What Should We Do Now? Necessary Information After a Diagnosis of Trisomy 18 or Trisomy 13.

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The Need for Information at Diagnosis
For parents facing decision-making following a prenatal diagnosis or diagnosis following the birth of a son or daughter with trisomy 18 or trisomy 13, information is essential, if informed decisions are to be made. Parents are being asked to consider termination of the pregnancy, or to prepare for the birth of a child with a poor prognosis, who will need medical intervention, and to prepare others for what will be not be routine, easily understood or easily accepted. Some will consider adoption, as birth parents or adoptive parents. After birth, the kind and degree of routine and aggressive interventions that will be requested must be understood and articulated. A “do not resuscitate” order may be requested by physicians or parents. Surgery may be considered.

The two most important questions in deciding what to do would seem to be: What will the quality of life be for my child with such a devastating diagnosis? What will the quality of life be for our family in meeting the needs of a child with both physical and cognitive limitations and the need for extraordinary care? Parents need to know they are making the decision that best serves their unborn child or newborn and
addresses their own emotional, practical, philosophical/theological, and financial reality. There is little time available to gather the necessary information and make critical, life altering decisions.

First, parents need to know there is nothing they did that resulted in the trisomy. It is a random occurrence in full trisomies, mosaic and some partial trisomies. More rarely, translocations involving part of a chromosome can be passed from a parent with a balanced translocation (the right amount of genetic material) to the child, who inherits too much or too little genetic material from specific chromosomes. Parents have no control in preventing the third chromosome that attached to another chromosome, rather than separating into different cells, and slipped into the first cell at meiosis. Every cell in the child’s body came from that first cell and carried the extra chromosome (two copies from one parent, one copy from the other parent). Nothing could have been done with earlier information to change the outcome. Nothing can change the cellular, organ and system changes resulting from the extra genetic material from the moment of conception. However, at diagnosis decisions will need to be made and with information, parents gain some control in approaching medical professionals as partners and making informed decisions that are best for their families.

Second, there is some preliminary factual information that must be given to parents facing a new diagnosis of trisomy 18 or trisomy 13, that is, a context for the devastating and confusing information that has just jolted them. It is believed that most conceptions of trisomy 18 or trisomy 13 do not result in live births. Many children diagnosed in the second or third trimester will be stillborn. For live-born children, survival is not assured. There have been many demographic studies, and there is agreement that many infants will die soon after birth, about half will die in the first week of life, and fewer than 10% will reach their first
birthday. Most of the children diagnosed with trisomy 18 or trisomy 13 will not survive long, a sad reality of each diagnosis. Girls with trisomy 18 do better than boys with trisomy 18, both prenatally and postnatally. For unclear reasons, females survive longer. There is no apparent gender difference with trisomy 13. Children succumb early to apnea and other conditions related to neurological immaturity, to structurally defective hearts that cannot keep up with the demands of a growing body, to respiratory and other infections, to complications from corrective surgery, and more rarely in time to renal or hepatic cancers. Nevertheless, there have been a number of long-term survivors with trisomy 18, some few females thriving into their fourth decade of life, some males and more females in the third decade. There have been long term survivors with trisomy 13 surviving into their fourth and fifth decade.

For parents newly facing such a diagnosis there is at least a glimmer of hope that their newborn will survive. This reality should be acknowledged by professionals treating their child. There are survivors. Despite the odds, some children with trisomy 18 and trisomy 13 do survive, develop, and become important members of their families. Trisomy 18 and trisomy 13 are not, as has been declared, “incompatible with life”, nor are the syndromes necessarily “lethal conditions”. Survival depends on the particular constellation of malformations resulting from the extra genetic material, the rest of the child’s genetic code, the medical care and interventions an infant, then child, receives in the hospital and at home, exposure and response to viruses, and probably, an element of chance. It is not understood why some children survive. Some with early risk factors and multiple medical crises during childhood, survive into adulthood.

Third, information is available from parents, who are caring for their children with trisomy 18 and trisomy 13. Trisomy 18 or trisomy 13 each represent a grim diagnosis requiring the
parents to grieve the expected child and embrace a child with a previously unknown diagnosis of limited developmental potential and medical fragility. Parents living with children with trisomy 18 and trisomy 13 have shared what is positive, along with what is stressful in parenting their child. These parents provide an important blueprint of what to expect that goes beyond anything in a medical textbook or on-line description. Reality, although shockingly devastating at diagnosis, is not as negative as many parents report doctors have indicated. Parents report important development in their child and in themselves, great joy along with grief, and delight in what their child accomplishes. They grieve the expected or imagined child and embrace the child with the previously unfamiliar diagnosis. Parents who initially live with a child they expect to die imminently, in time, turn to the business of focusing on a child who will be with them a while, focus on a child who they expect and hope will live. With each medical setback or crisis, these parents want one more year, one more week, one more day with their adored child, as much a part of their family as any child.

Results of a Study of Surviving Children with Trisomy 18 and Trisomy 13
An extensive study of 51 fathers and 66 mothers of 69 living children over the age of one with trisomy 18 or trisomy 13 or a related disorder of those two chromosomes helps to answer the two questions that will guide decision-making after diagnosis. In the study, parents completed the Parenting Stress Index, answered further questions about stress and coping, about the availability and value of social support and the possibility of positive reappraisal of what is initially an overwhelmingly negative situation. Parents also answered questions about services in the community, family interaction with their child, observations of their child at play, attitudes of others toward their child, and educational, medical and respite opportunities they had used. Sixty-five of the children were assessed by the Vineland Adaptive Behavior
Scale to determine developmental scores and age levels, and to compare functioning across scales and across syndromes. Both medical and school records were obtained for each child and coded for archival information in order to gain a fuller understanding of capabilities and constraints.

Of the study sample, 26% of children were ages 1-3, 26% ages 4-7, 25% ages 8-12, 22% ages 13-21 and one individual older than 21. The mean age of those with trisomy 18 was 7.92 years, of trisomy 13, 8.34 years, and of those with related disorders, 6.59 years. In the sample 54% had full trisomy 18, 17% full trisomy 13 and 29% had mosaic, partial or translocations involving chromosomes 18 or 13. In this study, reflecting increased morbidity of males with trisomy 18, 86.5% were girls, 13.5% were boys with trisomy 18, including one teenager. Boys and girls with trisomy 13 were more equally represented. Fifty-six per cent of the children were first born. Only children comprised 26% of the sample being only children; thus, nearly three quarters of the children at the time of the study had siblings. Parents were from 27 states, 3 Canadian provinces and the United Kingdom. Parental educational levels ranged from some high school to graduate degrees. At the time of the birth of their child with trisomy, 70% of the mothers and 59% of the fathers were ages 22 to 34. Only 6% of mothers and 10% of fathers were older than 40. This reflects the reality that although there is an increased chance of having a child with trisomy with advanced maternal age, most births in general are to younger parents, so even with a low percentage of trisomy births, this translates to more infants with trisomy 18 and trisomy 13 being born to younger mothers, who were not at increased risk for a child with trisomy.

What will the Quality of Life be for my Child? Parents want to know if by maintaining a pregnancy or insisting on aggressive treatment after the birth of their child with trisomy, they are making the best decision, when
they choose to support survival in an infant with a poor prognosis and that some doctors predict will have little chance of a meaningful and positive life. The situation is a wanted child with a previously unknown and now unwanted diagnosis. Parents want to do what is best for the child, themselves and their families, helped by making decisions after considering information from families who have faced the same situation, not from old textbooks or from doctors who have never spent time with an older child with trisomy 18 or trisomy 13. Children with trisomy 18 and trisomy 13 are considered to be at the genetic extreme with significant cognitive impairment and multiple physical anomalies, some of which have serious medical ramifications. This means physicians may encourage termination of the pregnancy or be less aggressive in treatment than they would be with other at-risk neonates. The decisions parents are being asked to make are sudden, complex, confusing and emotionally painful, and what they determine they want may be at odds with what physicians recommend or society expects.

Although some parents have been told that there is not the possibility for development or that their child will live in a “persistent vegetative state”, the reality is that children with full trisomy 18 and full trisomy 13 slowly achieve developmental milestones. The one year old is more capable than the two month old and the four year old is making important gains. These children are in Early Intervention programs, and at age three are placed on Individual Educational Plans for school programs that address their needs and foster growth. Most go to school when they are school age. The twenty-two year old is typically in a day program for adults with continued curriculum and meaningful activities. Parents who were told their newborns would not develop are shocked to meet surviving children who demonstrate surprising capabilities and distinct personalities.

Cognitive ability is universally low in those with trisomy 18
and trisomy 13, but they learn and develop as they get older and respond to interaction with others and to specific therapies. The results of the Vineland Adapative Behavior Scales (Interview Edition, Survey Form) for 65 children in this study indicate that overall the children had a mean age score of 13.8 months and mean standard score of 32.48 with Socialization (16.54 months/ SS43.22) significantly higher than Communication (13.68/ SS35.05), Daily Living (13.94/SS27.97), and Motor (11.46/SS30.42) scales. This sample of children demonstrated their strongest development in Socialization: showing affection, showing an interest in others, imitating others, demonstrating preferences, anticipating actions of others, and sharing.

It is Socialization which includes, play, leisure, interpersonal interactions and coping skills, that reflects the child’s ability for interaction with others, agency and enjoyment of life. Nearly a third of children with full trisomy 18 and more than half of children with full trisomy 13 had Socialization scores of at least a standard score of 50. With the widening gap between what the children can do and the age expectations for adolescents, by age 13 all children with full trisomy had Socialization scores below a standard score of 20. This does not represent a loss of ability or lack of on-going development, just a gap between what they can do and what their normally developing peers can do. This would include speech, independent excursions, understanding time, playing complex games and dating. Motor milestones are accomplished typically in the first decade, while attaining competence with socialization tasks extends through adolescence. Children with trisomy 18 attained a mean of 11.5 months in social development with all children functioning below 18 months on the Vineland. For those with trisomy 13 there was a shift toward stronger development with all children functioning at least at a six month level and a third functioning between a year and two years in their social development. This means these children do develop and interact
meaningfully with others, despite lack of verbal communication, limited mobility and for some reduced vision or hearing. To understand what this level of functioning means, think about interacting with a six month old, one year old and two year old. With children with trisomy 18 and trisomy 13, the verbal and some motor items on Socialization are missed because they are non-verbal with reduced mobility, but some social tasks beyond the achieved scale level are met. For instance, a child who cannot name relatives, may show clear concern when someone is sad, taking a hand or gently touching a face.

The widest range of performance on all scales of the Vineland was seen with those with related disorders, which would be expected. Ten percent functioned at a six month level or less, a quarter at a level commensurate with the second six months of life; therefore 35% functioned at a level commensurate with the lowest functioning children with full trisomy. The rest of the children with mosaic, partial or translocations of chromosomes 18 or 13 were evenly distributed up to four years of age in development, with a few higher. Nearly a third of those with less than a full extra chromosome in every cell were functioning overall at least at the level of a three year old and 10% of the sample of those with related disorders were functioning at a four year old level or better. Developmentally, there was a wider range for children with related disorders with some as disabled as those with full trisomy and others significantly more capable. Development is less predictable for those with related disorders of chromosomes 18 and 13, which relates to particular cell lines with the third chromosome, the part of the chromosome that is triplicated, and medical set backs and neurological damage related to surgery and illness.

The questions on the Vineland did not capture all the children could do, and in initial written responses and later in lengthy telephone interviews parents presented anecdotes and
observations that revealed evidence of important social functioning. The children respond differently with different people, understanding expected activity level and seeming to know what they can get away with. They direct specific emotional responses to specific people, especially the mother. Many act differently with siblings than grandparents. They make social choices in accordance with preferences and clearly enjoy themselves. They can be coy, jealous and charming. They flirt, resist, provoke, ignore and show concern. They laugh heartily, show affection and act contrite. As they age they make wants known, initiate interaction, indicate expectation, imitate others, show humor, respond emotionally to situations, and manipulate others to achieve what they want. Many school-aged children demonstrated a clear sense of agency, taking control to gain attention, objects or a preferred activity. Many parents reported the need for guidance or reprimand to stop negative behaviors, as with any child. Some reported a sharp, “no!” could be followed by intentional persistence and even a smirk. Obstinancy, including intent and agency, even in a usually compliant child, reflects welcome development. Social-emotional development is evident in children with trisomy 18 and trisomy 13, despite negative predictions made by those who have not spent adequate time with older children with trisomy.

Parents in the study reported good quality of life for children with trisomy 18 and trisomy 13, despite their many limitations. They described their children as happy, content and sociable, despite limited cognitive abilities, greatly reduced mobility and a reliance on rudimentary nonverbal communication. Parents described development across the lifespan, despite being told there would be no development. The children slowly reach important developmental milestones. Many of those with full trisomy 18 or trisomy 13 develop object permanence, work to master tasks and seem pleased when successful, gain a sense of agency seen in delayed but welcome “terrible 2s”, tease others, get bored, manipulate others, and
show disappointment. They understand cause and effect and try to bring about changes. They are curious, exploring their environment despite structural limitations of hands and feet, and for some sensory defensiveness, and sensory impairments in many. Their parents described them as adored and adorable, functioning members of their families, who make important contributions to family life, are loved, require and give affection and enrich the lives of parents, siblings and others. That is not to say that there is not a difficult process involved in coming to terms with what the infant, then child, cannot do. In time, often quickly, the child becomes more than a complicated diagnosis, and parents see the many capabilities of their child—abilities not disabilities. Having a medically fragile child with limited cognition and physical inabilities certainly raises questions of “why?” before there is acceptance and appreciation of the child’s unique personality and place in the family.

One mother reflected on her life with her daughter and wrote, “The heartaches, the disappointments, the grief is deep and raw. Yet, the love, the joy and choice to experience happiness while enduring the sorrow brings an exquisite clarity to what is happening, so that it can be fully appreciated. While I wouldn’t wish my daughter’s condition on anyone, I wouldn’t trade this time with her for anything in the world.”

Many children with full trisomy, as well as those with related disorders who are more likely to be higher functioning, clap hands, stamp feet, give high fives, hide their eyes, play turn-taking games, pull coverings away, share and sometimes pretend. They turn pages of books and point to pictures. They point to wants on communication devices. They are tickled, carried on shoulders, swung in the air, and they dance with others in and out of their wheelchairs. They enjoy pools, swings, carnival rides and music. They go on cruises, fly, go camping, go to sporting events, church, restaurants, Disney World, and national parks. They live life with their families
who pack up their gear, often in a van with a wheelchair lift, and go. Day to day they are typically in the thick of things with siblings with interactions ranging from spontaneous affection on both sides to sharing and even some rough and tumble play. They sit with groups that are socializing and laugh when others do. They crawl, roll, cruise, walk and wheel to join others and interact. They reach out and engage strangers. With age, children interact differently with different people, amuse themselves with favorite toys, musical devices and television and always thrive being with others. If children with autism tend to turn from others and exist in a world apart, children with trisomy 18 and trisomy 13 gravitate to where the action is, engage those around them and rise to any social occasion. With others, they are happy, animated, and at their best. Although some with related disorders show frustration with what they cannot do, and thus some irritability, those with full trisomy 18 and trisomy 13 do not typically react to what they cannot do, although they will show frustration to the agendas of others: too much time at the mall, too long in the doctor’s office. Parents reported they get bored by inactivity, but they tend not to get frustrated by their own inabilities.

There are times when children with trisomy 18 and trisomy 13 are ill, uncomfortable, and in pain, more often than with normally developing children, but most of the time their parents report they are enjoying a good quality of life as happy, engaged individuals. There are medical setbacks, crises and worry, but most of the time the children are content and happy with positive moods and no distress. One mother shared,

“When I was pregnant with him, and we learned of his condition I was most heartbroken thinking of my baby never smiling and responding to touch. This was the picture painted for us. If he was born alive he would be in such a negative state. I used to pray that my baby could at least be able to smile and recognize my face. Boy, were my prayers answered. He loves
life, his quality of life is terrific. He is a very happy and responsive child far from the picture painted of a baby whose condition could be incompatible with life.”

What Will be the Quality of Life for the Family?
Parental responses on the survey forms they completed indicated a paradox. On one hand, mothers and fathers are stressed by the demands of parenting children with medical uncertainty and challenges related to both unusual and constant daily routines. Sleep may be at a premium, and bathing, feeding, dressing, lifting, repositioning, and entertaining, are daily and lifelong. With trisomy 18 and trisomy 13 the baby or toddler never grows up, never achieves any degree of independence. Nevertheless, many described their life with their child as an unexpectedly positive experience, full of joy and a pathway to unexpected opportunities. Several parents described their child with trisomy as easier than their normally developing children. One mother declared her child, “a perfect child in an imperfect body.”

Stress levels are higher in parents with children with trisomy than in parents with typically developing children. In this study parents of children with related disorders reported the most stress. When elevated stress on The Parenting Stress Index was examined, 34% of mothers and 48% of fathers of children with full trisomy reported elevated stress on the Child Scale and 20% of mothers and 25.7% of fathers reported elevated stress on the Parent Scale. With children with related disorders (many of them ambulatory and verbal with minimal judgment) 73.7% of mothers and 62.5% of fathers reported elevated child-related stress. For the Parent scale, 31.6% of the mothers and no fathers reported parent-related elevated stress. This means stress was more likely to come from child characteristics such as demandingness, short attention span, adaptability and mood than from parent characteristics such as isolation, competence, role restriction, spousal support or health. The greatest area of
stress was acceptability, which relates to physical, emotional and intellectual characteristics of the child. No parent reported only minimal level of stress in that specific area. Demandingness refers to noncompliance and dependence in higher functioning children and dependence in lower functioning children unable to do things for themselves. Some parents acknowledged the presence of demands but added that they did not believe they were stressful for them, although marking them elevated the stress score. This might mean that G tube feedings would elevate the stress score, although parents came to find them routine and not a source of any actual stress. On the Parent scale both mothers and fathers reported feeling competent in their roles with only low levels of depression. Mothers felt stressed because of low spousal support when the child was young; fathers felt less attached to the young child. This is not surprising with 97% of the fathers in the study working full-time and only half the mothers working, some only part-time, and many mothers of young children staying at home.

Asked to comment on negative changes in themselves related to care of their child, themes emerged of fatigue, isolation and anxiety about the child’s health. Health concerns in the parent related to fatigue, strain with frequent lifting of the older child and lifting equipment, and little time for exercise. Although many described themselves as becoming more tolerant themselves, parents were bothered by intolerance in the medical community, in those advocating abortion, in those denying rights to the disabled and in those with materialistic values. Some reported a crisis of faith, while others reported a heightened spirituality related to their experience with their child. Isolation was often self-imposed but related to avoiding those uncomfortable with their child, to having difficulty finding competent sitters, and for mothers, to being at home all day with young children. Some parents reported in time feeling most comfortable with other parents with children with disabilities. Many parents reported that
unusual procedures, that at first seemed stressful, such as feeding procedures, using a wheel chair, and repositioning, in time become routine, second nature and thus not areas of stress.

Everything is Not Routine
There are concerns inevitable with a child with trisomy 18 or trisomy 13 that threaten to overwhelm parents also dealing with jobs, financial constraints, household tasks and often other children. Medical fragility is an on-going concern that becomes less an issue as the child ages and matures neurologically, benefits from surgical interventions and strengthens immunity. In the study, the number of hospitalizations in the past year correlated with maternal stress levels (Adaptability, Mood, Health of Parent). Parents were asked about hospitalizations in the previous year to determine the extent of medical intervention necessary for their child. This study did not include infants less than one year of age, but for the youngest children the early months would have been included. Of those with full trisomy 18, 51.4% had no hospitalizations in the previous year. Of those with full trisomy 13, 41.7% had no hospitalizations. Half of those with Related Disorders were not hospitalized. Hospitalizations decreased significantly with age. Of those ages 1-3, 89.9% were hospitalized at least once; of those ages 4-7, 50% were hospitalized during the previous year; of those ages 8-12, 29.4% had been hospitalized the previous year; and 31.2% of those at least age 13 were hospitalized the previous year. There were gender differences with 43.1% of females and 72.2% of males hospitalized in the last year, a function at least in part of more older females. Gender and age were significant for hospitalizations but diagnosis was not. Hospitalizations were not always emergency admissions. Hospitalizations for young children were often for surgical procedures such as G tube placement, Nissen fundaplication, and ear tube placement. There were also surgeries of cleft lip/palate, hernia, undescended testes, and strabismus. For some there was heart
surgery. An analysis of surgeries in this study sample indicates that 74.5% of females and 88.1% of males have had at least one surgery with a mean of 1.2 and 1.8 surgeries respectively. An analysis by diagnosis indicates 83.8% of those with trisomy 18, 83.3% of those with trisomy 13 and 65% of those with related disorders had had at least one surgery (means: 1.6, 1.4, 1.7, respectively). Those ages 8-12 were least likely to have had surgery, reflective of a trend toward more surgeries in children younger than this age group and in increase of age-related surgeries (scoliosis, cataracts, joint deterioration, tumors) with older individuals. There were also hospital stays for respiratory and other infections, and seizures. Hospitalizations were stressful for parents because of attendant uncertainty for survival, difficult decision-making and balancing work and child care demands.

On-going medical concerns that do not always require hospitalization that parents reported as stressful included: constipation, respiratory infections, scoliosis, reflux, ear infections, urinary tract infections, neurological conditions, the need for weight gain, pain management and difficulty related to heart conditions. Prevalence of these medical conditions was significantly higher than would be found in normally developing populations, reflective of physical anomalies and the chronicity of their management. These on-going medical concerns took their toll on parents in their chronicity, potential for a life threatening episode and need to be with the child, rather than sleep or attend to other demands. Parents reported that constipation with subsequent discomfort for the child, and the need for medication and diet changes, as well as, respiratory illness, feeding issues and sleep irregularity were areas of concern related to time and effort, as well as, emotional concern. These are areas of reported stress related to the child’s physical/medical management.
Although in this study a third of parents reported no difficulties with constipation (21% with trisomy 18, 33% with trisomy 13, 50% with related disorders), almost half report nearly constant concern, particularly for those with trisomy 18 and related disorders of the eighteenth chromosome. Respiratory infections tend to be frequent and potentially critical with children with motor limitations, a tendency in some to aspirate, thoracic malformations, scoliosis and weaker immune systems. A snuffle can quickly become serious. Although 27% of parents reported their children were fine in that respect, 20% of parents reported their child had had pneumonia and required intubation, 6% had required a tracheotemy and 7% had been hospitalized for a respiratory infection that was not pneumonia. Children with trisomy 18 were more likely to be hospitalized and have a tracheotomy and less likely to be fine (only 18%) in terms of respiratory illness.

In terms of feeding interventions, parents in this study reported 68% of those with trisomy 18 are fed by G-tube, 67% of those with trisomy 13 and 25% of those with related disorders. Without G-tubes feedings can be lengthy, aspiration common and weight gain minimal. Most children with G-tubes in this study eat orally as well, but G-tubes better ensure nutritional and hydration needs being met and subsequent weight gain and also simplification of administering medications. Oral defects are common. Parents reported 16% of those with trisomy 18 and 42% of those with trisomy 13 and 25% of those with related disorders did not have oral defects. Most notable is the 25% rate for cleft palate and 17% rate for both cleft lip and palate for those with trisomy 13, which complicates feeding, and ultimately may require surgical repair.

Sleep patterns in those with trisomy 18 and trisomy 13 can mean disrupted sleep, affecting two thirds of the children, although 20% of parents reported the children outgrew the problem. The pattern of wakefulness did not vary with
diagnosis or feeding method. Children can be wakeful, sleep intermittently, rise early or fall sleep late. Some parents reported night nurses to be a solution to their own subsequent sleep deprivation. The chronicity of disrupted sleep makes this stressful for parents without provided support, which varies by state.

Heart defects, which are common in these syndromes relate to more serious medical management. In this study of children who survived at least a year, only 20% of those with trisomy 18, 40% of those with trisomy 13 and 50% of those with related disorders were without heart defects. The heart abnormalities include most commonly ASD, VSD, both ASD/VSD, tetrology of Fallot, dextrocardia, and accompanying PDA. Heart surgery is a possibility, and many children have successfully undergone repair, but such surgery is a risk to a weak child. Physicians are not always willing to perform such surgery on a child with a poor prognosis, so parents wanting surgery may have to find a willing cardiologist.

There are other problems that add to parental stress reported by parents in this study. Seizures affected nearly half this sample of children, mostly those with full trisomy, less than 20% since infancy, 12% after age one and another 9% after age 5, the rest only a few times. Parents reported seizures are problematic when hiring babysitters, and because medication and continued seizures reduce alertness, focus and energy in the child. Scoliosis and sometimes kyphosis, curvatures of the spine, affect half of those with trisomy 18, 42% of those with trisomy 13 and 35% of those with related disorders. The curvature compresses, which compromises respiration and digestion, mobility and overall physical comfort, and necessitates frequent repositioning. Orthopedic jackets and surgery have been solutions to improve quality of life for the child.

Sensory impairments are related to diagnosis and affect the child’s ability to interact in the world. Although in this
study 65% of children with trisomy 18 had good vision, only 17% of those with trisomy 13 had adequate vision with 25% having no vision, 25% legally blind and 33% having partial sight. For related disorders, 70% have fine vision with those with mosaic and partial trisomy 13 most likely to have reduced vision. Children with trisomy 18 often have photosensitivity and need sunglasses. For hearing, 47% of those with trisomy 18 had fine vision, only 6% profoundly deaf, but 47% with reduced hearing. Two thirds of those with trisomy 13 were fine, but 17% are profoundly deaf, a quarter with partial hearing.

Parents reported that children with sensory impairments rely on other senses, including touch, to interact with others and rely on stronger senses to explore their world. Although sensory impairment can be stressful to parents, most children reportedly readily adjust to responding with their stronger senses.

Some of what parents in the study sample found stressful relates not to the characteristics of the child but to the response of others to the child. Some parents reported they are stressed by the negative attitudes by some physicians, but most manage to find physicians that are supportive and willing to take the child’s lead into new territory. Some parents have difficulty coordinating multiple appointments. Stress relates to lack of respite and nursing services in some states, judgmental relatives, and strangers who are less than accepting of a child who looks different and has cognitive and physical challenges. The stress is not so much in time demands with the child but in dealing with those who are not receptive to the child or the needs of the family. Parents can feel isolated by the attitudes and responses of those uncomfortable with their child.

Siblings must be considered in determining quality of life for the family. From interviews and panel discussions after the study, siblings shared what are overwhelmingly positive experiences. Siblings born after the child with trisomy
encourage young parents to have more children, not be afraid that life will be less for normally developing siblings. Siblings reported that having a brother or sister with trisomy 18 or trisomy 13 is overall a positive experience. They typically adore their sibling and become competent in handling daily routines. They rise to responsibilities, develop a sense of competence, and live their own life with sports, activities and jobs without feeling held back. On the downside, they have to be more careful when they are sick, so the germs are not spread, have to contend with others who are uncomfortable with someone with cognitive and physical limitations, and are disrupted by middle of the night emergencies. There was consensus by the siblings that the benefits far outweigh any inconveniences.

This information gathered from parents of surviving children with trisomy 18 and trisomy 13 should help those facing a recent diagnosis in understanding what they will face in parenting a child with trisomy 18 or trisomy 13. As in the case of those trying to understand a new diagnosis, this is not the journey these parents expected to take, but they learned about their own adaptability, resilience and growing expertise as they cared for their child. They became experts able to share what they have learned with the next generation of parents facing a similar diagnosis and its unique challenges.

If you are a parent newly facing a trisomy diagnosis and have questions, need someone to hear your concerns and need guidance, please contact your local SOFT contacts, who are all parents of children with trisomy. These parents are listed under chapters by state on the SOFT website. For more extensive information, three books: Trisomy 18: A Resource for Families, Trisomy 13, A Resource for Families, and the newly revised Care of the Infant and Child with Trisomy 18 or Trisomy 13, which all include extensive information from a number of studies, experts and articles from the newsletter,
are available for purchase at trisomy.org website. They are highly recommended as important reference guides. SOFT membership also includes informative articles in the quarterly newsletter. Requesting the family packet will provide families with newsletters and brochures with important information.

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