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DISABILITY

My 'Orphan Disease' Has Given Me a New Family

By Rosemarie Garland-Thomson

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The members of my family share a number of traits, but not one of them looks quite like me.

Many of us have the same sturdy physique, blue eyes, fine Northern European hair and skin that should never see bright sun. I look pretty much like that except for one very unusual characteristic. I was born with asymmetrical, atypical hands and forearms into a world where symmetry and typicality are the marks of good looks and proper function. In fact, I am so unusual that in the half century of my lifetime, I have encountered only one other person who looks precisely like me. In other words, I'm rare.

I recently learned that I am distinguished in this way because I have a rare genetic condition — complex syndactyly. Before I learned this, no medical doctor had ever presented a diagnosis more helpful than a shrug.

Before being able to identify and give a name to what I have, I was subject to plenty of unsavory terms: “freak of nature,” “funny-looking kid,” “deformed,” “birth anomaly,” “sporadic limb deficiency.” The most disagreeable and persistent was “birth defect,” the unfortunate outcome of whatever sin or defilement could be guessed at or imagined — alcohol consumption, pollution, environmental contamination. With no impressive diagnosis to offer to the perpetual query about “what happened” to me, I usually resorted to, “I was born that way.”

I'm glad to now have a “rare genetic condition” instead of a “birth defect.” Anything “rare” has prestige, suggesting something sought after and prized by important collectors, archaeologists or scientists. Indeed, so exceptional is my way of being that fewer than one in 90,000 people are enough like me to receive the same diagnosis. It's a bit like winning the lottery.

Before decades of scientific work gave us the genome map in 2003, our ways to explain unexpected human variation were limited. People like me were inexplicable. As often happens with the inexplicable, supernatural and superstitious reasoning rushed in. Mother-blaming and divine retribution were prominent. Mortal sins, erotic thoughts or any violation of the social code was thought to produce a disabled child.

Modern versions of this superstition haunt the mothers of congenitally disabled children; they experience corrosive guilt about exposure to toxins ranging from face cream and nail polish, alcohol and cigarettes, to thalidomide and the BPA in our plastic bottles. People with disabilities were then and are now taken as cautions or warnings of bad things past or future, canaries in the coal mine of human existence.

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But things have changed. “Rare” is now an inflection in my personal dignity tool kit, a status upgrade. Even “syndrome” has an air of sophistication, an augmentation rather than the lessening that “deficit” or “defect” brings. It’s something you *have* instead of something you *don’t have*. More important, my form is not a mistake, some random whack from a menacing outside world. My shape is intentional, manifesting deliberately from some mysterious purpose at the very core of my being. Evolution’s purposive caprice, some inexplicable force beyond our puny human imagination, is trying out a new design.

Today, identifying genetic diseases is a growth industry, powered by scientific-research funding and lucrative commercial interests. With testing available everywhere now, from private companies to clinics, people whose genetic diseases have been identified are an increasing population. All of us, it turns out, are carriers for at least eight to 10 common, significant genetic diseases and even more rare, enigmatic conditions.

What this tells us is that we are all related to one another through a system of genetic lineage that almost no one understood until recently. These genetic kinship circles are expanding and connecting us through networks of recessive, dominant and autosomal genes; mitochondrial DNA; and complex interactions with the environment that shape how genes express themselves as we develop. Medical science has discovered more than 7,000 genetic diseases, and new ones emerge every day.

People in my world of disability pride and advocacy sometimes call themselves by tribal names such as “a thalidomide” or “a polio” or even “crips.” Our new genetic identities now yield more complex affinities. Rare genetic conditions are also called “orphan diseases.” Now that I have an orphan disease instead of a birth defect, I’m no longer an orphan but instead newly a member of several distinctive tribes, a heretofore hidden web of kinship and clan affiliations. Resemblances are crucial to kinship, whether familial, tribal or ethnic. What we look like tells us and other people to whom we belong. Our distinctive traits gather us in kinship networks that often provide us with alternative families of mutual care and support.

In a recent conversation about Crispr, the newest genetic editing tool, the geneticist and Nobel laureate Mario R. Capecchi told me, “The purpose of evolution is to anticipate the unexpected.” Random genetic variation is what moves evolution forward, yielding new forms that are fresh solutions to changing environments both natural and human designed.

The short span of human life and imagination limits our capacities to anticipate the unexpected. Ways of being that meet the demands of human life as we live it here and now may not serve our distant descendants so well. Characteristics we think of as intelligence, strength, vision, uprightness, dexterity, body mass or whiteness will outlive their usefulness, morphing from advantages to disadvantages, counterintuitive though that seems today, especially perhaps to those who have those valued traits and benefit from them.

Capecchi’s framing of evolution’s purpose as anticipating the unexpected can give us progressive perceptions about living with disabilities and stand as a caution against hubris and narcissism in our aspirations to shape our human communities according to the traits valued by the majority.

People with disabilities are the unexpected made flesh. The challenges of living in a world not built for us are occasions for resourcefulness and adaptability, especially for those of us who start out disabled early in life. We are innovators, early adopters, expert users and technology hackers as we respond to the adversity that the built and natural environments present us.

We don’t know which human variations will be advantages and which will be disadvantages in the long arc of our struggle to prevail in an ever-changing environment. My blind friend with excellent orientation skills quips that she will be leading all of us out of burning buildings and planes when the lights go out. My deaf friends avoid the stress of noise pollution and the exhaustion of talking over the incessant din in fashionable bars in which social and professional life takes place now. Another friend, who is a small person, remarks that he consumes fewer resources and fits better into airplane spaces than big guys good at fighting and football. Some people with autism have a capacity to focus that boosts creativity. Expertise at composing with my voice instead of clunky keyboards puts me ahead in new communication technology.

The bright line between the healthy and the diseased, those who “have” a disease and those that don’t, grows dimmer every day. Each of us carries within us many “orphan diseases” — that faintly Dickensian phrase that typically severs the connection between people like me and the human family of ordinary people. All of us are anything but “orphans.” Instead, we are all second or third cousins, inextricably linked through a chain of quickly vanishing ancestors and descendants. We are all patients-in-waiting, bound together on a wait list of inheritance hidden deep in the elegant whirls of that double helix in every one of our cells — silently paused in its inscrutable determination to shape our lives. All those clichés of connectedness are now encoded in our genes. The human community is quite literally the human family.

We disabled are no longer orphans. We are instead a sturdy tribe, blood kin, navigating a changing world, living well, bonding fast and passing it on.

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