A journey through testing times

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"….burning with curiosity, she ran across the field after it, and fortunately was just in time to see it pop down a large rabbit-hole under the hedge. In another moment down went Alice after it, never once considering how in the world she was to get out again." (Lewis Carroll, Alice in Wonderland.)

Does autism run in your family? Why is your sister like that? Was she born that way? I have spent a life time answering these questions never knowing the answers myself. My youngest sister, Eesha, was diagnosed with autism when I was twelve and she was a three year old baby. Needless to say my earliest memories surrounding the words hereditary and familial weren't exactly pleasant, and they were often accompanied by a sense of confusion and a feeling of persecution. For the next few years Eesha remained a skeleton in the family closet. I entered the field of genetic research with this background baggage and rolled down the rabbit hole into the genetic wonderland filled with curiosity and wanting all sorts of answers.

My initial attempts at discussing genetic tests with families who had "pedigrees suggestive of a genetic predisposition" were clumsy and often awkward. I was scared of cancers and considered mutations a disease and had only begun adjusting into my role as a research officer who had a MBBS degree but was no longer a "doctor." I was taking consents for research that often had "bad" or confusing results and which many times translated into life-altering decisions for the research participants. I struggled and stumbled in the dark with Urdu explanations for genes, hereditary illnesses, single nucleotide polymorphisms and other genetic paraphernalia for the next few years. I would love to say that eventually I had some genetic epiphany but of course that didn't happen; however, over time some things did change.

On a personal level, over the years as I have entered further into the joyous and heart aching journey of raising an autistic sibling, I have become grateful for her silent, gentle presence in my life. Over the years autism has changed too. It now has many names and shades and many genetic links have emerged through research. The unpredictable future of our unborn children sometimes still worries me and my siblings. We try not to remain complete hostage to such thoughts. On the professional front, my encounters with genetic research participants have certainly changed over the course of time. The 70 year old "Amma," whom I met the other day, was concerned about passing on her cancer to her daughters just like she got it from her mother, but could understand only few things in my simplified genetics 101 lesson. She wanted to talk and my time was the only thing I could offer. She left with a pat on my head and I was left humbly educated by her ability to see disease, disability and death as a natural flow of life and her complete acceptance of human genetic "imperfections."

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Over time, I have also learnt that doing genetic research means entering into a “relationship” with the research participant families. Besides the requisite information that I collect for research records, I hear about marital discords, the troublesome in laws, the nafarman aulad, (disobedient offspring) the nalaik bahu, (useless daughter-in-law) and every possible imaginable personal and family life issue, and often end up giving lots of personal advice. It's also often hard to be just laying out clinical options for risk reduction without mixing it with some subjective often paternalistic advice. Lastly, I believe, with the ever expanding “unknown” in genetic research, one of the greatest assets for a researcher is a certain amount of “genetic humility,” recognition of the inherent haziness of the genetic crystal ball and the fine line that separates prediction from pure speculation.

Eesha turned 26 last month. She gingerly accepted my hug but smiled widely at the birthday feast we had laid out for her party. I also recently came across an article about genetic testing of embryos to screen out genetic diseases. We all discussed it, perhaps might even consider it someday, but for now, whatever will be, will be…

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