GENE PANEL STUDY FOR FAMILIAL PITUITARY ADENOMA

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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.

Results

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Medium result</th>
<th>[Min - Max]</th>
</tr>
</thead>
<tbody>
<tr>
<td>Growth hormone</td>
<td>13,9 ng/ml</td>
<td>[0.42 - 23,9]</td>
</tr>
<tr>
<td>IGF-1</td>
<td>1517 ng/ml</td>
<td>[439 - 1871]</td>
</tr>
<tr>
<td>Adenoma size</td>
<td>7 mm</td>
<td>[5.5 - 17]</td>
</tr>
</tbody>
</table>

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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