Variant of Rett Syndrome and CDKL5 gene: Clinical and autonomic description of 10 cases.
Neuropediatrics 2012.

This study from Italy describes features in 10 girls, 8 from Italy and 2 from Sweden, aged between 2 and 13 years. All had established CDKL5mutations (4 missense, 3 frameshift, 1 nonsense and 2 deletions). Early-onset epilepsy was the predominant feature with 9 developing seizures by the 4th month of life and one at 6 months. Speech was absent in all 10 girls, while 8 had some sort of disturbance of their vision. All 10 had experienced disturbances in their sleep patterns during the first 4 months of life and 7 continued to do so at the time of the review.

The autonomic aspect of the study focussed on breathing patterns. There are 4 recognised disorders of breathing in Rett Syndrome. Forceful breathing patterns were noted in 8 of the girls with CDKL5 while the remaining 2 girls exhibited the apneustic type.

Note - A study which again reinforces many of the features we are coming to recognise as part of the CDKL5 spectrum. The description of the breathing patterns is interesting as I have not encountered this in any other studies I have read (I may have missed something!). They also discuss other autonomic dysfunction such as cold hands and feet, all of which we recognise in Ellie.