Treatment of Primary Hyperparathyroidism in the Setting of Tatton-Brown-Rahman Syndrome

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BACKGROUND

Primary hyperparathyroidism (PH) is a common clinical diagnosis encountered by physicians specializing in the treatment of endocrine disorders. In rare cases, PH may be attributed to Tatton-Brown-Rahman syndrome (TBRS). TBRS is a rare overgrowth syndrome that is caused by de novo germline mutations in DNA cytosine methyltransferase 3A (DNMT3A) and is characterized by a distinctive facial appearance, tall stature, and intellectual disability.[1] There have been very few published descriptions of the treatment of PH in patients with TBRS.

METHODS

We describe the treatment of primary hyperparathyroidism in a 17-year-old male with TBRS who had previously undergone unsuccessful surgical exploration.

RESULTS

Our patient with PH was referred to our surgical oncology clinic for a parathyroidectomy evaluation. The patient had previously undergone a bilateral neck exploration (BNE), in which no parathyroid tissue was identified. A sestamibi scan was completed and showed increase uptake in the left mid upper thyroid lobe, concerning for a parathyroid adenoma (Figure 1). Computerized tomography scan and ultrasound of the neck were unable to be completed due to the patient's developmental delay and inability to cooperate. The patient was taken back to the operating room for a repeat BNE.

The left inferior and superior poles of the thyroid were explored first with no lesion identified. Next the tracheoesophageal groove, carotid sheath, thyrothymic ligament, and thymus were all explored on the left side. With no lesion identified, focus was shifted to the right side, where all areas were explored with no lesions identified. Blood was then drawn from the left and right internal jugular veins and sent for parathyroid hormone (PTH) levels. Results showed a slightly higher PTH level from the right internal jugular vein; thus, right sided neck dissection was continued. Tracing the recurrent laryngeal nerve inferior, an ectopic parathyroid adenoma was identified in the inferior portion of the upper mediastinum on the right side and resected. The patient had an appropriate drop in his PTH level after excision and has been seen back in clinic with normalization of his PTH and calcium levels.

CONCLUSION

With TBRS first being identified in 2014 and only about 200 individuals diagnosed, there is still minimal literature describing the congenital abnormalities and clinical features associated with this syndrome.[1,2] This case represents the importance of obtaining multiple imaging modalities during preoperative preparation for a parathyroidectomy in patients with TBRS. Additionally, it validates the importance of BNE in patients with TBRS, even with sestamibi positive imaging. Lastly, we advocate for further publications of anatomic anomalies associated with PH in patients with TBRS in the future.

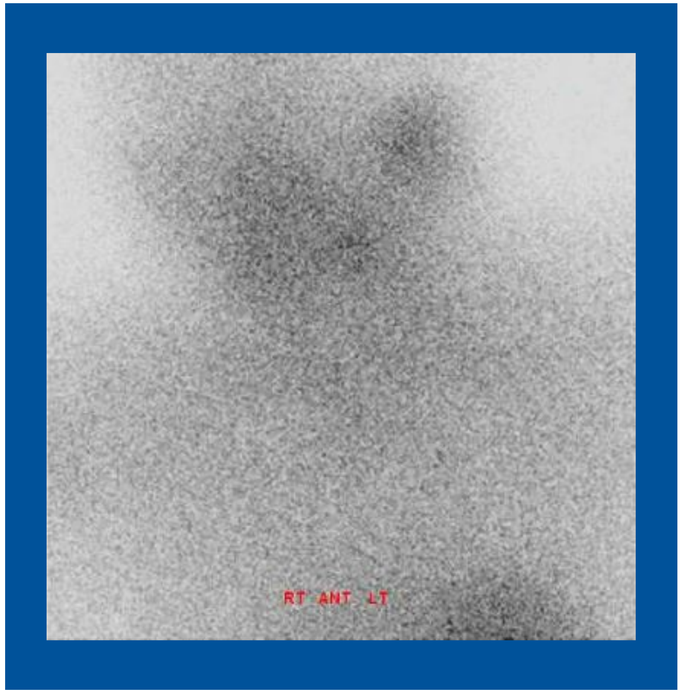

Figure 1: Delayed planar technetium 99m sestamibi image demonstrating faint persistent uptake over the left mid thyroid gland.

REFERENCES

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