Familial Isolated hyperparathyroidism caused by single adenoma

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ABSTRACT

Hyperparathyroidism is associated with increased secretion of parathyroid hormone. Primary hyperparathyroidism is due to adenoma or hyperplasia and very rarely carcinoma. Although various forms of familial hyperparathyroidism exist, they usually present with parathyroid hyperplasia. But a familial pattern of hyperparathyroidism caused by single adenoma also may be manifested rarely. A thorough pre operative evaluation including radionuclide scan is mandatory before contemplating surgery. Excision of the adenoma almost always cures the condition, but search for other parathyroid should also be made during exploration. We hereby present a 26 yr old lady with a familial hyperparathyroidism caused by a single adenoma.

Key words: Familial primary hyperparathyroidism, parathyroid adenoma, parathyroidectomy

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A familial pattern of hyperparathyroidism caused by single adenoma may be manifested rarely. Primary hyperparathyroidism is due to adenoma or hyperplasia and very rarely carcinoma. Although various forms of familial hyperparathyroidism exist, they usually present with parathyroid hyperplasia. But a familial pattern of hyperparathyroidism caused by single adenoma also may be manifested rarely.[1] We hereby present a 26 yr old lady with a familial hyperparathyroidism caused by a single adenoma.

CASE REPORT

A 26yr old patient presented with fracture of the neck of the right femur following a trivial fall. On investigation she had multiple fractures of the lower thoracic and lumbar vertebra. She had a clinically palpable nodule in the region of the thyroid which led to the suspicion of parathyroid adenoma. X ray of the right hand showed subperiosteal erosion over radial aspect of the middle phalanx of the index finger. Parathormone was greatly elevated to the level of 654 ng/ml with a serum calcium of 12.1 gm/dl. Ultrasound abdomen was done to rule out possibility of other neoplasms associated with MEN syndrome. The study was normal. FNAC of the swelling was unable to differentiate between an adenoma and hyperplasia. Radionuclide scan was done which confirmed the presence of an adenoma showing increased uptake in that region. Hence based on these findings bilateral neck exploration was carried out under general anesthesia. Patient had an enlarged left lower parathyroid gland which was excised after frozen section confirmed the diagnosis of an adenoma.

The other three parathyroid glands were found to be normal and were left alone. Following surgery the parathormone values dropped to 107.6 ng/ml in three weeks and serum calcium returned to 9.2 mg/dl. Histopathology report was that of a parathyroid adenoma. The patient recovered subsequently.

Two months later the patients elder sister (31 yrs) presented with complaints of lower backache. Knowing familial history she was also worked up for the pres-
ence of parathyroid adenoma. Even though the ultrasound of the neck did not pick up any abnormality the parathormone was raised to the level of 1314 ng/ml with serum calcium value of 12.2 mg/dl. Radionuclide scan confirmed the increased uptake in right lower parathyroid gland. X-ray hand also confirmed the presence of subperiosteal erosion. Ultrasound of the abdomen was normal.

The patient underwent bilateral neck exploration. The right lower parathyroid was enlarged and was excised for histopathological confirmation. The other glands were found to be normal. Histopathology confirmed the presence of parathyroid adenoma. The patient recovered after surgery.

**DISCUSSION**

In mild hypercalcemia, many patients are asymptomatic and this condition is frequently discovered accidentally during routine laboratory screening. Familial hyperparathyroidism (FHP) is a hereditary disease where hyperparathyroidism (HPT) is transmitted in an autosomal dominant fashion. FHP consists of a variety of diseases such as multiple endocrine neoplasia type1 (MEN 1) and type2 (MEN 2), familial isolated hyperparathyroidism (FIHPT) with single adenoma and with multiple adenomas (or hyperplasia), and FHP with jaw-tumor (FHP-JT). Isolation of the genes responsible for MEN1, and 2, i.e. MEN1 and RET, respectively, makes it possible to examine the relations among disorders constituting FHP.[1]

The disease may follow a recessive mode of inheritance or may be due to a dominant germ-cell mutation in one of the parents. Ultrasonography can be a sensitive and accurate method for preoperative localization of enlarged parathyroid glands in primary hyperparathyroidism, comparable in overall utility to sestamibi scintigraphy. Ultrasound and nuclear medicine scintigraphy provide complementary roles in the preoperative localization of parathyroid adenomas in patients with PHPT.[2]

DNA index by flow cytometry and image cytometry differentiates normal from abnormal parathyroids. DNA index might influence extent of resection in two- and three-gland hyperplasia and selection of the most appropriate gland for autografting and cryopreservation in patients with four-gland hyperplasia.[3]

Bilateral parathyroid exploration with thyroid mobilization by capsular dissection is the procedure of choice for PHPT. Patients undergoing a unilateral procedure have a lower incidence of biochemical and severe symptomatic hypocalcaemia in the early postoperative period compared with patients undergoing bilateral exploration. Unilateral neck exploration with intraoperative parathyroid hormone assessment is a valid surgical strategy in patients with primary hyperparathyroidism with distinct advantages, especially for patients with solitary parathyroid adenoma.

**REFERENCES**