Trends in Health Care for Trisomy 18 and 13

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Our youngest, Megan (1985-2004), was about two months old when diagnosed with trisomy 18. It was a time when little information about trisomy 18 was available. We were told she had a very small possibility of living to her first birthday and no information was provided about survivors. Her doctor had seen only one other patient with trisomy 18, who died in infancy.

One of the most significant developments in prenatal care is the maternal serum triple screen which tests for the possibility of increased risk of trisomy conditions. By the early nineties it was a routine part of prenatal care in the USA and likely also in other developed countries but it requires invasive follow-up testing for confirmation of a diagnosis. A decade earlier due to maternal age, we were offered but declined amniocentesis because of the small risk to the fetus. Had triple screening been available, a positive result, which means further testing is warranted, might have altered our decision about amniocentesis and most certainly would have added anxiety to what had seemed like a normal pregnancy. However, prenatal diagnosis of trisomy 18 or 13 has ramifications that affect the focus of care provided by an obstetrician and termination or continuation of the pregnancy of a fetus with developmental and physical disability is discussed.

The introduction in 2012 of non-invasive prenatal testing (NIPT) from a single maternal blood draw as early as ten weeks gestation has been found to be a reliable diagnostic test for trisomy 21, 18 and 13 without the need for follow-up invasive tests. With time the NIPT will likely become the new standard of care for all pregnancies; thus increasing prenatal detection of these syndromes.

A common trend in obstetrics is to not consider caesarean delivery an option for these babies. Thus begins parental realization that health care management might be an issue of concern. Most of these pregnancies end with elective termination (75% in the USA and 90% in Europe), miscarriage or stillbirth and in both disorders only 5-8% of live births (estimated at 1/7000 for trisomy 18 and 1/10,000-20,000 for trisomy 13) will survive to celebrate their first birthday. [Carey, JC (2012)] Current information about survivors and care allow parents to make informed decisions, but still today, some parents are not provided accurate information.

Perinatal Hospice is a relatively new trend in palliative care that has come about because of prenatal diagnosis and is now an option available to parents who choose to continue a pregnancy after a diagnosis of little to no hope of fetal survival. Perinatal palliative care focuses on a creating a birth plan, end of life wishes and if there is a live birth, comfort care. While planning for probable loss it is crucial that these palliative care providers also allow hope for the predicted small possibility of survival when advising these families. Perinatal and Pediatric Hospice/Palliative programs are still evolving. Approach to life extending cardiac surgery for trisomy 18 and 13 varies by region and provider.

In the mid-eighties doctors did not support aggressive treatment, such as cardiac surgery, for an infant with trisomy 18 or 13. Rather, the standard of care for these babies consisted of comfort measures only and parents who asked for intervention were likely told “not for this baby.” Hospice is recommended
when doctors do not expect a patient to survive and was suggested for our daughter but we declined. Nearly three decades later many of these newborns still receive comfort-only care and a referral to hospice or palliative care but since the early nineties a growing number of infants and children with trisomy 18 and 13 have benefitted from life-extending surgery. The Support Organization for Trisomy 18, 13 and related disorders (SOFT) has maintained a Surgery Registry since 1988, composed of information provided by parents on the membership registration form. This data includes cardiac repair of a number of children with full trisomy 18 and full trisomy 13 as well as mosaic cases of these disorders.

Paternalistic medicine (the doctor knows best) was the conventional standard of care delivered for decades in the USA and is still accepted practice today. In recent years a patient-physician partnership in decision-making, which considers the wishes of the patient or parent about interventions to be done or not done, has also become an accepted standard of care in the USA. Physicians who manage their patients’ health care with such a partnership are more likely to support, if appropriate, a parent’s desire for life-extending intervention for their child, but finding a willing surgeon and hospital can be difficult and even more so today with the health care focus on resources versus benefits.

In 1987 SOFT brought together, for the first time, a group of parents seeking information, help, and contact with other parents. These parents were and are advocates for their children and those who agree to participate in studies about these disorders contribute to educating interested professionals. Since 1960 there have been individual case studies of older children with trisomy 18 but it was SOFT that opened the opportunity to do studies involving a number of children. Pediatrics, the official journal of the American Academy of Pediatrics, was first to publish a study of a group of children with trisomy 18 from the SOFT membership (with parental permission). [Van Dyke DC, Allen M. 1990]

Medical conferences and journal articles discussing trisomy 18 and 13 educate physicians worldwide. While a majority of doctors still practice only comfort care for these babies, physician advocates are beginning to open discussions with their peers about these children and their health care management. Dr. John C. Carey, Medical Advisor and Co-founder of SOFT, speaks nationally and internationally on behalf of these children and has authored numerous articles about these syndromes, including Anticipatory Guidance and Health Supervision in Children with Trisomy 18 and 13, a summary of health care management suggestions for these children. [Carey JC, 1992] Hundreds of clinical reports and several studies about these conditions have been published throughout the world (Carey, 2010). In particular, Baty and colleagues collaborated with SOFT and published two studies in 1994 on medical problems, growth curves, and development, in the largest series of studies up to that time. (Cited by Carey, 2010) Publication of these first, and as yet only, growth charts was considered to be an impressive accomplishment by our daughter’s physician. Additionally, international SOFT support groups are now present in a number of nations and continents, including Japan, Australia, and Europe.

In the early nineties the Do Not Resuscitate (DNR) order form began to be used in hospitals to define what life support actions were to be done or not, if a cardiopulmonary event occurred to the patient while hospitalized. Concurrence from a parent is obtained by discussion. But, the DNR order can be misused either through error or purpose which is a breach of trust between physician and parent and can result in death. Some hospitals and physicians have the foresight to request a parent signature on a DNR to prove concurrence but unfortunately many do not, making it unlikely a parent will see what is
written on the order form. This vestige of paternalistic medicine is convenient for the doctor but risky for the patient, particularly the disabled. “Get it in writing” is common sense advice that is a safety precaution in any agreement between two persons. Changing the DNR laws to require signature from the physician and the parent or guardian, when possible, will help prevent misuse of this order. This is a safety issue and, if ever achieved, will improve the in-hospital care of all patients.

In recent years, social networks have opened a floodgate of comments from parents who have or had a child with a chromosome disorder, raising public awareness about issues surrounding trisomy 18 and 13 and they voice a common concern about the outdated “not compatible with life” label associated with trisomy 18 and 13 that some health care providers still use when speaking to parents about their newborns, which ignores the survivors and gives no hope to new parents. Carey estimates there are approximately 300-400 living persons with trisomy 18 or 13 over age one year living in the USA today. (personal communication, 2010) The SOFT web site can be found at www.trisomy.org and it allows instant access to information for parents and professionals. But perhaps it is not yet well known as there are still parents who are not informed about such resources by their health care providers.

“Health care is too important to leave to the health care provider.” (Terri Graedon, The Peoples Pharmacy, 2012). We have a personal responsibility to maintain our own and our dependents health and care information, and to provide it to each doctor we see. If we do not understand what has been said, or why a procedure needs to be done, delayed or is being denied, or the purpose of medication prescribed and the side effects, it is in our or the child’s best interest to question the doctor. If a patient is hospitalized, Graedon recommends asking for the Attending Physician to come and answer such questions. If dissatisfied with the answers, it is appropriate to seek a second opinion.

Health care is historically slow to change. Medical care of these children is complex and modern health care management is based on scientific evidence. Studies about long-term survivors definitely help to update doctors about these disorders. Significant is recent public awareness of Senator Rick Santorum’s three year old daughter, Bella, who has trisomy 18. His 2012 presidential campaign has educated the world about trisomy 18 and might well lead to a new trend of better health care for these special children.

References:


Van Dyke DC, Allen M (1990) Clinical Management Considerations in Long-term Survivor