Isolated TSH deficiency in a patient with Myotonic Dystrophy

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Myotonic dystrophy (MD) is a systemic genetic disorder predominantly affecting musculature. Although various endocrine disturbances have been described, data relating to thyroid and function are sparse. Isolated TSH (thyroid stimulating hormone) deficiency is uncommon and about 60 cases have been reported in the literature. However, isolated TSH deficiency in MD is very rare and described in only a handful of cases. We report a case of myotonic dystrophy presenting with secondary hypothyroidism.

Mrs AW is a 39 year old lady with myotonic dystrophy presented with tiredness, low energy, dry skin and weight gain in early 2008. The thyroid function test done by the GP showed both TSH and FreeT4 (FT4) were low at 0.33 mU/L (0.35 - 5.50) and 10.6 pmol/l (11.50 - 22.70) respectively. She was started on thyroxine 50 mcg a day by the GP and referred to endocrine clinic.

Past medical history includes hysterectomy with bilateral salpingo-oopherectomy in 1999 for ovarian cysts and endometriosis. Due to progressive swallowing difficulties as a result of myotonic dystrophy she has been on a PEG feeding since 2000. Her current medications are Progynova TS oestrogen patch and Fluoxetine 20 mg once a day.

Her subsequent TSH was persistently low 0.26 with a low FT4 of 9.7 and FreeT3 2.8 pmol/l (3.50 - 6.50). Assessment of rest of pituitary function showed, a high LH and FSH at 26.6 and 23.7 respectively, prolactin was normal - 168, IGF-1 was normal - 17.2 nmol/L (9.50 - 45.00). Short synacthen test was also normal. The serum was also analysed in a different laboratory which ruled out any assay interference. TRH stimulation test revealed a subnormal TSH response; [TSH - 0.68 (0 min), 3.1 (20 min) and 2.29 (60 min)]. A MRI pituitary did not show any pituitary lesion.

She felt very unwell when she came off her thyroxine for 3 weeks when awaiting TRH test. She was recommenced on 50 mcg of thyroxine. She felt significantly better remains well since initiation of thyroxine replacement.

Isolated thyroid stimulating hormone deficiency causing central hypothyroidism in patients with myotonic dystrophy though known is rare. MD is characterised by abnormalities in the dystrophia myotonica protein kinase (DMPK) gene. The defective protein encoded by the DMPK gene is thought to act differentially on hypothalamus-pituitary-endocrine organ axis, resulting in various endocrine abnormalities seen in this condition.