PRIMARY HYPERPARATHYROIDISM DUE TO A DELETION OF THE CDC73 GENEp2-74

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ESPE 2021
The 59th Annual Meeting Online
22-26 September 2021

INTRODUCTION

Primaryhyperparathyroidism(PHPT), oftencausedbya single adenoma (80%–85%) orhyperplasiaoradenomas involving multipleS glands, isthe majorcause of hypercalcemia. PHPT is common and occurs in individuals of all ages, but its prevalence is lower in young adults. Parathyroid tumors and PHPT can be caused by germline(hereditaryPHPT) or somatic mutations of tumor suppressor genes (e.g.multiple endocrine neoplasia type1 and CDC73) and proto-oncogenes. Approximately 5%–15% of PHPT cases occuring a familial setting, either in an isolated form (familial isolated hyperparathyroidism) or associated with other syndromal features(MEN1-2A, familial hypocalciuric hypercalcemic).I n 15-20% of sporadic parathyroid carcinomas a mutation in the *CDC73gene (HRTP2) is identified, which is why some authors, considerita tumor suppressor gene.*

AIM

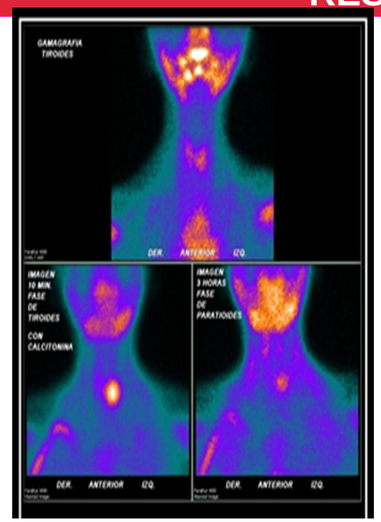
To present a patient with a parathyroid adenoma carrying a deletionof the *CDC73 gene in heterozygous state*

METHOD

We investigated the pathogenic mutations in the CDC73 gene and analyzed their relation ship with the phenotype in the patient with PHPT and parathyroid adenoma using Multiplex-ligation dependent probe amplification(MLPA**).**

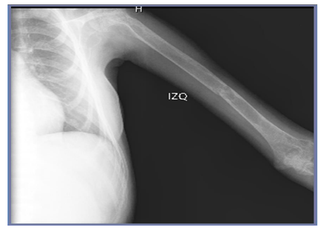
RESULTS

A17-year-old Female presented with pathological fracture of the middle third of the humerus with polyuria, polydipsia, radiographs with generalized osteopenia and osteolytic lesions and nephrocalcinosis.
Bone densitometry-4 SD.
Scintigraphy with sestamibi showed oval mass in the left lower lobe probable left atypical parathyroid adenoma.
Months later she became psychosis.
5 years later the tumor relapsed in the neck, showed cervical nodule reported a s metastasis o f parathyroid adenoma; with no evidence o f parathyroid injury. With calcium13mg / dl (vn8,5-10.5 mg/dl) , PTH> 2000pg/ml (vn10-55 pg/ml)
GENETIC study reported a HETEROZYGOUS DELETION OF THE CDC73 GENE. Mother without genetic alteration s .No family history of hyperthyroidism.
Satisfactory Clinical evolution.

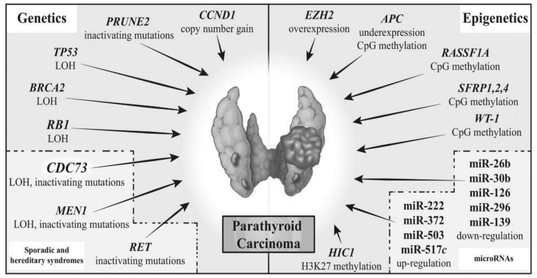


Scintigraphy with sestamibi showed oval mass in the leftl ower lobe

Laboratories	Normal value
Calcium 17 mg / dl	8,5-10.5 mg/dl
Ionic calcium 4.24 mmol / l	1,1-1,3 mmol/l
PARATOHORMONE 1834 pg./ml	10-55 pg./ml
Phosphorus 3.6 mg/ dl	4-7 mg /dl
Alkaline phosphatase 2037 UI/l	44-147 UI/l



Fracture of the middle third of the humerus



CONCLUSIONS

PHPT is the main cause of hypercalcemia. Although the most common disease is sporadic PHPT ,due to a single adenoma, t here are heritable forms in 10% of cases. Patients with parathyroid carcinoma should undergo a careful review oft heir family history and should be offered a genetic study ofthe CDC73 Gene.

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ACKNOWLEDGEMENTS

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