Case report

Bilobar agenesis of thyroid gland with hypertrophied isthmus

P. Sharma,[1] B. Karki, [2] P. Gautam Ghimire,[3] M. Shrestha,[2] A. Ghimire,[1] A. Pathak [2]

[1] Department of Radiology and Imaging, [2]Department of Surgery, [3]Department of Pathology, Nepalgunj Medical College Teaching Hospital, Kohalpur, Banke, Nepal

***Correspondence to**:* Dr. Prakash Sharma, Department of Radiology and Imaging, Nepalgunj Medical College Teaching Hospital, Banke, Nepal

Email: prakashshrm@ yahoo.com

Abstract

Bilobar agenesis of thyroid gland is a rare presentation of thyroid gland. Here we report an 18 years male presenting with history of midline swelling in the neck. Ultrasonography and CT neck revealed bilateral agenesis of thyroid gland with hypertrophied isthmus.

Keywords: Bilobar agenesis of thyroid, Hypertrophied Isthmus

Introduction

Thyroid gland is the first endocrine gland to develop in the embryo. The thyroid gland appears as an epithelial proliferation in the floor of the pharynx between the tuberculum impar and the copula at a point later indicated by the foramen cecum.[1]Subsequently, the thyroid descends as a bilobed diverticulum. It is well known for its developmental anomalies ranging from common to rare. Persistence of pyramidal lobe and thyroglossal duct cyst are the common anomalies whereas other rare anomalies are agenesis or hemi-agenesis of thyroid gland, agenesis of isthmus alone or aberrant thyroid glands.[1-3] Only one case of bilobar agenesis is described so far. Exact cause of bilobar agenesis is not well known. Here we report a case of agenesis of bilateral lobes of thyroid with hypertrophied isthmus presenting as a midline swelling in the neck.

Case report

Eighteen year male presented to surgical OPDS with history of midline swelling in the neck for two years with no clinical features of hypo- or hyperthyroidism. Systemic examination was within normal limit. Local examinations revealed well defined non tender firm mass about 4 x 3 cm in the midline of neck which moves upward with deglutition but no movement during protrusion of tongue. No cervical lymphadenopathy was found. High resolution ultrasonography of neck revealed hypertrophied isthmus with agenesis of both thyroid lobes (Fig. 1). CT scan of neck also showed similar findings (Fig. 2). FNAC from the mass showed thyroid follicles (Fig. 3). Thyroid function test was within normal limit. The patient is kept under regular follow up as no intervention was carried out.

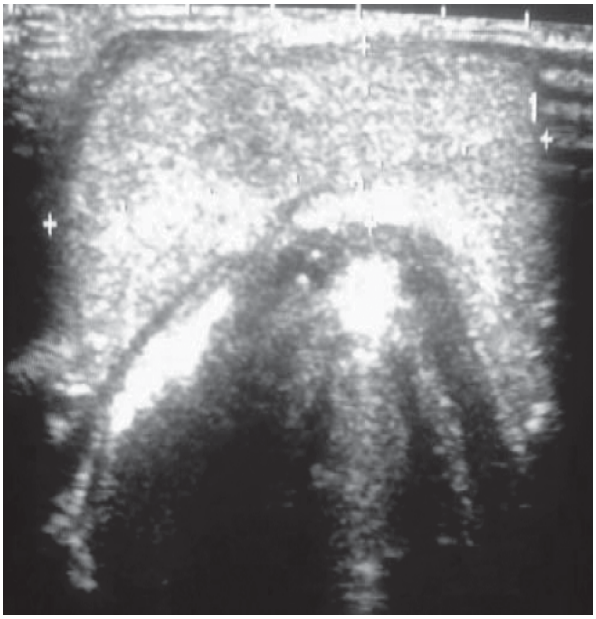

Fig. 1: High resolution ultrasound showing enlarged isthmus of thyroid with agenesis of both the lobes.

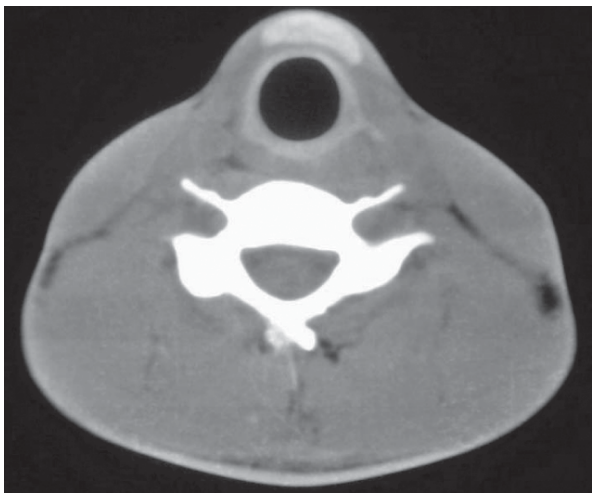

Fig. 2: CT scan of neck showing agenesis of both the lobes with hyperplastic isthmus.

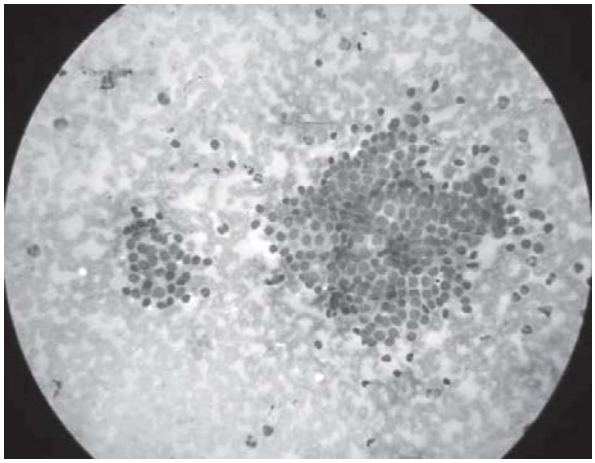

Fig. 3: Fine needle aspiration cytology showing thyroid follicular cells.

Discussion

Normal thyroid descends and develops as a bilobed diverticulum. Failure in such development leads to agenesis of the thyroid. In our case, there was agenesis of both lobes of the thyroid gland with hypertrophied isthmus.

Persistent growth stimulation causes diffuse hyperplasia in pubery.[4] In our case, since there was bilobar agenesis, whole of the functional activity was taken up by the isthmus resulting in its hypertrophy during the puberty. Attipou K et al reported a case of a 27-year-old female patient presenting with bilateral agenesis of the thyroid gland and a benign euthyroid adenoma of the isthmus and the pyramidal lobe.[5] In our case beside hypertrophy there was no focal lesion within the isthmus.

Such condition can present as a midline swelling in the neck which moves with deglutition. They can cause problem in diagnosis and further management. In such cases ultrasound scan can prove as useful and cheap modality. CT scan, although routinely not required, can further aid in the diagnosis. In our case both USG and CT scan was done which helped in the diagnosis of such rare case. Radionuclide scanning should be performed to know about the distribution of functional activity.[4] It was not done in our case due to the unavailability of such facility.

Management of such case can create problems. Simple excision can inadvertently lead the patient to become hypothyroid. Treatment is administration of exogenous thyroid hormone to suppress thyroid stimulating hormone and radioactive iodine ablation followed by hormone replacement.[6]

In our case the patient was euthyroid with no other complaints apart from the swelling for which he was not concerned about the cosmesis. So he is kept in regular follow up.

Conclusion

Bilobar agenesis of thyroid gland is a rare congenital condition which can be diagnosed by high resolution ultrasound.

References

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