



International Trisomy 13/18 Alliance

Newsletter ISSUE 3 2016

www.internationaltrisomyalliance.com

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Welcome to our 3rd International Trisomy Alliance (ITA) e newsletter

In this edition our Chief Medical Advisor, Dr. John Carey (MD), has looked at the evidence that intervention in trisomy 13/18 infants, including cardiac surgery, improves outcome. Dr. Debbie Bruns (Ph.D., ITA co-founder) also reported similar results with data from the Tracking Rare Incidence Syndromes (TRIS) project. She and her staff continue to recruit new project participants and respond to parent queries. Barb Farlow (ITA co-founder) represented ITA in Geneva Switzerland in March 2015 to end the phrase 'Incompatible with Life' for prenatal diagnoses that include trisomy 13 and trisomy 18. ITA is co-hosting a perinatal conference at the end of January in Dublin Ireland, Complex Prenatal Diagnoses: A Holistic Approach to Management and Treatment that Dr. Marty McCaffrey, medical advisor to ITA, and Barb will be speaking. Barb and Dr. Annie Janvier (MD) also presented their research at the Canadian Cardiovascular Congress in October 2014. As a member of Patients for Patient Safety Canada, Barb was the keynote speaker, sharing the story of her daughter and her research on the experience of parents who have a baby with trisomy 13 or trisomy 18 at the leader's conference of the British Columbia Health Service Authority and at the Ontario Practical Pediatrics Day for pediatricians. Dr. Siri Fuglem Berg (ITA co-founder) continued to support families in Norway and shares some positive changes taking place in Denmark.

Professor Joan Morris the former Director of the National Down Syndrome Cytogenetic Register (NDSCR) has written about the value of national databases. The NDSCR has collected information on trisomy 13 and trisomy 18 (Edwards and Patau syndrome) since 2004, and collects all cytogenetic or DNA reports of trisomies 21, 18 and 13 and their cytogenetic variants occurring in England and Wales.

Sara Simmons Chappell, Kristen Cook and Christopher Gomez and his wife Helen Roper share their experience of partnering with medical professionals to obtain the best care after a diagnosis of trisomy 13 or trisomy 18. In addition, Alison Pearson who wrote '*Never say never about my child*' in the prestigious British Medical Journal, has written about her daughter Isabel. Hospice provider and pediatric nurse Lanise shines a light into treatment options and says that babies with a rare trisomy condition do have options to enhance quality of life, and sometimes, these options can extend life.

My role is to maintain the ITA website and we have added a new webpage, Trisomy 13/18 In The News, to the main menu. SOFT UK, a charity I co founded in May 1991, celebrates their silver anniversary in 2016, and I have been invited to join the 25th Weekend organizing committee. I am looking forward to working with long standing friends including Dr. John Carey who is flying over from the USA to be the principal speaker. Jenny Robbins (ITA co-founder)



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

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



Debbie Bruns

In the first ITA newsletter (December 2013), I introduced the Tracking Rare Incidence Syndromes (TRIS) project. Since then the project has continued enrolling new families, analyzing data and sharing results with interested audiences.

The past year has been a busy one with new projects and data analyses on several fronts from topics generated by participating TRIS project parents and online queries via Facebook groups, and email messages such as prevalence of specific heart defects in children with full trisomy 18 and oral health care needs. If a topic is not part of project surveys, data can be collected informally, or TRIS project staff can develop a formal data collection instrument to gather the information from project participants. This has already resulted in a publication in

2013 and one of the manuscripts in review listed below (oral health needs). The TRIS project had two new publications in 2015 and two additional manuscripts under review:

Bruns, D. A., Martinez, A. & Campbell, E. (in review). Oral health needs in individuals with trisomy 18 and trisomy 13: Implications for professionals. Special Care in Dentistry.

Bruns, D. A. & Martinez, A. (in review). A review of cardiac conditions and surgical interventions in 84 cases with full trisomy 18. American Journal of Medical Genetics Part A.

Bruns, D. A. (2015). Developmental status of 22 cases with trisomy 18 and eight cases with trisomy 13: Implications and recommendations. American Journal of Medical Genetics Part A, 167(8), 1807-1815. DOI: 10.1002/ajmg.a.37102

Bruns, D. A. & Campbell, E. (2015). Twenty-five additional cases of trisomy 9 mosaic: Birth information, medical conditions, and developmental status. American Journal of Medical Genetics Part A, 167(5), 997-1007. DOI: 10.1002/ajmg.a.36977

New directions for data analysis are association of prenatal diagnosis with gestational age, birth weight and post-birth interventions in children with trisomy 18, prevalence of apnea (central and obstructive) and treatment and incidence of cardiac defects in infants living 60 days or less with trisomy 13 or trisomy 18.

I have also served as content expert on a Dissertation Committee. The resulting paper, Medical Interventions-Quality of Life and Survival in Months-Cognitive Development in Trisomy 18, utilized TRIS project data to determine longevity as related to aggressive medical interventions (n=83). Average survival age was found to be 69.78 months with 74.2% living at time of this study with one or more aggressive medical interventions such as cardiac surgery.

Looking ahead to 2016 offers more opportunities to share TRIS project findings with plans underway for greater outreach to international groups. TRIS project also continues to work with the Support Organization for Trisomy 13, 18 and related disorders (SOFT USA).

Finally, the TRIS project's website moved over the summer. The homepage is now located at <http://tris.siu.edu> - you will find links to how to enroll, download case studies to share with family members and medical professionals and review a list of project articles and presentations. The Facebook page remains at

<https://www.facebook.com/TRIS.Trisomy.project> with just over 1,100 likes.

The value of national databases

Professor Joan Morris Former Director NDSCR



Professor Joan Morris

Up until 2014 the UK National Health Service Fetal Anomaly Screening Programme printed leaflets in which 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.'

These leaflets were distributed to British women during pregnancy and could be downloaded from the Internet by the public.

Trisomy 13 and trisomy 18 are rare anomalies, with only 3 per 10,000 births having trisomy 13 and 7 per 10,000 having trisomy 18. In England and Wales in 2012, 229 trisomy 13 diagnoses and 526 trisomy 18

diagnoses were made [1], surely enough to quickly confirm or deny the statements above.

However the situation is not so simple. Both trisomy 13 and trisomy 18, even though up until recently they were not being formerly screened for, are often detected during prenatal screening for Down syndrome that is offered to all pregnant women in the UK. In 2012 93% of trisomy 13 and 88% of trisomy 18 diagnoses were made prenatally. A large proportion of women, on receipt of a prenatal diagnosis, decide to terminate the pregnancy. In addition, for both syndromes a proportion of fetuses will not survive to term. As a result in 2012, for trisomy 13 there were only 7 fetal deaths reported after 24 weeks gestation and 21 live births and for trisomy 18 only 27 and 66 respectively.[1] Such small numbers makes establishing the true facts much harder.

However the availability of a national database for 10 years, the National Down Syndrome Cytogenetic Register (NDSCR), has allowed valid research to establish the true estimates of the proportions of babies that will die before they are born or shortly after. The NDSCR collected information from all cytogenetic laboratories on all diagnosis of trisomy 13 and trisomy 18 made in England and Wales from 2004 to 2013. [1]The information included the gestational age at which the diagnosis was made and the gestational age at which the pregnancy resulted in a live birth, a fetal loss or a termination. This allowed researchers in 2008 to estimate what proportion of babies will survive to birth after a diagnosis at 18 weeks gestation [58% for trisomy 13 and 35% for trisomy 18].[2]

The second question about what proportion of live births will live for, say, three months is harder. Live births in the NDSCR were identified by the NHS Information Centre to obtain their date of death. This was done in 2013 for the all live births from 2004 to 2011 and resulted in estimates that 20% of children with complete trisomy 13 or trisomy 18 will survive for at least three months and around 10% will survive to their first birthday.[3] Over 70% of children with mosaic trisomy 13 or mosaic trisomy 18 lived beyond 1 year. [3]

There is still one vital step lacking. That is the translation of such information from scientific peer reviewed journals into information provided to pregnant women. The National Health Service Fetal Anomaly Screening Programme has delayed providing this information to women.

In 2015 the NDSCR was formerly moved from Queen Mary University of London into Public Health England. It is hoped that this unique resource will continue to be used for such valuable research despite the fact that all the original researchers are no longer involved in its running.

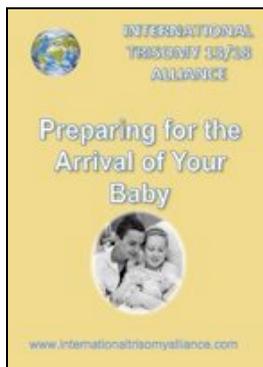
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Free booklets for families

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

The e booklets published by International Trisomy Alliance are for families and children who wish to know more about trisomy 13 and trisomy 18. They are free to download from www.internationaltrisomyalliance.com and links to other publications include the SOFT USA 'Care of the Infant and Child with Trisomy 13 or Trisomy 18.



Preparing for the Arrival of Your Baby



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

Much Ado About Something: Asking a Relevant and Timely Question

John C. Carey, MD, MPH
Professor
Department of Pediatrics
University of Utah

With sincere apologies to William Shakespeare for modifying a well-respected title, I would suggest that the “Something” is the important and now relevant question: What is the evidence that medical interventions improve outcomes in the care of infants with the trisomy 13 and trisomy 18 syndromes? As I have suggested in the past, there has been an emerging dialogue in the medical genetics, pediatrics, and bioethics literature in the last 6-7 years about this topic ¹.

Prior to the 2006, I would say that in most newborn intensive care nurseries in North America and Europe, there was the conventional dogma that intervention in the care of infants with the syndromes did **not** make a difference. Then the seminal paper by Dr. Tomoki Kosho and colleagues at Nagano Children’s Hospital in Japan arrived on the scene ².

This study reported the outcome after major interventions and respiratory care (not including cardiac surgery) in 24 newborns with trisomy 18; six of the 24 survived the 1st year of life showing the highest median survival of any previous series. While the number of children was relatively small, this work suggested that intervention did improve outcome in infants with trisomy 18. Given the similarities in early medical problems between the 2 syndromes, it was logical to infer similar outcome for trisomy 13 (although untested). Let us discuss this important question more.

There are 3 lines of evidence to suggest that intervention makes a difference and improves outcome in young children with both syndromes. First, there is the work of Dr Kosho (mentioned above) suggesting that full intervention, including ventilation and tracheostomies, improves survival compared to published population studies and other hospital series. Other recent evidence that confirms this idea includes 1. the recognition that some central apnea in newborns with trisomy 18 is, in fact, a seizure disorder that is potentially treatable, and 2. the observation that while central apnea is an important cause of serious illness in infants with trisomy 13 and 18, there are likely many children who have upper airway difficulties, which may be more amenable to treatment than central apnea ¹.

The second line of evidence comes from the recent studies reporting outcomes in children who have had cardiac surgery during the 1st year of life. Presently, there are over 160 children reported in these 8 series from North America and Japan who have had cardiac surgery with over 85% leaving the hospital. (This number does not include



John Carey

the greater than 100 children reported in the SOFT Surgery Database created by Ann Barnes that we are currently analyzing in detail.) While this figure represents decreased survival compared to children **without** trisomy 13 and 18, it does clearly demonstrate that children with the 2 syndromes do better than was predicted in earlier decades when the prevailing attitude was "we don't operate on these children." Some of these studies suggest improved survival in the 1st year of life in those who have had heart surgery compared to population study figures.

The last line of evidence is that recent figures in some population studies suggest improved survival over time. Most notably, this appears to be the case in the United Kingdom. The initial study by Young and colleagues (1986) showed no survivor of 1 year out of 21 infants, while 2 recent investigations (including Wu et al. where Dr. Joan Morris is an author), show 6% and 8% survival, respectively.

In addition, a recent paper currently in press in the *American Journal of Medical Genetics* from Meyer and associates, reports mortality data using population-based studies in 9 states in the U.S.; the number of children in this study far exceeded any previous work: 693 children with trisomy 13 and 1,113 with trisomy 18. One-year survival was higher than any previous studies, but, more importantly, 5-year survival was 9.7% in trisomy 13 and 12.3% in trisomy 18. This is the first study (other than one from the 1960s) that reported on 5-year survival. The authors suggest that there may be 'improved survival following more aggressive medical intervention for these children.'

While more investigation needs to be done addressing this relevant and timely question, the emerging evidence supports the idea that medical intervention does make a difference in outcome in the trisomy 13 and 18 syndromes.

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About hospice or palliative care

Different countries and medical systems have various ways of supporting families who care for a baby or child with trisomy 13 or trisomy 18. Hospice or palliative care services may provide pain relief, family support and respite care in addition to end-of-life care.

This care can be beneficial if providers are cognizant that some children with trisomy 13 or trisomy 18 do survive, and the service itself does not preclude the child from having beneficial interventions, like surgery.

International Trisomy Alliance

An ideal home care service from a mother's perspective

Sara Simmons Chappell



Nora trisomy 13

“Hospice. You’re being sent home on hospice. You will be discharged tomorrow with your ten day-old baby girl on hospice support until she passes. This is the best option for your situation.”

By the time those words were spoken, we were numb. We had already been told our firstborn child would soon die. We had been strong-armed into a DNR, and told that portions of her condition would never be fixable and that they would lead to imminent death. We were shuffled into a hurried meeting with a geneticist who promptly inquired if somehow Jerry and I were related? As if she could explain

Lanise came to our home almost every day. In the beginning her tone was very soft, almost hushed. I am fairly certain she could sense my hostility. I didn’t want her to be here checking my child for signs of her demise. Lanise did her best to make our conversations as normal as possible and to end each visit on a positive note.

away this unfortunate circumstance with a ‘distant cousin’ theory. We were told that because our daughter had a relatively normal appearance, the whole ‘situation’ (i.e. imminent death) would be far more difficult. According to her, the only trisomy 13 patient she had seen outside of the NICU was a vegetable. She looked down her nose... ‘Now you don’t want that kind of life, right?’ She handed us the Wikipedia page for trisomy 13 and sent us on our way.

The neonatology team was no better. They insisted that nothing about Nora’s condition was fixable. We were told that a hospice was our only choice. We went home on day 10 with our Nora. Our hospice nurse was waiting in the driveway when we got home. I hated hospice. I wanted normal. I wanted to be showered with the normality of bringing a baby home. Instead of arriving home to casseroles and balloons on the mailbox, we were met with oxygen tanks, morphine bottles, our nurse, Lanise, and our social worker, Stefanie.

I hated hospice. I hated Lanise and Stefanie. The NICU had told us that our time was short. We should go home and ‘enjoy’ our moments at home. Anyone who has ever told another person to go home and ‘enjoy’ a loved one who is about to pass into heaven, has clearly never done so themselves. We obliged. We accepted hospice and waited for Nora to pass. But Nora had other plans.

Over the course of our first several months, Lanise slowly became like part of our family. She transitioned from someone we saw as associated with Nora’s passing, to our biggest cheerleader. She leapt for joy the first time I mentioned the possibility of repairing Nora’s heart condition. We started to really open up about what

Nora's LIFE could and would look like if we treated the things that were causing her trouble.

HOSPICE has some pretty strong connotations in the lexicon of healthcare in the United States. It brings up instant images of elderly cancer-riddled patients, hooked up to oxygen unable to speak, eat or move. So what happens when those hospice patients are newborns? What happens when those needing hospice support were thought to have their whole lives ahead of them? What happens when the hospice social worker is helping couples plan for their child's funeral?

It is a whole different playing field and one that requires a very special skill set and approach. It requires an approach that allows hope; hope for today and, more importantly, hope for tomorrow. Despite all of the odds, parents need to be allowed to hope. Lanise and her team encouraged us to hope. She encouraged us to pursue full treatment for Nora, while still in their care.

Around Christmas of 2014, Lanise called to tell me that they would soon have to

drop Nora from their service. She had been unable to adequately document a 'decline' in Nora for many months. Our daughter was being lovingly kicked out of the pediatric hospice program she has been enrolled in since birth.

Instead of joy, I felt panic. You see, this team had become like family to us. They had laughed, cried, prayed, and celebrated Nora's accomplishments with us. They had watched Nora's first smiles, had taken part in her first belly laughs, and cheered her on during her many therapy sessions. They knew us and our situation better than virtually anyone.

They were our cheerleaders, and our shoulders to cry on for two years. The service that I started out resenting had become our lifeline and a constant source of positivity in our lives. Lanise and Stefanie threw Nora an amazing hospice graduation party. We all cried true tears of joy that this child who was never "supposed" to live, was thriving. We cried because Nora was a success that they rarely get to see.

Our lives will forever be connected and we will be forever blessed.



Nora with her baby brother

**See page 15 to read the article by Lanise Shortell
Pediatric Clinical Care Nurse Specialist who cared for Nora**

Partnering with professionals for optimal care

Kristen Cook

When I was 15 weeks pregnant, the results from an NIPT showed Hannah had a 95% likelihood of having trisomy 18. At the beginning of our journey, we were told that Hannah was incompatible with life and would be vegetative. Knowing this, our goal was for Hannah to live a good life, so we focused on simply keeping her comfortable and in our arms for the duration of her life.

As time went on we learned more about the positive outcomes for some children with rare trisomies. Since Hannah appeared to have no life threatening physical anomalies, our goals and focus shifted to not only giving Hannah her best life, but also her best *chance* at life.

We assembled a team of medical professionals to help us, including my obstetrician (OB), my maternal fetal medicine (MFM) specialist, a palliative care neonatologist and the neonatologist that would be present at her birth. My OB's only experience with rare trisomies was with stillbirths. In our initial discussions with her she recommended that I go into labor naturally, deliver vaginally and she would monitor Hannah's heartbeat intermittently so we'd know if she died during labor.

On my request for a recommendation of fetal Doppler brand, she handed me the one she was using and gave me the parameters that Hannah's heart rate should remain in. Meanwhile we continued to visit the MFM doctor for detail sonograms. In the third trimester, the umbilical cord blood flow had a higher than normal resistance.

In babies with typical chromosomes this can be a sign of imminent demise. Once the resistance gets too high, absent-end or reversed diastolic flow can occur and the baby needs to be delivered immediately. We didn't know if this meant the same thing in babies with rare trisomies. In fact, in the past most families carrying their trisomy child to term did not opt for, or had not been given the option for increased observation, so in a lot of ways we were flying by the seat of our pants. We began the tightrope walk of watching Hannah, carefully trying to figure out the right time to deliver her in order to give her best chance at life.

Based on the elevated blood flow resistance the team decided Hannah should be delivered by a C-section. At this point my OB had shifted gears to feeling confident that we would meet Hannah alive. She was happy to perform the C-section and I believe she was excited to potentially have a different end to this trisomy pregnancy. Based on Hannah's small size, and the blood flow issue we also decided to deliver her at 37 weeks. I learned that oftentimes trisomy baby's lungs aren't mature at the same gestation as 'typical' babies.



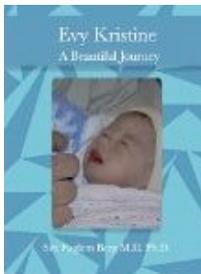
Hannah trisomy 18

After discussion with both my OB and MFM we agreed I should get antenatal steroids in the week before delivery in order to increase the production of lung surfactant, which is critical for proper lung function.

Hannah was born on March 19th by scheduled C-section. When my doctor pulled her out, she immediately started crying. A weak cry that sounded like a kitten, but a cry that meant she was breathing. She was alive. She was in the NICU for 8 days prior to discharge and lived for 9 months. While my medical team did not expect Hannah to live very long, they were all willing to hope with us and to support her, giving Hannah her best chance at life. I am currently working with two of my doctors in order to develop a blueprint that families and doctors can use to help other families with a prenatal diagnosis to achieve their unique goals for their child.

Evy Kristine A Beautiful Journey By Siri Fuglem Berg

Available Amazon USA
Amazon UK and Norwegian



Translations

Spanish, Italian, Turkish, Finnish, and Indonesian translations of children's books are now on the ITA website



BMJ Practice- What your patient is thinking:

Never say never about our child

<http://www.bmj.com/content/350/bmj.h1246>

Alison Pearson wrote movingly about her daughter Isabel for the British Medical Journal (*BMJ* 2015;350:h1246) monthly series in which patients and carers set the learning outcomes for professionals. She asked healthcare staff to reconsider the concept of offering 'false hope.'

Isabel has trisomy 18 and Alison wrote the following ITA article especially for anyone having a younger child with a recent diagnosis. She and her husband now realise how much quality of life their daughter could have been denied if the doctors had had their way!

A life less ordinary - Isabel Alison Pearson

Five years ago we experienced pretty well the worst day of our lives: our beautiful daughter Isabel was diagnosed with Edwards syndrome (trisomy 18) when she was three days old. We were given a bleak outlook delivered with complete certainty by the medical professionals: we were told to expect her to live “days, or weeks if you are lucky”. Nobody really briefed us about any kind of disability other than the fact she would not be able to feed properly, as I think they didn't see any point – after all, she wasn't going to be around long enough to worry about that.



Isabel trisomy 18 with her brother

Fast forward a few months and she was still here: we were starting to think that maybe she would be with us a little longer, but the worries hung over us like a huge black cloud. Not just the worry about whether she would live to the next milestone (oh please, just make it through Mother's Day, Easter time, birthdays, Christmas), but what our lives would be like. Would I ever work again? Surely we would never make it abroad on holiday? What oh what would we tell our son

Harry, then four? Would Isabel ever smile? How could we possibly cope with hospital admissions? And just how much or little would she be able to do?

So, five years on, how are things working out? Well, for a start Isabel is very much here and what a joy she is – I honestly think she is the happiest person I know. She has this radiant beam for all the special people in her life that makes you feel like the most important person in the world. She just exudes love in an unconditional, beautiful way. I work part time, thanks initially to two wonderful child minders, and now to the fact that Isabel is at school. Yes, really – school. Ok, it's a special school, with small groups and lots of therapy input: Isabel absolutely loves it. She also attends a mainstream pre-school one day each week, and the other children adore her.

We have been on holiday – in fact, that's downplaying our holidays significantly. We go on at least three overseas trips a year, including our annual 'once in a lifetime' trip to Disneyland Paris. Well the children love it so much, as do we, that how could we possibly only want to do it once? ! We have to be a bit more planned than previously, but not in an impossible way.

Isabel feeds completely orally, and enjoys her food although she is quite discerning and will refuse anything she doesn't like the smell or look of. Her food is of a coarse mash consistency, and we continue to work on challenging her with lumpier food. We are sure she could manage harder food, but chooses not to – she completely knows her own mind and is very strong willed, in a completely age-normal way.

Photos of Isobel trisomy 18

Developmentally she is making progress, and this is something we learned to focus on early. If one day she stands for a second, then the next day it might be two, and after a while a significant amount of time.

This year she has gone from not being able to sit unsupported to being able to sit on the floor for 15 minutes or so. She plays with toys, and reaches and grabs, especially things she is not supposed to! She communicates her wishes very clearly, and really understands cause and effect: communication milestones that we continue to help her to build on.

Isobel has clear likes and dislikes: she loves water so swimming or having a bath are favourite. She also loves music and really enjoys playing random notes on a piano. Isobel also enjoys movement, so she likes sitting in a swing, and in our newly acquired bike trailer. She loves being in the thick of the action and will protest if she thinks she is being left out. It's not all been easy. Whilst Isobel needs no ongoing nursing she is medically fragile and a respiratory infection can easily lead to a hospital admission: some of these have been terrifying and she has been really poorly. There have been various battles with 'authority', first medically and then educationally, as Isobel does not fit the rule book. But life has a whole range of challenges and we just keep going through the tough times with as much emotional support as we can rally.

Most of all Isobel is a child, not a diagnosis: a much loved daughter, sister and granddaughter who radiates joy and unconditional love. We are indeed privileged to have her in our lives.

You can read Alison's blog on: <https://ourmiracledaughter.wordpress.com>



Denmark - towards a brighter future? Dr. Siri Fuglem Berg

In Scandinavia, Denmark has been regarded as the most eugenic of the Scandinavian countries ever since they introduced a national pregnancy screening program for chromosomal abnormalities in 2005. Ever since, the number of births of babies with Down syndrome has plummeted, and this has been celebrated by the medical profession as a great success. The willingness by obstetricians and neonatologists to offer advanced medical interventions to babies with trisomy 13 or trisomy 18 has, as in Norway, been as good as absent. With the recent birth of one little boy with trisomy 18, that has changed.



Mika trisomy 18

Mika was diagnosed in his mother's womb at 12 weeks of pregnancy, full trisomy 18. His parents decided against the common pathway in Denmark, which is to terminate the pregnancy. They wanted to keep Mika for as long as possible, and they wanted a chance to meet him. Later in pregnancy, Mika was diagnosed with a VSD and an omphalocele. The doctors agreed to do the necessary surgeries if he lived to his birth. They also promised intensive care treatment if necessary, including ventilator. They promised to treat him like any other baby.

Mika was born on November 10th, in the 34th week of pregnancy, delivered with C-section after receiving antenatal steroids for treatment of fetal lung immaturity. He was intubated, and his omphalocele was operated on later.

Sadly Mika passed away from complications to the lungs when he was ten days old. His parents are preparing for his funeral as I write this. But Mika did get ten days to enjoy his parents and siblings, and his family had ten days to enjoy him. Ten days may not seem like a whole lot to most people, but when life is expected to be short, ten days is an eternity. It makes all the difference.

Having physicians who found Mika's life worth saving, doctors who did not give up on him, but respected his parents' wishes, was of the greatest importance. Mika's parents can go on living, knowing they did everything possible for their son. To many parents, that really matters.

Being denied treatment for your baby on account of the extra chromosome only adds to the pain and grief. Mika's parents did not have to endure that extra burden, and Mika had a full, albeit short, life. He did not suffer. He was loved, unconditionally. Our thoughts go to his parents who are now going through the greatest grief – the grief of losing a child.

And Denmark – well, Denmark may not be as bad as we thought when it comes to giving babies with disabilities a chance to life. Perhaps there is hope.

An ideal home care perspective from a provider experience

Lanise Shortell RN CHPPN CPLC

Pediatric Clinical Care Nurse Specialist Atlanta, GA USA

I am a pediatric hospice nurse and would like to share my experience with babies with trisomy 13 and 18 and the families that champion them. My hope is that you find encouragement and empowerment here.



Hospice providers, Lanise and Stephanie with Nora, trisomy 13

Our pediatric hospice team is often requested to speak with families when a child is diagnosed in utero or shortly after birth. We realize we are the last people that families want to meet, especially after receiving a diagnosis that many medical providers, often erroneously, consider lethal.

However, in our little corner of the world, we offer hope, we provide guidance, and we champion each baby that we are privileged to care for. In fact, our hospice has had three babies with trisomy 13/18 graduate from hospice care in the last two years. For our hospice team, we see each T13 and T18 babies as miracles just waiting to unfold.

Full disclosure for the skeptics that may be reading this — there is a full spectrum

of T13 and T18. There are many babies that are born sleeping, there are babies that pass away hours after birth, or just a few short weeks after entering our world. However, we cannot dismiss the facts that there are babies that are surviving. Nora is a beautiful toddler with trisomy 13, nearing her 3rd birthday.

A diagnosis of T13 or T18 does have challenges, but I stand up for the children we care for and proclaim that each baby deserves a chance to tell their story. Each baby has a different experience and it is imperative that medical professionals around our world honor this fact when discussing treatment options. Yes, life honoring treatment options. Babies with trisomy do have options to enhance life quality, and sometimes, these options

can lead to longer lives.

When a baby with trisomy is born and admitted to our hospice program, we make automatic referrals to a program that consists of physical and occupational therapists who go to the family home to provide muscle strengthening and comfort.

Early intervention, energy conservation, and parent education on how to physically care for a medically fragile child helps parents develop into confident caregivers. We feed our babies with trisomy just as we would feed babies without the complexities associated with trisomy 13 and trisomy 18. Nutrition is required for strength, energy, and basic metabolic needs.

Now for the skeptics, we monitor the feedings closely so as not to overwhelm often compromised cardiovascular systems. We provide supplemental oxygen for the children on our program to decrease the workload on their hearts.

Our goal is energy conservation and effective symptom management. If babies are kept nutritionally sound, comfortable, and introduced to a primary pediatrician for well-baby check ups, there is a better likelihood of life continuing.

Hospice care for children is very different from adult hospice care. In 2010 the United States enacted a federal law called concurrent care that allows families to seek aggressive care for terminally ill children while receiving expert symptom management, psychosocial support, and spiritual guidance that hospice provides. This is the best of both worlds. Parents are given the option of leaving no treatment stone unturned while caring for their children. Concurrent care does not promise a cure. There are children that have received concurrent care that have been overcome by their medical condition. However, parents that who have accessed concurrent care report they did not feel care was withdrawn or that hope was extinguished simply due to a their child's diagnosis.

I have witnessed babies with trisomy 13/18 overcome the odds, reach milestones, and enjoy a lives that medical professionals still profess is unlikely to occur. I wish there was more awareness of the lives being lived by children that were once considered incompatible with life.

The Internet is often the only source of information for parents with newly diagnosed babies. We can change the lack of information for new parents by writing articles, sharing stories of T13 and T18 babies that are enjoying life, and offer a different perspective than the outdated textbooks. We can join together, educate communities, work alongside physicians and share stories of babies with T13/18 that are not just living, but thriving. Every baby deserves a team in their corner to champion them and their families. Our hospice team is honored to fill that role for each baby in our care.



Lanise and Stephanie also celebrated the graduation of baby Brayleigh who was thirteen months old and had trisomy 18

ITA to co-host Perinatal Conference in Ireland



We are proud and excited to announce that we are co-hosting a conference entitled, Perinatal Conference 2016: Complex prenatal diagnoses, a holistic approach to management and treatment, on January 30, 2016 at the Royal College of Surgeons in Dublin, Ireland. The conference will draw international speakers including neonatologist, Dr. Martin McCaffrey who is a medical advisor to ITA and specialists from Johns Hopkins and Boston Children's Hospital all whom will discuss novel treatment for the care of conditions such as trisomy 13 and trisomy 18 that were once considered to be 'lethal'. Barb Farlow, co-founder of ITA will be presenting her research based on the experience of over 350 parents who had a baby born with trisomy 13. Conference delegates will be made aware that almost all parents reflect on their child's life as a positive event, regardless of longevity and that surviving children are happy and an enriching influence on their families. More information is available on:

<http://www.perinatalconference2016.com>

Geneva Declaration to end 'Incompatible With Life'

On March 11th, 2015, Barb Farlow joined a group of enthusiastic physicians, parents and Grégor Puppincq from the European Centre for Law and Justice at the United Nations in Geneva, Switzerland to launch the Geneva Declaration. The Declaration aims to end the phrase 'incompatible with life' as a description for diagnoses that are associated with shortened life spans. Parents of children with these diagnoses feel that the description insults and devalues their child's life. Furthermore, it can be inaccurate as some children live for much longer than predicted. Most importantly, when a child is described as having a condition that is "incompatible with life" physicians are reluctant to provide medical care, erroneously believing that medical care and interventions are always futile. The trisomy 13 and 18 community can certainly relate to this experience. [The Geneva Declaration: http://www.genevaperinatalcare.com](http://www.genevaperinatalcare.com)

