Isolated hypoaldosteronism is a rare endocrinopathy in a limited number of patients who secrete normal level of cortisol, due to mutation in CYP11B2. In some cases clinical diagnosis can be late and genetic analysis showed difficulties. There are some cases with clinically and biochemically appropriate AS deficiency but determination of mutation could be difficult as our patients.

Case-I
- A 7 month-old girl infant vomiting, failure to thrive and severe hyponatremia,
- Unexplained neutropenia
- Birth weight: 2440 gr.
- There was no consanguinity between parents.
- **Physical examination:**
  - Dehydration
  - She had atopic eczema.
  - Urine Na: 64 mmol/L (<20)
  - Plasma renin activity (PRA): 80,5 ng/ml/hours (0,48-4,88)
  - Aldosterone: 13,3 pg/ml (5,38-38)
- **Laboratory evaluation:**
  - Na: 122 mmol/L (136-146)
  - Urine Na: 64 mmol/L (<20)
  - Potassium: 5,9 mmol/L (3.8-5.5)
  - Ca: 11,7 ng/dl (9-11)
  - Plasma renin activity (PRA): 80,5 ng/ml/hours (0,48-4,88)
  - Aldosterone: 13,3 pg/ml(5,38-38)
- NaCl and fludrocortisone was initiated. LCMS profile was studied (Table 1).

A heterozygous change in the CYP11B2 (c.554 C>T (p.T185I)) was detected. Advanced genetic analyses has not been completed.

### Table 1: LCMS Profile of case I

<table>
<thead>
<tr>
<th>Steroid</th>
<th>Concentration</th>
</tr>
</thead>
<tbody>
<tr>
<td>Aldosterone</td>
<td>0,326 ng/ml</td>
</tr>
<tr>
<td>Cortisol</td>
<td>82,806 ng/ml</td>
</tr>
<tr>
<td>Cortisolone</td>
<td>20,21 ng/ml</td>
</tr>
<tr>
<td>11deoxycortisole</td>
<td>2,985 ng/ml</td>
</tr>
<tr>
<td>11deoxycorticosterone</td>
<td>0,822 ng/ml</td>
</tr>
<tr>
<td>17 OH-pregnenolone</td>
<td>1,224 ng/ml</td>
</tr>
<tr>
<td>17 OH-progesterone</td>
<td>0,326 ng/ml</td>
</tr>
<tr>
<td>21deoxycortisol</td>
<td>0,211 ng/ml</td>
</tr>
<tr>
<td>Androstenedione</td>
<td>0,08 ng/ml</td>
</tr>
<tr>
<td>Testosterone</td>
<td>0,02 ng/ml</td>
</tr>
<tr>
<td>DHEAS</td>
<td>10,427 ng/ml</td>
</tr>
<tr>
<td>Progesterone</td>
<td>0,18 ng/ml</td>
</tr>
<tr>
<td>Cortizone</td>
<td>24,21 ng/ml</td>
</tr>
</tbody>
</table>

### Discussion
- Aldosterone synthetase deficiency is a rare cause of persistent hyponatremia.
- Clinical findings vary with age.
- The association of eosinophilia and neutropenia has not been reported so far. In order to explain for these rare associations and make clear the heterozygous mutation an advanced genetic analyses is needed in rare cases such ours.

Case-II
- A 2 month-old male infant severe hyponatremia with suspected diagnosis of congenital adrenal hyperplasia.
- Birth weight of 2700 gr.
- Parents were first degree cousin.
- **Physical examination**
  - Dehydration
  - Weighgt: 3190 gr (<-2SDS)
  - Height: 52 cm (<-2SDS)
  - Head circumference 35.5 cm was (<-2SDS).
  - Severe eczema, protein-losing diarrhea, dissemine squamatos lesion
  - Eosinophilia
- **Laboratory evaluation:**
  - Na: 124 mmol/l (136-146)
  - Urine Na: 34 mmol/L(<20)
  - K: 6 mmol/L (3.8-5.5)
  - PRA: 100ng/ml/h (0,48-4,88)
  - Aldosterone: 5.4 pg/ml(5,38-38)

A heterozygous change was detected in the CYP11B2 gene c.763 G>T (p.Glu255Ter)
- Advanced genetic analyses (Next Generation Array) determined a homozygous change on CYP11B2 gene.