

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

"The Care Book" for families

4th edition, January 2018

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

<https://softtrisomy.wpengine.com/>

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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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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.

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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 three 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.**

This eBook, Care of the Infant and Child with Trisomy 18 or Trisomy 13 (2017), **can be downloaded as a PDF file and printed on your computer.** We hope this book will be helpful but **it is not a substitute for the care and advice of your medical practitioner.** 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 and more.

Other SOFT e-Books See Publications at <https://softtrisomy.wpengine.com/>

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:

Preparing for the Arrival of Your Baby by Berg et al., [2015]

http://www.internationaltrisomyalliance.com/uploads/1/4/8/3/14838122/march_6_ita_revised_book.pdf

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

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

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

Video: SOFT Mom & Me Mother's Day Tribute

created by Terre Krotzer, past SOFT Board Member, and mother of Krissy, (trisomy 18, 3/25/00)

Medical textbook recognition of SOFT :

Kyle T; Carman S. 2017. Essentials of Pediatric Nursing, 3rd edition, pg 1087. "SOFT is a support organization for families who have had a child with a chromosome abnormality."



Kari
9/8/1977 – 6/3/1988
Trisomy 18

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



Stacy
5/21/1981
Trisomy 18

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



Joey
9/28/1977 -1/7/1984
Trisomy 13

"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.



Conor
4/2/1986 – 4/9/1986
Trisomy 18

Pam Healey,
 author of
 What Should
 We Do Now (2003)
 (for parents of newly
 diagnosed infants with
 trisomy 18 or 13)
 is the mother of
 Conor.

See
 Publications>
 Pam's Papers
[https://softtriso
 my.wpengine.co
 m/](https://softtriso.my.wpengine.com/)



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**
Balloon Celebration.



Ecaterina, "Cati"
4/26/2011
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

"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]

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 much more serious 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 most will not survive their first year of life. However, **these disorders are not universally lethal, as sometimes described; 5-8% of these infants live past their first birthday without extraordinary measures.** And, once a child's age is greater than a year there is a 60% chance to live beyond age 5 years. (personal communication, Dr. John C. Carey, medical advisor for SOFT, 2011) **A recent 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 and 5-year survival was 12.3% for trisomy 18 and 9.7% for trisomy 13.** [Myer et al., 2016]

Beginning in the sixties, chromosome analysis was done by Karyotype (cells arranged in a specific order, photographed and counted). Karyotype remains useful today for trisomy 18 or trisomy 13. Since the mid-nineties the maternal serum triple/quad SCREENING analyzing a mother's hormone level for the **possibility** of a trisomy condition has been a 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)** 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 doctors might not know about **PPV** and thus it would be appropriate to share the following link about the calculator with your doctors.

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. Currently some companies state in their literature that NIPT is only a SCREENING and **a positive result needs confirmation by invasive testing.** 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. 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. See [Preparing for the Arrival of your Baby](#) [Berg et al., 2015] to help discuss perinatal care with your doctor. In recent years, expectant parents on Facebook post about having to search for a supportive Obstetrician or Maternal-Fetal-Medicine 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. **A referral to the new perinatal palliative care programs, offering guidance for creating a birth plan, end of life wishes and care options/decisions for the live born infant is occurring more often for those who continue their pregnancy.** These programs are advisory, sometimes done by phone, and participation is **optional**.

Diagnosis and Parents

The plight 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 deal 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 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 judgement 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 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., 2015; Kosho & Carey, 2016]

Surgeries are being done today that were not available thirty years ago, such as cardiac repair. Finding a surgeon willing to do a cardiac repair is difficult in many locations but is becoming more available. See current list of hospitals that provided cardiac repair/procedure in last decade on page 50. Some parents report having to meet with a **Hospital Ethics Committee** to obtain approval for a heart repair for their child. Ethics committees are composed of a bioethicist, physician, clergy, and other pertinent health care professionals. Consensus for or against a procedure is a responsibility shared by the committee and doctor presenting the case. Parents should also be a part of this meeting. **It is necessary and appropriate for parents to voice their opinion as their wishes are significant to the decision making.**

"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 Times, 2006]



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

Joanna lived 38 years, 3 months, 20 days; **one of** the longest lived SOFT members with trisomy 18.



Erin
2/22/78 – 3/19/2013
Trisomy 13

Erin lived 35 years, 25 days; the longest lived female SOFT member with trisomy 13.

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



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

Donnie lived 22 years, 9 months, 11 days; the longest lived male SOFT member with trisomy 18.

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 approximately 250 living persons with trisomy 18 and 13 older than one year in the USA. (personal communication, 2014) 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, 1992]

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 guidebooks for families referenced on page 8.

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. See Diagnosis page 11.

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, 2010] 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, 2010] 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

Deviation of hand at ulna or radius (forearm bones)

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)

Cleft lip or palate or both (10-20%)

Low Occurrence (<10%)

Diaphragmatic hernia

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 2013; 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, 2013]

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, 2010]



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

Barb Farlow, one of the founding parents of the **International Trisomy 13/18 Alliance (ITA)**, is the mother of Annie.

Annie's mother co-authored:
The Experience of Families with Children with Trisomy 13 and 18 in Social Networks. Janvier A, Farlow B, Wilfond B. 2012. Am Academy of Pediatrics

Preparing for the Arrival of your Baby

by Berg S, Farlow B, Robbins J, Bruns D.
(An ITA e-Book for expectant parents)

Developmental Achievements

Individual case studies published during 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 first 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, 2010]

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 most of 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:

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.

[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 one child with trisomy 18 has been seen at SOFT conferences.

Although individuals with trisomy 18 and trisomy 13 were clearly functioning in the severe to profound range of developmental disability, they did achieve many skills of childhood, 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 that 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, 2010]

“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

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

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 page 19, Table 3.

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]

Healey also discussed her survey of 20 children with related disorders of **mosaic or partial trisomy 18 or 13**, age range of 25 months - 17 years, and noted better overall functioning than those with full trisomy 18 or 13, but some functioned at levels consistent with full trisomy. In those with related disorders 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 therapy are available to children who meet criteria for **early intervention** programs or **special education** at public school. Those 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 will need a referral from the child's doctor and should check with the clinic billing office to determine how these services will be reimbursed.

Available in English and Spanish, SOFT books Trisomy 18 Handbook for Families and Trisomy 13 Handbook for Families provide information about early intervention and public school education. Parents can contact their local department of education for details about registration and location of the school their 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. Arrangements to visit the classroom, meet the teacher and whoever else may interact with their child can be made by parents. Most families have mixed feelings about sending their child to school.

The uncertainty of survival complicates the parent's decision to enroll their child in an early intervention program. Yet, most will find that participation in these services benefits both the child and parent. Parents in these programs are often supportive of one another and share helpful information. **Referral to early intervention is recommended in the ongoing care of infants and children with trisomy 18 and trisomy 13. [Carey, 2010]** With guidance from these programs, along with time and love these children develop their own personalities learn to respond positively to caregivers, learn to indicate preferences in toys and slowly acquire skills that allow achievement of some milestones.

"Sending Joseph off to school was something we never dreamed we would see. After five years of holding so tight, it was difficult at first to relinquish him to school. But he loves it, and I have more freedom." M.S., Joseph, trisomy 18

"We asked the school to send a teacher to our home after a long hospital stay. She would have gone into high school when she recovered, but we decided it was best to continue with the teacher and therapist coming to our home and the school agreed to do so." A.B., Megan, trisomy 18



Morghan
9/14/1996
Trisomy 18

"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

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



Lunah
12/3/2015
Trisomy 18

Table 4**Developmental Achievements in Trisomy 18 and Trisomy 13**

Trisomy 18	Av. Mos. (SE)	Range	N	Normal Range
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	
<hr/>				
Trisomy 13	Av.mos. (SE)	Range	N	Normal Range
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 & Sons, Inc.

Table 5 Common Illnesses and Health Problems

Prevalence of Common Illnesses & Health Problems in Trisomy 18 or Trisomy 13	
<u>Frequent Incidence</u>	
Apnea	Photophobia (18)
Congenital Heart Defects	Pneumonia
Constipation	Pulmonary Hypertension
Ear Infections	Scoliosis
Elevated Blood Pressure (13)	Seizure
Feeding difficulties	Sinusitis
Gastroesophageal (GE) Reflux	Urinary Tract Infection
<u>Less Frequent Incidence</u>	
Cataracts in children with trisomy 13	
Glaucoma in children with trisomy 13	
Wilms Tumor (kidney cancer) in children with trisomy 18	
Other anomalies	
<u>Low Incidence</u>	
Hepatoblastoma (liver tumor) in children with trisomy 18	
[source: SOFT Registry, the SOFT touch, The SOFT times, 1988-2014]	

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

The three main categories of apnea are Central, Obstructive (see page 51) **or Mixed; a combination of both central and obstructive 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, cyanosis, pallor, and/or marked hypotonia." **Prematurity is the most common cause of infant central apnea but it is important to consider other causes.**

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] 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)

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Physicians should keep epileptic apneas in mind when treating apneas in neonates with trisomy 18. [Fukasawa et al. 2015] In the September 2016 SOFT Times newsletter Dr Steven Cantrell lists **visual triggers** mentioned by adult patients with photosensitive epilepsy **that cause seizures or occasional apnea.** He suggests the same triggers might affect children with trisomy 18 or trisomy 13 and advises that these children wear protective glasses when with family around computers, television, cell phones and other LED technology. These are known triggers for children at risk. (See page 35 below)

Seizures

Seizures are a functional nervous system problem occurring in about 25% to 50% of older infants and children with trisomy 18 or trisomy 13. [Carey, 2010] Photosensitive epilepsy (PSE) is common in trisomy 18 and trisomy 13. [Cantrell, SOFT Times newsletter 2016] At [https://softtrisomy.wpengine.com/openPublications> Newsletter Library >September 2016, page 35 for Photophobia and Photosensitive Epilepsy.](https://softtrisomy.wpengine.com/openPublications%20NewsletterLibrary%20September%202016%20page%2035%20for%20Photophobia%20and%20Photosensitive%20Epilepsy)

It is possible that a small amount of stomach contents might be refluxed or vomited during a seizure, temporarily obstructing the airway or risking aspiration. 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 appropriate, referral to a pediatric **neurologist** can be made by the child's doctor. 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.

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 brains hemispheres are partially divided) or alobar (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, Dr.

Steven Cantrell, 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 often report the use of a custom-made orthopedic body jacket to prevent or slow further progression of scoliosis, ankle foot orthotics (**AFOs**, also known as shin splints), and eye patches or glasses to strengthen eye muscles as prescribed by specialists for their child. Physical, occupational, speech, music, and vision therapies are reported by parents as benefitting their child.

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 food and milk. 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 28)

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.**

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). See page 26. 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 often 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." 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, 2013] 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 > 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 (SOFT-SR)** 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]



Sofie Marie
1/24/2014
Trisomy 13

"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



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

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

Head Lag

Many infants/babies with trisomy 18 or trisomy 13 frequently hyperextend their head or let their head lag towards their back. Such posturing can make swallowing more difficult. 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.

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 29) causes discomfort and is a common problem in trisomy 18 or trisomy 13. Gastroesophageal (GE) reflux (see page 28) 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 below) 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. **Very few of these infants and children 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, some 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) and some kinds of nourishment, when given by mouth, 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 currently 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 62. **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**", was signed by Kansas Governor Brownback on April 7, 2017 to take 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. Texas Governor Abbott signed a similar law August 16, 2017 to take effect April 1/2018.



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.

See page 8 for link to purchase

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, 2013]. 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 JC 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.

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

“When about 8 months she was prescribed Lactulose, but sometimes, she also needed Babylax. In her teens, she was prescribed MiraLax and it worked well for her.” A.B., Megan, trisomy 18

“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.

“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 75% success for voiding and 80% for BM's." AB, Megan, trisomy 18

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. [DDHealth.info.org] 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-SR which is found on the SOFT website at <https://softtrisomy.wpengine.com/>. For example: bowel obstruction repair or resection, and bowel malrotation repair show a combined total of 22 children with trisomy 18, and 37 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 9 children with trisomy 18, and one with trisomy 13.

Be aware that 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.) [DDHealthInfo.org] **When soft stool is achieved it is important to continue daily maintenance, as prescribed, to prevent reoccurrence 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 small possibility of being caretakers of a child with disabilities. "Hope is what got us through it all." [SOFT mother D.D., 2013] Keep in mind that even those in palliative care or Hospice programs might live longer than predicted. A few SOFT families report their child was discharged from these programs 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. Documented information from a well-baby or child physical is referenced by health providers when asked to complete health forms. 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 44-46. 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.** See Resources at <https://softtrisomy.wpengine.com/>.

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

"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]

"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, 2005] Dr. Steven D. Cantrell, optometrist, reports that many non-verbal vision tests exist that can accurately estimate the prescription for glasses and parents should **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 annual 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 and SOFT newsletter article Vision, Light Sensitivity, Headaches and Seizures, [SOFT Times, September 2016]. Go to <https://softtrisomy.wpengine.com/> >Publications to find the SOFT Times Newsletter Library.

Photophobia and Photosensitive Epilepsy

Photophobia (painful eye response to bright light) occurs in trisomy 18 from early infancy on, as these children have reduced levels of macular pigment, which leads to painful eye sensitivity. Cantrell reports recent study findings that some children with trisomy 13 were diagnosed with Photosensitive Epilepsy (**PSE**). Spagnoli et al., (2015) <https://www.ncbi.nlm.nih.gov/pubmed/25459971> 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]

Known triggers, some listed below, of seizures and occasional apnea in adults with PSE might have a similar effect on children with trisomy, if eyes are not protected. [Cantrell, 2016]

- LED bulbs (Big box stores often have eye offensive white, or blue/white LED lighting.)
- Turning bright lights on and off in a room or flashing lights
- Flickering light through ceiling fans, or sunlight through trees
- New flat screen LED televisions, and all technology screens (Reduce screen brightness, if possible.)
- Baby/child enjoying family time together around TV and other technology, **if eyes not protected**

Cantrell encourages eye healthy nutrition, eyewear lenses (see box), and Ophthalmics to protect eyes. DAILY intake of the antioxidants lutein and zeaxanthin which naturally filter harmful high-energy blue wavelengths of light protects eye health. Leafy green and yellow vegetables provide these antioxidants. One SOFT mother juiced these greens for her daughter with trisomy 18 and her photophobia went away.

BluTech Dark Medical Lenses Are Best for Children with Trisomy 18 or 13

Cantrell recommends **BluTech medical sunglass lenses** to be worn outdoors and when around indoor technology such as televisions and computers. All technology may trigger a seizure with unprotected eyes. In addition these lenses should be worn while traveling with parents in a car, in the mall, or other brightly lit big box stores. Unprotected eyes are more likely to develop insomnia and increase risk for night time seizure with potential apnea. Wearing **darker BluTech sunglass lenses** indoors will reduce symptoms and increase comfort. Adults report severe **headaches** after a seizure which can be assumed occurs after a seizure with children born with Trisomy 18 and 13. Ask your child's doctor about safe headache remedies. (personel communication Cantrell, 2017)

Kodak BluTech: www.BluTechLenses.com or call for a local source at 800-258-5902

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]

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 the SOFT-SR 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 foods such as spinach, kale, and collard greens naturally reverses light sensitivity and cataracts. Cataracts will interfere with normal vision, and if advanced, may cause a substantial loss of vision. [personal communication, Cantrell, OD, 2007] 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-SR for trisomy 18 is strabismus surgery; reported 6 times more for trisomy 18, than trisomy 13. "Uncorrected high prescription hyperopia 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 developed in her early months. When she lived past her first birthday, the ophthalmologist said she was a "survivor" and did strabismus repair. He cautioned that another form of strabismus, exotropia (eye turn outward), could result from the repair which did happen in one eye about 4 months after her surgery." A.B., Megan, trisomy 18

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-SR lists glaucoma surgery for 9 children with trisomy 13; 0 for trisomy 18.

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.

Micro-Ophthalmus (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 U.S. require mandatory screening of hearing in a newborn. Infants who do not pass screening receive referrals for follow up testing. In states or regions where universal newborn screening of hearing does not occur, referral to a hearing specialist is important. Testing with a pediatric audiologist would be best for those who are difficult to evaluate. **In trisomy 18 and trisomy 13, a hearing evaluation after 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 and 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.

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

Hearing

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.

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.

Mosaic or Partial trisomy 18 or 13 (of 20 children)

25% had partial hearing and a few had profound hearing loss.

Vision

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.

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.

Mosaic or Partial Trisomy 18 or 13 (of 20 children)

Vision was fine in nearly 75% of those with mosaic or partial trisomy.
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]

A few SOFT Facebook moms report their child uses Soft Bands (**Baha and Pronto Soft**), a head band containing hearing devices.

"...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

"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 have noticed 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 health care provider should refer to the AAP recommendations. Medicaid has a list of required immunizations for their recipients that follows AAP recommendations.

Immunizations should be determined on an individual basis. [Bruns, 2014] Some children may be exempted due to religious reasons or adverse reaction to a prior vaccine or need a delay due to illness or other concerns. Two SOFT mothers wondered if their infants low weight (<5 pounds) might have contributed to their infants post vaccine reaction. 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 (SOFT-IR) maintains information provided by parents about their child's vaccine history. Some parents delayed or declined vaccines but of those vaccinated the majority received the standard immunizations and a yearly flu vaccine. Parents also reported if any reactions occurred. Most reactions were typical such as fever (only), redness or lump at site or not-specified but more concerning or adverse reactions also happened. Very few named the vaccine that might have caused the reaction. None mentioned if their child's reaction had been reported to the Vaccine Adverse Event Reporting System (VAERS) - CDC.

SOFT Immunization Registry (SOFT-IR) of parent reported reactions as of Dec 2016

- Of 204 with T18 who received a vaccine 25 had a reaction; 12 typical, 13 concerning or adverse.
- Of 82 with T13 who received a vaccine 6 had a reaction; 3 typical, 3 concerning or adverse.
- Also one child with mosaic trisomy 13 had a concerning reaction.

Reported Concerning or Adverse Reactions:

- Illness- T18 = 8; T13 = 2
- Seizure T13 =1
- Apnea/breathing episode- T18 = 4
- Fever, seizure T13M = 1
- Seizure, death- T18 = 1 (Japan)

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.

Muscle and Skeletal

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 16-17.) 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.

Scoliosis

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, 2005]** 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. Curvature > 20 degrees is usually treated with a custom fit orthopedic brace called a body jacket; used to delay or arrest progression, sometimes worn for years with refitting for growth. **If curvature progresses >40 degrees, surgery is usually discussed.** Further progression to more severe curvature, gradually diminishes lung capacity leading to long term heart and

lung complications. A number of children in the SOFT Surgery Registry had a spinal fusion at <10 years of age, but most orthopedic surgeons prefer delaying to 10 years or greater due to continued growth that might cause need for further repair. In recent years a few parents mentioned via Facebook that **Adjustable Growing Rods (spine or rib) were used to delay a spinal fusion until their child was older.** 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., Eleanor, trisomy 18, 2012

*Vertical Expandable Prosthetic Titanium Rib (VEPTR) **Growing Rods.**

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

Several 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. Those with significant renal defects should be followed for infection and renal insufficiency. [Carey, 2010] See page 55 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.

Dr Carey suggests asking each professional who treats 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.



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. 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

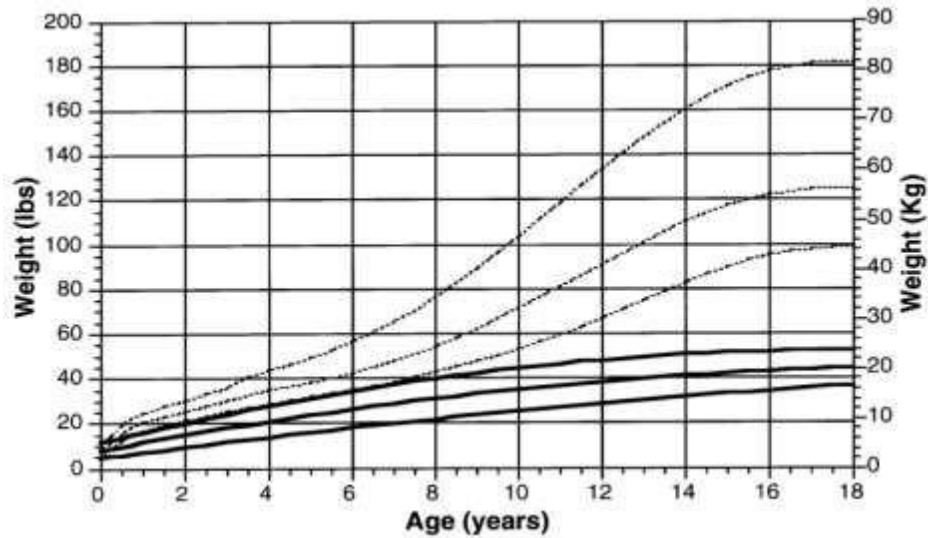


Emerson
6/23/2011
partial Trisomy 18q and partial Monosomy 13p translocation

Table 8a Growth Curves for Trisomy 18 and 13

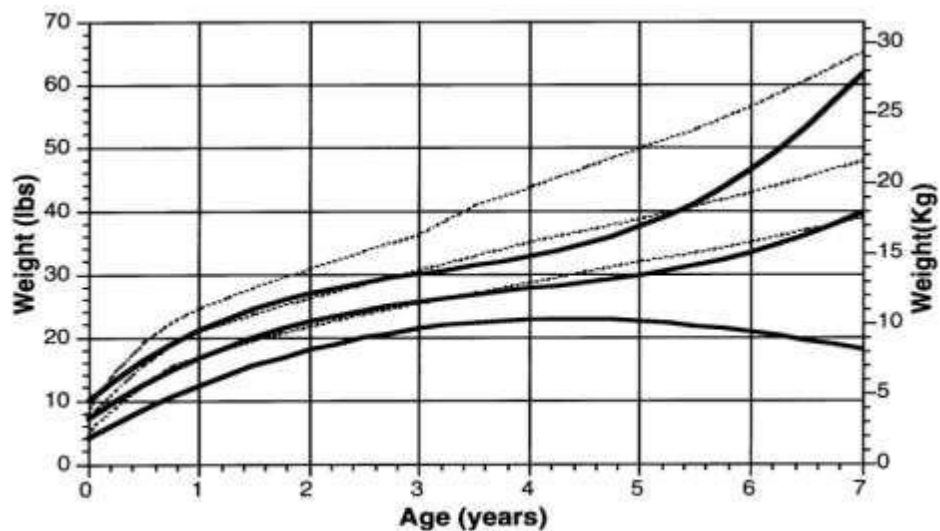
Trisomy 18 Weight Curve

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)

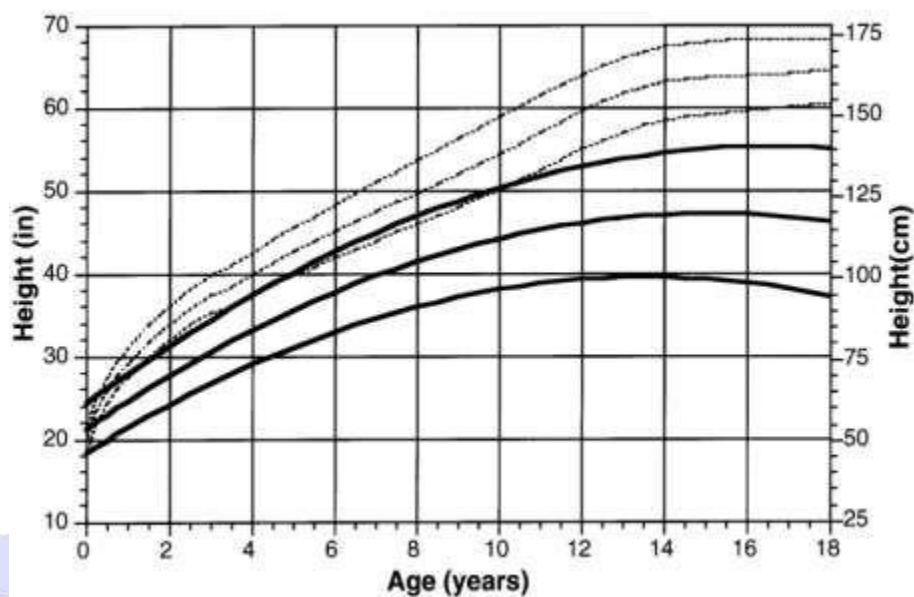


Source for weight curves: Baty BJ, Blackburn BL, Carey JC (1994) Natural history of trisomy 18 and trisomy 13: I. Growth, physical assessment, medical histories, survival and recurrence risk. Am J Med Genet 49: 175-188, © 1994 Wiley-Liss, Inc. Reprinted by permission of John Wiley & Sons, Inc.

Table 8b Growth Curves for Trisomy 18 and 13

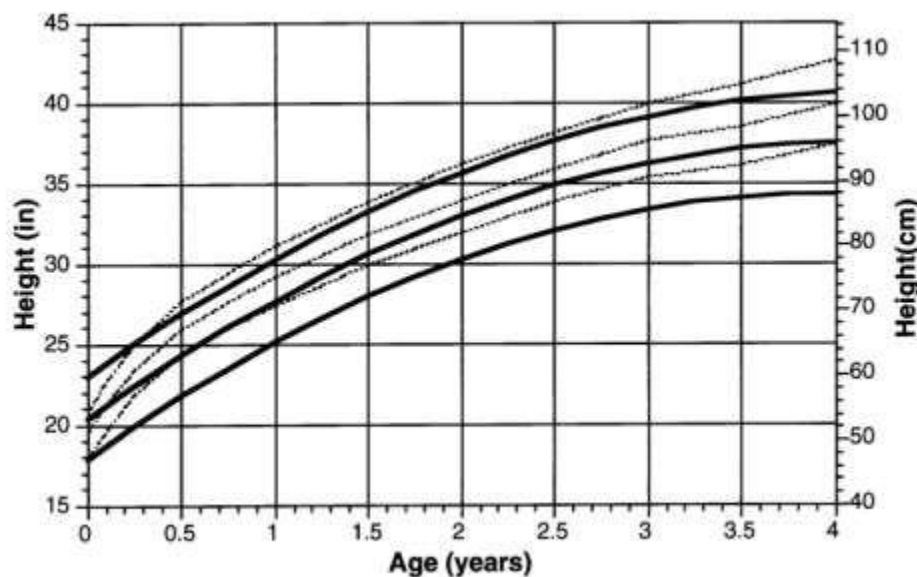
Trisomy 18 Height Curve

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)

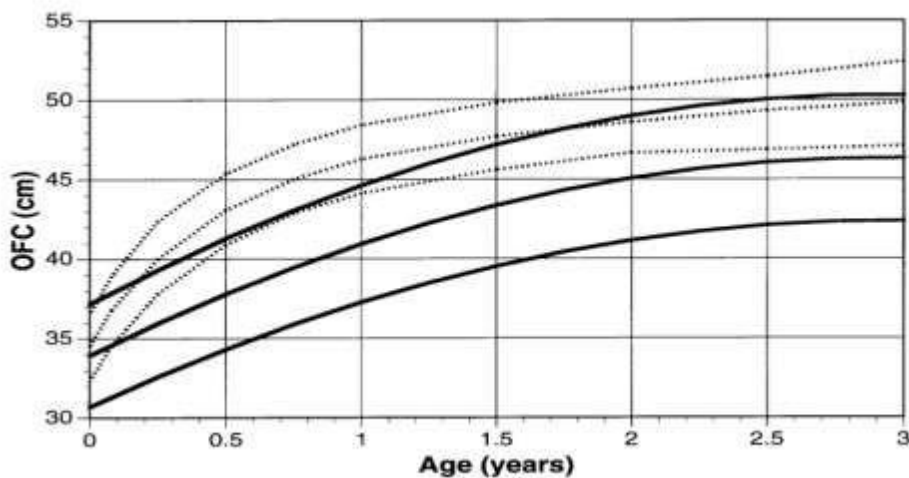


Source for height curves: Baty BJ, Blackburn BL, Carey JC (1994) Natural history of trisomy 18 and trisomy 13: I. Growth, physical assessment, medical histories, survival and recurrence risk. Am J Med Genet 49: 175-188, © 1994 Wiley-Liss, Inc. reprinted by permission of John Wiley & Sons, Inc.

Table 8c Growth Curves for Trisomy 18 and 13

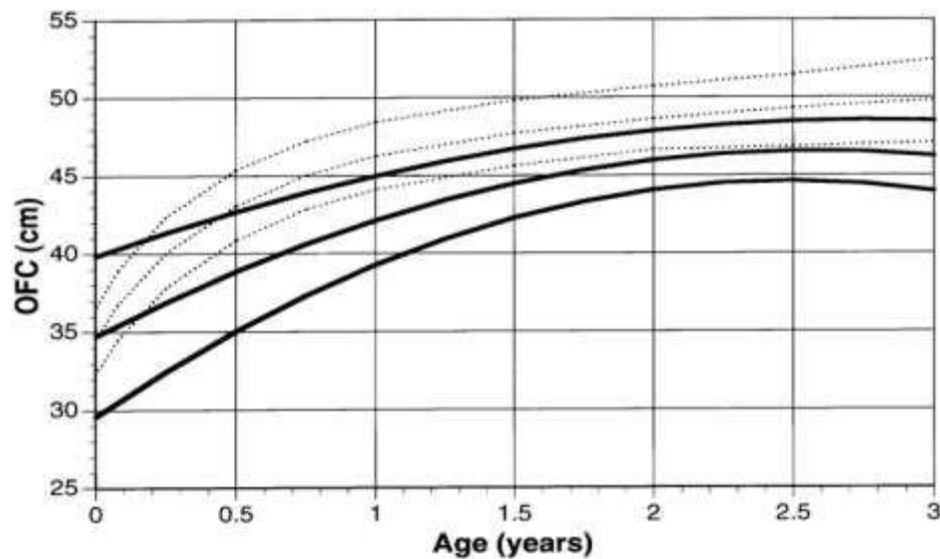
Trisomy 18 Head Circumference Curve

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

"An Ethics doctor said the attitude is changing for the positive toward trisomy 18." A.S., Janessa, trisomy 18, 2014

"The many surgeries that were done to Joey during his six short years were always to improve the quality of his life. We believe they did." P.F., Joey, trisomy 13

Table 9 Cardiac Surgery

The SOFT Surgery Registry (SOFT-SR Oct 2017)						
Repair or procedure & number done by diagnosis	T18	T18M	T13	T13M	RD	Total
CARDIAC: AORTA WIDENED	1	0	0	0	2	3
CARDIAC: AORTIC COARCTATION REPAIR	9	0	3	1	1	14
CARDIAC: AORTIC VALVE STENT	0	0	1	0	0	1
CARDIAC: ASA REPAIR (atrial septum aneurysm)	1	0	0	0	0	1
CARDIAC: ASD REPAIR	33	8	10	3	17	71
CARDIAC: AVSD REPAIR (Atrioventricular Septal Defect)	3	0	0	0	1	4
CARDIAC: BALLOON DILATATION	3	0	0	0	0	3
CARDIAC: BICUSPID AORTIC VALVE SURGER	1	0	0	0	1	2
CARDIAC: BLAYLOCK-TAUSSIG	3	0	1	0	3	7
CARDIAC: CARDIAC CATHETERIZATION	17	2	2	2	1	24
CARDIAC: CARDIAC COIL PROCEDURE	2	0	2	0	1	5
CARDIAC: DOUBLE OUTLET RT. VENTRICLE	3	0	0	1	1	5
CARDIAC: ECMO	1	0	0	0	0	1
CARDIAC: FONTANA PROCEDURE	1	0	0	0	0	1
CARDIAC: GLENN PROCEDURE	0	0	0	0	1	1
CARDIAC: HLHS PDA STENT	1	0	0	0	0	1
CARDIAC: MITRAL VALVE REPAIR/LEAK CORRECTION	1	0	1	0	1	3
CARDIAC: PACEMAKER	6	2	0	0	0	8
CARDIAC: PDA REPAIR*	47	4	7	5	15	78
CARDIAC: PERICARDIECTOMY	0	0	0	0	1	1
CARDIAC: PFO CLOSURE	2	0	0	0	0	2
CARDIAC: PULMONARY ARTERY BAND REMOVAL / LOOSENING	5	1	1	0	1	8
CARDIAC: PULMONARY ARTERY BANDING	19	2	4	0	3	28
CARDIAC: PULMONARY ATRESIA	5	0	0	0	1	6
CARDIAC: PULMONARY STENOSIS REPAIR	3	1	0	1	1	6
CARDIAC: PULMONARY VEIN REPAIR	1	1	0	0	0	2
CARDIAC: ROSS PROCEDURE	0	0	0	1	0	1
CARDIAC: SHUNT PLACEMENT	2	0	1	0	1	4
CARDIAC: SURGERY, UNSPECIFIED	5	2	0	3	5	15
CARDIAC: TAPVR**	0	0	1	0	1	2
CARDIAC: TETROLOGY OF FALLOT REPAIR	7	2	5	0	6	20
CARDIAC: TRUNCUS ARTERIOSUS REPAIR	0	0	0	0	1	1
CARDIAC: VSD REPAIR	66	10	9	3	22	110

T = trisomy, M = mosaic, RD = related disorders (SOFT-SR)

*Patent Ductus Arteriosus

**Total Anomalous Pulmonary Vessel Return

Surgery table above and Hospital table below are dynamically updated on SOFT website.

"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]

Table 10 Trisomy 18, 13 - Cardiac Surgery Hospitals

Hospitals that provided cardiac surgery or procedure since 2007			***
for full T18 or 13 and most recent year done			
Some hospitals listed might no longer provide repair for T18 or 13.	year	state	
STOLLERY CHILDREN'S HOSPITAL, EDMONTON, AB, CANADA	2014	AB	
CHILDREN'S OF ALABAMA, BIRMINGHAM, AL	2017	AL	
ARKANSAS CHILDREN'S HOSPITAL, LITTLE ROCK, AR	2007	AR	
SAN DIEGO CHILDREN'S HOSPITAL, SAN DIEGO, CA	2012	CA	
LUCILE PACKARD CHILDREN'S HOSPITAL - STANFORD, PALO ALTO, CA	2014	CA	
UCSF BENIOFF CHILDREN'S HOSPITAL, SAN FRANCISCO, CA	2016	CA	
UC DAVIS MEDICAL CENTER, SACRAMENTO, CA	2013	CA	
CHILDREN'S HOSPITAL COLORADO, AURORA, CO	2017	CO	
CHILDREN'S NATIONAL MEDICAL CENTER, WASHINGTON, DC	2012	DC	
A.I. DUPONT CHILDREN'S HOSPITAL, WILMINGTON, DE	2012	DE	
ST JOSEPH'S CHILDREN'S HOSPITAL, TAMPA, FL	2007	FL	
WOLFSON CHILDREN'S HOSPITAL, JACKSONVILLE, FL	2011	FL	
CHILDREN'S HEALTHCARE OF ATLANTA EGGLESTON, ATLANTA, GA	2013	GA	
TUBINGEN KINDERKLINIK, TUBINGEN, GERMANY	2017	de	
KAPIOLANI MEDICAL CNTR FOR WOMEN & CHILDREN, HONOLULU, HI	2013	HI	
ST FRANCIS MEDICAL CENTER, PEORIA, IL	2009	IL	
ADVOCATE LUTHERAN GENERAL HOSPITAL, PARK RIDGE, IL	2009	IL	
PAYTON MANNING CHILDREN'S - St VINCENT'S, INDIANAPOLIS, IN	2016	IN	
NORTON CHILDREN'S HOSPITAL, LOUISVILLE, KY	2016	KY	
CHILDREN'S HOSPITAL OF NEW ORLEANS, NEW ORLEANS, LA	2016	LA	
BOSTON CHILDREN'S HOSPITAL, BOSTON, MA	2011	MA	
JOHNS HOPKINS HOSPITAL, BALTIMORE, MD	2014	MD	
C.S. MOTT CHILDREN'S HOSPITAL, UM, ANN ARBOR, MI	2014	MI	
CHILDREN'S HOSPITAL OF MICHIGAN, DETROIT, MI	2010	MI	
CHILDREN'S HOSPITAL OF MINNESOTA, MINNEAPOLIS, MN	2007	MN	
CARDINAL GLENNON MEDICAL CENTER, ST LOUIS, MO	2017	MO	
CHILDREN'S MERCY HOSPITAL, KANSAS CITY, MO	2017	MO	
CAROLINAS MEDICAL CNTR-LEVINE CHILDREN'S, CHARLOTTE, NC	2015	NC	
CHILDREN'S HOSPITAL & MEDICAL CENTER, OMAHA, NE	2017	NE	
PRESBYTERIAN HOSPITAL, ALBUQUERQUE, NM	2011	NM	
SUNRISE HOSPITAL - CHILDREN'S HEART CENTER, LAS VEGAS, NV	2017	NV	
NATIONWIDE CHILDREN'S HOSPITAL, COLUMBUS, OH	2016	OH	
CHILDREN'S HOSPITAL OF PITTSBURGH, PITTSBURGH, PA	2008	PA	
CHILDREN'S HOSPITAL OF PHILADELPHIA, PHILADELPHIA, PA	2013	PA	
PENN STATE HERSHEY CHILDREN'S HOSPITAL, HERSHEY, PA	2013	PA	

CHILDREN'S HOSPITAL OF PHILADELPHIA, PHILADELPHIA, PA	2014	PA
GREENVILLE MEMORIAL CHILDREN'S HOSPITAL, GREENVILLE, SC	2007	SC
MEDICAL UNIVERSITY OF SOUTH CAROLINA, CHARLESTON, SC	2017	SC
LeBONHEUR CHILDREN'S HOSPITAL, MEMPHIS, TN	2017	TN
TEXAS CHILDREN'S HOSPITAL, HOUSTON, TX	2016	TX
COOK CHILDREN'S MEDICAL CENTER, FORT WORTH, TX	2014	TX
CHILDREN'S HOSPITAL OF SAN ANTONIO, SAN ANTONIO, TX	2009	TX
MEDICAL CITY DALLAS HOSPITAL, DALLAS TX	2014	TX
METHODIST CHILDREN'S HOSPITAL, SAN ANTONIO, TX	2015	TX
UNIVERSITY HOSPITAL, SAN ANTONIO, TX	2013	TX
TEXAS CHILDREN'S HOSPITAL, HOUSTON, TX	2010	TX
BIRMINGHAM CHILDREN'S HOSPITAL, WEST MIDLANDS, ENGLAND	2015	UK
PRIMARY CHILDREN'S MEDICAL CENTER, SALT LAKE CITY, UT	2015	UT
UNIVERSITY OF VA MEDICAL HOSPITAL, CHARLOTTESVILLE, VA	2012	VA
ROANOKE MEMORIAL HOSPITAL, ROANOKE, VA	2008	VA
CHILDREN'S HOSPITAL OF THE KING'S DAUGHTERS, NORFOLK, VA	2011	VA
SEATTLE CHILDREN'S HOSPITAL, SEATTLE, WA	2017	WA
CHILDREN'S HOSPITAL OF WISCONSIN, MILWAUKEE, WI	2016	WI
RUBY MEMORIAL HOSPITAL, MORGANTOWN, WV	2013	WV
SUNNINGHILL HOSPITAL, GAUTENG, SOUTH AFRICA	2015	ZA
SOFT-SR Nov 2017 www.trisomy.org		



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

"How can any kind of hope be false, when it is all you've got? Hope cannot be false as far as I am concerned. Having that little bit of hope got us through it all." D.D., 2013

Mortality

"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. 2015; Kosho & Carey, 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

"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]

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 22.
- **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**. Lack of adequate sleep contributes to irritability in the infant. Sleep-deprived parents are tired parents and the daily life of the family is affected.

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. However, tolerance for breathing through a mask or nasal pillows can be a discomfort issue, especially for children.

It is of note that reports to SOFT of tracheostomy surgery for both trisomy 18 and trisomy 13 has 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 have chosen 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.



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

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 learn 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. See <http://www.medgadget.com/2013/01/philips-respironics-coughassist-t70>

Scoliosis is a common development in trisomy 18 and those with trisomy 13 might also develop this problem. Scoliosis often begins in early childhood but signs were also noted in an older infant with trisomy 18. Scoliosis usually progresses with time, and growth; resulting in restriction of the lungs and poor pulmonary function. Pneumonia is a serious illness for someone with restrictive lung disease and can be life threatening. See page 42.

Cardiac

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" or cardiac 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 present at birth. 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 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 is an impression that this may develop early for those with trisomy 18. [Carey, 2010] 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 might 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 slowly 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-SR maintains a list of parent reported cardiac repairs and hospitals where these repairs were done. See pages 48-50. **Be aware-** Hospital policies can change or a willing surgeon might relocate so some hospitals listed might no longer do 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]

"Her cardiologist was surprised because her large VSD had closed on its own." [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]

Using the SOFT-SR, 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. See entire paper at <https://softtrisomy.wpengine.com/> under Professional tab. "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 Castello 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]

Neoplasia (Tumor)

Older infants and children with trisomy 18 are at increased risk to develop Wilms tumor (kidney tumor) and hepatoblastoma (liver tumor). Development of Wilms in trisomy 18 is typically after 5 years of age and the oldest known child was 13 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]

In recent years Carey recommended screening for tumor by abdominal ultrasound in trisomy 18. Screening should start after the age of six months and be done every six months. The actual risk of developing Wilms tumor is small (likely <1%). It is not known when to stop screening so performance of abdominal sonogram until 15 years seems prudent. [Carey, 2010]

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 7th birthday and Hepatoblastoma through the 4th 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 4th birthday. Trisomy 13 was not looked at in the AACR study. These recommendations are simply a carry-over from guidelines for screening in other conditions and **not** specific for trisomy 18. **See current recommendations below in Table 11.** These will be revised in the future as current research is published. (personal communication, Carey, 2017)

Tumor Profile in Patau Syndrome (trisomy 13) by Satge D et al., (2017), and Tumor Profile in Edwards Syndrome (trisomy 18) Satge D et al., (2016), were recently published in the American Journal of Medical Genetics.

“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]

Issues in Care

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.

“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]

“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

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. **In the next chapter Parents and Life Support Order Decisions are discussed.**

The existence of three advance directive documents plus the in-hospital life support order form might create confusion for the public. The fact that forms and form titles can vary from place to place adds to the lack of understanding about end-of-life directives and what constitutes a life support order that will be followed by medical persons. 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, such as Uniform Do Not Resuscitate, or Portable DNR. 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, to show proof of consent. 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 Order 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, might be only briefly addressed as “Do you want her on a ventilator?” Ideally, 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.

When the question about life support is poorly asked, without an explanation about procedures or without showing the form to the parent, **a parent might not even be aware an order is being created.** 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 would be wise 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.

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.

A positive prenatal screening or diagnosis results in expectant parents searching the internet for information about these syndromes. Finding resources such as SOFT and ITA, 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

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

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



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

"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.



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

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

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

Infants (0-1)

Manifestation/Theme	Diagnostic or Screening Test/ Referral
Congenital heart defects	Cardiac evaluation, including echocardiogram in newborn period
Developmental disability	Referral to early intervention program 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
Chronic Constipation	Referral to Gastroenterology where appropriate
Hearing loss	Audiology 6-8 months of age; follow-up as needed
Neurologic/ Central nervous system	Neuroimaging in newborns and infants with trisomy 13 as needed
Ocular malformations	Routine in trisomy 13; as indicated in trisomy 18
Neoplasia (Wilms tumor/ Hepatoblastoma)	Abdominal ultrasound at birth or when diagnosed, done every 3-4 months to age 7 years and every 6 months until age 12; AFP every 3-4 months up to age 4 years for those with trisomy 18
Other respiratory difficulties	Referral to Pediatric Pulmonology where appropriate
Genetic issues	Referral for medical genetics consultation and genetic counseling
Family coping	Family counseling as needed; referral to support group, local, national SOFT; distribute written information

Older Children

Developmental disability	Referral to special education and other educational resources (PT, OT, speech)
Urinary tract infection	High risk of suspicion; evaluate when needed
Scoliosis (especially in trisomy 18)	Follow closely; referral to Orthopedics as indicated

[Carey, 2017]

Carey also recommends other well-child care as would occur in any infant or child. This would include routine immunizations including varicella. RSV immunoglobulins are recommended at the beginning of the RSV season in appropriate infants with trisomy 18 or 13. This particular approach is usually carried out for infants with chronic disorders and history of prematurity. [Carey, 2005] Recommendations in table 11 for health care providers are also in Carey's chapter in Allanson and Cassidy's book (2010) and in Cerda and Carey (2012). Wilson and Cooley provide a similar working checklist.

Managing Total Care

No matter what their age, those with trisomy 18 or trisomy 13 require continual supervision and total care. Often one parent is the primary caregiver with the other assisting. While family and friends can be helpful, for many caregivers their first time with any significant hours away from their child is when their child starts public school. It can be difficult for a primary caregiver to relinquish care, yet "caring for the caregiver" is 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.

"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]

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

It is common 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 significant 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 provides only comfort care to ease symptoms but no interventions for a cure. 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 small 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.

Social Services

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 as the **Association for Retarded Citizens (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

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 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 his 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 rollercoaster 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. 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



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 highpoint 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 (NDNR) 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 18th 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.

This 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
Trisomy 13

Loss, Grief and Support

"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]

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 **Balloon Celebration**, a memorial event, where each child is named and a balloon is released 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.

"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)

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**. 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 who understand. Many find it helpful to also join a group, specifically for those who've lost a child, at **Trisomy Angel Parents** <https://www.facebook.com/groups/TrisomyAngelParents/>

"There was silence. Silence during the pregnancy, silence after she was born, and silence when she died. There's 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, (2018)

Resources for Parents

Resources

The Support Organization for Trisomy 18, 13 and Related Disorder (SOFT) <https://softtrisomy.wpengine.com/>
International Trisomy 13/18 Alliance (ITA) www.internationaltrisomyalliance.com/
Tracking Rare Incident Syndromes (TRIS) <http://tris.siu.edu/modules/index.php>
Chromosome 18 Registry and Research Society www.chromosome18.org

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

MediGuard medication monitoring made simple www.MediGuard.org
National Institute of Health (NIH) drug, supplement and herbal information
<http://www.nlm.nih.gov/medlineplus/druginformation.html>

Support

Prenatal Partners for Life www.prenatalpartnersforlife.org for those with an adverse prenatal diagnosis wanting to carry to term
Now I Lay Me Down to Sleep 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/
Share Pregnancy & Infant Loss Support Inc www.nationalshare.org/ serves those who had pregnancy loss, stillbirth, or infant loss of life in early months.

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