Unilateral Thyroid Agenesis—Curiosity or Predictor of Future Pathology?

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Abstract

Unilateral thyroid agenesis (thyroid hemiagenesis) is a rare disorder with less than 300 cases currently in the world literature. The current clinical importance of this condition lies in the possibility that such patients may be at an increased risk for a wide range of future pathology in the contralateral lobe. If this indeed proves to be the case in future, these patients need to be identified.

A case of incidentally discovered unilateral thyroid agenesis is presented and the clinical importance discussed.

Keywords: Thyroid; Agenesis; Hemiagenesis

Introduction

Unilateral thyroid agenesis (thyroid hemiagenesis) is a disorder where one lobe of the thyroid gland fails to develop normally and this is believed to be the rarest of all the developmental thyroid anomalies [1].

The first reports of this entity appeared in 1866 by Handfield-Jones, in 1876 by Luschka and in 1886 by Ehlers [2]. Structural, congenital thyroid anomalies can be caused by either abnormal genesis or abnormal descent of part of the thyroid gland [2]. Thyroid hemiagenesis is a rare condition with only about 270 cases reported in the world literature up to 2003 [2] and still only about 300 cases, most as case studies by the year 2010 [3].

Here, a young male patient with some unusual features of thyroid hemiagenesis is presented and discussed.

Case Report

A 21-year old Caucasian male was referred for a comprehensive examination due to the presence of asymptomatic, microscopic hematuria. The clinical examination was unremarkable with a blood pressure of 110/70 mmHg and no abnormalities noted on repeat urinalysis. Both his echocardiogram and renal ultrasound study was perfectly normal.

During thyroid ultrasonography complete absence of the isthmus and a severely hypoplastic left sided thyroid lobe was noted (Figures 1 and 2) with a normal right sided lobe (Figure 3). No clinical stigmata of thyroid disease was present. Biochemically he was in an euthyroid state (TSH=2.97 uIU/mL and free T4=15 pmol/L) and no antithyroid antibodies could be detected. The possible clinical implications and uncertainties related to this diagnosis will be discussed.

Discussion

There are several aspects of this case that merits discussion. Unilateral agenesis of a thyroid lobe is a rare congenital anomaly which affects women three times more commonly than men and in 80% of cases the left thyroid lobe will be the one affected [4]. Furthermore, in these cases of left thyroid lobe hemiagenesis about 50% will also have an absent thyroid isthmus [4]. In this young male the isthmus is completely absent (Figure 1), however some rudimentary tissue of the left lobe can be seen in Figures 1 and 2.

There are two clinical questions relating to unilateral thyroid agenesis:

The true incidence of this condition is unknown and probably completely underestimated, as it is usually an incidental discovery when the patient’s thyroid is examined due to the presence of pathology in the remaining lobe [5]. A vast array of pathologies have been described in the contralateral lobe and these include: carcinoma [6], subacute and chronic thyroiditis [7], Graves disease with ophthalmopathy [8], adenoma [5], multinodular goiter [5] and both hypo-and hyperthyroidism [5].
This of course leads to the second and most important clinical question: Does the presence of thyroid hemiagenesis predispose the afflicted patient to disease and malignancy in the remaining lobe? At present, this is not known, but it is a valid and concerning issue that needs to be clarified. Another valid clinical question emanating from this case is why a thyroid ultrasonographic examination was performed on a young man with hematuria? Kocak et al. [9] has shown a significant association between renal glomerular disease and autoimmune thyroid disease, thus the ultrasonographic examination in this patient referred with microscopical hematuria.

Conclusion

In conclusion, a case of a healthy 21-year old Caucasian male with incidentally discovered unilateral thyroid agenesis (thyroid hemiagenesis) is presented. Based on current literature, there is a real possibility that these patients may be at an increased risk for malignancy and/or functional disturbances in the remaining lobe. This important clinical question can only be answered by a properly powered study which will need a large population of screened cases with a proper duration of follow-up.

References


Figure 2: Rudimentary left-sided thyroid lobe.

Figure 3: Normal right-sided thyroid lobe.