Atlas of Genetics and Cytogenetics in Oncology and Haematology

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Cancer Prone Disease Section

Mini Review

Hyperparathyroidism-jaw tumor syndrome (HPT -JT)

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Identity

Note

Some Familial Isolated Primary Hyperparathyroidism (FIHP) families have been mapped to this locus.

Inheritance

Autosomal dominant, early onset, highly penetrant, reported in caucasians and Japanese so far.

Clinics

Note

- Phenotypic spectrum variable;
- Growth: normal;
- Head: normal;
- Skin: normal;
- Hyperparathyroidism, jaw tumors, and kidney cysts;
- Hyperparathyroidism develops in about 95% of affected individuals due to parathyroid adenomas which can occur singley or multiply, with some patients undergoing several parathyroidectomies over the course of their lives;
- Parathyroid carcinoma develops in about 5% of patients (compare to 1 in 5 million in the general population);
- The jaw tumors consist of trabeculae of woven bone set in a cytologically bland fibrocellular stroma;
- About 50% develop fibro-osseous tumors of the maxilla or mandible, which may recur and are independent of the course of the parathyroid adenomas;
- Some families display an increased risk of developing kidney cysts (nephroblastomas or hamartomas) or adult Wilms tumors;
- Surgical removal of neoplastic tissues;
- Regular serum calcium level screening is a cost effective method to catch the development of parathyroid adenomas at an early stage;
- Dental x-rays are not recommended; may increase risk of development of jaw tumors.

Evolution

Usually develop parathyroid adenomas by age 40 years old (range: 10-66 years).

Cytogenetics

Inborn conditions

Normal.

Cytogenetics of cancer

Loss of heterozygosity of all of chromosome 1 has been seen in some HPT-JT parathyroid adenomas.

Genes involved and proteins

HRPT2 (not yet cloned)

Location

1q25-31

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