Clinical Spectrum of Congenital Adrenal Hyperplasia due to 3-Beta-Hydroxysteroid Dehydrogenase Deficiency: A Case Series

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INTRODUCTION

- 3-beta-hydroxysteroid dehydrogenase type 2 ($3\beta HSD_2$) deficiency is a rare cause of congenital adrenal hyperplasia (CAH) in which synthesis of all active steroid hormone is impaired (Figure-1).
- Testosterone deficiency results in genital ambiguity in males (46,XY) at birth, however, females (46XX) are usually born with mild to moderate clitoromegaly.

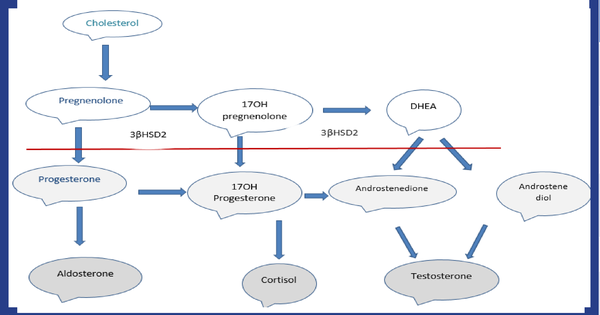

Figure 1 ;showing steroid hormonogenisis

AIM

- To determine the clinical spectrum of 3-beta-hydroxysteroid dehydrogenase type 2 ($3\beta HSD_2$) deficiency.

METHODS

- Retrospective review of all children with genetically confirmed $3\beta HSD_2$ presented at a tertiary care unit over a period of one year.

RESULTS

- Total 6 patients (4 males) from 3 different families of CAH due to $3\beta HSD_2$ deficiency c.818_819del p.(Lys273Argfs*7) were identified.
- All born to consanguineous parents with history of sibling death in two families (F1 and F2).
- Mean age of presentation was 21 (7-45) day of life.
- Failure to thrive was the most common presenting complain (n=6), followed by recurrent vomiting (n=5) and hypoglycaemia (n=1)
- Both female presented with normal female like genitalia with suspicion of mild clitoromegaly.
- 2/4 male born with penoscrotal hypospadiasis along with bifid scrotum, one with mid-shaft hypospadiasis and one with normal male genitalia.

	Case 1 (F1)	Case 2 (F1)	Case 3 (F1)	Case 4 (F1)	Case 5 (F2)	Case 6 (F3)
Age of presentation (Day of life)	7	15	8	31	20	45
Gender rear up at presentation	Female	Male	Male	Female	Male	Male
Presenting complaints	Vomiting FTT	Hypoglycaemia FTT	Vomiting FTT	Vomiting FTT	Vomiting FTT	Vomiting, FTT
Affected siblings	5	5	5	5	2	
Genital scoring Prader/EMS scoring	1 *mild clitoromegaly*	10 *mid-shaft hypospadiasis*	12 *normal male*	1 *mild clitoromegaly*	2 *microphallus, bifid fold, opening in perineum, gonads in folds*	2 *microphallus, bifid fold, opening in perineum, gonads in folds*
Karyotype	46,XX	46,XY	46,XY	46,XX	6,XY	46,XY
USS	Uterus + Ovaries+ Testes –	Testes +(scrotum) Uterus - Ovaries-	Testes +(scrotum) Uterus - Ovaries-	Uterus + Ovaries+ Testes -	Testes +(scrotum) Uterus - Ovaries-	Testes +(scrotum) Uterus - Ovaries-
Hyponatremia	Yes	Yes	No	Yes	Yes	Yes
Hyperkalemia	Yes	Yes	Yes	Yes	Yes	Yes
17-OH P (nmol/l)	63	56	252	762	45	968
DHEAS	13	08	15	15	09	13
Renin	128	180	150	500	350	500

CONCLUSION

- CAH patients due to $3\beta HSD_2$ deficiency can present with normal male or female genitalia.
- We need to have a strong suspicion of it in the region where there is increased consanguinity and no screening tests, to avoid preventable deaths due to this variant.

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