Open Peer Review on Qeios

National Cancer Institute

Source

National Cancer Institute. <u>Torsion Dystonia 6</u>. NCI Thesaurus. Code C156361.

An autosomal dominant condition caused by mutation(s) in the THAP1 gene, encoding THAP domain-containing protein 1. It is characterized by dystonic craniofacial movements, dysarthria, and dysphagia. Limb involvement is common.