Thyroid Function in Rett Syndrome
Hormone Research in Paediatrics 2015.

This is an interesting piece of research from Italy looking at thyroid function. Although Rett syndrome appears in the title, a number of children with CDKL5 are included in the study.

By way of introduction, the thyroid gland controls the basic metabolic rate of the body through the secretion of hormones, namely T3 and T4. T4 is converted into T3 which is the more active of the two. The thyroid is stimulated into action through the effects of another hormone imaginatively called thyroid stimulating hormone (TSH). If you have too much thyroid hormone the increased activity of your body cells or body organs can cause an increase in heart rate or activity of the intestines (hyperthyroidism) while too little hormone can cause the body to slow down causing the heart rate to become slower than normal and the intestines to work sluggishly (hypothyroidism).

In this study, the authors recruited 45 females with features of Rett syndrome. Various tests of thyroid function were measured through blood tests, including levels of TSH, free T3 and T4, and these results compared to those obtained from 146 age-matched healthy controls.

Overall mean values for free T3 were not significantly different between the study group and controls. However, the mean free T4 levels were significantly higher in the study group. Of the 45 females in the study group, 39 had MeCP2 mutations while 6 had CDKL5 mutations. The ages of the 6 with CDKL5 ranged from 2 to 12 years. Differences were noted when the results between these 2 groups were compared.

	CDKL5	MeCP2
Raised free T3	2/6 (33%)	10/39 (25.6%)
Raised free T4	3/6 (50%)	5/39 (12.8%)

Overall, proportionally more of the CDKL5 group had elevated levels of free T3 and T4 although none of them exhibited features of hyperthyroidism, perhaps because their levels of TSH were found to be within normal limits. The authors discuss the role of thyroid hormones and potential mechanisms of interaction with MeCP2 but are unable to discuss any potential significance in relation to CDKL5.

Note – an odd paper. None of the 45 girls in the study group appear to have symptoms of an overactive thyroid. This is not discussed in any great detail, so it is difficult to be sure what prompted the study and what the significance of the raised hormone levels is. Some of the group with MeCP2 mutations are said to have subclinical hypothyroidism so that makes even less sense..! However, although the numbers in this study are small – so we have to be very cautious about the differences seen - this is one of the first, if not the first study looking at hormone levels in CDKL5, and the abnormalities identified might just be the tip of a hormonal iceberg.