Rare Turkish neurodegenerative disorder caused by a single mutation

International teams of researchers using advanced gene sequencing technology have uncovered a single genetic mutation responsible for a rare brain disorder that may have stricken families in Turkey for some 400 years.

The discovery of this genetic disorder, reported in two papers in the journal Cell, demonstrates the growing power of new tools to uncover the causes of diseases that previously stumped doctors.

Besides bringing relief to affected families, who can now go through prenatal genetic testing in order to have children without the disorder, the discovery helps lend insight into more common neurodegenerative disorders, such as ALS, also known as Lou Gehrig’s disease, the researchers said.

Both teams identified a new neurological disorder arising from a single genetic variant called CLP1. Children born with this disorder inherit two defective copies of this gene, which plays a critical role in the health of nerve cells.

Read the full, original story: Scientists discover new rare genetic brain disorder