A novel transcript of cyclin-dependent kinase-like 5 (CDKL5) has an alternative C-terminus and is the predominant transcript in brain. Human Genetics 2012.

The molecular characterisation of the CDKL5 protein is still largely unknown. However, previous research has identified 3 different variants of the CDKL5 protein. This paper describes the theoretical existence of a 4th variant which the authors then go on to show not only exists, but is likely to be the predominant form expressed in the human brain. Although the CDKL5 gene contains 21 coding exons, this novel variant is shorter, only incorporating amino acids coded in exons up to 18. They conclude that this novel variant is likely to be of primary pathogenic importance in CDKL5-related conditions.

Note - This is quite a technical-based paper and took me several malts to get through!! It is estimated that the human body is made up of over 100,000 different proteins yet only contains some 25 - 30,000 genes. This means that on average, one gene may code for perhaps 4 - 5 proteins, and this seems to be the case with CDKL5. The authors of this paper are suggesting that they may have identified one particular form of the CDKL5 protein which is slightly shorter in length, amino acids coded in exons 19 - 21 are not incorporated, and may be the relevant form of the protein in CDKL5-related conditions. This therefore needs to be considered when screening children who present with features of CDKL5.