

International



Trisomy Alliance

A Cherished Pregnancy

For parents who continue a pregnancy



www.internationaltrisomyalliance.com

International Trisomy Alliance A Cherished Pregnancy

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Dear Parent,

By the time you have been given a prenatal diagnosis of trisomy 13 or trisomy 18, your unborn baby is a tiny miracle who has overcome tremendous odds.

Your life will become a rollercoaster of mixed emotions, good days and bad days, but you and your family can love and enjoy this special baby during your pregnancy, and create some wonderful memories.

Whatever happens in the future, your pregnancy can be a positive experience that you and your family will always treasure.

Siri Fuglem Berg MD Ph.D

and

Jenny Robbins



Front cover, Siri and husband Odd with Evy Kristine who had trisomy 18 and lived for 3 days



Jenny and her family with Beth who had trisomy 13 and lived for 3 months

A Cherished Pregnancy is for parents who continue a pregnancy and includes medical terms, but International Trisomy Alliance does not recommend or advise any particular course of action or medication if a medical problem arises, and families should always consult their doctor before changing or introducing treatments.

Trisomy 13 and 18 Patau and Edwards syndrome

A baby with trisomy 18 has three number 18 chromosomes in every cell instead of the usual pair, and a baby with trisomy 13 has three number 13 chromosomes in every cell.

Many women conceive a baby with a chromosome defect but it is estimated most of these pregnancies end naturally before a woman knows she is pregnant. No one knows why some babies continue to develop and live.



Annie trisomy 13

Mosaic trisomy and translocation

Mosaic trisomy is when only a proportion of cells in the body have three chromosomes and the remaining cells have the usual pair. The outlook for mosaicism can be a very wide spectrum ranging from near normal to that of a full trisomy. It has been known for blood tests to show full trisomy, and other tests to identify mosaic cells. An incomplete diagnosis of full trisomy may affect the treatments that are offered.

An extra piece of chromosome attached to one of the chromosomes is called a translocation. Most of the clinical data refers to full trisomy 13 and 18, and may be inappropriate when applied to children with a mosaic trisomy or translocation. Doctors do not always know how a child diagnosed with a mosaic trisomy or a translocation will be affected until they grow older.



Bryn trisomy 18



Mia mosaic 18

Screening for risk

Scans and blood tests are screening tests to indicate if a pregnancy is in a low or high-risk category but do not provide a definitive diagnosis. Parents are rarely prepared for a result indicating something may be wrong with their unborn baby. An amniocentesis or placental biopsy remains the only way at present to confirm a chromosome problem.

Ultrasound scans

Nuchal translucency scans at around 11-13 weeks measure the fluid area at the back of the baby's neck, and check for the presence of nasal bone, estimate the due date, assess the baby's growth and development, and detect some neural tube defects.

Ultrasound scans at around 18-20 weeks show flat images of the internal organs, the position of the placenta and the growth of the unborn baby.

3D scans show three-dimensional external images. For example, when a cleft lip is detected, the parents are prepared before the birth and can discuss what treatments may be available when the infant grows.

4D scans produce a moving image.

Marina Alyssa at 25 weeks...so beautiful!!! It was this ultrasound where we first saw her completely open her left hand...the following week she completely opened her right hand. It was also this ultrasound where we saw a bit of her personality. We caught her lying with legs extended, ankles crossed, feet propped on the placenta like it was a footstool, and one hand and arm behind her head, like she was resting on them. We had to giggle...it was so precious!!! Tamara



Angelo Jack smiling (top)
and playing peekaboo



Marina Alyssa

Diagnostic tests

Genetic amniocentesis and chorionic villus sampling (CVS) are diagnostic tests that have a very slight risk of miscarriage, and they may not be advised after previous miscarriages, placental problems or an incompetent cervix.

Before you decide to have a diagnostic test that will confirm whether your unborn baby has trisomy 13 or 18, ask your doctors what the consequences will be and why it is in the interests of your baby to do so.

Will there be increased pressure to end your pregnancy?

Will your medical team be more likely to discriminate and withhold monitoring during labor?

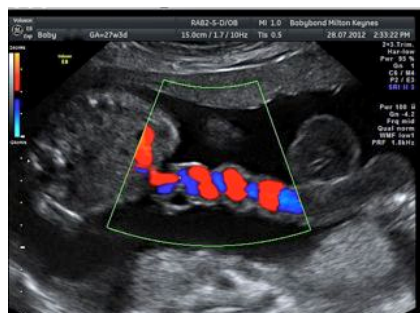
Will your obstetrician fail to consider a caesarean for fetal distress if requested?

Will your baby be refused life saving treatments immediately after birth?

In the absence of a diagnostic test, close monitoring by ultrasound scanning will allow you to discuss treatment levels with neonatal doctors should your baby survive, and doctors should treat your baby accordingly.



Heart beat 27 weeks trisomy 18



Blood flow 27 weeks trisomy 18

How you are given the diagnosis

If you have a diagnostic test and the results show your baby has trisomy 13 or 18, how you are given the diagnosis will affect both the decisions you make and your emotional wellbeing. Doctors may have little experience of your diagnosis, and what they know may be based on old and over simplified information. This will be reflected in what they say to parents as the examples below indicate.



'Babies with trisomy 13 or trisomy 18 often have a shorter life expectancy, but some babies thrive beyond any medical professional's expectation. They interact, they smile and recognize the people around them, and they respond differently to their mother and closest family.'



'Trisomy 13 and 18 are lethal and incompatible with life. Babies who survive birth suffer and have no quality of life. They are like vegetables and will never communicate, interact or respond to other people.'



'We had good days and bad days, and we enjoyed every scan, kick and hiccup.'

*Stacey, mom of
Angelo Jack
trisomy 18
27th October 2012*

Questions you need to ask:

Does my child have full trisomy, mosaic or a translocation, and what does this mean?

What major medical problems does the ultrasound confirm?

Can I discuss these specific problems with neonatal specialists?

If you feel your doctors do not respect your decision or value your baby, you might decide to choose another doctor.

How long will your baby live?

Parents may be told that a baby diagnosed with trisomy 13 or trisomy 18 is incompatible with life or will have no quality of life. This is not true as some babies can live for months, years, even decades in a few cases. Length of life is impossible to predict as the same medical issues and will not affect all babies in the same way, and not all children will have similar access to medical care, treatments and surgeries.

When children receive medical care, it is beneficial for some and they can live longer. These factors, together with the number of babies that might survive if pregnancies were not terminated, prevent accurate statistics for the life expectancy of children with trisomy 13 or 18.

Studies with babies and older children who have had heart surgery have shown survival rates over a year for 10-40%. Recent studies show more than 40% will survive over a year when they receive intensive medical care and surgeries. A child will experience some developmental delays, but they can communicate, show preferences, and reach developmental milestones such as smiling, sitting, rolling, standing and walking.



Annie trisomy 18 14 years old



Natalia trisomy 13, birth to 13 years



Angelo Jack trisomy 18

Your pregnancy

After the initial shock of the diagnosis of trisomy 13 or 18, your pregnancy can be a precious time for the family to get to know your baby. When your baby moves, your children can put their hands on your tummy and the times they share with the baby will be wonderful memories of their sibling. Enjoy your baby during your pregnancy, and don't let your worries for the future make you miss out on precious moments.

Make happy memories and take pictures. Be proud of your little fighter. Relax, talk and play music to your baby, and respond by massaging your tummy gently when baby kicks.



Evy Kristine's siblings feeling
Evy kicking in mom's tummy.

Placental failure is common and a cause of stillbirth, so women should be closely monitored towards the end of the pregnancy.

Ask to talk to pediatricians with experience of your child's chromosomal condition, and discuss possible active and palliative care after the baby has been born. They can give advice on where to give birth, how the birth should take place and more.

If you do not agree with their views on treatment, you might wish to choose another doctor who is more sympathetic to your values, hopes and wishes.

Telling other people

It can be upsetting to tell people your unborn baby has a trisomy condition. It can take away some of the stress to let a few trusted people explain the situation to others. You can print out cards for friends with a short message, for example, 'We are pleased to tell you we are expecting a much loved baby (name) but sad to share the news that (name) has a serious condition (trisomy 13 or 18), and his/her future is uncertain.

Your birth plan

Studies have shown that babies carried full term and with a greater birth weight, tend to live longer. The baby can be monitored frequently toward the end of the pregnancy to reduce the risk of placental failure leading to stillbirth, and to ensure the well being of the baby.



Aimee trisomy 13
lived 20 minutes

Questions you may wish to ask your doctor:

Are you aware that some children with trisomy 13 and 18 do survive, and there are children and adults living with the condition today?

Some doctors agree to a planned C-section if this increases the possibility of a live birth. If fetal distress occurs in labor, is a C-section an option, and what is the increased risk of surgery and for later pregnancies?

How frequently will I be monitored for placental failure? Do you want to help my family to achieve some precious time with our baby?

Your child should not be denied resuscitation purely because they have one of these syndromes. Ask about monitors and respiratory support options: bagging, blow by oxygen, nasal prong oxygen, intubation and cpap, a mask on the nose blowing air to help keep the airways open (continuous positive airway pressure).

After delivery, can we have a room away from new mothers?

Miscarriage, stillbirth and if baby lives a short time

Continuing your pregnancy is about filling your baby's life with love for as long as possible. Having a miscarriage or stillbirth does not mean you have no happy memories, and this is also true when the baby lives only for a short time.

Practical steps to collect memories may include making a photo album, and starting a blog or a diary. You do not have to start right away, but you might like to collect photographs of your tummy and ultrasound scans. You can connect to parents who have had a similar experience through the organizations listed on the ITA website.

I wanted to take a moment and say to the mommy's still carrying their angels that my worst fear was losing Alessandra before I could say hello to her. And when it was confirmed I cried hard. But I stopped and then dealt with labour, and as soon as I had her in my arms, I thought I'd be sad knowing she wasn't 'here'.

It didn't matter at all. It was still the happiest moment of my life. I cuddled and held her, kissed her and told her I loved her for hours. Everyone close to me got a chance to do the same. It made my entire journey worthwhile. Yes there were tears. Though for me the smile didn't leave my face until the time came I had to hand her over to the nurse. That was the single hardest moment of my life, kissing my angel and handing her over knowing I wouldn't see her on this side of heaven again.

But I have zero regrets. I loved every minute of carrying her and welcoming her into the world. So don't live in fear of it, just live each day with the goal of making sure your baby knows nothing but your love and you won't have any regrets either. I hope this helps someone out there. Erika



*Evvy with her sisters
In our pregnancy we felt the need to prepare for all outcomes. We had time to prepare for the possibility of a funeral and burial, and had several conversations with our faith leader on that matter. Siri –Co author*



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When a baby lives

Before a baby leaves hospital parents should discuss the optimal care plan with their doctor. They might wish to monitor breathing with a pulse oximeter or apnea monitor. About 50% of babies will have apnea, a pause in breathing of 20 seconds or longer, and this can usually be treated, unless the child is otherwise ill with pneumonia or heart failure.

Ask your doctor about resuscitation techniques you can use if your child does not breathe after stimulation. Conversely, some parents may decide on non-intervention.

Babies, whether or not they have a cleft lip and palate, will normally need to be tube fed as they lack the energy and coordination to suck effectively, and you can learn how to provide tube feedings. Supplements may be prescribed to assist your baby to gain weight.

Babies are quite vulnerable especially in their first year of life, and it is a good idea to keep them away from sick children and people who have coughs, colds and other infections.



Saskia T18 -PEG feed



Mille 7 years trisomy 13



John trisomy 13



Vera trisomy 18

Medical problems

The list of potential problems associated with trisomy 13 and 18 is overwhelming, but your baby will likely experience only some of them. The more common conditions such as flexed fingers, rocker bottom feet and vision issues are not life limiting.

Heart, brain and other anomalies may range from mild to severe, and you and your doctor will have discussed any problems identified by ultrasound scanning and the different care options.

Your obstetrician will tell you if treatments are available for conditions such as spina bifida, omphalocele, or a diaphragmatic hernia, and the medical team can monitor your baby closely.

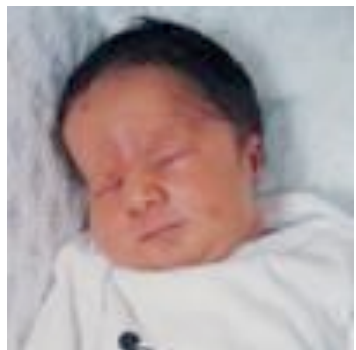
Developmental delay and medical problems such as scoliosis are not normally an issue in young babies.



Lewis (left) has microcephaly and mosaicism



Antony had mosaic trisomy 13 and omphalocele



Beth had trisomy 13 and holoprosencephaly

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Children with trisomy



Saskia and brother Scott



Emeli trisomy 18



Lily - front with siblings and mom Sue



Parents with a baby or a surviving child report that these children lead valued and fulfilled lives regardless of how long or short they live, and they enrich the lives of their siblings.



Amy Kate trisomy 18

Small feet leave large traces.
Even though you lived for three days and I wish I could have kept you longer.
The days I got to meet you Evy, were the best days of my life.
Einar

Brother of Evy Kristine



Jonas mosaic 18



Mo with Noah



In our pregnancy we felt the need to prepare for all outcomes. We had time to prepare for the possibility of a funeral and burial, and had several conversations with our faith leader on that matter.

We also had time to make plans in case our daughter's life was short - plans to ensure our children were present immediately after birth, plans to take pictures and collect memories. The three days we had with our daughter were wonderful, albeit tough.

Having a seriously ill baby is difficult, but we were as prepared as we could be. You can never really prepare for the death of your baby, but you can plan for the little time you have, to make it as good as possible, to build memories.

Siri – Co author

Bereavement

The loss of a child is life changing. There is no right way or length of time to mourn, and each person grieves and copes in a different way.

Grief remains with you, but with time it becomes less painful, and sharing happy memories during a funeral or Service of Remembrance is a valuable part of this process. A Service can also be held after a miscarriage or a stillbirth.

Opportunities to grieve can also give siblings insight in how to cope with change and loss throughout their lives.

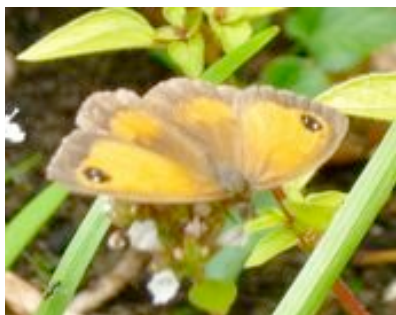


A service for Annie



Saying goodbye to Angelo

Remembrance



Some websites have a memorial gallery where you can post a photograph and details, or the name of your baby with a butterfly or a star if there are no photographs. A number of companies make jewelry with hand/footprints and the name of the baby.

Here are a few ways families have remembered their baby or child.

- Remembrance jewelry
- Framed foot or handprints
- Writing a blog or a book
- Making a website or Facebook page
- Releasing balloons or lighting candles
- Raising money for charity
- Designing a tattoo
- Keeping a memory box



Other ways to remember a child can include naming a star, planting flowers or a tree in a special place, or placing a memorial bench somewhere peaceful where you like to walk and sit.

Founders of ITA

Jenny Robbins

Her daughter Beth had trisomy 13 with holoprosencephaly and lived for three months. Jenny co founded and was a trustee of SOFT UK for over twenty years, and she wrote several publications for families affected by trisomy 13 or trisomy 18.

Siri Fuglem Berg MD Ph.D.

Siri is an anesthesiologist who was given a prenatal diagnosis of trisomy 18, and Evy Kristine lived for three days. Siri established a Norwegian website to support families in similar situations:

www.trisomi18.com

Barb Farlow MBA

Barb's daughter Annie had trisomy 13 and lived for 80 days. Barb's driving goal has been to improve communication with parents and professionals about the experience of having a child with trisomy.

Debbie Bruns Ph.D.

Debbie is the Principal Investigator of the Tracking Rare Incidence Syndromes (TRIS) project. She worked with three preschool aged children with trisomy 18 when she was a classroom teacher.

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