

TRISOMY 13 AND 18



WHAT YOU
NEED TO KNOW



YOUR **BABY** AND **YOU**

If you have just received a diagnosis of Trisomy 13 or Trisomy 18 for your baby, it is important that you understand the diagnosis and the possible outcomes it can bring.

Recent research has led to an improved understanding of these conditions, and a better awareness of the experiences of families in this situation.

WHAT IS **TRISOMY 13** OR **TRISOMY 18**?

These are rare conditions which arise when the baby is developing in the womb. **Trisomy** means three chromosomes.

A baby with Trisomy 18 has three copies of the number 18 chromosome in every cell instead of the usual pair. With Trisomy 13, the 13th chromosome is affected in the same way. Trisomy 18 and Trisomy 13 are less common than Down Syndrome, which is Trisomy 21.

There are differences in the way Trisomy 13 and Trisomy 18 affect children, but many medical issues are similar for both trisomies.

WHAT IS **PARTIAL** OR **MOSAIC TRISOMY**?

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.

International Trisomy Alliance

WHAT DOES THIS MEAN FOR MY BABY?

Children with Trisomy 13 or Trisomy 18 can have a range of medical problems, which can be severely life-limiting.

Survival is difficult to predict as every baby born with Trisomy 13 or 18 is unique. However, survival rates are increasing, almost certainly because of decisions to provide more treatment.¹



Children who live until birth with Trisomy 18 may have an average lifespan of 14 days; with 39% of babies living for more than a month, and 10% living for more than a year.^{2,3}



Research has also found that children with Trisomy 13 may have an average lifespan of 12 days after birth, with almost 31% living for more than a month, and up to 13% living for more than a year.^{2,3}

The risk of miscarriage is significant with both Trisomy 13 and 18 – the chance that a baby will miscarry or be stillborn is high in early pregnancy, and decreases as time goes on.⁴

Good care should ensure that your needs are met, that you know your baby's life is valued, and that you are assisted in making memories and making the most of the time you have with your child.



¹. Lantos J, (2016) Trisomy 13 and 18 – Treatment Decisions in a Stable Gray Zone, Journal of the American Medical Association (JAMA), July 26, Volume 316, Number 4 397

². Rasmussen SA, Wong LY, Yang Q, May KM & Friedman JM (2003). Population-based analyses of mortality in trisomy 13 and trisomy 18. *Pediatrics* 111(4 Part 1):777-84.

³. Nelson KE, Rosella LC, Mahant S, Guttman A. Survival And Surgical Interventions For Children With Trisomy 13 And 18, Journal of the American Medical Association. doi:10.1001/jama.2016.9819.

⁴. Morris JK, & Savva GM (2010), The Risk Of Fetal Loss Following A Prenatal Diagnosis Of Trisomy 13 Or Trisomy 18, American Journal of Medical Genetics Part A. 146A(7): 827-832.



"Muireann was born, two fists in fighting position... ready to take on the world. And fight she did, for six whole weeks. She was an amazing little girl who made a huge impact on us all."

Mandy Dunne on her daughter Muireann who lived for 6 weeks with Trisomy 13.

"I look back knowing I did everything I could possibly do to make sure she got here and knew that she was so loved."

Grace Sharp on baby Lilly Joy who lived for two hours after birth with Trisomy 18.



"She brought nothing but joy, nothing but love."

Derbhillie McGill on her baby girl Clodagh who lived for 33 days after birth with Trisomy 13.

Seán Hynes lived for two days after birth with Trisomy 18. His mother, Sarah, says:

"So we wrapped him in love for those two days, two wonderful days we will never forget. Seán has made us the proudest parents in the world. He has made us better people, and for that we are forever grateful."





"She had a life inside me for 32 weeks. And I got to hold an angel in my arms".

Gemma Bradley on her daughter Lily Rose, who was stillborn with Trisomy 18.

Elaine Fagan, given just days to live after birth with Trisomy 18, spent 25 years with her loving family before her death in 2011, confounding all medical expectations.



"She is at the very heart of our family and has made us better parents and better people."

Tracy Harkin on her daughter Kathleen Rose, living for 10 years with Trisomy 13.



"We were so proud of our beautiful little girl. We will never forget her first smile".

Barbara Farlow on her little girl Annie, who lived for 80 days with Trisomy 13.



WHAT ARE THE EXPERIENCES OF OTHER FAMILIES?

Research has found that almost all parents interviewed felt that their baby enriched their lives and the lives of their families, and that almost all families of children with Trisomy 13 and 18 reported that their time together was happy and rewarding.¹

Where the baby continued to live, 97% of parents described their child as a happy child, despite their severe disability.¹

Parents said that their aim was to fill 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 if the baby passed away.²

IS MY BABY IN ANY PAIN?

There is no evidence that your baby's condition causes them any pain or discomfort while in the womb. It is a very safe environment.

Any discomfort your baby has after birth can be best managed by palliative care.

1. Janvier A, Farlow B, & Wilfond BS. The experience of families with children with trisomy 13 and 18 in social networks. *Pediatrics*. 2012; 130(2):293-298

2. Lathrop, A., & VandeVusse, L. (2011). Affirming Motherhood: Validation And Invalidation In Women's Perinatal Hospice Narratives. *Birth*, 38(3), 256-265

BEST CARE AROUND BIRTH

The **International Trisomy Alliance** advises:

- ♥ Your birth plan can focus on what is best for and your baby. Numerous studies have shown that babies carried to full term and with a greater birth weight tend to live for longer.^{1,2,3}
- ♥ However, because of the risk of placental failure, careful monitoring with ultrasound scans might indicate a requirement to deliver baby before term. It is important you discuss with your neonatologist 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 baby is distressed during labour, as well as the associated risks of C-section for both present and any subsequent pregnancies.
- ♥ 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.

After birth, your baby should be treated as a unique person, and his or her needs objectively assessed. Japanese studies show that medical interventions such as intensive care and surgery can prolong life, and increase the one-year survival rate for some babies with Trisomy 13 and Trisomy 18 to over 40%.^{4,5,6}

See more at www.internationaltrisomyalliance.com

1. Bruns, D. A. & Campbell, E. (2014). Twenty-two long-term survivors with full trisomy 18: Presenting and current medical conditions. *American Journal of Medical Genetics Part A*, 164A(3), 610-619.
2. Wu J, Springett A, & Morris JK. (2013). Survival of trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome) in England and Wales: 2004-2011. *American Journal of Medical Genetics Part A*. 161A(10): 2512-2518.
3. Janvier A, Farlow B, Barrington KJ. 2016. Parental hopes, interventions, and survival of neonates with trisomy 13 and trisomy 18. *Am J Med Genet Part C Semin Med Genet* 9999C:1-9.
4. Nelson KE, Rosella LC, Mahant S, Guttman A., Survival and surgical interventions for children with trisomy 13 and 18, *Journal of the American Medical Association (JAMA)*, doi:10.1001/jama.2016.9819.
5. Kaneko Y, Kobayashi J, Achiwa I, Yoda H, Tsuchiya K, Nakajima Y, Endo D, Sato H, & Kawakami T. (2009). Cardiac surgery in patients with trisomy 18. *Pediatric Cardiology*. 30(6):729-734.
6. Kosho, T., Nakamura, T., Kawame, H., Baba, A., Tamura, M., & Fukushima, Y. (2006). Neonatal management of trisomy 18: Clinical details of 24 patients receiving intensive treatment. *American Journal of Medical Genetics Part A*, 140(9), 937-944.

WHAT SUPPORT IS AVAILABLE FOR ME?

The HSE Standards on Bereavement Care (2016) affirm that the best perinatal care should be available when baby has a condition which may be life-limiting.

In practical terms, that means obstetricians, nurses, neonatologists, social workers, bereavement counsellors, chaplains and photographers work together to ensure that parents are given the gift of time with their children. This ensures:

- ♥ **Continuity of care**, so that you have a dedicated team who are aware of the baby's diagnosis and don't have to explain your situation at every appointment
- ♥ That a **special, separate room** is made available for you for when your baby is born
- ♥ That **counselling** is available for you throughout your pregnancy and **bereavement counselling** is available when needed
- ♥ That **palliative care** ensures that any discomfort your baby may feel is managed and minimised
- ♥ That a **special photographer** is there for you and your family
- ♥ That a **chaplain** is available to baptise the baby if that is desired

All of this means that you have the gift of time with your baby, however short his or her life may be, and that you can focus on spending time with your child, before and after birth.

WHERE CAN I FIND OUT MORE?

For practical advice, more information and support from parents who have also been in this situation please see:



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