**Prevalence of familial isolated pituitary adenomas (FIPA)**

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**Background**

- Clinically relevant pituitary adenomas have a prevalence of approximately 1:1000.
- While pituitary adenomas usually occur as sporadic disease, an increasing number of patients are being recognised with a family member also suffering from a pituitary adenoma.
- If no other syndromic features are present, these families are categorised as familial isolated pituitary adenoma (FIPA).
- Germline mutations of the AIP (aryl-hydrocarbon receptor interacting protein) gene are associated with a predisposition to pituitary adenomas.
- In published studies, 20% of the FIPA families, 20% of sporadic childhood and 13% of sporadic young-onset (≤30y) acromegaly patients carry a germline AIP mutation.

**Patient cohort and methods**

- Retrospective and prospective audits were performed in a tertiary referral centre for pituitary diseases for a family history of pituitary adenoma.
- Known MEN1 patients were excluded.
- Retrospective data were gained from reviewing the notes of 225 patients with acromegaly.
- Prospective data were gained from a questionnaire of 222 pituitary adenoma patients.

**Results – family history**

- Retrospective acromegaly patients (n=225): 3% AIP positive, 96.4% AIP negative.
- Prospective acromegaly patients (n=64): 10.93% positive family history.
- All prospective patients (n=222): 7% positive family history.

**Results - Young onset disease and AIP mutations**

- Retrospective young onset acromegaly patients (diagnosis ≤ age 30) n=225: 15% positive family history.
- Prospective young onset acromegaly patients (diagnosed ≤ age 30) n=64: 27% positive family history.

**Summary**

- Our data suggest that 7% of an unselected pituitary patient population of a tertiary referral centre have a family history of pituitary adenoma.
- There is a considerable difference in the percent of patients with family history between the retrospective and prospective acromegaly cohorts (3.6% vs. 10.9%), suggesting that careful history taking increases the proportion of patients with a family history of acromegaly nearly 3-fold.
- Active inquiry may reveal previously unknown familial connections in all types of pituitary adenoma.
- Increased awareness and genetic screening, when available, could provide the possibility of early tumour detection and treatment.

http://www.fipapatients.org/