



# TRIS project online module

## Trisomy 18: Overview of the syndrome, care and management, parent experiences and resources

Deborah A. Bruns, Ph.D.

Southern Illinois University Carbondale

Carbondale, IL



# TRIS project Mission statement

The Tracking Rare Incidence Syndromes (TRIS) project seeks to increase the knowledge base on rare incidence trisomy conditions, and to make this information available to families and interested educational, medical and therapeutic professionals. Related outcomes will be the development of appropriate services, advance treatment options, and supports during the prenatal, newborn and subsequent developmental periods. Children affected by trisomy 18, trisomy 13, and other rare syndromes possess unique needs requiring education of those involved in their daily, educational and medical care.

# Continuing Education (CE) information

This module will provide a historical review and current perspective on cases diagnosed with trisomy 18 (full trisomy 18 is the focus). Information includes prevalence and incidence, overview of the literature, phenotypic characteristics, common medical issues and their treatment as well as ongoing management and care. Developmental outcomes will also be shared. A list of resources and a glossary of terms are also provided.



Maristella  
DOB 12/31/09



# Module overview

This module will provide results from the medical literature along with findings from the TRIS project, materials from organizations such as the Support Organization for Trisomy 18, 13 and related disorders (SOFT), media reports (e.g., newspaper articles) and parent experiences. Photos are also provided to illustrate phenotypic features and highlight long-term survivors with this genetic condition.

Taken together, this module offers a synthesis of care and management for cases diagnosed with trisomy 18 within a context of professional recommendations and those involved in day-to-day care.



# Trisomy 18

## Edwards syndrome

The first published account was by Edwards and colleagues in 1960.

The authors described the infant as follows:

"birth weight was 5 lb. 1 oz., length 17 in., head circumference 13 1/2 in. Some unusual features were immediately obvious: an odd-shaped head with wide occipitoparietal and narrow frontal diameters; a broad and flat bridge of the nose; low-set ears; a small mouth, inadequate for breast-feeding; webbed neck...During the first week of life she had several cyanotic attacks..." Presence of VSD was also noted. Infant lived from May 22-October 2, 1959.

Edwards JH, Harnden DG, Cameron AH, Crosse VM, & Wolf OH. (1960). A new trisomic syndrome. *The Lancet*, 275(7128), 787-789.

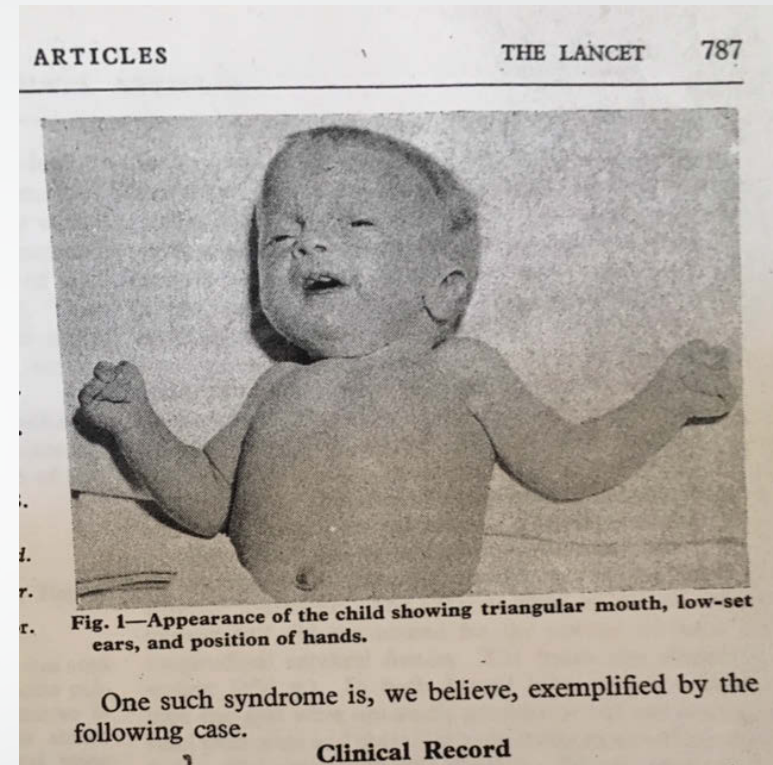


Fig. 1—Appearance of the child showing triangular mouth, low-set ears, and position of hands.

One such syndrome is, we believe, exemplified by the following case.

Clinical Record

# Historical overview

An early large scale study described 27 cases highlighted the following:

- - Incidence of 1 in 6,766 births
- - Very few survived to six months of age (mean = 70.85 days)
- - Low set ears, elongated skull, short neck, ocular hypertelorism and micrognathia in majority of cases. Feeding difficulties were also noted.
- - Most cases diagnosed with congenital heart disease, short sternum and limited hip abduction.

Taylor AI. (1968). Autosomal trisomy syndromes: A detailed study of 27 cases of Edwards' syndrome and 27 cases of Patau's syndrome. J Med Genet, 5(3), 227-252.

A study published in 1985 described 48 cases from a population study over 10 years based on a primary population of 2.2 million. Median life expectancy for live-born infants was five days (range one hour to 18 months). Mean age at death was 48 days. Annual incidence is 14 per 100,000 total births.

Carter PE, Pearn JH, Bell J, Martin N, & Anderson NG. (1985). Survival in trisomy 18: life tables for use in genetic counselling and clinical paediatrics. Clin Genet, 27(1), 59-61.

# Historical overview continued

In 1994, Root and Carey found 64 live-born cases with trisomy 18 of 388,563 total births over a 10-year period in Utah. The prevalence rate was 1 in 6,071 with median survival of 4 days 9% survival at 6 months and 5% at 1 year.

Root S, & Carey JC. (1994). Survival in trisomy 18. Am Journal Med Genet, 49(2), 170-174.

More recently, results on 84 cases of trisomy 18 born between 1974-1997 in Scotland indicated median survival of 6 days with an increased trend for survival for infants who lived through the first 48 hours. Few reached one year of age.

Brewer CM, Holloway S. H, Stone DH, Carothers AD, & Fitzpatrick DR. (2002). Survival in trisomy 13 and trisomy 18 cases ascertained from population based registers. J Med Genet, 39(9), e54-e56.

**In contrast, drawing from hospitalization data from the U.S. Kids' Inpatient Database for 1997, 2000, 2003, 2006, and 2009 (1036-1616 cases per year), Nelson et al. reported over one third of cases were over one year of age and successfully underwent procedures including cardiac surgery and tendon release.**

Nelson KE, Hexem, KR, & Feudtner C. (2012). Inpatient hospital care of children with trisomy 13 and trisomy 18 in the United States. Pediatrics, 129(5), 869-876.

# Historical overview continued:

## Case studies focusing on trisomy 18

The 88.5 day course for an infant with trisomy 18 is described. Markers were noted prenatally. Definitive diagnosis was made with FISH testing shortly after birth. The infant never left the NICU. Infant presented with multiple cardiac anomalies and experienced central apnea. He received prostaglandins but dosage was reduced after review by hospital's Ethics Committee. Lorazepam was given twice during his final 24 hours.

Bruns DA. (2014). Caring for an infant with trisomy 18: A case study and guidelines. Clin Nurs Studies, 2(1), 30-36.

### **Case studies describing long-term survivors with trisomy 18:**

Kelly M, Robinson BW, & Moore JW. (2002). Trisomy 18 in a 20-year-old woman. Am J Med Genet Part A, 112(4), 397-399.

Petek E, Pertl B, Tschernigg M, Bauer M, Mayr J, Wagner K, & Kroisel PM. (2002). Characterisation of a 19-year-old "long-term survivor" with Edwards syndrome. Genet Couns, 14(2), 239-244.

Shanske AL. (2006). Trisomy 18 in a second 20-year-old woman. Am J Med Genet Part A, 140(9), 966-967.



# Current thoughts on trisomy 18

In a 2017 publication addressing care for infants with a prenatal diagnosis, the authors say arguments for withholding treatment are no longer valid as studies and anecdotal reports point to positive outcomes (e.g., cardiac surgery).

The authors also describe patient-centered care as guided by dignity and respect, information sharing, participation, and collaboration and focusing on the parent and child's best interest. This outlook is echoed by Brosco and Feudtner.

Haug S, Goldstein M, Cummins D, Fayard E, & Merritt TA. (2017). Using patient-centered care after a prenatal diagnosis of trisomy 18 or trisomy 13: A review. *JAMA Pediatrics*, 171(4), 382-387.

Brosco JP, & Feudtner C. (2017). Shared decision making for children with trisomy 13 and 18. *JAMA Pediatrics*, 171 (4), 324-325.

# Current thoughts on trisomy 18 continued

Although mortality was the most common outcome, approximately 10% survived to 10 years of age (n=254 live born between 1991 and 2012 in Ontario, Canada). **Among children who underwent surgical interventions (minor procedures to cardiac repair at mean of 205.5 days), one year post-surgery survival was close to 70%.**

Nelson KE, Rosella LC, Mahant S, & Guttman A. (2016). Survival and surgical interventions for children with trisomy 13 and 18. JAMA, 316(4), 420-428.

**“...increasing evidences about efficacy of intensive treatment, slow but constant development in survivors, and positive parental feelings.”** (p. 330)

Nishi E, Takamizawa S, Iio K, Yamada Y, Yoshizawa K, Hatata, T, ... & Nakamura T. (2014). Surgical intervention for esophageal atresia in patients with trisomy 18. Am J Med Genet Part A, 164(2), 324-330.

# Prenatal markers

Although some conditions may resolve prior to birth, the following are considered “**major**” prenatal markers for trisomy 18:

- One or more cardiac defects (most common are VSD and ASD)
- Choroid plexus cysts (*may resolve prior to birth*)
- Neural tube defect (e.g., spina bifida)
- Ventriculomegaly or hydrocephalus
- Omphalocele
- Diaphragmatic hernia
- Anorectal atresia
- Urethrovesical obstruction
- Horse shoe kidney
- Hydrops
- Cystic hygroma
- Two vessel umbilical cord
- Intrauterine growth restriction (small for gestational age)



# Prenatal markers continued

The following are considered “**minor**” prenatal markers for trisomy 18 and primarily related to phenotype (physical characteristics):

- Abnormal head shape (e.g., “strawberry”)
- Cleft lip
- Micrognathia
- Clenched hands
- Overlapping digits or contractures
- Unilateral radial aplasia
- Club feet / Rocker bottom feet



# Prenatal testing and diagnostics

- It is critical to fully explain results of non-invasive prenatal testing (NIPT; also termed Cell Free DNA Screening) to prospective parents including the occurrence of false positives and results are not definitive as this testing only screens for possible genetic issues.
- Testing can begin as early as 10 weeks gestation.
- Parents with NIPT results can use the Predictive Value Calculator to view the risk of trisomy chromosome conditions by maternal age.  
(<https://www.perinatalquality.org/Vendors/NSGC/NIPT/>).

*This is one option for measuring risk of a chromosomal condition, there are others. Important to note that prevalence defaults may not be the best estimates of an individual patient's prior risk for a chromosome condition. Also, sensitivity of the screening test contributes to determining the risk percentile.*

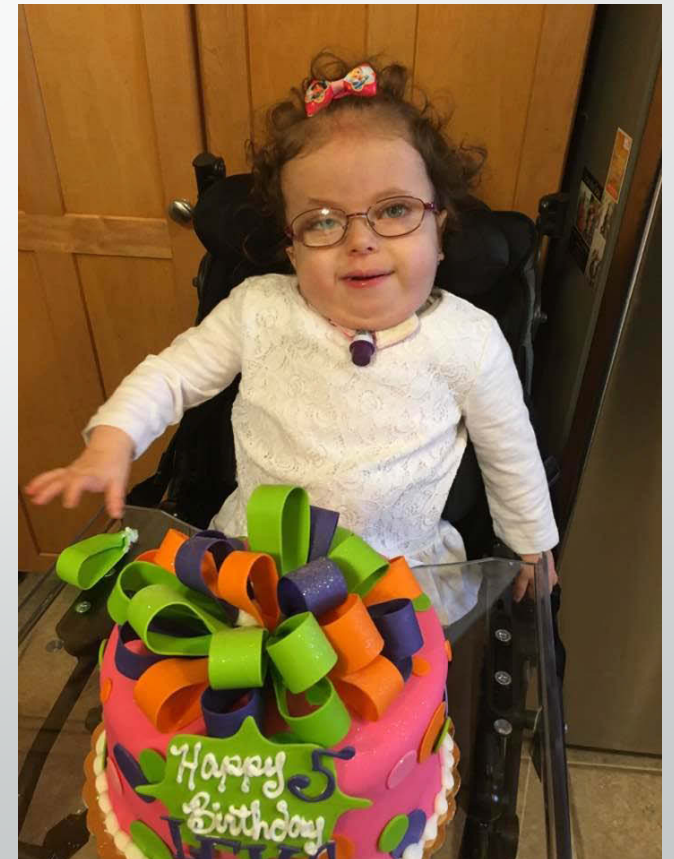
# Prenatal testing and diagnostics continued

- Diagnostic testing is required for a definitive diagnosis. These tests are more invasive than NIPT with a nominal risk to the fetus.
- If trisomy 18 is confirmed prior to approximately 24 weeks gestation, termination of the pregnancy may be recommended. Prospective parents should also be assisted to plan for a live birth including development of a birth plan and active care options. Many hospitals offer a perinatal palliative care program for this purpose. An end of life plan may also be developed.
- **AND** parents and family members are increasingly turning to social media including Facebook groups and blogs to ask questions and gather recommendations (e.g., “trisomy friendly” hospitals and physicians, details about identified anomalies, care options at birth, perinatal course).

# Incidence of trisomy 18

- Based on available data, best overall estimate is 1 in 6,000 live births (Cereda & Carey, 2012)
- Larger prevalence of females at birth and beyond is uniformly cited
- Multiple reports state only 5-10% reaching first birthday BUT anecdotally and review of TRIS project data indicate a larger percentage especially when provided with interventions such as placement of a tracheotomy to assist with airway issues.

Leila  
DOB 12/28/11



# Phenotype

Facial features including:

- Prominent occipital bone
- Short palpebral fissures
- Hypoplasia of orbital ridges
- Low-set ears
- Micrognathia
- Cleft lip and/or palate (*low incidence*)

Karson  
DOB 07/23/13

Jones KL, Jones MC, & Del Campo M. (2013).  
*Smith's recognizable patterns of human malformation*. Elsevier Health Sciences.





# Phenotype continued

Other features including:

- Ulnar or radial anomaly
- Hypoplastic or absent thumb
- Hypoplastic nails (fingers and/or toes)
- Clenched hands
- Short sternum
- Small pelvis
- Rocker bottom feet

Lane  
DOB 12/8/08



Jones KL, Jones MC, & Del Campo M. (2013). *Smith's recognizable patterns of human malformation*. Elsevier Health Sciences.

# Key medical information

- Common cardiac conditions include ventricular septal defect, atrial septal defect, patent ductus arteriosus, and polyvalvular disease (see <http://www.webmd.com/heart-disease/congenital-heart-disease#1> for a general description). Less frequently diagnosed anomalies include pulmonary stenosis, coarctation of aorta, and bicuspid aortic valve. Pulmonary hypertension may occur as well.
- Diuretics and other cardiac medications may be prescribed (see <http://www.webmd.com/heart-disease/tc/congenital-heart-defects-medications> for an overview). Palliative and corrective surgeries are options with the latter being increasingly shown as effective.

# Key medical information continued

- A common respiratory condition is apnea. There are two types (central and obstructive) and may be difficult to determine which is causing breathing difficulties. Some infants may experience both; termed mixed apnea. Conditions such as laryngomalacia or tracheobronchomalacia may contribute to apnea. Supplemental oxygen may be necessary. Providing caffeine during the period after birth is also an option to treat central apnea.
- There is increasing agreement for placement of a tracheotomy to assist respiration. Anecdotally, parents report improved respiratory status and overall quality of life. Some children are able to be successfully decannulated.

# Key medical information continued

- Common renal conditions include horseshoe-shaped kidney, double ureter, hydronephrosis and polycystic kidney. Wilm's tumor (kidney cancer) may occur.
- Reports of hepatoblastoma (liver cancer).
- Omphalocele may occur as well as Meckel's diverticulum and malrotation of colon (requires surgery).
- Defects to male and female genitalia is possible.



# Key medical information continued

- Variety of feeding difficulties due to anatomical anomalies, low muscle tone and/or impaired ability to coordinate suck, swallow and breathing in neonates and infants.
- Anatomical issues may include esophageal atresia with or without tracheoesophageal fistula. Successful surgery has been documented to correct these conditions.
- Concerns about aspiration and reflux affecting intake, weight gain and overall health. Often need supplemental nutrition or fully tube fed to address conditions. For tube feeding, can be blended diets and/or commercially available formula (e.g., Pediasure, Compleat).

# Key medical information continued

- Low incidence of neurological conditions including brain malformations such as hydrocephalus, Dandy Walker malformation and absence of corpus callosum.
- Potential for vision and/or hearing issues. For the latter, narrow ear canals can pose difficulties as well as possibility of sensorineural hearing loss to varying degrees.

# Key medical information continued

In recent years, there has been concern about epilepsy and seizures in children with trisomy 18. Studies have examined affected areas in the brain and effectiveness of medication. Reported results have been mixed.

Kobayashi Y, Ishikawa N, Fujii Y, Nakamura K, Kobayashi M. (2014). A case of trisomy 18 with exacerbation of seizures triggered by administration of valproic acid. *Am J Med Genet Part A*, 164A, 285–286.

Kumada T, Maihara T, Higuchi Y, Nishida Y, Taniguchi Y, Fujii T. (2013). Epilepsy in children with trisomy 18. *Am J Med Genet Part A*, 161A, 696–701.

Matricardi S, Spalice A, Salpietro V, et al. (2016). Epilepsy in the setting of full trisomy 18: A multicenter study on 18 affected children with and without structural brain abnormalities. *Am J Med Genet Part C*, 172C, 288–295.

# Seminal articles

- Carey J. (2012). Perspectives on the care and management of infants with trisomy 18 and trisomy 13: Striving for balance. *Curr Opin Pediatr*, 24, 672–678.
- Cereda A, Carey JC. (2012). The trisomy 18 syndrome. *Orphanet J Rare Dis*, 7, 81.
- Edwards, J. H., Harnden, D. G., Cameron, A. H., Crosse, V. M., and Wolff, o. H. (1960). A new trisomic syndrome. *The Lancet*, 275 (7128), 787-789.
- Pont S, Robbins J, Bird TM, Gibson J, Cleves M, Tilford J. (2006). Congenital malformations among live born newborns with trisomies 18 and 13. *Amer J Med Genet Part A*, 140A, 1749–1756.
- Van Dyke DC, Allen M. (1990). Clinical management considerations in long-term survivors with trisomy 18. *Pediatr*, 85, 753–759.



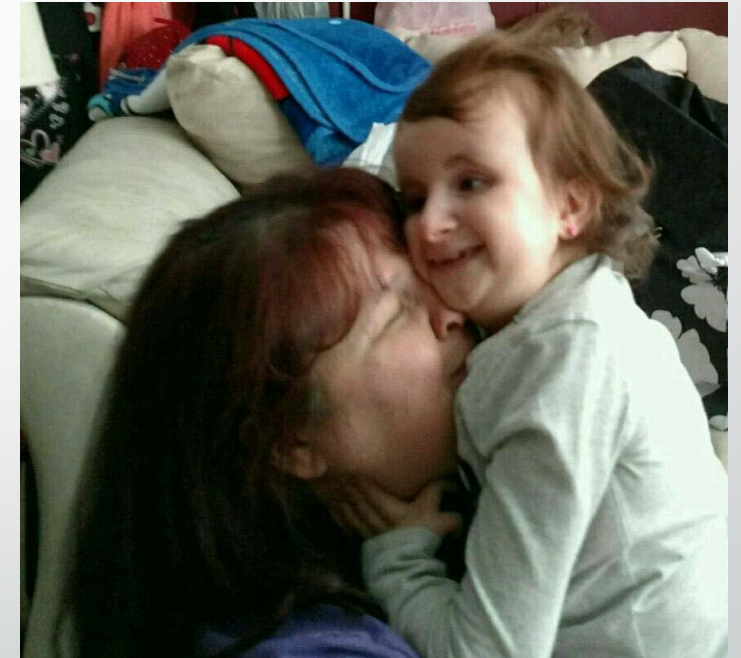
# Additional information

- Braddock B, McDaniel J, Spragge S, Loncke F, Braddock SR, Carey JC. (2012). Communication ability in persons with trisomy 18 and trisomy 13. *Augment Altern Comm* 28, 266–277.
- Costello JP, Weiderhold A, Louis C, Shaughnessy C, Peer SM, Zurakowski D, Jonas RA, Nath DS. 2015. A contemporary, single institutional experience of surgical versus expectant management of congenital heart disease in trisomy 13 and 18 patients. *Pediatr Cardiol* 36, 987–992.
- Derrington SF, Dworetz AR. (2011). Confronting ambiguity: Identifying options for infants with trisomy 18. *J Clin Ethics* 22, 338–344.
- McGraw, M. P., & Perlman, J. M. (2008). Attitudes of neonatologists toward delivery room management of confirmed trisomy 18: potential factors influencing a changing dynamic. *Pediatrics*, 121(6), 1106-1110.

# Developmental milestones

A recent article described developmental milestones; some key findings:

- Strengths in language and communication including purposeful eye gaze or vocalization to make choices
- Indicates preferences for parents, siblings and other family members and caregivers, smiles, and engages in social play
- Demonstrates independent sitting for several minutes at a time
- Able to ambulate with gait trainers and similar mobility devices
- Explores objects with mouth and/or hands



Bruns DA. (2015). Developmental status of 22 cases with trisomy 18 and eight cases with trisomy 13: Implications and recommendations. Am J Med Genet Part A, 167, 1807–1815.

Joey  
DOB  
6/17/09

# Care and management: Special issue of American Journal of Medical Genetics Part C

Carey, JC & Kosho, T. (Editors) (September 2016). Perspectives on the care and advances in the management of children with trisomy 13 and 18, 172(3), 247–308.

Topics include:

- Shared decision making in pre and postnatal management
- Medical interventions and survival by gender
- Treatment of epilepsy
- Treatment of Wilms tumor

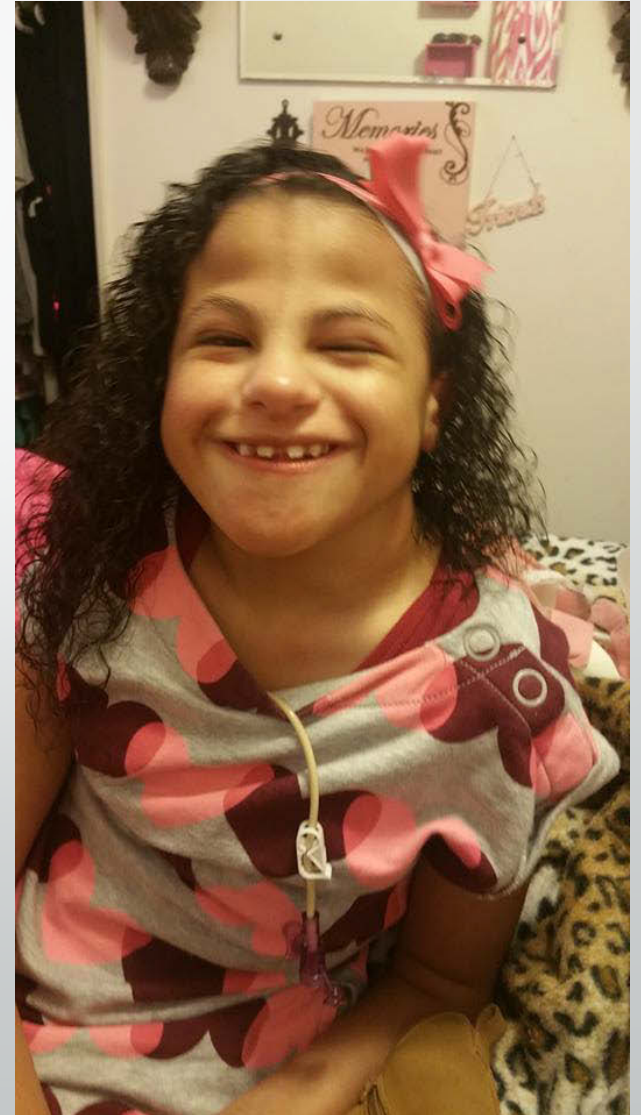
All children at right  
are enrolled in the  
TRIS project



# Care and management

- Monitor cardiac conditions and determine treatment as/if needed. As noted, literature provides positive outcomes for palliative and more intensive care.
- Work with medical team to provide treatment for apnea such as continuous positive airway pressure (CPAP) or bi-level positive airway pressure (BiPAP). Sleep study may be necessary.
- Use of a “Shake vest” to loosen mucus and secretions can assist to decrease likelihood of pneumonia.

Ava  
DOB  
2/26/07





# Care and management continued

- Ongoing monitoring of growth; refer to trisomy 18 growth charts at [http://trisomy.org/?page\\_id=4203](http://trisomy.org/?page_id=4203) or review the SOFT Care Book located at <http://trisomy.org/wp-content/uploads/2014/08/Carebook-updated-8-15-14.pdf>
- Monitor feeding for dysphagia and reflux. There are several tests to determine these conditions such as a videofluoroscopy or upper GI series.
- Work with child's team to maintain oral feeds if appropriate and amount and type of tube feeding (blended diet, commercially available formula).





# Care and management continued

- Monitor for urinary tract infections and constipation.
- Monitor for Wilms tumor and hepatoblastoma; currently, recommendation is every six months.

Satge D, Nishi M, Sirvent N, & Vekemans M. (2016). A tumor profile in Edwards syndrome (trisomy 18). *Am J Med Genet Part C* 172, 296–306.

- Monitor for scoliosis and kyphosis along with orthotics for feet and ankles and/or hands. Some children may need elbow splints to address behavioral issues.



# Care and management continued

- Many children have sensitivity to light; require sunglasses and/or hat for outdoor activity. Many also require glasses to interact with their environment. There is also possibility for cataracts and glaucoma.
- Many children have small ear canals; monitor for ear infections and redo audiological testing as hearing range and acuity can change over time; some children use hearing aids.
- Monitor neurological status; report all suspected seizure activity. Possibility of photosensitive epilepsy has been raised.





# Care and management continued

- Early intervention services for infants and toddlers to encourage gains in developmental areas (in home, clinic, child care setting).
- Preschool-aged children should attend school as tolerated (e.g., one hour session, half day) for developmental activities and socialization.
- Schooling continues for children age five and older. Focus on functional skills such as choice making and assisting with dressing, sensory stimulation, and developing age appropriate interests.

Kammie  
DOB 5/8/97



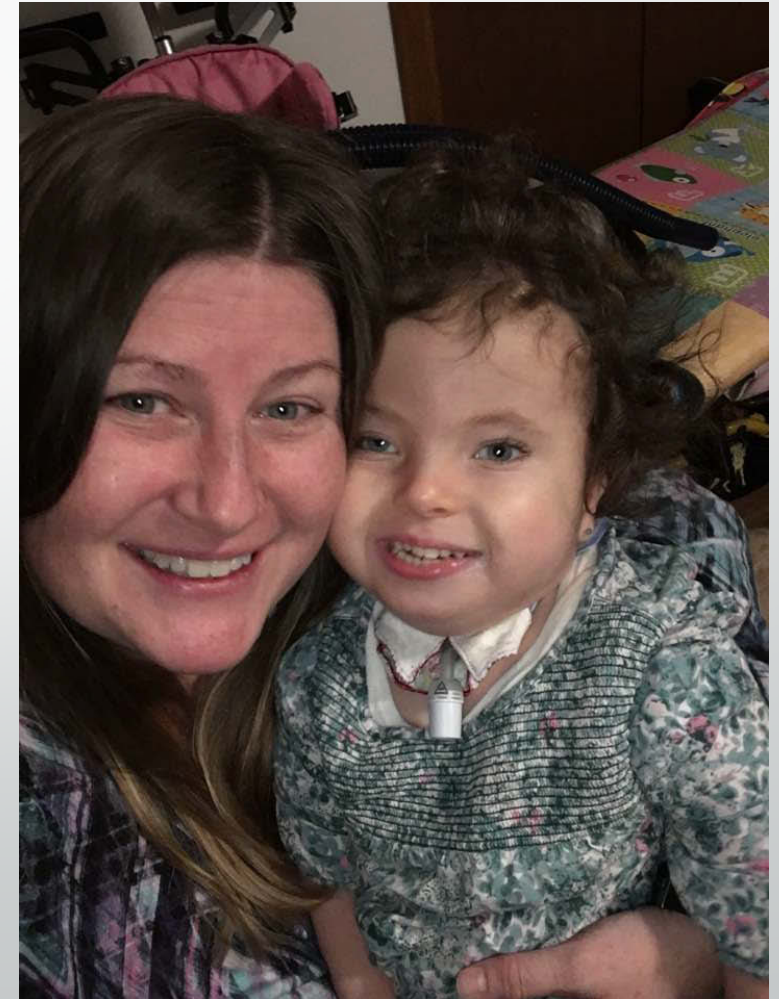
# Special considerations: Medically related

- Immunization schedule adjusted based on child's weight than chronological age as option as well as one per visit to monitor response than multiple at one visit per established recommendations
- Many parents note surgical placement of a tracheotomy to address obstructive apnea offers positive outcomes and ability to provide necessary daily and long-term care. Some children have been successfully decannulated as well.
- Blended diet via gastrostomy tube rather than prepared formulas; collaboration among parents, caregivers and child's team to meet caloric and nutritional needs. Anecdotal reports of positive outcomes. In addition, quality and quantity of oral feeds weighed against aspiration risk, reflux etc.; importance of repeated evaluation.
- Changes at puberty (may be early onset) including start of seizures (may need to reevaluate all medications) and moodiness and other symptoms in girls (majority of long-term survivors reaching adolescence are female)
- Photophobia is sensitivity to light, which can be addressed with use of a hat and sunglasses when outdoors



# Special considerations: Non-medical needs

- Therapy and educational goals including short and long-term outcomes related to parent and caregiver preferences and in range of functional skills (e.g., purposefully activate toy, choice making, ambulation in gait trainer)
- Long-term survivors may need planning in the following areas: guardianship, community and regional adult services, and application to state and national programs for adults with disabilities



Leila  
DOB 12/ 28/11



# Parent and family experiences



Madison  
DOB: 04/25/08

# Parent and family experiences: Children living two months or less

- We [child's parents] leaned on each other and found a new appreciation for one another.
- He [husband] understood me and held me for hours every day while I cried.
- He [significant other] has been so comforting and understanding.
- It's been very hard on us, especially since we are military and live so far away from our families.
- She [mother's sister] was always there and put up with my moods.
- She [maternal grandmother] often speaks of Elise and tells me that she misses her.
- Complete and total understanding and love for us and our situation [from maternal grandparents].
- They [paternal aunt and paternal cousin] were respectful and compassionate.

# Parent and family experiences: Long-term survivors

- We are experiencing this journey together.
- He [husband] shares all responsibility.
- He [husband] does not try to deny or play down her condition. He stays positive always.
- He is my husband and has been very hands on through the whole experience. Although it can be tough we try to understand it is hard to be the parent of a trisomy child and we try to understand each other when we are overwhelmed by the experience.
- We handled the information very differently but found ways to each cope in our own way and still try to work together as a team when needed.
- He [son] is the only other one here, and he assists me and helps me and loves his sister with everything he has.

# Parent and family experiences: Long-term survivors continued

- She [maternal grandmother] loves my daughter as if she were her own. She is the closest person who is as familiar with my daughter's needs as I am.
- She [maternal sister] always listened, offered support and would even take over researching something for me.
- They [maternal sisters] are there whenever we need them, whatever time of day or night.
- She [maternal aunt] has been someone to talk to, helped get information on medical issues, helped to care for Kaiya.
- She [paternal great aunt] listened to my problems and was kind and would babysit if needed.
- They [paternal grandmother and sister-in-law] have been here the most, learned the most about her care and her condition.

# Parent and family experiences: Parent descriptions of unmet needs

## Social and/or emotional support

- We need support, not just hospice, from medical professionals, who do and say things in the best interest of our daughter, not their opinions on her trisomy diagnosis.
- We are not able to spend time as a couple.
- People really understanding what we went through and continue to go through.
- Need help what happens to my son if something happens to me. Nobody knows how to give him all this medicine and do all these things and his sibling just aren't responsible enough.
- I would like to get together with some parents that are going through what I am going through. I am friends on Facebook with trisomy mommies.



# Parent and family experiences:

## Parent descriptions of unmet needs continued

Services, equipment needs etc.

- Respite care is rarely available to me and I don't live close enough to my family to use them for babysitting much.
- Medical insurance doesn't cover needs and cannot get Medicaid due to income.
- There is always equipment or something insurance won't purchase that we want. but we don't publicize these needs because they are more “wants” then true “needs”.
- I still feel scared that the medical community may not give her the care she needs.
- Financially set behind on medical supplies, diaper wipes, gloves. Always have to buy, had to buy cash \$1,000 scooter Medicaid wouldn't pay.
- Juggling a full-time job or career while caring for a child with disabilities is a challenge that is not being addressed by our society.

# Trisomy 18 in the news

"...when she was six months pregnant, doctors diagnosed Brayleigh with a genetic disorder known as trisomy 18...They basically told us that trisomy 18 meant she would be 'incompatible with life'...After six months, she kept getting stronger, she kept growing, she kept thriving...Last month, a huge milestone the Richards weren't sure Brayleigh would reach: her first birthday." *(Georgia toddler born with genetic disorder celebrates major milestone: Her first birthday, Fox 5, Atlanta, GA, November 16, 2015)*

"When you Google this condition, the first thing you'll see in quotes is 'incompatible with life'. Doctors said Ella Grace wouldn't live to see her first birthday...after open heart surgery and with the help of several doctors, the infant's health improved...She may never walk and she may never be able to verbalize but that doesn't mean she doesn't have a purpose here." *(Toddler with genetic disorder defies odds, KATC, Acadiana, LA, January 3, 2017)*

# Trisomy 18 in the news continued

"He's definitely a fighter...Jordan requires around the clock nursing care... She's [Jordan's mother] become well-versed and knowledgeable about the medical world...He makes you look at life differently and appreciate it so much more."  
*(Team Jordan helps Virginia Beach family tackle a serious health issues, The Virginian-Pilot, Norfolk, VA, October 25, 2016)*

"Vera was delivered full term via elective Cesarean section...A week later while still in NICU, Vera was diagnosed with trisomy 18...We brought Vera home assuming she would live at most a year...The first six months were exhausting...We started to enjoy each moment with Vera as if it were our last with her...Against the odds, she began to thrive. When she reached her first birthday, it became clear that she was not going anywhere, anytime soon...Vera is now 20kg and growing...Earlier this year, shortly after she turned eight, Vera had a severe lung infection. We nearly lost her...But this determined little girl is not giving up, so neither can I." *(Caring for a special child: A working mother's experience, Today Online, Singapore, January 7, 2017)*

# Trisomy 18 in the news continued

"Hope survived liver cancer, pneumonia twice, a hernia, three holes in her heart, a club foot, feeding difficulties, breathing difficulties, sight issues and hearing loss...Bobby and Rachel are focused on giving Hope a full life as long as they have her. She goes to the lake, takes scooter rides with her dad and her family can't count how many baseball games she's attended...Hope is the so-called angel in the outfield for her three older brothers' baseball teams."

*(Hope's journey includes beating cancer, outlasting trisomy 18 prognosis, NBC 5 KXAS, Fort Worth, TX, August 27, 2016)*

"Three days later test results showed Alex had trisomy 18...Doctors said Alex might live a couple weeks...Death at our doorstep is what it felt like. Just this looming sense of doom. We just didn't know when it was going to happen...He smiles all the time...Alexander turns 7 months old this week. Alexander is not out of the woods just yet, but his parents are determined to prove a doctor's statistics are just statistics...But doctors don't hold crystal balls. They don't know the future. So I say take it a day at a time and hope until there's no reason to hope." *(Every day a milestone for Alexander the Great, KCCI, Des Moines, IA, July 11, 2016)*

# Additional information: SOFT and ITA

Support Organization for Trisomy 18, 13 and Related Disorders (SOFT) provides information, support and opportunities for face-to-face meetings.

The following are links to some active SOFT groups around the world:

- SOFT USA: <http://trisomy.org>
- SOFT UK: <http://www.soft.org.uk>
- SOFT Australia: <http://www.trisomyaustralia.com>
- SOFT Italy: <http://www.trisomia.org>

The International Trisomy Alliance (ITA) offers resources for medical professionals and family members as well as booklets related to topics including preparation for delivery and supporting siblings. Booklets are available in multiple languages. Homepage is located at <http://www.internationaltrisomyalliance.com>



# TRIS project links

TRIS project website links:

Homepage: <http://tris.siu.edu>

Enrolling in the project: <http://tris.siu.edu/survey/form/PreEnroll.php>

Publications and presentations list:

<http://tris.siu.edu/publications-presentations/index.php>

Case studies: <http://tris.siu.edu/case-studies/index.php>

TRIS project Facebook page:

<https://www.facebook.com/TRIS.Trisomy.project>

