

## Supporting CDKL5



### **Historic, clinical and prognostic features of epileptic encephalopathies caused by CDKL5 mutations. Paediatric Neurology 2012.**

This is a retrospective study from the Mayo Clinic, looking at the clinical features and treatment of seizures in 6 children (4 girls, 2 boys) with CDKL5. The onset of seizures ranged from 1 to 3 months of age. Particular features are discussed including dysphagia (difficulty swallowing) and cortical visual impairment. It is also noted that the 2 males in the group appeared less affected by certain features than the females. No particular treatment eliminated seizures, but topiramate, vigabatrin and the ketogenic diet were the most helpful at reducing their frequency.

Note - This is a study of a small number of children with a relatively short follow-up period, less than 3 years in 2 children and no more than a year in the other 4. The children studied are all young (eldest less than 3 years of age at review). The prognostic features referred to in the title seem to be the presence of the CDKL5 mutation itself and the resultant early-onset of seizures. Vagal Nerve Stimulation (VNS) does not appear to have been used as a treatment option, possibly because of the age of the children involved, although a recent [study](#) has suggested that VNS is a safe and effective treatment for resistant epilepsy in young children. The authors of the above study rightly conclude that more research is needed.