



## POSTER PRESENTATION

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# Clinical and molecular characterization of patients with classic $3\beta$ -hydroxysteroid dehydrogenase deficiency

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## Background

$3\beta$ -hydroxysteroid dehydrogenase type 2 ( $3\beta$ HSD2) is the key enzyme converting  $\Delta^5$ -steroids to  $\Delta^4$ -ketosteroids in adrenal and gonadal steroidogenesis. Severe loss-of-function mutations of HSD3B2 gene encoding for this enzyme cause the rare form of congenital adrenal hyperplasia, " $3\beta$ HSD deficiency". Affected individuals have salt losing, adrenal insufficiency and ambiguous genitalia in both sexes. Patients with  $3\beta$ HSD deficiency may have elevated  $17\alpha$ -hydroxyprogesterone (17OHP) levels due to normal peripheral type 1,  $3\beta$ HSD.

## Aims

To describe two unrelated patients with  $3\beta$ -hydroxysteroid dehydrogenase deficiency and perform mutation analysis of the HSD3B2 gene.

## Patients and Methods

Patient 1 (Thai) and 2 (Indian) are 46,XY male newborns with ambiguous genitalia (micropenis, penoscrotal hypospadias) who developed salt-losing since early infancy. They were stabilized with normal saline resuscitation and high dose hydrocortisone replacement. Patient 2 was initially misdiagnosed as 21-hydroxylase deficiency due to elevated 17OHP until he was referred for genitoplasty at the age of 2.5 years and the patient were re-evaluated. The ACTH tests revealed low cortisol response, moderately elevated 17OHP, elevated  $\Delta^5/\Delta^4$  steroids, suggestive of blockage at the level of enzyme  $3\beta$ HSD. Patients' leukocyte genomic DNA was extracted and the entire coding regions of the HSD3B2 gene were assessed by

polymerase chain reaction (PCR) and sequencing analysis.

## Results

Patient 1 was homozygous for T259M (c.776C>T) mutation in the HSD3B2 gene. Patient 2 was homozygous for the novel nonsense mutation Y180X (c.540C>A) and his parents were heterozygous carrier.

## Conclusion

We report the mutations of HSD3B2 gene, T259M and Y180X (novel) responsible for classic  $3\beta$ HSD deficiency. The clinical and hormonal phenotypes can be complicated in this disorder. These cases emphasize the importance of confirming the specific enzyme deficiency with molecular genetic analysis.

*Written informed consent was obtained from the patient for publication of this abstract and any accompanying images. A copy of the written consent is available for review by the Editor of this journal.*

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