

# TECPR2 Gene

Subjects: Genetics & Heredity

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Tectonin beta-propeller repeat containing 2: The TECPR2 gene provides instructions for making a protein that is involved in a cellular process called autophagy.

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## 1. Normal Function

The *TECPR2* gene provides instructions for making a protein that is involved in a cellular process called autophagy. Cells use this process to recycle worn-out or unnecessary cell parts and break down certain proteins when they are no longer needed. During autophagy, materials that are no longer needed are isolated in compartments called autophagosomes. The autophagosomes are then transported to cell structures that break the materials down. The TECPR2 protein is thought to be important for the formation of autophagosomes.

## 2. Health Conditions Related to Genetic Changes

### 2.1. Spastic paraplegia type 49

At least five mutations in the *TECPR2* gene have been found to cause spastic paraplegia type 49. This condition is characterized by gradually worsening muscle stiffness (spasticity) and paralysis in the lower limbs (paraplegia), intellectual disability, and other neurological problems that lead to trouble regulating involuntary body processes, such as digestion and breathing. Many of the mutations that cause spastic paraplegia type 49 change single protein building blocks (amino acids) in the TECPR2 protein. Others lead to production of an abnormally short protein that is likely broken down quickly. Alteration or loss of the TECPR2 protein is thought to impair autophagy, making cells less efficient at removing unneeded materials. Researchers suggest that nerve cells (neurons) may be particularly vulnerable to impaired autophagy because it is especially difficult to transport waste materials through their long extensions (axons and dendrites) for breakdown. The waste materials can build up in neurons and damage them. Damage to neurons results in the neurological problems that occur in spastic paraplegia type 49.

## 3. Other Names for This Gene

- KIAA0329
- SPG49
- tectonin beta-propeller repeat-containing protein 2 isoform 1
- tectonin beta-propeller repeat-containing protein 2 isoform 2

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