



INTERNATIONAL
TRISOMY 13/18
ALLIANCE

Preparing for the Arrival of Your Baby



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Trisomy 13 and Trisomy 18

**Preparing for the
Arrival of Your
Baby**

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PREPARING FOR THE ARRIVAL OF YOUR BABY

About this book

If you have just received a diagnosis of trisomy 13 or trisomy 18 for your unborn or newborn baby, you are probably in a state of shock. You may feel anger and anguish together with moments of hope. These are natural emotions. This book includes the most recently published research and provides a broad overview of trisomy 13 and trisomy 18, pregnancy, possible outcomes, planning and caring for a baby. Many women are overwhelmed by negative information and grim statistics about lifespan and quality of life. It is important to realize that your baby is not a statistic or a genetic label. Seldom can the life of a baby be predicted with certainty. Each child deserves to be cared for as the unique individual they are.

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Message from Dr Martin McCaffrey, Neonatologist

If your baby has trisomy 13 or trisomy 18 there is a higher risk of loss during pregnancy. Children with trisomy 13 or trisomy 18 can be born with a range of medical problems, some of which can be life threatening, but as you will see here, there is always hope. Sadly many of my doctor colleagues, for whatever reason, are unwilling to offer the hope that can exist for such babies. A significant number of babies with trisomy 13 and trisomy 18 will survive to birth, and can live for varying periods up to years thereafter. That is not all, but a number do. None of us has the crystal ball but you should know that you have been blessed with a challenging and special gift regardless of lifespan.

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DIAGNOSIS OF TRISOMY 13 OR TRISOMY 18

What are trisomy 13 and trisomy 18?

Trisomy means three chromosomes. A baby with trisomy 13 has three number 13 chromosomes in every cell instead of two, and a baby with trisomy 18 has three number 18 chromosomes in every cell. Trisomy 18 and trisomy 13 are the second and third most common trisomy conditions, respectively, after Down syndrome, which is trisomy 21. There are differences in the way trisomy 13 and trisomy 18 affect children both medically and in their appearance, but many medical issues, ability levels and survival rates are similar.



Amy, trisomy 18, Germany



Cati, trisomy 13, Rumania

How prevalent (common) are trisomy 13 and trisomy 18?

In the past decade, various studies from different countries show that trisomy 18 occurs in one in every 1,500-2,000 pregnancies, but only one in approximately 5,000-12,000 live births. The numbers for trisomy 13 are one in every 3,500-5,000 pregnancies, but only one in approximately 7,000-20,000 live births (Cereda & Carey, 2012; Crider et al., 2008; Irving et al., 2011; Rasmussen et al., 2003; Vendola et al., 2010; Wu et al., 2014; Loane et al., 2013). The difference between the number of fetuses (babies before birth) and the number of babies born is due to termination of pregnancies and stillbirths.

What is the survival rate for babies with trisomy 13 or trisomy 18?

Parents may be told that a baby diagnosed with trisomy 13 or trisomy 18 is incompatible with life or that the condition is lethal (Koogler et al., 2003). This is not universally true as some babies can live for months, years, and in a few cases, for decades. Generally the statistics quoted for one-year survival are 5-11%. There are several important things to realize about survival. The studies reporting these low survival rates do not include any information about the care that babies were offered (Burke et al., 2013; Rasmussen et al., 2003). For example, did the babies receive nutrition, and did they receive oxygen for respiratory (breathing) distress or was respiratory support withheld? Length of life is impossible to predict as the same medical issues will not affect all babies in the same way, and not all children will have similar access to medical care, treatments and surgeries (Cereda & Carey, 2012).



Delaine, trisomy 13, Canada

A baby with a prenatal diagnosis (diagnosed before birth) is often not provided with medical interventions at birth or in the immediate post birth period. The withholding of this care likely plays a role in survival rates. Studies indicate that many survivors received interventions at the time of birth such as monitoring and delivery by cesarean section (sometimes called a C-section, a term used when a surgical cut is made through the abdomen and womb to deliver one or more babies), and in the immediate post birth period such as respiratory support (Bruns & Campbell, 2014; Janvier et al. 2012). In addition, recent studies show that one-year survival increases to over 40% with intensive care and cardiac surgery for selected cases (Wilkinson et al., 2014). The rate of death is highest in the first week but declines thereafter. In fact, a baby that is thriving at 6-8 months of age has a higher probability of living for several years (Imataka et al., 2007; Wu et al., 2013).

Do babies with partial and mosaic types of trisomy have a better outcome?

Partial trisomy occurs when only part of the chromosome is extra due to a translocation (a rearrangement of the chromosomes, where parts of one chromosome are reattached to another chromosome). The mosaic form of a trisomy condition occurs when a proportion of cells in the body have three

chromosomes and the remaining cells have two. Children with partial or mosaic trisomy 13 or 18 often have a much larger spectrum of outcomes than those with full trisomy. Information found in articles, books, and on the Internet about full trisomy 13 and full trisomy 18 may be inappropriate if applied to children with a partial or mosaic trisomy.

There have been instances when a baby was initially diagnosed with full trisomy 13 or full trisomy 18 by amniocentesis (a needle inserted through the abdomen to obtain amniotic fluid with fetal cells) or blood tests, but was later re-tested and found to have the mosaic form. If doctors suspect the diagnosis of full trisomy could be wrong, they might order a blood test to analyze a larger number of cells and/or test a tissue sample (usually the skin). Results of these tests may reveal mosaicism (Banka, 2013). In the absence of a significant medical condition such as a serious cardiac defect, it is impossible to know before or at the time of birth how a child with the mosaic or partial form of trisomy 13 or trisomy 18 will develop.



Lily, mosaic trisomy 18, UK



Ivory, mosaic trisomy 13, UK

Why is the information about trisomy 13 and trisomy 18 so negative?

Trisomy 13 and trisomy 18 were originally identified in the early 1960s, and pictures and descriptions in texts were primarily of babies exhibiting complex symptoms of the condition. Unfortunately these cases became associated with the genetic label of trisomy 13 and trisomy 18. Yet, early studies on children born with trisomy 13 or trisomy 18 revealed that children lived longer than at present (Magenis et al., 1968; Weber, 1967). The survival rates for infants with trisomy 13 and trisomy 18 that are often provided to parents reflect survival of large groups of children during a time when withholding care to newborn babies, or withdrawal of care after a postnatal diagnosis was common. These studies only discuss death rates; they do not provide information about medical care that was provided or withheld (Courtwright et al., 2011; Koogler et al., 2003; McGraw & Perlman, 2008; Rasmussen et al., 2003).



Mikayla, trisomy 18, South Africa

As a result, one-year survival rates are typically reported as between 3-10% (Bos et al., 1992; Wu et al., 2013). Further, when the diagnosis of trisomy 13 or trisomy 18 was made during pregnancy, the practice in some hospitals was to question whether to resuscitate (revive) new-born babies in distress (i.e. not breathing, slow/ no heart rate), and not to offer the level of standard or intensive care that would be provided for babies without a genetic diagnosis. The result of these practices is that the incompatible with life label became a self-fulfilling prophecy (McCaffrey, 2011; Morrison et al., 2010). This misled physicians into believing that trisomy 13 and trisomy 18 are always lethal conditions. International guidelines were developed that recommended no newborn resuscitation for babies with trisomy 13 or trisomy 18 due to this mistaken view of a short life and few survivors (Bos et al., 1992; McCaffrey, 2011; McGraw & Perlman, 2008; Morrison et al., 2010).

An article entitled Lethal Language, Lethal Decisions (Koogler et al., 2003), suggested that doctors were imposing their own view of an acceptable quality of life on parents: *Although many of the congenital syndromes that used to be lethal no longer are, they are still routinely referred to as lethal anomalies. But the label is not only inaccurate, it is also dangerous: by portraying as a medical determination what is in fact a judgment about the child's quality of life, it wrests from the parents a decision that only the parents can make* (p 37).

When did research start to reflect the views of parents?

Finally, in 2012, research was published which for the very first time asked parents about their experience of having a child with trisomy 13 or trisomy 18 (Janvier et al., 2012). Even when the baby did not live for long, the time the parents and the siblings enjoyed with baby was considered a positive event for their families. This study also revealed that treatment is beneficial in prolonging life for some children. Around the same time, Japanese studies were published which showed that medical interventions such as intensive care and surgery including heart surgery can prolong life, and increase the one-year survival rate for some babies with trisomy 13 and trisomy 18 to over 40% (Kaneko et al., 2009; Kosho et al., 2006; Maeda et al., 2011).

There is now greater awareness of the range of trisomy 13 and trisomy 18, and more is known about the life and experience of children with these conditions and their families (Bruns & Campbell, 2014; Kosho et al., 2013). Out-dated attitudes and definitions still exist but there is evidence of change: *Healthcare management approaches or policies that reject out of hand the goal of prolonging the life of any infant/child with trisomy 18 are not defensible* (Lorenz & Hardart, 2014).



Ole aged 2 years, trisomy 18, Norway

What is daily life like for children with trisomy 13 and trisomy 18?

After a diagnosis of trisomy 13 or trisomy 18, the majority of parents are told their baby is 'incompatible with life' and worse. Baty and colleagues (1994) were the first to study developmental milestones. Although children with full trisomy 13 or trisomy 18 typically experience significant developmental delays, parents in their study reported that their baby smiled and recognized and interacted with them. While most children will not speak beyond a few words, they do communicate their needs through facial expressions and other signals. Children were also described as showing awareness of their surroundings and exploring the world around them. Some were able to crawl and walk with assisted devices such as a walker (Baty et al., 1994). Parents of seventy-nine older living children with full trisomy 13 or trisomy 18 were asked for their views, and 95% reported that their child could communicate needs to them, while 99% described their child as 'happy' (Janvier et al., 2012).



John Paul, trisomy 13, USA

The TRIS project is reviewing developmental milestone data and will share the results with parents and medical professionals see <http://www.internationaltrisomyalliance.com/tris-project-ongoing-research.html> Preliminary findings indicate children showing preferences, smiling, sitting, and standing independently. Exploration of hand-held items and looking at pictures in books was also noted for many of the children in the analyzed samples. There are challenges to caring for a child with trisomy 13 or trisomy 18, and many parents tell of significant financial sacrifices, but almost all parents reflect on the lives of their children as being positive and enriching (Janvier et al., 2012).



Saskia aged 23 years and Lexi have trisomy 18, UK

PREGNANCY AND BIRTH

What types of screening tests are available?

Screening tests are non-invasive and do not cause a miscarriage. They consist of ultrasound scans and blood tests that indicate if a pregnancy is in a low or high-risk category for certain medical conditions including chromosome defects, but they do not provide a definitive diagnosis. Parents are rarely prepared for a result indicating there may be a possibility that their unborn baby has a chromosome disorder, and an amniocentesis or chorionic villus sampling (CVS—a needle inserted through the abdomen into the placenta and used to remove a sample of tissue, or a thin catheter inserted through the cervix and a sample of chorionic villi cells are gently suctioned into the catheter) currently remains the only way to confirm a chromosome problem, see: <http://webmd.com/baby/chorionic-villus-sampling-cvs>



Annika, trisomy 13, Netherlands

Should I have a diagnostic test to confirm the diagnosis?

Genetic amniocentesis and chorionic villus sampling (CVS) are the only tests that can confirm a trisomy or other chromosomal problems. The diagnostic tests have a slight risk of causing a miscarriage, and may not be advised after previous miscarriages, placental problems or an incompetent cervix. If you decide not to have a diagnostic test, close monitoring will enable your physician to highlight any medical problems identified by ultrasound scanning, and you can discuss different care options. If an ultrasound scan shows a medical problem such as a cleft lip or cardiac defect, your obstetrician can tell you what treatments are available, and the medical team can keep close watch on your baby during the pregnancy.

If you decide to have a diagnostic test, you should be aware that a baby with a diagnosis of trisomy 13 or trisomy 18 may not be cared for in the same manner as a baby without a trisomy diagnosis. You might consider asking your obstetrician if a baby without a diagnosis of trisomy 13 or trisomy 18:

1. Would be monitored more actively during labor and delivery?
2. Would have a greater chance of being delivered by Caesarean section (C-section) if requested for fetal distress?
3. Would be offered treatments if needed immediately after birth?

Why do mothers continue their pregnancy?

After receiving a diagnosis of trisomy 13 or trisomy 18 parents often experience a rollercoaster of mixed emotions with good days and bad days as they learn about the condition. Many parents say they choose to continue their pregnancy because they love their child unconditionally and want to give them a chance of life (Guon et. al., 2013). Other parents believe in destiny and that the child was meant to be. Some religions forbid pregnancy termination.

Parents who continued their pregnancy said they did so with the aim of filling the life of the baby with love for as long as possible. Women described continuing their pregnancy when life was expected to be brief, as a positive experience, in spite of the sorrow and grief (Lathrop & Vandevusse, 2011). Regardless of how long the baby lived, moms also said their pregnancy was a time to love their baby and create wonderful memories for their family. Siblings could feel the kicks when the baby moved, and parents told us they enjoyed these precious moments of being able to care for and interact with their baby, and they tried not to focus on worries for the future.



Lilliana, trisomy 18, USA

When perinatal palliative care (a formal service that was developed to support parents who choose to continue pregnancy after a prenatal diagnosis) was offered as an alternative to termination, 37-87% of women decided to continue their pregnancy (Balaguer et al., 2012). Even the lower figure of 37% is quite high when you consider that large scale statistics show more than 90% of women terminate a pregnancy after a prenatal diagnosis of trisomy 13 or trisomy 18. Sometimes, those who provide this service assume or encourage a limitation in care for the baby when born. It is important to inquire so that you can ensure that the care provided to your baby is consistent with your wishes.

What are the chances of a miscarriage or stillbirth?

The chance that a fetus will miscarry or be stillborn is high in early pregnancy, and decreases as time goes on because babies with the most serious medical needs are more likely to die in utero (in the womb before birth). For trisomy

13, the risk of fetal loss before birth estimated from a recent study was 49% at 12 weeks, 42% at 18 weeks and 35% at 24 weeks gestation. For trisomy 18, the risk of fetal loss before birth was approximately 72% at 12 weeks, 65% at 18 weeks and 59% at 24 weeks gestation (Morris & Savva, 2008). In addition there is a higher risk of fetal loss during labor. One study showed that 17% of fetuses with trisomy 18 died during this time (Yamanaka et al., 2006).

When there is no prenatal diagnosis, approximately 50% of babies with trisomy 18 and 25% of babies with trisomy 13 were delivered by C-section. These rates are higher than average, and may indicate that a baby with trisomy is more likely to experience fetal distress during delivery (Baty et al., 1994; Courtwright et al., 2011). Even when the pregnancy ends in a miscarriage or stillbirth, women report positive memories and value the time given to bond with their baby. This was also true when the baby lived only a few hours or days. Every study has reported that the majority of women who continued their pregnancy describe the experience of their child's life as positive, regardless of length of the baby's life (Janvier et al., 2012; Balaguer et al., 2012; Walker et al., 2008; Wool, 2011).

Should I be induced, request a C-section or aim to have a natural birth?

All birth plans are personal and will include instructions for different scenarios that might unfold during or after delivery. Tell your physician how important it is for you and your family to have time with your baby. Numerous studies have shown that babies carried to full term and with a greater birth weight, tend to live for longer (Bruns & Campbell, 2014; Wu et al., 2013).



Sophya, trisomy 13, Bulgaria

Placental failure is a known cause of stillbirth in babies with trisomy 13 and trisomy 18. It is often identified during ultrasound scans by growth retardation, and can alert physicians to closely monitor and sometimes deliver a baby before term (40 weeks gestation) by C-section or induction. This is a reason why some women might request to be monitored frequently in the final weeks of pregnancy for signs that the baby's placenta is failing or other signs that might indicate it would be safer for baby to be delivered early. It is important you discuss with your neonatologist (a doctor who cares for newborn babies)

whether it might be beneficial to be given steroids to help to develop your baby's lungs before birth. Some doctors agree to a planned C-section if this increases the possibility of a live birth. It is also important to ask whether a C-section is an option if fetal distress occurs during labor, as well as the associated risks of C-section for both present and for subsequent pregnancies. You need to discuss the options thoroughly with your physician.

Most parents like to work with their medical team on a plan that takes account of specific medical issues suspected or identified by scans, as well as their values and choices on necessary interventions. Seek advice on whether to choose a hospital with a higher level of neonatal intensive care (NICU) unit to give birth. It is critical to check that your baby will not be denied the care that is usually given to a newborn baby, such as help to breathe, solely on the basis of a diagnosis of trisomy 13 or trisomy 18.

Also ask about monitors and respiratory support options: bagging (breathing for the patient using a mask and a bag of air often with extra oxygen), blow-by oxygen and nasal prong oxygen (extra oxygen to aid breathing), CPAP (continuous positive airway pressure delivered by a mask over the face of the baby), and intubation (inserting a tube to keep airways safe from aspiration, collapse etc.). You can request a room away from other new mothers if you wish.



Evvy Kristine with CPAP mask, trisomy 18, Norway

What if my baby is stillborn or dies shortly after birth?

Sometimes a baby will not survive birth. The reality of a prenatal diagnosis of trisomy 13 or trisomy 18 is that you need to prepare for the possibility of stillbirth, the death of your child shortly after birth, and a living baby. If you want to be prepared for a stillbirth, you should ask your midwife or obstetrician to describe the procedure of stillborn delivery, and tell you what help and keepsakes they can offer. For example, many parents are glad they held their baby after a stillborn delivery. Some parents appreciate having photos of their stillborn baby, footprints, and a lock of hair if possible. Most families choose to name their baby, and some do not. Some like the hospital clergy to perform a service. If your baby lives only for a short time and you have shared him/her with friends and family, this may help with the grieving process.



Alessandra, trisomy 18, USA

Parents may have the choice whether to take the baby home before a funeral while others ask the funeral home to hold the baby until burial or cremation. Some parents prefer a funeral for their baby and invite family and friends. Others prefer to be alone. There are several options: an individual or shared gravesite, a memorial site for stillborn babies, or deciding where to place the ashes after a cremation. You and your family can choose. There is no right or wrong way to collect your memories, or the type of burial, rituals and ceremonies you select. Grieving takes time. Periods of grief may recur even years after your loss. It can be an emotional rollercoaster, and couples often grieve differently and at different times. Ask what the hospital has to offer, whether they provide seminars for bereaved parents, or if they can help your grieving process in other ways. Most parents appreciate the short time they spent with their baby, whether only in the womb or as a brief life. The memories you make and love for your baby will stay with you.

CARING FOR A NEWBORN BABY

What special care may be needed at the hospital?

You should discuss a care plan for your baby. First ask whether the diagnosis is full trisomy 13 or full trisomy 18, or a related disorder. If the diagnosis is mosaic or partial trisomy 13 or 18, ask to speak to a geneticist for more information. You may wish to speak with another specialist about specific medical problems your baby might have. If you do not agree with their views, consider asking to speak to another doctor who is more sympathetic to your values, hopes and wishes. Treatments and intensive care should be an option to give a baby time. Do not accept a refusal to treat based on the trisomy label, and beware of recommendations to 'only keep baby comfortable'. They may be based on the presumption that life is not worth living when a baby has trisomy 13 or trisomy 18. As parents it should be up to you to discuss with your doctors the level of care you want for your baby.



Bristol, trisomy 18, USA

What types of breathing problems and feeding difficulties can be expected?

Babies with trisomy 13 and trisomy 18 are often unable to suck effectively and tire quickly making them difficult to breast or bottle-feed. They may need to be fed through a nasogastric tube (n-g tube, a tube inserted through the nose), and gavage is also an option (tube from the mouth to the stomach). Oral feeding (bottle/ breast/ syringe) can be tried even if there is a need to supplement with an n-g tube for example, unless there is a concern about aspiration (food, vomit or other substances breathed into the airway). Many children with trisomy 18 also experience reflux (gastro-esophageal reflux, or reflux, is when fluid in the stomach flows back up the food pipe) causing an acidic burning feeling that can complicate oral feeding (Bruns & Springer, 2013). Some older babies are able to eat solid or mashed food, but may need additional nutrition via feeding through an n-g tube or gastrostomy (g-tube, a tube inserted directly into the stomach). Parents can learn how to give n-g and g-tube feedings and the supplements that may be prescribed to help a baby to gain weight.



Jonas tube feeding, partial trisomy 18, Norway

Apnea is the medical term for a pause in breathing lasting more than 20 seconds. About 50% of babies with trisomy 13 or trisomy 18 will experience apnea, especially during early infancy.

Central apnea (when the brain does not signal the baby to breathe) might be overcome by delivering babies at full term if prenatal monitoring shows no need to be delivered early. It may be helpful to start caffeine therapy after birth. This is the medication that is given to some premature babies with apnea and you should discuss caffeine therapy with your neonatologist. Central apnea is frightening for parents as breathing stops suddenly and a baby may become limp and blue. Breathing usually restarts after a few moments when a baby is given gentle stimulation such as stroking their face or moving them slightly. These actions can encourage a quick recovery unless the child is otherwise ill.



Anaya, trisomy 18, USA

Many parents have discovered that their child has obstructive apnea not central apnea. This means there is something obstructing the airway such as tonsils and adenoids, or a floppy trachea that makes it difficult to breathe. Obstructive apnea due to floppy or under developed upper airway may be treated with surgery, CPAP, nasal trumpet, or by a tracheostomy (a tube in the neck for the air to pass through to bypass airway obstruction).

Before a baby leaves the hospital, discuss with the medical team if you wish to monitor the breathing of your baby with a pulse oximeter (a non invasive way to monitor the levels of oxygen in the blood, by a sensor placed on a thin part of the body, often the foot) or apnea monitor as monitoring your baby

might be a wise precaution. You can request information about resuscitation techniques if your child does not breathe after stimulation. Some parents might complete a CPR course before leaving the hospital.

Is surgery an option for my baby?

Your baby should not be denied resuscitation, medication, surgery or treatment solely because he or she has trisomy 13 or trisomy 18. There will be several ways to confirm prenatally suspected medical issues after the birth including blood tests, echocardiograms (a sonogram/ultrasound of the heart), abdominal (stomach) scans, and other tests according to the needs of the baby. If medical issues are confirmed, the parents and doctors will need to discuss effective treatments that are available.



Ivory, mosaic trisomy 13, before and after surgery for a cleft, UK

After discussing the results with a supportive paediatrician and specialists as needed, parents can make informed decisions based on accurate information. A neonatal specialist will tell you the types of surgeries or treatments that are available for conditions such as omphalocele, a diaphragmatic hernia, or esophageal atresia (see <http://www.internationaltrisomyalliance.com/ita-booklets-for-families> for more information on these medical conditions). There is research that describes babies surviving cardiac surgery (Graham et al., 2004; Maeda et al., 2011; Muneuchi et al., 2010).

Surgeries for children with trisomy 13 or trisomy 18 may not be offered in all hospitals, and this can vary by country, state and region. Yet, it is important to stress to doctors that when children are considered to be suitable candidates for surgery, most survive the procedures and thrive as a result of the surgery (Bruns & Campbell, 2014; Cereda & Carey, 2012; Nelson et al., 2012).

Babies with trisomy 13 or trisomy 18 do tolerate anaesthesia, but their airways are often narrow and/or floppy which can be problematic. Surgery should be performed at a hospital with experienced pediatric anesthesiologists and pediatric surgeons, and preferably with staff who have experience working with children with genetic syndromes. Each baby is unique and surgery may not be the right course of action for every baby. Some babies with cardiac defects can be supported to live longer with heart medications and oxygen in cases where surgery is not an option. Physicians should describe all options

including benefits and risks to your baby. It is best for everyone involved to keep an open mind about the alternatives.



Felicia 6 months after heart surgery, trisomy 18, Norway

How should I plan for discharge?

It is important to look ahead to services that may be needed if you are able to take your baby home. Home support services can vary widely between and within countries. It is best to check with your insurance provider, hospital or medical system about the types and amount of support that would be available. Generally, the amount of nursing care offered depends on the medical complexity of the baby. A discharge plan should concentrate on support for mother and for baby and include practical help, medical advice and access to emergency care should it become necessary.

Hospital support should be available 24/7. Ask for details of a direct telephone number with named members of staff, and agree the details you need to give in the event of an emergency. Ensure the information is updated regularly. There is nothing worse than making an urgent telephone call during a weekend or at night, and having to repeat the medical history of your baby to several different members of staff who appear to know nothing about trisomy 13 or trisomy 18. Always check you have sufficient supplies of essential equipment and medications before leaving the supportive environment of the hospital.

Medical support at home can be a lifesaver. There should be home visits during which a doctor or nurse can check the progress of your baby, answer any questions you might have, and advise on problems that may have arisen. Different countries have widely differing support systems. In some countries night nurses, financial help for the family, respite care or equipment may be available for a baby with special needs, and specialist nurses should be able to give you the details.

Practical support should be in place before discharge. Caring for a new baby is tiring and time consuming, and more so when that baby has trisomy 13 or trisomy 18. Small feeds may be required more frequently, feeding your baby can take longer, and equipment such as n-g tubes may need extra

preparation. You might consider asking people you can trust to occasionally help with baby especially when you have other children at home, and show them how you like things to be done. There may also be the option of out-of-home care to give the family a break. Parents are reluctant to talk about respite care but we recommend investigating what is on offer well before this type of help may be wanted. Remember, even the most devoted of parents need time to recharge batteries, relax and sleep.



Vera, trisomy 18, Singapore

In conclusion

We hope that this booklet has provided you with some of the background to support your discussions with your physicians and help you to make informed decisions about your baby. Remember that every baby is unique but equally special. Plans and experiences are personal. The challenges of preparing for your baby can cause families to forget to enjoy time with baby, even before birth. Make memories, take pictures, and create a diary or a blog. This journey is life altering and one that few regret.

RESOURCES

Resources produced by ITA

The following books produced by ITA and details of research can be read and downloaded free of charge from the ITA website:

Trisomy 13 and Trisomy 18 for Younger Children

Trisomy 13 and Trisomy 18 for Older Children

A Family Dictionary of Medical Terms

A Cherished Pregnancy

Preparing for the Arrival of Your Baby

Newsletters



International Trisomy Alliance (ITA) has produced the two e-books for younger and for older children to explain trisomy 13 and trisomy 18 in an age appropriate way. Parents can decide which book to read with their child. The ITA book for younger children has topics that include A Special Baby and Memories. The ITA book for older children covers trisomy 13 and trisomy 18 in more depth with an explanation of chromosome types. There are translations of the books for children into Italian, Spanish, Norwegian, Indonesian, Turkish and Finnish on the ITA website.

Resource books produced by other organizations

Care of the Infant & Child with Trisomy 18 or Trisomy 13 SOFT USA

Special Delivery - A grief support book for kids

CONTACTS

Parent groups and organizations

Trisomy 13 and trisomy 18 groups and organizations can provide valuable support and information. Connecting with other families who have a similar experience can give you the confidence to advocate for your child.

International Trisomy 13/18 Alliance has details of groups and organizations in the following countries: America Australia France Germany Ireland Italy Japan Norway Romania UK.

See <http://www.internationaltrisomyalliance.com/trisomy-support-groups.html>

Facebook

Families often use social networks like Facebook to ask questions and share with other parents. Many SOFT and other trisomy 13 and trisomy 18 groups also have links to Facebook pages.

Hope for Trisomy

LEONA e V

Leve med trisomi 13 og trisomi 18 kontakt sirifberg@gmail.com

Prenatal Partners for Life

Sindrome-de-Edwards-Trisomia-18

Sindrome Patau Trissomia

SOFT Australia Support for Trisomy 13/18

SOFT Ireland

SOFT Italia

SOFT UK

SOFT USA

SOFT USA Groups

Trisomia 13 habla hispana

Trisomy 18 Mommies

Trisomy Families

Trisomy 18/13 Support UK

Trisomia 18/13 (apoyo para la comunidad hispana) los estados unidos

Trisomy13 - Patau Syndrome - Living with Trisomy 13 Community

Trisomy Angel Parents

TRIS Research Project

Valentin- Association des Porteurs d'Anomalies Chromosomiques

UNIQUE Chromosome Disorders

PLEASE NOTE International Trisomy Alliance has no control over the nature, content or availability of the Facebook pages listed above. Groups are included if they provide support and information for families affected by trisomy 13 and trisomy 18. The inclusion of any links does not necessarily imply a recommendation or endorsement of the views expressed within them, and the views or opinions expressed by individuals do not necessarily reflect those of ITA.

GLOSSARY

Abdominal

Stomach

Apnea

Central apnea is when the brain does not signal the baby to breathe, Obstructive apnea is when something obstructing the airway of the baby such as tonsils and adenoids, or a floppy trachea that makes it difficult to breathe

Amniocentesis

A needle inserted through the abdomen to obtain amniotic fluid with fetal cells

Anomaly

Not standard or expected

Aspiration

A condition in which food, vomit or other substances is breathed into the airway

Bagging

Breathing for the patient using a mask and a bag of air often with extra oxygen

Blow by oxygen, nasal prong oxygen

Extra oxygen to aid breathing

Caesarean section (C-section)

Surgical cut is made through the abdomen and womb to deliver one or more babies

Chorionic villus sampling (CVS)

A needle inserted through the abdomen into the placenta and used to remove a sample of tissue, or a thin catheter inserted through the cervix and a sample of chorionic villi cells are gently suctioned into the catheter

Congenital

Present from birth

CPAP

Continuous positive airway pressure delivered by a mask over baby's face

Echocardiogram

A sonogram/ultrasound of the heart

Fetus

Baby before birth

Gastro-esophageal reflux (reflux)

When fluid in the stomach flows back up the food pipe

Gavage

Tube from the mouth to the stomach

Intubation

Inserting a tube to keep airways safe from aspiration, collapse etc.

In utero

In the womb before birth

Nasogastric tube

N-g tube – tube inserted through the nose

Neonatologist

A doctor who cares for new born babies

Perinatal palliative care

A formal service that was developed to support parents who choose to continue pregnancy after a prenatal diagnosis

Postnatal

After a birth

Prenatal diagnosis

Diagnosed before birth

Pulse oximeter

A non-invasive way to monitor the levels of oxygen in a baby's blood, by a sensor placed on a thin part of the baby's body, often the foot

Respiratory

Refers to breathing

Resuscitate

Revive

Tracheostomy

A tube in the neck for the air to pass through to bypass airway obstruction

Translocation

A rearrangement of the chromosomes where parts of one chromosome are reattached to another chromosome.

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