are not always listened to in terms of what treatment they want for their baby or that medical teams feel they should not offer interventional treatment.

'We felt we had to fight for her all the way through. It was like they only saw this syndrome and not our child.' (Parent)

This can be an incredibly difficult situation to be in. Keep talking to your baby's doctors, ask questions and ensure that you fully understand the reasons for their views. Ask for a second opinion if appropriate.

CARING FOR A BABY WITH SPECIAL NEEDS AT HOME

You may have the option of taking your baby home when hospital care is no longer required. Specialised support should be arranged together with a structured care plan to prepare you for any problems that may arise, and the care plan must be flexible to the changing needs of the baby or your family. Always feel that you can ask for information or advice – as many questions and as often as you need. SOFT UK can provide you with information and support. You might want to think about joining SOFT UK's Families Facebook group – a private group of families who all have experience of trisomy 13 or 18 and are willing to share their experience and things that have worked for them.

Feeding problems are not unusual and can cause slow growth and low weight gain because babies lack the co-ordination to suck and swallow properly. *(Cereda A. & Carey J., 2012).* Mothers who want to breast feed can express their milk. Some babies may need to be tube fed,

if they are not able to suck or swallow sufficiently. You will be taught how to manage this yourself and you can use expressed breast milk if you choose.

'He never cried for his feed. In fact, we had to keep a close watch and wake him when a feed was due. He was successfully tube fed with my expressed breast milk.' (Parent).

Apnoea or pauses in breathing are not uncommon in babies with trisomy 13 or 18. *(Cereda A. & Carey J., 2012).* The use of breathing monitors may be considered, and it can be helpful for parents to learn resuscitation techniques.

Families taking a baby home should be informed about Special Care Baby Unit staff home visits, Community Paediatric nurses, physiotherapy and other sources of help available to them, including how to manage return visits to hospital if required.

Some parents prefer to care for their baby at a hospice. This is usually planned ahead of the birth and parents can be offered the opportunity to visit the hospice before the baby is born. The whole family can be supported at the hospice, with other family members visiting and with support in creating memories together. The hospice can also provide respite care.

CARING FOR A CHILD WITH PARTIAL TRISOMY OR RELATED DISORDER

The outlook for a baby with partial, mosaic or other related disorder is much more variable depending upon the precise nature of the chromosomal defect and how many cells are affected and where they are in the body. A full genetic assessment should be carried out.

'She has partial trisomy 13 and we were given very little hope of her survival in the early days. We only learned the truth when we insisted we saw a genetic consultant. She does have various health problems and her development is slow, but she attends school happily.' (Parent)

'Our son has a deletion on chromosome 13. He has small stature, hearing and visual difficulties and a learning disability. However, he is a happy, lively and well child.' (Parent)

'He has a rare form of partial trisomy 18 and when he was diagnosed his consultant stressed how serious his particular condition was. He is now 10 years old and although he does have a profound disability, he goes to a special centre daily and is a special member of our family.' (Parent)

'Before she was born, we were told to expect the worst. She had a heart problem that required surgery. The surgeon was fantastic and said he would treat her symptoms not her chromosomal disorder. Today she is still small for her age but otherwise completely fine. She attends school, keeps up with her siblings and loves life. Her mosaic trisomy appears to have been isolated to her heart, which has now been successfully treated.' (Parent)

BEREAVEMENT

The loss of a baby is a heart-breaking experience. No two people react in exactly the same way.

The diagnosis that your baby is going to have a genetic disorder can also feel like a bereavement. Parents want their child to be healthy and the knowledge that a baby has a serious life limiting condition is devastating.

There are various stages of grief, but we don't always follow a set pattern and not everybody needs expert counselling. However, everybody does need to share the sorrow of losing their child with someone. At whatever point your baby dies no one can really prepare you for the

great sense of loss that follows.

Some of the feelings you experience may include:

IN SHOCK

The initial shock brings a numbness that could be described as being on 'auto pilot'. You can function but are shielded from the full impact of what has happened.

ANGER AND GUILT

It is not uncommon to feel anger and guilt focused against other people; for example, doctors and other family members, also resentment against those who don't appreciate their own healthy babies. Such bitterness is quite normal but can become destructive if you are unable to share it. Parents turn to each other for support, but it is unlikely two people will experience exactly the same emotions at the same time, and this can put a strain upon a relationship. Finding someone a little more independent to talk to may be helpful.

DEPRESSION

Depression isn't straightforward or well defined. Like the other stages of grief, depression can be difficult and messy. It can feel overwhelming. You may feel exhausted, hopeless, confused, unable to take on the most usual of daily tasks.
Depression may be an inevitable part of any loss. However, if you feel you can't seem to move past this stage, seek expert help from your doctor to help you work through this period of coping.

MEMORIES

Losing your baby during pregnancy or shortly after birth does not have to mean you have no memories. There are many things you can do to make these memories, such as keeping a diary or even an online blog of your pregnancy, or start a scrapbook with the scans, cards or photographs you have.

You may want to plant a special tree or flower for your child, or have their name written beautifully in the snow or sand captured in a photograph forever, or maybe you would like to name a star after your baby or have a remembrance star posted on the SOFT UK website, there are so many alternatives. If your baby died before they were born, you could turn a pregnancy scan photograph into a charm you can wear on a necklace or keyring.

Taking a clipping of your baby's hair that you can keep close can help you feel as if part of them is still with you. Footprints and handprints can be made, or casts taken before or after your baby has died. These can then be made into jewellery or framed, and ashes can become diamonds, a permanent and beautiful reminder of your little one. Memory boxes and chests are widely available, and you can store mementos of your child to keep as precious memories.

It is important to do what you feel is right and keep whatever you want to. There is no 'proper' way to grieve, and everyone is different.

'I don't think there is a day that goes by when I don't think of her and long to hold her; but that all-encompassing pain has lessened over the years and the ache that remains is actually a comfort.' (Parent)

THE OLDER CHILD

Although many babies diagnosed with trisomy 13 or trisomy 18 die during pregnancy or shortly after birth, some children do survive for longer. A recent, large population-based study found that 11.5% of babies with trisomy 13 and 13.4% of babies with trisomy 18 who were born alive, survived to their first birthday. 80% of those who survived to 1 year of age, went on to their fifth birthday. Survival rates at 5 years were 9.7% for children with trisomy 13 and 12.3% for children with trisomy 18. (*Meyer R.E. et al., 2015*). SOFT UK is in contact with an increasing number of families who have an older child with trisomy 13 or 18.

Despite their complex needs, children with full trisomy 13 or 18 can make progress with their development, albeit slowly. Many children are reported to be able to communicate their needs, show awareness of surroundings, sit and stand, sometimes with some support. Older children may attend school.

Feedback from parents suggests a positive quality of life for their child as a valued member of the family.

Because children with the mosaic or partial form of trisomy 13 or 18 have fewer cells carrying the additional chromosome, they may be much less severely affected or even have no observable effect, but that is not always the case. The impact can be extremely variable from one child to another. Unfortunately, it is not possible to predict this variance before the baby is born.

SOFT UK SUPPORT

We know that a diagnosis of trisomy 13 or trisomy 18 can feel isolating. SOFT UK aims to ensure families can access the support they need, when they need it.

The SOFT UK Support Service provides a safe, confidential place for anyone whose pregnancy or child is affected by trisomy 13 or trisomy 18. Whether your experience is recent or happened long ago, we aim to support you for as long as you need us.

You can contact us at:

Email: support@soft.org.uk

Phone helpline: 0300 102 7638

Our website www.soft.org.uk has a wealth of information and family stories. We can help in finding information and answering the many questions you will have.

REFERENCES

Cohain J.S. et al., *Spontaneous first trimester miscarriage rates for women amongst parous women with 1 or more pregnancies of 24 weeks or more.*, BMC Pregnancy and Childbirth. (2017)

Cereda A. & Carey J., *The Trisomy 18 Syndrome*, Orphanet Journal of Rare Diseases., (2012)

Goel N., Morris J.K. et al. *Trisomy 13 and 18 prevalence and mortality: A multi registry population-based analysis* (1974-2014: Total birth prevalence including live births, miscarriage/ stillbirth and elective termination of pregnancy). AM Journal Medical Genetics., (2019)

Meyer R.E. et al., *Survival of children with trisomy 13 and trisomy 18: A multi-state population-based study*., AM Journal of Medical Genetics (2015)

Morris J.K. & Savva G., *The risk of fetal loss following a prenatal diagnosis*., AM Journal Medical Genetics (2008)

National Congenital Anomaly and Rare Disease Registration Service, Congenital anomaly statistics (2017)

Peterson J.K. et al., Long term outcomes of children with trisomy 13 trisomy 18 after congenital heart disease intervention., Ann Thoracic Surgery (2017)