



Common Problems of Babies with Trisomy 18 or Trisomy 13 (Revised 2012)

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Parents often learn from other parents. For nearly three decades, new parents and seasoned parents of the Support Organization for Trisomy 18, 13 and Related Disorders (SOFT) have shared information about their child with these syndromes at SOFT conferences, in newsletters and now on Facebook. A consistent list of problems and concerns has been reported by parents through the years. Viability of their newborn is their first concern, followed by decisions about management of care. Feeding difficulties and constipation are common, often lifelong problems; care of infants with these issues follows below. There is a high frequency of cardiac abnormality in both disorders, and other internal or external abnormalities often occur. Congenital heart disease, apnea (interruptions in breathing) and risk of pneumonia are common and can be life-threatening. With time, motor and mental delays become apparent and chronic health issues such as seizures can develop. The care of infants and children with trisomy 18 or 13 is complex and usually involves many specialists as well as a primary health care provider.

Feeding Difficulties

Newborns too ill or premature to feed are sustained temporarily by intravenous (IV) nourishment until they are able to tolerate feedings. Most infants with trisomy 18 or 13 begin life with feeding problems related to difficulty coordinating their breathing, sucking and swallowing, and often experience choking and sometimes vomiting. *Reflux* (the upward movement of small amounts of stomach contents into the esophagus or throat), *aspiration* (inhalation or trickle of fluids into the lungs), and oral facial clefts further contribute to feeding difficulties and health issues. If appropriate, a baby with these issues might be referred to a dysphagia clinic or feeding specialist to help with feeding problems, or to a gastroenterologist for evaluation.

When feeding orally (by mouth):

- Ask the nursery staff to show you how to position your baby's head up, in good body alignment, because hyperextension of the head, a usual position of infants with trisomy 18 or 13 who have not yet developed head control, makes swallowing more difficult.
- Keep your baby's head elevated about 30 degrees or more during feeding and, if possible, about 30 minutes or more after, while digestion occurs, to help decrease possible reflux.
- Try feeding with pre-softened premie nipples and give only small amounts, offered frequently.

- The help of a lactation consultant will likely be needed, to breast feed. More often than not, these infants are unable to be fed by breast or bottle; however, some parents reported success with bottle feeding, and more than a few reported successes with breast feeding.
- Ask the nursery staff to show you how to burp your baby during and after feeds.
- Babies with oral facial clefts usually require special nipples and instruction from the nursery staff. The Haberman feeder (now called Special Needs feeder) is noted for this purpose in *Trisomy 13, A Guidebook for Families; a parent handbook*, which is available for purchase on the SOFT website. Medela, Inc. makes these feeders and can be contacted at 800-435-8316.

Because of feeding issues, most babies with trisomy 18 or 13 are fed by a nasogastric (n.g.) tube inserted through the nose, or an orogastric (o.g.) tube through the mouth, down through the esophagus into the stomach. Some babies eventually learn to coordinate their suck and swallow, but Dr. John C. Carey, medical advisor for SOFT, notes that those not able to do so by about 6 months of age, are unlikely to progress to bottle or breast feeding. Eventually many parents opt to have a gastrostomy tube (G-tube) placed abdominally. Some babies and children are fed only by tube; others are fed both by mouth and by tube. Some parents feed their babies and children only by mouth. A few parents report that their older child has/had some self-feeding skills.

Reflux

Reflux, a frequent problem in infants and children with trisomy 18 or 13, is usually treated with medication prescribed by a doctor, but referral to a gastroenterologist may be needed if reflux persists. The American Academy of Pediatrics recommends that healthy infants sleep on their backs to decrease the risk of sudden infant death syndrome (SIDS). But *reflux can cause pneumonia due to aspiration* so it is important to ask the doctor what sleep position is safe for your baby. Some children with reflux need consideration of surgical correction.

Surgery

Prior to any surgery, a cardiac evaluation will be needed to determine the safety of doing the procedure. The most frequent surgery done for babies and children with trisomy 18 or 13 is placement of a gastrostomy tube for feeding. The SOFT Surgery Registry maintains a list of surgeries reported by parents on the SOFT membership forms. This list can be seen on the SOFT web site in the Family Support section at the bottom of the home page and in the "Professionals" tab of the main menu.

Slow Growth

Children with feeding difficulties and cardiac problems grow slower and in general are smaller than other children. Ask your doctor about formulas or supplements to help your baby gain weight. A referral to a nutritionist might be helpful. Growth charts for children with trisomy 18 or 13 were published in 1994; a number of participants in this study were from SOFT. These charts are available on the SOFT website, to copy and add to your child's medical record for use in monitoring your child's growth in comparison to others with the same disorder.

Irritability and constipation

All babies cry but frequent crying and fretting is stressful for parents. Some infants might be experiencing colic (crying for no apparent reason) but a common problem of infants with trisomy 18 or

13 is discomfort due to gas, constipation (firm stool) or difficulty in emptying their bowel, no matter what the consistency of the stool.

Feeding problems make it difficult for an infant to get enough nourishment for comfort and growth which in turn results in the baby waking because of hunger. Crying, fretting and feeding problems contribute to a baby swallowing air and any baby experiencing the bowel discomforts mentioned above will be less willing to eat.

Ask your doctor:

- about anti-gas medication and give it enough time to work by using it consistently, as directed.
- for a baby formula that helps form soft stool.
- for advice about stool softener medication (frequently needed)
- about using Pedia-Lax (formerly Babylax). Pedia-Lax is a liquid glycerin suppository designed to relieve *occasional constipation for the 2 to 5 year old; consult with the doctor for those under 2 years.* Dr. John C. Carey, medical advisor for SOFT, advises all parents to check with the doctor before using Pedia-Lax.

Caution: Enemas can deplete a baby of electrolytes and alter body fluids in children. Enemas should only be used with caution and under the guidance of a physician.

Low muscle tone and neurological problems

Problems of muscle tone and nervous system abnormalities affect the development of motor skills. Related to this, many babies and children with trisomy 18 or 13 eventually develop chronic health problems such as scoliosis (curvature of the spine), esotropia (crossed eyes), and constipation. A referral to a specialist is often needed for evaluation and help with these problems. Further, your doctor will likely refer your baby to a physical, occupational, or vision therapist and a speech pathologist. Profound developmental delays affect children with trisomy 18 or 13. They learn slowly, but with time and therapy do attain some developmental milestones. Early intervention programs for infants with special needs are available in most locations as well as special education in the public school starting at age 3 years.

Common Chronic Illnesses and Problems

It is common for those with trisomy 18 or 13 to have chronic illnesses that need monitoring by a doctor, such as congenital heart disease, the eventual development of pulmonary hypertension, elevated blood pressure, obstructive apnea issues, and seizures, as well as frequent pneumonias, and other infections such as sinusitis, cystitis (urinary tract infection), otitis (ear infections) and eye infections that should be evaluated by a doctor.

Varying degrees of vision and hearing deficits are common and need evaluation. Hearing aids, glasses and sometimes surgeries may be of help. Prior to any surgical correction, a cardiac evaluation will be needed to assess the risk versus the benefit. Hypersensitivity to sunlight is common and the use of sunglasses helps when in bright sunlight. See *Helpful Links* on the SOFT web site for information about sunglasses.

Congenital Anomalies

Trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome) have multiple congenital anomaly

patterns (many abnormalities present at birth). Multiple anomalies alert the doctor to the possibility of a syndrome but diagnosis must be confirmed by genetic testing. A new genetic test is now available that can confirm the diagnosis of these syndromes using only maternal blood; this test will likely become the diagnostic standard of care for expectant mothers. For infants born with anomalies but without a prenatal diagnosis, rapid diagnosis can be done with a small amount of the infant's blood. Each syndrome has its own specific set of findings but also some common anomalies such as the presence of cardiac defects. Some findings do not affect the infant's health, making treatment optional. However, some risk the viability of the infant; and, if appropriate, might need consideration of medical or surgical intervention.

Reported Causes of Death

Approximately 1 in 7,000 live births of trisomy 18 and 1 in 10,000 live births of trisomy 13 occur each year in the USA. Survival studies show a similarity in the life expectancy of infants born with trisomy 18 or 13 in that only about 5% to 8% will survive past their first birthday. Dr. John C. Carey, medical advisor for SOFT, notes that apnea (interruption in breathing) in combination with other health factors is thought to be a primary reason for death in the infant. Parents sometimes don't know why their baby died but the usual reported causes are cardiac failure, apnea and pneumonia.

Long Term Survivors

Perhaps in the case of trisomy 18 or 13 the long-term survivor is any child who has lived past their first birthday. SOFT records list many of these children surviving into their teens and several into adult years; with a few being in their 3rd decade. Many of the early problems become more manageable with time however new problems can occur with the progression of heart or lung disease and the development of scoliosis. Periodic health check-ups are needed to help prevent or treat new problems; often different specialists, such as a cardiologist, monitor specific problems. With time, parents shift from waiting for their baby to die to focusing on the many challenges of caring for a handicapped child and finding ways to help them develop their potential. However, if a medical crisis occurs, parents again face decisions and emotions related to the very real possibility of their child dying.

For the Professional

The internet and social media have changed the way people research information and connect with one another. Newly diagnosed parents find other parents through these means and see *positive* comments about these babies and long term survivors. Yet, a common problem for many (but not all) parents of the newly diagnosed fetus or newborn is to hear *only negative* information from their health care providers.

To make informed decisions about continuing a pregnancy, and decisions about care of their newborn, parents need accurate and current information about Trisomy 18 or 13; not only the prognosis but also the fact that 5% to 8% survive beyond one year. Parents will likely be told these infants have significant mental and physical disabilities but also need to know that these infants do respond to comforting, and with time, will come to know their caregiver and begin to enjoy attention.

The conventional view that trisomy 18 and 13 are "incompatible with life" is refuted by the many families living with their child who is a survivor. However the conventional view often influences care options available to these infants.

A common problem for parents wanting cardiac repair is finding a hospital or doctor that will intervene, if needed and appropriate. SOFT has a list of hospitals where cardiac surgeries occurred for infants and children with trisomy 18 and 13. From the www.trisomy.org menu, see the Resources>Surgery section for information. But with recent health care concerns about resources and benefits, some hospitals listed might no longer provide cardiac repair for those diagnosed with these disorders. Recently published studies from several countries, about the outcome of cardiac surgery for infants and children with trisomy 18 or 13, show that approximately 90% survived the surgery and post-operative period and were discharged to home. Parents appreciate the health care provider who helps them find doctors and hospitals (local or elsewhere) that allow consideration of cardiac surgery for infants and children with trisomy 18 or 13.

With nearly three decades of helping families, SOFT is uniquely able to educate parents and professionals about long-term survivors and the issues of care and coping. A check list for routine [*Health Supervision and Anticipatory Guidance for Infants and Children with Trisomy 18 or 13*](#) for use in the primary care setting was first published in the medical literature in 1992 and an updated 2005 revision is available on the SOFT website.

See the [SOFT Store](#) for books about these syndromes. *Care of the Infant and Child with Trisomy 18 or 13; a care book for families* provides a more in-depth discussion of problems and issues of care and living that are addressed in this common problems paper.

Recommended reading on-line at www.trisomy.org includes:

[*What Should We Do Now?*](#) ...after receiving a diagnosis of Trisomy 18 or 13

[*New Trends in Health Care for Trisomy 18 and 13*](#) ...new prenatal test and care option, yet paternalistic care persists and more research is needed

[When in the Hospital](#) ...tips for a safer hospitalization

www.trisomy.org/family-support/ The SOFT website *Family Support Section* lists many helpful articles.

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