Knowledge Is Power When It Comes to Quality of Life

My name is Harriet Schleifer, and I am hoping you will join me in the quest to find the diagnosis of your yet-undiagnosed loved ones who have special needs. My 36-year-old son, David, captured our hearts, but changed our lives forever. His diagnosis eluded us until only three years ago.

His syndrome was only recently identified and is known as Transketolase Deficiency or TKT Deficiency. Unbeknownst to us, both my husband and I are carriers, as is my older son, but David inherited both copies of the TKT gene, thus resulting in the disease.

We, along with others, including medical professionals, hope to identify affected individuals whose families don't know the etiology of their symptoms. We need to be able to identify the natural history of our affected loved ones and get TKT Deficiency on the prenatal screening panel. Knowledge is power.

With modern technology it is now possible to determine the full genetic sequence of all the genes one has inherited. This has led to rather amazing scientific and medical advances. And this is especially true for Jews, as our historically somewhat insular population, from a genetics perspective, has resulted in an increased frequency of genetic variants—both good and bad—in our gene pool. For example, it is well known that there is a genetic predisposition to increased lifespan inherited by some lucky Ashkenazi Jews. They seem to have been blessed genetically to live to 100 or more while retaining all their faculties. Perhaps Moses was the first to have this longevity gene!!

There are also many genes that cause disease when an individual inherits TWO defective copies of a gene. Gaucher's disease, cystic fibrosis, Tay-Sachs disease, and spinal muscular atrophy are examples with a high prevalence in Ashkenazi Jews compared to the rest of the population. The reason for this high prevalence is that the frequency of being a carrier of one defective copy of the gene is between 1 in 10 and 1 in 50. That means many of us are carriers of one of these genetic diseases.

TKT is much more common in Jews than non-Jews. Even though this defective gene is far more common in Jews it is still relatively rare. Only about 1 in 500 Jews have a defective copy of this gene. If two carriers have children, then there is a 1 in 4 chance of their children having this disease. Thus far fewer than a dozen individuals in the world have been diagnosed because it has only recently been identified.

The characteristics of TKT deficiency include intellectual disability and developmental delay, short stature, and heart defects. There may also be delayed or absent speech, ophthalmologic abnormalities (strabismus, cataracts, uveitis), facial dysmorphism, hyperactivity and hypotonia among other challenges.

If you are interested in further information, or just want to talk, please do not hesitate to contact me at hpschleifer@gmail.com.

My gratitude to JCC Association of North America for circulating this PSA and helping to possibly advance science and the quality of lives.