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Prevalence of familial isolated pituitary adenomas (FIPA)

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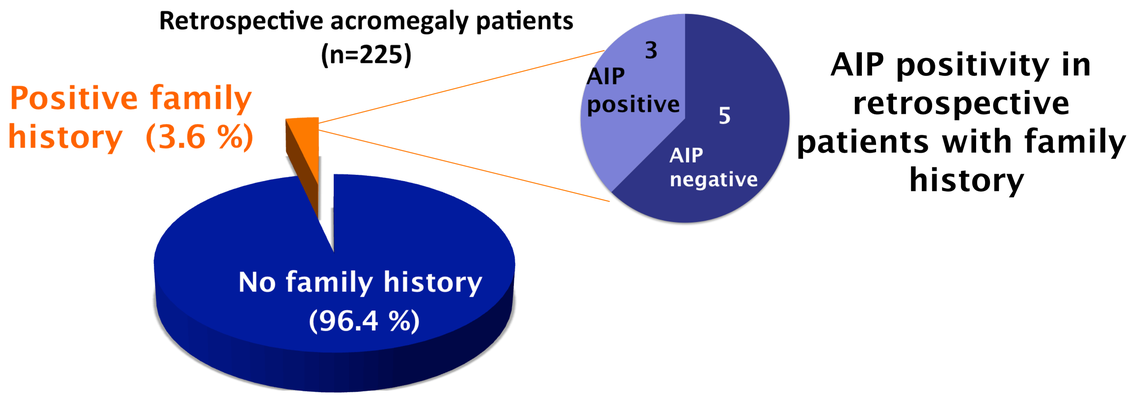
Background

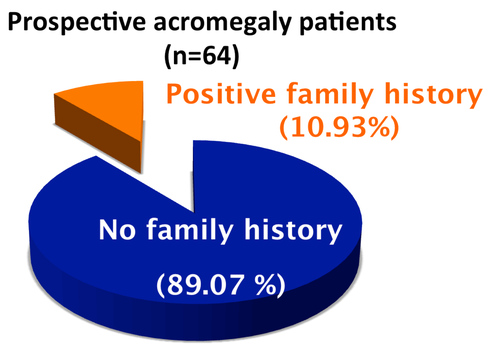
- Clinically relevant pituitary adenomas have a prevalence of approximately 1:1000
- While pituitary adenomas usually occur as sporadic disease, an increasing number of patients are being recognised with a family member also suffering from a pituitary adenoma
- If no other syndromic features are present, 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

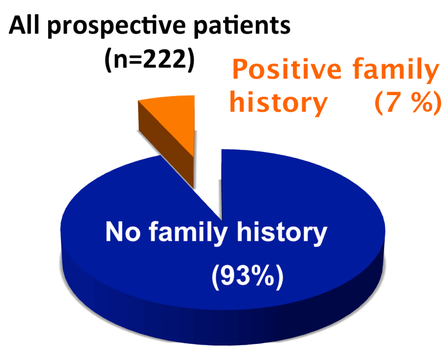
Patient cohort and methods

- 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

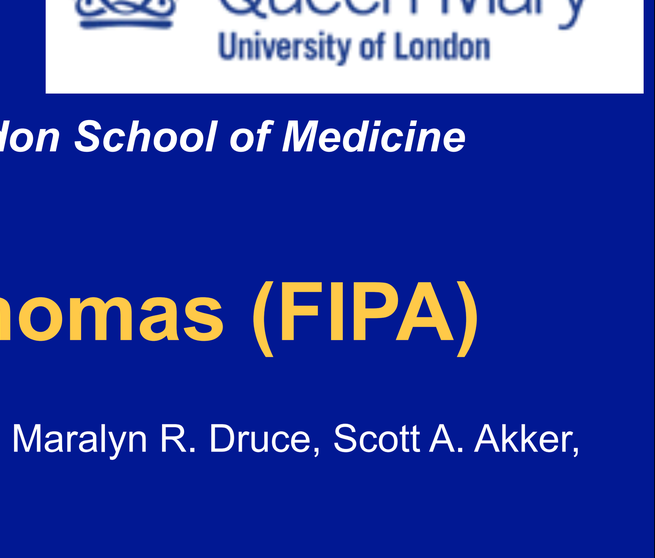
Results – family history

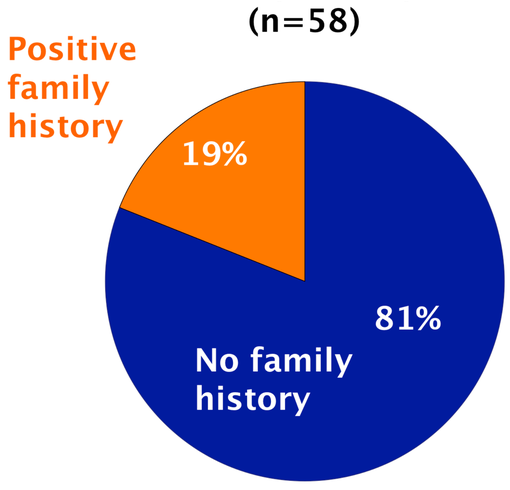


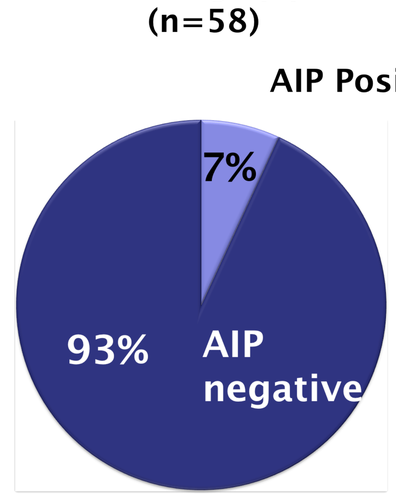


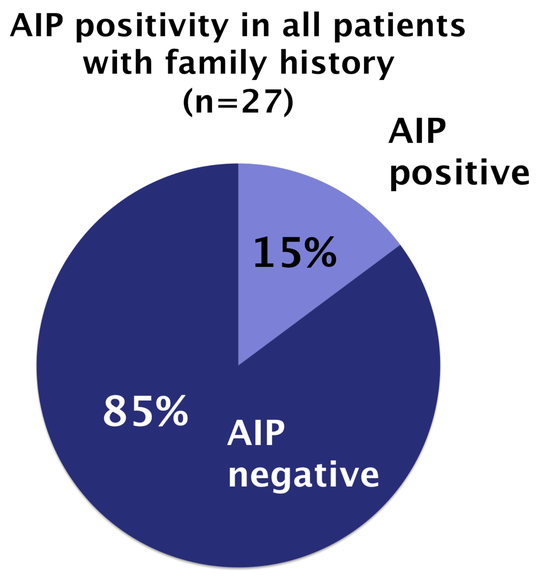


Results - Young onset disease and AIP mutations









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