INTERNATIONAL TRISOMY ALLIANCE

e Newsletter ISSUE 2

www.internationaltrisomyalliance.com December 2014

Welcome to our second ITA e newsletter

In this issue we have updates by three of our founders, Barb Farlow, Debbie Bruns and Siri Fuglem Berg, and reviews of our publications. These reviews include the free-to-download ITA publications and Dr Siri Fuglem Berg's recently published book about her daughter 'Evy Kristine', now available from Amazon (USA and UK Amazon in English and a Norwegian translation).

There are articles by ITA Chief Medical Advisor Professor John Carey MD., and a new and highly regarded addition to our advisors, Professor Martin MacCaffrey, a neonatologist and paediatrician. Valued contributions have been received from Hana Sroka, a genetic counsellor, and Lanise Shortell, a paediatric clinical care nurse specialist who writes from a hospice perspective.

There are parent advisor updates from Italy, Romania, Norway and the United Kingdom sharing some of the work they do to support families.

Over the last twelve months ITA has posted more translations of booklets on their website and added research papers published in 2014.

The founders wrote and produced 'Trisomy 13 and Trisomy 18: Preparing for Your Baby's Arrival, a research based parent-friendly book which John Carey described as "a landmark in information for prospective parents of a baby with trisomy 13 and 18." This can now be downloaded free from the website.

Our mission is to offer trisomy 13 and trisomy 18 support groups, physicians and other professionals, accurate information and resources, and to be a mechanism for sharing between groups.

To show how we are fulfilling our aim of providing information about trisomy 13 and trisomy 18 to the international community, ITA has added two new pages to the ITA website, Founders Publications and Books and Founders Presentations and Seminars

Finally, I have the pleasure of maintaining the website, co authoring the ITA books, and editing the magazine.

Best wishes Jenny Robbins

Founders of I.T.A. Barb Farlow and husband, Jenny Robbins, Debbie Bruns and husband, and Siri Fuglem Berg MD and family

Contents

Proposed Research Agenda: Trisomy 13 and Trisomy 18 by John C Carey, MD, MPH

John C Carey is the Chief Medical Advisor of ITA

Recognizing the recent proliferation of original research on the biologic, medical, and social aspects of the trisomy 13 and trisomy 18 syndromes, I considered it timely and relevant to propose a **Research Agenda** for the remainder of this decade. This proposal is simply one person's view and is humbly and respectfully submitted as a topic for discussion and reflection. I recognize the multiplicity of medical issues involved in the care of persons with these two medically complex conditions.

The following list includes what I would consider the highest priority areas of future investigation, again acknowledging the subjectivity of this one person's opinion. The order is not by the highest priority as all 10 belong at the "top."
My proposal for priorities of research centers on the causal and management aspects of the two syndromes and consists of the following:
1) Detailed analysis of the outcome of heart surgery in infants with both trisomy 13 and trisomy 18;
2) Update and expansion of growth curves for infants, children and adolescents with both syndromes including the original data by Baty, et al 1994 (available on the US and UK SOFT web sites);
3) Precise estimation of the risk for Wilms tumor and hepatoblastoma in children with trisomy 18 and determination of any tumor risk in children with trisomy 13;
4) Determination from population studies of survival in children with trisomy 13 and with trisomy 18 over the age of 1 year;(it is vital in these studies that the ascertainment of survival after 1 year come from infants ascertained near birth and living past 12 months of age);
5) Comprehensive analysis from the population sources of the variables that enter into survival in infants with trisomy 13 (including the absence of holoprosencephaly) and trisomy 18;
6) Development of international registry of cases with trisomy 13 mosaicism and trisomy 18 mosaicism for the study of natural history and outcome of these conditions;
7) Characterization of individuals with partial trisomy 18, including 18p and the various forms of 18q in collaboration with Chromosome 18 Registry;
8) Characterization of individuals with partial 13q trisomy;
9) Investigation of the quality of life of parents and siblings of children with trisomy 13 and with trisomy 18 using validated and standardized quality of life instruments;
10) Development of novel approaches to investigation of nondysjunction and related predisposing factors to the events of altered segregation. And an un-numbered priority: development of mouse models for both trisomy conditions.

Articulation of one person's "wish list" of research into the rare disorders of trisomy 13 and trisomy 18 is naive at best. The challenge is to precisely pose the research questions, hypotheses, and study designs and to pursue the appropriate funding and resources to complete the studies.

John C Carey, MD., M.P.H.


John C Carey

John C. Carey, MD, MPH, is Professor and Vice Chair of Academic Affairs, Department of Pediatrics, at the University of Utah. Throughout his career, Dr. Carey has been interested in birth defect syndromes and the care of children with these conditions. Dr. Carey graduated from Villanova University in 1968 with an A.B. and obtained his M.D. from Georgetown University School of Medicine in 1972. He trained in pediatrics, genetics and dysmorphology as a resident and fellow at the University of California San Francisco, 1972-1979. Dr. Carey obtained an M.P.H. from the University of California at Berkeley in 1976 in between his residency and fellowship years. Dr. Carey joined the faculty at University of Utah Health Sciences Center in 1979. He became Chief of the Division of Medical Genetics in 1985 and remained in that leadership position until 1999 when he stepped down to assume the role as Editor-in-Chief of the *American Journal of Medical Genetics*. He has held that editorial position since 2001. Dr. Carey established the Medical Genetics Fellowship Program at the University of Utah and continues as Program Director.

Dr. Carey's research focus has been in congenital malformations, neurofibromatosis and syndrome delineation. He has authored or co-authored over 280 papers, chapters, invited articles and editorials for scientific journals. He also co-authored the textbook, 'Medical Genetics,' by Jorde, Carey, and Bamshad, now in its 4th edition. The book is a widely used text in schools of medicine throughout North America and Europe. Dr. Carey has served as medical adviser and 'founding professional' for the Support Organization for Trisomy 18/13 and Related Disorders (SOFT) since 1980. The medical and ethical aspects of care of infants and children with these conditions are currently some of his major academic interests.

Founders - Publications Presentations Books and Seminars in 2014

Publications authored or co authored - Debbie Bruns

Nine children over the age of one year with full trisomy 13: A case series describing medical conditions. American Journal of Medical Genetics Part A, 164A(12), 2987-2995.
Maternal and paternal age at pregnancy for low incidence trisomy groups: Preliminary findings and implications. Journal of Genetic Disorders and Genetic Reports, 3(2).
Twenty-two long-term survivors with full trisomy 18: Presenting and current medical conditions. American Journal of Medical Genetics Part A, 164A(3), 610-619.
Caring for an infant with trisomy 18: A case study and guidelines. Clinical Nursing Studies. 2(1), 30-36.

Publications authored or co authored - Barb Farlow

CPS position statement for prenatal counseling before a premature birth: Simple rules for complicated decisions. Paediatrics and Child Health, 19(1):22-24.
Arrogance-based medicine: guidelines regarding genetic testing in children. American Journal of Bioethics, 14(3):15-6.
Communications with parents concerning withholding or withdrawing of life-sustaining interventions in neonatology. Seminars in Perinatology, 38(1):38-46.
End-of-life decisions for extremely low-gestational-age infants: why simple rules for complicated decisions should be avoided. Seminars in Perinatology, 38(1):31-37.
Our children are not a diagnosis: The experience of parents who continue their pregnancy after a prenatal diagnosis of trisomy 13 or 18. American Journal of Medical Genetics A, 164A(12), 308-318.

Publications co authored – Siri Fuglem Berg

Ethics and etiquette in neonatal intensive care. JAMA Pediatrics, 168(9):857-8.

Presentations presented or co presented - Debbie Bruns

TRIS project findings: A resource for parents and professionals. Invited session presented at the Support Organization for Trisomy 18, 13 and related disorders (SOFT) Conference, Norfolk VA.

Presentations presented or co presented - Barb Farlow

Harvard Hospital System, Grand Rounds, June 2014
Massachusetts General Hospital, Presentation, June 2014
Syracuse University, Symposium, April 2014

Books

Siri Fuglem Berg author **Evy Kristine, A Beautiful Journey**
Available Amazon USA http://www.amazon.com/EVY-KRISTINE-Siri-Fuglem-Berg-ebook/dp/B00NQ78YX4/ref=sr_1_1?ie=UTF8&qid=1414422334&sr=8-1&keywords=siri+fuglem+berg
Available Amazon UK http://www.amazon.co.uk/EVY-KRISTINE-Siri-Fuglem-Berg-ebook/dp/B00NQ78YX4/ref=sr_1_1?ie=UTF8&qid=1414426367&sr=8-1&keywords=siri+fuglem+berg
Available in Norwegian: http://www.z-forlag.no/produkt/evy-kristine-retten-til-et-annerledesbarn/

Seminars - Dr Siri Fuglem Berg

Sept 14th, **Trisomies**, Norwegian Parliament, invited speaker
June 11th, **Prenatal diagnostics**, Aarhus University Hospital, Denmark; invited speaker
April 10th, **Dialogue around the unborn life**, Salvation Army, Oslo, Norway; invited speaker
April 4th, **Personal meeting with Norwegian Minister of Health**, invited speaker
March 12th, **Ethics seminar,** Grefsen Church, Oslo, Norway; invited speaker
February 16th, **Leader training for medical students,** Norwegian Christian Medical Association, Hurdal, Norway; invited speaker
February 5th, **Trisomy 18/13**, Frambu Center for rare diagnosis, Oslo, Norway; invited speaker
January 30th, **Ethics seminar, Medical Philosophic Forum North**, Bodø, Norway; invited speaker
January 18th, **Southern and Western Norway´s Conference for KrFU**, Christian Party´s Youth Organization, Bergen, Norway; invited speaker

Martin J McCaffrey MD, CAPT USN (Ret), Professor of Pediatrics

The founders of the International Trisomy Alliance are very pleased that Doctor McCaffrey has accepted our invitation to be a medical advisor. His extensive experience in neonatology as well as his compassion and understanding of parents who give birth to a child with trisomy 13 or trisomy 18 will be a wonderful asset.

It is an honor to be invited to be a member and advisor for the International Trisomy Alliance. As a neonatologist I have been blessed to work with a variety of heroic parents and patients on a regular basis. Many of our most challenging patients often survive, and many of these survivors may lead what the world accepts as a reasonable quality of life.

This perception of quality of life changes everything. When a neonatologist counsels a family with an impending delivery of a 23 week old baby that has not received a full course of antenatal steroids, the prognosis delivered is one that is grim, with perhaps a 10% chance of survival, and only a 10% chance of survival without profound neurodevelopmental impairment. Despite the challenges such a baby will face, if the parents state their desire to fully support their baby, we are all in as neonatologists.

We have seen some of these babies recover and attain a reasonably normal level of life functioning. In my experience, certainly within the first week of life as such infants start to declare their ability to survive; no one withholds therapeutic interventions without open and clear conversations with the family members.

The case for infants with a known diagnosis of trisomy 18 or 13 is decidedly different. Many of us as neonatologists were raised with the understanding that infants with trisomy 13 or 18 had a lethal disorder. I cannot remember spending any significant time considering the literature on these infants beyond what was written in iconic textbooks. I don't recall ever having a parent challenge the dismal prognosis for a baby with trisomy, nor do I recall willfully suppressing knowledge that trisomy 13 and 18 were not fatal diagnoses.

It has been a long, dark journey for families dealing with these conditions over the last twenty years. Despite an increasing prevalence of trisomy 13 and 18 diagnoses, there have been significant reductions in live born infants with trisomy 13 and 18.[1] As a result of newer prenatal testing, the spread of ultrasound evaluations and the development of newer pregnancy termination methods, the termination rates for trisomy babies have risen dramatically. These changes in obstetrical practice, coupled with landmark publications in 1990s pediatric literature recommending limitations in the care for such infants, have fortified the characterization of trisomy 13 and 18 as "lethal".[2, 3] This classification has been a critical element in the significant reduction of live born infants with trisomy 13 and 18, and the withholding of life support measures which has led to the generally reported short life span of such infants.

Dr. Carey in his inaugural remarks for the newsletter suggested that over the last decade a dialogue on the approach to infants with the diagnosis of trisomy 13 and 18 has evolved. I believe this is the case. The opportunity for parents of these children, and these children, to be heard and seen, has opened as a result of the heroic efforts of a few researchers demonstrating that trisomy 13 and 18 are challenging and difficult diagnoses but not lethal. [4] As important has been the rising tide of increasingly more informed parents coping with the challenges of a child with a trisomy diagnosis. This tide has risen as a consequence of social media and the creation of advocacy groups such as International Trisomy Alliance. This tide will need to continue to rise. Neonatologists have been asked about their views on resuscitating trisomy 18 infants and a study reported that only 44% would be willing to do so.

Professor Martin McCaffrey with daughter Shea

What is most remarkable is the language used by the authors to describe their finding. These observations raise the concern that some neonatologists are abandoning the best-interest standard, which would require that providers agree only to treatment strategies that are consistent with furthering the good of the infant, and instead are adopting an 'ethic of abdication' in their approach to difficult treatment/non-treatment decisions.

A second factor opposing those advocating for patients with trisomy 13 and 18 is the infusion of the concept of 'quality of life' into care of patients who will have what society views as 'limitations' in their life. Quality of life is regularly applied to patients who are at high likelihood of suffering severe cognitive deficit. There appears to be for many a bar for worldly interaction that, if not met, leads to judging a life as painful, incomplete and without the capacity to be happy. Continued on next page

Martin J McCaffrey continued from previous page

It is not necessarily the medical challenges that the individual faces that make the life less meaningful, it is the inability to interact and experience joy in an obvious way that is a primary consideration in judging such lives to be less meaningful.

The AMA has actually codified this in their Code of Medical Ethics, Opinion 2.215 - Treatment Decisions for Seriously Ill Newborns. “Care must be taken to evaluate the newborn’s expected quality of life from the child’s perspective. Life-sustaining treatment may be withheld or withdrawn from a newborn when the pain and suffering expected to be endured by the child will overwhelm any potential for joy during his or her life. When an infant suffers extreme neurological damage, and is consequently not capable of experiencing suffering or joy, a decision may be made to withhold or withdraw life-sustaining treatment.”

Those waving the banner of “quality of life” are a powerful force opposing anything but the most basic care for infants with trisomy 13 and 18. In a 2012 survey of neonatal providers, the overwhelming majority underestimated the data reported survival of infants with trisomy 13 and 18 to discharge. Ninety one percent reported quality of life for such infants to be “non-viable” or poor. Ninety two percent recommended only comfort care. Forty one percent of providers were willing to override parents request for maximum intensive care. The study concluded that maximum interventions were recommended when quality of life by the providers was perceived to be good.

I am at an extreme disadvantage in engaging the quality of life conversation with many of my more intuitive and knowledgeable colleagues. I believe I was absent the day they gave out the quality of life crystal balls in medical school. As a result I have been forced to resort to more plebian tactics in counseling and caring for families with a child with trisomy13 or 18. Using the best available data regarding outcomes for infants with trisomy 13 and 18, and asking the fully informed family what their goals are for their child has served me well in my clinical practice.

Clearly quality of life is in the eyes of the beholder but the most important verdict on an acceptable quality of life for an infant with trisomy should be rendered by the family that will be caring for the child. There has been recently published work to help us better understand the view families have on the quality of life for their children with trisomy 13 or 18. A web-based survey was tabulated from 332 parents on 272 children. Parents of children who died described the overall experience of their child’s life as being positive (88%); 68% had no regrets and 31% regretted that they did not consider more interventions. The children had significant developmental delays but gained milestones over time (Table 4). Almost all parents (95%) reported they communicated with their children and understood their needs; 99% of parents described their child as a happy child. [9]

These are the times that try the souls of many parents experiencing pregnancy with a child with trisomy 13 or 18. This diagnosis is a difficult one but it is not lethal, just as birth at 23 weeks is challenging but not lethal. There is increasing willingness on the part of some providers to offer fetal based maternity care (monitoring, steroids and cesarean section for fetal distress), resuscitation and intensive therapies such as intubation, heart surgery and tracheostomy to infants with trisomy, but parents often have to resort to support groups or social media to identify such providers and facilities.

In a world that prizes perfection, the desire to support the life of infants with Trisomy 13 and 18 is often met with rebuke; and the simple truth is that labelling a condition lethal, and saying it enough times, generally does make it lethal. Lethality begets lethality. [10] Advocating for children with trisomy 13 and 18 is a privilege. The innocence of such spirits, whether they live for 10 days or 10 years, is something we will never be capable of experiencing. Yet we call these children handicapped? If we had eyes to see we might realize who the handicapped truly are.

Bibliography

1. Irving C1, Richmond S, Wren C, Longster C, Embleton ND. Changes in fetal prevalence and outcome for trisomies 13 and 18: a population-based study over 23 years. J Matern Fetal Neonatal Med. 2011 Jan;24(1):137-41
2. Bos AP, Broers CJM, Hazebroek FWJ, et al. Avoidance of emergency surgery in newborn infants with trisomy 18. Lancet 1992; 339:913–917.
3. 5. Paris JJ, Weiss AH, Soifer S. Ethical issues in the use of life-prolonging interventions for an infant with trisomy 18. J Perinatol 1992; 12:366–368. [Context Link]
4. Carey J. Perspectives on the care and management of infants with trisomy 18 and trisomy 13: striving for balance. Curr Opin Pediatr 2012; 24: 672–8.
5. Guon J, Wilfond BS, Farlow B, Brazg T, Janvier A. Our children are not a diagnosis: the experience of parents who continue their pregnancy after a prenatal diagnosis of trisomy 13 or 18. Am J Med Genet A. 2014 Feb;164A(2):308-18.
6. Janvier A, Farlow B, Wilfond BS. The experience of families with children with trisomy 13 and 18 in social networks. Pediatrics. 2012 Aug;130(2):293-8.
7. McGraw MP and Perlman JM. Attitudes of neonatologists toward delivery room management of confirmed trisomy 18: potential factors influencing a changing dynamic. Pediatrics. 2008 Jun;121(6):1106-10.
8. AMA Code of Medical Ethics, Opinion 2.215 - Treatment Decisions for Seriously Ill Newborns. http://www.ama-assn.org/ama/pub/physician-resources/medical-ethics/code-medical-ethics/opinion2215.page?
9. Perspectives on anticipated quality-of-life and recommendations for neonatal intensive care: a survey of neonatal providers. Salmeen K, Janvier A, Sayeed SA, Drey EA, Lantos J, Partridge JC. J Matern Fetal Neonatal Med. 2014 Sep 18:1-6.
10. McCaffrey MJ. Lethality begets lethality. J Perinatol. 2011 Sep;31(9):630-1; author reply 631-2.

Evy Kristine, A Beautiful Journey by Siri Fuglem Berg

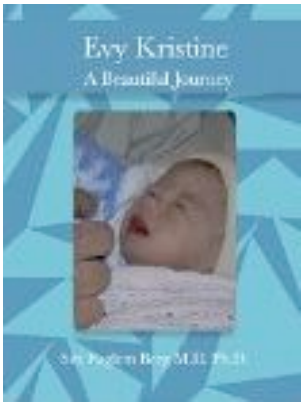
Dr. Siri Fuglem Berg is a founder and a medical advisor of ITA.

"As a physician I want to share my experience of carrying to term my daughter with trisomy 18. Our journey was beautiful and enriching, in spite of the obstacles we met in the health care system - the pressure to terminate, and the lack of medical care for babies with trisomy 18. I hope my book can be a support for other women carrying to term a baby that did not turn out as expected."

'Evy Kristine, A Beautiful Journey' is available from Amazon USA, Amazon UK and in Norwegian: see Books on page 3 of this e newsletter for links.
http://www.internationaltrisomyalliance.com/other-publications.html

Dr Siri Fuglem Berg

Book Review - Evy Kristine, A Beautiful Journey by Karin Kaaen USA



I have just finished reading the book, 'Evy Christine; A Beautiful Journey', written by a doctor who had a child with trisomy 18. It was enlightening to read from the perspective of a trisomy parent in the medical field. Not only were her personal feelings towards carrying a child to term with this diagnosis very comparable to many of us in the same situation, but her findings from a doctor's perspective were interesting as well.

Dr Fuglem Berg was asked numerous times about termination, which she knew from the start was something she did not want to do. Yet, the medical field seemed to offer no alternative for a parent who wants to carry a trisomy baby to term.

As a woman who is 28 weeks pregnant with a trisomy 13 baby, I have been equally shocked at the medical community's blatant recommendations NOT to carry to term. They use 'quality of life' as an argument for these children to not be treated, yet every child I have seen who has survived is smiling and happy and loved.

Siri Fuglem Berg M.D. Ph.D. also points out that children who have been diagnosed prior to birth do not receive the life saving care that undiagnosed children do, therefore the odds of survival go down dramatically simply because doctors refuse to treat a child due to the diagnosis of trisomy. Siri compares this to the way Down Syndrome (a trisomy 21 diagnosis) was once treated not too long ago, but now there are doctors and specialists who work with children with Down syndrome, and many lead very productive and satisfying lives.

This book is a very good read for anyone, whether a family member of a trisomy baby, or someone looking to learn more and get a real perspective of the process we as parents go through after a diagnosis of trisomy.

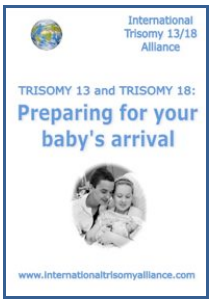
Free Booklets for Families

www.internationaltrisomyalliance.com/ita-booklets-for-families

The International Trisomy Alliance booklets for families and children are free to download on the ITA website, and the links to other publications include the USA 'Care of the Infant and Child with trisomy 13 or 18'.

A Cherished Pregnancy, Family Dictionary, and booklets for younger and older children

ITA Translations



TRISOMY 13 and TRISOMY 18
Preparing for your baby's arrival

Hope for More Newborns to Go Home by Barb Farlow MBA

A founding member of International Trisomy Alliance

One of the questions asked in my research of 107 parents who received a prenatal diagnosis of trisomy 13 or trisomy 18 was, “What was your hope when your baby was first diagnosed?” The majority of them responded that they hoped their baby would be born alive and that they might come home. Sadly, most babies with a prenatal diagnosis do not live long enough to go home. However, research might allow more parents to achieve their hopes.

Regarding stillbirth, from my experience it seems that few parents are offered the opportunity to monitor their baby as carefully as any baby at risk for stillbirth would be monitored. Monitoring might avert a stillbirth. Research into the rate of stillbirth in the last weeks of pregnancy for women who are being monitored is needed.

Decades ago, babies born with trisomy 13 or trisomy 18 had a longer median survival, even though medicine was not as advanced. Over the same time that median survival decreased, prenatal diagnoses increased and recommendations were developed to withhold resuscitation to newborns and limit care to comfort only. Historically, trisomy babies were born at a much greater gestational age (half of babies with trisomy 18 were born after 42 weeks) but now, as the recent population study from the UK (England and Wales) reveals, many babies are induced and are born much sooner. Furthermore, articles have reported that babies with a prenatal diagnosis face much bleaker outcomes than those with a postnatal diagnosis

It is possible that inducing labor and withholding respiratory support leads to early death for a baby that might have needed only a day or two of support before being discharged home from hospital. Caring for baby at home is an important desire of parents.

Preliminary examination of the data from my research and a recent poll of parents in support groups reveal that the need for respiratory support (oxygen, CPAP, ventilation) is highly sensitive to gestational age, even after 37 weeks when non-trisomy babies would be considered to be full term. Babies born after 37 weeks with a postnatal diagnosis received full care at birth and very often needed only a couple of days of respiratory support before being discharged home. Yet, if that support had been withheld, the baby would likely have died in the first day or two.

Important research is needed regarding the effect of induction, the benefit of prenatal steroids before birth and surfactant after birth for the possibility of premature lungs on babies that are otherwise considered to be mature.

The co-founders of ITA plan to undertake or inspire this research to help more parents achieve their hope of bringing baby home.

Translations of ITA Books

http://www.internationaltrisomyalliance.com/ita-booklet-translations.html

The books for children on the ITA website have been translated into Italian, Norwegian, Turkish, Spanish, Indonesian and Finnish. The new ITA book, “Preparing for your baby’s arrival’ is currently in the process of being translated into Norwegian and Italian. We are reliant on volunteers and would love to include other translations, especially German, French, Spanish and Japanese. Offers to translate ITA books into other languages would be gratefully received.

Tracking Rare Incidence Syndromes (TRIS) project update by Debbie Bruns, Ph.D.

A founding member of International Trisomy Alliance

Debbie Bruns

In the first ITA newsletter (December 2013), I introduced the Tracking Rare Incidence Syndromes (TRIS) project. The past year has continued with enrolling new families, analyzing data and sharing results with interested audiences. For this entry I'd like to shift the focus to what I bring to the International Trisomy Alliance and, hopefully, to all of you reading this newsletter.

My background is as an educator. I taught for six years in New York City (USA). Most of the children I worked with had severe, multiple disabilities and intensive medical needs. I knew very little when I started my position and this was after four years of university training.

My students represented a very, very small group in relation to those with learning disabilities and so called 'high incidence conditions'. Neither my courses or teaching placements included many children with significant needs so I learned by trial and error, consulting with my colleagues including other teachers, therapists, social workers and nurses, and researching information.

In fact, when I first looked up trisomy 18 in the early days of the Internet, most of what I found was autopsy photographs. This was when I was working with not one, not two, but three preschool-aged girls with trisomy 18. Autopsy photographs? What I needed was information about encouraging oral feeding, developing problem solving skills, motivating language etc.

I feel akin to the other ITA Founders in this way. I started my journey feeling very ill equipped to meet my students' educational, social and behavioral needs. Funny thing was my students still made gains. It was a team effort and we celebrated progress, no matter how large or small. We figured things out with these three wonderful children leading the way. I think Siri, Barb and Jenny would agree with this thought about their girls. No matter the length of time each was here, the adults in their lives followed their lead.

What I am also learning on my journey with the TRIS project and ITA, is to be an unrelenting advocate for those who cannot speak whether it is the children with trisomy 18 or trisomy 13 or their parents. I am reminded on an almost daily basis of the poor treatment of pregnant mothers and new parents who are told their child is 'incompatible with life'. My staff and I prepare data for publication highlighting data on children who are living and thriving, but that is not enough. I respond to parent questions so they can take data to their appointments with doctors, but that is not enough. I look for new ways to do outreach for the TRIS project and ITA, but that is not enough. I don't know if it will ever be enough but I won't stop trying. The children are too precious; their voices must be heard and valued.

Please visit the TRIS project homepage at http://www.coehs.siu.edu/tris/ for additional information about the project, how to enroll, download case studies and review articles and presentations. Also, please like the project's Facebook page located at https://www.facebook.com/TRIS.Trisomy.project

SOFT Italia by Daniela Fitzgibbon ITALY

Soft Italia was founded by families with children with trisomy 13 and trisomy 18, puts parents in contact with one another, and offers support during prenatal diagnosis during the life of the child and after. Soft Italia has an annual general meeting every year, a fantastic opportunity for families to share their life experience. The aim is to encourage support from any hospitals in each region of Italy, and despite the very hard journey, families are always there to support one another.

Daniela Fitzgibbon is one of many members of Soft Italia who gives support and will continue to share information available from all the SOFT organizations worldwide, helping with the translation of booklets and medical documents to make them available for families in Italy. Daniela is a mum of three beautiful girls, and one of them lived with trisomy 18.

Genetic Counseling: Changing Perception of Trisomy 13 and 18

By Hana Sroka Genetic Counselor

Hana Sroka

Hana Sroka *is from Toronto Canada. In this piece, she shares her experience and knowledge about the impact trisomy support groups have had on providing hope to newly diagnosed parents.*

Why do I do what I do? That's a question I get asked many times. I actually love my job. It allows me the opportunity to meet some incredible people. I am privileged to intimately witness their vulnerabilities and resilience. I am reminded daily of what it is to be human….and share compassion.

As you can imagine, I am often meeting individuals at a particularly difficult and vulnerable time in their lives. I am not afraid to cry with them and I am also known to celebrate their victories alongside them. I am that extra person to lean on when they feel others may not understand what they are going through. If they need further support or express interest in talking to parents who have been there, I help them find that too. I look to them for direction, not the other way around.

I am so grateful for parent organizations as they provide easy access to information about not only what the condition is, the medical facts, but what is typically more important for parents- information on what it is truly like to raise a child with a specific genetic difference. Obviously each condition is variable and no one can predict exactly which challenges and which triumphs any given child will face, but such information is vital as it provides context. Sure, some health care providers will argue that such information is biased, but it is also fair to say, all information is biased, we just need to be open and transparent of those biases. All genetic conditions are variable, with some children more mildly affected than others and ultimately each child is unique, just like children without any known genetic alteration.

When someone shares their decision to continue a pregnancy with a life limiting or other condition, I suggest they prepare themselves with knowledge regarding the specifics of the condition and demonstrate to their health care providers that they truly understand what they are facing, and I serve as a resource and advocate. Unfortunately, some health care providers have outdated information or will often assume a parent is in denial or does not adequately understand their own situation. I believe that parents will find more support within their health care team if they are armed with the information and thus can 'speak the language' and demonstrate what the quality of their child's life means to them. Only then can some see beyond the label of the diagnosis and begin to understand. And once they have, they will become more adept to seeing quality of life through the lenses of the families they serve.

Having worked as a genetic counselor in prenatal genetics for almost 15 years now, I have witnessed a change in attitude towards pregnancies with trisomy 18 and trisomy 13. At a recent genetic counseling conference, a study was presented with data documenting what I suspected, that today more and more couples are continuing pregnancies with various chromosome differences diagnosed during pregnancy, including trisomy 18 and trisomy 13.

I believe this change is not because health care providers in general have become more optimistic about outcomes (as much as I wish that were true). It is because parents have opened their eyes by opening your hearts, sharing their experience, their stories and their children. These parents have challenged preconceived ideas of what quality of life means, and spread much needed hope.

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http://www.internationaltrisomyalliance.com/ita-advisors.html

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Incompatible with Life?

First published by ICPCN 12-8-2014. This article has been reproduced with the kind permission of Lanise Shortell, RN CHPPN Paediatric Clinical Care Nurse Specialist – Atlanta.


Lanise Shortell

Introduction: *Our team cares for many trisomy families. I am burdened by the brokenness we witness after our families speak to medical experts. We are called in to consult with families when a life limiting diagnosis is made. Some may think we are providing false hope to the families we meet. I often ask myself how hope can be false? I am available for any questions and offer support from a hospice perspective. Kindest Regards Lanise*

Incompatible with life. These three words have the power to bring life to a screeching halt for new parents.


Nora full trisomy 13

The terms *incompatible with life* can fast-forward a family of a seriously ill child into a future they cannot imagine. Hearing the words *incompatible with life* can quickly deplete hope, cause feelings of isolation, and lead a family to despair. Regrettably, these three words remain in common use today.

One would think these terms are outdated. One would hope that with international efforts for quality of life, life would be the focus. However, these three words continue to find themselves in our current medical records, in our professional conferences, and within our intimate family meetings discussing treatment options. The unfortunate reality is that we care for children who will die. However, we all witness children far exceed their initial life expectancies. We realize life for these children can be a bit more complicated and will require more specialized support. Thankfully, we are learning that there is a large spectrum for life limiting illnesses. In spite of established challenges, the collaborative efforts of care team's focus on the importance of children living a quality life … not being incompatible with it.

Avoiding the label of *incompatible with life* and introducing a family to a *life limiting illness* can redirect a conversation and will highlight the true meaning of individualized, dignified, and comfort care. Life can have quality, no matter how brief. Intentional use of the terms *life limiting illness* provides space to devote our efforts to the quality of the life with illness, not dismiss it. I am honored to share photos of brave children who are proving to be *compatible with life*, despite having significant life limiting illnesses. Logan and Nora represent others around our world; they represent life. Both of these children were labeled *incompatible with life* within the last two years. These terms were used to label Logan and Nora, and these terms have been proven false. As much as we would like to think we have, we have not transitioned these words out of our language.

Medical professionals claim to be in support of this terminology turnaround. Why, then, are these terms still in use? Why haven't we made the total terminology shift from 'incompatible with life' to life limited illness? Unfortunately, I do not have these answers.

I do, however, hear the frustrations of parents and families as they question the validity of medical professionals that speak these words. I do hear and see the confusion of families as they are forced to move past being told their child is *incompatible with life* to parenting a medically fragile child, often for years.

We have a duty to eliminate this short-sighted terminology. We have a duty to prepare families for what the future may hold. Continued use of incompatible with life is irresponsible and, ultimately, can be harmful.

The time is now for us to stop using outdated, black-or-white terminology that funnels us into black-or-white thinking. This binary thinking results in only two labels for children: life or death. Instead, we need to embrace the in-between.

Because this outdated terminology is still in use, we have much work to do. Hospice and palliative care professionals have put much effort in to pioneer this change. We have tried. However, we have failed.

To complete this task, medical professionals, parents, friends, and family members must consistently advocate for the shift in terminology and, hence a change in thinking, to occur. Truth is our duty.


Logan

Review of ITA Publications by Renate Tønnessen NORWAY

First of all thank you to ITA for the amazing job they have done for parents who have a need for information about trisomy 13 and trisomy 18.

Our world changed when our now 16 months old son was diagnosed with trisomy 18 prenatally, and we searched for information about his condition. The doctors doomed him. I was fully aware of the horrible statistics regarding trisomy 13 and trisomy 18. I knew all about the malformations these babies were born with. It was impossible to miss, nearly all my searches on the Internet were glowing with grim facts about trisomy 13 and trisomy 18. Abortion was not an option for us. It was up to our baby so we continued our pregnancy, and we wanted to give him the best chance as possible.

But how could we do that? What should we prepare for? What should we ask the doctors? What about the possibilities, outcomes and so on? How could I be sure that my baby was in best hands from before his birth and for rest of his life? How could I ask for invasive care for my baby - without being neglectful of his well being?

What about the siblings, how could I tell them about Jonas condition? How could I prepare them for all the possibilities.

It was really frustrating searching for information I could use. Finally I found information I needed.

I really appreciated the booklet 'A Family Dictionary of Medical terms'. It was so much easier to understand what the doctors were talking about. My two oldest children loved the booklets 'Trisomy 13 and Trisomy 18 for Younger Children', and there is a booklet for older children as well explaining in more detail. Also 'A Cherished Pregnancy' and 'Preparing for Your Baby´s Arrival' are very informative. Fifteen months later I still use the booklets to inform the professionals and other caregivers about trisomy 13 and trisomy 18. We have had issues with the doctors, and we have to fight for our son's right for invasive care, but it has been worth every penny of our worries.

These booklets have been a lifesaver for us. The professionals could not reject them, since professionals with an objective view wrote them, and the booklets have up-to-date facts. The booklets are very useful for professionals wanting to know more about these conditions, but most important, parents on an emotional rollercoaster can read and understand more about trisomy 13 and trisomy 18, and make decisions that they feel are right for them, whether it is comfort care or invasive care. They are given a voice and a foundation they can use to obtain the best care for their child when they head towards some of the hills they (almost for sure) will meet during their contact with the health care service.

Please keep up the good work, and continue to give our children a voice. They deserve it. Thank you so much again for all help and everything you have done for us and other families.

Support Groups

http://www.internationaltrisomyalliance.com/trisomy-support-groups

The ITA website lists trisomy support groups and organizations in America, Australia, France, Germany, Ireland, Italy, Japan, New Zealand, Norway, Romania, United Kingdom, and Facebook support groups around the world. Please contact us via the website if you belong to or know of an organization that provides support to families affected by trisomy 13 and 18, and you would like your group added to the ITA Trisomy Support Groups or Links pages.

România by Raluca Manea Parent Advisor ITA ROMANIA

Raluca and family

My daughter Ecaterina, also known as Little Strawberry Cati, was born in April 2011 with trisomy 13. From the very beginning we suspected it was a case of full trisomy 13 as she showed 80% of symptoms, and this was confirmed by blood tests. However, her heart, brain and internal organs were not severely affected, and she only needed oxygen in the first weeks of life. This makes Cati a natural survivor. Had it not been so, I don't think she would have stood a chance at life as she was born in a medium sized Romanian hospital, where only one doctor in the NICU was aware of her condition.

Cati was soon moved to an even smaller hospital in our hometown, with the label ‘incompatible with life’ attached to her. It was a friendly nurse that told me, "Yes, she might die, but she is not dying now". That gave us hope and we took her home. Two months later Cati was still alive and gaining weight.

We started medical investigations: had her eyes and ears checked, later her heart and kidneys. Although Cati has cataracts and a lot of other eye problems, an ARSA and some minor heart issues, all the tests came back better than we expected.

Cati was also gifted with a sweet spirit and a charisma that goes beyond our human nature so most of the doctors we met loved her and tried to help her as much as the Romanian health system allowed. I should say this is not the case with every trisomy child in Romania. Teodora Dragomir who has full trisomy18 was denied heart surgery both in Romania and Germany. The state refused to support her surgery, so the parents had to raise funds and find a hospital willing to perform this surgery far way in Austria. Teodora is now a happy and thriving girl of four and a half years old.

Since Cati was born, I benefitted so much from the support of the international trisomy community that I wanted to do a similar thing for the Romanian parents. Thus, I began a search for rare trisomy children in our country, or Romanian children living abroad. We made great friends with Lizuca (partial trisomy 13, four years old) and Natalia (partial trisomy 13, thirteen years old) and their families, and Cati was able to celebrate her third birthday party with them.

We also got to know Teodora (the little four year old with trisomy18 mentioned above). We met Phillp, trisomy 9 mosaic, seven years old, on-lin. He was of Romanian origin living in the USA, and Rares, a little boy with partial trisomy 9 who is doing very well at two and a half years old.

We have a small Facebook group and try to help each other with support, information and useful advice.

Our goal is for Cati (photographs below) to bring hope and inspiration in the trisomy community all over the World, and the special needs community in Romania.

Photographs of Ecaterina (Cati) below.

Exciting Changes in Norway by Siri Fuglem Berg M.D. Ph.D

A founding member International Trisomy Alliance

In Norway, politicians have received letters from trisomy parents complaining about the lack of medical care for their babies. There have been articles in newspapers and news reports on television of trisomy children who were denied life saving interventions. As a result, politicians from various political parties have directed questions about treatment of trisomy babies to our Minister of Health. In his replies, focus is on palliative care, on relieving suffering as the primary goal of care.

Felicia, 6 months after cardiac surgery, one year old, trisomy 18

The Norwegian Directory of Health points out, through him, 'The importance of acknowledging the fact that most of these children die early due to respiratory or circulatory failure'. They have been ordered to develop guidelines for palliative care for children in Norway. The survival statistics that the Directory of Health point out in the Minister of Health´s response letters, are based on population studies where the norm is not to offer advanced medical interventions to babies with trisomy 18 or trisomy 13, resulting in 5-10% one-year survival.

Now, where else do we discuss survival statistics without treatment?

Parents of children with trisomy were not too happy about the response. They know that recent medical literature shows increased survival rates when intensive care and cardiac surgery is an option. They seemed disgusted by the focus on palliative care. As one mother said: "All our children are ever offered is palliative care. We want focus on proper medical, including advanced care and treatment, according to their needs, not refused on account of the trisomy label."

Further communication ensued between various politicians from a broad spectrum of parties – from the farthest left to the farthest right – and parents of a child with trisomy. As a result they decided to hold a mini-seminar on trisomy 18 and trisomy 13 in Parliament. The seminar was held on October 14th and Professor Ola Didrik Saugstad, a renowned neonatologist, was the main speaker at the event. He summed up the recent literature, stating that trisomy should no longer be called lethal, that attitudes need to change, that the lethal language has to change, that health care workers need to admit that they have been mistaken about the prognosis with these children, and that active care would be as appropriate as palliative care.

Most importantly he apologized to children with trisomy and their parents, both on behalf of himself and on behalf of the Norwegian health care system. As a committee member of the International Liaison Committee on Resuscitation (ILCOR), he is ashamed that he did not question the guidelines that have recommended for over a decade that newborn resuscitation should be withheld for children with trisomy 13 and similar conditions.

He said that the Norwegian health care system treats children with trisomy the way they treated children with Down syndrome 40 years ago, and that most colleagues probably share the view he had 10 years ago – that resuscitation of babies with trisomy is unethical. He stated that guidelines listing diagnosis where life-saving interventions should be withheld, should be removed completely, and that each patient should be treated individually, with focus on parental preferences with regards to level of treatment.

Emeli, 15 years, trisomy 18

Ivar Stokkereit, a legal expert on children´s rights, also an invited speaker to the seminar, stated the importance of not making guidelines restricting the level of care for specific diagnosis. He said, not only was it unethical, it was illegal. The Department of Health´s speaker, Dr. Jens Grogaard, stated the importance of palliative care, and that one should respect the parents of children with trisomy. On questions from parents at the seminar on why babies with trisomy 18 and trisomy 13 should be included in a palliative care program from the day the diagnosis was made, his response was that, "trisomy parents misunderstand the meaning of palliative care".

Three more physicians told of their experience with children with trisomy who lived for a while. They had all changed in their view on the diagnosis, admitting that intensive care and cardiac surgery may be appropriate for some babies with trisomy. We were told that so far one baby with trisomy 18, Felicia, has received corrective cardiac surgery in Norway.

Continued on next page

Exciting Changes in Norway continued from previous page

This girl is now eighteen months old. She is doing very well and would have passed away without the surgery. Three parent speakers, including me, spoke last. The two other parents told of their living children with trisomy 18. Emeli is 15 years old and Frida is one and a half years. They are happy and content, easy to please, and spreading both joy and worry in their families.
The entire seminar showed a changing attitude amongst the physician speakers. It resulted in newspaper headlines with the apology from Saugstad. I am thrilled, but I worry that most colleagues don´t share his view. We still have a huge job ahead of us in informing health care workers and changing their attitudes, in making them realize that life with trisomy 18 or trisomy 13 actually is worth living. And that parents, in the grey zones between life and death where medical outcome is unpredictable, just may be better suited to make balanced decisions for their baby.

Frida, 18 months, trisomy 18

Newspaper articles in Norway that appeared after the seminar

SOFT UK Charity 1002918 (1990)

by Chris Rose Co founder and Trustee

As a membership organisation, families can share their experiences through the SOFT UK website, member's magazine and regular family days and conferences. The support is offered to all members of the family, including siblings of the affected child, for as long as required. Over more than 20 years, SOFT UK has built networks with many professionals, including health professionals, family support workers and hospice staff. Our information is evidence based, prepared in collaboration with our professional advisors, and is always given to compliment the advice of clinicians who work with individual families. In line with our charitable mission, we do not offer medical advice.

The main aim of the charity is to provide support for families with a child who has a diagnosis of Patau syndrome (trisomy 13) or Edwards syndrome (trisomy 18) and their related disorders of; partial trisomy, mosaicism and deletions. We run a SOFT UK website which enables families to access dedicated volunteer befrienders via an Enquires function. (SOFT UK has no manned office). Professionals can refer families or families can self-refer. Our trained volunteers offer a supportive, listening ear and evidence based information to support families during a challenging time. Newsletters are sent out twice a year. All support is provided free of charge. Families can also obtain support and information by phoning the 24hr Helpline 0121 3513122 that will refer them on to a trained volunteer or suggest they access the SOFT UK Website. Volunteers trained by ARC and CRUSE handle prenatal queries.

SOFT UK supports families prenatally, those caring for a baby or child and those who are bereaved. Our volunteers do not express their personal opinions regarding continuing or terminating a pregnancy. Information is given so that they can make an informed choice that is right for their situation. SOFT UK has funded and developed, and is now delivering a bespoke training package to support volunteers. The training package can be delivered at development days and via distance learning. Written information is provided via booklets, available in print or electronic copy; and medical professionals update them regularly.

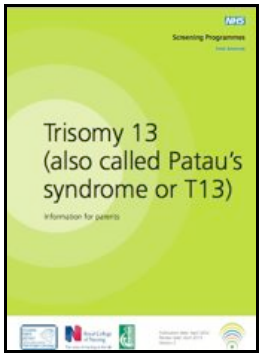
SOFT UK is presently sponsoring research into understanding the impact upon siblings - *The impact of genetic life limiting and life-threatening conditions on non-affected brothers and sisters.* SOFT UK is making a financial contribution to the first phase of the research and a number of SOFT families will contribute to the fieldwork data collection. The outcome of this work will enable SOFT to plan to better support siblings.

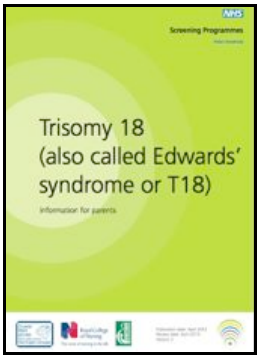
And in conclusion

I would like to thank everyone who found the time in their very busy schedules to write the articles that have made this a hopeful yet thought-provoking issue, and an essential read for families and professionals. We appreciated Hana Sroka's article in which she wrote, 'All information is biased, we just need to be open and transparent of those biases'. Yet some information that is circulated is not just biased, it is factually incorrect.

Before I retired from the SOFT UK Board in December 2011, our Medical Advisor (a geneticist) wrote to the UK National Health Service Fetal Anomaly Screening Programme about printed leaflets that were being distributed to British women during pregnancy, leaflets that could be downloaded from the internet by the public.

They stated:
Complete trisomy 13 and 18 are fatal.
Most babies with complete trisomy 13 and 18 die before they are born or shortly after birth.
Babies with partial or mosaic trisomy 13 and 18 can live beyond a year, but this is rare.





It is tragic to consider the number of UK families whose 'informed choice' was influenced by the misinformation in these leaflets.

Major population studies including a recent study by highly respected British researcher Professor Joan Morris reported a survival rate as high as 10% and 11% for full term babies with trisomy 18 and trisomy 13 respectively, with almost half of these babies surviving over one month. The survival rate for children with mosaic conditions was found to be over 70% in the British study.

The risk of trisomy is now being identified as early as 10 weeks gestation and parents need accurate information on which to base the critical decisions that are expected of them by the medical profession. These factually incorrect leaflets blatantly contradicted the UK National Health Service principle of ensuring parents would be able to make informed choices before and after antenatal screening and testing. However, the Programme ignored the SOFT UK genetic advice and the leaflets remained unaltered for several years and throughout 2014.

As Renate Tønnessen says, 'parents are on an emotional rollercoaster,' and at ITA we want parents 'in the grey zones between life and death where medical outcome is unpredictable' (described by co founder Dr Fuglem Berg) to have the facts before being asked to make important decisions and plans.

We understand the vulnerability of parents, and strive to ensure that those who seek resources will find information that is both accurate and unbiased on our website if and when they need it. This is the right of parents and they deserve no less.

SOFT UK has informed us that the leaflets are being revised and we will update readers in a future issue.

Best wishes Jenny Robbins

A Happy New Year

from everybody at International Trisomy Alliance