Proceeding S.Z.P.G.M.I. vol: 21(1): pp. 45-47, 2007.

Familial Primary Hyperparathyroidism

Pervaiz Iqbal and Rabail Javed
Department of Orthopaedic Surgery, Sheikh Zayed Postgraduate Medical Institute, Lahore Pakistan-54600

ABSTRACT

A case of primary hyperparathyroidism in a 17 years old female is described. Patient presented with severe abdominal and pelvic pains associate with discomfort. Diagnostic testing revealed multiple upper and lower limb fractures and renal stones. Case was self limiting. Patient's condition resolved on parathyroidectomy for adenoma and partial thyroidectomy. Follow-up showed familial links.

Key Words: Primary Hyperparathyroidism, Multiple Fractures, Familial Hyperparathyroidism

INTRODUCTION

Parathyroid regulates calcium. In hyperparathyroidism excessive calcium gets removed from the bones causing bone deformities and fractures in severe cases. Primary hyperparathyroidism is an intriguing condition. The incidence of primary hyperparathyroidism is suggested to be in 1:1000 of the population[1].The commonest pathology, a solitary adenoma, forms in about 80% of the cases. The remaining causes are equally distributed between multigland adenomata and diffuse hyperplasia. Carcinoma is the rarest type and forms less than 2% of cases[2]. The commonest presentation currently is thought to be asymptomatic hypercalcemia. Fifty percent of the symptomatic patients present with renal stones and these form 5% of all those managed with renal calculi[3]. The vast majority of cases occur in people with no familial history of the disorder. Only about 3-5% can be linked to an inherited problem.

CASE REPORT

A 17 year old girl was admitted on February 2007 in our unit. At the time of admission she had all apparent features of primary hyperparathyroidism (pHPT). She was suffering form multiple upper and lower limb fractures, abdominal pain, psychosis and renal stones. The radiological examination of extremities revealed marked generalized osteopenia and coarse trabeculations; fracture shaft of femur on both sides, segmental fracture of shaft of humerus on both sides (Fig. 1), subperiosteal erosions in tubular bones of hand and radiolucent area in proximal phalanx of left ring finger (Fig. 2). The X-ray abdomen, Ultrasound Examination and IVU reports confirmed multiple renal stones. Ultrasound and MIBI scan of neck for parathyroid revealed adenoma. A complete blood examination was carried out. Her serum parathormone was 934pg/ml as against in control of 11-67 pg/ml. Her serum calcium level was 16.7 mg/ml (control of 8.8-10.5mg/ml).

Excision of parathyroid adenoma with left lobe of thyroid and partial isthemectomy was performed by the department of general surgery. The fractures were managed conservatively. The histological examination revealed appearances of a parathyroid adenoma. Her serum calcium levels and parathormone were monitored regularly during recovery and postoperative period. Headaches, episodes of psychiatric disorder, depression, vomiting and pains slowly diminished. Her condition was stabilized within two months after surgery. She was discharged with normal vital signs, calcium, and parathyroid levels.

Familial risk factor was also investigated. Blood test reports suggested familial links. She is fourth out of six sisters with three of her elder sibling neonatal expiries (two elder brothers with intra-uterine deaths: IUD). All other family members had normal calcium and parathyroid levels except her father. Serum calcium of her father was

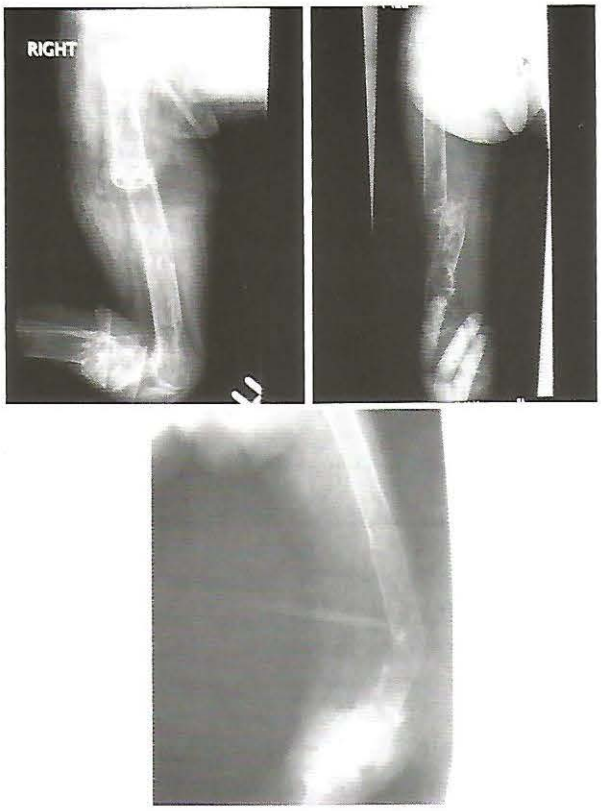

Fig. 1: Healing fractures at different stage and deformities.

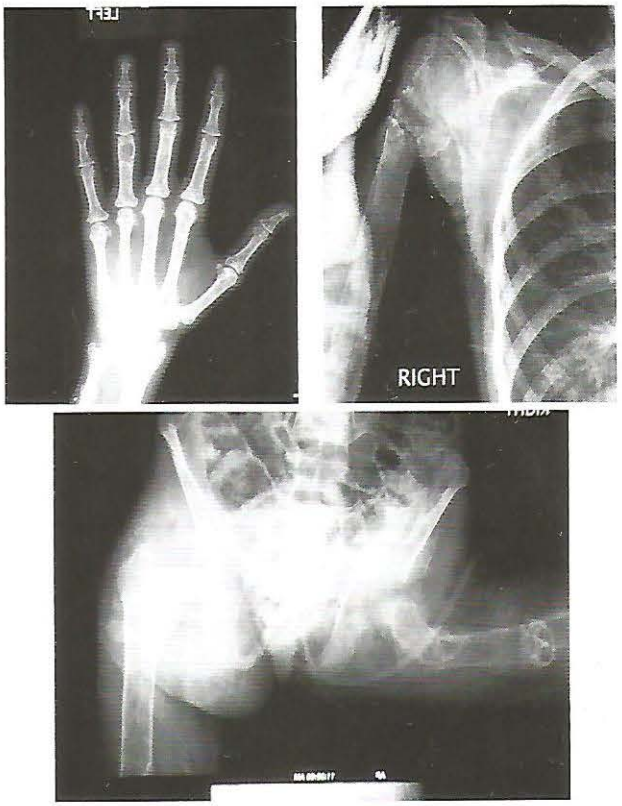

Fig. 2: Subperiosteal erosions in tubular bones.

found to be 11.7mg/ml. The parathyroid level was 90pg/ml. He also had a 15-20 years history of complex renal stones. Ultrasound examination of abdomen revealed bilateral renal stones but no evidence of multiple endocrine neoplasia (MEN). Doppler ultrasound examination of neck revealed a hypoechoic lesion with peripheral calcification at the lower pole of left thyroid measuring 1.6x1.0 cm with high probability of parathyroid adenoma.

DISCUSSION

Familial hyperparathyroidism is a rare disorder. In this case, we found a familial link with adenoma of parathyroid. This makes it an important case to describe. Good results have been obtained in early diagnosis, treatment and follow up of hyperparathyroidism and progress has been made in search for genes involved in dysgenesis and dyshormonogenesis of thyroid gland. However a few cases of hyperparathyroidism with familial links have been reported from developing countries.

In our study we focused on familial trend of this young girl. Her family profile related her hyperparathyroidism with inheritance. Her parents were consanguineous. While patients father was a chronic patient of hypercalcemia and renal stones. This may interpret an autosomal inheritance of the trait for hyperparathyroidism in the patient. It has been suggested that the clinical expression of familial hyperparathyroidism within a particular family tends to remain constant towards either largely bone or renal pathology.[4]

A study explains as inherited syndromes to be minor causes of pHPT (<5%).It includes multiple endocrine neoplasia type1 and type2a (MEN-1, MEN-2a), familial hypocalcuria hypocalcaemia (FHH) and neonatal severe pHPT.MEN-1 is inherited as an autosomal dominant trait[5,6].

In other studies, it is described that hyperparathyroidism with single adenoma may follow a recessive mode of inheritance or may be due to a dominant germ-cell mutation in one of the

parents[7,8]. Careful long term follow up is required because metastasis of parathyroid carcinoma occurring as long as 15 years after the resection of a parathyroid 'adenoma' has been reported[9,10]

We describe from our study as primary hyperparathyroidism with inherited links to occur at early life stage. Primary hyperparathyroidism is reported in females above the age of 30, but in our study we presented a young girl with parathyroid adenoma as well as family links. This focuses on hyper parathyroid condition in young girls and boys due to familial and acquired conditions. Much research and investigation still needs to be done on such cases especially in developing countries. We feel that in all cases of primary hyperparathyroidism below the age of thirty years, possibility of familial links should be kept in mind.

X-Rays pictures of patient showing pronounced bony changes of hyperparathyroidism, like generalized osteopenia, coarse trabeculations, subperiosteal erosions, cyst formation, cortical thinning, and multiple fractures.

REFERENCES

1. O' Riordan JLH. Hormonal control of mineral metabolism. In: O'Riordan JLH, ed *Recent Advances in Endocrinology & Metabolism (No. 1).* Edinburgh: Churchill Livingstone.1978; 194.
2. Arnaund CD. The parathyroid glands:primary hyperthyroidism. In Wyngarden JB, Smith JH. eds. *Cecil Textbook of Medicine. Part XI, 16th edn).* Philadelphia: WB Saunders, 1982; 1289-97.
3. Kanis JA. Disorder of calcium metabolism.In: Weatherall DJ, Ledingham JGG, Woorell DA eds. *Oxford Textbook of Medicine (Vol. 1,2nd edn)* Oxford: Oxford University Press, 1987; 59-62.
4. Miedlich et al 2001b
5. Peters N, Chalmers TR, Rack JH, et al. Bone and renal pathology in familial hyperparathyroidism. *Post Graduate Medical Journal* 1966; **42:**228.
6. Marsden P, Anderson J, Doyle D, et al. Familial hyperparathyroidism. *Br Med J,* 1971; **3:**87-90.
7. Joshua CC, Shenoy J, Alfred AJ, Gaurav M, Keshav P. Familial Isolated hyperparathyroidism caused by single adenoma. *Indian J Surg* 2005; **67:**265-266.
8. Scheiner JD, Dupuy DE, Monchik JM, Noto RB, Cronan JJ. Pre-operative localization of parathyroid adenomas: a comparison of power and colour Doppler ultrasonography with nuclear medicine scintigraphy. *Clin Radiol* 2001; **56:**984-8.
9. Megumi F, Ken O, Kaori S, et al. Familial isolated hyperparathyroidism due to multiple adenomas associated with ossifying jaw fibroma and multiple adenomyomatous polyps. *J Endocrinol* 1998; 138:557-61.
10. Fujimoto Y and Obata T. How to recognize and treat parathyroid carcinoma. *Surgical Clinics of North America* 1987; **67:** 343-57.

The Authors:

Pervaiz Iqbal
Assistant Professor
Department of Orthopaedic Surgery
Sheikh Zayed Postgraduate Medical Institute, Lahore Pakistan -54600
Email: piqbal50@gmail.com

Rabail Javed
Research Officer
National Health Research Complex
Sheikh Zayed Postgraduate Medical Institute, Lahore, Pakistan -54600
Email: rabailjaveed@hotmail.com

Address for Correspondence:

Pervaiz Iqbal
Assistant Professor
Department of Orthopaedic Surgery
Sheikh Zayed Postgraduate Medical Institute, Lahore, Pakistan-54600
Email: piqbal50@gmail.com