Cushings syndrome due to carneys complex – Case series and report of a new mutation from a South Indian tertiary care centre

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Introduction

Carneys complex is an Autosomal Dominant condition characterised by spotty skin pigmentation, myxomas, endocrine and non endocrine tumors. Cushings syndrome due to PPNAD occurs in 25-45% of patients with carneys complex.

Clinical presentation

3 patients in our series were diagnosed between second and third decade. All the 3 patients had diabetes, hypertension, cushingoid features. Diagnosis was clinically suspected by the presence of lentigenes on the face.

Method

We report case series of 3 patients diagnosed with cushings syndrome associated with carneys complex. Their clinical, biochemical, radiological and genetic details are described below.

<table>
<thead>
<tr>
<th>Sl.no</th>
<th>Sex</th>
<th>Age of onset (Years)</th>
<th>Age at diagnosis (Years)</th>
<th>Sr.k (meq/l)</th>
<th>Basal cortisol (ug/dl)</th>
<th>ONDST (ug/dl)</th>
<th>LDDS (ug/dl)</th>
<th>ACTH pg/ml</th>
<th>Imaging CT Abdomen and MRI Brain</th>
<th>HPR</th>
<th>Genetic testing</th>
</tr>
</thead>
<tbody>
<tr>
<td>1.</td>
<td>M</td>
<td>21</td>
<td>25</td>
<td>3.7</td>
<td>25.8</td>
<td>26</td>
<td>45</td>
<td>&lt;1</td>
<td>Nodular Left Adrenal Gland</td>
<td>PPNAD</td>
<td>Positive (C769G&gt;A;E257K) New variant</td>
</tr>
<tr>
<td>2.</td>
<td>F</td>
<td>13</td>
<td>18</td>
<td>4.4</td>
<td>10</td>
<td>14.4</td>
<td>17.5</td>
<td>&lt;1</td>
<td>Both Adrenals Normal Pituitary micro adenoma on MRI</td>
<td>PPNAD</td>
<td>Negative</td>
</tr>
</tbody>
</table>

Conclusion: Multiple lentigenes, a classical hike in serum cortisol after dexamethasone administration, suppressed ACTH and radiological evidence of nodular adrenals can help us suspect carneys complex in a patient presenting with cushings syndrome. Genetic testing could be negative in 30-40% of patients.

Follow up: None of them had cardiac myxomas on screening. USG Neck and Scrotum were normal on screening. All the 3 patients underwent bilateral adrenalectomy. Patients 1 and 3 were under follow up and patient 1 on follow up had High grade olfactory Neuroblastoma (Esthesioneuroblastoma)