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Introduction

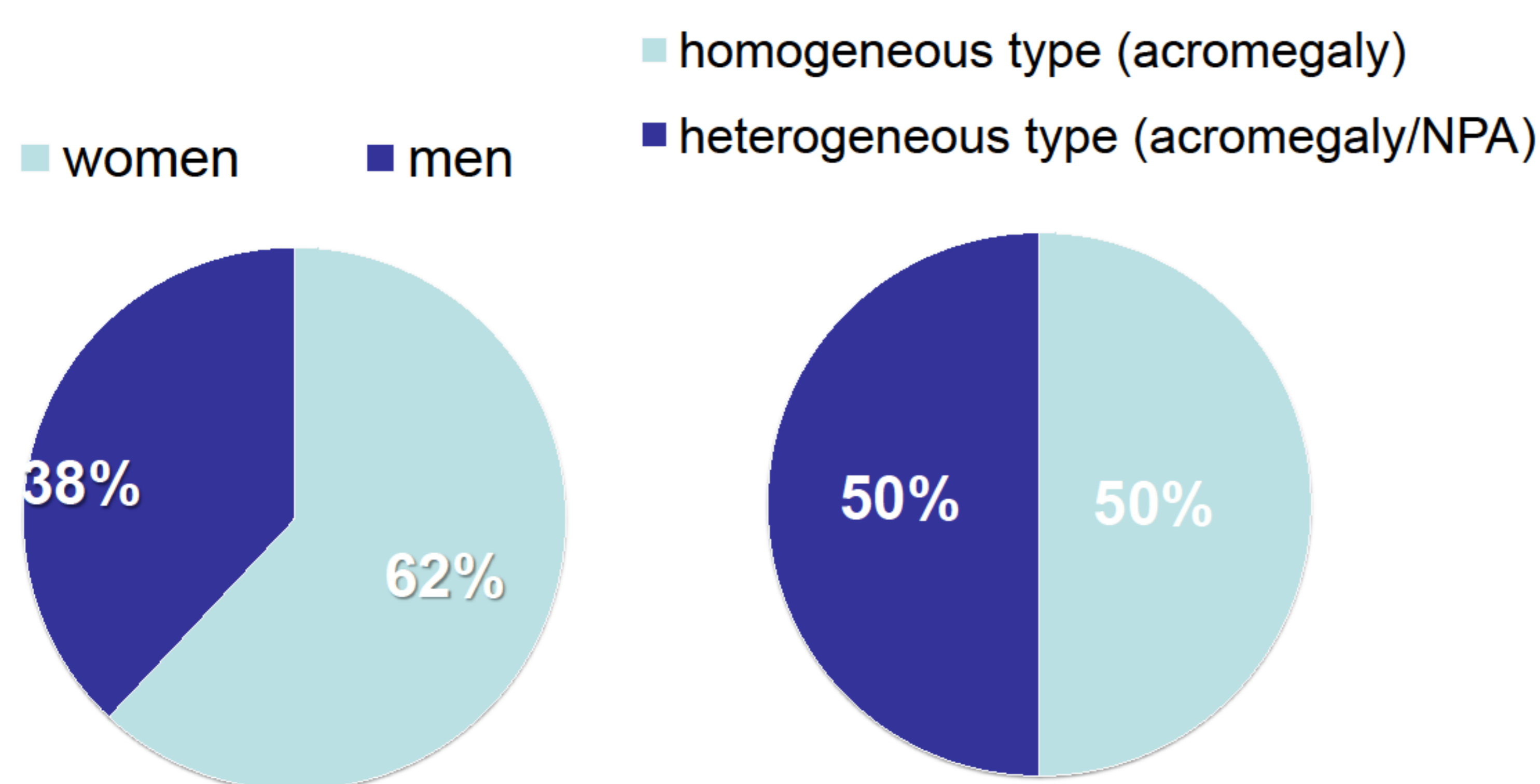
Several genetic syndromes are associated with familial pituitary adenomas. The penetrance of clinical manifestations of these syndromes is not ubiquitous and this might be the reason for the lack of detection of genetic mutations when only one or few genes are studied.

Aim

Clinical characterization and molecular genetic study of a panel with 10 genes involved in formation of pituitary adenomas in familial setting.

Materials and methods

Study included 6 families (13 patients) with familial pituitary adenomas with no other features of known genetic syndromes.

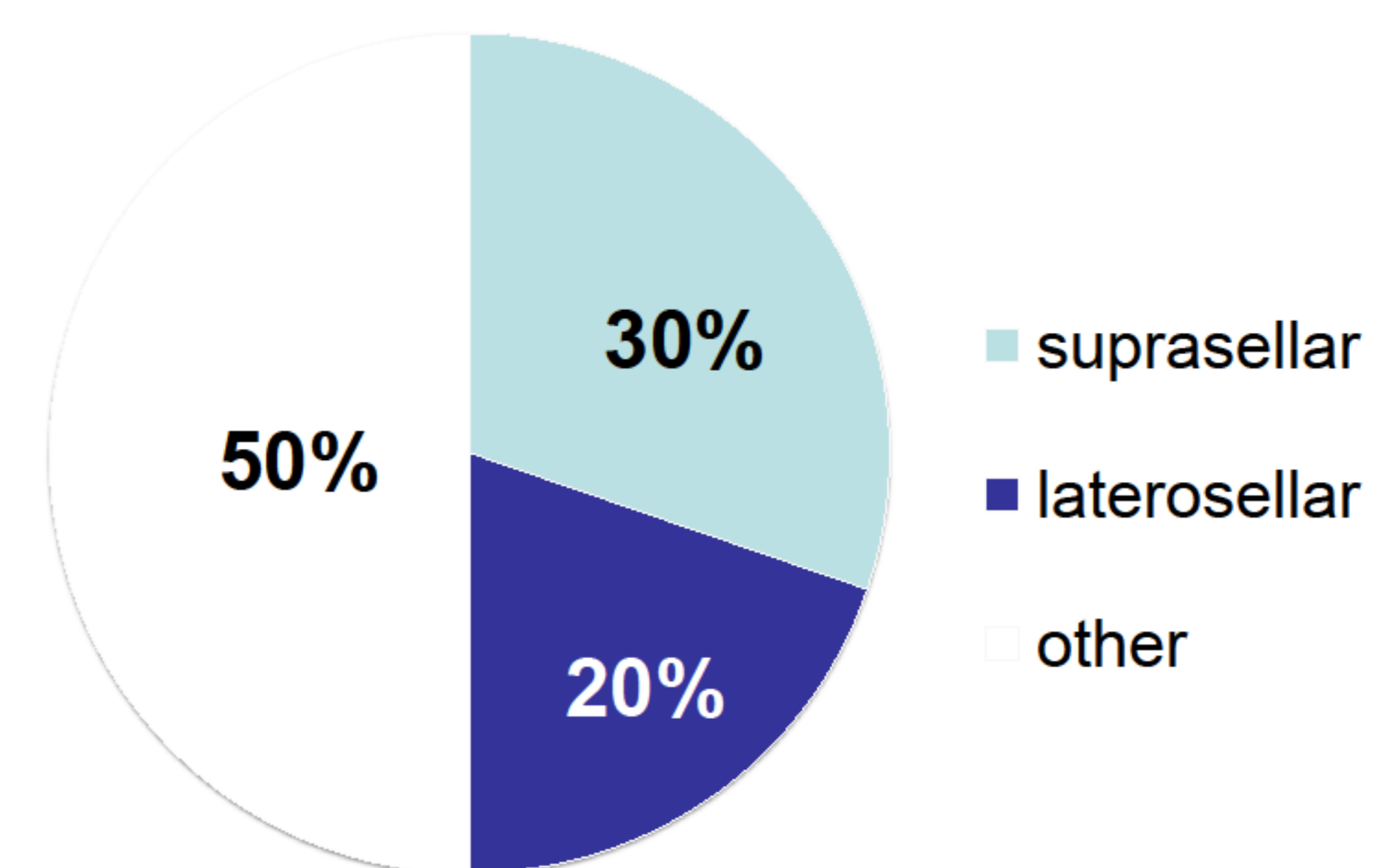


Genomic DNA from a blood samples of patients (probands) underwent high-throughput sequencing on the Ion Torrent Personal Genome Machine (Life Technologies, USA) using a custom-designed AmpliSeq™ panel for the sequencing of a panel of genes (MEN1, CDKN1B, PRKAR1A, GNAS, AIP, SDHA, SDHB, SDHC, SDHD, PRKCA, CDKN2C, CDKN2A, POU1F1, PTTG2) but no pathological mutations were detected.

Results

Parameter	Medium result	[Min - Max]
Growth hormone	13,9 ng/ml	[0,42 - 23,9]
IGF-1	1517 ng/ml	[439 - 1871]
Adenoma size	7 mm	[5,5 - 17]

Tumor extensions



In most patients, adenomas were extended in more than 2 directions.

Median age was 55.6 years (40 to 69 years), the average height for females was 160 cm, for males 170 cm.

Most adenomas were GH-producing and non-secreting. The diagnosis of acromegaly was confirmed by hormone testing (GH > 2.5 ng/ml, no suppression on OGTT below 1 ng/ml and high IGF-1).

Conclusions

Families with hereditary pituitary adenomas can have tumors with homogenous and heterogeneous types of secretion. We were not able to show any genetic alteration in the group of patients studied.

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