A Patient With Multiple Endocrine Neoplasia Type 1 Presented With Precocious Puberty

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Background:
Multiple Endocrine Neoplasia Type 1 (MEN1) is an autosomally transmitted hyperplastic or neoplastic disorders of some endocrine and non-endocrine organs. Pituitary tumors develop in 30-70% of patients with MEN1. Mean age at onset of MEN1 associated pituitary tumors is the 4th decade. It is very rare before and during puberty. Although there are two case reports about MEN1 and delay puberty , early and rapidly progressive puberty with MEN1 has not been reported yet.

Case:
Eight year and 5 month old girl whose father has MEN1, applied with pubic and axillary hair that were detected 10 months ago. At that time, her tanner stage was 2. The patient was diagnosed as central precocious puberty as a result of LHRH test **(Table 1).** Bone age was advanced (2.5 years more than chronological age). Her predicted adult height was calculated considerably shorter than her target height and GnRH analogue treatment was given to patient. In pituitary MR, hypointense region which was stable in size during the follow up, was seen in intermediate lob but it was not interpreted in favor of the adenoma. The biochemical tests at admission are given in **Table 2**. Serum calcium level was high whereas serum phosphorus, PTH and 25-OH vit D levels were normal. PTH increased and hyperparathyroidism was detected during follow up. The ultrasonography (USG) also showed adenomas in parathyroid gland and patient was diagnosed as MEN1 same as her father. Two parathyroid adenomas with diameter 3.6 mm and 5.9 mm respectively were detected by ultrasonography. Familial pedigree is given in **Figure 1.** Her father (case 3 in figure) has a heterozygous mutation c.1699_1671delinsC in the exon 10 of MEN1 gene. Our patient molecular analysis is stil studying. Since growth velocity decreased during the GnRH analogue treatment, growth hormone (GH) treatment was also started . The last assessment was done at 11 year and 9 month old. The GnRH analogue treatment was stopped where the GH treatment had been continued for 8 months more. The serum calcium levels were between 10.4 to 10.9 whereas PTH levels decreased to normal range during the followup. Urine calcium/creatinin was normal. In abdominal USG, nephrolithiasis has not been observed and pancreas, surrenal glands were also detected normal. Other pituitary hormones, blood glucose remained in the normal range. She has not needed any medical treatment for hypercalsemia. The hypointense region in pituitary MRI has been stable during the yearly control MRIs (4 years).

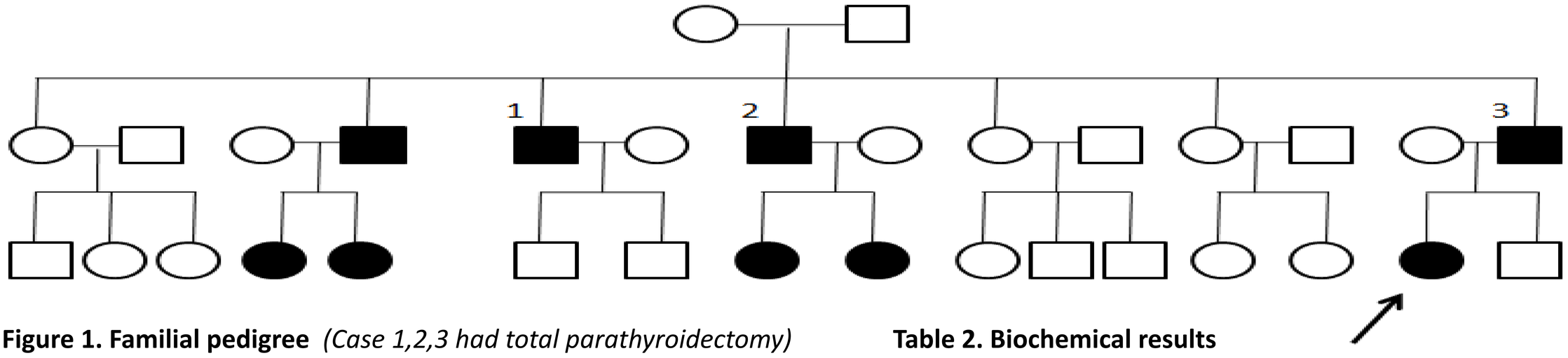

Figure 1. Familial pedigree *(Case 1,2,3 had total parathyroidectomy)*

Table 1. LHRH test results

Laboratory		
LHRH Test	FSH(mIu/ml)	LH(mIu/ml)
0	4.6	0.5
30	10.3	5.6
60	12	4.3
90	12.5	2.2

Table 2. Biochemical results

Laboratory	
Glucose (mg/dL)	77
Ca (mg/dL)	10.7↑
P (mg/dL)	4.03
ALP (U/L)	284
PTH (pg/mL)	68.78↑
1-25 OH(nmol/L)	117
25-OH (pmol/L)	19.2
PRL (ng/dl)	5.71
Insulin (uIU/mL)	15.22
TSH (uIU/mL)	1.91
FT4 (ng/dL)	1.49
Cortisol (ug/dL)	9.8
ACTH (pg/mL)	5.2
IGF-1 (ng/mL)	780
IGFBP3 (ug/mL)	3.6

Conclusion:
This case emphasize relevance of early screening of endocrine disorders for members of families with MEN1 because of diversity of endocrine disorders and also it should be kept in mind that rare endocrine presentations as precocious puberty can also be detected in the follow up of patients with MEN1.

References
1. Giri D, McKay V, Weber A, Blair JC. **Multiple endocrine neoplasia syndromes 1 and 2: manifestations and management in childhood and adolescence.** Arch Dis Child. 2015 Oct;100(10):994-9.
2. Walls GV. **Multiple endocrine neoplasia (MEN) syndromes..**Semin Pediatr Surg. 2014 Apr;23(2):96-101.

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