

Abstract

Primary hyperthyroidism (pHPT) is common endocrine disorder. In more than 80% of the patients with pHPT, a single parathyroid adenoma can be found, in 15-20% of the cases parathyroid hyperplasia or multiple adenoma and very rare parathyroid carcinoma (1%). Primary hyperparathyroidism occurs mainly as a sporadic disorders (90%), however 10% of the patients present a hereditary form of disease in which pHPT is either the main or associated feature. Based on both clinical and molecular characterisation, hereditary pHPT can be divided into several subgroups:

1. hyperparathyroidism-jaw tumor syndrome (**HPT-JT**)
2. **MEN 1**
3. **MEN 2**
4. familial isolated hyperparathyroidism - (**FIHP**)
5. familial hypocalcaemic hypercalcaemia (**FHH**) and neonatal severe primary hyperparathyroidism (**NSHPT**)

The environmental risk factors that predispose to or promote parathyroid neoplasia are:

- previous neck irradiation
- chronic application of lithium
- calcium and vitamin D deficiency
- female gender are more affected (ratio female/male = 2-3 :1)
- menopause

It is very important to note that patients with pHPT deserve further investigation to some hereditary syndrome if:

- patients younger than 40 years
- patients have multiple parathyroid adenomas or parathyroid hyperplasia(MEN 1, FHH)
- patients have atypical parathyroid adenoma or carcinoma (HPT-JT)
- family history for some hereditary pHPT syndrome

Key words: primary hyperparathyroidism, jaw tumor, MEN 1, MEN 2, FIHP.