**INTRODUCTION**

Congenital Adrenal Hyperplasia (CAH) is an autosomal recessive disorder caused by impairment of one of the enzymes involved in the steroidogenesis pathway. 3β-hydroxysteroid dehydrogenase type 2 deficiency (3βHSD2 deficiency) is a rare form of CAH (<0.5%) due to pathogenic variants in the HSD3B2 gene encoding for the enzyme Type 2 3β-hydroxysteroid dehydrogenase Δ4–Δ5 isomerase (3βHSD2).

**CASE PRESENTATION**

A 15-day-old female neonate was referred from a local hospital to the Pediatric Department of “G. Gennimatas” Hospital of Thessaloniki due to dehydration with hyponatremia and hyperkalemia. The biochemical and hormonal profile was consistent with CAH (Table 1).

Table 1. Biochemical and hormonal profile of patient

<table>
<thead>
<tr>
<th>Biochemical and Hormonal profile</th>
<th>Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>17-OH progesterone (ng/ml)</td>
<td>69.5</td>
</tr>
<tr>
<td>Cortisol (µg/dl)</td>
<td>3.9</td>
</tr>
<tr>
<td>Aldosterone (µg/dl)</td>
<td>621</td>
</tr>
<tr>
<td>Potassium (mEq/l)</td>
<td>7.1</td>
</tr>
<tr>
<td>Sodium (mEq/l)</td>
<td>123</td>
</tr>
<tr>
<td>Testosterone (ng/dl)</td>
<td>309</td>
</tr>
<tr>
<td>DHEAS (µg/dl)</td>
<td>&gt;1500</td>
</tr>
<tr>
<td>Androstenedione (ng/ml)</td>
<td>18.9</td>
</tr>
<tr>
<td>Plasma Renin Activity (PRA) (ng/ml/h)</td>
<td>40.4</td>
</tr>
<tr>
<td>Aldosterone (µg/dl)</td>
<td>289</td>
</tr>
</tbody>
</table>

She was immediately commenced on hydrocortisone (15mg/m²/day) with a good response. The biochemical and hormonal profile was consistent with CAH (Table 1).

**METHOD**

- PCR and bidirectional sequencing for the CYP21A2, CYP11B1 and HSD3B2 genes was carried out sequentially.
- MLPA analysis was also performed for the identification of Copy Number Variations (CNVs) in the CYP21A2 gene (P050-C1 CAH).

**RESULTS**

**Molecular investigation of the CYP21A2 gene**

The index patient carried the p.Q318X pathogenic variant on a duplicated CYP21A2 gene in heterozygosity.

Both patients were found to have the p.Q318X pathogenic variant on a duplicated CYP21A2 gene in heterozygosity.

**Molecular investigation of the CYP11B1 gene**

No pathogenic variants were detected in the CYP11B1 gene on the index patient.

**Molecular investigation of the HSD3B2 gene**

The index patient carried the homozygous 2bp deletion at codon 273 (p.[Lys273Argfs*7];[Lys273Argfs*7]). Parents were found to harbor the p.Lys273Argfs*7 in heterozygosity (Figure 1).

**CONCLUSIONS**

In this study

We present a patient with a homozygous deletion in the HSD3B2 gene (p.Lys273Argfs*7) in heterozygosity.

- In compound heterozygosity with p.Thr318fs variant in a female patient presenting with salt wasting CAH, pigmentation and mildly enlarged clitoris at birth. This case is of the first female patient with a homozygous p.Lys273Argfs*7 in the HSD3B2 gene.

Molecular investigation is important

- to confirm clinical diagnosis
- to provide proper treatment and genetic counseling to the family.

**REFERENCES**


**ACKNOWLEDGEMENTS**

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**CONTACT INFORMATION**

Eirini Fylaktou
First Department of Paediatrics, Medical School National and Kapodistrian University of Athens, “Agia Sofia” Children’s Hospital
E-mail: efylaktou@med.uoa.gr