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CASE OF AUTOSOMAL DOMINANT HYPOCALCAEMIA

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We are presenting a 40 years old man with history of asymptomatic hypocalcaemia which was found during routine testing , calcium level 1.97 mmol/l, phosphate 0.94 mmol/l , PTH of 1.4 pmol/l (1.3 -9.4) and vitamin D of 27 ng/l , 24 urinary calcium of 4.6 mmol/l (2.5- 7.5) with urine volume of 2.5 L. The reason for low calcium was not clear ,but during the course of the investigation patient told us that his daughter who is 10 years old also found to have hypocalcaemia this together with the inappropriate low normal PTH low trigger the possibility of likely genetic cause of the hypocalcaemia this was confirmed with the presence of CaSR mutation, and the diagnosis of autosomal dominant hypocalcaemia was established. Prevalence of autosomal dominant hypocalcemia is unknown. The condition is likely underdiagnosed because it often associated with no signs or symptoms; however patient can presented with severe symptomatic hypocalcemia and seizures. Probably in the absence of stigmata of autoimmune hyperparathyroidism genetic causes should be investigated even without significant family history which is sometimes difficult to elicit. The condition usually doesn't require treatment, this patient was initially treated with calcium and vitamin D but this was subsequently stopped as the urinary calcium started to rise, calcium level remains within accepted level.

Biography

Dr Ali Ahmed is final year specialist registrar at Hull royal infirmary, Dr Najeeb Shah is specialist registrar in endocrinology , Dr Kmarudeen Mohammed is senior endocrinology and honorary lecturer at HYMS university , he also the programmer director of the endocrinology training at Yorkshire deanery in UK .

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