



Dystonia and the Jewish Community

Dystonia is a painful, disabling disease for which there is no cure. For almost three decades, a small but growing group of scientists has labored to understand how and why dystonia occurs—and how to stop it.

The answers needed to cure this disease may very well lie in the study of one specific form that is particularly prominent in persons of **Ashkenazi Jewish descent**, called **early onset torsion dystonia**. This community has suffered greatly from this disease, and it is believed that better treatments and ultimately a cure are possible by paying close attention to how dystonia affects Ashkenazi Jews both as a population and as individuals.

- In 1911, the disease was originally called, “dystonia musculorum deformans”
- This form of dystonia was discovered to be caused by a mutation in a gene called **DYT1**.
- Thousands of Ashkenazi Jewish families in North America are estimated to carry the mutated DYT1 gene
- A genetic test is available to determine if a person is a carrier of the DYT1 gene, though not all individuals who have the gene ultimately develop symptoms of dystonia.

What is known?

- We know that there is a higher prevalence of DYT1 dystonia in Ashkenazi Jews. Medical literature is lacking in definite data regarding the frequency and prevalence of this disorder, but the latest sources state that **less than 1 percent of the overall population are carriers of the DYT1 mutation. Among Ashkenazi Jews, the frequency is at least 3-5 times higher.**
- We know that a mutation in the DYT1 gene is associated with early onset dystonia, but not all persons who develop this disease have the mutation.
- **90% of Ashkenazi Jews who develop early onset disease have the DYT1 mutation**, but only about 50% of non-Jews with the same disorder have the mutation. Therefore, there are probably other genes that are involved in some individuals, especially non-Jews.

Why is the DYT1 mutation more prevalent in Ashkenazi Jews?

- The Ashkenazim accounts for 80% of all Jews. The Ashkenazim is a group in which a number of genetically inherited diseases occur at an unusually high frequency as compared to non-Jewish populations. For many of these disorders, a causative gene has been identified and 2-3 mutations are found to cause most cases (90%+) of the disease within the group.
- There are several explanations for why a given population may have a higher prevalence of a genetic disorder than the general population. The **most plausible explanation is a “founder effect.”** A disease may become common within a population because all individuals within the population are descended from a small number of ancestors, and one or a few of the ancestors were affected by the disease.

What is the Importance of Studying DYT1 dystonia?

A better understanding of DYT1 dystonia will contribute hugely to the greater field of dystonia research. These studies provide a helpful model of dystonia in terms of:

- How the DYT1 gene mutation **biochemically affects cells and the nervous system.**
- How the familial forms of dystonia are **inherited**, and why symptoms within a family may vary
- Achieving a **better understanding of how dystonia affects populations**, and especially the Jewish community.

All of these elements will illuminate a route to better treatments and a **cure**.