CASE REPORT

Familial Occurrence of Thyroglossal Duct Cyst

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Abstract Thyroglossal duct cysts are one of the commonest benign midline neck swellings. These are the remnants of the embryologic duct that fails to regress by the 10th week of fetal development. Generally, these are sporadic, slowly enlarging midline neck lumps, with half of them presenting before the age of 30. However, despite their relative frequency, reports of familial inheritance are rare. A review of literature reveals a total of 25 patients from 8 families showing a familial inheritance pattern worldwide. We describe here a 9th family with 5 affected patients taking the total number of cases to 30. Also, this is the 1st such reported case of familial inheritance of thyroglossal cyst from the Asian continent.

Keywords Thyroglossal · Cyst · Familial · Occurrence

Introduction

Thyroglossal duct cysts are one of the commonest benign midline neck swellings. These are the remnants of the embryologic duct that fails to regress by the 10th week of fetal

development. Review of literature reveals a total of 25 patients from 8 families till now showing a familial inheritance pattern worldwide. We are describing the 9th family with 5 affected patients taking the total number of cases to 30. Also, this is the 1st such reported case from the Asian continent.

Case Report

Case 1 A 9-year-old girl presented with an asymptomatic congenital midline neck swelling, located just below the hyoid bone, measuring 2 cm × 2 cm, moving with deglutition and with protrusion of tongue. History was not suggestive of any infection or any thyroid gland abnormality. A Sistrunk operation was performed and histopathology revealed features of a thyroglossal cyst.

Case 2 During the hospital stay of case 1, it was observed that her 42-year-old father had a similar asymptomatic midline neck swelling measuring 3 cm × 3 cm. Sistrunk operation was performed on him as well and the histopathology revealed features of thyroglossal cyst along with focal collection of thyroid follicles. Both cases 1 and 2 recovered uneventfully and did not showed any signs of recurrence till 1 year of follow-up.

Case 3, Case 4, and Case 5 On elicitation of detailed history, it was found that the mother of case 2 also had a similar midline swelling for which she underwent surgery. Cases 3 and 4 were the maternal uncle and maternal uncle's grandson of case 2, respectively (refer no. 8 of Fig. 1). Additionally, the elder sister of case 2 was a known case of hypothyroidism.

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Fig. 1 Pedigrees of familial occurrences of thyroglossal cyst (all published cases). Other thyroid associated diseases are indicated by an *asterisk*. AR autosomal recessive. 1 Ashworth JT [1], 2 Millikan JS et al. [2], 3 Issa MM, De Vries P-AR [3], 4a Klin et al. family1-AR, 4b Klin et al. family 2 [4], 5 Castillo TS, Castillo P [5], 6 Greinwald JH et al. [6], 7 Schader I et al. [7], and 8 present case

Discussion

Out of 25 cases reported till now, 14 are from the USA, 3 Moslem Arabs, 2 Jews, 2 Hispanics, and 4 from New Zealand. We are presenting five cases of Asian origin. Although a definitive pattern of inheritance is yet to be established, most of these cases have exhibited an autosomal dominant pattern with incomplete penetrance and only two families (no. 3 and 4a of Fig. 1) seem to be autosomal recessive. In Issa et al. case (no. 3 of Fig. 1), male to male transmission rules out X linked transmission of the disease. Millikan et al. case (no. 2 of Fig. 1), though basically autosomal dominant one, but environmental and nutritional influence may be associated. In our case (no. 8 of Fig. 1), amongst the five members, the sister, her son, and her granddaughter, all affected are consistent with autosomal dominant mode of inheritance whereas the brother and his grandson being affected are more consistent with autosomal dominant mode of inheritance with incomplete penetrance.

Of all the reported cases till now, associated thyroid abnormalities have been found only in the families studied by Millikan et al. and Issa et al. Millikan et al. (no. 2 of Fig. 1) reported one case with hypothyroidism and one with anaplastic thyroid carcinoma. Issa et al. reported maternal grandmother of patient with autoimmune thyroiditis (Hashimoto's

thyroiditis) and maternal grandfather with anaplastic carcinoma of thyroid. In our study, one of the sisters of case 2 was hypothyroid. Issa et al. documented the occurrence of ectopic thyroid tissue in the thyroglossal duct cyst as was found in case 2 in our study. Normally, 10% of ectopic thyroid tissue is found in thyroglossal cyst. Out of total 30 cases, 10 are male and 20 are female indicating female preponderance though in our study males are more affected (3:2). In sporadic cases, male preponderance is slightly greater. Age of manifestation in sporadic cases is usually less than 30 years but in familial cases no definite age of manifestation is mentioned. In our study, it was less than 40 years. We observed first child of younger generation to be affected whereas third, fourth, or fifth child was affected in older generation. Literature has not shown any relation of thyroglossal cyst with birth order. Consanguineous marriage is reported in only one family studied by Klin et al. (no. 4a and b of Fig. 1). In our study, no consanguineous marriage was present.

Conclusion

Familial occurrence of thyroglossal duct cyst though rare but is very much under reported worldwide more so in the Asian

continent. Secondly, its correlation with age of manifestation, gender, geographical distribution, and association with other thyroid anomalies is yet to be established. So, we suggest that a detailed family history of the patients presenting with thyroglossal duct cyst should always be elicited taking into consideration all these aspects to reveal other such hidden cases in the family or in nearby blood relations with reporting of each and every case and then only we will be able to comment on its actual incidence worldwide along with its association with other factors.

Compliance with Ethical Standards

Conflict of Interest The authors declare that they have no conflict of interest.

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