

9 months, his motor and mental development were noted to be delayed. Array comparative genomic hybridization analysis was planned to show Xp21 deletion syndrome.

Conclusion: Serum CPK and TG levels should be measured in all male patients who present with an adrenal hypoplasia. These simple tests may help early diagnosis and appropriate genetic counseling for next pregnancy.

Pseudohypoparathyroidism Type 1a: A Case Report

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Objective: Pseudohypoparathyroidism (PHP) is a group of disorders characterized by end-organ resistance to the parathyroid hormone (PTH). PHP type 1a includes multi-hormone resistance syndrome, Albright's hereditary osteodystrophy, and obesity and is caused by mutations in GNAS exon 1 through 13. Characteristic features of disease are hypocalcemia, hyperphosphatemia, elevated PTH, obesity, round facies, and subcutaneous calcification. The disease is inherited from affected mother. On the other hand, pseudopseudohypoparathyroidism (PPHP) occurs if the mutation is paternally inherited. In PPHP, calcium and phosphorus levels are generally normal. In this report, we want to present a boy with PHP type 1a who has normal calcium and elevated thyroid stimulating hormone (TSH) levels, which is a rare event.

Case: The 12-10/12-year-old boy was admitted with the complaint of short stature. On physical examination, brachydactyly, round facies, and short neck were observed as well, indicating PHP 1a. However, serum calcium, phosphorus, alkaline phosphatase, vitamin D, and PTH levels were normal. These results were compatible with PPHP. On the other hand, TSH levels were found to be high (8 µIU/mL, normal range 0.5-4.8 µIU/mL), free thyroxine levels slightly low (0.7 ng/dL, normal range 0.8-2.3 ng/dL), urine iodine level normal, and thyroid antibodies to be negative. These features were compatible with PHP type 1a. Therefore, genetic analyses were performed and p.D826H (C2476G>C) heterozygous mutation was found in GNAS. The genetic analyses of parents revealed maternal inheritance. As far as we know, this mutation was not reported before and was found to be high risky for being a cause of the disease according to mutation taster and human splicing finder.

Conclusion: In this report, we want to emphasize that normocalcemia can be a finding of PHP type 1a.