MOLECULAR GENETIC ANALYSIS IN FAMILIAL ISOLATED PITUITARY ADENOMA PATIENTS

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Introduction

FIPA – is a syndrome which includes pituitary adenomas with any kind of secretion in two or more members in a family in the absence of MEN or Carney complex; it also includes isolated family somatotropin syndrome (IFS).

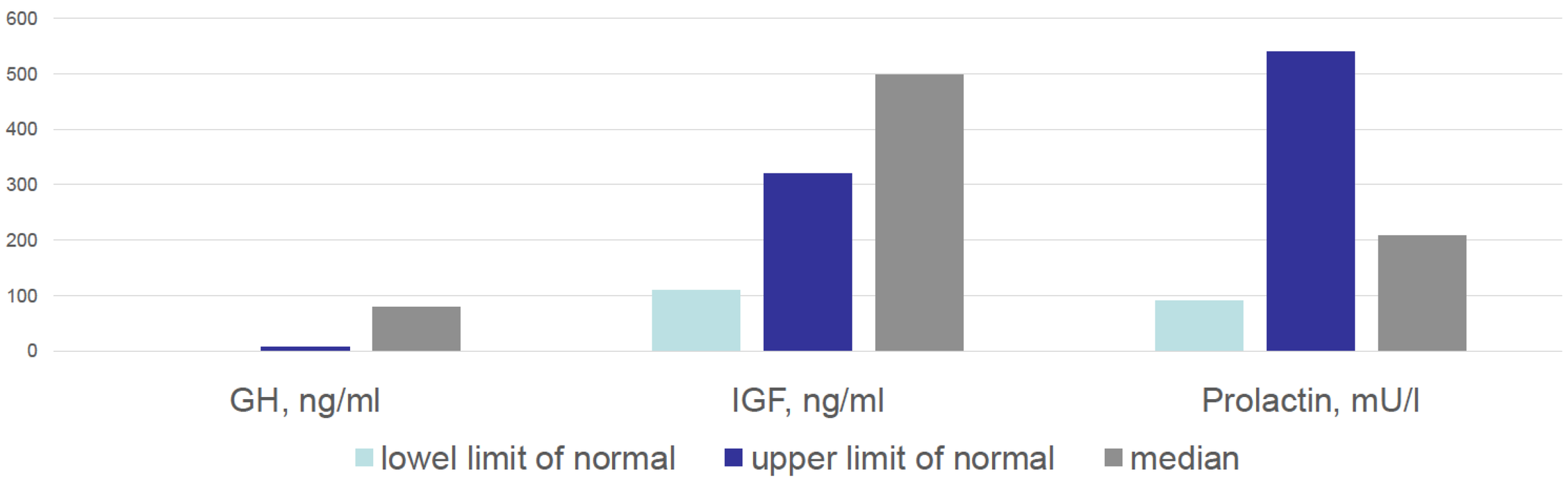
Aim

Molecular genetic study of a gene panel in FIPA patient.

Materials and methods

Study included 1 family (2 men, 24 and 58 years) with pituitary adenomas with homogeneous secretion type — somatotropinomas.
Brain MRI: macroadenomas (maximum size of 39 mm). One patient after combination therapy (somatostatin analogs in the maximum dose and primary surgery, without radiological therapy), another patient without any treatment at the time of the study. There were no normalization of GH and IGF.

Genomic DNA from a blood samples of patients underwent high-throughput sequencing on the Ion Torrent Personal Genome Machine (Life Technologies) using a custom-designed AmpliSeq panel for the sequencing of a panel of genes (MEN1, CDKN1B, PRKAR1A, GNAS, AIP, SDHA, SDHB, SDHC, SDHD, PRKCA, CDKN2C, CDKN2A, POU1F1, PTTG2).



Results

Direct sequencing revealed mutation in exon 6 of the gene AIP p.R271W, which was not previously described in studies.

Conclusion

We were able to identify a new AIP mutation in FIPA family.

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