

Care of the Infant and Child with Trisomy 18 or Trisomy 13

The Care Book for Families

5th edition 2025

Kari Simon Krissy Megan Patrick Emerson Stacy Mary
Bella Hayley Gavin Leila Ashton Lunah Conor Lyndsay
Morghan Saskia Ella David Karson Sofie Natalia Hope
Della Nicholas Samuel Zion Rosalie Taylor Phillip Kyle
Avianna Morganne Ashlyn Aniella Eleanor Vivian Faren
Elizabeth Greta Sarah Arianna Joey Shelby Tiffany Ava
Natalee Lillian Emma Kara Eden Akiaya Annabel Ryan
Annie Leilani Cary-Ann Lilly Aaron Zoe Claire Madison
Brandon Alexander Payton Devon Mark Harley Ryleigh
Abigail Brooklyn Natasha Giuliana Remi Becca Hannah
Summer Erin Jillian Nora Faith Darina Kimberly Angela
Lilliana Laurel Joseph Dawson Kameron Madeline Caleb

www.trisomy.org

Care of the Infant and Child with Trisomy 18 or Trisomy 13

"The Care Book" for families

5th edition, 2025

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Cover & last page first names of children born with trisomy 18.13 or related disorders who have been a part of the SOFT family; names shown limited by available space.

SOFT
The Support Organization for Trisomy 18, 13 and Related Disorders
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www.trisomy.org

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While it is unusual for an author to acknowledge a co-author, the many medical papers written by Dr John C Carey are so significant to this book that he is acknowledged for his life-long passion to improve the quality of care for those born with trisomy 18 or trisomy 13.

Kris Holladay, mother of Kari (9/8/77 - 6/3/88) is recognized and honored as the founding parent of the Support Organization for Trisomy 18, 13 and Related Disorders (SOFT).

Barb Van Herreweghe, mother of Stacy (5/21/81) is acknowledged for her many years of leadership as the President of SOFT. Barb reviewed the book and provided comments about care and trusts.

We are grateful to the SOFT families for sharing their stories of parenting a child with these disorders. Some parents sent photographs and many quotes are from the SOFT newsletters, *The SOFT Times*, formerly *the SOFT touch* and Facebook..

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Pamela J Healey, PhD, Special Educator and Developmental Psychologist, Past Board Member of SOFT, and mother of Conor (4/2/86 - 4/9/86) reviewed the text and contributed research information.

Scott Showalter, MD, MPH Pediatrician, Past Board Member of SOFT, and father of Patrick (2/6/87 - 4/18/87) reviewed and added comments to this book.

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**This book is dedicated to
Megan Elizabeth Barnes**

7/8/85 - 12/29/04

Trisomy 18

Beloved daughter of Frank and Ann Barnes

"Every life has value and purpose"

Preamble

This book is a labor of love and gratitude for the almost 20 years that our life was graced by our youngest daughter, Megan, who was born with trisomy 18. Megan was a little teacher who gently guided our hearts.

"Take your baby home and love her for the time you have her". Such words are often said to parents who are fortunate enough to be able to take home their newborn with trisomy 18 or trisomy 13. Loving these precious little babies is the easy part. The fear of her dying was the hard part.

With the gift of time we learned about many health issues, resources, special education, and more. But there was a lack of information about survivors with her diagnosis, and we longed to find other families. It was near her 2nd birthday that SOFT held its first conference.

Finally meeting other children and their families and feeling the sense of worth given to each child by parents and doctors who were at the conference was an uplifting experience that encouraged us to follow our hearts.

We wanted to give our daughter every chance to survive and to be as well as possible. She was content and knew she was loved, and it was obvious that what she valued most was being with the people who loved her.

Megan has touched the hearts of many; even some who never met her. She taught us about the joy of unconditional love and the sorrow of losing a child. She is a part of who her father and I are and always will be.

It is a privilege to share the lessons that have become Megan's legacy, along with the stories that other SOFT parents have related about their children.

MEGAN

The bicycle girl
Red wheels turning
You give it a whirl.

In your stand you smile,
You giggle,
Stretching,
You're very smart!

Megan, dear sweet Megan,
With the red hair, Megan
You touch my heart.

By Joy Acey, classroom teacher aide

MEMORIES OF MEGAN

Only memories remain
Of our lovely Megan
We savor the good ones

Only memories remain
Of dark auburn hair
The ladies loved it

Only memories remain
Green eyes, long lashes
How pretty she was

Only memories remain
Expressive eyes
Said what her voice could not

Only memories remain
Of her sweet disposition
We miss it

Only memories remain
These last forever
They will have to do

By Frank Barnes, Megan's dad

Introduction

We welcome the reader to learning about the daily issues of living for those who have trisomy 18 or trisomy 13 and the challenges encountered by parents who love and care for these infants and children; some are adults. We have drawn from a combination of the available literature regarding these syndromes and information provided by parents who are members of the Support Organization for Trisomy 18, 13 and Related Disorders (SOFT). SOFT membership consists mainly of families who have or had a child with trisomy 18, 13 or related disorders and includes expectant parents and those whose pregnancy ended early. Physicians and other health care providers are also members of SOFT, as well as interested supporters.

SOFT has supported families for nearly four decades and a consistent list of problems and concerns has been reported through the years. **The primary concern of all parents is viability of their newborn, and then if their infant goes home, how to manage the care of a baby with health and disability issues and a prognosis of an uncertain tomorrow.**

We hope this book will be helpful, but **it is not a substitute for the care and advice of your medical practitioners.** Your child should be under the care of a provider who can help you with your child's individual needs. Although we use the title of doctor, we acknowledge the importance of the many health care providers who tend to the health care needs of our children; the pediatrician, the family practice doctor, the physician's assistant, the nurse practitioner, the internal medicine and pediatric certified (Med-Peds) specialist, the comprehensive care specialist, and more.

Other SOFT e-Books See Information >Expecting a Child at www.trisomy.org
Trisomy 18 – A Handbook for Families [Stenson et al., 1993] English or Spanish
Trisomy 13 – A Handbook for Families [Stenson et al., 1992] English or Spanish

Recommended:

Trisomy 18 New and Expectant Parent Resource Guide [by the Support Organization for Trisomy 18, 13 and Related Disorders, 2024] can be downloaded from the SOFT website and is also available on Amazon!

Trisomy 13 New and Expectant Parent Resource Guide [by the Support Organization for Trisomy 18, 13 and Related Disorders, 2024] can be downloaded from the SOFT website and is also available on Amazon!

Preparing for Your Baby's Arrival [Berg et al. 2014] for the parent expecting a baby with trisomy 13 or 18. <https://trisomy.org/wp-content/uploads/2022/08/ITA-Preparing-for-Your-Babys-Arrival.pdf>

I Am Not a Syndrome - My Name is Simon [Crosier S, 2012] Order at www.Simonismyname.com.

Born Abel! The SOFT Landing, by Abel Isaac {2024} a "kid friendly" trisomy awareness book illustrating children with trisomy. A series of Born Abel books are available on Amazon.

The Complex Child E-Magazine <http://complexchild.org/>

Genetics Home Reference <https://ghr.nlm.nih.gov/>



Kari
9/8/1977 – 6/3/1988
Trisomy 18

Kris Holladay, SOFT Founding Parent, is the mother of Kari.



Stacy
5/21/1981 - 4/29/2018
Trisomy 18

Barb VanHerreweghe, SOFT President, 1993- 2023 is the mother of Stacy.



Joey
9/28/1977 -1/7/1984 **Trisomy13**

*"I cried the first time
Joey called me 'Mama'.
I was told he would never
know me. He certainly
knew his 'Papa', when he
came into the room."
P.F., 2007*

Joey is the inspiration for the **SOFT
Conference Attendance Assistance fund.**



Pam Healey, mother of Conor, wrote ***What Should We Do Now*** (2003), for parents of newly diagnosed infants with trisomy 18 or 13. Link below box.



Conor
4/2/1986 – 4/9/1986
Trisomy 18

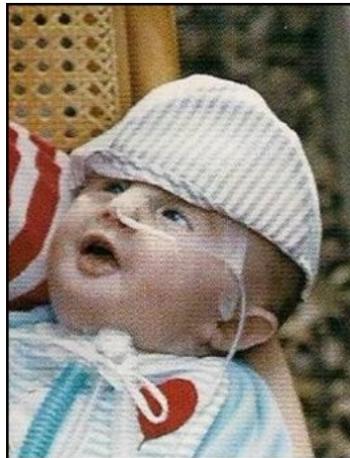
<https://trisomy.org/shall-now/#/>

Leilani
5/3/2000
Trisomy 18



Zion
5/2/2001 – 2/16/2010
Trisomy 13

Zion is the inspiration for the **SOFT conference Parade of Stars**.



Ryan
10/4/1985 – 6/15/1986
Trisomy 18

Ryan is the inspiration for the **SOFT conference Memorial Celebration**.



Ecaterina, "Cati"
4/26/2011 - 2/26/2024
Trisomy 13

"One thing I can say about Cati is that she is loved beyond imagination." R.M., 2013

*"Maggie was like a prism: taking all the love our family shared, making it more, turning it into rainbows."
 J. Z., trisomy 18*

Care of the Infant and Child with Trisomy 18 or Trisomy 13

Parenting an infant, child or adolescent, and in some cases a young adult, born with trisomy 18 or trisomy 13, is a profound learning experience of the heart. The child's unique needs provide an education about a variety of health problems and care issues. While this book relays the reported experiences of infants and children with trisomy 18 or trisomy 13, many of the topics discussed represent challenges that might also affect persons with related disorders.

Diagnosis

The diagnosis of trisomy 18, Edwards syndrome, or trisomy 13, Patau syndrome, is more medically serious and complex than the more commonly known trisomy 21, Down syndrome. Developmental and physical delays are present in all three syndromes, but in trisomy 18 and trisomy 13 these delays are usually greater, and the prognosis includes a much shorter life span as many will not survive their first year of life. **However, these life limiting disorders are not universally lethal.** And, once a child's age is greater than a year there is an 80-90% chance to live beyond age 5 years. (Meyer et al., 2016)

A multi-state population study found 1- year survival improved among children with trisomy 18 to 13.4% and trisomy 13 to 11.5% following more aggressive medical intervention, more than double of the prior figures of 5-6%; 5-year survival was 12.3% for trisomy 18 and 9.7% for trisomy 13. [Meyer et al., 2016]. Other studies have confirmed these improved survival figures (Nelson et al., 2016).

Perspectives on the Care and Management of Children with Trisomy 13 & 18 Syndromes
SOFT Conference, Grand Rounds at Gillette Children's, St Paul, MN, July 12th 2024
Current survival range summary; a slide presentation by Dr John C Carey

Summary

3 – 13% of children survive to 1 year

4-12% >5 years, 10 years

*1-year survival with intensive intervention

35-50%

Beginning in the 1960s, chromosome analysis was done by a karyotype (cells arranged in a specific order, photographed and counted). Karyotype remains useful today for trisomy 18 or trisomy 13. In the mid-nineties the maternal serum triple/quad SCREENING analyzing a mother's hormone level for the **possibility** of a trisomy condition is/was the standard of care. In the first and/or the second trimester of pregnancy a combination of maternal blood or serum tests and fetal ultrasound may detect the possibility of a fetus having trisomy 21 or trisomy 18. When a possibility of trisomy 13 is suspected, it is usually through ultrasound findings and sometimes might be detected through a first trimester blood test called pregnancy associated plasma protein (PAPP). [Best, 2006]

A **negative result** of screening **does not exclude the possibility of a trisomy condition** but means there **is no increased risk detected** for a possible trisomy condition from that particular test. Unless there are other risk factors such as maternal age or previous pregnancy with a genetic disorder, further testing is usually not recommended.

A **positive result from the screening does not mean that a fetus has a trisomy condition**. It does mean there **is an increased chance** that the pregnancy is affected with these problems and further diagnostic testing is warranted. If a screening test shows an increased risk for a possible trisomy condition, diagnostic tests to identify trisomy 18 or trisomy 13 are available but invasive. These tests (*cytogenetic testing*) study the genetic make-up of cells in amniotic fluid obtained through amniocentesis, or tissue from chorionic villus sampling (CVS), or fetal blood. Some parents choose not to do follow-up invasive testing due to a possible 1% or less risk to the fetus and/or personal beliefs.

A new **non-invasive prenatal test (NIPT, often called cell-free DNA testing)** that can detect the **possibility** of **trisomy 21, 18, or 13**, with a single blood draw, as early as the tenth week of pregnancy, was **marketed in 2012**. The first of these tests were advertised as more than 98% accurate with less than 1% False/Positives. (A False/Positive reports a test result as a positive when it actually is not.) After 5 years in use, it has become apparent that a negative result is nearly always accurate, but **a significant number of positive NIPT results have actually been found to be false**. And, to apply a 98% chance for all positive screenings, no matter the woman's age or specific trisomy, can be misleading. Concerned obstetricians and genetic counselors developed a tool based on a woman's age, occurrence of a specific trisomy disorder (trisomy 21 is the most frequent occurring and trisomy 13 the least), and sensitivity of the test to more accurately determine NIPT results, called a **Positive Predictive Value (PPV)** calculator. Some health providers might not know about **PPV** <https://ppv.geneticsupportfoundation.org/>

NIPT looks at cell-free DNA fragments (cfDNA) from the fetus, circulating in the mother's blood and can detect the **possibility** of trisomy 21, 18 or 13 with higher accuracy than the traditional maternal serum screening. Some companies state in their literature that NIPT is only a **SCREENING** and **a positive result needs confirmation by invasive testing, such as amniocentesis**. NIPT benefits and limitations, and the fact that screening and/or invasive testing to confirm chromosomal abnormality is **optional** are usually explained to the expectant parent by their prenatal care provider. **Because NIPT is a screening test, the medical genetics community suggests calling it non-invasive prenatal screening (NIPS). It is predicted to completely replace maternal serum triple/quad screening in the upcoming years.**

If a positive screening is not confirmed, perinatal care as would be done for an unaffected pregnancy is generally provided. **A confirmed diagnosis of trisomy 18 or 13 can limit pregnancy care, delivery, and post birth options for the infant depending on the view of a health provider about these syndromes.**

More than a few SOFT Facebook group posts are from parents searching for a **supportive** Obstetrician (OB) or Maternal-Fetal-Medicine (MFM) specialist.

Another recently approved genetic test that can detect trisomy conditions and more, **chromosomal microarray (CMA)**, is done from samples obtained during amniocentesis or chorionic villus sampling or postnatally from a blood draw. CMA looks at submicroscopic changes within a chromosome, finding information not routinely identifiable by karyotype and gives more information than NIPT, including

findings of unknown significance that can create anxiety in a parent. [Stokowski and Klugman, 2013] Genetic counseling is needed. Check about insurer coverage of CMA, if considering this test.

Genetic counseling is recommended for all women with “at risk pregnancies.” If a diagnosis of trisomy 18 or trisomy 13 is identified before 24 weeks of pregnancy, the provider will discuss the option of termination of the pregnancy. Many who continue their pregnancy mention via Facebook how upsetting it feels to be asked repeatedly about termination by various health providers. **Referral to perinatal palliative care, offering guidance for creating a birth plan, end of life wishes, and care options/decisions for the live born infant is common for those who continue their pregnancy.** These programs are advisory, sometimes done by phone, and **participation is optional.**

Diagnosis and Parents

The journey of families having infants with trisomy 18 or trisomy 13 is particularly unique. First, the family must deal with the low survival rate and then the family must cope with the prospect of significant disability if their infant survives. Mixed feelings about what is best for the infant are a natural occurrence. Ongoing support as well as validation of the **uncertainty** of the situation is crucial at this time for parents. The physician has the unique opportunity of providing this ongoing support. **It is crucial for the doctor to help parents cope with this paradox of preparing for both the probability of death and the possibility of living.** [Carey, 1992]

“We learned of our daughter’s diagnosis in the fourth month of my pregnancy. We knew of Down syndrome, but what was this? After hearing the diagnosis we left the doctor’s office heartsick and numbed by the grim facts.” [the SOFT touch, 1990]

“I felt hopeless and lost. I was torn between being afraid she would die and being scared to death that she would live.” [The SOFT Times, 2002]

Vital to discussion between the doctor and parent is accurate and current information about mortality risk, and developmental outcome of older infants and children with trisomy 18 or 13. All children will progress in (some) developmental milestones, although slowly. [Carey, 2010] **A published study about quality of life in families of children with these disorders found 97% of parents describe their child as enriching the family.** [Javier et al., 2012; Carey, 2012]

Diagnosis and Care

Rapid diagnosis in newborns with suspected trisomy 18 or trisomy 13 can be helpful in making decisions regarding surgical intervention and care. [Carey, 2010] **Medicine recognizes there is increased patient/parent satisfaction in shared-decision making.** [Brosco et al. 2017]. Andrews and colleagues advocated for a shared decision making approach in caring for infants with trisomy 13 and 18 [Andrews et al., 2016] A recent publication in the Journal of the American Medical Association (*JAMA*) advocates for **patient-centered care** after a diagnosis of trisomy 18 or 13 based on the best interest of the parent and child. [Haug et al. 2017]. Yet, for these syndromes, many parents must search to find health providers willing to include them in the decision making prenatally, and/or after the birth of their child. [Andrews et al., 2016]

Most often today, a need for care decisions begins with prenatal diagnosis and continues at birth and during times of health crises for children living with these disorders. The physician able to provide facts about the syndrome in non-judgmental language, explain the specific health concerns of the affected child, and collaborate with the parents in care decisions is greatly appreciated. In an on-line parent survey by Janvier et al., [2012] **more parents chose comfort care than intervention but a greater percent of children treated with interventions lived past their first birthday, compared to those on comfort care.** Some parents choose comfort care at birth, but change their mind later and ask for intervention, such as heart repair. Others choose interventions from birth, as needed, (respiratory, cardiac, nourishment and hydration, even surgery) to give their child every chance possible. Occasionally some have made the decision to cease intervention efforts.

"One of the positive things I remember being said when Joseph was born was, 'Don't be afraid to love this baby'." Advice from the pediatrician who cared for Joe all his life (almost 22 years) and in the end spoke at his funeral. M.S., Joseph, trisomy 18

"Conor was born at a large teaching hospital in 1986 and not diagnosed until three days after he was born. It seemed more was done for him before his trisomy 18 diagnosis than after. At six days old he was sent home to die and lived less than a day. He remains a presence in our lives." P.H., Conor, trisomy 18

There are hospitals that have **futility policies** to protect their physicians from obligation to provide care deemed futile; **a value judgment** related to outcome benefits, and costs when applied to treatment of conditions with very low survival statistics or considered universally lethal. It is appropriate to ask for an explanation of a hospital's futility policy and to convey, to health providers, that trisomy 18 and trisomy 13 are not universally lethal and that **a number of medical studies about trisomy 18 and trisomy 13 indicate improved outcomes and survival following interventions (respiratory, nutrition, cardiac) in the early neonatal period and later as needed for viability.** [Meyer et al., 2016; Kosho & Carey, 2016; Nelson et al., 2016; Cortezzo et al., 2022]

Surgeries are being performed today that were not available thirty years ago, particularly cardiac repair. Finding a willing cardiac surgeon for those with trisomy 18 or 13 is still difficult in some locations. **SOFT has a parent-reported surgery registry of hospitals where cardiac surgery or procedure occurred for those with full trisomy 18 or 13.** See link on page 59.

Hospital Ethics Committees were utilized in the not too distant past for consideration of approval for a heart repair to be done for a child with trisomy 18 or 13. In recent years such meetings are less common in the USA as more and more centers provide surgeries.

"We wanted her to have every chance possible. We met with the Ethics committee. They agreed to do the heart surgery. [The SOFT Times, 2003]

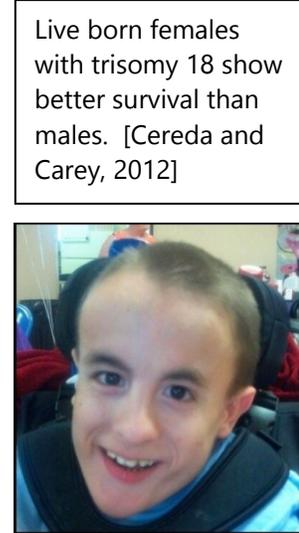
"We asked if the anesthesiologist would like to hold Aaron and Aaron became a human being with a name who responds to touch and cuddling and love. He agreed to assist in the surgery." [The SOFT Touch, 2006]



Joanna
10/14/1974 - 2/4/2013
Trisomy 18



Erin
2/22/78 – 3/19/2013
Trisomy 13



Donald, "Donnie"
9/10/1992 – 6/21/2015
Trisomy 18

Live born females with trisomy 18 show better survival than males. [Cereda and Carey, 2012]

Because of the small numbers who survive past age one year, there are few doctors who have had experience in long term care of a child with trisomy 18. [VanDyke, 1990] Carey estimates there are about 500 living persons with trisomy 18 *older than one year* in the USA. (personal communication, **2024**) Most doctors do not expect these infants to survive; an expectation that might guide a doctor's approach to care. The doctor who **treats each problem as it occurs**, advising interventions to promote wellness and developmental potential, helps prepare the infant and parents for the possibility of living.

Congenital Anomalies

Trisomy 18 (Edwards syndrome) is the second most common autosomal trisomy syndrome and trisomy 13 (Patau syndrome) is the third most common autosomal trisomy syndrome with trisomy 21, Down syndrome, being the most common. [Carey, 2012]

Autosomal refers to any one of the chromosomes that is not a sex chromosome. The autosomal chromosomes are numbered from the largest in size as number 1 to the near smallest, number 22. Every normal human cell contains 46 chromosomes composed of **22 pairs** of autosomes, plus two X chromosomes for a female or an X and Y chromosome for a male. [Stenson et.al., 1993]

Trisomy refers to three copies of a chromosome. When three copies of any one of the chromosomes are present, rather than the normal two, the outcome is 47 chromosomes in the cell. In the case of trisomy 18 and 13, this **extra chromosome results in congenital malformations, serious developmental and**

motor delays, and a high incidence of mortality. A more in-depth discussion of chromosomes can be found in the SOFT Handbooks for families at www.trisomy.org

Syndrome refers to a group of signs and symptoms. Findings of several abnormalities by prenatal ultrasound or present in a newborn alert the doctor to the possibility of a chromosome disorder. With examination the physician may suspect a specific trisomy condition, but a diagnosis requires confirmation.

There are different patterns of malformation in trisomy 18 and trisomy 13. This difference includes the facial characteristics, type of cardiac defect and the potential presence of holoprosencephaly in those with trisomy 13. [Carey, 2012] Those with related disorders such as partial trisomy or mosaic trisomy may also have congenital anomalies. Their prognosis varies depending on the degree of involvement, but it is more optimistic than for those with full trisomy, and they generally achieve greater developmental milestones.

Cardiac defects occur in about 90% of those who have trisomy 18 with the most common defect being ventricular septal defect (VSD) with polyvalvular disease, which is a thickening of the heart valves. About 10% of those with trisomy 18 who have heart defects have a more complicated cardiac malformation. [Carey, 2012; Kosiv et al., 2023]

Cardiac defects occur in about 80% of those with trisomy 13 with the most common defect being shunt lesions such as atrial septal defect (ASD) and ventral septal defect (VSD). **The majority of heart lesions in both syndromes are usually not those that produce neonatal death but occasionally more serious defects can occur.** [Carey, 2010]

Many anomalies do not affect the infant's health, making treatment optional. However, some risk viability and immediate decisions may be needed. **For parents there is often a struggle within themselves and sometimes between the doctor and themselves in deciding what approach to care is in the best interest of their infant. A second opinion might be helpful.**

Anomalies affecting every organ system are noted in studies of those with trisomy 18 or 13, with some overlap between syndromes. Yet, **both syndromes have identifiable patterns of malformation.** [Carey, 2010]. An infant may be born with several abnormalities yet, another with the same syndrome might have only a few. See Tables 1 and 2.

[Table 1 Frequency of Selected Anomalies in Trisomy 18](#)

Frequent Occurrence (> 50%)

Cardiac Defects (VSD, polyvalvular heart defects)

Clenched hands with the index finger overlapping the 3rd and 5th over 4th finger

Hernias (inguinal, umbilical etc.)

Joint contractures (including club foot)

Low arch dermal ridge (shallow finger prints)

Occipital prominence of the back of the head

Shortened big toe (sometimes bent backwards)

Short sternum (breastbone)

Small mouth and jaw

Less Frequent Occurrence (10 – 50%)

Absent or defective thumb development
Cleft lip or palate or both
Deviation of hand at ulna or radius (forearm bones)
Talipes equinovarus (a form of club foot)
Kidney defects
Omphalocele (hernia of the navel)
Ptosis (droopy eye lid)
Scoliosis (curvature of the spine, primarily seen in older children)

Low Occurrence (<10%)

Diaphragmatic hernia
Tracheo-esophageal fistula with esophageal atresia
Dislocated hip
Hydrocephalus (excess fluid in the brain)
Meningomyelocele (a form of Spina Bifida)
Ocular defects (eye)
Radial Aplasia (lack of a bone that is part of the forearm)
_____ [adapted from Jones 2021; Cereda and Carey 2012]

Table 2 Frequency of Selected Anomalies in Trisomy 13

Frequent Occurrence (> 50%)

Brain abnormalities, especially **holoprosencephaly** (the forebrain fails to divide properly)
Cardiac defects, ASD, VSD, PDA (patent ductus arteriosus)
Capillary hemangioma (birthmark of tiny blood vessels close to skin surface)
Cleft Lip or Palate or both
Dextrocardia (reversed or right sided heart)
Hernias (inguinal or umbilical)
Microcephaly (moderately small head, sloping forehead)
Ocular abnormalities (such as small or absent eyes)
Polydactyly (extra 5th finger beside little finger or extra 5th toe beside little toe)
Posterior prominence of heels
Scalp Defects

Less Frequent Occurrence (< 50%)

Cystic Kidneys or other kidney defects
Meningomyelocele (a form of spina bifida)
Omphalocele (abdominal wall defect-opening at navel allows organ protrusion)
Radial Aplasia (lack of a bone that is part of the wrist)
_____ adapted from Jones, 2021]

Frequency of trisomy 18 or 13 is similar in all cultures and nations. The number of affected pregnancies in both disorders is greater than the number of live births, due to stillbirths and elective terminations. **Estimates of termination are about 90% in Europe and about 75% in the USA.** The 1994 Utah study indicated 1 in 6000 live births in trisomy 18. [Root and Carey, 1994] With the increase in use of prenatal diagnosis and average maternal age, recent studies indicate the **over-all** frequency of trisomy 18 pregnancies has increased to 1 in 2500 while the **number of live births has decreased** to 1 in 7000. Updated 2004-2009 data from the UK indicates live births of trisomy 18 are 1:10,000. (Carey conference workshop, 2012) The best estimate of live births of trisomy 13 is about 1 in 10,000. [Carey, 2021]



Annie
5/25/05 - 8/12/05
Trisomy 13

Annie's mother, Barb Farlow, co-authored:

***The Experience of Families with Children
with Trisomy 13 and 18 in Social Networks***

Janvier A, Farlow B, Wilfond B, 2012

Preparing for Your Baby's Arrival

Berg S, Farlow B, Robbins J, Bruns D 2014

Developmental Achievements

Individual case studies published in the 1970-80s have some information about the development of an older child with trisomy 18 or trisomy 13 based on single case reports, but after the first annual gathering of SOFT in 1987 the opportunity arose to study a group of children with these disorders. Baty and colleagues note that the 1st work in pediatric literature to discuss the challenges of parents of children with trisomy 18 used the SOFT support group for case identification.

This study directed physicians to approach long term management of care of children with trisomy 18 as they would for any child with disabilities. [Van Dyke and Allen, 1990; Carey, 2012]

A 1994 publication of a study by Baty et al., involved SOFT parents who filled out questionnaires about the psychomotor development of their child with trisomy 18 or trisomy 13. Medical records for these children were also obtained for this study with permission of the parents. Common activities reported for both disorders were as follows and show a progression of skills by gain in age:

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**First year:** cooing, rolling, smiling responsively, reaching and recognizing close adults.

**Two & three years:** sitting supported, object permanence, imitation, playing baby games, sitting independently and recognizing words.

**Four to six years:** commando crawling, independent playing, following simple commands, helping with hygiene tasks, standing, understanding cause and effect, and use of signs.

**Older children:** identified common objects, used a walker, crawled, understand words and phrases. Walking and some toileting skills were also reported for trisomy 13.

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[Baty et al., 1994]

Reported developmental milestones for both disorders were tabulated by Baty et al. (See Developmental Achievements in Trisomy 18 and Trisomy 13, Table 4). Within SOFT newsletters, a few parents of children with trisomy 18 note successful assisted toileting skills. Two children with trisomy 13 appropriately use(d) one-word utterances, as reported by parents contributing to this book. Independent standing by a couple of children with trisomy 18, with a parent for safety near by, has been seen at SOFT conferences.

Although individuals with trisomy 18 and trisomy 13 were clearly functioning in the significant (severe) to profound range of developmental disability, they did achieve many skills of childhood even beyond their developmental age, and always continued to learn. [Baty et al., 1994] Children with trisomy 18 and trisomy 13 do progress even though there is significant disability, and it is not appropriate to use the term "vegetative" as is sometimes used in life support decisions in intensive care situations. [Carey, 2012] **Baty reported developmental ages in older children with trisomy 18 averaging at 6 to 7 months, and in those with trisomy 13 averaging at 13 months.** [Carey, 2012]

*Joey's tenacity eliminated all doubts any of us had about him! He did sit up, crawl and walk! He learned to say 'Papa, Mama, up and go' and he used them appropriately!" P.F., trisomy 13*

Pamela and Michael Healey had a son, Conor (4/2/86 - 4/9/86) who was born with trisomy 18. A 2001-2 study by Pamela J. Healey, PhD, involved SOFT parents, who completed questionnaires and interviews about their child's social, motor, communication and adaptive behavior. See Table 3 on page 22.

"Development continues across the lifespan of each syndrome with some loss of skills, particularly motor, with age related or illness-related constraints". [Healey, 2003] Those with progressive illnesses eventually might have more difficulty with skills, and hospitalization and/or surgery can result in setbacks with long time periods needed to regain skills, if regained at all.

**The gap between children with trisomy conditions and their typically developing peers widens as they get older, so development quotients decrease, but skill acquisition continues. [Healey, 2003]**

Baty et al. noted this drop in the developmental curve for those with trisomy compared to the average developing child **does not represent a loss of skills**, but rather greater distance from the normal curve. An on-line survey of parents belonging to support groups for trisomy 18 and 13, described significant disability for surviving children but report the children do learn and progress at their own pace. [Janvier et al. 2012]

**Table 3**

**Trisomy 18 and Trisomy 13 Development and Skill Study**

**Children studied**

37 children with full trisomy 18, age range of 18 months-21 years

12 children with full trisomy 13, age range of 13 months-24 years

**Over-all development** of those with trisomy 18 was below 18 months. Those with trisomy 13 were a few months higher.

**Social development** was the area of **greatest strength** for both trisomy 18 and trisomy 13. Most with trisomy 18 demonstrated social development at 7-24 months of age with a few scoring lower. Those with trisomy 13 functioned within 7-24 months and more children with trisomy 13 approached the two year level than those with trisomy 18.

**Communication skills** showed functioning for both disorders with 50% at 6-12 months. Some with trisomy 13 and a few with trisomy 18 scored in the 12-18 month level. Nearly a third with trisomy 18 but fewer than 10% with trisomy 13 functioned below 7 months in communication.

**Motor skills** showed 50% of those with trisomy 18 functioning below 7 months with 90% scoring at 12 months or lower. Those with trisomy 13 were slightly stronger with 75% at or below 12 months.

**Daily living skills** showed that nearly 20% of those with trisomy 18 functioned higher than age 12 months. In trisomy 13, a third demonstrated skills higher than 12 months.

*Social Development of Children with Trisomy 18 and Trisomy 13 in the Context of Family and Community [Healey PJ, 2003]*

20 children with **mosaic or partial trisomy 18 or 13**, (25 months - 17 years) had better overall function than those with full trisomy 18 or 13, but some functioned at levels consistent with full trisomy. 20% demonstrated social skills of at least 36 months with some higher than 48 months. More than 50% had communication skills of at least 12 months with a few at 36 months and higher. [Healey, 2003]

Physical, Occupational, Speech, and Vision therapies are available to children who meet criteria for **early intervention** programs or **special education** at public school. Some with trisomy 13 who have both vision and hearing impairment might need a more comprehensive program. Parents who take their child for extra therapy in a clinic setting need a referral from the child's doctor.

SOFT's *Trisomy 18 Handbook for Families* and *Trisomy 13 Handbook for Families* includes information about early intervention and public school education. Contact the local department of education about registration and location of the school your child will attend. Special education might not be provided in every school setting, but public law mandates availability within a district, and bus service is to be provided.

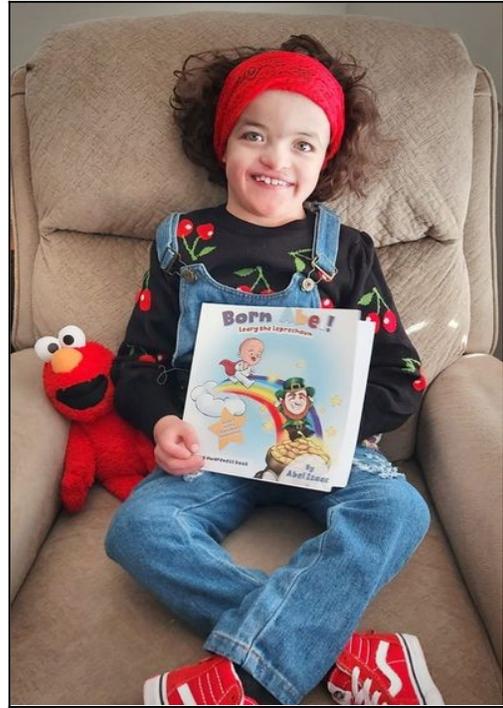
**Referral to early intervention is recommended in ongoing care of infants and children with trisomy 18 and 13. [Carey, 2021]**

*"Stacy understands much more than she is often able to communicate to us." B.V., Stacy, trisomy 18*



**Morghan**  
**9/14/1996**  
**Trisomy 18**

Morghan had a VSD repair when eight weeks old and weighing 6 pounds, 8 ounces.



**Lunah**  
**12/3/2015**  
**Trisomy 18**

Lunah is one of many children illustrated in the Trisomy Awareness *Born Able* series of books.

*"Morghan started using a walker at about age seven for mobility purposes at school. However, she walked holding your hand at about four and we still hold her hands to walk at home." M.K., 2007*

*"I'd begun to think that maybe she was one of the small percentage that would make it to her first birthday." [The SOFT Times, 2004]*

*"After 5 years of holding so tight, it was difficult at first to relinquish him to school. But he loves it!" M.S., Joseph, trisomy 18*

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**Table 4**

| <b>Developmental Achievements in Trisomy 18 and Trisomy 13</b> |                      |              |          |                     |  |
|----------------------------------------------------------------|----------------------|--------------|----------|---------------------|--|
| <b>Trisomy 18</b>                                              | <b>Av. Mos. (SE)</b> | <b>Range</b> | <b>N</b> | <b>Normal Range</b> |  |
| Smiled responsively                                            | 4.7 (0.5)            | 0.5-24       | 54       | 0-2                 |  |
| Held head up                                                   | 9.0 (1.5)            | 0.3-36       | 33       | 0-2.5               |  |
| Watched toy or face                                            | 4.4 (0.6)            | 0.2-24       | 57       | 0-1                 |  |
| Reached for object                                             | 9.6 (1.2)            | 2.5-36       | 38       | 3-5                 |  |
| Laughed out loud/giggled                                       | 13.0 (3.1)           | 2.3-96       | 36       | 1.5-3.3             |  |
| Sat up with help                                               | 20.4 (2.9)           | 3.5-60       | 25       | 1.6-4.3             |  |
| Sat up alone                                                   | 38.5 (6.3)           | 7.5-72       | 12       | 4.8-7.8             |  |
| Said consonant sounds                                          | 23.0 (6.2)           | 8.0-52       | 8        | 5.6-10              |  |
| Rolled over                                                    | 30.5 (16.5)          | 0.2-540      | 32       | 2.2-4.7             |  |
| First tooth                                                    | 11.5 (0.7)           | 4.0-20       | 30       | 4.0-17              |  |
| Balanced on hands and knees                                    | 53.7 (18.1)          | 12-204       | 10       | -                   |  |
| Walked in walker                                               | 39.5 (7.4)           | 24-60        | 5        | -                   |  |
| Cruised furniture                                              | 72                   |              | 1        | 7.4-12.7            |  |
| Walked alone                                                   |                      |              | 0        | 11.2-14.4           |  |
| Used signs                                                     | 61.5 (9.9)           | 36-84        | 4        | -                   |  |
| Number of signs                                                | 2 (0.4)              | 1-3          | 4        |                     |  |
| Number of words                                                | 3.4 (0.7)            | 1-5          | 5        |                     |  |
| -----                                                          |                      |              |          |                     |  |
| <b>Trisomy 13</b>                                              | <b>Av.mos. (SE)</b>  | <b>Range</b> | <b>N</b> | <b>Normal Range</b> |  |
| Smiled responsively                                            | 5.5 (1.3)            | 0.5-1.5      | 12       | 0-2.0               |  |
| Held head up                                                   | 9.5 (2.4)            | 0.7-24       | 10       | 0-2.5               |  |
| Watched toy or face                                            | 8.4 (3.3)            | 0.9-40       | 12       | 0-1.0               |  |
| Reached for object                                             | 14.2 (2.3)           | 4.5-30       | 10       | 3.0-5.0             |  |
| Laughed out loud/giggled                                       | 10.4 (2.0)           | 4-20         | 9        | 1.5-3.3             |  |
| Sat up with help                                               | 22.4 (3.1)           | 15-36        | 7        | 1.6-4.3             |  |
| Sat up alone                                                   | 31.0 (5.7)           | 23-42        | 3        | 4.8-7.8             |  |
| Said consonant sounds                                          | 19.4 (7.6)           | 11.8-27      | 2        | 5.6-10              |  |
| Rolled over                                                    | 11.2 (1.9)           | 4-24         | 10       | 2.2-4.7             |  |
| First tooth                                                    | 10.0 (1.3)           | 4-18         | 10       | 4.0-17              |  |
| Balanced on hands and knees                                    | 41.5 (6.5)           | 35-48        | 2        | -                   |  |
| Walked in walker                                               | 32.5 (12.1)          | 9-58         | 4        | -                   |  |
| Cruised furniture                                              | 56.5 (15.5)          | 41-72        | 2        | 7.4-12.7            |  |
| Walked alone                                                   | 112                  |              | 1        | 11.2-14.4           |  |
| Used signs                                                     | 72                   |              | 1        | -                   |  |
| Number of signs                                                | 6                    |              | 1        |                     |  |
| Number of words                                                |                      |              | 0        |                     |  |

Source: Baty BJ, Jorde LB, Blackburn BL, Carey JC (1994) Natural history of trisomy 18 and trisomy 13. II. Psychomotor Development. Am J Med Genet 49:190 c 1994 Wiley-Liss, Inc. Reprinted by permission of John Wiley & Son

Table 5

| <b>Prevalence of Common Illnesses &amp; Health Problems in Trisomy 18 or Trisomy 13</b> |                         |
|-----------------------------------------------------------------------------------------|-------------------------|
| <u>Frequent Incidence</u>                                                               |                         |
| Airway problems, including tracheal/bronchial abnormalities                             |                         |
| Central apnea                                                                           |                         |
| Obstructive apnea                                                                       | Photophobia (18)        |
| Congenital Heart Defects                                                                | Pneumonia               |
| Constipation                                                                            | Pulmonary Hypertension  |
| Ear Infections                                                                          | Scoliosis               |
| Elevated Blood Pressure (13)                                                            | Seizures                |
| Feeding difficulties                                                                    | Sinusitis               |
| Gastroesophageal (GE) Reflux                                                            | Urinary Tract Infection |
| <u>Less Frequent Incidence</u>                                                          |                         |
| Cataracts in children with trisomy 13                                                   |                         |
| Glaucoma ( 13)                                                                          |                         |
| Wilms Tumor (kidney cancer) in children with trisomy 18                                 |                         |
| <u>Low Incidence</u>                                                                    |                         |
| Hepatoblastoma (liver tumor) (18)                                                       |                         |
| Other anomalies                                                                         |                         |

## **Central Nervous System Problems and Abnormal Muscle Tone**

Abnormalities of the brain and dysfunction of the central nervous system contribute to health issues common for those with trisomy 18 or 13; apnea, seizures and muscle tone problems.

### **Central Apnea**

Infant sleep apnea is defined by the American Academy of Pediatrics as "an unexplained episode of cessation of breathing for 20 seconds or longer, or a shorter respiratory pause associated with bradycardia (slow pulse), cyanosis (bluish discoloration of skin, nail beds), pallor (loss of color, pale), and/or marked hypotonia (floppy muscle tone)."

**Prematurity is the most common cause of infant central apnea but it is important to consider other causes. Physicians should keep epileptic apneas in mind when treating apneas in neonates with trisomy 18.** [Fukasawa et al. 2015]

Traditionally it is suggested that **central apnea**, either alone or in combination with other health issues, where breathing pauses due to central nervous system dysfunction of the respiratory center in the brain, is the most common cause of death in those less than one year of age with trisomy 18 or trisomy 13. [Root

and Carey, 1994; Embleton et al., 1996; Wyllie et al., 1994; Carey, 2010] **However this premise has been recently questioned (see below). Upper airway difficulties always need to be assessed before concluding central apnea alone.**

Some parents post on social media that **caffeine treatment** for central apnea has helped; others say it had no effect. Dr Carey has seen caffeine prescribed a few times for infants with central apnea but with no definite benefit. "Treating central apnea with caffeine is an option about which parents can ask their doctors for their opinion." (Carey, 2012, personal communication)

The primary categories of apnea are Central apnea, Obstructive apnea or Mixed apnea which is a combination of both central and obstructive apnea. See more about obstructive apnea on page 61.

Dr Martin McCaffrey, Neonatologist, UNC Chapel Hill, NC and SOFT Medical Board member recently responded to a SOFT Facebook question, with **current thinking in 2024 about apnea.**

**"It is increasingly clear that trisomy related apnea is very often associated with airway findings leading to obstructive apnea. Central apnea might occur with seizures, maybe severe brain anomalies, but generally the apnea we are seeing in trisomy patients is obstructive. The way to diagnose is a sleep study and an airway evaluation."** [McCaffrey M, SOFT Facebook 2024]. **The airway evaluation is done by an Ear-Nose -Throat doctor or a pulmonologist.**

**The various causes of obstructive apnea are treatable and thus making this distinction between central and obstructive apnea vitally important.**

## Seizures

Seizures are a functional nervous system problem occurring in about 25% to 30% of older infants and children with trisomy 18 or trisomy 13 [Jaspersen et al., 2023]. At the 2024 SOFT conference Grand Rounds for healthcare professionals, Dr Meghan Candee, Pediatric Neurologist, Primary Children's Hospital, UT and coauthor on the above cited paper, presented a summary of a *Tracking Rare Incidence Syndromes* (TRIS) database survey of 186 children with trisomy 18 which reported 30% experienced seizures. See *Perspectives on the Care and Management of Children with Trisomy 13 & 18 Syndromes* on SOFT's YouTube Video Channel under Resources at [www.trisomy.org](http://www.trisomy.org).

Steven Cantrell, OD, notes **visual triggers** mentioned by his adult patients with photosensitive epilepsy, that cause seizures. He suggests the same triggers might affect children with trisomy 18 or trisomy 13 and advises that these children wear protective glasses when around family using computers, television, cell phones and other LED technology. These are known triggers for children at risk. [Cantrell, SOFT times Newsletter, 2016] See page 40 for more from Cantrell on *Photophobia and Photosensitive Epilepsy*.

Parents should inform the doctor, if their child is experiencing unusual repetitive motions, shaking, a blank stare, eyes fluttering, head or eyes turned to one side, or any other concerning behavior, as **parental observation, as well as testing, help determine if a child is experiencing seizures.**

If seizures occur, a referral to a pediatric **neurologist** can be made by the child's care provider. Testing is done with an electroencephalogram (EEG) that measures electrical activity in the brain by using sensors attached to the scalp, done in an overnight sleep study.

There are a variety of different medications to treat seizures, and the neurologist will need the parent's observations to help determine if a medication is effective or if there are problems with side effects. Difficulty in finding the right medication has been reported by a number of parents in the SOFT Facebook group. A few parents posted other measures to help prevent seizures, (e.g., Ketogenic diet, Vagus nerve stimulation), were tried with various outcomes. This is an area worthy of investigation.

### [Holoprosencephaly](#)

Seizures tend to be more complicated in children with trisomy 13, possibly related to the presence of a structural defect of the brain called **holoprosencephaly (HPE)**. HPE is a failure of the embryonic forebrain to sufficiently divide into the double lobes of the cerebral hemispheres. **HPE malformations range from mild to severe and are commonly present to some degree in about 60-70% of infants with trisomy 13.** [Carey, 2005] Neuroimaging can determine the presence of holoprosencephaly and is important in predicting prognosis in those with trisomy 13. Although it is known that children with trisomy 13 *without* holoprosencephaly have central apnea, those who survive the first year of life do *not* have semilobar (the brain's hemispheres are partially divided) or lobar (the brain has not divided at all) holoprosencephaly. [Reynolds et al., 1991; Morelli et al., 2000; Carey, 2010], **suggesting that the presence of HPE is a risk factor for survival in trisomy 13.**

### [Abnormal Muscle Tone](#)

Hypertonia refers to abnormally high muscle tone. Hypotonia means abnormally low muscle tone. Secondary or functional neurological findings in both trisomy 18 and trisomy 13 include hypotonia in infancy and hypertonia later in childhood. Older children with trisomy 18 usually have a mixture with low toned trunk and arms and with legs that eventually increase in tone. [Carey, 2010]

Central nervous system problems and abnormal muscle tone affect the development of motor skills. Related to this, children with trisomy 18 or trisomy 13 can develop chronic health problems such as **scoliosis** (curvature of the spine), hip dislocation and tightening of their tendons (contractures). The development of **strabismus** (eye(s) turn inward or outward) can result from unequal ocular muscle tone. **Nystagmus** (involuntary eye movements) can be of neurological origin. (personal communication, Steven Cantrell, OD, 2011)

If appropriate, the child's doctor can make referrals to a pediatric **orthopedic** doctor for skeletal problems, and a pediatric **ophthalmologist** for eye problems. Parents mention **preventive orthotics** prescribed for their child; a custom-made orthopedic body jacket to slow further progression of scoliosis, ankle foot orthotics (AFOs, also known as shin splints to prevent contractures), and eye patches or glasses to strengthen eye muscles as prescribed by specialists. **In more recent years parents report that Adjustable Growing Rods were used to delay a spinal fusion until their child was older.** See more about scoliosis page 49.

## Head Lag

Infants/babies with trisomy 18 or trisomy 13 frequently hyperextend their head or let their head lag towards their back. Such posturing is a sign of poor muscle tone. The nursery staff can show parents how to position and support the baby's head for a feeding. When appropriate, the child's doctor can refer the baby to a physical therapist for advice about positioning, supportive seating equipment for proper body alignment and therapy to improve muscle tone. With time and therapy, head control usually improves.

At Grand Rounds for healthcare professionals during the SOFT conference at Gillette Children's, St Paul, MN, **2024**, Dr Glenn Green, Professor Otolaryngology (ENT) **mentioned head lag along with subtle retractions as signs of obstructive apnea.** See *Perspectives on the Care and Management of Children with Trisomy 13 & 18 Syndromes* on SOFT's YouTube Video Channel under Resources at [www.trisomy.org](http://www.trisomy.org).

## Feeding Problems

A baby born with trisomy 18 or trisomy 13 usually begins life with feeding problems which require patience and persistence from parents and help from health care providers. Some newborns are too ill or premature to feed and are sustained temporarily by intravenous (IV) nourishment until able to tolerate feedings.

Infants and children need to consume enough calories for energy, comfort, and growth plus enough fluid for adequate hydration. Infants are usually nourished through their action of sucking but newborns with trisomy 18 or trisomy 13 often have a suck that is weak and they tire quickly from the effort of breast or bottle feeding. A few parents reported their infant had no ability to suck at all. **These infants are at risk for aspiration of nourishment.** Aspiration is an inhaling or trickling of a small amount of liquid from the esophagus through the trachea into the lungs which could precipitate aspirational pneumonia. **(See page 32 for more about gastroesophageal reflux and aspiration.)**

Poor co-ordination of the muscles used to breathe, suck and swallow is a common problem for these infants, causing inadequate intake, choking and sometimes vomiting. Such problems make it difficult and time-consuming for caregivers to feed an infant enough formula for nourishment and hydration. Feeding issues often continue for the older baby and child. If appropriate, referral to a dysphasia (difficulty swallowing) clinic, or feeding specialist for advice about oral exercises, feeding techniques and adaptive devices to aid feeding and/or referral to a pediatric **gastroenterologist** for evaluation might be done. **It may be necessary to perform a radiographic (x-ray) dye study to confirm that an infant can protect his (or her) airway from aspiration. "Even a child who appears to be able to bottle or breast feed should have an evaluation of feeding to evaluate airway competency." [Carey, 2010]**

Most infants/babies with trisomy 18 or trisomy 13 are gavage fed by a **nasogastric (NG)** tube inserted through the nose or **orogastric (OG)** tube inserted through the mouth, down through the esophagus into the stomach. Eventually many parents opt to have a permanent gastrostomy tube (G-tube). The SOFT Surgery Registry records indicate that the **most commonly reported surgery** for infants and children with trisomy 18 or trisomy 13 is the placement, through the abdomen into the stomach, of a permanent gastrostomy tube (G-tube). A few parents reported using a pacifier in their infant's mouth for oral stimulation whenever they gavage fed their baby. Eventually some infants learned to take a bottle.

Breast feeding is more difficult than bottle feeding for all babies. To breast feed, the help of a lactation consultant will likely be needed. More often than not, infants with trisomy 18 or trisomy 13 are unable to learn how to bottle or breast feed; however some parents report success with bottle feeding, and more than a few report success with breast feeding.

*"She had not yet been diagnosed and was discharged to home at 7 days old. It took two weeks of trying before she finally learned to latch and suck. She was totally breast fed her first year and gained 10 pounds which tripled her birth weight. Going from almost 5 lbs to 15lbs!" A.B., Megan, trisomy 18*

*"The nurse and I worked on her sucking and we would put a pacifier in her mouth during feeding times. She ...finally started to drink from a bottle." [The SOFT Times, 2001]*

## Clefts

Orofacial clefts are present in about 10% of infants with trisomy 18 and 60% - 80% of those with trisomy 13. [Jones, 2018] Cleft lip might prevent an infant from getting the mouth closure around a nipple that is needed for effective sucking. Cleft palate can allow some nourishment to seep into the nasal passages through the roof of the mouth. These babies usually require special nipples and specific guidance from the nursery staff or feeding specialist for oral feeding. The **Haberman Feeder** has been mentioned by parents of those born with cleft palate as enabling their child to learn to bottle feed. This product has a new name and is now called a **Special Needs Feeder**. The company that markets this item finds it to be beneficial for infants with other issues as well as cleft palate and also have a mini version for premature infants. Special Needs Feeders can be ordered from Medela, Inc. at [www.medela.com](http://www.medela.com) > search *special needs feeder* or (1-800-435-8316 English or Spanish). Parents should check with their insurer about coverage as these expensive nipples are needed for a medical reason.

The **SOFT Surgery Registry** shows more cleft lip and/or palate repairs were done for those with trisomy 13 compared to repairs for those with trisomy 18. Repairs can involve more than one surgery. If the child has a cardiac defect, the plastic surgeon must consult with the child's cardiologist and risks versus benefits of surgery should be discussed with the parents. Other specialists, such as an ear, nose and throat (ENT) surgeon might be consulted in this type of repair. Some parents opt for no surgery.

*"Grace was drinking out of a **Haberman** bottle. It is a special bottle designed for babies with cleft palates." [The SOFT times, 2004]*

*"She was initially fed by tube but four days later she was able to take formula and breast milk by the **Haberman** feeder." [The SOFT Touch, 2002]*

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"Sofie Marie feeding from her Haberman bottle is also fed by naso-gastric tube. Consults will be done for bilateral cleft lip and palate repairs"
K.H., 2014



Sofie Marie
1/24/2014 - 10/29/2024
Trisomy 13

Patrick
2/6/1987 – 4/18/1987
Trisomy 18

"Patrick taught us so much about love in so little time." V.S., 1986

Irritability

Some parents report that their babies with trisomy 18 or trisomy 13 are often fretful. Babies swallow air, especially while being fed, which can cause abdominal discomfort. It is helpful to burp a baby during a feeding and when the feeding is finished to burp the baby again. Constipation (see page 34) causes discomfort and is a common problem in trisomy 18 or trisomy 13. Gastroesophageal reflux (GERD) (see page 32) contributes to discomfort and is a known problem in trisomy 18 or 13. Babies with feeding difficulties may also be uncomfortable and irritable due to hunger.

All babies have fussy times and for first time parents it may be hard to know if your baby is excessively fretful. Inform your child's doctor if you are struggling frequently with trying to comfort a fussy baby. Medications for symptoms can be discussed with the baby's doctor and formula changes might also be considered, if needed.

A small percentage of all babies have **colic**. When babies cry without being sick, hungry, uncomfortable, or in pain it is called colic. Colic usually begins around three weeks of age and resolves around three months for the average baby. Unexplained crying in a baby is very stressful for parents. Let your pediatrician know if your baby is crying excessively.

Fatigue

Congenital heart defect or disease, abnormal muscle tone, and low weight are issues that contribute to fatigue in an infant. The work of coordinating breathing, sucking and swallowing for nourishment uses calories and some babies tire quickly with the effort. Small amounts of formula, given frequently, and using pre-softened preemie nipples may be less tiring for the infant. Feeding a newborn infant every two or three hours will exhaust a parent, thus **it is important for the parent to let their spouse or some other person help as much as possible**. Weight gain is slow for these infants. Gavage feedings are given by some parents as a supplement to oral feeding. Some infants might need to be totally fed by gavage. A few

newborns with trisomy 18 or 13 had a gastrostomy placed for tube feeding (see next topic) before discharge to home from birth.

~

"Nothing about her care has been easy. ...she was not able to nurse or take a bottle without tiring excessively." [The SOFT Times, 2003]

Tube Feeding

For some parents a decision for surgical placement of a permanent gastrostomy tube (G-tube) is made shortly after birth and for others a decision is made weeks, months or even years later. The primary care giver (usually the mother) assumes she will be able to feed her infant or child and might feel a loss of normalcy or even a sense of failure when realizing a feeding tube is needed. **Few infants and children with trisomy 13 or 18 can be adequately nourished and hydrated without the help of a feeding tube.** Many parents find use of the feeding tube benefits both the child and the parent. While most utilize commercial enteral formulas to tube feed their child, in recent years, quite a number of parents post via Facebook about home blenderized feeds. Talk with your child's doctor if considering this and **be aware** of the **importance of consulting with a dietitian before using home blenderized feedings.**

After the surgical placement of a gastrostomy tube, through the skin, abdomen and into the stomach, the opening is allowed time to form a tract around the G-tube for four to six weeks, and then, the doctor removes the G-tube and instead inserts a gastrostomy button (G-button) into the opening. The G-button placement is done in the doctor's office or clinic. The G-button provides a skin level opening to the stomach that allows attachment of a tube for feedings. Having the G-button is more convenient as it is easier to put clothing over it and the caretaker doesn't have to worry about inadvertently catching or pulling on a G-tube that extends out for many inches. There are several kinds of G-buttons but availability may depend on which company a hospital has a contract with at the time of surgery.

If a child with trisomy 18 or trisomy 13 is not bottle or breast feeding by 6 months of age, consideration of gastrostomy placement is indicated. Even a child who appears to be able to bottle or breast feed should have an evaluation of feeding to evaluate airway competency. [Carey, 2010]

When a child has a cardiac defect or disease, a pediatric cardiologist must be consulted prior to any surgery and the benefits versus the risks of surgery should be discussed. Some parents prefer to avoid surgery and, if needed, feed by gavage. Hospital staff can teach parents how to safely insert the tube and how to check to be sure the tip is into the stomach. Sometimes the feeding tube is taped to the infant's face to keep it in place to use for more than one feeding but parents still must check for safe placement of the tube before each feeding. A stethoscope is used for this purpose. Skin irritation from tape can be problematic. Let the doctor know if this is happening.

Some babies or children fed through a tube are also fed by mouth. Many are totally tube fed, and a few are fed only by mouth. A few eventually learn some self-feeding skills with the help of an occupational therapist or speech pathologist. **A diagnosis of trisomy 18 or trisomy 13 makes your child eligible for early intervention and special education programs that provide therapies to those who meet the criteria.**

"After much perseverance (and luck) Joseph now eats table food with his fingers and sometimes a spoon. He drinks from a cup and even a juice box." M.S., Joseph, trisomy 18

"Feeding him was difficult at first. But it got better and he learned to eat finger foods and drink from a cup unassisted." P.F., Joey, trisomy 13

~

"The G-button (gastrostomy opening through the abdomen) at 5 years old was the best thing we ever did." B.V., Stacy, trisomy 18

Gagging

Babies occasionally gag, spit up and sometimes vomit while being fed, but in trisomy 18 or trisomy 13, these problems often occur for several reasons. Difficulty handling oral secretions such as mucus (phlegm) draining into the throat from upper respiratory areas is common and can result in gagging and/or vomiting. Upper respiratory infections (colds) increase the production of phlegm. **Inadequate fluid intake and certain medications** may cause dryness, which thickens mucus secretions. Guidance of a nutritionist about adequate hydration and a feeding specialist about gagging and other feeding issues can be helpful. Gagging may be a behavioral issue or a sign of some other health problem such as gastroesophageal reflux. See Gastroesophageal Reflux below. **If a child has frequent gagging episodes, parents should inform their child's doctor.**

"She definitely showed signs of something different going on. She began to gag much more and didn't seem to tolerate her feedings as well." [the SOFT touch, 1993]

"She occasionally had gagging episodes, but then it increased to one to two times daily. The pediatrician listened to my concerns and with testing found a problem that needed surgery. A.B., Megan, trisomy 18

Gastroesophageal Reflux and Aspiration

Usually the parents of the infant or child with feeding issues will be questioned by the doctor about the possibility of reflux. Gastroesophageal reflux is a backward flow of a small amount of stomach contents upward to the throat. **Gastroesophageal reflux is a consistent finding in infants with trisomy 18 and 13. In older infants, it is a potential explanation for irritability and recurrent pneumonias. Assessment of the presence of gastroesophageal reflux as a potential factor in feeding problems should occur.** [Carey, 2010]

Chronic acid reflux over time can irritate the esophagus and put a baby at risk for aspiration. Aspiration is an inhaling or trickling of a small amount of liquid from the esophagus through the trachea into the lungs. It can contribute to the development of an aspirational pneumonia. **Aspiration during feeding or from reflux may precipitate an early death.**

Simple measures can help decrease the occurrence of reflux such as keeping a baby's head elevated about 30 degrees or more after a feeding while digestion occurs, and raising the crib mattress on one end by 30 degrees for sleep. Do not use pillows as a firm mattress surface is safer. It is very **important to ask the doctor about recommended sleep positions** as reflux or frequent spitting up problems can result in pneumonia due to aspiration. Reflux often occurs for infants in general and can be treated with medication prescribed by the doctor but, if persistent, referral to a pediatric gastroenterologist for testing and treatment recommendations will be needed.

A fundoplication surgery is done to block stomach acid from backing upwards into the esophagus and might be suggested, if medication for reflux fails. The SOFT Surgery Registry as of **2024** shows that 23% of children with trisomy 18 and a G-tube also had a fundoplication; 68% with trisomy 13 and a G-tube also had a fundoplication. These procedures might not be done during the same surgery. If a child has a heart defect and a surgery is being considered, a pediatric cardiologist must be consulted. Some parents decline surgery.

Typically, hospital staff work with parents to assure their newborn can take nourishment either orally or by gavage feeding or a gastrostomy tube before the infant is discharged to go home. Infants able to be discharged are sent home with appointments to follow up with their doctor, and/or **pediatric hospice** or the new **pediatric palliative care** for those with life limiting conditions. **Participation in these programs is optional.** See page 73. **Be aware** that problems noted in this chapter may not be apparent in the newborn but with time might need evaluation and treatment by a healthcare provider.

Comfort feeds

Sucrose (sugar water) might be fed to an infant in the NICU as it gives an infant temporary comfort and hydration but it is not adequate nourishment. **Be aware** that a different approach to feeding has occurred for some newborns with trisomy 18 or trisomy 13, as seen in the following quote.

*"I was pumping breast milk but the hospital care givers were only giving our baby minimal feeds of sucrose (sugar water) for comfort. **I was shocked** when they informed us that some parents don't feed these babies. A nurse went to bat for us and finally our baby began to receive breast milk via a nasogastric tube." S.C., Simon, trisomy 18 [Global Genes advertisement (2014) Our "Labeled" Child's Name is Simon: Fighting for Treatment by Sheryl Crosier]*

A legislative bill called "**Simon's Law**", signed by Kansas Governor Brownback took effect July 1, 2017. This bill prevents a healthcare professional from withholding or restricting life-sustaining measures or authorizing a DNR (Do Not Resuscitate) order for a minor without a parent's permission. And if a parent asks, an explanation about a hospital's futility policy must be provided. **As of 2024, Simonslaw.org lists a dozen "Simon safe" states and continues to advocate for Simon's Law to become effective in every state.**



Simon 9/7/2010 – 12/3/2010, Trisomy 18

Simon is the inspiration for:
Simon's Law
I Am Not a Syndrome; My Name Is Simon,
authored by his mother, Sheryl Crosier.
Hello My Name Is Simon
A reflection of surviving siblings love, written
by Samuel and Sean Crosier with Sheryl
Crosier.

Constipation

Constipation refers to difficulty emptying the bowel and it is a common problem for those of all ages born with trisomy 18 or trisomy 13. Constipated stools are described as dry or hard but in the case of trisomy 18 or trisomy 13 may also refer to difficulty emptying the bowel no matter what the consistency of the stool. Abdominal discomfort, a sense of fullness, and appetite decrease can occur with constipation. As a result, an infant tolerates a smaller volume of feeding than usual, thus may be waking frequently because of hunger.

Congenital defects affecting the bowel are present in a small number of babies with trisomy 18 or trisomy 13 [Jones, 2018]. Let the doctor know if your infant or child is having difficulty with bowel movements. If appropriate, referral to a pediatric gastroenterologist for evaluation and testing can be done, to rule out abnormalities of anatomy or function of the bowel. Numerous causes and comorbidities are associated with constipation and findings are significant to care and management.

Daily Routine

Constipation is a **daily concern** for those with trisomy 18 or trisomy 13. Standard advice for anyone with constipation problems is to increase fiber in the diet, increase fluid intake and increase exercise. An increase in fiber adds bulk to the stool but can be counterproductive, if fluid intake is not also increased. Guidance of the pediatric provider or a nutritionist, about formulas, enteral products for tube feedings or diet, plus the amount of free water needed for adequate hydration is important. Further, the advice of a feeding specialist might benefit those who are difficult to feed; supplemental or full tube feeds might be needed for some children.

Preventing constipation with exercise is not achievable in those with full trisomy 18 or trisomy 13, thus use of a laxative is common. There are many kinds of laxatives with different actions that your doctor can advise you about. **MiraLax** (polyethylene glycol 3350 (PEG) without electrolytes) is discussed here as it is considered the **“Gold Standard of Care” for constipation in children**. (personal communication, Dr John C Carey, 2013) MiraLax retains water in the stool and is absorbable in only trace amounts with a very low risk of electrolyte imbalance. [Walia et al., 2013] It is indicated *for occasional constipation of adults* but is prescribed off-label for children. The best studied laxative medication is PEG without electrolytes, for which there is moderate quality evidence for both efficacy and safety. [Ferry, 2013]

Polyethylene glycol (PEG) 3350 came under FDA scrutiny for neuropsychiatric adverse side effects with an FDA decision in 2011 of *no action necessary based on available information*. See the FDA homepage at www.fda.gov. Check with your child’s doctor before starting MiraLax or any laxative. **When discussing MiraLax, let the doctor know if your child has a history of kidney disease, bowel obstruction, or irritable bowel syndrome**. Adverse effects are **dose related** and include diarrhea (10%), bloating or gas (6%) abdominal pain (2%) and should be reported promptly to the doctor. [Ferry GD, 2013]

Pedi-Lax liquid glycerin suppository (formerly Babylax) is another product familiar to SOFT parents. It is designed to *relieve occasional constipation for the 2 to 5 year old; consult with the doctor for those under*

2 years. **Dr. Carey advises all parents to check with the doctor before using Pedi-Lax**, which attracts water into the stool to soften it, promoting a bowel movement usually within 15 minutes to an hour.

"When about 8 months she was prescribed Lactulose, but sometimes, she also needed a Babylax. When older, she was prescribed MiraLax and it worked well for her." A.B., Megan, trisomy 18

"Nothing works forever for Stacy. Medication and techniques needed to be changed every few months." B.V., Stacy, trisomy18

"While a patient of multiple GI doctors, she experienced dangerous and some lasting side effects from increased (doctor ordered) doses of MiraLax." J.S., Giuliana, mosaic trisomy 18

Toileting Skills

Placement on the potty chair, following meals and after sleep, establishes a routine at times that are generally beneficial for potty-training success. One parent, noted below, recently posted in the SOFT Facebook group that her child (age 9 years) indicated a need to go.

"She is put on the toilet at routine times for voiding. She hardly ever wets or has a BM in her diaper." [The SOFT Times, 2003]

"She did not indicate a need to go but responded to praise and routine times for potty use after feedings and sleep with about 80% success!" AB, Megan, trisomy 18

"We got her a potty, where she can sit like a big girl on the toilet and we can proudly say that for the past 3 weeks she is potty trained. She will let us know when she needs to go by a deep humming, growling sound. Then we ask her: "Lunah needs to go poopy?" And she would nod 'yes". When she's done, she would reach towards us." LR, Lunah, trisomy 18, 2024

Impaction

A decrease in the frequency of stooling and a gradual increase in stool retention results in stool drying in the rectum, and constipation. When this happens the retained stool becomes firm, making it more difficult for the infant to push it out and sometimes the stool can become immovable or impacted. It is possible for loose stool to leak around the impaction and the child appears to have diarrhea. Guidance of a physician is needed for treatment of impaction and management of constipation.

Constipation is a frequent complaint accounting for 5% of all pediatric office visits. [Ferry GD. edited by Post TW, (accessed Nov 2014).] Most cases are diagnosed as functional (not caused by abnormality or disease) but occasionally, if chronic, constipation can be a symptom of an underlying problem. If appropriate, x-rays and tests to rule out bowel abnormality, obstruction or disease might be needed.

Numerous gastrointestinal (GI) surgeries are listed in the SOFT surgery registry. For example: bowel obstruction repair or resection, and bowl malrotation repair show a combined total of 28 children with

trisomy 18, and 44 with trisomy 13 (One had malrotation surgery twice at five years apart). Meckel's diverticulum, a congenital anomaly that can cause obstruction, was removed in 13 children with trisomy 18, and one with trisomy 13.

Enemas should only be used with caution, under the guidance of your child's health care provider, as enemas can deplete a baby of electrolytes and alter body fluids in children. This is especially true of tap water enemas. [Marks JW, 2012. Constipation, MedicineNet.com, WebMD]

Laxatives and fiber (*with adequate fluids*) are effective in improving bowel movement frequency unless the constipation is caused by an underlying disorder or a slow GI transit problem. (Slow motility in the large colon allows for more water absorption and drying of stool.) [Ferry GD reprint UptoDate 2014 www.uptodate.com] **When soft stool is achieved it is important to continue daily maintenance, as prescribed, to prevent recurrence of constipation.**

Routine Medical Care, Growth, and Other Themes in Care

Medical care for those with trisomy 18 or trisomy 13 is often complex as multiple health issues are common. Newborns able to be discharged to home from the hospital will need to be under the care of a pediatrician or other health care provider. Preparing parents for the probable loss of their baby is a part of this doctor's job, but it is crucial to also prepare parents for the possibility of being caretakers of a child with disabilities and/or medical complexity.

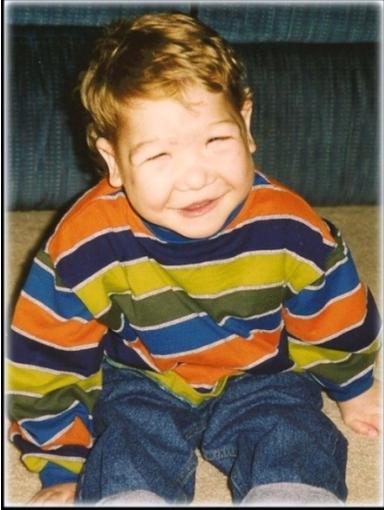
"How can any kind of hope be false, when it's all you've got? Hope cannot be false as far as I am concerned. Hope is what got us through it all." [D.D., SOFT parent, 2013]

"Decisions could not be made on the specifics of his disorder, because, quite frankly, Andrew was writing his own book, rather than following on." [Andrew's pediatrician, *The SOFT times*, 2002]

Keep in mind that even those in palliative care or Hospice might live longer than predicted. A few SOFT families report their child was discharged from Hospice because they were doing better than expected.

Routine medical care and some potential health problems for an infant or child with trisomy 18 or trisomy 13 are addressed in this chapter. Routine well-baby or child visits include measuring and charting growth measurements, discussing and recording the developmental progress of the child, providing immunizations, and screening for potential problems. Entrance to early intervention or public school usually requires a physical, vision and hearing exam, and documentation of up-to-date immunizations.

~



Jonathan
4/3/1996 – 8/27/2006
Partial trisomy 3,5,13 unbalanced translocation

*"Jonathan was a tissue donor. We didn't know until we were asked to donate, that organ or tissue donation was an option for those with trisomy conditions. His helping others gives us comfort."
C.C., 2011*



Arianna
6/19/2006
Trisomy 13

Growth

Baty et al. notes that infants and children with trisomy 18 and trisomy 13 grow slowly and are generally smaller than other children, especially those with trisomy 18. Information follows, from the study by Baty et al., which explains where these newborns plot on a growth curve when using a standard growth chart.

Of 96 infants or children with trisomy 18, the average (mean) birth weight was 4.84 pounds. On a standard growth chart they consistently plot below the lowest centile lines for weight and height (length) except at birth where they had an overlap with standard growth curve lines.

Of 31 infants or children with trisomy 13, the average (mean) birth weight was 5.90 pounds. On a standard growth chart they plot for weight and height (length) with more overlap onto standard growth chart curve lines than trisomy 18.

The head circumference for both conditions shows overlap with the standard growth curve, although the medians are lower.

Natural history of trisomy 18 and trisomy 13: I., Growth, Physical Assessment, Medical Histories, Survival and Recurrence Risk, [Baty et al., 1994]

Plotting the growth of these children on a standard growth chart has problems similar to trying to plot the growth of a premature infant on a standard growth chart. A more accurate assessment of growth of the premature infant is available when using growth charts that compare them to other premature infants.

From information provided by parents and medical records, Baty et al. developed the first charts of growth curves for weight, height and head circumference from birth to 18 years of age for trisomy 18, and birth to 7 years for trisomy 13. Unlike standardized male or female growth charts, these charts represent data combined from both male and female infants and children and provide a means for growth comparison to

others with the same disorder. **See pages 54-56** It is suggested these charts be copied and placed in your child's medical records, for monitoring the growth of your child.

The growth charts are available for download and printing from the SOFT website
<https://trisomy.org/medical-professionals/#/>

The Medical Advisory Board of SOFT is currently facilitating the development of updated growth charts for children with trisomy 18 and trisomy 13.

There is an increase in caloric need for any baby with heart disease, and congenital heart anomalies are present in about 90% of babies with trisomy 18 and 80% of those with trisomy 13. The child's health care provider might recommend consultation with a nutritionist, to determine caloric need and discuss diet. Consideration may be given to using high calorie formula or nutritional supplements added to formula. Guidance of the child's doctor, as well as the nutritionist, is important for children who have difficulty tolerating feedings. Tube feeding, discussed in *Feeding Problems*, allows more control over the amount of fluid and calories consumed.

"He stayed at 19 pounds for a year and then his doctor put him on a supplement to mix in his milk that is simply extra calories. It has really worked." [The SOFT Times, 2004]

"Despite her failure to thrive, she continued to grow in length, stretching to 21 inches at 5 ½ pounds." [The SOFT Times, 2001]

A few older children with these disorders, who are fed some or all of their feedings by tube, have a higher weight centile compared to their height (length) centile.

"I never in my life thought I would have to say Stacy needed to be on a diet." [The SOFT Times, 2007]

"After an extended hospitalization she became less active than we realized. Totally tube fed with no increase in calories, she gradually put on weight in the year that followed. With her cardiac issues, we thought it was fluid retention but the doctor determined it was fat. The nutritionist calculated calories plus free water replacement needed to lose about a pound a month." A.B., Megan, trisomy 18

Puberty

Three young women with trisomy 18, and one with trisomy 13 have (or had) menstrual cycles as reported by their parents for this update. Secondary signs of puberty occurred such as growth of pubic and axillary (underarm) hair and breast development. Menarche (onset of menses or menstruation) spanned 13 years to 18 years in the three with trisomy 18.

Usually, an increase in body fat is needed for any girl to reach puberty. The adolescent female, who is underweight and very small for her age, as is sometimes seen in trisomy 18, might not reach puberty. One young woman, with trisomy 18, who survived 25 years showed no signs of development of puberty.

She plotted on the lowest or below the weight centile curves on the trisomy 18 growth chart, fluctuating between 30 to 37 pounds in the last seven years of her life.

A few parents of older girls report behavior issues such as pinching themselves or others and subdued moods. **“Attitude” is mentioned by a few parents; at all ages.** Following are selected accounts of puberty in two females and one male with trisomy 18, along with one female and one male with trisomy 13.

She was 13 years old, with trisomy 18, and weighed about 60 pounds at menarche. She is now 27 years old and weighs 80 pounds. She has an obvious menstrual flow for several days but the cycles are irregular. At the onset of puberty, she began having seizures and was placed on seizure medication. Her mother reports that her seizures are hormonally induced.

She was 15 years old, with trisomy 18, and weighed 55-60 pounds at menarche. Currently, she is 32 years old and weighs 75 pounds. She has normal flow for 2 days and light flow for another 1-2 days. Her cycles are very irregular, sometimes monthly for 5-6 months, sometimes skipping for 6-7 months. Seizures started when she was 8 years old. Her mother reports that her seizures are not related to hormonal changes.

A young man with trisomy 18, who survived for 21 years, grew a small amount of pubic hair and a few axillary hairs when he was about 18 or 19 years of age but never grew facial hair. Onset of seizures occurred in his late teens and his mother believes the seizures were related to hormone changes that occur with puberty.

*She was 11 years old, with trisomy 13, and weighed about 50 pounds at menarche. She is now 25 years old and weighs 85 pounds. Her cycles are regular. She becomes irritable before her periods. She had a seizure due to high fever at age 4 years and was put on phenobarbital. She stayed seizure free until age 11 years, when she began having monthly seizures. Her mother is not sure if the breakthrough seizures were related to her beginning menses. It was found that her phenobarbital dose needed to be increased and she needed the addition of another seizure medication. **Also, calcium plus vitamin D supplements were started because the long term use of phenobarbital had depleted her bones of these nutrients.***

He was 11 years old, with trisomy 13, and weighed about 90 pounds when he began puberty. He is now 16 years old and weighs 128 pounds. He has a light growth of beard and is shaved. In the first year of puberty his behavior became more agitated but in the past few years that behavior is gone. Acne issues have been a problem for about 3 years but the bigger issue has been the clogging of sweat glands which causes boils. Recently, one was surgically removed. Seizure episodes began when he was younger and are under control with medication. There has been no change in his seizures with puberty.

Vision

Eye malformations commonly occur in trisomy 13 and have a low occurrence in trisomy 18. **It is recommended that an eye exam be done by a pediatric ophthalmologist for all infants with trisomy 13 and when signs and symptoms are present in those with trisomy 18. For both conditions, periodic eye examination is recommended for those over one year of age.** [Carey, 2010]

Dr. Steven D. Cantrell, optometrist, St Louis, MO, reports that many non-verbal vision tests exist that can accurately estimate the prescription for glasses and **parents ought to ask their eye care specialist if their child needs glasses**. Vision problems may be detected that could be corrected with prescription eyewear. Quality of life is enhanced if a child can see faces and expressions, and engage with parents. Steve and his wife, Peggy, had a son, Ryan (10/4/85 - 6/15/86), who was born with trisomy 18.

Cantrell has observed the eye health of children he sees at SOFT conferences annually since 1987. **In recent years he has “become convinced that these children are affected by photosensitive epilepsy to some degree”**. Descriptions of selected ocular problems common to trisomy 13 and seen in some children with trisomy 18, are detailed below, as well as Cantrell’s recent observations in a SOFT newsletter article *Vision, Light Sensitivity, Headaches and Seizures*, [SOFT Times, September 2016].

Photophobia and Photosensitive Epilepsy

Photophobia (painful eye response to bright light) occurs in trisomy 18 from early infancy on. Children born with trisomy 18 can have deficient levels of protective antioxidants, Lutein (L) and Zeaxanthin (Z) which leads to painful eye sensitivity. This is easy to reverse and replenish by adding a nutritional supplement containing lutein and zeaxanthin such as OcuVite or PreserVision, once a day with food or juicing vegetables rich in Lutein and Zeaxanthin added to food such as green leafy vegetables and yellow/orange fruits.

Cantrell reports study findings of some children with trisomy 13 that were diagnosed with Photosensitive Epilepsy (PSE). Spagnoli et al., (2015) <https://www.ncbi.nih.gov/pubmed/25459971>

At www.trisomy.org scroll SOFT’s You Tube Video Channel to watch ***Finding Solutions: Managing Photosensitivity, Seizures, Light Sensitivity & Ocular Health*** [Cantrell, 2021]

The diagnosis of Photosensitivity Epilepsy means that we know what type of epilepsy and what triggers are around the house that could cause a seizure.

Before your child’s next eye exam tell your doctor your child might be sensitive to bright lights shined in their eyes and could cause a seizure. Mention that photosensitive epilepsy is common to trisomy 18 and 13. [Cantrell, 2016]

The Photosensitive Epilepsy seizure triggers are the same in adults and many children born with trisomy 13 and 18.

It’s helpful for parents to know where these triggers are in the home to cover and eliminate.

Where are the triggers?

- Direct sunlight, or sunlight through trees when in a moving car.
- LED bulbs (Big box stores with many banks of florescent tubes and direct LED bulbs.
- Emergency vehicle flashing lights
- Flickering light through ceiling fans, flickering flashing strobe lights
- Video games

How to reduce episodes:

- Tinted car windows and eye protection help reduce episodes.
- Diet may reverse sensitivity but children with Trisomy should always wear dark sunglasses indoors to help prevent seizures.
- Children with trisomy should wear dark eye protection morning to night until the effect of nutritional improvement is seen. Your child's body language will tell you when they are no longer reacting at the same level.
- Your child should avoid all Digital Screens including TV, computer monitors and cell phones. You can reduce screen brightness which reduces the amount of high energy light emitted. FLUX is a free download for computers with variable settings.

Overnight EEG studies indicate that insomnia and an increased risk for night time seizures increases with screen exposure. Eye protection indoors as well as outside will reduce symptoms and increase comfort. [Cantrell 2017]

BluTech outdoor lenses are science based and helpful. They should be available everywhere but are difficult to find. BluTech: www.BluTechLenses.com or call for a local source at 800-258-5902

Several years ago Cantrell looked into the possibility of a blue light protective contact lens being developed for children with trisomy 18 and 13. Now in 2024 on a trial basis, there are 2 or 3 young children from SOFT with trisomy 13 wearing blue light protective contacts, and also taking supplements of the antioxidants Lutein and Zeaxanthin to aid in decreasing visual triggers of photosensitivity. Results for these children has been positive in preventing or decreasing seizures. The lens was created by Cantrell.

Cantrell was contacted about this "notch filtering contact lens" by Dr Thiele, pediatric neurologist, of Boston, MA. She works with children who develop a rare seizure disorder called Sunflower Syndrome. Dr Thiele headed an institutional review board study of these lenses at Massachusetts General that found these contact lenses to be "*positive for efficacy*". Dr Thiele plans to submit this lens to the FDA for clinical trials as a new drug to allow insurance to cover the cost.

Cantrell recommends a daily consumption of lutein and zeaxanthin to aid in decreasing visual triggers of photosensitivity (see above). Vegetables such as spinach, kale, and collard greens and corn are high in these antioxidants. One SOFT mom juiced these greens for her daughter and her photosensitivity went away. Nutritional supplements, FloraGlo lutein 10-12mg and zeaxanthin 2 mg, ensure a proper daily dose. These supplements are non-toxic and our children cannot overdose. As mentioned above, **daily supplements of lutein and zeaxanthin can be given using Preservision or OcuVite in a G-tube.** [Cantrell S, Slide presentation to the Sunflower Syndrome Families, [2024]

Lutein and zeaxanthin now have been found in the placenta with a 3rd trimester transmission when the placenta has matured and grown to adapt to the increased nutritional needs of the fetus and are important for brain function. Moran Eye Center study using mice, 2023 <https://doi.org/10.1167/iovs.64.4.9> Lutein and zeaxanthin are important during pregnancy while carrying a child with Trisomy 18 or 13. [Cantrell, 2024]

Benefits for mother and child of Lutein and Zeaxanthin supplements were found in the Moran Eye Center's recent study of 47 expectant mothers in *Ophthalmology Science* [2024]. <https://www.sciencedirect.com/science/article/pii/S2666914524000733>

Cataracts

Cataracts are cloudy, rather than clear, crystalline lenses of the eye. **The incidence of cataracts is greater in trisomy 13, than in trisomy 18 and those with trisomy 13 are more likely to have cataracts at birth.** Cataract removal is the most frequent eye surgery reported to SOFT for an infant or child with trisomy 13. Those with trisomy 18 are less likely to be born with cataracts but develop them with age, possibly due to antioxidant deficiencies, which cause light sensitivity and are needed to protect the eye from ultraviolet damage.

A diet rich in the antioxidants lutein and zeaxanthin, either from nutritional supplements or vegetables such as spinach, kale, and collard greens naturally reverses light sensitivity and cataracts. Our body does not make lutein and zeaxanthin. Only dietary lutein and zeaxanthin are deposited in the macula at the center of the retina in the back of the eye. The macula contains rods and cones which are light sensing receptors in the eye. Macular pigment filters light internally.

Cataract surgery replaces the cloudy lens with a clear intraocular implant. Prior to surgery, consultation with a cardiologist about risk versus benefit of surgery should be done for those with congenital heart defects.

Strabismus

One of the common forms of strabismus is esotropia (crossed eyes) which often appears in infants or very young children. As noted in Central Nervous System Problems, strabismus is a problem in those who have abnormal muscle tone. The most frequent eye repair in the SOFT surgery registry for trisomy 18 is strabismus surgery; reported 6 times more for trisomy 18, than trisomy 13.

"Uncorrected high prescription hyperopia (difficulty focusing on nearby objects and easier to see objects far away) may trigger crossed eyes. Infants are more likely to be Far Sighted or Hyperopic which when trying to focus will often cause a crossed eye that might disappear with glasses. Sometimes surgery is needed but it is a good idea to determine if glasses are needed first." (personal communication, Dr Steven Cantrell, 2017)

Strabismus surgery is done for a medical reason, to align both eyes, and also has positive cosmetic results. The eye surgeon must consult with the child's cardiologist and the benefit versus risk should be discussed prior to any surgery for those who have cardiac anomalies. Some parents choose to not do surgery. Covering the better seeing-eye with a patch to improve vision in the weaker eye has been done for some SOFT children who have strabismus, and corrective lenses have also been prescribed.

"Esotropia (eye turned inward) developed in her early months. When she lived past her first birthday, the ophthalmologist said she was a "survivor" and did strabismus repair on both eyes. He cautioned that another form of strabismus, exotropia (eye turned outward), could result from the repair which did happen in one eye about 4 months after her surgery." A.B., Megan, trisomy 18

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Glaucoma

Glaucoma is an eye condition in which fluid builds up inside the eye, increasing pressure on the optic nerve. **Some infants with trisomy 13 develop congenital or early onset glaucoma resulting in increased intraocular tension and could be a reason for unexplained irritability.** [Carey, 2005] Treatment with topical drops to maintain optimal pressure is common for anyone with glaucoma. The SOFT surgery registry lists glaucoma surgery for 10 children with trisomy 13; 1 child with trisomy 18.

Eye Irritation

Often these children **do not fully close** their eyelids when sleeping. The exposed surface of the eye becomes **dry and irritated**. Long eyelashes along with **incomplete lid closure** can cause a red, itchy eye condition called **Blepharitis**. Prescribed antibiotic/steroid drops or ointment treats blepharitis

Many particles in the air like dust, mold, pollen and sand can get into our eyes and harm them. Eyelashes help to sweep these particles out of the way. Children with trisomy have very long eye lashes that catch and hold everything contributing to red goopy eyes.

- Cleansing over and around eyelashes with **OcuSoft Infant Lid Scrubs**, washes away debris.
- Hydrating eye drops are also needed daily. Cantrell suggests **Natural Ophthalmics Ortho K** thin eye drops 3x per day and Ortho K thick at bedtime.

OcuSoft Infant premoistened lid scrubs are available in drug stores without prescription.

Natural Ophthalmics products: Ortho K thin eye drops and Ortho K Thick eye drops are available through a doctor's office. Call 1-877-220-970 for an office near your home. [Steven Cantrell, OD, 2016]

Cryo-Epilation or cryotherapy of a lash line was done to permanently remove eye lashes that were scratching the sclera (the white outer area of the eyeball) for a young woman with trisomy 18.

"My recommendation to families would be to skip electrolysis and entropion correction altogether, and go straight to cryotherapy. The results were almost instant improvement." GM, mother of Leilani, 2024

Table 6 Possible Eye Conditions Seen in Trisomy 18, Trisomy 13, or in Any Child

| Eye Conditions |
|--|
| Amblyopia One eye develops less vision than the other and can cause crossed eyes. |
| Astigmatism Either nearsighted or farsighted and can affect both far and near vision. |
| Blepharitis (irritated eyelids which may be painful) May be goopy and stuck shut in the AM with dandruff like appearance. Treat with lid scrubs, warm washcloth, and hydrating eye drops or may require prescription drops and ointment.) |
| Cataracts (common with trisomy 13, the clear lens within the eye is cloudy in one or both eyes, surgery may not be necessary if one eye sees well) There may be problems getting an intraocular lens implant in the size needed |

for a child.

Corneal Opacities (blotchy spots throughout the cornea which may interfere with vision) Often harmless and does not require therapy. Consult with physician for possible treatment.

Dry eyes (eyes not tearing sufficiently) Can cause an infection, treat with artificial tears or ointment or placement of a tear duct plug may help.

Epicanthal Folds (extra fold of skin next to nose) Harmless but can give the appearance of a crossed eye.

Glaucoma (elevated intraocular pressure) Treated with doctor prescribed drops and/or surgery may be needed.

Hyperopia (far sightedness, difficulty seeing close) Uncorrected may cause strabismus. Treat with corrective lenses.

Incomplete Lid Closure Eye lids do not close completely while sleeping. May be painful and exposure will cause red, watery eyes. Treat with artificial tears, lid scrubs, ointment at bedtime, or surgery may improve closure.

Intraocular Cartilage (connective tissue dispersed randomly throughout the eye) This may interfere with normal vision but no treatment is necessary.

Iris Coloboma (incomplete closure of the round colored iris) May resemble a keyhole and allows too much light to enter eye, increasing light sensitivity)

Jaw-Wink Reflex (eyes roll back when nursing, taking a bottle or eating) is harmless.

Long eye lashes Usually does not cause any problem but when eye lashes turn inward may scratch the eye causing continually watery red, irritated eyes and infection. Also may cause discomfort if lashes touch glasses.

Microphthalmus (one eye is smaller (underdeveloped) in size than the other) Vision may be normal in the smaller eye but usually is not and may be blind.

Myopia (near sightedness, difficulty seeing far) Treated with corrective lenses

Nystagmus (involuntary eye movements) Corrective lens may help

Optic Nerve Hypoplasia (underdeveloped optic nerve) May interfere with normal vision. No treatment necessary.

Photophobia (extreme light sensitivity, very common in trisomy 18) Treat with Blutech sunglasses, hats, tinted auto windows and extra Lutein & Zeaxanthin through diet or supplement. Go to www.BluTechLenses.com or call 800-258-5902

Ptosis (upper eyelid droops and is lower than the other) Does not require surgery unless interferes with vision.

Strabismus (muscle imbalance, one eye is not straight, turns in most often or out and may alternate from one eye to the other) Glasses can correct or surgery may be required.

[Cantrell, SOFT Times 2001, updated 2017]

Hearing

Most states in the US. require mandatory screening of hearing in a newborn. Infants who do not pass screening receive referrals for follow up testing. In countries or regions where universal newborn screening of hearing does not occur, referral to a hearing specialist in the first weeks of life is important. Testing with a pediatric audiologist would be best for those who are difficult to evaluate.

"The audiologist said that much of the transmission gets lost in her ear canals because they are so incredibly tiny. But as Lilly grows, hopefully her ear canals will grow. And then she will hear better." LH, Pray for Lilly Blog, 2011

In trisomy 18 and trisomy 13, a hearing evaluation before 6 months of age performed by a trained pediatric audiologist is indicated [Carey, 2010]. Parents are usually aware of responses that indicate their infant can hear sounds. Their observations offer valuable input to the evaluation of their child's hearing.

Brain Stem Auditory Evoked Response (BAER) or Auditory Brain (stem) Response (ABR) measures brain wave response to sounds heard through earphones. Electrodes are placed on the scalp and sedation will likely be needed to keep a child still. Another test called a behavioral response hearing test can usually be done on children older than one year and no sedation is needed. This test is done in a quiet sound room and the children are evaluated by their response such as a change in breathing pattern or eye movement to tones or other sounds introduced by the audiologist. Sometimes the initial hearing test and a later follow-up test show different results. Moderate to severe hearing loss has been reported in some older children in trisomy 18 and trisomy 13.

A few SOFT Facebook moms report their child uses Soft Bands (**Baha and Pronto Soft**), a head band containing hearing devices. **Cochlear Implant** surgery was reported to SOFT for 2 children with trisomy 13, 1 child with mosaic trisomy 13 and 1 child with trisomy 18.

Table 7 Sensory Impairments in Trisomy 18, Trisomy 13, and Mosaic or Partial Trisomy 18 or 13

| |
|--|
| <p>Hearing</p> <p>Trisomy 18 (of 37 children) Hearing was fine in nearly 50% of those with trisomy 18. Nearly 50% had partial hearing and a few had profound hearing loss.</p> <p>Trisomy 13 (of 12 children) Hearing was fine in 66% of those with trisomy 13. 16.5% had partial hearing and 16.5% had profound hearing loss.</p> <p>Mosaic or Partial trisomy 18 or 13 (of 20 children) 25% had partial hearing and a few had profound hearing loss.</p> <hr/> <p>Vision</p> <p>Trisomy 18 (of 37 children) Vision was fine in nearly 66% of those with trisomy 18. More than 25% had partial sight and a few were legally blind.</p> <p>Trisomy 13 (of 12 children) Vision was a greater problem for those with trisomy 13. 33% had partial vision, 25% were legally blind, and 25% were totally blind.</p> <p>Mosaic or Partial Trisomy 18 or 13 (of 20 children) Vision was fine in nearly 75% of those with mosaic or partial trisomy.</p> |
|--|

A few were partially sighted, and few more were legally blind.

Social Development of Children with Trisomy 18 and Trisomy 13 in the Context of Family and Community [Healey, P.J., 2003]

"Jack received hearing aids to try out for 30 days but we haven't seen any reaction to noise yet." [The SOFT Times, 2003]

"She has hearing in each ear and unit conductive loss in each ear, moderate to severe. Hearing aids are expensive and our insurance doesn't cover them." [The SOFT Times, 2004]

*"Ella finally got her hearing aids after **Easter Seals** and our audiologist agreed on what was causing her profound hearing loss. We notice she is aware of sound when wearing them." [The SOFT Times, 2005]*

Immunizations

The American Academy of Pediatrics (AAP) has firm guidelines regarding immunizations for infants and children with neurological problems. The healthcare provider ought to refer to the AAP recommendations. Medicaid has a list of required immunizations for their recipients that follows AAP recommendations.

"We know that vaccines are critically important for all babies. There is strong evidence they prevent routine illness and life threatening illness, and that is no different for premature babies." Dr Therese Grover, NICU medical director, Anschutz Medical Campus, Aurora, CO.

See more from Dr Grover in this parent guide, ***Vaccines for Babies with Medical Conditions*** at <https://www.childrenscolorado.org/conditions-and-advice/parenting/parenting-articles/vaccines-babies-medical-conditions/>

Immunizations should be determined on an individual basis. [Bruns, 2014] Some children may be exempt due to religious reasons or adverse reaction to a prior vaccine or need a delay due to illness or other concerns. A few SOFT mothers have voiced concerns about their baby's low weight (<5 pounds) and a vaccine reaction. It is important for a parent to talk with their child's healthcare provider if they have any concerns about vaccines.

"Reported side effects or (as stated in some papers) 'adverse reactions; are observations. These events do not show or prove cause and effect." [John C Carey, personal communication, 2024]

Immunization records of the children in the study by Baty et al., were looked at, and no evidence was found for an increase in adverse reactions to immunizations in children with trisomy 18 and trisomy 13, compared to chromosomally normal children, although the numbers studied are small [Baty et al.,1994].

The SOFT Immunization Registry maintains information provided by parents about their child's vaccine history. 863 children had their vaccinations reported to SOFT as of January 2024. 552 of these are children with a full trisomy 18 or 13 diagnosis.

Of the 863 vaccinated 90.7% were reported as being up-to-date for their age. 58% received 1 or more Flu vaccine, and 58.6% received 1 or more of the Synagis antibody injections to prevent RSV.

But the name of a vaccine or antibody related to an event was reported by only 30 parents. Synagis and Flu were each named 5 times, followed by Varicella and Covid each named 4 times. DTaP was named 3 times and other vaccines 1-2 times.

53.4% of events were typical: fever, tiredness, site redness or lump, rash post Varicella or MMR and some indicated an event occurred but did not specify it, totaling 47 children (5.4% of all children vaccinated).

46.6% had adverse events following the vaccine: seizure 4; apnea 5; breathing episode 4; need of O2 increase 4; illness 18 or cellulitis 2, totaling 41 children (4.7% of all children vaccinated). As mentioned above, having an event after the vaccine does not mean the vaccine caused the event,

The total number of typical and adverse events reported, following a vaccine, is 88 (10.2% of the 863 children vaccinated).

Percentages of all typical and adverse events, by diagnosis were greater for T18 and mosaic T18, compared to T13 and mosaic T13.

Number of children vaccinated and number of all reactions reported by diagnosis:

379 children with T18- events reported = 49 (12.9%)

63 children with mosaic T18- events reported = 7 (11.1%)

173 children with T13- events reported = 13 (7.5%)

43 children with mosaic T13- events reported = 3 (6.9%)

205 children with other Related Disorders- events reported = 16 (7.8%)

The total of all diagnoses with a report of seizure post vaccination is 12.

Seizure after vaccine by diagnosis: 3 with T18, 4 with T13, 1 with mosaic T13, 4 with Related Disorders

A parent in Japan joined SOFT USA in 2014 and reported that her infant with trisomy 18 weighed 3lbs 4 oz at birth. At 6 weeks of age her infant experienced an event to an unspecified immunization with "fever, twitch and died 24 hours later". It is unknown to SOFT if this sad event was caused by the vaccine. **"It is known in the medication side effect literature that about 70% of reported side effects turn out to not be related to the medication."** [John C Carey, personal communication, 2024]

A small number of parents from 18 nations outside the US are members of SOFT USA. Some provided their child's immunization information on their registration, along with reactions they observed, if any. They are included in the *SOFT Immunization Summary* above. Below lists their country, the vaccine named and diagnoses.

1 child with T18 in Indonesia (BCG for Tuberculosis)

2 children with T18 and 1 child with mosaic T18 in South Africa (BCG, multi-vaccines, unknown)

2 children with T18 from France (BESCRO for meningitis B, unknown)

1 child with T18 from Japan (unknown)

1 child with T13 from Italy (H1N1 Flu)

Vaccine schedules are not exactly the same from country to country. Disease prevalence needing public health intervention with a vaccine for prevention is also not the same globally.

It is important for a parent to talk with their child's healthcare provider if they have any concerns about vaccines.

Covid-19 immunizations are included in the above numbers of vaccinations and reactions.

The number of children who received Covid-19 vaccination = 48. The 2 youngest, born in 2022, have T13. Recipients diagnoses: T18 = 20; T13 = 8; mosaic T18 = 7; mosaic T13 = 1; Related Disorders = 12

Reactions to Covid-19 vaccination are reported for 8 children or 16.6% of the 48 received. Half had typical events. Tiredness being the most common reported following the vaccination. Adverse events reported for the other 4 follow:

1 child with T13 was irritable, unwell x 2 days post the 1st Moderna vaccine and had diarrhea post the 2nd.

1 child with T18 was lethargic post the Pfizer vaccine and 5 days after was ill for 24 hours with fever.

1 child with mosaic T18 became ill with Covid post receiving the Johnson & Johnson booster.

1 child with partial T18 had 4 seizures, 3 hours post the 2nd dose of Pfizer.

All of these reported events resolved.

RSV

Respiratory syncytial virus (RSV) is a serious lung infection for premature infants and infants with chronic lung disease. For older children and adults it usually results in a cold or upper respiratory illness but an older child with trisomy 18 was hospitalized with RSV. RSV occurs during the winter and early spring as an annual epidemic.

RSV is easily spread by direct contact with infectious secretions on the hands, by droplets in the air, and the virus can survive for hours on surfaces such as countertops. Preventive measures would be to limit exposure to child care centers, if at all possible, and to encourage good hand washing at home and in any setting where you take your child. [Showalter, *The SOFT Times*, 2004-2005] Dr. Scott Showalter, pediatrician, has attended SOFT conferences annually since 1987 to attend to the children who might become ill at the conferences. Scott and his wife, Vivian, had a son, Patrick (2/6/87 – 4/18/87), who was born with trisomy 18.

Antibodies to prevent RSV can be given by an intramuscular injection of Synagis® (Palivizumab).

Synagis® is started prior to the beginning of the RSV season, and given once a month until the end of the season. The season may vary in different parts of the country. There are specific guidelines for who is eligible to receive Synagis®. It is only for those infants and children under two years of age who are considered to be at increased risk for RSV. Chronic lung disease or congestive heart failure requiring treatment or pulmonary hypertension may qualify infants to be considered at an increased risk. It is appropriate for families of children with trisomy 18 and trisomy 13 under the age of 2 years to discuss with their child's doctor the option of their child receiving these antibodies to prevent RSV and to ask if their child might still qualify the following year. This will sometime be approved by health insurance with a letter from your doctor.

Musculoskeletal system

A variety of muscle and skeletal abnormalities occur in trisomy 18 and trisomy 13, including medically significant malformations and minor anomalies of limb and skeleton. [Carey, 2010] (See Tables 1 and 2 on pages 18-19.) If appropriate, the child's doctor can make a referral to a **pediatric orthopedic** doctor for evaluation of anomalies that might need casting or surgical intervention. X-rays allow for diagnosis of degree of involvement and help in the decision as to what will be in the best interest of the child. Some parents choose to not intervene, others defer interventions until their child is older and some choose correction. If a child has a cardiac defect, surgical decisions require consultation from the child's cardiologist about benefits versus risks of surgery.

Orthopedic, Scoliosis and Risk of Fracture

Curvature of the spine (scoliosis) is a potential problem for older children with trisomy 18 and may also occur in trisomy 13. **Scoliosis should be evaluated clinically at routine health supervision visits in children with trisomy 18 starting at age two years. Usually, over time, a series of x-rays of the spine are done when scoliosis is suspected. [Carey, 2010]** If curvature progresses >40 degrees, surgery is usually discussed. Progression to a more severe curvature, gradually diminishes lung capacity leading to long term heart and lung complications.

The early stage of scoliosis can be seen by looking at the child's back for misalignment of hips or shifting of the spine. Parents should ask their child's doctor to check for these problems. If appropriate, the doctor can refer the child to a pediatric orthopedic specialist for examination and x-rays to determine the type of scoliosis, to measure the degree of curvature and to develop a plan of care. In the recent past curvature > 20 degrees was treated with a custom fit orthopedic brace called a body jacket to delay or arrest progression, sometimes worn for years with refitting for growth. Surgeons prefer waiting to do fusion surgery until a child is 10 years or older as a fixed fusion can stunt growth in a younger child.

In recent years parents began to report a treatment for scoliosis that allows the child to continue to grow. Adjustable Growing Rods are a surgical treatment for scoliosis in children who have not reached their skeletal maturity. The rods are attached to the spine above and below the curve with screws, and then lengthened during follow-up procedures every six to 12 months. This allows the spine to continue growing while managing the curve until the child is old enough for spinal fusion, which is typically around age 10 for girls and age 12 or 13 for boys."

There are several types of growing rods, including traditional growing rods, Vertical Expandable Prosthetic Titanium Ribs (VEPTR), and MAGnetic Expansion Control (MAGEC) rods. The type of rod used depends on the curve type and any other underlying conditions that are contributing to the scoliosis.

When a child has a congenital heart defect the orthopedic surgeon should consult with the pediatric cardiologist when surgery is considered. Parents need to be informed of risks versus benefits.

"She had a Boston brace for many years; when it stopped being effective VEPTRs were installed and she went in every 4 months for 5 years to have them expanded. We just had the VEPTRs removed and the spinal fusion done." J.V., Elanor, trisomy 18, 2012

Risk of Fracture

Older children with trisomy 18 and trisomy 13 have been diagnosed with osteopenia or osteoporosis. Osteopenia refers to bone mineral density (BMD) lower than the normal but not low enough to be classified as osteoporosis. Osteoporosis refers to a very low BMD. Decreased bone density is an increased risk for fracture. Testing and treatment for these conditions is available.

Weaver and colleagues at the Omaha Children's Hospital reported fractures in 13 of 29 children (45%) with trisomy 18 at their hospital from a record review of hospital encounters involving children of all ages. The authors considered this to be due to an intrinsic predisposition to osteopenia and lack of mobility.

One young woman with trisomy 18 receives Zometa® for osteoporosis, once a year by IV infusion. She cannot tolerate the oral medication. **Always ask how a medication should be given and about possible side effects**

A number of parents report that their child suffered a broken bone as a result of being picked up or from a fall. Lack of activity needed to develop strong bones, inadequate calcium and vitamin D intake, and side effects of some medications increase the incidence of fracture.

*"CPR on him caused multiple fractures and one needed surgical repair and casting. He started fracturing bones at 11 ½ years and had 12 fractures in the last 9 months. We are seeing an **endocrinologist**." (later **he was treated with bisphosphonates**) J.W., Nicholas, trisomy 13, 2007*

Genitourinary

A variety of defects of the genital and urinary tracts have been described in those with trisomy 18 or trisomy 13. Urinary tract infections (UTIs) occur with frequency in both disorders. Symptoms such as frequent voiding of small amounts of urine, discomfort when voiding, strong smelling urine, and fever should be reported to the doctor. **An adverse effect of constipation is urinary tract infection, especially for girls.** [Dr Liptak, SOFT conference workshop, 1999]

Because of the high frequency of renal defects, abdominal ultrasound is recommended in those with trisomy 18 and trisomy 13. Those with significant renal defects should be followed for infection and renal insufficiency. [Carey, 2010] See page 65 about screening those with trisomy 18 for Wilms (kidney tumor).

"We're told the probable cause of the kidney stone was Lasix®. She took it only for a month in infancy." JD, Giuliana, mosaic trisomy 18

"He had another urinary tract infection. The cause is a diverticular pouch on his bladder that traps urine. So, now he is on a daily maintenance dose of antibiotic." [The SOFT Times, 2007]

Dental

The American Academy of Pediatric Dentistry (AAPD) recommends the first visit to the dentist be no later than age 12 months or within 6 months of the infant's first erupted tooth. Bacterial infection related to developing dental cavities can be acquired at a very young age and guidance about **preventive** dental care is important. Usually those who have congenital heart disease will need antibiotic protection given prior to dental care procedures. Dental care is essential as cavities and gum disease can lead to infection in the blood stream, which might affect the heart. Parents can ask their child's doctor for referral to a pediatric dentist. If appropriate, the child's doctor or dentist can prescribe a prophylactic antibiotic to use prior to dental exam and care.

The many reminders of poor prognosis might delay a decision to start dental care. Oral hygiene is necessary. A pediatric dentist can give helpful advice on how to clean your child's teeth. Children with trisomy 18 or trisomy 13 are often orally defensive, resistant to anything in their mouth except their own fingers. Brushing their teeth can be a challenge. One SOFT parent recommends a dental product called Biotene®, a gel-toothpaste that is tolerated well by her daughter because it is low foaming. Biotene® contains enzymes to help reduce symptoms of dry mouth. **Some medications have side effects of dry mouth or overgrowth of gum tissue.**

A number of SOFT parents report their child had a dental procedure done in a hospital under anesthesia. The department of social services in a state will have a list of dentists who accept Medicaid. Dental visits are every 6 months for cleaning and check-up. Parents should check with the dental billing office about reimbursement.

Safety

Taking an adult, and an infant and child **cardiopulmonary resuscitation (CPR)** class and learning what to do in an emergency will benefit the whole family. Contact the local fire department and/or emergency transportation to let them know there is a child with special health care needs living in your home. This will help them find your home more quickly, if ever needed.

"I am one proud mama listening and watching my boys describe/demonstrate how to do CPR. Thank you to the American Heart Association for free family day!" T.G., SOFT Facebook, 2014

Children with trisomy 18 or trisomy 13 see many specialists. They may need tube feedings, oxygen, medications, wheel chair transportation and more during a trip to the doctor. The caretaker has many details to remember and must also inform the doctor about her child's history and the reason for the visit.

Each specialist will ask for a medical history, list of surgeries done, medications with strength and dosage that the child receives and if the child has any allergies. Record and keep this information up to date with changes, to take to clinic or hospital visits to assure all providers are given the same information.

Today, a parent can provide such information prior to the visit, at facilities and offices with eCheck-In. Some facilities also provide a secure online portal allowing parents access to their child's health information, communication with the health provider, and to manage appointments and view tests.

If a facility does not provide parents access to their child's electronic records, ask each professional treating your child to briefly summarize their findings and recommendations in a log book. A tech savvy SOFT member recommends **an app called Genius Scan** to keep track of medical history. The app turns your phone into a scanner and files into PDF's. You can store them in the app as well as export them to the cloud or email.

"My biggest fear is that she would be treated differently because she is 'special'; that the doctors would base their decisions on her trisomy 18 and prognosis." [the SOFT touch, 1992]

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**Megan**  
**5/15/1980**  
**Trisomy 18**

Megan is one of the 10% of those with Trisomy 18 born with a normal heart.

*"Trisomy 18 and 13 were talked about at birth. Blood work came back with Trisomy 9. However, Greta's diagnosis was changed to SLO when she was 21 years old."  
M.T., 2008*



**Greta**  
**7/15/1980**  
**Smith Lemli/Opitz (SLO)**

SLO, sometimes confused with Trisomy 13, is related as a condition with many similar problems.



**Ryan**  
**12/23/1999**  
**Trisomy 18 Mosaicism**

*"We take it one day at a time with Ryan and thank God for each day we have".  
M.S., **SOFT Family Stories***



**Kameron "Kammie"**  
**5/8/1997**  
**Trisomy 18**

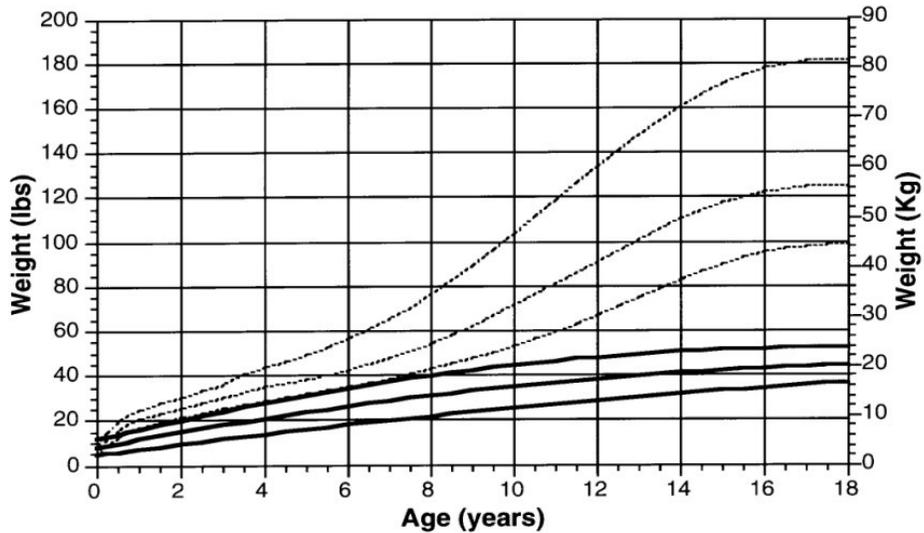


**Emerson**  
**6/23/2011**  
**partial Trisomy 18q and partial Monosomy 13p translocation**

**Table 8a Growth Curves for Trisomy 18 and 13**

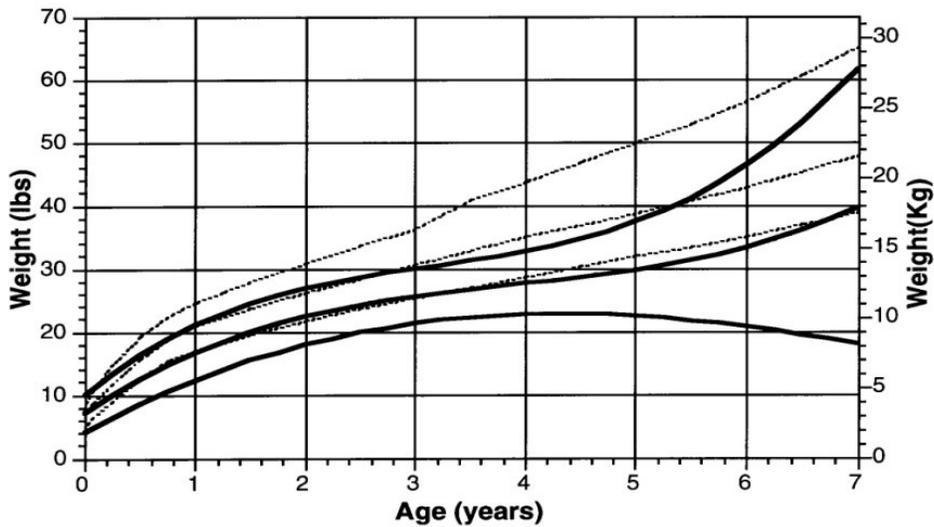
**Trisomy 18 Weight Curve**

Regression and 95% confidence limits  
(trisomy 18 = solid lines: normal = dotted lines)



**Trisomy 13 Weight Curve**

Regression and 95% confidence limits  
(trisomy 13 = solid lines: normal = dotted lines)

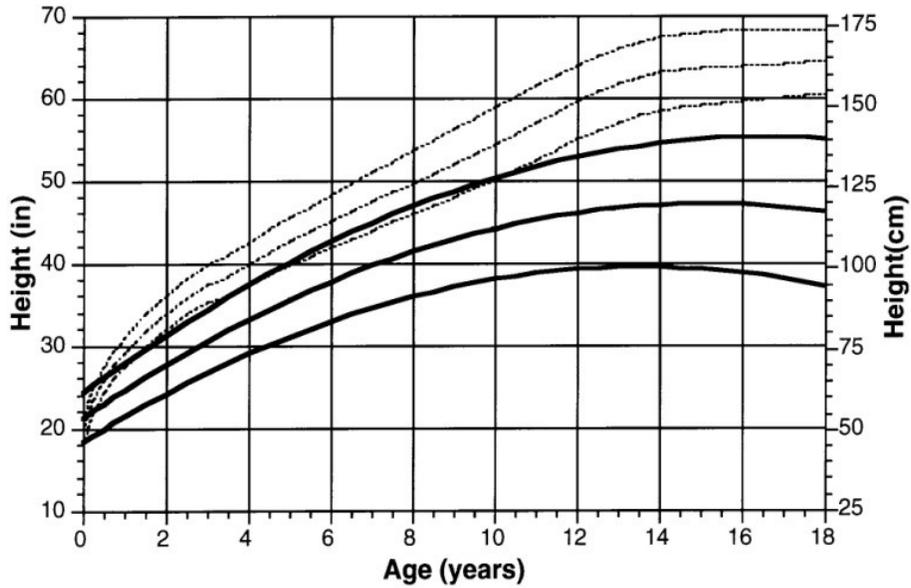


**Source for weight curves:** Baty BJ, Blackburn BL, Carey JC (1994) Natural history of trisomy 18 and trisomy 13: I. Growth, physical assessment, medical histories, survival and recurrence risk. *Am J Med Genet* 49: 175-188, © 1994 Wiley-Liss, Inc. Reprinted by permission of John Wiley & Sons, Inc.

**Table 8b Growth Curves for Trisomy 18 and 13**

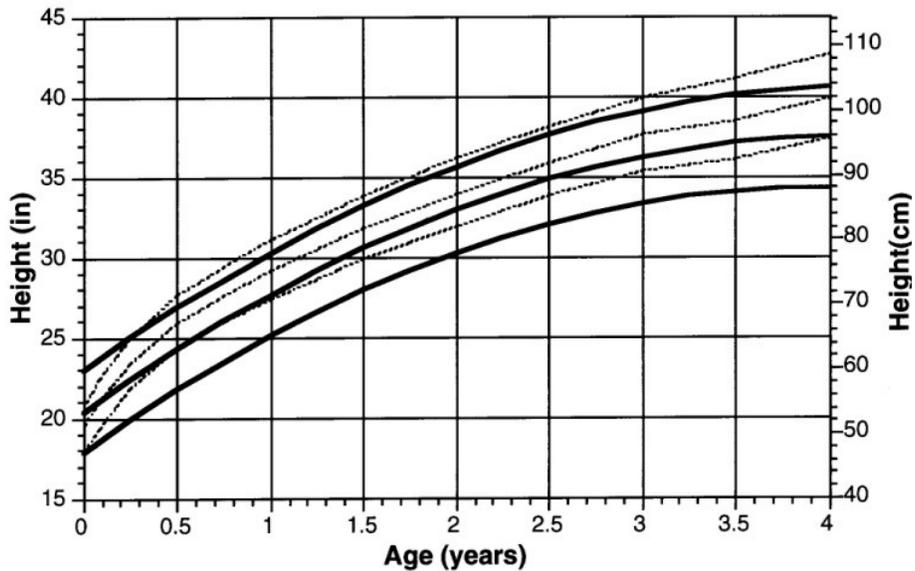
**Trisomy 18 Height Curve**

Regression and 95% confidence limits  
(trisomy 18 = solid lines: normal = dotted lines)



**Trisomy 13 Height Curve**

Regression and 95% confidence limits  
(trisomy 13 = solid lines: normal = dotted lines)

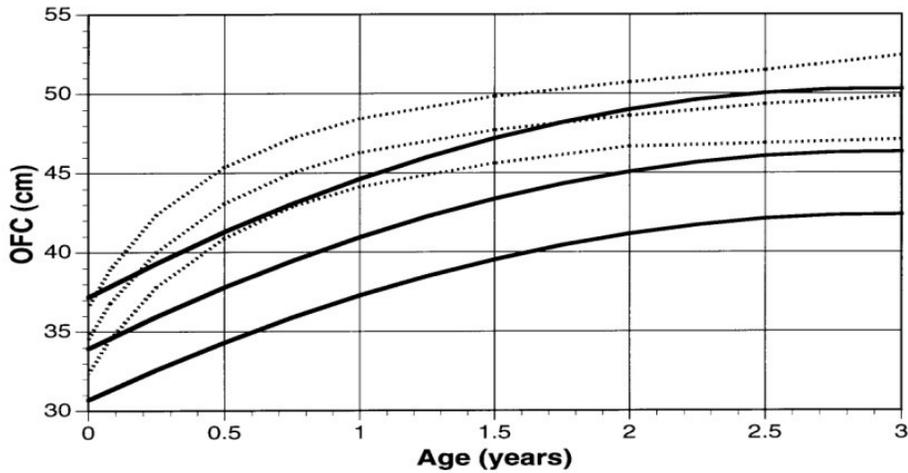


**Source for height curves:** Baty BJ, Blackburn BL, Carey JC (1994) Natural history of trisomy 18 and trisomy 13: I. Growth, physical assessment, medical histories, survival and recurrence risk. *Am J Med Genet* 49: 175-188, © 1994 Wiley-Liss, Inc. reprinted by permission of John Wiley & Sons, Inc.

**Table 8c Growth Curves for Trisomy 18 and 13**

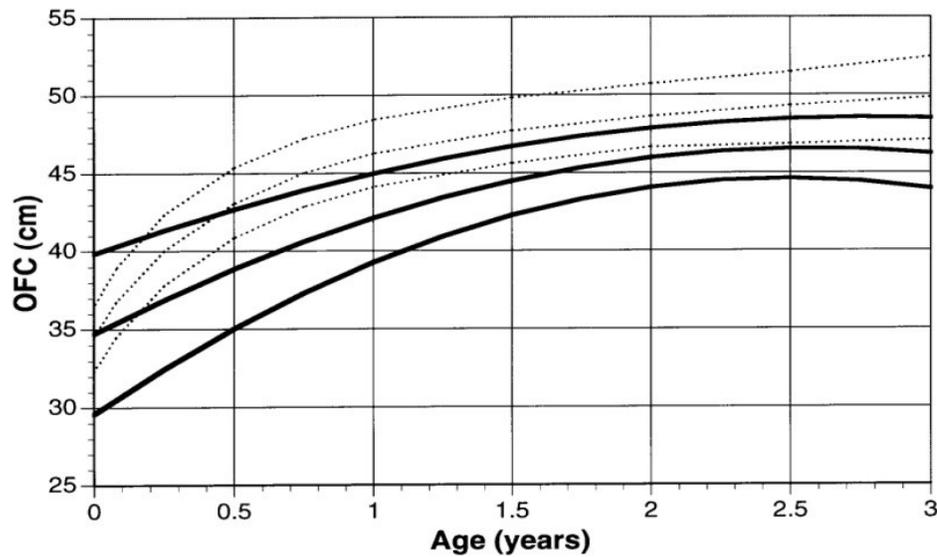
**Trisomy 18 Head Circumference Curve**

Regression and 95% confidence limits  
(trisomy 18 = solid lines; normal = dotted lines)



**Trisomy 13 Head Circumference Curve**

Regression and 95% confidence limits  
(trisomy 13 = solid lines; normal = dotted lines)



**Source for head circumference curves:** Baty BJ, Blackburn BL, Carey JC (1994) Natural history of trisomy 18 and trisomy 13: I. Growth, physical assessment, medical histories, survival and recurrence risk. *Am J Med Genet* 49: 175-188, © 1994 Wiley-Liss, Inc. Reprinted by permission of John Wiley & Sons, Inc.



**Lyndsay**  
**7/20/2000**  
**Trisomy 18**

*"Dr Carey told us Lyndsay's heart repair was done at the youngest age, known to him, for an infant with trisomy 18."*  
*L.S., 2014*

Lyndsay had cardiac surgery for a large VSD, small ASD and PDA when 2 weeks old and weighing only 4 pounds and 3 ounces.



**Leila**  
**12/28/2011**  
**Trisomy 18**

Leila had a tracheostomy at 2 months old and cardiac repair of TOF at 9 months.



**Nora**  
**2/9/13**  
**Trisomy 13**

Nora had ASD and VSD cardiac repairs at 8 months of age.



**Milan**  
**11/2/2010**  
**Trisomy 18**

Milan had VSD repair at age 2 months and 3 weeks.



**Alexander**  
**1/10/2014 - 3/8/2014**  
**Trisomy 13,**  
**Robertsonian translocation**

*"Should we do surgery? Can I put him through that? What if he dies? Will I ever forgive myself? What kind of quality of life will he have if we don't do surgery?" [The SOFT Times, 2001]*

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**Table 9 Cardiac Surgery**

| <b>The SOFT Surgery Registry</b>                                                            |            | <b>(Spans 3 decades + to 2024)</b> |            |             |           |              |
|---------------------------------------------------------------------------------------------|------------|------------------------------------|------------|-------------|-----------|--------------|
| <b>Name of Surgery or Procedure &amp; Number Done, by Diagnosis</b>                         | <b>T18</b> | <b>T18M</b>                        | <b>T13</b> | <b>T13M</b> | <b>RD</b> | <b>Total</b> |
| <b>T = Trisomy, M = Mosaic, RD = Related Disorders</b>                                      |            |                                    |            |             |           |              |
| CARDIAC: AORTIC COARCTATION , patch / AORTA widened / Interrupted AORTIC ARCH / Aortoplasty | 20         | 0                                  | 4          | 1           | 3         | 28           |
| CARDIAC: AORTOPEXY Aortic Arch fixated to sternum                                           | 1          | 0                                  | 0          | 0           | 0         | 1            |
| CARDIAC: ASA REPAIR- Atrial Septum Aneurysm                                                 | 1          | 0                                  | 0          | 0           | 0         | 1            |
| CARDIAC: ASD REPAIR / STENTS                                                                | 58         | 13                                 | 15         | 5           | 18        | 107          |
| CARDIAC: AVSD REPAIR (Atrioventricular Septal Defect)                                       | 3          | 0                                  | 0          | 0           | 1         | 4            |
| CARDIAC: BALLOON DILATATION                                                                 | 1          | 0                                  | 0          | 0           | 0         | 1            |
| CARDIAC: BLAYLOCK-TAUSSIG SHUNT                                                             | 4          | 0                                  | 1          | 0           | 3         | 8            |
| CARDIAC: CARDIAC CATHETERIZATION to diagnose or treat                                       | 27         | 5                                  | 7          | 3           | 5         | 47           |
| CARDIAC: DOUBLE OUTLET RIGHT VENTRICLE                                                      | 4          | 0                                  | 1          | 1           | 1         | 7            |
| CARDIAC: ECMO                                                                               | 3          | 0                                  | 0          | 0           | 0         | 3            |
| CARDIAC: FONTANA PROCEDURE                                                                  | 1          | 0                                  | 0          | 0           | 0         | 1            |
| CARDIAC: GLENN PROCEDURE                                                                    | 0          | 0                                  | 0          | 1           | 1         | 2            |
| CARDIAC: HLHS PDA STENT                                                                     | 1          | 0                                  | 0          | 0           | 0         | 1            |
| CARDIAC: LINQ CARDIAC MONITOR                                                               | 0          | 1                                  | 0          | 0           | 0         | 1            |
| CARDIAC: PACEMAKER                                                                          | 8          | 3                                  | 0          | 0           | 1         | 12           |
| CARDIAC: PDA REPAIR / LIGATION / STENT Patent Ductus Arterius                               | 85         | 9                                  | 14         | 6           | 17        | 131          |
| CARDIAC: PERICARDIECTOMY                                                                    | 0          | 0                                  | 0          | 0           | 1         | 1            |
| CARDIAC: PFO CLOSURE                                                                        | 7          | 0                                  | 0          | 0           | 1         | 8            |
| CARDIAC: PULMONARY ARTERY BAND REMOVAL / LOOSENING                                          | 6          | 2                                  | 1          | 0           | 0         | 9            |
| CARDIAC: PULMONARY ARTERY BANDING                                                           | 34         | 3                                  | 8          | 1           | 1         | 45           |
| CARDIAC: PULMONARY ATRESIA                                                                  | 6          | 0                                  | 2          | 0           | 1         | 9            |
| CARDIAC: PULMONARY STENOSIS REPAIR / VALVULOPLASTY                                          | 8          | 3                                  | 2          | 1           | 2         | 16           |
| CARDIAC: PULMONARY VEIN REPAIR/ INNOMINATE VEIN REPAIR                                      | 2          | 1                                  | 0          | 0           | 0         | 3            |
| CARDIAC: TAPVR Total Anomalous Pulmonary Venus Return                                       | 0          | 0                                  | 1          | 0           | 1         | 2            |
| CARDIAC: TETROLOGY OF FALLOT REPAIR                                                         | 13         | 3                                  | 9          | 1           | 6         | 32           |
| CARDIAC: TRUNCUS ARTERIOSUS REPAIR                                                          | 0          | 0                                  | 0          | 0           | 2         | 2            |
| CARDIAC: UNSPECIFIED CARDIAC SURGERY/PROCEDURE                                              | 8          | 2                                  | 1          | 3           | 7         | 21           |
| CARDIAC: VALVE Surgery- BICUSPID (Mitral), AORTIC, TRICUSPID                                | 7          | 1                                  | 3          | 0           | 3         | 14           |
| CARDIAC: VALVE SURGERY ROSS PROCEDURE                                                       | 0          | 0                                  | 0          | 1           | 1         | 2            |
| CARDIAC: VSD REPAIR                                                                         | 121        | 18                                 | 15         | 4           | 25        | 183          |

**Table 10 – Click to Link to Trisomy 18, 13 - Cardiac Surgery Hospitals**

SOFT parents report their child's surgeries and procedures on the SOFT membership form. The link below shows hospitals that provided cardiac surgeries of those with a diagnosis of full trisomy 18 or 13. New reports to the surgery registry are automatically added to this link and will be viewable.

<https://trisomy.org/resources/hospitals-that-have-performed-cardiac-surgeries-or-procedures/#/>

The compiled list is sorted by state or province. It's important to pay attention to the latest year a surgery was done at that hospital. **Facilities that provided recent surgery are more likely to have a surgeon willing to assess a child with trisomy 18 or 13 for a cardiac repair. 2<sup>nd</sup> opinions are always an option.** A number of parents have posted in the SOFT Facebook group about having their infant's records sent to an out of state surgeon for a 2<sup>nd</sup> opinion. Parents can also directly contact a surgeon's office. **It is okay to ask for more than one 2<sup>nd</sup> opinion.**

Still in 2024 there are facilities unwilling or unable to offer cardiac repair to a newborn affected by trisomy. Parents in private Facebook groups for Trisomy such as <https://www.facebook.com/groups/TrisomySOFT/> readily share with other parents asking questions and can be a vital resource when seeking a friendly hospital or surgeon. Parents may also contact SOFT with inquiries at [www.trisomy.org](http://www.trisomy.org).

In 2024 an important landmark consensus report was published by the American Association of Thoracic Surgeons (St Louis et al., 2024). The consensus group stated nine different positions regarding cardiac surgery in trisomy 18 and trisomy 13. Most importantly the authors stressed the information showing that cardiac surgery for many conditions increased hospital discharge rates and long-term survival. The authors based their recommendation on multiple published studies cited in the paper showing these better outcomes after cardiac surgery. Additionally the report suggested a team approach to care in children with trisomy 18 and 13. Weaver and colleagues (2021) had previously advocated for a multidisciplinary team approach and documented their experience.

The SOFT link below illustrates the number of children with full trisomy 18 or 13 and also mosaic trisomy 18 or 13 by diagnosis who had a cardiac surgery and the number of different cardiac repairs done, during the same surgery. For example: Patent Ductus Arteriosus (PDA) and Ventricular Septal Defect (VSD) were reported as done during one surgery. Some children had just a single defect repair, while others had 2 or 3 different cardiac repairs in the same surgery. It is common for children with trisomy to have more than a single cardiac defect. <https://trisomy.org/reports/cardiac-procedures-by-diagnosis/#/>

*"An Ethics doctor (Canada) said the attitude is changing for the positive toward trisomy 18."*  
[A.S. Janessa, trisomy18, 2014](#)

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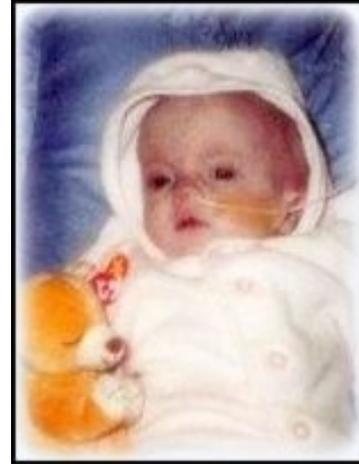


Ashton
10/11/1999
Trisomy 18

"Ashton met Dr Carey at 12 hours old. He gave us statistics and information to help us make the type of decisions no parent should ever have to make."

R. & S.W., 2013

Ashton had a VSD repair when she was about 2 ½ years old.



Morganne
10/9/1998 – 5/13/1999
Trisomy 18

Mortality and possible causes

"People told us having him would be the hardest thing we ever did. But they were wrong. The hardest part is not having him." P.F., Joey, trisomy 13

The sorrow of anticipatory grief is experienced by all parents of those diagnosed with trisomy 18 or trisomy 13. The risk of miscarriage or stillbirth, or fetal loss during labor is greater than in unaffected pregnancies. [Morris & Savva, 2008] Survival studies of affected live born infants, without mention of any interventions, indicate in both syndromes; 50% die in the first week of life and 5-8% live beyond the first year. [Rasmussen et al., 2003; Carey 2012; Cereda & Carey 2012] More recent population studies show improved survival with interventions. [Meyer et al. 2016; Kosho & Carey, 2016; Nelson et al., 2016] Approximately half of the infants in the Meyer study were born premature and **gestational age at birth was the strongest predictor of mortality**. Health care providers remind parents at every visit that their baby's situation is "grave", even when a baby is doing well. **Most parents hope that their baby will be the exception to the statistics but fear that the predicted loss will happen.** Parents of those who live beyond infancy, at some point, begin to shift from the sadness of waiting for their baby to die to finding out what can be done to help their baby develop her potential and enjoy life. This transition is emotionally uplifting for parents. However, the fear of predicted loss returns with each health crisis. For those who reach childhood and older there is less emphasis from their doctors about dying and a common reference to their child "writing her own book".

"I was prepared for a new baby. But I wasn't prepared for seizures, tubes, medicines, doctors, special foods and the fear of never knowing if he would die." P.F., Joey, trisomy 13

"Our son is doing great and we are gradually shifting from grieving to appreciating the joy he brings us and believing that he will be with us for a while." G.T., Garrett, trisomy 13

~

Apnea (Obstructive)

Only about 50% of those born with trisomy 18 or trisomy 13 survive longer than a week and apnea is reported by a number of SOFT parents as cause of death. There are three main categories of apnea.

- **Obstructive apnea** is a pause in breathing due to obstruction of the upper airway
- **Central apnea** is a pause in breathing due to central nervous system dysfunction of the respiratory center in the brain. See page 25.
- **Mixed apnea** is a combination of both central and obstructive apnea.

Obstructive apneas are the most common occurring. Enlarged tonsils or adenoids, and malformations such as **tracheomalacia** (softening of the cartilage of the trachea) can narrow or temporarily **obstruct** the airway, especially during sleep, as muscle tone decreases during sleep. Reflux, or vomiting can briefly occlude, or positioning might narrow the airway. Parents should inform the doctor if their infant or child experiences restlessness during sleep, excessively sweating, flaring their nostrils, or gasping for air. Snoring is another sign to be reported but it needs to be noted that respiratory effort decreases during sleep and for those who are not moving enough air, snoring may not be evident.

Often parents hear from other parents about how little their child sleeps and accept this as part of parenting these children. It is important to **look for an underlying problem**.

If appropriate, the child's doctor can refer the child to a **pediatric pulmonary doctor and sleep specialist for sleep studies** to be done at a sleep clinic. Other relevant specialists include a **pediatric neurologist** for evaluation of possible seizures, a **pediatric gastroenterologist** to test for reflux, aspiration or structural issues or a pediatric **otolaryngologist** (ear, nose and throat doctor commonly called **ENT**) for evaluation of possible tonsil and/or adenoid problems.

"The pulmonologist said it looked like Lilly's tongue may be blocking her air passage so to have her sleep on her side. I still am amazed at the difference it has made for Lilly. We went from her waking up gagging and gasping almost every hour to sleeping easily through the night." L.H., Lilly, trisomy 18 [Pray for Lilly blog, 2011]

A number of parents report use of a **Bi-level Positive Airway Pressure (BiPAP)** machine at night to maintain an open airway during sleep. The BiPAP, using airflow, keeps the airway open with greater pressure during inhalation and lesser pressure during exhalation. A Continuous Positive Airway Pressure (C-PAP) machine has one pressure setting. These machines are **non-invasive breathing aids** that can be set for different modes of action depending on the child's needs, usually evaluated by a sleep study. It is a commonly used machine for adults with snoring and apnea issues. Finding the right size mask can be difficult, perhaps more so for infants and children with small jaws as is often seen in trisomy18, and tolerance for breathing through a mask or nasal pillows can be a discomfort issue.

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**Kristina "Krissy"**  
**3/25/2000**  
**Trisomy 18**

*"At that time there wasn't a neonate size C-PAP mask. Krissy had a trach placed when 3 months old to keep her safe during sleep because of obstructive apnea." T.K., 2017*

Krissy is the inspiration for **Trisomy Talk** webinars created and hosted by her mother, Terre Krotzer.



**Summer**  
**10/21/2011**  
**Trisomy 13**

Reports to SOFT of tracheostomy surgery for both trisomy 18 and trisomy 13 increased significantly in the last several years. A tracheostomy surgery creates an opening through the neck into the trachea (windpipe). A tube through this opening provides an airway and access for removal of secretions from the lungs. The SOFT Surgery Registry shows a couple of children had surgery to close their tracheostomy. A few parents chose to keep the tracheostomy, even when no longer necessary, as they find it beneficial when their child is ill or if need of future surgery.

### **Pneumonia**

Pneumonia is often reported as cause of death by parents of those with trisomy 18 or trisomy 13. Reflux with aspiration can result in an aspirational pneumonia; a frequent illness in both disorders. Pneumonia usually requires hospitalization for those with trisomy 18 or trisomy 13.

Preventive breathing treatments, given through a **nebulizer machine** that dispenses a medicated mist to inhale and directly treat the lungs, often becomes a daily or more at-home treatment for those with chronic respiratory problems. For those without chronic respiratory issues the recommendation for treatment might only be when the child has symptoms of a cold.

**Chest percussion following treatment** helps aid pulmonary drainage and parents can be instructed how to do this at home. A relatively new therapeutic tool called the **Vest™ is an Airway Clearance System** used to help clear mucus from the lungs. It is an inflatable vest, with air chambers that pulse off and on, giving chest percussion simultaneously to all areas of the chest during a treatment. Call for Vest™ information at 1-800-426-4224.

**Phillips Cough Assist T70 is a mechanical device for those unable to effectively cough.** Ask the doctor if your child would benefit from using this machine. <http://www.medgadget.com/2013/01/philips-respironics-coughassist-t70>

Pneumonia is a serious illness for someone with restrictive lung disease and can be life threatening. See page 61. Scoliosis is a common illness in trisomy 18 and those with trisomy 13 might also develop this

problem. Scoliosis often begins in early childhood but parents of a couple toddlers with trisomy 18 have reported an earlier onset of scoliosis. Scoliosis usually progresses with time, and growth; resulting in restriction of the lungs and poor pulmonary function.

## Cardiac Conditions

On the SOFT registration form a frequent response from parents about the cause of death for their child with trisomy 18 or trisomy 13 is "heart" failure. In a 1994 study of the natural history of trisomy 18 and trisomy 13, researchers noted that when they looked at the causes of death reported by SOFT parents and by medical records of the children, they were surprised by the lack of variety of the diagnosis, as they found that **most causes of death were a variation of cardiopulmonary arrest.** [Baty et al., 1994]

**Prenatal ultrasound findings are not always accurate.** A number of parents were told their fetus had a severe heart defect, but at birth, a less serious defect was found. Newborns with these disorders should be evaluated by a pediatric cardiologist and also have an echocardiogram. It is appropriate for parents to request this evaluation if it has not been done. **Carey recommends that infants with trisomy 18 or trisomy 13 have an echocardiogram in the newborn period.** [Carey, 2010; Cerda & Carey, 2012]

**Pulmonary hypertension (PH) related to heart defects is common in infancy in both disorders and there are studies showing development of PH early for those with trisomy 18.** [Kosiv et al., 2023] PH is abnormally high blood pressure in the small arteries of the lungs resulting in the right side of the heart working harder to pump blood to the lungs and eventual heart failure. **Once an infant is past 2-3 months and is thriving, the issue of the development of pulmonary hypertension emerges.** [Carey, 2010] There is no cure for PH. Medications can help and sometimes a cardiac repair improves PH. PH can be the reason a cardiac surgery is denied.

*"Her PH diagnosed when 4 months old developed quickly, in a 3 week span between echoes, with shunting so pronounced on the echo there was no need to confirm by cath. After 3 months of treatment with oxygen and medications, and when evidence of shunting on her echo was no longer seen, a cath was done. Her pressures were found to be low enough for it to be safe to close her ASD & VSD. She weaned off all PH and cardiac meds within 6 months of open heart" S.C., Nora, trisomy 13, 2017*

**Cardiac surgery has become more common in recent years for infants with trisomy 18 or trisomy 13** yet it is still not done in many hospitals. Parents who find a surgeon in another hospital must ask the cardiologist at the birth hospital to provide findings to this surgeon. If the surgeon is willing to attempt a repair, the infant must then be stable enough for transport. It is reasonable and appropriate for parents to ask the NICU attending if medical interventions can be done to stabilize their infant for this purpose.

The SOFT surgery registry maintains a list of parent reported cardiac repairs and hospitals where these repairs were done. See pages 58 -59. **Be aware-** Hospital policies can change or a willing surgeon retires or sometimes relocates so some hospitals listed might no longer provide cardiac repair for those with trisomy 18 or trisomy 13.

About 10% of infants with trisomy 18 and 20% of those with trisomy 13 are born **without** cardiac anomalies. Survival can be longer for some with a normal functioning heart but other problems common to these disorders still risk their viability. **The greatest gift of successful heart repair is less about life**

**extension and more about preventing or relieving the discomfort of progressive heart disease and improving quality of life.** If a cardiologist recommends cardiac surgery, then a surgeon is consulted about doing the repair. The benefits versus the risks of cardiac surgery need to be discussed with the parents. Some parents choose not to do surgery. Treatment of cardiac symptoms with medications is reported by most parents of infants and children with both disorders. Sometimes cardiac anomalies become less significant or resolve on their own.

*"An echocardiogram showed that not only had the PDA closed but also the VSD we did not know he had was almost closed." [The SOFT Times, 2002]*

*"The cardiologist knew the surgeons at our hospital would not repair Lyndsay's heart as they had a bias and refused to do heart surgery on infants with trisomy 18 or 13 as 'parents were not realistic to their long term outcomes'. So off we went to another hospital where the pathway had been forged months earlier by the family of another little girl with trisomy 18. Her heart was repaired with her weighing only 4 pounds, 3 ounces. [The SOFT Times, 2002]*

*"Her cardiologist was surprised because her large VSD had closed on its own." [The SOFT Times, 2002]*

Using the SOFT surgery registry, Hansen et al., (2000) performed a study of cardiac repair in those with trisomy 18 or trisomy 13 and reported 25 of 29 children (86.2%) who had a cardiac surgery were discharged to home. There were 4 surgical mortalities (13.8%). Graham et al summarized a 91% discharge to home in their 2004 published study. **It is evident that these children can survive anesthesia, and survive to go home, making surgery an appropriate option in certain cases.** [Carey, 2010; Cereda and Carey, 2012]

*Care of Trisomy 18 Children in Japan* published in the *American Journal of Medical Genetics [2008]* by Dr. Tomoki Kosho, medical advisor for trisomy 18 and trisomy 13 societies in Japan, discusses comprehensive care for newborns with trisomy 18 that includes cardiac surgery. "To my knowledge this is the first series, which attempts to discuss efficacy of intensive cardiac treatment including surgery for patients with trisomy 18 or 13 though the sample size is small" Dr. John C. Carey, [The SOFT Times, 2010]

In a single institution study of patients with trisomy 18 and trisomy 13 Costello et al. found longer survival in those who had cardiac repair compared to those who did not. Costello "advocates" offering surgical repair over palliative care for moderately complex congenital cardiac anomalies considered as operable to parents of children with these syndromes. [Costello et al. 2015]

**The American Association for Thoracic Surgery (AATS)** – Expert consensus document mentioned above, *Recommendations for the care of children with trisomy 13 or 18 and a congenital heart defect* conclusion stated "Based on our analysis of recent data, we recommend that decisions should not be based solely on the presence of trisomy but, instead, should be made on a case-by-case basis, considering both the severity of the baby's heart disease as well as the presence of other anomalies. These recommendations offer a framework to assist parents and clinicians in surgical decision making for children who have trisomy 13 or trisomy 18 with congenital heart defect." [St Louis,, et al. 2023 ]

## [Neoplasia \(Tumor\)](#)

In 2017 the American Association of Cancer Research (AACR) published research about syndromes with a predisposition to developing Wilms tumor or Hepatoblastoma (HB). The AACR recommends screening in trisomy 18 for Wilms tumor through the 7<sup>th</sup> birthday and Hepatoblastoma through the 4<sup>th</sup> birthday by abdominal ultrasound, starting from birth (or diagnosis) and done every 3 months. Screening for HB in trisomy 18 should also include Alpha-Fetoprotein (AFP) checks, every 3 months through the 4<sup>th</sup> birthday.

These recommendations were simply a carry-over from guidelines for screening in other conditions and **not** specific for trisomy 18. These will be revised in the future as current research is published. (personal communication, Carey, 2017)

Older infants and children with trisomy 18 are at increased risk to develop Wilms tumor (kidney tumor) and hepatoblastoma (liver tumor). Development of Wilms tumor in trisomy 18 is typically after 4 years of age and the oldest known child was 12 years old. Clinical reports of hepatoblastoma in trisomy 18 report occurrence between the ages of 4 months to 3 years. [Carey, 2005; Cereda and Carey, 2012, Satge et al., 2016; Satge et al., 2017].

**A study of tumor in trisomy 18** included cases reported by parents to the SOFT surgery registry and resulted in the seminal publication of guidelines specifically for tumors in trisomy 18. (Farmakis et al., 2019) Because of the risk for the tumor of the kidney and liver (which are treatable and curable) in children with trisomy 18, the guidelines include: abdominal ultrasounds every 3-4 months until age 4; these detect changes in the liver and kidney; blood levels of alpha feto-protein (AFP) every 3-4 months until age 4 (screening for the liver tumor); at age 4 years the AFP screening is stopped because liver tumor risk is now nil and the imaging is switched to kidney ultrasounds (easier than abdominal which require fasting before study); continue kidney ultrasound until age 8 years. [Farmakis et al., [2019]

### **Health Supervision and Anticipatory Guidance for Children with Trisomy 18.**

|                                               |                                                                                                                                                                                            |
|-----------------------------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Neoplasia<br>(Wilms tumor/<br>Hepatoblastoma) | Abdominal ultrasound at birth or when diagnosed, performed every 3 -4 months to age 4 years and then renal ultrasound every 3-4 months until age 8; AFP every 3-4 months up to age 4 years |
|-----------------------------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|

**Dr John C Carey's response to a parent's question on SOFT Facebook about when to stop screening:** (with permission as shared by parent BK, SOFT Facebook group. Aug 2024):

"Dr Stephen Braddock has been investigating a revision of the guidelines. The age when to stop the kidney ultrasound is being discussed with tumor experts. All of the children who developed Wilms tumor were under 8 years, except one who was 12; therefore we are planning to increase the age of kidney ultrasound to 14 years." [Carey, 2024]

"Additionally because of a child who had the liver tumor (hepatoblastoma) after age 4 we have considered extending abdominal ultrasound until 10 and then switching to kidney ultrasound at age 10 until 14." The actual risk of either a liver tumor or Wilms tumor is likely less than 1%. [Carey, 2024]

"Led by Dr Braddock, we hope to publish a revision by early 2026 after discussion with tumor specialists." [Carey, 2025]

## There are no well established tumor risks in trisomy 13.

*"They gave us choices ...start chemotherapy first... or remove the tumor first and then do chemotherapy, or do nothing at all. They treated our daughter with respect and dignity. They told us they were treating her as they would any child with a hepatoblastoma." [the SOFT touch, 1993]*

*Perhaps the most profound legacy our daughter left is about caring. Family, friends and most surprising, doctors, nurses and therapists dared to care about her..." [the SOFT touch, 1993]*

## Life Support Directives

Decisions regarding life support in the care of infants and children with trisomy 18 and trisomy 13 are complex. In the box below is a general review of advance directives. **On the next page, Parents and Life Support Order Decisions are discussed.**

The existence of three advance directive documents plus the in-hospital life support order form can create confusion for the public. See a general explanation of Advance Directives in the box below.

### Advance Directives

Competent persons, age 18 years or older, can document their health care decisions in advance to be used as a declaration of their intent when a life support order needs to be written. A Living Will is **not** considered a legal medical order and will **not** be followed by medical personnel without a concurring, signed physician order. The only Advance Directive that is a legal medical order is #3 (below) because it is signed by a physician.

#### 1. Living Will

This document is a declaration of an adult person's directive about life-saving efforts to be done for him/her in the case of terminal or incurable illness or persistent vegetative state.

#### 2. Health Care Power of Attorney

This document names another individual to manage healthcare decisions of a person, if that person is determined by a physician to be unable or no longer able to make decisions on his/her own.

#### 3. Advance Directive with a signed physician order

*(This is the only advance directive that can be used for a minor)*

The title often differs from state to state. **It is an advance directive that is signed by a physician, which makes it a legal medical order** and it is used in cases of end-of-life terminal conditions for adults and **children** as defined by each state. Some states require the patient or representative also sign this directive, as well as the physician. This form is to be kept in a visible place with the patient at home to inform emergency personnel of the patient's directives in an end-of-life situation. **It must be seen if it is to be followed by medical persons.** If the patient is transported to the hospital, the form should go with him to be used as information in completing an in-hospital Life Support order form.

## DNR/Life Support Order

The title on the form used to write the **in-hospital Life Support order** for a patient may vary from hospital to hospital. To be valid, this order must be signed by the hospitalized patient's attending physician. A Life Support Order directs the amount of **cardiopulmonary resuscitation (CPR)** to be done or NOT done in the event of cardiopulmonary arrest. **A DNR (Do Not Resuscitate) is a Life Support order to withhold CPR.** One SOFT mother recently explained that the life support order documented on her child's in-hospital chart was termed **Allow Natural Death (AND).**

There is a provision in some states for a doctor to make a **unilateral** decision to code a patient as a DNR; when there is no directive, no one to speak for the patient and the patient is unable to speak for herself in a "hopeless" terminal situation. A second written opinion from another doctor unconnected to the case is usually required. Your state law statutes pertaining to life support directives can be checked on-line.

## Parents and Life Support Decisions

It is part of the admitting doctor's job to ask the parent or guardian what to do in a cardiopulmonary event **each time** a child is brought to the emergency room or hospitalized. It is an uncomfortable question and in the midst of many other admission questions, is often only addressed as "Do you want her on a ventilator?" **But some parents might not be aware a Life Support order is being created.**

A discussion about Life Support would provide an explanation about procedures used during cardiopulmonary resuscitation (CPR), such as intubation to open the airway and the use of a ventilator that will do the work of breathing, chest compressions to stimulate the heart or the use of a defibrillator that shocks a heart back to beating in a normal rhythm, and medications used to stabilize a patient in crisis.

**The purpose of the question "Do you want her on a ventilator" is not clear,** especially for new parents, without a discussion about Life Support procedures or without showing the hospital Life Support Order form to the parent. A Life Support order tells the medical staff if the child should be given full CPR, or partial CPR interventions, or no CPR in the case of the child's heart or respirations stopping.

Some doctors prefer to ask for a parent or guardian signature when they write a DNR order to show proof of agreement, and there are hospitals that require physician and parent or guardian signature when a DNR order is written for a child. If a parent signature is not required, a Life support order would likely not be seen by a parent or guardian; unless they ask to see it.

Parents of newborns have shared, via social media, that their infant was coded a DNR in the NICU because of their diagnosis of trisomy 18 or trisomy 13; sometimes without parental knowledge or consent. **It is important and appropriate for parents of newborns to ask about their child's Life Support order.** If parents are not comfortable with the order, they can ask for it to be changed. Parents considering discharge to Hospice or Palliative care ought to **ask about** the program's policy pertaining to Life Support Orders and options for care.

## Changing Life Support Orders

A child's Life Support order is written and signed by the doctor and **should** reflect parent or guardian wishes. Parents or guardians can change their mind at any time about what is to be done and ask for a new order to be written. A new Life Support Order signed by a doctor invalidates all previous orders. All in-hospital life support orders ever written for your child are part of your child's permanent records forever.

A permission to release information form signed by parents, makes these records available for legal action or other stated purpose.

**Most people are unaware that in-hospital Life Support orders are reassessed at specific times. This means they are looked at for appropriateness and re-written whenever a patient is:**

- moved to a new unit, such as admission to the PICU from the ER
- when a new attending physician takes over the care of a patient
- if there is a change in the patient's condition
- when there is a change in parent/family wishes.

It is very important to check the status of your child's the Life Support Order, if any of these changes happen, as **it is possible for a Life Support order to be written or changed without parental knowledge or consent**. Parents can ask to see or request a copy of their child's life support order.

*"The outcome was that the actions in the final 24 hours did not represent appropriate forms of care. A DNR was entered in her chart without consent". B.F., Annie, trisomy 13, personal communication, [2007]*

*"In the end, the PICU Attending **overrode our wishes for life support**. We learned this after her death when we obtained her records and found entries on two consecutive days, written by the resident that said "DNR per Attending." AB, Megan, trisomy 18 [2004]*

## Making Decisions

Be it a prenatal diagnosis or child in a health crisis, the parent struggles with hope and fear and comes to a decision based on the doctor's explanation about their child's illness and options, their personal values and beliefs, and what they feel is in their child's best interest. Crucial to parent or guardian comfort with a life support decision is physician-parent trust. Respect by the parent of the doctor's knowledge and limitations AND respect by the doctor of the parent's values and directives are vital to trust.

*"...I knew then, and I know now that her death was not the result of a (physician-parent) partnership failure." [The SOFT Times, 2004]*

*"It was our decision not to take heroic measures to prolong his life..." [Stenson et al., 1992]*

A positive prenatal screening or diagnosis results in expectant parents searching the internet for information about these syndromes. Finding resources such as SOFT and connecting with other families via social media, enables learning about variable outcomes, depending on the unique circumstance of

each infant, options for interventions, and care decisions. Members of Facebook groups for parents at all stages of the trisomy journey ask questions and/or share about problems and solutions. The SOFT private Facebook group is at <https://www.facebook.com/groups/TrisomySOFT/>

**Parents often comment about being advised and feeling pressured to code their child with trisomy 18 or trisomy 13 as a DNR.** If there is not a DNR order, medical personnel are required to administer CPR, if the child's heart stops or breathing ceases. The concern expressed by the medical community is that the child might be unable to be weaned off a ventilator. The worry for parents is that the conventional view, of limiting extraordinary treatment for these children, will result in withholding interventions that might give their child a chance to live.

### **Disagreement about Life Support Order Decisions:**

Any child who is acutely ill does not present at their normal baseline. Photographs taped on the wall by the hospital bed, enable healthcare providers to see the child when well; *a picture really is worth a thousand words.*

A doctor's view of trisomy 18 or trisomy 13 could contribute to the recommendation of a DNR; thus **patients with these syndromes can be at increased risk for the writing of a DNR order.** In most states the laws governing Life Support orders are protective of the physician in his decision making. If there is unresolved disagreement between or among the healthcare providers and/or the patient's family about a Life Support designation, a physician might ask the hospital **Ethics Committee** to help with resolution. A doctor may consider transfer of the patient to another physician, or a parent may request a different physician. Possibility of transfer to another healthcare facility or judicial resolution might also be considered.

*"Later we found out there was a DNR. That explains why they did nothing. **We never signed a DNR.**" S.C. Simon (t18) **Global Genes** ad *Our "Labeled" Child's Name is Simon: Fighting for Treatment [2014]**

*"The doctor said 'it's too risky' to take her down for a CAT scan that would confirm the problem. We now know that **we should have asked 'Too risky for whom?'**" A.B., Megan, trisomy 18*

Parents with concerns about their child's hospitalization and care have a right to request the child's medical records but will be charged for the service of copying and mailing. Some hospitals provide requested records to authorized persons by download from the hospital web site.

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"Kimberly is a blessing. She is in our hearts, our memories, and is still very much a part of our lives." E.R., 2007

Kimberly lived longer than any person known in scientific literature with full trisomy 18. She was the oldest known to SOFT, until 2011.

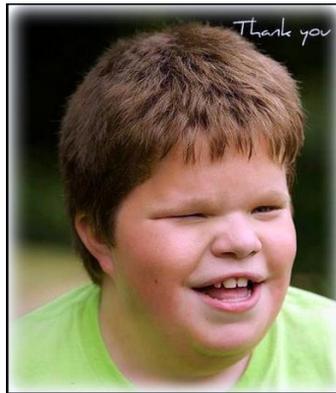


Kimberly
12/11/1968 – 8/3/2005
Trisomy 18

Mary
3/27/1996 – 8/21/2016
Trisomy 18



Nicholas
4/6/1995 – 11/4/2013
Trisomy 13



Samuel, "Sam"
11/28/1998
Trisomy 13 Mosaicism



Natalia
8/25/2000
Trisomy 13

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**Table 11 Health Supervision, Surveillance, and Anticipatory Guidance for Infants and Children with Trisomy 18 and 13**

**Infants (0-1)**

| <b>Manifestation/Theme</b>                    | <b>Diagnostic or Screening Test/ Referral</b>                                                                                                                                                              |
|-----------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Congenital heart defects                      | Pediatric cardiac evaluation, including echocardiogram in newborn period and follow-up as needed per pediatric cardiology                                                                                  |
| Developmental disability                      | Referral to early intervention program<br>Referral to other programs for children with disabilities (OT, PT speech/hearing)                                                                                |
| Growth delay                                  | Measure length, weight, head circumference at every visit; plot on trisomy 18/13 growth curves                                                                                                             |
| Feeding difficulties                          | Referral to dysphagia or feeding team; assessment for GE reflux, consider G-tube at 6 months or sooner if required                                                                                         |
| Chronic Constipation                          | Referral to Pediatric Gastroenterology where appropriate                                                                                                                                                   |
| Hearing loss                                  | Newborn screening; Audiology 6 months of age; follow-up as needed                                                                                                                                          |
| Neurologic/<br>Central nervous system         | Neuroimaging in newborns and infants with trisomy 13; in trisomy 18 as indicated                                                                                                                           |
| Ocular malformations                          | Newborn Ophthalmology evaluation in trisomy 13; as indicated in trisomy 18 in the newborn period; then ophthalmology evaluation by 6 months in trisomy 18                                                  |
| Neoplasia<br>(Wilms tumor/<br>Hepatoblastoma) | For trisomy 18: Abdominal ultrasound at diagnosis, done every 3-4 months to age 4 years and renal ultrasounds every 3-4 months until age 8; AFP every 3-4 months to age 4 years                            |
| Other respiratory                             | Pediatric ENT or Pulmonology evaluation of airway in the newborn period and then follow-up as needed; consider sleep study in the newborn period; high index of suspicion for obstructive apnea in infancy |
| Genetic issues                                | Referral for medical genetics consultation and genetic counseling                                                                                                                                          |
| Overall care                                  | Referral to <b>comprehensive care team</b> , usually at a children's Hospital; or Medical Home in primary care practice if applicable                                                                      |
| Palliative care                               | Guidance in decision-making and overall support                                                                                                                                                            |



important. Finding reliable help and being able to count on certain times when their child will be competently cared for decreases caregiver burnout and allows some time for the caregiver to do other things. Parents report that it takes a while to find the right helper and to adjust to having another caretaker in the home.

Too many parents of newborns with trisomy 18 or 13 are not told about the **Medicaid Waiver** by hospital staff. Parents can ask the social worker about applying for the Medicaid Waiver for a child with trisomy. **The Medicaid Waiver for those who do not qualify for regular Medicaid, and regular Medicaid cover doctor ordered medical care cost, either as the main insurer or supplemental for those with private insurance. See <https://medicaidwaiver.org/>**

*"My child is disabled and I need help to do all the things he needs done. Your agency sent you. I can't always tell if you are real, but my son can. So I watch him. ...My husband resents people coming in and out of our home. ...You tell me you are coming, and then you call and tell me you have to cancel!!!" [The SOFT Times, 2002-2003]*

*"I just can't do this alone any more, what can I do? How do I find a good nurse? I won't let just anyone take care of my son." [The SOFT Times, 2001]*

## **AAP Report 2025**

In July 2025 the American Academy of Pediatrics (AAP) issued an important Clinical Report entitled "Guidance for Caring for Infants and Children with Trisomy 13 and Trisomy 18". The report was authored by Dr Alaine Pyle and coauthors representing the AAP Committee on Bioethics, the Section of Neonatal-Perinatal Medicine, and the Committee on the Fetus and Newborn. This document represents another landmark publication along with the AATS Consensus document on cardiac care (see page 64). The AAP report indicated that there are "no ethical justifications" for treatment of children with trisomy 18 and 13 differently than other children with medical complexity based solely on the genetic diagnoses. They summarize their recommendations by stating that "Pediatric and obstetric clinicians participating in the care of these patients should maintain current knowledge of literature to provide adequate counseling through a shared decision-making process and offer the broad range of support that is needed for families throughout the continuum of care from the prenatal to postnatal time and extending throughout the child's life." The Report goes a long way in removing any bias toward declining treatment or surgery in children with trisomy 18 and 13 based simply on the diagnosis of the condition.

## **Pediatric Hospice, Pediatric Palliative Care, and Perinatal Palliative Care Programs**

It is common now for parents of newborns with trisomy 18 or trisomy 13, to be offered the option of Pediatric Hospice or Palliative Care when their infant is discharged. These programs have become more available in recent years, as well as, Perinatal Palliative Care, for the expectant parent carrying a fetus with a life-limiting disorder. Those struggling with accepting the diagnosis and predicted prognosis might be reluctant to be in programs often associated with dying. Participation is optional.

Both **Hospice** and **Palliative Care** provide palliative care to ease discomfort and improve quality of life. But **there is a difference in management of care in each program.**

**Pediatric Hospice Care** is for patients considered to be in their last six months of life as determined by a doctor. Hospice focuses on comfort care to ease symptoms for those no longer seeking interventions for a cure. However, **Pediatric Hospice still lets kids have treatment oriented care.** Parents ought to ask if this is true in their local hospice program. Hospice also focuses on emotional and spiritual support for patients and their families. Hospice provides nurse visits or nurse aide and volunteer hours in home care and in some communities a hospice facility might be available. Hospice might also supplement services a family is receiving through their state, such as nurse's aide hours in the home.

*"We had a **Hospice** nurse coming to the house every other day. Mandy was Rebecca's nurse, and my new and dear friend! Thank you Mandy." [The SOFT Times, 2001]*

*"It was time for him to come home and he was still having bouts of apnea. I requested an apnea monitor and the neonatologist would not approve one. We gathered up our baby and left. We were hooked up immediately with **Hospice**, who by the way got us an apnea monitor." [The SOFT Times, 2002]*

**Pediatric Palliative Care** focuses on patient comfort but also allows interventions such as therapies, treatments and even surgery but cardiac repair for those with trisomy 18 or trisomy 13 might not be approved. Cardiac surgery approach varies by region and team. (Carey JC, personal communication 2013) **Palliative care helps parents with decision-making about care for their child with a life limiting diagnosis. It can begin early in a diagnosis and extend for years.**

**Perinatal Palliative Care** (PPC) is a care concept for parents who choose to continue a pregnancy after learning their expected baby has a life-limiting diagnosis. Prenatal diagnosis prompted the development of this program. PPC helps families with making choices about the birth and end of life care. A birth plan is created to also include parent's wishes about care and interventions, if their baby survives birth.

**Survival statistics for trisomy 18 and trisomy 13 will likely influence PPC guidance to anticipate loss but it is crucial that care providers also allow hope for the predicted possibility of survival.** See <https://www.perinatalhospice.org/list-of-programs> for listings of perinatal/palliative care programs in the USA and internationally. Parents should find out how PPC is reimbursed, confirm that Medicaid is accepted and if privately insured, also check with their insurer. Grants and donations allow service to those in financial need.

## **Other Important Care and Support**

Most states have programs to help families care for their special needs child at home but some states provide more services than others. Contact the local Department of Social Services (DSS) to inquire about services for families with special needs children and about Medicaid and the Medicaid WAIVER. Be aware these services might be affected by federal changes to the Affordable Care Act and state rules.

**A Medicaid Waiver evaluates only the assets of the child** and the income of the parents is not a consideration for qualification. **The child must not have any assets in order to qualify.** Usually a child's name goes on a waiting list to get into this program. There are states that do not have a Medicaid Waiver

program. Medicaid generally covers the cost of services documented to be medically necessary. Prior approval may be needed for medical equipment or home health nursing or aide services. Medicaid is the primary payer but if the child has private insurance, then private insurance is the primary payer and Medicaid becomes secondary. See Medicaid Waivers by state- <http://medicaidwaiver.org/>.

*"I called in her first monthly supply order for the year and was told that insurance is no longer covering her food (enteral formula). We have great insurance **but times they are a changing**." T.K., Facebook, 2014*

*"The Medicaid Waiver has made a difference in our pocketbook and the flexibility of our lives." [the SOFT Times, 2002]*

*"We put her name on the waiting list for the Community Alternative Program (CAP), funded by Medicaid, which helps families of disabled children." A.B., Megan, trisomy 18*

## Respite Care

Organizations such **the ARC** or **Easter Seals** provide respite and other services within their communities for families with special needs children and adults. It is important to ascertain if the respite service offered is safe for your child's care needs. Families report difficulty in finding appropriate programs, particularly for the young adult who ages out of the public school system.

*"Our daughter never went to a public school; instead she attended Arc programs for her education and still attended the Arc when 32 years old." C.H., Joanna, trisomy 18*

## Placement

While rare in current times, a few parents of newborns with trisomy 18 or trisomy 13 were advised by the doctor to consider placing their infant in a facility. Occasionally, parents of older infants and children are offered a placement option by the doctor, because their child's care needs have increased. Families with any older child, adolescent or adult requiring continual supervision and care, might at some point, choose to seek such placement.

Facilities for children and young adults with special needs are few in number and have waiting lists and requirements as to who can be admitted. Currently in the USA Medicaid covers the cost for those who qualify but parents should check with the business office about how reimbursement is obtained. A child generally ages out of these facilities at twenty-one years old and his court appointed guardian(s), usually the parent(s), bring their young adult home or find an adult facility. Guardianship is discussed later in this chapter. It is important to meet staff, see the accommodations, and learn how daily care and activity such as special education, therapy and recreation are provided; plus confirm how health care is obtained.

Parents can change their mind, make arrangements to bring their child home and arrange to restart home services. A young man with trisomy 18 lived in group home for a few years but his medical issues increased and his care needs could no longer be met at that facility. His family brought him home and cared for him through the final years of his life.

The family of an adolescent with trisomy 18 had several negative experiences with home health and decided on placement in a facility for special needs youth, not far from their house. They bring their daughter home for weekends and other special days. She is now an adult and still resides at the facility.

An adult with trisomy 18 was in a skilled nursing facility for the last several years of her life. She was tube fed, used oxygen and Bi-PAP which made her level of care too involved for a group home. In the beginning this was an emergency placement due to her mother being injured and hospitalized. This young woman with trisomy 18 eventually became a resident at this facility as it was decided that placement was best for their family.

*"We put Aaron's name on a waiting list at age 11 years, thinking it would be several years before a slot opened for placement but we got the call when he was 12. Making the decision was a roller-coaster event with one of us saying yes to placement but the other saying no and then each reversing our decision several times but finally we decide to give it a try." S.B., Aaron, trisomy 13*

*"It was difficult not having him home but we see benefits for Aaron. He has a friend who seeks him out and that didn't happen at home. He has gained skills because therapy is available there, that could not be done at home. We did not realize how little attention his sister got and our whole family is healthier this way. It is best for our family". S.B., Aaron, trisomy 13*

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**Elizabeth, "Liz"**  
**8/11/1986**  
**Trisomy 18 Mosaicism**

*"Saskia is the oldest known person in England living with trisomy 18."*  
*H.H., 2014*

Heidi Herdman, co-founder of **Trisomy 18-13 Support-UK** is the mother of Saskia.



**Saskia**  
**2/18/1992**  
**Trisomy 18**

*"Attending the SOFT conference is the high point of "Liz's" year! She attended Kingsborough Community College and volunteers in the recreation department of a local nursing home. She is an "honorary mom" on the SOFT Facebook Group page." A.B.-C., 2014*

## **Legal**

Most states have an agency that advocates for persons with disabilities. Legal disputes regarding education rights, access rights or possible medical negligence can be presented to this agency and, if there is merit to the complaint, legal advice or aid might be given to the parents by the agency. There is usually no charge for these services but parents might be asked to contribute to the costs involved in making their case. Cost should be clarified in the initial meeting between the state advocate and the parents. National Disability Rights Network (NDRN) lists state protection and advocacy agencies and client assistance programs (P&A & CAP) at <http://www.ndrn.org/en/ndrn-member-agencies.html>

## **Guardianship**

Parents need to apply for guardianship by or before the child's 18<sup>th</sup> birthday. In this process, the child will be declared incompetent and a parent(s) will be made the court-appointed legal guardian(s). This allows the parent(s) to continue making decisions for their child after they reach legal age. The service of an attorney is helpful.

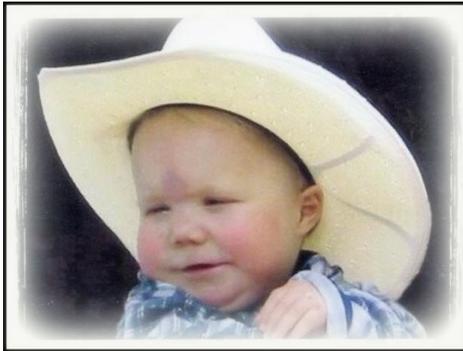
Barbara Van Herreweghe, President of SOFT, reports that in the state of New York parents can print a guardianship application form from the internet, have a notary witness signatures, and take the form and any other required documentation to a Surrogates' Court to request guardianship of an incompetent dependent. Doing this is less expensive than using the services of an attorney. Check the internet to find information about the availability of a Surrogates' court within a specific state. Barbara and her husband, Dave, are the parents of Stacy (5/21/81) who was born with trisomy 18.

## **Supplemental Care Trust Fund**

Trisomy 18 and 13 survival statistics indicate it is unlikely that there will be a need for a Supplemental Care Trust Fund to be in place, yet it is a possibility. Parents of long term survivors, who are doing well, and of those with related disorders should consider arranging a supplemental care trust fund. Van Herreweghe reports there are three types of trusts for this purpose. The following discusses one; a Testamentary trust.

A Testamentary trust fund is established and will be activated in the event of the death of the only parent/guardian or both parents/guardians of a surviving dependent or incompetent child. This legal document is created as a part of the parents' wills. It is used to provide for the needs of the surviving child that are not covered by other means such as Medicaid. It also covers the expenses incurred by the person named to make care decisions or the person who manages the funds in fulfilling their obligations.

If the child dies before this fund is used, it is no longer valid and the funds remain in the parent's estate. If it is activated and the child dies prior to using all the funds, then, in some states such as North Carolina, any funds left from the set amount in the trust will be used to reimburse Medicaid. Van Herreweghe reports that in the state of New York any funds left are reimbursed to the parent's estate and intended heirs. It is important for parents to obtain information about their state requirements pertaining to disbursement of trust funds.



**Tucker**  
**4/5/2001 – 5/3/2003**  
**Trisomy 13**

*"After Tucker died we talked about adoption... we brought Joey home 5/21/07." [The SOFT Times, 2007]*



**Joey**  
**3/6/2007 - 8/20/2020**  
**Trisomy 13**

Lori Reite, mother of Tucker and Joey, authored *Child of my Heart, the Trisomy 13 Journey, 2024*, available for purchase on Amazon. It is about life with these two boys born with trisomy 13. Quite a few SOFT parents have written books about their trisomy journey. **Their child's story could be a guide to those who come behind.** Many parents find purpose in helping others. For those whose child is no longer here, it is a way of honoring the memory of their child.

### [Loss, Grief and Support](#)

Laden by predicted loss, families wonder how they will endure such heartbreak. Parents who might never before have thought about funerals will eventually have decisions to make. Caring for their child through the end of life, and in memory beyond life, is an inherent part of parenting these children. **It is immensely important to parents that their child be remembered.** Many parents find solace in gathering with other families, who have lost a child, at the **SOFT** conference annual memorial event, where each child's name is read and a candle is lit in their memory.

Knowing ahead of time does not make the devastation of losing a child any easier. Grief is such a difficult/painful journey and each person goes through it in their own way. It is hard to adapt to such a life-altering change and life is never quite the same.

*"Why did he have to die? I wanted him to live. When will the pain end? Will I ever feel joy again? How do I learn to live without him?" [The SOFT Times, 2001]*

*"She will always be Mama's baby and Daddy's Little Girl." K.&R.R., Michelle, trisomy 18*

**Hospice** has classes for bereaved parents and also **loss of a sibling** classes for children too. Places of worship hold memorial services and hospital chaplains provide group memorials. Often teachers arrange a remembrance event with the child's class. Individual counseling or medications for sleep or sadness may be needed, as **grief can be all-consuming.**

*Grieving parents need to allow some time to pass (at least 4 months) after such loss, before joining a bereavement support class, as they will be unable to hear beyond the heartbreak of their own pain.” (advice from a Hospice bereavement counselor)*

Eventually... sorrow softens, wishful “*if only*” thoughts recede... and sweet memories bring comfort. Special dates can be difficult and sometimes the “*if only*” thoughts come and go. It is helpful to talk about your child, especially with other parents who have been there. Some families already belong to a Facebook group with other trisomy families. Many find it helpful to also join a group, specifically for Trisomy Angel Parents [https://www.facebook.com/groups/TrisomyAngelParents/.](https://www.facebook.com/groups/TrisomyAngelParents/)

*“If you are lucky enough to have not lost a child, you don’t understand how hard it is” Beyond Goodbye blog “Grief Shared is Grief Lessened” [www.trisomy.org](http://www.trisomy.org)*

*“There was silence. Silence during the pregnancy. Silence after she was born. Silence when she died. There is still silence around me...but I promised my baby girl I would be a better person, and I’ve promised myself I’d start breaking the silence. **I don’t want other families to go through what we went through. The “incompatible with life” paradigm must change and I’m sure it will change. There’s hope**”. N.K., Maria Paz, t18 (5/19/2016 - 5/26/2016) Paz translates to peace in English.*

The mother of Maria Paz, translated the 2018, 4<sup>th</sup> edition of the Care book from English to Spanish and continues to help Spanish speakers contacting SOFT. Parents who reach out to help other parents, often find it helps themselves too.

## Advocacy

Numerous SOFT parents, at all stages of the trisomy journey, have advocated for changes to the outdated narrative about trisomy 18 and 13, by telling their child’s story, participating in studies, and reporting their child’s surgeries to the SOFT surgery registry, a data source for research about surgery and outcomes.

“SOFT witnessed a landmark moment for our community with the American Academy of Pediatrics’ new guidance on caring for children with trisomy 13 and trisomy 18. This report affirms what SOFT families have long known—that medical care should be individualized and grounded in hope.” NH, {SOFT Newsletter, July 2025)

## [Resources for Parents](#)

### Resources

**The Support Organization for Trisomy 18, 13 and Related Disorder** (SOFT) [www.trisomy.org](http://www.trisomy.org)

**Tracking Rare Incident Syndromes** (TRIS) <http://tris.siu.edu/modules/index.php> The TRIS project seeks to increase the knowledge base on rare incidence trisomy conditions, and to make this information available to families and interested educational, medical and therapeutic professionals. Over 1000 families from around the world have enrolled in the TRIS project, including many SOFT families.

**Chromosome 18 Registry and Research Society** [www.chromosome18.org](http://www.chromosome18.org) This is an advocacy organization with a mission to help people with chromosome 18 abnormalities overcome obstacles they face so they may lead happy, healthy and productive lives. The majority served are those with related chromosome 18 disorders, though those with full trisomy 18 are welcome.

## Facebook groups

**Support Organization for Trisomy** <https://www.facebook.com/groups/TrisomySOFT/>

**Trisomy Angel Parents** <https://www.facebook.com/groups/TrisomyAngelParents/>

**Trisomy 13 Life** <https://www.facebook.com/groups/Trisomy13/>

## Find medication information

**National Institute of Health** (NIH) drug, supplement and herbal information  
<http://www.nlm.nih.gov/medlineplus/druginformation.html>

## Support

**Be Not Afraid** (BNA) <https://benotafraid.net/> a private non profit supporting parents with a prenatal diagnosis carrying to term. Provides individualized support and care of parents at diagnosis, during pregnancy, at birth and during post partum up to one year. BNA Facebook  
<https://www.facebook.com/BeNotAfraid.Net/>

**Now I Lay Me Down to Sleep** [www.nowilaymedowntosleep.org](http://www.nowilaymedowntosleep.org) gives private photo sessions at hospital or hospice to create memory photos at parent request.

**The Compassionate Friends** (TCF) is a national non-profit which offers grief support after the death of a child of any age. [www.compassionatefriends.org/](http://www.compassionatefriends.org/)

**Share Pregnancy & Infant Loss Support Inc** [www.nationalshare.org/](http://www.nationalshare.org/) serves those who had pregnancy loss, stillbirth, or infant loss of life in early months.

**Simon's Law** <https://simonslaw.org/about-us/> 501(c)4 to create national awareness and protection for medically endangered pediatric dependents with life-threatening diagnosis. **Simon's Kids** 501(c)3 advocates for fair and equal access to life saving medical treatment for pediatric dependents with life-threatening diagnoses.

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