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

A care book for families, 3rd edition, updated May 2014

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A care book for families

3rd edition, updated May 2014



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

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

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This book is dedicated to

Megan Elizabeth Barnes

7/8/85 - 12/29/04

Trisomy 18

Beloved daughter of Frank and Ann Barnes

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, teacher’s aide

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.

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 e-Book is the 2014 update to the 2008 edition of *Care of the Infant and Child with Trisomy 18 or Trisomy 13*. The 2008 edition can still be purchased at minimal cost, as a printed soft-cover book.

We hope this information 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.

See **other SOFT books** about chromosomes at www.trisomy.org

Trisomy 18 - A Handbook for Families [Stenson et al., 1993]

Trisomy 13 - A Handbook for Families [Stenson et al., 1992]

Now available: an e-Book (Spanish version) of the Trisomy 13 Handbook

Coming soon: an e-Book (Spanish version) of the Trisomy 18 Handbook

Recommended Resource:

Genetic Home Reference www.ghr.nlm.nih.gov/handbook

Video: SOFT Mom & Me Mother's Day Tribute <http://trisomy.org/mothers-day-video-2014/>

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

Kari
9/8/77 - 6/3/88
Trisomy 18



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

Stacy
5/21/81
Trisomy 18





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

See Publications>
Pam's Papers
on the
SOFT website at
www.trisomy.org

Conor
4/2/86 - 4/9/86
Trisomy 18



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

Ecaterina
4/26/2011
Trisomy 13

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 infants and children 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)

Since the mid-nineties the maternal serum triple/quad screening for the **possibility** of birth defects has been a standard of care for the expectant mother. 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 positive result from the screening does not mean that a fetus has a trisomy condition. It does mean there **is an increased risk** that the pregnancy is affected with these problems and further diagnostic testing is warranted.

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. 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.

Recently, four laboratories marketed new **non-invasive prenatal testing (NIPT)** that can detect a trisomy condition, with a single blood draw from the expectant mother as early as the tenth week of pregnancy. While it has not yet replaced other prenatal diagnostic tests, it is likely to eventually become the prenatal diagnostic standard of care. NIPT is discussed in more detail on page 16.

The health care provider will explain how procedures are done; discuss the risk and benefit of the tests, and the possible chance for error. 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. 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, August/September/October, 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. About **5-8% of these infants survive to one year, without extraordinary intervention**, and all children will progress in (some) developmental milestones, although slowly. [Carey, 2010] Recent studies of offering intervention for those with trisomy 18 showed improved survival. [Kosho et al, 2006. [Carey, 2012] And, 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

Medical care of newborns with trisomy 18 or trisomy 13 was traditionally (and often still is) managed as comfort care only, yet there has been a shift in the last decade regarding the medical management of these infants by **some** health care providers to **include consideration of parental decisions** for treatment.

"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. Twenty - one years later he remains a presence in our lives." P.H., Conor, trisomy 18

Rapid diagnosis in newborns with suspected trisomy 18 or trisomy 13 can be helpful in making decisions regarding surgical intervention and care. [Carey, 2010] SOFT families report surgical interventions that

would not have been available twenty years ago, such as cardiac repairs. But finding a surgeon willing to do a cardiac repair was and still is difficult for some parents. Recent concerns about resources versus benefits have contributed to a policy, at more than a few hospitals, of no cardiac repair for these infants.

A number of parents report going before the **Ethics Committee** of a hospital. Ethics committees are usually composed of a bioethicist, physician, and clergy, and also include other pertinent health care professionals. Thus the recommendation for or against a procedure is a responsibility shared by the committee members and the doctor who presented the case. Parents are also a part of this meeting, and 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, May/June/July, 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]

Because of the small numbers who survive past age one year, there are **few doctors who have had experience in long term care** of a child with trisomy 18. [VanDyke, 1990] Carey estimates there are about 200 living persons with trisomy 18 and 13 *older than one year* in the USA. (personal communication, 2013) Most doctors do not expect these infants to survive and this expectation might guide a doctor's approach to care. A doctor who **treats each problem as it occurs, advising interventions** to promote wellness and the child's developmental potential is helping to prepare the infant and parents for the possibility of living.

"A new doctor, watched her rocking, smiling, giggling and responding to attention in a hospital crib. He looked at us and said 'She can't have trisomy 18'." A.B., Megan, trisomy 18

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

Arianna
6/19/06
Trisomy 13



In 2011
Joanna
became
the oldest
living person with
trisomy 18
known to SOFT.
She lived 38 years,
3 months and 20
days.

Joanna
10/14/74 - 2/4/13
Trisomy 18



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 handbooks for families referenced on page eight.

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. A chromosome analysis is performed and cells are arranged in a specific order (karyotype) and counted. This procedure has been in use since the 1960's. Many SOFT families have a photo of their child's karyotype and this diagnostic tool remains useful today for trisomy 18 and 13.

A reliable **non-invasive prenatal test (NIPT)** was marketed in 2012. It is very appealing as optional follow-up invasive testing (ie amniocentesis) is not necessarily needed, and results are obtained earlier than with other tests. NIPT looks at cell-free DNA fragments (cfDNA) from the fetus, circulating in the mother's blood and can detect trisomy 21, 18 or 13 with higher accuracy than the traditional maternal serum screening. (See page 11). Studies find NIPT is effective with a false positive rate (calling the result a trisomy 18 or 13 when it was not) of less than 1%. [Carey, JC Professional Viewpoint: Journal Club, The SOFT Times [Feb/Mar/Apr, 2013] There is a **lack of agreement** within medicine as to whether NIPT is a **screening or diagnostic test** and to

whom it should be offered; all pregnant women or only to women at high risk of fetal aneuploidy (abnormal number of chromosomes). A pretest explanation should be provided to the expectant woman of NIPT benefits and limitations and the fact that **testing for chromosomal abnormality is optional**. If a physician views this test as only a screening and not a diagnosis, invasive testing might be suggested for confirmation of a positive result or a parent may request further testing.

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. 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, as explained above, is needed. Check about insurer coverage of CMA, if considering this test.

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
Cleft lip or palate or both
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)

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 (eye)
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 (hernia of the navel)
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]



Barb Farlow,
one of the
founders of
**International
Trisomy 13/18
Alliance (ITA)**
began in 2012,
is the mother
of Annie.

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



*"I cried the
first time
he 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 SOFT's
**Conference
Assistance
Fund.**

Joey
9/28/77 - 1/7/84
Trisomy 13

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.

Commonly documented activities reported for both disorders in this study 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]

The reported developmental milestones for both disorders were tabulated by Baty et al. (See Developmental Achievements in Trisomy 18 and Trisomy 13, Table 4). In addition, within SOFT newsletters, parents of a few 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 was observed at the 2013 SOFT conference.

In discussion at the end of her paper Baty wrote that 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] In reference to this study and to individual case reports, Carey notes that 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 et al. reported developmental ages in the older children with trisomy 18 averaging at 6 to 7 months, and in those with trisomy 13 averaging at 13 months. [Carey, 2010]

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. The results are summarized in Table 3. Pamela and her husband, Michael, had a son, Conor (4/2/86 - 4/9/86), who was born with trisomy 18.

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, with the age range of 25 months - 17 years, and noted there was 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]

Healey notes that development continues across the lifespan of each syndrome with some loss of skills, particularly motor, with age related or illness-related constraints. Those with progressive illnesses eventually may have more difficulty with skills, and hospitalization and/or surgery might result in setbacks with long time periods needed to regain skills, if regained. 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.

“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

Physical therapy, occupational therapy, speech therapy and vision therapy are available to those who meet criteria for early intervention programs or special education at public school. For those with trisomy 13 who might have both vision and hearing impairment, a more comprehensive program might be needed. Some parents take their child for extra therapy in a clinic setting which usually requires a referral by the child’s doctor. Parents should check with the clinic billing office to determine how these services will be reimbursed.

The *Trisomy 18 Handbook for Families* and the *Trisomy 13 Handbook for Families* mentioned in the Introduction provide helpful information about early intervention and public school education. Parents can contact their local department of education to obtain details about registration and location of the school their child will attend. Special education may not be available in every school setting, but public law mandates the

availability within a district, and bus service will be provided for the child. Parents can make arrangements to visit the classroom and meet the teacher(s), aide(s) and whoever else may interact with their child. Most parents have mixed feelings about sending their child to school.

“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

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**. The parents of children in these programs are often supportive of one another and are information resources. 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, parents report that their 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.



“Donnie” is
the oldest male
living with
trisomy 18, in the
SOFT membership.

Donald
9/10/92
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		

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.



Erin was the oldest person known to SOFT with full trisomy 13. She lived 35 years and one month.

Erin
2/22/78 - 3/22/13
Trisomy 13



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

“Dr. Carey told us her heart repair was done at the youngest age, known to him, for an infant with trisomy18.”
L.S., 2014

Lyndsay
7/20/00
Trisomy 18

Common Illnesses and Health Problems

Infants and children with trisomy 18 or trisomy 13 can become sick with the common illnesses that all children have. *Acute* illness, such as pneumonia, usually has a rapid onset and short duration but frequently occurs in many infants and children with trisomy 18 or trisomy 13. *Chronic* illness, such as congenital heart disease or seizures, is ongoing and is common in both disorders. Table 5 indicates the incidence of illnesses and common health problems in those with trisomy 18 or trisomy 13.

Table 5

Prevalence of Common Illnesses and Health Problems in Trisomy 18 or Trisomy 13

Frequent Incidence

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

Less Frequent Incidence

Cataracts in children with trisomy 13
Glaucoma in children with trisomy 13
Wilms Tumor (kidney cancer) in children with trisomy 18
Other anomalies

Low Incidence

Hepatoblastoma (liver tumor) in children with trisomy 18

[source: SOFT Registry, the SOFT touch, The SOFT times, 1988-2014]



Terre Krotzer,
creator and host of
**Trisomy Talk
Webinars**,
is the mother of
“**Krissy**”.

Kristina
3/25/00
Trisomy 18



Zion is the
inspiration
for the SOFT
Parade of Stars.

Zion
5/2/01 - 2/16/10
Trisomy 13

Central Nervous System Problems and Abnormal Muscle Tone

Abnormalities of the brain and dysfunction of the central nervous system contribute to health problems common for those with trisomy 18 or 13.

Central Apnea

It is suggested that central apnea, either alone or in combination with other health issues, where breathing stops temporarily 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] Dr Carey has seen caffeine prescribed a few times for these 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) Only about 50% of those born with trisomy 18 or trisomy 13 survive longer than a week. Some parents report apnea as the cause of death of their infant on the SOFT membership forms. Another type of apnea, called obstructive apnea (see page 70), is due to a blockage of the airway and is discussed within the chapter on Mortality.

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] 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, the doctor can refer the child to a pediatric neurologist. Testing is done by an **electroencephalogram (EEG)** that measures electrical activity in the brain by using sensors attached to the scalp and might be done in an overnight sleep study. There are many 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. See Photophobia on page 51 for information about **Photosensitive Epilepsy** by Dr. Steven D. Cantrell.

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 refers to 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. Steve Cantrell, 2011)

If appropriate, the 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**, often called shin splints), and eye patches to strengthen eye muscles as prescribed by specialists for their child. Physical therapy, occupational therapy, speech therapy, music therapy, and vision therapy are reported by parents as beneficial for their child

*“Patrick
taught us
so much
about love
in so little
time.”*

V.S., 1986

Patrick
2/6/87 – 4/18/87
Trisomy 18



*“Morghan
started using a
walker at about
age seven years
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 ½ pounds.

Morghan
9/14/96
Trisomy 18





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

See Clefts, page 34

Sofie Marie
1/24/14
Trisomy 13



Ryan
is the
inspiration
for **SOFT's**
annual
**Balloon
Celebration**

Ryan
10/4/85 - 6/15/86
Trisomy 18

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** (see page 39) is discussed later in this chapter under **Gastroesophageal (GE) reflux**.

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. These 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 might be helpful and/or referral to a 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) placed. See page 36. 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).

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 reported success

with bottle feeding, and more than a few reported success with breast feeding.

"She had not yet been diagnosed, so no one advised against breast feeding. It took about two weeks of trying before she finally learned to latch on and suck, and she was breast fed for two years. Her weight increased by ten pounds that first year when totally breast fed. But the second year she only gained 2 pounds as we began spoon feeding her jarred baby foods and likely breast fed less." A.B., Megan, trisomy 18

In the SOFT newsletters, a few parents reported using a pacifier in their infant's mouth for oral stimulation whenever they gavage fed their baby by a tube inserted through the nose or mouth, down the throat, through the esophagus and into the stomach. Eventually their infant learned to take a bottle.

"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." [adapted from The SOFT Times, February/March, 2001]

Clefts

Oral clefts are present in about 10% of infants with trisomy 18 and 60% - 80% of those with trisomy 13. [Jones, 2013] Cleft lip may prevent an infant from getting the mouth closure around a nipple that is needed for effective sucking. Cleft palate may 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 at 1-800-435-8316. Parents should check with their insurer about coverage as these expensive nipples are needed for a medical reason.

The SOFT Surgery Registry shows more cleft lip and/or palate repairs were done for those with trisomy 13 compared to repairs for those with trisomy 18. Repairs can involve more than one surgery. If the child has

a cardiac defect, the plastic surgeon must consult with the child's cardiologist and risks versus benefits of surgery should be discussed with the parents. Other specialists, such as an ear, nose and throat (ENT) surgeon or dental 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, February/March/April, 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, February/March/April, 2002]*

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 41) causes discomfort and is a common problem in trisomy 18 or trisomy 13. Gastroesophageal (GE) reflux (see page 38) 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 before two 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.

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 an infant or 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 is often problematic. Let the doctor know if this is happening. There are a variety of medical tapes.

Survey questions for parents, asking how their child is (or was) fed are on the annual SOFT membership form. Responses show that some are fed through a tube and are also fed by mouth. Some are totally tube fed and a few are fed only by mouth. Parents also report that a few children learned 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

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 putting 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 for sleep, elevating the crib mattress on one end by 30 degrees. 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 for consideration, if medication fails.

The SOFT Surgery Registry of all diagnoses shows that approximately 1/3 of those with a G-tube also had a fundoplication. These surgeries were not always done at the same time. 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** (if available) or the new **pediatric palliative care**, for those with life limiting conditions. Participation in these programs is **optional**. See page 85. Problems noted in this chapter may not be apparent in the newborn but with time might need evaluation and treatment by a healthcare provider.

"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]



Leilani
5/3/2000
Trisomy 18



ThereseAnn,
one of the
administrators
of the SOFT
Facebook page,
is the mother
of Natalia.

Natalia
8/25/00
Trisomy 13

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 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 issues 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 may 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 usually 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 within 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, 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, August/September/October, 2003]

"She did not indicate a need to go but responded to praise and routine times for potty use with 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 Surgery Registry which is found on the SOFT website at www.trisomy.org. For example: **bowel obstruction repair or resection, and bowl malrotation repair** show a combined total of 14 children with trisomy 18, and 32 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 seven children with trisomy 18 and one with trisomy 13.

Enemas

Be aware that use of enemas to facilitate bowel action can lead to problems. Enemas can deplete a baby of electrolytes and alter body fluids in children. (Dr Jay Mamel, Professor Gastroenterology, USF, personal communication, 2000) **Enemas should only be used with caution and under the advice of a doctor.**

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. [DDHealthInfo.org] **When soft stool is achieved it is important to continue daily maintenance, as prescribed, to prevent reoccurrence of constipation.**



Nicholas
4/6/95 - 11/4/2013
Trisomy 13



Samuel
11/28/1998
Trisomy 13
Mosaicism

Routine Medical Care, Growth and Other Themes in Care

Before newborns are discharged to home from the hospital an appointment is made for follow-up with a pediatrician or other health care provider. Or if available, discharge to pediatric palliative care or hospice is an option for infants with a life limiting diagnosis. It is a part of this doctor's job to prepare parents for the probable loss of their baby with trisomy 18 or trisomy 13 but it is also important that the doctor prepares parents for the possibility of being caretakers of a child with disabilities. It is appropriate for parents to ask the doctor to help them find current information about the health and care issues of the older infant and child. Contact with a support group such as SOFT is beneficial. In recent years, social media has greatly enabled contact between parents. See www.trisomy.org for facebook links.

"The most frustrating part for us has been the inability to ascertain exactly what to expect in the future." J.K., Hayley, trisomy 18

"We soon realized Andrew was going to live life his way, no book or expert could tell him what to do. ...Andrew taught me more about trisomy 13 than any book could." [Andrew's pediatrician, The SOFT Times, 2002]

This chapter discusses routine medical care and some potential health problems that might occur for those with trisomy 18 or trisomy 13. 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 referred to by health care providers when they are asked to complete health forms. Entrance for any child to early intervention or public school usually requires a physical, vision and hearing exam, and documentation of up to date immunizations. Medical care for those with trisomy 18 or trisomy 13 is often complex as multiple health issues are common.

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 of those with trisomy 18 and trisomy 13, 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 60-62. The charts are also provided as removable pages in the front pocket of the 2008 printed book. 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 also available for printing, on the SOFT website. See Resources at www.trisomy.org.**

There is generally 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.

"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. He has put on 5 pounds since Christmas." [The SOFT Times, May/June/July, 2004]

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

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

"I never in my life thought I would have to say Stacy needed to be on a diet." [The SOFT Times, February/March/April, 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 first 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. "An 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

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 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**. Cantrell noted that some SOFT parents have indicated their child's vision test went no further than the disabilities and side effects rather than maximizing abilities. 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.

Ocular (eye) Problems

Eye malformations commonly occur in trisomy 13 and have a low occurrence in trisomy 18. Cantrell has provided information about selected ocular problems common to trisomy 13 and as seen in some children with trisomy 18.

Cataracts

Cataracts are cloudy, rather than clear, crystalline lenses of the eye and may occur in all children born with trisomy. However, the incidence of cataracts is much higher with trisomy 13. Infants with trisomy 13 are more likely to have cataracts at birth. The most frequently reported eye repair for children with trisomy 13 is cataract removal. Those with trisomy 18 are less likely to be born with cataracts but develop them with age. This could be 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, corn 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. The SOFT Surgery Registry lists 15 children with trisomy 13 and two with trisomy 18 had cataract surgery. Prior to surgery, consultation with a cardiologist about risk versus benefit of surgery should be done for those with congenital heart defects. An alternative non-surgical eye drop to reverse cataracts is currently in clinical trials and has shown promise. As with any therapy, carefully consider the risk versus the benefit. (personal communication, Dr Steven Cantrell, 2007)

Glaucoma

Glaucoma is an eye condition in which fluid builds up inside the eye and increases pressure on the optic nerve. Some infants with trisomy 13 will develop congenital or early onset **glaucoma** resulting in increased intraocular tension. This could be a reason for unexplained irritability. [Carey, 2005] Treatment with topical drops to maintain optimal pressure is a common treatment for anyone with glaucoma and has been reported by SOFT parents. The SOFT Surgery Registry shows children with trisomy 13 had glaucoma surgery. New approaches to surgery for glaucoma are recently in the news but not all patients are candidates.

Strabismus

Strabismus (crossed eyes) is a common disorder that can be seen in any child. As noted in the chapter on Central Nervous System Problems, strabismus is a common problem in those who have abnormal muscle tone. Strabismus surgery is reported in the SOFT Surgery Registry as the most frequent eye surgery for children with trisomy 18 while only one child with trisomy 13 and one with mosaic trisomy 13 had strabismus repair. “An eye examination for glasses is suggested prior to surgery. Uncorrected high prescriptions may also trigger crossed eyes.” (personal communication, Dr Steven Cantrell) Strabismus surgery is done for a medical reason, to align both eyes for normal vision, but also has positive cosmetic results. The eye surgeon must consult with the child’s cardiologist and the benefit versus risk should be discussed with the parents prior to any surgery for those who have cardiac anomalies. Some parents choose to not do surgery.

Photophobia

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 recommends a dark polarized gray sunglass for those without seizures. Children experiencing seizures along with light sensitivity may be subject to **Photosensitive Epilepsy** caused by flickering light. **Glasses tinted with #2 or #3 Colbalt Blue tint** have been shown to **reduce or eliminate this trigger** and should be worn full time. The antioxidants lutein and zeaxanthin, found in green leafy and yellow vegetables or supplements, listed under Cataracts, are also helpful in reducing light sensitivity.

Eye Irritation

Often children with both conditions **do not fully close** their eyelids when sleeping and the exposed surface of the eye becomes dry and irritated. Some parents report use of an ophthalmic ointment to protect the surface of the eye during sleep and sometimes artificial tear drops during the day as prescribed by their child's doctor. One parent reported that gold weights were implanted in her child's eyelids. The weights close her eyes by gravity when she sleeps and are working well. Cantrell notes that **long eyelashes** are common to children with trisomy and along with **incomplete lid closure** may cause a red eye condition (inflammation) called **blepharitis**. The eye doctor may prescribe an antibiotic/steroid drop or ointment to treat the blepharitis and help prevent red eyes.

In table 6 (pages 51-53) Cantrell lists eye conditions that might be seen in those with trisomy 18, trisomy 13, and related disorders but notes **these problems could also occur in any child.**

Table 6

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 artificial tears 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 sunglasses, hats, tinted auto windows and extra Lutein through diet or supplement.

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 2011]

Hearing

Most states in the U.S. have a requirement for mandatory screening of hearing in a newborn. Infants who do not pass screening will 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 might show different results. One parent was happy to report a follow-up behavioral response hearing test showed improved auditory response. Moderate to severe hearing loss has been reported in some older children in trisomy 18 and trisomy 13.

"...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, May/June/July, 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]*

A few SOFT Facebook moms report their child uses Soft Bands (Baha and Pronto Soft) worn like a head band containing hearing devices. Links to these products can be found under Resources on the SOFT website.

Table 7

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

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

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

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 are exempted for religious reasons or adverse reaction to a prior vaccine or need a delay due to illness or other concerns. 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 or trisomy 13, compared to chromosomally normal children, although the numbers studied are small [Baty et al., 1994]. Two decades later, the majority of parents reported on the SOFT membership form that their child received the standard immunizations and a yearly flu vaccine. They also reported any reactions. Reactions considered typical for any child occurred in 17 of 181 with trisomy 18 **but illness also occurred in a few** of the 17. In those with trisomy 13 only 1 of 110 had a reaction (non-specified). A few parents chose to delay or decline vaccines. SOFT parent reports about immunizations and reactions concur with the findings of Baty et al.

RSV

Respiratory syncytial virus (RSV) is a serious lung infection for premature infants and infants with chronic lung disease. For older children and adults it usually results in a cold or upper respiratory illness but one 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*, November/December/January, 2004-2005] Dr. Scott Showalter, pediatrician, attends the annual SOFT conferences and is available to care for 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 18 and 19.) If appropriate, the child's doctor can make a referral to a **pediatric orthopedic** doctor for evaluation of anomalies that might need casting or surgical intervention. X-rays allow for diagnosis of degree of involvement and help in the decision as to what will be in the best interest of the child. Some parents choose to not intervene, others defer interventions until their child is older and some choose correction. If a child has a cardiac defect, surgical decisions require consultation from the child's cardiologist about benefits versus risks of surgery.

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 with visual inspection 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 beyond 20 degrees is usually treated with a custom fit orthopedic brace called a body jacket, used to delay or arrest progression. Several parents report their child wears or wore a body jacket for many years. When the curvature progresses beyond 40 degrees, surgery is usually discussed. Further progression to more severe curvature gradually diminishes lung capacity leading to long term heart and lung complications. One child in the SOFT Surgery Registry had a spinal fusion done at seven years but most orthopedic surgeons prefer to wait until the child is ten years old, because continued growth is an issue that might result in a need for more surgery. Some parents choose to delay surgery. When a child has a congenital heart defect the orthopedic surgeon should consult with the pediatric cardiologist and risks versus benefits of surgery should be discussed with the parents.

There are several types of rods used for spinal fusion which the surgeon can differentiate for the parents. A few parents reported their child had a successful repair done with a **Luque™** rod which requires a longer time under anesthesia for placement than some other rods do.

“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).

Risk of Fracture

Several older children with trisomy 18 and trisomy 13 were 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. **Ask how medications should be given and about possible side effects.** See Resources page 97.

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]

“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 doctor or dentist can prescribe a prophylactic antibiotic to use prior to dental 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 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

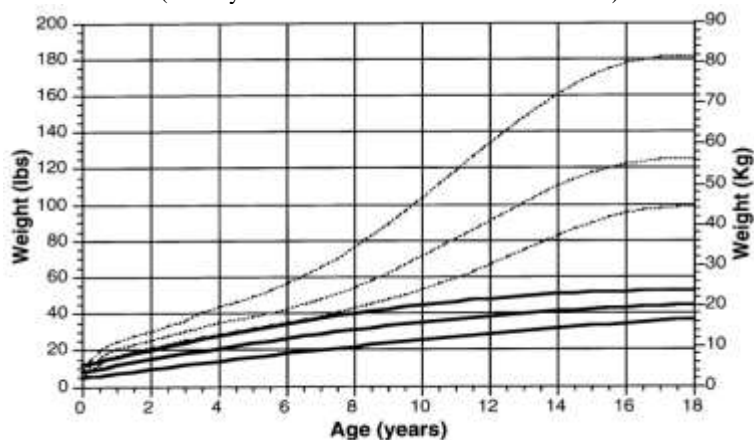
Over time a child with trisomy 18 or trisomy 13 will see many specialists. These children may need tube feedings, oxygen, medications, wheel chair transportation and more during a trip to the doctor. The caretaker has many details to remember and must also inform the doctor about her child's history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

It is necessary for each specialist to 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. Parents who have recorded and kept this information up to date with changes should take the recorded information to the clinic or hospital visits to assure that all providers are given the same information. A log book format could be used. Dr Carey suggests asking each professional who treats your child to briefly summarize their findings and recommendations in the log book.

Table 8a Growth Curves for Trisomy 18 and 13

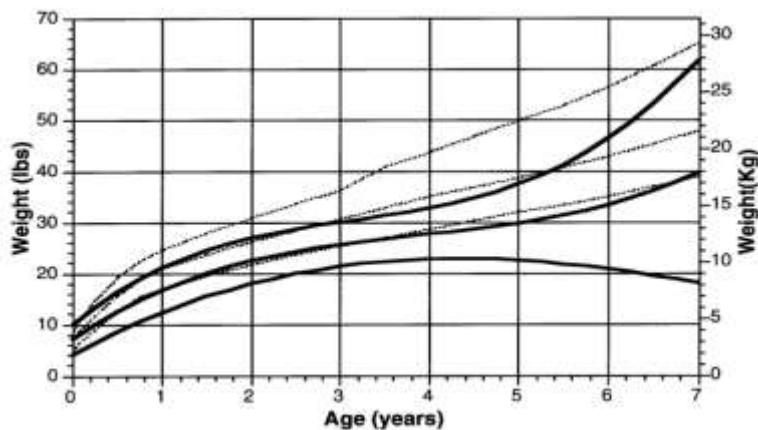
Trisomy 18 Weight Curve

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



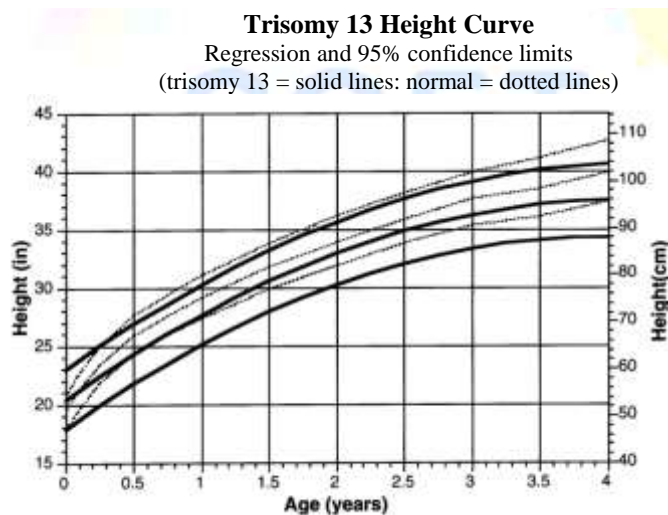
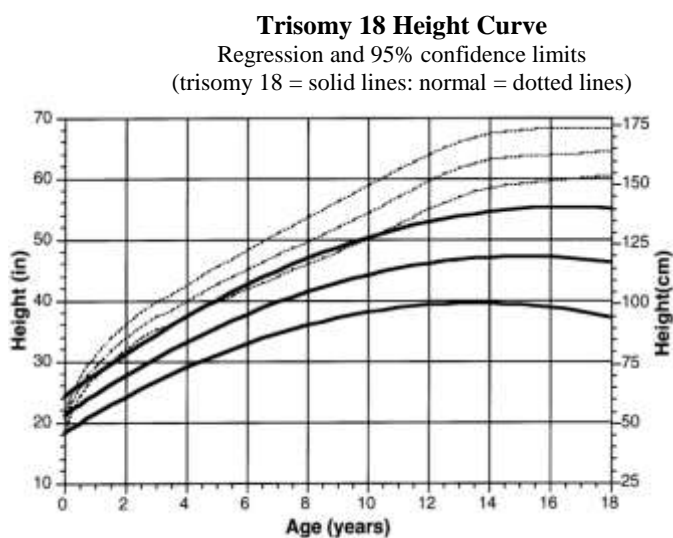
Trisomy 13 Weight Curve

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

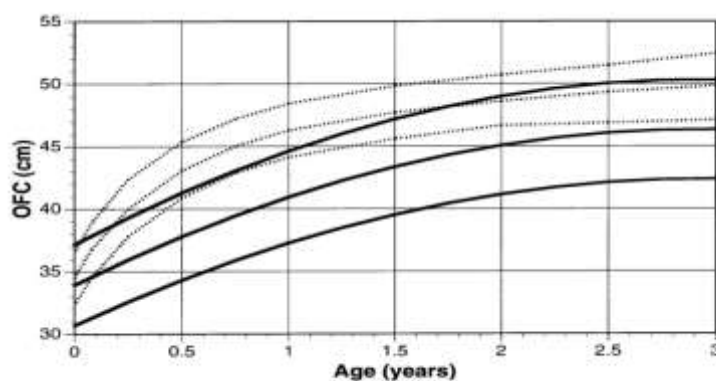


Source for height curves: Baty BJ, Blackburn BL, Carey JC (1994) Natural history of trisomy 18 and trisomy 13: I. Growth, physical assessment, medical histories, survival and recurrence risk. Am J Med Genet 49: 175-188, © 1994 Wiley-Liss, Inc. reprinted by permission of John Wiley & Sons, Inc.

Table 8c Growth Curves for Trisomy 18 and 13

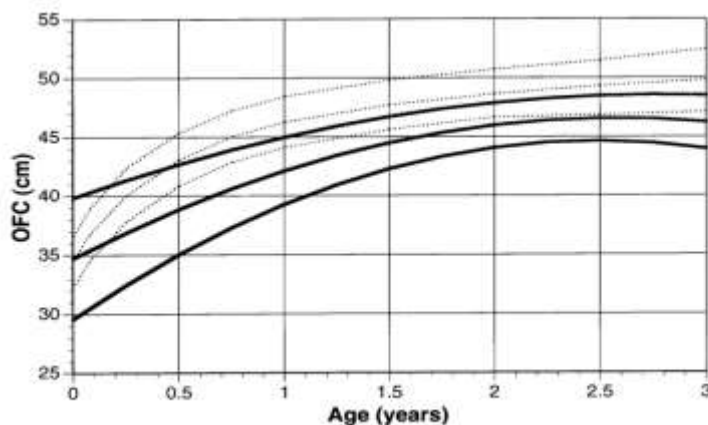
Trisomy 18 Head Circumference Curve

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.

Abir Schmidt,
a member of
the 2014
SOFT Conference
planning committee,
is the mother of
Summer.

Summer
10/21/11
Trisomy 13



Terra Garst
welcomes
expectant
families to SOFT
and is the mother
of Emerson.

Emerson
6/23/11
partial
Trisomy 18q and
partial
Monosomy 13p





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

Nora
2/9/13
Trisomy 13



*"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

Morganne
10/9/98 - 5/13/99
Trisomy 18

The SOFT Surgery Registry

Other surgeries/procedures, besides cardiac, are in the SOFT Surgery Registry. Go to www.trisomy.org > Resources > SOFT's Surgery Registry to see all surgeries and who to contact with a surgery inquiry.

Table 9

The SOFT Surgery Registry					
Cardiac surgeries & procedures (t18,t13 & mosaic)		Number of surgeries, by diagnosis			
Outcomes Vary	T13	T13m	T18	T18m	RD
Aorta angioplasty/widened /reconstruction	0	0	1	0	1
Aortic Coarctation repair	3	1	5	0	1
Aortic Valve surgery	0	0	1	0	1
ASD repair (Atrial Septal Defect)	4	2	25	5	11
AVSD repair (Atrioventricular Septal Defect)	0	0	1	0	0
Balloon Dilatation	0	0	2	0	0
Blaylock -Taussig	1	0	3	0	2
Cardiac Catheterization	1	1	10	1	1
Double Outlet Right Ventricle	0	1	2	0	1
Fontana procedure	0	0	1	0	0
Mitral Valve repair/leak correction	1	0	0	0	1
Norwood procedure	0	1	0	0	0
Pacemaker	0	0	5	1	1
PDA* Cath w/coil/occluder repair	1	0	2	0	1
PDA* repair/ligation	4	3	34	3	15
Pericardiectomy	0	0	0	0	1
Pulmonary Artery Band removal/loosening	1	0	2	0	0
Pulmonary Artery banding	1	0	12	1	2
Pulmonary Artery- patch/arterioplasty/reconstruction	1	0	2	0	0
Pulmonary Stenosis repair	0	1	2	1	1
Pulmonary Valve Atresia Valvuloplasty	0	0	3	0	1
Pulmonary Vein repair	0	0	0	0	1
Shunt placement	1	0	2	0	0
Surgery, unspecified cardiac	0	2	4	2	3
TAPVR**	1	0	0	0	1
Tetralogy of Fallot repair	4	0	6	2	4
Truncus Arteriosus repair	0	0	0	0	1
VSD repair (Ventricular Septal Defect)	4	2	40	9	14

T = trisomy, m = mosaic, RD = related disorders (SOFT Surgery Registry 2014)

* Patent Ductus Arteriosus,

**Total Anomalous Pulmonary Venus Return

Table 10

The SOFT Cardiac Surgery Registry of Hospitals

Hosp list April 2014- page 1 of 2

Hospitals where cardiac surgery occurred for full T18, T13
(names reported by SOFT parents 1990 - May 2014)
Some listed might no longer provide cardiac surgery for T18 or 13
<p>Arkansas Children's Hospital, Little Rock, AR</p> <p>BC Children's Hospital, British Columbia, Canada</p> <p>Boston Children's Hospital, Boston, MA</p> <p>Brackenridge Children's Hospital, Austin, TX</p> <p>Cardinal Glennon Children's Hospital, St Louis, MO</p> <p>Carolinas Med Center-Levine Children's Hospital, Charlotte, NC</p> <p>Children's Hospital Colorado, Aurora, CO</p> <p>Children's Hospital of Philadelphia, Philadelphia, PA</p> <p>Children's Hospital of Pittsburgh, Pittsburgh, PA</p> <p>Children's Hospital of the King's Daughters, Norfolk, VA</p> <p>Children's Hospital of WI, Milwaukee, WI</p> <p>Children's Hospital, Los Angeles, CA</p> <p>Children's Hospital, Orange County, CA</p> <p>Children's Hospitals Minnesota, Minneapolis-St Paul, MN</p> <p>Children's Medical Center of Dallas, TX</p> <p>Children's Memorial (Lurie Children's), Chicago, IL</p> <p>Children's Mercy Hospital, Kansas City, MO</p> <p>Children's National Medical Center, Washington, DC</p> <p>Cook's Children's Medical Center, Fort Worth, TX</p> <p>CS Mott Children's Hospital, Ann Arbor, MI</p> <p>Dayton Children's Hospital, Dayton, OH</p> <p>Edmonton Stollery Children's Hospital, Canada</p> <p>Egleston Children's Hospital, Atlanta, GA</p> <p>Emanuel Hospital, Portland, OR</p> <p>Johns Hopkins Baltimore, MD</p> <p>Kapiolani Medical Center for Women & Children, Hawaii</p> <p>Le Bonheur Children's Hospital, Memphis, TN</p> <p>Legacy Health System, Portland, OR</p> <p>Loma Linda Children's Hospital, Loma Linda, CA</p> <p>Lucile Packard Children's Hosp.-Stanford, Palo Alto CA</p> <p>Medical University of South Carolina, Charleston, SC</p> <p>Mercy Hospital, Des Moines, IA</p> <p>Methodist Children's, San Antonio, TX</p> <p>N.C. Baptist Hospital, Winston-Salem, N.C.</p>



Nationwide Children's Hospital, Columbus, OH
 North Memorial, Robbins Clark, MN
 OSF St. Francis Medical Center, Peoria, IL
 Payton Manning Children's-St Vincent's, Indianapolis, IN
 Penn State Hershey Children's Hospital, Hershey, PA
 Presbyterian Hospital, Albuquerque, NM
 Primary Children's Medical Center, SLC, UT
 Rady Children's Hospital, San Diego, CA
 Riley Children's Hospital, Indianapolis, IN
 Royal University, Saskatoon, Canada
 Ruby Memorial Hospital Morgantown, WV
 San Diego Children's Hospital, San Diego, CA
 Seattle Children's Hospital, Seattle, WA
 Shands Teaching Hospital, UF, Gainesville, FL
 Sick Children's Hospital, Toronto, Canada
 St Francis Hospital, Tulsa, OK
 St Francis Medical Center, Peoria, IL
 St Joseph Children's Hospital, Tampa, FL
 St Luke's Hospital, Boise, ID
 St Mary's Hospital, Rochester, MN
 Tampa Children's Hospital, Tampa, FL
 Texas Children's Hospital, Houston, TX
 Tulane Medical Center, New Orleans, LA
 UC Davis, Sacramento, CA
 University of Alabama Hospital, Birmingham, AL
 University of Chicago Hospital, Chicago, IL
 University of Miami, Jackson Memorial, Miami, FL
 University of Mississippi Medical Center, Jackson, MS
 University of VA Medical Hospital, Charlottesville, VA
 Wolfson's Children's Hospital, Jacksonville, FL
 Yale Children's Hospital, New Haven, CT

"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]

"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

"An Ethics doctor said the attitude is changing for the positive toward trisomy 18... they want to be on the front of the curve." A.S., Janessa, trisomy 18, 2014



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

Megan
5/15/80
Trisomy 18



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

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

Greta
7/15/80
Smith Lemli/Opitz
Syndrome (SLO)

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. Survival studies indicate similar outcomes 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] 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.

“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

“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, February/March/April, 2004]

Parents of those babies who live beyond early 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”.

“Decisions could not be made on the specifics of his disorder, because, quite frankly, Andrew was writing his own book, rather than following one.” [Andrew’s pediatrician, The SOFT Times, Feb/Mar/Apr, 2002]

Apnea

Approximately half of those born with trisomy 18 or trisomy 13 do not survive the first week of life and often the immediate cause of death is not known. Some parents report apnea in which breathing stops temporarily, as the cause of death. **Central apnea** (page 29), where there is a reduction in stimulus to or impulses from the respiratory center of the brain, is discussed under Central Nervous System. Recent investigations suggest its presence combined with other health problems as a primary reason for infant death. [Root and Carey, 1994; Embleton et al., 1996; Wyllie et al., 1994; Carey, 2010] Another type of apnea is **obstructive apnea** where the airway becomes temporarily blocked. Gagging, vomiting or reflux might result in obstructive apnea episodes. Seizures sometimes affect respirations or contribute to reflux/vomiting. Enlarged tonsils or adenoids, and malformations such as tracheomalacia (softening of the cartilage of the trachea) can narrow or temporarily block the airway, especially during sleep, as muscle tone decreases during sleep. Parents should inform the doctor if their infant or child is experiencing 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 an 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. It is a **non-invasive breathing aid** 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.

Pneumonia

Pneumonia is often reported as the 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 CoughAssist 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 for those with trisomy 18 and children 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 that can be life threatening. Scoliosis is also discussed on page 56.

Cardiac

On the SOFT registration form one of the most frequent responses from parents about the cause of death for their child with trisomy 18 or trisomy 13 is simply written as “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]

All infants with these disorders should be evaluated by a pediatric cardiologist as it is important for health care decisions. 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.

Since 1990 discussion of cardiac surgery as an option has been more common yet such surgery is still denied in many hospitals for infants with trisomy 18 or 13. 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 or to go home to wait for repair.

Smith's Recognizable Patterns of Human Malformation has guidelines significant to neonatal, pediatric and genetics care. Earlier editions about trisomy 18 or trisomy 13 recommended withdrawal of all treatment “to prolong life” once diagnosis has been made. Recommendations about newborns with trisomy 18 or trisomy 13 in recent editions have changed as follows. “Once the diagnosis has been established, limitations of

*extraordinary medical means for prolongation of life should be seriously considered. **Parents and individual circumstances of each infant must be taken into consideration***". [Jones, 2013; Carey, 2012]

The SOFT Surgery Registry maintains a list of parent reported cardiac repairs and hospitals (see pages 66- 67) where these repairs were done. However, **policies can change** and 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 the feeding, respiratory and 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, May/June/July, 2002]

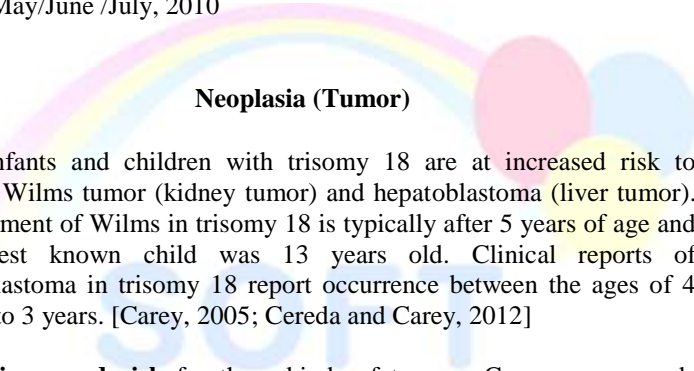
"Her cardiologist was surprised because her large VSD had closed on its own." [The SOFT Times, May/June/July, 2002]

*"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, May/June/July, 2002]

Using the SOFT Surgery Registry, Hansen et al., (2000) performed a study of cardiac repair in those with trisomy 18 or trisomy 13 and reported 25 of 29 children (86.2%) who had a cardiac surgery were discharged to home. There were 4 surgical mortalities (13.8%). Graham et al summarized a 91% discharge to home in their 2004 published study.

It is evident that these children can survive anesthesia, and survive to go home, making surgery an appropriate option in certain cases. [Carey, 2010; Cereda and Carey, 2012] The three cardiac surgery studies done to date for trisomy 18 and 13, from Japan, Canada and the USA show an average success rate of 90% surviving the surgery and discharged to home.

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 www.trisomy.org 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, May/June /July, 2010



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]

Due to **increased risk** for these kinds of tumors, Carey recommends screening 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] The SOFT Surgery Registry shows 5 children with trisomy18 had Wilms tumor surgery; 3 with trisomy18 and 1 with mosaic trisomy18 had liver tumor removal.

“Fortunately we caught it in the early stage. One month ago his kidney was removed and he is currently undergoing mild chemotherapy and so far tolerating it well.” [the SOFT touch, 1993]

“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]

Issues in Care

A health care provider’s **view** of trisomy 18 or trisomy 13 can influence the management and care these children receive. When a child needs intervention in a critical health situation, and it is withheld by a care provider without consultation with the parents, then the **care provider** has “**allowed the child to die**”. Some parents will never know that this has happened while others suspect it might have happened. Obtaining the child’s medical record and going through it with a trusted doctor can answer questions about what occurred. Parents have a right to request their 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.

When a child is hospitalized it is important for parents to ask questions about what is happening and why. Understandable communications from the physician to the parent about the child’s medical condition, the options for treatment and care, and the risks and benefits of intervention are vital to a parent making informed decisions. **If parents feel confused or uncomfortable with the answers, it’s critical to ask the doctor the question again or request a second opinion.**

“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 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

“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

“We were informed of her condition and options for care. Clearly, our choice was that Kari should receive every opportunity to get well. Our question to her doctor was direct. ‘Can you support this?’ His response was ‘we can support this decision’.” [The SOFT Times, February/March/April, 2004]



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

Milan
11/2/2010
Trisomy 18



Mary
3/27/96
Trisomy 18

Life Support Directives

Decisions regarding life support in the care of infants and children with trisomy 18 and trisomy 13 are complex. In this section we begin with a general review of documents used in end of life directives and discuss the requirements for a DNR order to be valid. **In the next chapter life support order decisions and parental rights are discussed.**

The existence of three advance directive documents plus the in-hospital life support order form create confusion about life support directives for the public. The fact that forms and form titles can vary from place to place adds to the confusion. A general explanation of Life Support Directives follows.

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/directives when a life support order needs to be written. Advance directive documents described in number one and two below are **not** considered legal medical orders and will **not** be followed by medical personnel without a concurring, signed physician order. Advance Directives consist of the following.

1. Living Will

This legal 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 legal document is usually completed along with a living will and it 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 of this document often differs from state to state, such as Uniform Do Not Resuscitate (DNR), or Portable DNR and more. Such titles refer to 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 to sign this directive as well as the physician to show proof of consent. Instead of an advance directive, a number of states have two separate forms; one is only a DNR, the other form outlines a plan of care including when and how to attempt resuscitation and the desired medical interventions.

One of these portable forms signed by a physician is 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

When there is no DNR order signed by a doctor, it is mandatory for medical personnel to do CPR for respiratory or cardiac arrest.

The title on the form used to write the **in-hospital Life Support order** for a patient may vary from hospital to hospital. This order is signed by the hospitalized patient's attending physician and directs the amount of cardiopulmonary resuscitation (CPR) to be done or not done in the event of cardiopulmonary arrest. If the patient is unable to speak for herself and does not have an advance directive, concurrence with this order must be obtained from the patient's representative or a relative, if available. If no one is available, the doctor can make the decision. This document is also often referred to as the DNR form.

Unilateral DNR

In the absence of a directive and non-availability of a patient representative and when the patient is unable to speak for herself in a "hopeless" terminal situation, there is a provision in some state statutes for a doctor to make a unilateral decision to code a patient as a DNR. A second written opinion from another doctor unconnected to the case is usually required.

It would be prudent for parents to check their state law statutes about life support directives and the DNR order. This can be done on-line.

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 sometimes in the midst of many other admission questions, might be only briefly addressed as "Do you want her on a ventilator?" If Life Support Order decisions for cardiopulmonary resuscitation (CPR) are not clearly discussed, the parent or guardian needs to raise this topic with the physician.

A discussion about Life Support should provide the parent an explanation about procedures used during 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.

It is very important that parents know what is written on the Life Support Order form but sometimes the question about life support is poorly asked by the health care provider, without an explanation about procedures or without showing the form to the parents. Under these circumstances **parents might not 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 if the child is coded as a **DNR (Do-Not-Resuscitate)** in the case of the child's heart or respirations stopping.

A DNR is a Life Support order to withhold CPR. 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 both a physician *and* parent or guardian signature for a Life Support order to be valid. In these circumstances a parent or guardian will see the order when signing consent on the in-hospital order form. In hospitals that do not require parent or guardian signature, it is unlikely the DNR order will be seen by a parent or guardian unless they ask to see it.

Changing Life Support Orders

After a discussion about life support for the child between the parent or guardian and doctor, Life Support orders are written by the physician and **should** reflect parental or guardian directives. The physician's signature

makes it a valid order. Thus, the Life Support order should be both a physician's order and a patient or representative's directive. Parents or guardians can change their mind at any time about what is to be done and can ask for a new order to be written. A new Life Support Order signed by a physician 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, make 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:

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

Always check the status of your child's the Life Support Order, if any of these changes happen. **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

Some parents within SOFT report being advised and **many felt pressured**, by hospital staff to code their child born with trisomy 18 or trisomy 13 as a DNR. If there is not a DNR order, medical personnel are required to administer CPR if your child's heart stops or breathing ceases. The concern expressed by many in the medical community is that those with chronic heart disease and or lung conditions or other serious health problems might be unable to be weaned off a ventilator used for resuscitation. The worry for parents is that the conventional view, of limiting extraordinary treatment for these children, may result in a withholding of interventions that could give the child a chance to survive.

In a medical crisis parents struggling with hope and fear come to a decision based on the doctor's explanation about their child's illness and options, their values and beliefs, and what they understand to be in their child's best interest. The guidance of a caring professional can help a parent understand what is happening to their child and what, if anything can be done towards recovery or for comfort care. Parents have a right to ask that full care and intervention be done for their child. Parents also have a right to ask for no intervention but comfort care. In states or hospitals where it is an option, parents may ask that only specific CPR interventions be done or not done. Parents expect their decisions will be respected and appreciate the supportive physician. Parents can ask for a copy of their child's Life Support order to be certain their wishes are being followed. **For ongoing reassurance a parent can request further copies of their child's life support order at any time during their child's hospitalization.**

Disagreement about Life Support Order Decisions:

The physician in charge of a child's care in the pediatric intensive care unit (PICU) may be someone unfamiliar with the child or family. Any child who is acutely ill does not present at their normal baseline. As mentioned previously, few physicians have cared for a long term survivor with trisomy 18 or trisomy 13. *A picture is worth a thousand words* and photographs of this child, taped on the wall by the hospital bed, allow healthcare providers to see the child when well.

A physician's view of these syndromes or mental and physical disability, and quality of life could contribute to his advising a DNR; thus patients with these disabilities 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 physician 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.

In the last decade, end-of-life directive issues have been in the news in which life sustaining measures have been withheld. In these few cases a physician diagnosed the patient as "vegetative" and/or with no hope of

recovery. These cases have made the public aware of the importance of advance directives and have increased acceptance of a DNR order.

Physician - Parent Trust

Crucial to parent or guardian comfort with a life support decision and their acceptance of outcome 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. Physician-parent partnership in Life Support decisions, with **open communication** about what is happening and why, **includes parents** in the recovery of or the letting go of their child.

"A devoted team of doctors... stood by her side and our sides from the time we learned of her diagnosis of cancer, through the surgery that followed and long into the night, agonizing hours of hope and despair and faith, the holding on and finally letting go." [The SOFT touch, 1992]

"...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]

*"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 "Labled" Child's Name is Simon: Fighting for Treatment (2014)*



Simon is the inspiration for
***I Am Not a Syndrome;
My Name is Simon,***
authored by his mother,
Sheryl Crosier.
(See page 92 for link to
purchase.)

Simon
9/7/10 - 12/3/10
Trisomy 18

Jude Wolpert, creator
and administrator of
Faces of Trisomy,
is the mother of Kammie.
See one
of her slideshows
on the SOFT website
under Family Support.

Kammie
5/8/1997
Trisomy 18



*"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

Jonathan
4/3/96 - 8/27/06
Partial trisomy
3,5,13
unbalanced
translocation



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, November/December/January, 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, June/July, 2001]

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. Parents can contact their local Department of Social Services (DSS) or Office of Mental Retardation and Developmental Disabilities (OMRDD) to inquire about services for families with special needs children.

It is important for parents to ask if their state has a **Medicaid Waiver** that **evaluates only the assets of the child** and the income of the parents is not a consideration for qualification. In some states this is referred to as the Katie Beckett Waiver. **The child must not have any assets in order to qualify.** There might be a waiting list. Federal funding of social services decreased in 2010 and budget constraints, in some states, resulted in cuts to these programs. 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 payor but if the child has private insurance, then private insurance is the primary payor and Medicaid becomes the secondary payor. Parents should ask if a Medicaid co-payment is required in their state. The Affordable Care Act of 2010 affects private insurers and thus the insured.

"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, May/June/July, 2002]

"SOFT friends told us about the Community Alternative Program (CAP-NC), funded by Medicaid, which helps families of disabled children. We put her name on the waiting list. CAP provided many hours of nursing services that made a difference in our lives." 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 for parents or guardians to ascertain if the respite service offered is appropriate for their child's care needs. Families report difficulty in finding appropriate programs, particularly for their young adult who ages out of the public school system, but such programs might be worth investigating, if available.

"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

Pediatric Hospice or Pediatric Palliative Care or Perinatal Palliative Care

Because of the infant mortality associated with trisomy 18 or trisomy 13, parents of infants who survive birth to go home, report being advised to utilize the services of Hospice or Palliative Care. In recent years

Palliative Care programs have become more widely available and for the expectant parent, Perinatal Palliative Care is a new care option. Parents still struggling with accepting the predicted prognosis of trisomy 18 or trisomy 13 might be reluctant to use these programs which are often associated with dying. However, these services are usually beneficial to the patient and the family.

It is important to be familiar with these different services and designations. The word *palliate* refers to easing discomfort by treating symptoms. Both Hospice and Palliative Care provide palliative care to ease discomfort and improve quality of life. **The similarities of these programs make for confusion but there is a difference in approach to care:**

Pediatric Hospice Care is for patients who are considered to be in their last six months of life as determined by a doctor. Many physicians do not expect those with trisomy 18 or 13 to survive and referral to Hospice is common for these infants, and also for older survivors who are not doing well. 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 the option of a hospice facility is available. Parents, if wanting facility care, likely need to ask if pediatric placement is available. Hospice might also supplement services a family is receiving through their state, such as nurse's aide hours in the home.

SOFT families have reported using Hospice and their infants did not survive long, but one parent reports that her son with trisomy 18 was discharged from Hospice and he is now 21 years old.

*"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, February/March, 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, May/June/July, 2002]

Pediatric Palliative Care is often recommended for newborns with trisomy 18 or 13 who are able to be discharged from the hospital. Palliative care focuses on patient comfort but also allows interventions such as therapies, treatments and even surgery to promote comfort. Cardiac repair for those with trisomy 18 or 13 will likely not be approved. However, cardiac surgery approach varies by region and team. (Carey JC, personal communication 2013) There are health care providers who are unaware of survivors with these disorders which can affect their advice to parents. It is important to **ask those taking care of your child if they are aware** that 5-8% of these children survive to their first birthday. 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 new care concept for parents who choose to continue a pregnancy after learning their expected baby has a life-limiting diagnosis. Prenatal diagnosis has 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 13 will likely encourage guidance to anticipate loss but it is **crucial that care providers also allow hope** for the predicted small possibility of survival.

These programs are billed the same way a visit to a doctor is billed. Parents should find out how this bill is to be 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.

See http://perinatalhospice.org/Perinatal_hospices.html#U.S._listings for listings of perinatal/palliative care programs and support in the USA and internationally.

*"She was released from the hospital on **Palliative Care** as our wish for her was to pass away peacefully at home." [The SOFT Times, November/December/January, 2003-2004]*

Placement

Parents of newborns with trisomy 18 or trisomy 13 might be advised by the health care provider to consider placing their infant in a facility.

Occasionally, parents of older infants and children are given the option of placement by the doctor, because their child's care needs have increased. Those with any older child, adolescent or adult requiring continual supervision and care, might choose to seek placement.

"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

Parents who make a decision for placement seek a facility that can meet their child's needs and is located close to home. 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. 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 usually ages out of these facilities at twenty-one years of age and his or her court appointed guardian(s), usually the parent(s), bring the young adult home or find an adult facility. Guardianship is discussed later in this chapter.

Parents need to ask about their own rights, obligations, and visiting hour rules. 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 have a right to change their mind and make arrangements to bring their child home. They can apply to the DSS or OMRDD to resume at-home services. A young man with trisomy 18 lived in group home for a few years but his medical issues increased and his needs were no longer being met. 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. The facility is not far from their house and they bring their daughter home for weekends and other special days.

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. The daughter became a resident at this near-by facility as it was decided that placement was best for their family. They visited their daughter often and were pleased with the care she received.

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 should 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, reviewed this chapter and added 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 accurate information about the requirements of their state pertaining to disbursement of trust funds.



"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

Elizabeth
8/11/86
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

Ryan
12/23/1999
Trisomy 18
Mosaicism



"Ashton met Dr. Carey at 12 hours old. He gave us statistics and information (about trisomy 18) to help us make the type of decisions no parent should ever have to make." R.&S.W., 2013

Ashton
10/11/1999
Trisomy 18



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, June/July, 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 the gathering of families at the annual **SOFT Balloon Celebration**, a memorial event, where each child is named and a balloon is released in their memory.

“She will always be Mama’s Baby and Daddy’s Little Girl” K&RR, Michelle, trisomy 18

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. Involvement with others who understand is beneficial. Bereavement support classes, some specifically for the **loss of a child**, can be found in most communities through **Hospice**; 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. Social media provides the opportunity to connect with others who understand.

SOFT Facebook Page

<https://www.facebook.com/Trisomy18.Trisomy13.Awareness.SOFTrelatedDisorders>

SOFT Facebook Group <https://www.facebook.com/groups/TrisomySOFT/>

Trisomy Angel Parents <http://www.facebook.com/groupsTrisomyAngelParents>

See more Grief Resources on page 97.

*"After Tucker
died we talked
about
adoption..."*

Tucker
4/5/01 - 5/3/03
Trisomy 13



*...and we brought
Joey home
5/21/07."*
*[The SOFT Times,
August/September/
October, 2007]*

Joey
3/6/07
Trisomy 13



Summary of Recommended Routine Medical Care

Discussion between parents and their child's doctor is very important to decisions about initiating evaluations and treatments, and the understanding of expected outcomes.

Consider treating each problem as would be done for other children with disabilities, according to the degree of involvement and best interest of the child.

It is always appropriate to provide nourishment and fluids. The help of a feeding specialist or feeding by gavage or gastrostomy tube is often needed. Assessment for reflux should occur even for those children who appear to be able to bottle or breast feed.

Cardiac evaluation is important at the time of diagnosis in all infants with trisomy 18 or 13 and should include an echocardiogram in the newborn period. Early onset pulmonary hypertension after the age of 2-3 months can be an issue for infants with trisomy 18 or 13 who have a heart defect. A predisposition for pulmonary hypertension is suggested in trisomy 18.

Anesthesia risk for **any** surgical procedure for those with trisomy 18 or 13 and who have cardiac anomalies requires consultation with a cardiologist.

Referral to pediatric pulmonology for evaluation and/or sleep studies is appropriate as well as referral to a dysphasia team for those who have apnea or other respiratory issues and feeding difficulties common to trisomy 18 or 13.

Ophthalmology (eye) consultation is recommended for all older infants with trisomy 13 and when signs and symptoms occur for those with trisomy 18. Routine eye exam is recommended for both disorders for those over age one year.

Neuroimaging to aid prognosis (if important) should be done in newborns with trisomy 13. Neurological examination is recommended for all infants and young children with trisomy 18 or 13. Referral to pediatric neurology is appropriate for evaluation and treatment of seizures.

Pediatric Gastroenterology assessment for chronic constipation is appropriate for infants and children with trisomy 18 or 13.

Abdominal ultrasound screening for Wilms tumor or hepatoblastoma in trisomy 18 is recommended starting at 6 months of age and re-screening every 6 months into adolescent age.

Abdominal ultrasound screening for renal defects is indicated in older infants with trisomy 18 and 13 or those with frequent urinary tract infection.

Audiological (hearing) evaluation is recommended in all newborns and infants with trisomy 18 or 13 older than 6 months. Behavioral testing for hearing can be done in those older than 1 year.

Scoliosis evaluation is a part of routine medical care screening and is recommended for those at age 2 years and older. Referral to orthopedics is recommended, when appropriate, for management of scoliosis or limb anomalies.

Immunoglobulin treatment (Synagis®) for respiratory syncytial virus (RSV) should be considered for infants with trisomy 18 or 13 prior to RSV season.

The study of children with trisomy 18 or trisomy 13 by Baty et al. indicated no unusual reactions to immunizations. Immunizations should be determined on an individual basis. The American Academy of Pediatrics (AAP) has recommendations for children with neurological problems and immunizations. The child's doctor should refer to the AAP recommendations.

The American Academy of Pediatric Dentistry (AAPD) recommends that the first visit to the dentist be no later than 12 months of age or within 6 months of the infant's first tooth erupting. Preventive antibiotic for those with cardiac abnormality, prior to dental procedure, is recommended.

Developmental screening, early intervention, special education and referral to therapies as needed, particularly physical therapy, are recommended for children with trisomy 18 or 13.

Social services, Pediatric Palliative Care or Hospice, respite, and on-line parent support groups, such as SOFT, can help in managing total care.

*"Saskia is the
oldest known person
in England,
living with trisomy 18."*
H.H., 2013

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

Saskia
2/18/92
Trisomy18



Table 11 (for Professionals)
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
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)	Every 6 month abdominal ultrasound at 6 months in children 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, 2005]

Other well-child care recommendations would occur as 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]

Carey's 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. Other professional articles about trisomy 18 and 13 and a checklist for routine health supervision guidelines for use in the primary care setting developed by Carey and Wilson (2005) can be seen at www.trisomy.org.

"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 always the oldest known to **SOFT**, until surpassed in 2011.

Kimberly
12/11/68 - 8/3/05
Trisomy 18



Resources for Parents

The Support Organization for Trisomy 18, 13 and Related Disorder (SOFT)
www.trisomy.org

International Trisomy 13/18 Alliance (ITA)
www.internationaltrisomyalliance.com/

Chromosome 18 Registry and Research Society www.chromosome18.org

Tracking Rare Incident Syndromes (TRIS) <http://web.coehs.siu.edu>

Trisomy Talk Chat <https://www.facebook.com/groups/trisomytalkchat>
SOFT Mom & Me Mother's Day Tribute! <http://trisomy.org/mothers-day-video-2014/>

Living with Trisomy 13 (LWT13) <http://livingwithtri13.org/>

Noah's Never Ending Rainbow (NNER) www.noahsneverendingrainbow.org

Genetic Home Reference www.ghr.nlm.nih.gov/handbook

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>

I Am Not a Syndrome - My Name is Simon by Crosier S, 2012. AllStar Press
www.allstarpress.com Authored by Simon's mother who believes every life has purpose and worth. Go to www.Simonismyname.com for more information.

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/

Mothers in Sympathy and Support (M.I.S.S. Foundation), is an international organization for grieving families. <http://www.missfoundation.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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Care of the Infant and Child with Trisomy 18 or Trisomy 13 [2014]

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