Primary Empty Sella-Is it familial??

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Background

Empty sella syndrome is a condition in which sella turcica is partially or completely filled with CSF resulting in a displacement of normal pituitary gland. Primary empty sella is due to inherent weakness of diaphragm sella or to an increase in intracranial pressure which promotes herniation of arachnoid membranes in to pituitary fossa. More common in middle aged obese female and headache is the most common presenting symptom. It may be associated with one or more pituitary hormone deficiency.

There are a few cases of familial association in the empty sella has been described in the literature. We would like to present cases of mother and daughter who presented with primary empty sella.

Case history

49 years old lady with a background of type 2 diabetes, obstructive sleep apnoea, asthma, Osteoarthritis, obesity presented to neurologists in 2009 with a history of recurrent headaches, no history of head injury. In her family there was a history of strokes and migraine. Neurological examination was normal. MRI of the brain revealed primary empty sella. A GnRH test revealed normal gonadotropin axis. Insulin tolerance test has revealed growth hormone deficiency. She has been commenced on growth hormone replacement. 6 years after commencing the growth hormone replacement, a glucogon stress test has revealed a subnormal cortisol response.

34 year old daughter (a neuropsychologist) of the above lady presented to the endocrine services with a history of weight gain, hair loss. She had a background of renal stones, endometrial polyp. Partial empty sella was diagnosed on MRI done when she participated in research. Pituitary dynamic function test have revealed growth hormone deficiency with normal hypothalamo-pituitary-adrenal axis and gonadotropin axis.