The Strength in Small Numbers

Hannah Jacobs was going to be a doctor.[1] Twenty-three years old, the Australian-born student was in the process of finishing up her undergraduate degree in New York City. She was a diligent student with a passion for human biology, so applying to medical school seemed a logical next step. As it happened, she was also recently married and pregnant with her first child.

When Asher was born, everything changed.

Hannah remembers, immediately after Asher's birth, one of the doctors raising a red flag.

"He noticed Asher's jaw," she explains, showing me a baby photo of her son, a cherubic, light-haired infant with wide eyes. Though barely noticeable, there is a slight slackness around his mouth. The doctor thought it could be a sign of something, some strange-sounding illness Hannah had never heard of.

"I didn't believe it," Hannah says. "I wasn't ready to hear that."

Soon after bringing Asher home from the hospital, however, it became clear that something was wrong. He seemed lethargic and weak; he wasn't able to feed.

Hannah and her then-husband, Gabriel, were of Ashkenazi Jewish descent, a lineage known for carrying a higher than average risk of hereditary diseases. So before marrying, before even announcing their engagement publicly, they underwent genetic screening. The tests came back clear, showing no indication of genetic risk, and they carried on with the wedding feeling reassured. But both Hannah and her husband were in fact carriers for a rare muscle disease—so rare that it hadn't been included on the screening panel. Shortly after Asher was born, he was diagnosed with nemaline myopathy (NM), a debilitating genetic illness that disrupts muscle development.

Even these first moments of Asher's story are an example of exclusion; of existing unseen, on the outskirts. It's a theme that defines much of his experience in the world of rare disease, which, as I've learned during my time with him, seems to carry with it a form of

[1] All names have been changed to protect the privacy of individuals mentioned.

invisibility. There are no public marches for NM, no ribbon campaigns, no ice-bucket challenges or celebrity backers. Instead, like most rare diseases, it is combatted in the wings, out of sight of the wider audience.

I first met Hannah and Asher at their home in uptown Manhattan, an apartment punctuated with the paraphernalia of healthcare: filters, feeding tubes, and wheelchair parts. Hannah and Gabriel divorced when Asher was young, and Asher visits his father in a neighboring state on occasional weekends. Asher is now almost sixteen years old, though he looks younger, a fair-haired teenager weighing just over sixty pounds. Because of NM, he has very little bulk on his frame.

Of the estimated one in fifty thousand people who are born with NM, some may die in infancy, while others experience only mild weakness that can go undiagnosed.

Asher's condition is on the severe end of the spectrum. He uses a wheelchair, unable to walk or stand on his own, and it can be a challenge to speak clearly since the fine motor control of his lips and tongue is disrupted. His swallowing reflex has also been compromised, meaning that not only is eating virtually impossible (he receives most nutrition through a feeding tube in his abdomen), but also that secretions are constantly building up in his throat that need to be suctioned off every hour. He breaths through a ventilator attached to a tracheostomy tube in his throat.

The most life-threatening aspects of Asher's NM have involved disruptions of this breathing apparatus, like when, on the thankfully rare occasions, his airway becomes clogged with mucus.

Hannah describes the abject terror of this event: her son suddenly turning blue as he begins to suffocate, unable to call for help. The possibility that this could happen again at any moment is a source of constant fear, meaning Asher can never be left alone for a single moment of the day or night. For Hannah, now working two jobs as a nurse practitioner, and without family support nearby, the stress can be immense.

Listening to Hannah speak about their lives, it occurs to me how often we take for granted these fundamental needs: the ability to walk, eat, speak, and breathe. The ability to be

alone. But the thing that surprises me the most about Asher is that, despite all of this, Asher is in many ways a typical teenage boy.

When we meet, he's preparing to write the Grade 9 Regents at school, with the hopes of going to college after graduating. Typical of his age, his personality is a mix of shyness, wry humor, and half-serious grandiloquence. He's a Broadway fanatic, a practicing drummer (fortunately, he retains dexterity in his hands), and an enthusiast of professional wrestling and basketball. But perhaps his greatest passion is video games, particularly the role-playing game *Destiny.*

Within the medical community, video games are often spoken of dismissively as mindless and unhealthy. But for Asher, gaming is meaningful source of entertainment, not to mention a flourishing social life. In *Destiny*, Asher's character is a powerful warrior equipped with gleaming, golden armor. He plays online with friends from school, each of whom suffers from their own disability or illness; facetiously, they've dubbed themselves "The Crippled Crew Clan."

"He's kind of the mayor," Hannah tells me, as we watch from the sofa. "He leads the group."

"I'm a legend," Asher adds with a shrug.

Wheeling up close to the screen, the fantasy world fills Asher's line of sight. Massive Sony headphones connect him with his teammates, blocking any outside noise.

"Jaaamie," Asher calls into his mic. "Jamie! You ready? Ok, let's roll."

On Asher's cue, the team of warriors runs and leaps across the screen towards their next mission.

Asher plays with people from all over the world, Hannah tells me, including others dealing with NM. Watching Asher banter with his friends, stopping every now and then to return a text on his phone, it occurs to me how incredible it is that he was able to forge this unconventional community, to connect with others even when physical closeness isn't possible.

Hannah, too, relies on online communities, like the NM Support Group on Facebook.

"It's a huge support system for me," she says. "Emotionally, socially, financially, everything."

Online, a few hundred parents of kids with NM exchange stories about their latest emergency or dilemma, and others who have lived through the experience weigh in.

Even just a few years ago, it must have been inconceivable for people dealing with such a rare disease to find one another. Now, Hannah describes them as family.

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In *Illness as Metaphor*, Susan Sontag writes: "Everyone who is born holds dual citizenship, in the kingdom of the well and in the kingdom of the sick."

Medical school is, at its outset, a cartography of the kingdom of the sick: we divide illness into provinces and territories, each with its own distinct character and history. We become anthropologists, trying to understand the experience of inhabiting these spaces.

Prior to this experience, I'd imagined rare disease as an uninhabited province; a gaunt landscape, lacking in resources and influence, without a city, or even a village, around which to centralize.

But the landscape isn't as sparse as I'd imagined. Advances in technology and social networking have evolved into new communities and advocacy groups. Those previously uninhabited regions are developing infrastructure, marshalling their resources. The result has been remarkable, with rare disease research and awareness increasing dramatically in recent decades.

About a month after our first meeting, Hannah arranged for me to visit Asher's school on Long Island. Walking through the wide front doors of the school, we are immediately cut off by three elementary-aged students zipping by in electric wheelchairs.

The school is unique: a specialized, K through 12 center with on-site medical care, dedicated solely to students with severe physical disabilities.

The hallways of the school are wide and bright, covered with student-made artwork. We pass a large "aquatic therapy" swimming pool where, with specialized harnesses, some students who are otherwise wheelchair-bound are able to walk and support their own weight.

Throughout the day, I notice several students who, unable to talk, make use of speech generating devices controlled by joystick or eye movement.

Even some of the teachers are living with disabilities. The principal waves a prosthetic hand as we pass.

The philosophy of the school is simple: that, given the opportunity, children with disabilities can go on to achieve full, independent, and productive lives.

Hannah tells me about other schools Asher attended prior to coming here, places where his education was neglected so badly that, by the age of eight, he could barely read. It took months of conferring with other parents, making phone calls, advocating, and wading through bureaucratic red tape to secure Asher a place at this school. But he's now flourishing.

The cost is substantial; the state pays about $72,000 a year for most of the 185 students who attend the school. But the benefit of investing in these kids is everywhere on display. About 80% of graduates go on to higher education, many of them continuing on to careers that might otherwise have been impossible. Older students receive vocational training and are encouraged to give back to the community.

On our way out, we walk by a banner listing some of the colleges attended by recent graduates: CUNY, University of Michigan, Harvard.

Asher's is just one story out of countless rare disorders that affect millions of people the world over. Mobilizing the resources to attack these illnesses individually seems almost impossible at first glance. But the benefits Hannah and Asher gain from their respective social networks suggests the power of these platforms to elicit change.

Social networks for rare diseases have become communities for advocacy and information-sharing; a way to connect, to ask questions, to provide financial support, and to learn from each other's experiences. I believe there could be significant untapped potential in expanding this concept to the healthcare community: creating a "medical home" in the online space, and encouraging medical professionals to build deep and enduring online relationships with their patients.

Connecting online could give researchers the opportunity to communicate directly with the people most affected by their disease. They could elicit input, like in deciding which research questions could do the most good for the community, and, importantly, raise funds to conduct more research by rallying the community together. This model has been shown to work; one online association for a disease called fibrodysplasia ossificans progressiva has, through grass-roots efforts, raised millions of dollars for dedicated labs, dramatically advancing research, despite this being one of the rarest diseases in the world. Such a partnership required sustained effort on both sides to build relationships and trust, including, for instance, email chains sustained for decades between researchers and organizers. Scientists may need to rethink their role, to step outside of the lab and clinic. But it is wholly possible, given the right motivation.

Clinical care could also benefit from real-time communication between doctors and patient groups, as patients often notice new manifestations of rare diseases before the medical community itself. For example, Asher has for years felt a build-up of mucus in the back of his throat where his suction tube can't reach; it's uncomfortable and constantly irritating. He mentioned this to his pulmonologist, but the concern was brushed off—the buildup wasn't going to impede Asher's breathing in any way. Conversing online, Asher realized that this was a universal complaint throughout the NM community. It was immediately validating; this was a real symptom. Collecting information like this could undoubtedly help physicians better direct their care. And social networks can assist in recruiting for research studies, creating tissue repositories, and fostering better communication overall. A stronger partnership online could mutually benefit both patients and physicians.

In 1657, William Harvey wrote that there is no better way to advance medicine than "to give our minds to the discovery of the usual law of Nature by careful investigation… of rarer forms of disease." The intervening centuries have validated this view: throughout medical history, the insights gained through the study of rare disease have frequently led to breakthroughs into far more common conditions.

Research into the bizarre disorder called Tangier disease, for example, in which a build-up of cholesterol causes people's tonsils to turn orange, is now leading to the creation of drugs

to prevent heart disease. Studies of cleidocranial dysplasia, a rare genetic disease in which children are born without clavicles, have uncovered key signaling pathways for normal bone development, with significance for osteoporosis and bone repair. It isn't a far stretch to imagine how NM research could reveal medical insights for the treatment of countless other muscle disorders.

This view narrows the gap between rare and common diseases: they may not be separate entities, after all, but different paths to understanding the same fundamental truths of human health and biology.

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My most recent visit to Asher's home found him, unsurprisingly, entranced in a video game, this time with a Western-theme. Having recently finished Grade 9 and written his Regents Exams (he scored honors in several), Asher was looking forward to summer.

Getting to know Asher and Hannah, I realized that my previous vision of healthcare, as the exclusive realm of IVs and blood pressure cuffs, was far too narrow. If the goal of medicine is to reduce suffering and improve wellbeing, physicians need to take a broader view, acknowledging that health doesn't exist in isolation of its social environment.

I've seen how robust social programs, including education and job training, can be essential for the mental health and quality of life of those dealing with serious illness; how internet access, social networks, and support groups can be as relevant to patients' health and wellbeing as medications. I've witnessed the considerable psychosocial benefits of seemingly frivolous entertainment like video games. A doctor's responsibility doesn't end when a patient walks out their door; to really make a difference, we need to take a more holistic approach to improving people's lives.

With all the stress and exhaustion of Hannah's work and of Asher's condition, I asked why they decided to open their lives to a medical student, what message they'd hoped to share.

"We'll do anything to help," Hannah said, simply.

Any small part she and Asher could play in raising awareness, in educating those in the medical community about NM might, Hannah hoped, mean more effort to find treatments. More understanding of the challenges they are facing.

It is the moral imperative of the medical community to build bridges into these remote regions of illness; to venture out with new research and innovation, to establish lines of communication. There is strength in community, even if the community numbers in the hundreds rather than millions. Asher and Hannah have managed to find and connect with others like themselves, to join their voices toward a common cause.

I am hopeful that we can learn to listen, and to act.