Taking It Day by Day: Melissa's Story

When I received the email in April notifying me that my patient match was a child with Trisomy 18, I thought for a second that it was a mistake. I had learned this past fall during my first semester of medical school that Trisomy 18 is lethal. *There's a mnemonic for that disease - rocker-bottom feet, overlapping digits, cardiac problems, mental retardation. Cause is usually maternal nondisjunction, mosaicism more rare. Prognosis is death before 1 year of age.*

Clearly I would be learning something new.

Through our initial emails, I could tell that Sara* was excited to share Melissa's* story and I was already grateful and excited to have this experience to intimately learn about a rare disease. I met Melissa and Sara a couple days later at a pediatrics clinic for a mix of appointments – hematology-oncology, renal, and prosthetics. I was able to meet Melissa and Sara nearly every week during my last two months of the school year for dozens of appointments and once after an in-patient hospital discharge, before I returned home for the summer. During this time, I was able to hear Melissa's story from Sara, continue reading online about Trisomy 18, and discovering the social media world of trisomies.

I knew that Trisomy 18 could affect every organ system in the body, but I quickly learned that Trisomy 18 is unique for each patient; one of the first things Sara told me was that Melissa will write her own story. Melissa's healthcare team has included a cardiologist, hematologist-oncologist, nephrologist, audiologist, home nurse, physical and occupational therapists, prosthetist, neurosurgeon, and over this summer, an orthopedist. Throughout our many conversations about Melissa's healthcare and progress, Sara's role as her primary caregiver, and how having a special needs child has affect Sara's and her family's life, I am no longer learning from a textbook about Trisomy 18 but trying to understand and navigate the complicated world that is a rare disease.

Sara's pregnancy became a tidal wave of information upon prenatally learning that her baby had Trisomy 18 – she had never heard of a trisomy before despite knowing about Trisomy 21 by its more common name, Down syndrome. She was informed of the lethal nature of the chromosomal abnormality and decided to continue with the pregnancy

**Names and details have been changed to protect patient and family privacy.*

and spend the unknown amount of time she would have with her baby. Melissa was born and eventually went home on hospice care.

However, Melissa did not become sicker. In the average family, a child's first birthday is exciting and new. For Melissa, reaching 1 month of age was truly a milestone, and reaching 1 year of age was amazing considering that 90%-95% of babies with Trisomy 18 die before their first birthday. Over these few short months having known Melissa, I have seen her triumph by transitioning off needing oxygen and transitioning to a blenderized diet in her gastrostomy tube. She has also overcome fevers, infections, cancer, and surgery thus far.

Each harrowing event is not only a stress on Melissa's body, but also a further stress on Sara and her family. I had so many questions for Sara: how does she feel about being Melissa's primary caregiver? How do you find support when your child has a rare disease? Where does a parent start after the first time they hear about Trisomy 18 in the doctor's office?

The Internet is filled with technical information, misinformation, and a lack of information when it comes to rare diseases. Additionally, social media is an abundance of stories and experiences.

Unlike cancer and Alzheimer's disease, for example, which are familiar to the public and are known intimately by millions, those with rare diseases have the added challenge of finding support. One of the first resources Sara was given was the website for SOFT, Support Organization for Trisomy (trisomy.org), but found that the forums weren't exactly what she needed.

Meanwhile, Facebook is filled with thousands of support groups and pages dedicated to diseases, caregivers, and personal experiences. Groups like these are key in providing support for parents of children with rare diseases. For example, the Trisomy 18 Foundation Facebook page states that it "sponsors and moderates this Private Group to connect parents at any stage in their personal journey that began with a diagnosis of Trisomy 18 for a much-wanted child." But because every patient with Trisomy 18 is unique, it is not enough to simply say that any Trisomy 18 group will provide adequate support. A parent of a child who is a few weeks old, medically unstable, and on hospice

care is having a much different experience than the parent of a child who is over 1 year old and medically stable, despite both children having the same syndrome in name.

The scope and significance of Trisomy 18 for Sara has extended beyond the traditional medical world of doctors, nurses, and hospitals. Sara is responsible for keeping Melissa alive beyond what we may consider normal parenting; she monitors Melissa's oxygen levels and temperature, feeds Melissa through her gastrostomy tube, and gives Melissa her medications. Currently, Sara gives Melissa 14 doses of medication each day - lorazepam, clonidine, furosemide, sildenafil, spironolactone, ranitidine, and potassium and sodium supplements.

Not only is she the primary caregiver for Melissa, but she also shares her experiences in the world of social media. Sara contacted other caregivers on Facebook soon after Melissa was born. Her experience has morphed into one where she keeps in regular contact with other caregivers who have a child with Trisomy 18 and are around the same age as Melissa.

The first time I was on Sara's Facebook community page for Melissa, the first and second posts were dedicated to two children who had passed away within the past two weeks. I was hit with sadness - the sadness of knowing a life had been lost and there were parents, siblings, and friends grieving the loss of a beloved child. But then I scrolled down to the third post, which included "Let's celebrate!" and noted the first birthday of a child with Trisomy 18. This community is strong and resilient and provides an environment in which to both grieve the loss of life and to celebrate life – a crucial outlet for emotion in the world of an unforgiving disease.

In addition to Sara's Facebook page for Melissa, Sara began to blog nearly a year ago. Her blog serves as a window into the life of a caregiver of a special needs child and is a first-hand account of the roller coaster ride of emotions that come with a Trisomy 18 diagnosis, the ever-lingering despair of future heartbreak that will arrive when she outlives Melissa, and the worry about the stress of a sick child on her marriage and other children. Blogs like Sara's give caregivers an outlet to describe their feelings, educate others, and help validate the emotions of caregivers, parents, and family members in similar positions.

As a future physician, I have learned how supportive social media can be for caregivers of a child with a rare disease. Physicians may be experts in managing patient care and treatment, but they generally do not have first-hand experience as the primary caregiver of a special needs child or a child with a rare disease. For this type of insight and information, parents new to the world of rare diseases can turn to social media – perhaps the only place where one could ask a question and receive multiple answers and perspectives within a few hours. Support for caregivers is crucial in ensuring that those they care for are taken care of.

I have reflected upon how my experience with Sara and Melissa has shaped my delivering of information to patients and caregivers as a future physician. The combination of the lethality of Trisomy 18, the desired outcome of a Trisomy 18 pregnancy for parents, and opinions of the physician make for difficult conversations. When Sara told me about her initial conversations with some of Melissa's physicians after her birth, she did not have the most ideal experience with all of her healthcare providers, some of whose suggestions leaned towards Melissa's life with Trisomy 18 being one that should not be lived due to the known severe impairments she will have as she grows. This situation poses an ethical dilemma with no "right" answer.

However, I have realized that the diagnosis of Trisomy 18 as "lethal" may sometimes be self-fulfilling in the sense that parents may elect palliative care for their baby. How would patient management change if data were more accurate in regard to how many Trisomy 18 patients were advancing beyond 1 year of age? What if the expectation were that more Trisomy 18 babies had the potential to survive if more aggressive treatment came first and was followed by hospice and palliative care when these interventions failed?

These questions are difficult to answer without accurate data. I had a hard time finding updated statistics online regarding the survival rates of Trisomy 18 in the United States, due to a lack of a national registry and different data collected by each state. Another issue is how many children with Trisomy 18 have access to excellent medical care - does this contribute to a higher survival rate? Sara is fortunate to live near a hospital capable of taking care of Melissa's many health issues. Much like how cardiovascular disease is paradoxically more prevalent because we have the medicine and

technology to keep adults alive longer, some Trisomy 18 babies with "lethal" diagnoses are becoming toddlers and toddlers are becoming young children. What can we do to support families' decision making? How can I as a healthcare provider help the patient and their caregiver make the best decisions for their specific situation when there are so many unknowns? These are questions I am still unsure of how to answer, but hope to continuing thinking about as I progress through my medical training.

Had I not met Melissa, I would have continued to be under the impression that Trisomy 18 is a diagnosis of no hope. Melissa's smile, her being able to roll onto her side, and her not being on oxygen anymore are all testaments of her strength and growth. And as much as I empathize with Sara's experiences from the start of her and Melissa's journey, I also realize that I will never be fully able to understand the uncertainty, fear, and life changes that come with a diagnosis of Trisomy 18 or any other rare disease. However, I can arm myself with other tools to enable caregivers to deliver great medical care.

These tools do not need to be new pieces of equipment or technology. One strong tool is empathy and compassion in understanding the special challenges that parents who act as caregivers face. They are experts at assembling machines, scheduling appointments, detangling tubes and wires, managing medications, and ordering supplies – they are a little bit support service, handyman, pharmacist, nurse, and secretary all in one. In many ways, their child's life is truly in their hands, unlike the average experience of a parent. Physicians should be checking in with caregivers to ensure that they are not too overwhelmed or distressed to take care of their child and should be able to offer encouragement, support, and guidance.

At the same time, physicians should also understand the excitement and happiness of parents of children with rare diseases. With Trisomy 18, reaching the first birthday is a significant milestone and in an emotionally different way than for the average healthy child who is generally expected to reach this point. Melissa's first birthday party was a huge celebration, with over a hundred people in attendance. Other causes for celebration exist, too - for Melissa, these included moving off oxygen via nasal cannula, moving to a blenderized diet, and rolling over. Each child, rare disease or not, is unique, but those

with rare diseases each have their own causes for celebration that physicians should recognize.

Outside of the doctor's office, I had wondered if Sara's home would be different than a "normal" home given Melissa's medical conditions, but found that their home was as ordinary as anyone else's. Sara graciously invited me for a delicious dinner shortly before my first year of school ended. Melissa had a play area on the floor in the family room and was held on Sara's lap while we ate dinner. Though Sara's life revolves around Melissa, her routine with Melissa was like that for any other child, though with different items like tube feedings instead of cooked meals.

As I left Sara's home, I realized how I had grown as a student and person since the beginning of my school year. I know I had thought, *why are we learning about these rare genetic diseases? They're rare so we probably won't see them anyways.* Learning about Trisomy 18 has led me to see how treatment of this rare disease is truly unique and individualized for each patient. There is no handbook to follow or predetermined medications to give. Sara takes care of Melissa day by day and cherishes the time she has with her.

In an ideal medical world, there would be a cure for Trisomy 18. But as a syndrome, treatment deals with the abnormalities caused by the extra chromosome on a case-by-case basis. Gene therapy that could correct trisomy by silencing the extra chromosomes holds hypothetical promise, but this technology and innovation is still decades in the future.

To further share Melissa's story and to educate my peers, I have shared Sara's blog and Facebook page with my classmates and other friends in healthcare fields who would benefit from learning about a rare disease and the unique aspects of Trisomy 18. In the upcoming semester, I have invited Sara to speak to a group of my classmates, particularly those interested in pediatrics. My goal is that my peers will learn about the experience of a parent and affected child dealing with one of many rare diseases in which the textbook description does not always give a complete, or even accurate, picture.

On a final note, I extend my utmost gratitude to Sara for letting me into some of the most intimate aspects of her life and Melissa's life. Her honesty, optimism, and warmth have inspired me to share Melissa's story to further educate my peers, family,

and friends on how I as a future physician, as well as the greater community of physicians, can best treat and care for those with rare and chronic conditions. Melissa has left an indelible mark on me as a person and future physician, and I look forward to keeping in touch with Sara and Melissa throughout my remaining time as a student and beyond. Each child has his or her own journey, or as Sara would put it, their own story - I have been so honored and privileged to be a small character in Melissa's.