

Supporting CDKL5



Adult Phenotypes in Angelman- and Rett- Like Syndromes. Molecular Syndromology. Published online: January 13 2012.

This paper looks at the evolution of the clinical characteristics (phenotype) in adults with various syndromes, and includes a female who was diagnosed with a [splice site mutation](#) in intron 7 of [CDKL5](#) at the age of 47. She had developed seizures after 6 months and a subsequent delay in psychomotor development. Her motor development had only been mildly delayed, but she hadn't learned to speak and she had profound intellectual disability. Her seizures had persisted despite multiple drug treatments. She was small, being at the lower end of [centile charts](#) and displayed a number of facial characteristics including prominent cheekbones, mild [prognathism](#), deep-set eyes and [blepharochalasis](#).

A number of other characteristics are also described. The authors state that this is the oldest reported patient with a [CDKL5](#) mutation. They also then review adult phenotypes reported in other published studies, which, with the adult in this study comprises a group of 10 adults with an age range from 18 to 47 years. From this review they surmise that apart from difficulty with seizure control, adults with [CDKL5](#) may also have medical problems such as secondary scoliosis, contractures, feeding difficulties and airway infections.

Note - It is probably with some caution that we should apply the findings in this relatively small group of adults to every child now growing up with [CDKL5](#), because, as many of us are aware, although our children share many characteristics, they also display variable levels of function and development. Furthermore, the fact that [CDKL5](#) is now recognised as a separate condition may improve their longer-term outcome through earlier diagnosis and subsequent improvement in treatment and care. Although factors that may influence their longer term development are still to be determined, we are sure to learn more as longitudinal studies continue.