

Rare Disease Day: I'm a circus freak and I'm in great pain

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Photo credit: Shalayne Smith Needham

Well, it's official—I can be featured in the circus sideshow. I can touch my tongue to my nose, my thumb to my forearm, and hyper flex all of my fingers upwards to a 90 degree angle, along with many other weird bendy, contortionist body positions. Turns out these party tricks aren't just amusing to bystanders, but are causing me extreme pain and physical dysfunctionality. That's because I have Ehlers Danlos Syndrome hypermobility type (hEDS).

EDS is considered a rare disease, and February 28th is Rare Disease Day, which is represented by zebra stripes to show that people with rare diseases are unique but not uncommon – there's a whole herd of us! One in ten Americans has a rare disease. In fact, rare diseases affect more people than AIDS and cancer combined.

It took me 30 long years to get the diagnosis, after begging and pleading with doctors that something was not right. Lack of research, reliable information and general awareness often means misdiagnoses and long diagnosis times. hEDS is around 30 years. I almost can't wrap my head around it.

hEDS is an inherited connective tissue disorder characterized by defective collagen in the joints and between internal organs. Symptoms include joint hypermobility affecting large and small joints, frequent joint dislocations and partial dislocations, soft and smooth skin that may be slightly elastic and bruises easily, chronic musculoskeletal pain, early-onset osteoarthritis, gastrointestinal issues, dysfunction of the autonomic nervous system, and increased risk of pelvic prolapse, painful menstruation, and painful intercourse in women. Needless to say I'm constantly in pain, fatigued, and experiencing severe physical discomfort. hEDS also causes poor proprioception—my sense of my body in physical space and how to move it appropriately. I'm a legit, certified klutz. Let's just say I break a lot of stuff...

It's easy to deduce that rare diseases negatively impact people financially, socially in and their overall quality of life. I can certainly attest to absurdly high ER bills, overpriced medication, and a decreased stamina for how many hours I can work in a day. Socializing is a genuine treat for me as it is not a weekly or even monthly activity I can participate in because I'm so busy surviving. Doing daily tasks for me is often a burden, leaving me exhausted and in pain. Eating, sleeping, and just brushing my teeth (made difficult by jaw joint dysfunction) are usually a serious challenge. This is all of course exacerbated by my Fibromyalgia and Gastroparesis. Every night I'm honestly amazed I survived another day.

There are few drug treatments or therapies for people with a rare disease. Ninety-five percent of rare diseases do not have a single FDA approved drug treatment and there are no cures. This is unfortunately the case for hEDS. I have started physical therapy, and it is going fantastically. If only the dozens of doctors I'd begged over the years had recommended it when I asked, I'd be further ahead of the curve in terms of joint stabilization. I rely on prescription NSAIDs, muscle relaxers, anti-nausea and anti-convulsants, SSRI-class drugs, and some other things to keep me going. I'm operating my own small pharmacy at this point.

Perhaps the most difficult part of it all for me is the invisibility aspect. I look 'fine', so when I'm unable to stand in the checkout line at the store for more than a few moments or put the shopping cart back, I see the judgement on people's faces. I use the accessible bathroom stall whenever possible because getting up and down from low toilets is difficult for me. This leaves people wondering why I've taken a stall from a wheelchair user. I have a big list of things I can't eat because of the GI issues, so feeding me isn't always easy. I'm not doing some Millennial cleanse trend, I'm just trying to not get in

a cyclical vomiting episode. And if I had a dollar for every bloody 'cure' I was offered I'd be able to actually fund a cure. Mysterious shakes, supplements, yoga, meditation, and positive thinking are constantly recommended to me as a way to thoroughly treat or cure my conditions. I admit that I've grown impatient with these responses, as I now see them as victim blaming and/or predatory.

So what can you say or do for someone with a rare disease? Here are my recommendations:

1. Plan friends' date night and make it accessible for that person – whatever it means. It gets real boring real fast being sick all the time.
2. Drive or accompany them to a doctor's appointment. Moral support can't be underestimated when you are dealing with busy or condescending medical professionals.
3. Invite them to your parties, family outings, etc. They may not be able to go, but just knowing they remembered you means so much.
4. Offer to help with chores or personal care. I would die of joy if someone braided my hair for me since styling it is so excruciating in terms of pain and fatigue. Are you one of those people that love to vacuum or weed? Someone out there needs your help!
5. Simply tell them you appreciate them, love them, etc. We deal with a lot of negative self-perception and it helps to be reminded you have worth.

Celebrate Rare Disease Day February 28th by wearing zebra stripes, sharing awareness posts on social media, and telling those you love that they are awesome for the daily effort it takes to live with a rare disease.

Sources:

<http://thezebranetwork.org>

<https://rarediseases.org/rare-disease-day>

<https://www.ehlers-danlos.com/what-is-hsd/>

<https://rarediseases.info.nih.gov/diseases/2081/hypermobility-ehlers-danlos-syndrome>