

“It’s All In The Name: Undiagnosis in a Diagnostic Age”
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I got to know Katherine through several disability leadership trainings over the course of my fieldwork. Her son, Jonathan, is nine years old. He is blind, nonverbal, has intellectual disabilities, and walks with the help of a cane; he’s considered to be on the deafblind spectrum, although he still has some hearing. He loves horseback riding and rides regularly at a therapeutic center in the city where he lives. When Jonathan meets someone, he slowly traces their hands with his fingers, paying careful attention to any identifying features, such as scars or jewelry.

Jonathan was born at 26 weeks gestation, a full three months premature. He was his parent’s first and only child. A so-called “million-dollar baby,” he spent 14 months in the neonatal intensive care unit (or NICU), and his mother was diagnosed a decade later with post-traumatic stress disorder from the ordeal. Jonathan’s survival is largely the result of stunning advances in newborn care in recent decades, yet he embodies a curiously modern paradox: his disabilities are inextricably linked to medical progress itself. The same technologies that have enabled his survival have, at the same time, ensured his bodily and intellectual difference. Or so we think. In the past, children like Jonathan simply did not live. Experts suspect he might have an underlying and unidentified genetic disorder; there might be more to his story than just prematurity. Lacking a diagnosis, his doctors simply refer to it as “the Jonathan Syndrome.” His family has undergone extensive genetic testing in the hope that, one day, they will receive a diagnosis. For now, however, they wait.

Disability holds an increasingly prominent role in the contemporary U.S. Recent decades have seen spikes in different disability populations, perhaps most highly publicized in regard to autism (or ASD). In fact, the trend is much broader and overall rates of diagnoses of neurodevelopmental disabilities and mental illness in children have jumped significantly in the last decade. It is critical to note that these include children with dual or multiple diagnoses. Diagnostic categories now proliferate, simultaneously naming and codifying difference in new ways and engendering previously absent disability worlds. Yet, for this growing population of children, their parents' questions often far outpace the current knowledge, both in the case of the rare and undiagnosed populations with whom I work and – as illustrated in regular news coverage – in terms of some of the more prevalent, widely-recognized diagnoses.

This paper uses data gathered from fieldwork with parents of undiagnosed children to rethink the practical and affective possibilities of diagnosis. It takes as its starting point that the aspirational category of the “typical” or “non-disabled” child is increasingly precarious. What does it mean – ethnographically, practically, and politically – when more children than ever have a diagnosis? Today, previously unexceptional aspects of childhood draw scrutiny. As an anthropologist and disability studies scholar I am curious about what this reveals about broader notions of risk, conformity, and potentiality. I am particularly interested in the potential impact of this diagnostic upswing on those who often get left out of popular discussion: people whose bodies and minds are already – from the outset – deemed to fall outside or at the borders of diagnostic common sense.

Much of my research examines the diagnostic journeys – or “diagnostic odysseys” – of parents whose children have undiagnosed disabilities. (Families like Katherine and Jonathan, whom I mentioned earlier.) The National Institutes of Health estimates that up to 40% of children with disabilities in the United States are undiagnosed. There is no label for their recognized impairments, behaviors, medical issues, or symptoms. This is a staggering figure, yet there is a surprising lack of research on this topic. Appearing to be made up of mismatched pieces to different diagnostic puzzles, these children do not make sense – medically, politically, or socially. These are the kids who, until quite recently, were met by specialists with a shrug and a placeholder designation of “intellectual or multiple disabilities.”

Within the constraints of the resources available, these parents seek help from all possible angles, often from Day 1. Families visit specialists in their hometowns, states, and at medical and research hubs across the country. They join listservs and Facebook groups, attend conferences, read PubMed articles religiously, and network with other families. Like Jonathan’s parents, they hope to eventually secure a diagnosis – most likely one for a rare and sometimes little-known disorder, perhaps with a tiny diagnostic population, maybe even only in the single digits worldwide. Children who are undiagnosed elude the commonly recognized labels we all know. Their families have to dig deeper – often, whenever possible, setting their sights on genetic testing. In many cases, biomedical technology and contemporary diagnostics cannot (yet) meet their needs. Their search for an answer continues.

To be undiagnosed, I was told, is to be neither “typical” nor diagnostically marked. There is no easy explanation or canned answer to the inevitable questions; no

elevator speech. One is diagnostically stateless, nameless, protected by fewer resources, rules, or practices. Each visit to a specialist becomes an application for diagnostic asylum, a request to be ushered in from a stateless refugee status to a more concrete transitional stage with the promise of a clear label, group, and biomedical claim.

During my fieldwork, I began to wonder how this ambiguous and little-discussed, albeit quite sizeable, population connected to the broader explosion of attention to disability in the media and in public life. There is a tendency to speak of disability in concrete terms, as if the labels are fixed, static, and universal. (As we know here, this makes disability an ideal area of study for anthropologists.) Disability has emerged on the public scene full force, and diagnoses for the parents I worked with continue to be equated with hope, curative potential, and new futures. There have been dramatic shifts in what childhood disability means today, compared to 20, 50, or 100 years ago (as has been well documented), including the fact that rates of physical disability among children have gone down while diagnoses of neurological and developmental disabilities have risen significantly. The bodies and faces have changed.

We are beginning to see the spectrums involved with autism, Sensory Processing Disorder, language delays and disorders, fetal alcohol syndrome disorders, and more. But what about the rare genetic conditions I tend to encounter? M-CM, Angelman's syndrome, CHARGE, Mowat-Wilson, Rubenstein-Taybi, Prader-Willi, Trisomy 13. Each of these designations can refer to a diverse range of disability portraits, and yet *a diagnosis remains a thing*. It makes sense out of bodily, intellectual, and mental difference. It groups patients under umbrellas and, perhaps especially in our

digital age, those umbrellas can give way to full-on communities. A diagnosis can give families hope for futures once thought unattainable. It can give a window into life expectancy, comorbidity/dual diagnosis, risks, and needs. And, for children, it can secure necessary educational and therapeutic services that would otherwise be out of reach, since a diagnosis is still thought to be the cornerstone of meaningful and appropriate interventions – particularly when subsidized.

Multiple parents I worked with lamented that their children’s disabilities weren’t of the easily recognizable variety, often mentioning Down syndrome and autism. Some attended mother’s groups or local parent support groups, but their children – and, by extension, their families’ narratives – were the outlying cases. As one woman told me: everyone in her moms’ group was nice to her, but their children all had autism; her daughter was the only one with a feeding tube. This example is significant, in that it illustrates the central role of bodies in the undiagnosed and rare disability communities, something that has been woefully pushed to the side in much of disability studies. In these cases, bodies emerge center stage as split, fragmented, and visibly quite different. Their tracheostomies, feeding tubes, seizures, crashes, special diets, chronic pain, and repeated surgeries. Here, it becomes increasingly more complicated to focus on the social or historical facets of disability experience and not also the physical.

Without local access to families in similar situations, these parents of undiagnosed children tended to connect via social media, using Facebook, blogs, disability websites to share and gain insights regarding undiagnosis. I spoke with several who literally helped diagnose their own children this way. As one mother described, the diagnostic journey was very “DIY.”

In most of my conversations, the diagnosis was the end goal. It was a journey with many hiccups and wrong turns on all sides, sometimes lasting well into a child's teens. But why was it so paramount – particularly when the special ed services were already in place and medical complexity was minimal? Parents tended to speak of two benefits. First, they wanted a window into their child's future –the unanswered questions of life expectancy, health issues, possibilities, the good and the bad. Second, they wanted to build communities. Networks of families like theirs, sharing experiences and information. Diagnostic knowledge was equated with power. This was taken as fact.

And yet, I came across parents of adults with rare conditions who had little interest in connecting with the new diagnostic generation. It simply was not on their radar. Perhaps it was too painful and they were too tired after years of struggling with school systems, physicians, therapists, and now the embattled world of adult services. Perhaps they simply did not identify, these families whose children came of age in a pre-digital era that was, in terms of disability information and access, a world away from where we are now. These families – seemingly at the vanguard of helping to understand certain diagnoses over the life course – were anything but excited about their position on the front lines.

I think, too, of a mother I met. I'll call her J. "He's a good project." This is what the specialists say about her child, Owen, who is still a baby. He has the typical list of seemingly haphazard symptoms: low muscle tone; developmental delay; poor eye contact; little to no speech. I hear these same terms rattled off casually by parents.

Her son was diagnosed at the age of three weeks, although she tells me could have known sooner had the hospital run a genetic panel after his birth. Three weeks, though. It's one of the fastest diagnostic stories I encounter. No goose chases or misadventures, no "Owen Syndrome" designations. Owen has a diagnosis. Or, rather, genetic testing identified the gene mutation behind what could not be explained previously. Owen has a highly specific and rare gene mutation.

I'm struck by the potential differences of knowing so early. "I feel lucky for the diagnosis," J., the mom, tells me. "Not to have *this* diagnosis, but to have a diagnosis." I nod. She says that the worst possible thing for a parent is not knowing, a sentiment I've heard again and again. *But what does the diagnosis mean*, I push. "I don't think it means anything," she replies. "Nothing...If you can't tell me what my child's going to look like in one, two, four, 10, 12, 15, 20 years, than you're not of any use to me. But I think it gives people some peace that it's nothing they did and it's nothing they can undo. That internal peace for people is really the best thing that this has done."

Hearing such stories, I began to think seriously about the pitfalls of diagnoses in the rare and undiagnosed disability community. There are clear logistical, medical, and affective benefits, to be sure, yet I cannot help but wonder if these new and often quite obscure labels – many without a name, referencing only the identified gene mutation – actually undermine broader possibilities for mobilization and meaning within the undiagnosed community.

Disability studies scholar Lennard Davis has written that disability continues to fall outside of dominant diversity paradigms, those shiny new ways of ordering and affirming difference. He argues that this boils down to one thing: medicalization.

Disability remains perceived as a medical category (outside of certain academic circles) and diversity does not apply to the disabled, ill, or dying. There is no room for such bodies. Diversity works, he says, because people are all different and equal as long as they are “not *that kind* of different” (2013, 14). Here, the old distinctions of normal versus pathological continue to reign. As a de-historicized bodily condition perceived of as needing a cure, disability cannot be welcomed by a diversity paradigm.

I would argue that something similar happens when today’s diagnostic fervor unfolds in cases of undiagnosis. What could be more medical than receiving a genetic diagnosis that no one else has? One is pulled from the diversity of undiagnosis. Individualized, named, and separated.

In contrast, I see a *real* and potentially transformative potential in reconceptualizing undiagnosis as more than just a liminal state between unknowns and biomedical truth. And I wonder how such reframing might give way to a new and newly meaningful disability politics of undiagnosis for the millions of American families with undiagnosed children. Within disability anthropology, it is critical to interrogate the baseline assumptions of diagnostic power, asking instead how a diagnosis (and, indeed, undiagnosis itself) both emancipates and constrains. What is at stake for the many families I spoke to who knew of only a handful of cases of their child’s diagnosis around the world? Or, in a few instances, none? And what potential might exist for scholars, along with families and allies on the ground, to help reframe undiagnosis as a powerful platform for a distinctive embodied and affective disability experience?

THANK YOU!!!