Discovery of gene mutations for motor disorder leads to treatment in children

Doctors have discovered a new genetic disorder that robs children of the ability to walk normally and makes it hard for them to control their other limbs.

…

The analysis revealed that the patients had mutations in a single gene called KMT2B which is thought to play a central role in switching on other genes needed for the proper control of body movements. The corruption of the gene leads to what doctors call dystonia, when muscles contract involuntarily.

…

Doctors can already refer people with dystonia for what is called deep brain stimulation (DBS), but the treatment is a last resort, used only when drugs have failed.

…

When it is used to treat all forms of dystonia, DBS works well in some, but not in others…The younger ones seemed to fare best, to the point that some regained the ability to walk unaided. “So far all of the children we have seen seem to have a positive response,” said [Manju Kurian , a specialist in movement disorders who led the research].

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion, and analysis. Read full, original post: Discovery of genetic disorder allows breakthrough in treating children