Review of basic research areas for dystonia

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Basic research typically refers to work that does not use patients. Of course, patient symptoms provide the most complete measure of the progression, severity and nature of a disease. However, this can be difficult to assess because of the complexity and variability of humans. Patient research also cannot easily identify the molecular or cellular mechanisms of disease, and it is very difficult to distinguish causative events from phenomena that are an indirect response to the disease process. Basic research uses model systems to study individual aspects of a disease, and these model systems are typically chosen for their simplicity, homogeneity and because they are easily manipulated. A challenge for dystonia is that the relevance of basic research depends on studying an event unequivocally linked to the disease, but yet we do not know the cause of most dystonia. This underlies why basic dystonia research often uses genetic mutations that cause certain rare forms of dystonia. In essence, these are known triggers of dystonia and we can study how they impact molecules and cells to identify the mechanisms that underlie symptom development. Most basic research has focused on the first-identified dystonia mutation (DYT1/ TOR1A) but, with the identification of many more dystonia-causing mutations, basic researchers now have new tools they can use to identify the mechanisms underlying this disease.