

## Discordant Calcium and Parathyroid Hormone with Presumed Epileptic Seizures

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### CASE DESCRIPTION

A 33-year-old woman presented for the first time at the age of 9 years with recurrent seizures and was subsequently treated for epilepsy for 2 years. At 11 years of age, it was noted that the patient had hypocalcemia with an increased parathyroid hormone (PTH)<sup>4</sup> measured with use of an intact PTH assay. Serum calcium was 5.73 mg/dL [1.43 mmol/L; reference interval (RI) 8.82–10.42 mg/dL (2.2–2.6 mmol/L)]; serum phosphate was 10.42 mg/dL [3.36 mmol/L; RI 1.86–4.34 mg/dL (0.6–1.4 mmol/L)]; and PTH was 319.68 pg/mL [33.9 pmol/L; RI 8.49–68.84 pg/mL (0.9–7.3 mmol/L)]. Vitamin D (Total 25-OH; 25-OH D2 and D3) concentrations, magnesium concentrations, liver and renal function tests were all normal. No other endocrine abnormalities such as thyroid or gonadotropin resistance were detected. The patient also was of short stature and was overweight. However, she became overweight after the age of 13 years (body mass index at 13 years was 22.8 kg/m<sup>2</sup>), and the short stature was not significant, as she was at the lower end of the RI for height at 1.55 m. There was no cognitive impairment. Positive Chvostek sign was observed but not Trousseau sign.

The patient was placed on active vitamin D (alfacalcidol) and calcium supplements. The patient was followed up for 13 years; selected laboratory results are presented in Table 1. Although the patient was managed on calcium supplements and active vitamin D and followed up at a tertiary endocrine clinic, serum phosphate concentrations remained persistently high (Table 1). This was attributed to intermittent noncompliance and unavailability of active vitamin D. The patient had also been prescribed calcium carbonate as a phosphate binder.

QUESTIONS TO CONSIDER
• What are the causes of hypocalcemia with an increased PTH concentration?
• What is the explanation for the persistently increased PTH concentrations?
• What is the role, if any, of molecular testing in a case like this?

Dates	Calcium (mg/dL)	Phosphate (mg/dL)	Intact PTH (pg/mL)
11/18/2004	7.57 (8.82–10.22)	4.58 (2.70–4.50)	394 (12.3–65.1)
03/10/2005	7.45 (8.82–10.22)	5.61 (2.70–4.50)	358 (12.3–65.1)
06/30/2005	Ionized 3.29 (4.57–5.17)	6.48 (2.48–4.34)	330 (16.0–86.8)
11/17/2005	6.85 (8.22–10.26)	5.67 (2.48–4.34)	225 (16.0–86.8)
06/22/2006	7.82 (8.22–10.26)	4.90 (2.48–4.34)	303 (16.0–86.8)
11/30/2006	8.78 (8.22–10.26)	4.65 (2.48–4.34)	335 (16.0–86.8)
11/06/2008	7.94 (8.22–10.26)	4.74 (2.48–4.34)	300 (16.0–86.8)
09/02/2015	6.25 (8.62–10.02)	6.54 (2.42–4.40)	179 (12.3–87.7)

<sup>a</sup> Values in the table are reported as concentrations (reference interval) expressed in traditional mass units. Conversion factors to standard international units: PTH pg/mL to pmol/L = 0.106X; calcium mg/dL to mmol/L = 0.250X; phosphate mg/dL to mmol/L = 0.323X.

### Final Publication and Comments

The final published version with discussion and comments from the experts will appear in the March 2018 issue of *Clinical Chemistry*. To view the case and comments online, go to <http://www.clinchem.org/content/vol64/issue3> and follow the link to the Clinical Case Study and Commentaries.

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