



Consumer Corner: The Future of Genetics

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Seven years ago we embarked on a journey for which we were completely unprepared. We couldn't imagine that our little girl's first years of life would be wrapped in misery and diagnostic mystery. Fast forward to today and you will find us as a family empowered with knowledge, advocacy skills, and the hope we have gathered along the way. So what does the future of genetics look like from our perspective?

We have been told that our daughter is suspected of having a mitochondrial disorder, which is genetic and congenital. Many told us there is no single test to diagnose it and there is no cure. Others told us that diet and environmental changes will have no impact on her disease progression. But, as parents, what we saw in our daughter did not match what we were being told. We saw her health drastically decline with each well-baby visit. We saw her exhibit autistic-like behaviors after we introduced cow's milk into her diet and we witnessed her neurological reactions to household products (hairspray, window cleaner, etc.). We began to "lose" her around 18 months, while multiple sub-specialists continued to be perplexed as to what could be causing her symptoms.

Currently, no genetic cause for our daughter's symptoms has been found, after years of testing. As parents we have had to actively research reasons

for our daughter's condition and coordinate her care. No one has connected either the "dots" or her many specialists, so we had to step up to do that. Based on these personal experiences, we believe that in order for the future of genetics to lead to successful outcomes for every patient, three areas need to be addressed:

- Environmental exposures must be considered in every case. Environmental factors do impact a patient's life. "Environmental" includes everything from the microbiome to air quality, from medications to pesticides on food, and everything in between.
- Epigenomics. It is crucial to recognize that this "turning on and off of genes" can be modified through environmental factors, specifically diet and nutrition. It is also critical to acknowledge that "inherited" does not equal "genetic."
- Patients/family need to become equal members of the care team. Parents prefer personalized care to "one-size-fits" all medical practice.

If a concerted effort is made to integrate these three areas into genetics practice, many patients can experience an improved quality of life, regardless of their disease prognosis. By empowering the patient and his/her family to take an active role in care, it is possible to uncover creative, cost-effective, and disease-improving solutions for that patient, and potentially others.

Our daughter's future could not wait. We followed our instincts regarding diet and environmental changes, from what food we fed her to what chemicals we allowed her to be bathed in, rubbed with, and injected with. What we witnessed is nothing short of a miracle. Our daughter re-emerged and returned to us. Today, she is a happy, engaged 6-year-old with a very bright future.

For more information about our story, visit our blog at:

<https://babyfoodsteps.wordpress.com/about/our-story/>

