Respiratory and sleep disorders in female children with atypical Rett syndrome caused by mutations in the CDKL5 gene.
Developmental Medicine & Child Neurology 2012.

This is a small study from the Netherlands looking at sleep and respiratory disturbances in 4 females with CDKL5 mutations. Their ages ranged from 2 to 15 years. The 3 younger children (ages 2, 4 and 5) had mutations affecting exons 5 and 9 and had severe phenotypes. The 15 year old had a mutation affecting exon 18 and a milder phenotype, being able to walk and having less severe seizure activity. The study involved the use of a questionnaire (Sleep Disturbance Scale for Children) and Polysomnography. REM sleep is thought to be the most important phase of our sleep cycle. In young children about 50% of sleep is spent in the REM phase while this drops to about 20% in adults. The 3 youngest in this study all spent less than 20% in REM with one recorded as having no REM sleep. Sleep efficiency - the percentage of time in bed spent asleep - was reduced in all 4 females - mainly due to frequent and long-lasting awakenings. Apnoea is the term for cessation of breathing, and it has a number of causes. Respiratory centres in the brain are responsible for controlling and driving the effort to breath, and central apnoea occurs when these control centres don't function properly. Two of the females, the youngest and oldest, had central apnoea, which occurred during the awake state. The authors conclude by suggesting that Polysomnography should more-or-less be mandatory for the assessment of sleep and respiratory disturbances in children with a CDKL5 disorder.

Note - Whilst we recognise the pattern of sleep disturbances described in this study (yawn...don't we just...), we have not seen respiratory disturbances in Ellie, although I think others have in their children. As the authors themselves admit, this is a very small study which also has incomplete data. It is therefore difficult to draw any wider conclusions. It does, however, establish some baseline data for children with CDKL5mutations that may be of use in later studies. Just one other thing - "atypical Rett syndrome" caused by CDKL5 mutations is now called the CDKL5 disorder.