

Supporting CDKL5



Validation of high-resolution DNA melting analysis for mutation scanning of the CDKL5 gene:

Identification of novel mutations. *Gene* 2012.

This study from France describes the validation of a technique called high-resolution melting analysis (HRMA) used to detect genetic mutations. HRMA was developed in the US by the University of Utah in conjunction with Idaho Technology, and introduced in 2003. The authors point out that hitherto, identification of mutations in CDKL5 has been time consuming, laborious and expensive. Their study reports the validation of HRMA and the results of a comparison with another technique called denaturing high performance liquid chromatography (dHPLC). Their results showed that point mutations and small insertions and deletions can be reliably detected by HRMA, and compared to dHPLC, HRMA profiles are more discriminating. They conclude for mutation screening, HRMA appears cost-effective, easy to set up, highly sensitive, non-toxic and rapid. The technique is ideally suited for large genes with mutations located along the whole coding sequence, such as occurs with the CDKL5 gene.

Note - I read this with interest because Ellie, like many other children who were eventually diagnosed with a CDKL5 disorder, initially had a false-negative result. As technology advances we would hope that testing for all mutations becomes more sensitive and reliable, quicker and cheaper.