**Background**

- Familial hyperaldosteronism type I (FH-I, OMIM #103900) is often characterized by severe hypertension, variable hyperaldosteronism, low plasma renin activity (PRA) and normal or decreased serum potassium due to unequal cross-over between genes that encode the steroid 11β-hydroxylase (CYP11B1) and aldosterone synthase (CYP11B1) enzymes, which results in a chimeric CYP11B1/CYP11B2 gene (CG) with aldosterone synthase activity regulated by plasma ACTH.

- An early diagnosis and treatment is important, not only to manage hypertension but also to avoid possible deleterious effects of aldosterone on the endothelium and cardiovascular diseases.

**Clinical case**

- A 3 months old boy was referred for evaluation because his mother, grandfather and uncle have FH-I confirmed by presence of chimeric CYP11B1/CYP11B2 gene.
- He was born at 36 weeks gestation, cesarean delivery due to intrauterine growth restriction, birth weight 2365 g. (<p10th) and birth length 44 cm (<p10th).
- He was admitted to the hospital during his first week of life due to transient tachypnea; without electrolytes or blood pressure disturbances during hospitalization.
- At initial evaluation his was normotensive (75/54 mmHg, reference <106/62 mmHg) and his physical exam was unremarkable.
- Laboratory tests were consistent with hyperaldosteronism:
  - Aldosterone 120 ng/dL, (reference: 5-90 ng/dL)
  - PRA = 0.39 ng/mL/h-1, (reference: 2.35-37 ng/mL/h-1)
- Genetic study was performed by XL-PCR and confirmed chimeric CYP11B1/CYP11B2 gene.
- The patient began treatment with cortisol (10 mg/m2/d) despite he had normal blood pressure.

**Molecular test**

![Chimeric gene](image)

- Positive control
- Negative control
- Patient

**Table 1. Clinical characteristics**

<table>
<thead>
<tr>
<th>Age (months)</th>
<th>New born</th>
<th>4</th>
<th>10</th>
<th>12</th>
<th>18</th>
<th>24</th>
<th>28</th>
</tr>
</thead>
<tbody>
<tr>
<td>Weight (g)</td>
<td>2365</td>
<td>5250</td>
<td>7320</td>
<td>7700</td>
<td>9000</td>
<td>9390</td>
<td>9950</td>
</tr>
<tr>
<td>Weight (SDS)</td>
<td>-1.78&lt;sup&gt;1&lt;/sup&gt;</td>
<td>-1.60&lt;sup&gt;1&lt;/sup&gt;</td>
<td>-1.78&lt;sup&gt;1&lt;/sup&gt;</td>
<td>-1.64&lt;sup&gt;1&lt;/sup&gt;</td>
<td>-1.75&lt;sup&gt;1&lt;/sup&gt;</td>
<td>-2.86</td>
<td>-2.28</td>
</tr>
<tr>
<td>Height (cm)</td>
<td>44</td>
<td>60.5</td>
<td>69</td>
<td>70</td>
<td>77.5</td>
<td>80.5</td>
<td>85</td>
</tr>
<tr>
<td>Height (SDS)</td>
<td>-1.50&lt;sup&gt;2&lt;/sup&gt;</td>
<td>-2.44&lt;sup&gt;2&lt;/sup&gt;</td>
<td>-1.32&lt;sup&gt;2&lt;/sup&gt;</td>
<td>-1.44</td>
<td>-1.65</td>
<td>-2.17</td>
<td>-1.64</td>
</tr>
<tr>
<td>DBP (mmHg)</td>
<td>80</td>
<td>68</td>
<td>99</td>
<td>---</td>
<td>---</td>
<td>90</td>
<td>90</td>
</tr>
<tr>
<td>DBP (SDS)</td>
<td>0.0</td>
<td>0.6</td>
<td>0.76</td>
<td>---</td>
<td>---</td>
<td>0.50</td>
<td>0.50</td>
</tr>
<tr>
<td>Cushing signs</td>
<td>No</td>
<td>No</td>
<td>No</td>
<td>No</td>
<td>No</td>
<td>No</td>
<td>No</td>
</tr>
</tbody>
</table>

SDS, standard deviation score; DBP, diastolic blood pressure.

<sup>1</sup> Corrected by gestational age (GA weeks).

**Conclusion**

- The early treatment with hydrocortisone (10 mg/m2/d) resolves the biochemical hyperaldosteronism in this normotensive infant with FH-I.
- As hyperaldosteronism has been associated with adverse cardiovascular, cerebrovascular, metabolic and renal sequel independently of its effects on blood pressure. We suggest genetic counsel and early diagnosed in high risk patient to have FH-I.

**Supported by FONDECYT 1130427 and 1150437, CORFO 13CTI-21526-P1 and IMII P09/016-F(ICM) Chilean Grants**