

Three Cases of Familial Papillary Carcinoma of Thyroid in a Family and its Management in Rural Hospital

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Abstract: Papillary thyroid cancer (PTC) is one of the most common thyroid malignancies, with an increase in incidence rates over the past few decades. The exact cause of thyroid cancer in most patients is still unclear but the possibility of genetic predisposition to PTC cannot be overlooked. The literature review on familial papillary thyroid carcinoma was done. Its occurrence at an advanced stage in closely related individuals in early life, suggests that underlying genetic factors may predispose this malignancy. We had speculated that some gene-related factor might play an important role in familial occurrence PTC. This report describes three sisters of first degree relation with papillary thyroid carcinoma.

Keywords: Papillary thyroid cancer (PTC), Genetic predisposition, three sisters.

INTRODUCTION

Papillary thyroid cancer (PTC) is one of the most common thyroid malignancies, with an increase in incidence rates over the past few decades [1]. The exact cause of thyroid cancer is still unclear, but the possibility of genetic predisposition to PTC cannot be overlooked [2]. Here, we report three cases of papillary carcinoma of thyroid in three 1st degree relative sisters of PTC and the management in rural hospital setup.



Fig. 1: Three sisters

CASE REPORT

A 35 year old female came our surgical opd with complaints of swelling in the neck which lasted for 3years. The patient didn't have any complaints of hyper or hypothyroid symptoms, no features initially suggestive of any malignancy it was noticed that the patient who had come with her sister had a similar neck

swelling on further investigating into their familial history it was found out that she had another sister who also had a similar complaints, after the first sister who came initially was admitted the other sisters were also asked to come to our hospital and get admitted. Once all the three sisters were admitted in our ward we began investigating them in detail. Written informed consent obtained from patient.

On examination

Two of the sisters had encapsulated nodule on the right lobe and a hard, ill-defined nodule on the left lobe of the thyroid and the third sister had left sided diffuse enlargement of the thyroid.

Hematological workup came uneventful for all three patients, which included basic blood and urine routine, liver and renal function tests and the thyroid function tests.

Ultrasound of the neck didn't show any obvious signs of malignancy and there was no evidence of any neck nodal metastasis.

But FNAC of the swelling of the entire three patient showed features suggestive of papillary carcinoma of the thyroid. So a working diagnosis of familial papillary carcinoma of thyroid was made and the first sister i.e. the patient with the largest swelling was initially operated on and total thyroidectomy was done under GA and the sample sent for

histopathological studies. The histopathological examination revealed Follicular variant of papillary carcinoma of the thyroid. Later the other two sisters were also operated on, total thyroidectomy was done. Similar to the previous case these two HPE also came as Follicular variant of papillary carcinoma of thyroid.



Fig. 2: Excised mass after thyroidectomy

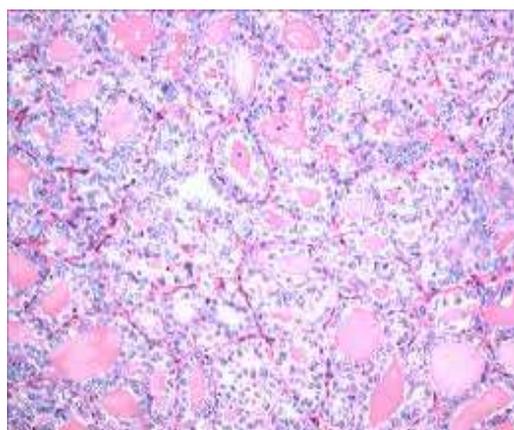


Fig. 3: Histopathological examination

Post op days uneventful and the patients were discharged on the 10th day. They were asked to follow up in opd after 1 week and the patients dint follow through.

DISCUSSION

The literature on familial papillary thyroid carcinoma was reviewed. The occurrence PTC at an advanced stage in closely related individuals in early life, suggests that underlying genetic factors may be responsible for predisposing this malignancy [3]. In 1967, Smith first discussed PTC in a family. He felt it was coincidental to familial polyposis of the colon. There may be two groups of familial papillary carcinomas. In one group multiple polyposis is associated with papillary carcinoma, while in the other familial papillary carcinoma is free of accompanying disorders [4].

Familial PTC may have a hereditary basis, independent of its association with the syndromes of multiple polyposis and of multiple hamartomas. Thus, it may represent a new entity with characteristics; distinguish it as a distinct subset of the more common disease [3]. Studies about modern genetic linkage have shown that familial PTC is genetically distinct from familial adenomatous polyposis coli [5].

Fearon ER reported that more than 20 different hereditary cancer syndromes have been defined and are attributed to specific germline mutations in various inherited cancer genes [6]. The majorities of mutations in cancer is somatic and are found only in individual cancer cells [7]. Somatic mutations in sporadic cancers frequently alter the inherited cancer genes. Study of the affected genes illuminated the functions of cell signalling pathways [6]. Although rare, the inherited cancer syndromes are of vast biological importance [7]. Studies of the specific mutations responsible for these syndromes and the cellular signalling pathways disrupted by the mutant proteins have begun to provide unprecedented insights into the molecular origins and pathogenesis of inherited and sporadic forms of cancer [8]. The proteins encoded by inherited cancer genes have been implicated in a diverse array of cellular processes that include proliferation, differentiation, apoptosis, and maintenance of genomic integrity [6].

Exposure to ionizing radiation is associated with papillary carcinomas and RET activation has been suggested in relation to it [9]. The RET proto-oncogene encodes a protein receptor tyrosine kinase. Mutations of RET are associated with dominantly inherited cancer syndromes multiple endocrine neoplasia (MEN), familial medullary thyroid carcinoma and also, with sporadic thyroid carcinomas [10]. Molecular characterization of specific genetic lesion could provide information regarding the association between the ionizing radiation and the RET oncogene activation [9]. The RET staining could be a useful marker for papillary thyroid carcinoma [11].

CONCLUSION

We speculate that some gene-related factor might play an important role in familial occurrence of papillary thyroid carcinoma. This report describes three sisters of first degree relation with papillary thyroid carcinoma.

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