Management dilemmas in a genetically female child with congenital adrenal hyperplasia raised as a male P2-P77

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BACKGROUND

- Female infants with congenital adrenal hyperplasia (CAH) are conventionally reared as females, even if severely virilised at birth(1).
- Recently, male gender of rearing for such children, is receiving consideration (2).
- There is however, a lack of evidence-based guidelines to direct management in such instances.

OBJECTIVES

- To report on a 9-year-old genetically female child with salt wasting CAH reared as a male from birth, presenting with central precocious female puberty
- To describe current and future management issues and management strategies planned

CASE HISTORY

- Initial presentation in new born period with ambiguous genitalia (Prader stage 1V)
- Karyotype 46 XX, ultrasound scan showing both ovaries , infantile uterus and bilateral adrenal hyperplasia
- Elevated serum 17-OHP, hyponatremia and hyperkalemia
- Diagnosed as salt wasting CAH due to 21 hydroxylase deficiency
- Long term oral hydrocortisone and fludrocortisone therapy commenced
- Parental decision to rear baby as a boy, despite extensive counselling
- Hypospadias repair performed at 2 years of age
- Treatment compliance and follow up suboptimal
- At 8 ½ years of age, developed cyclical bleeding per urethra
- Bone age significantly advanced (14 years)
- Ultrasound scan –haematocolpus
- Elevated serum FSH and LH

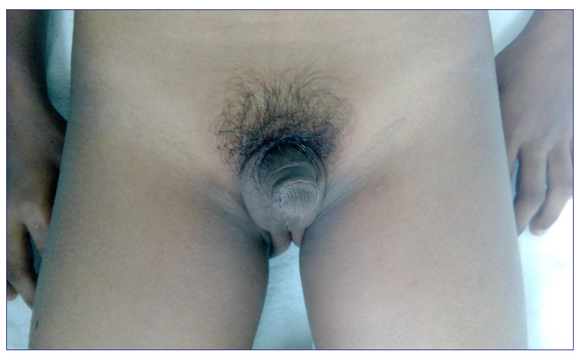

Figure 1- Image showing degree of genital virilisation in the child at 9 years of age

MANAGEMENT

- Psychological assessment- well adjusted male gender role
- Long acting GnRH agonists commenced to suppress puberty
- Hydrocortisone and fludrocortisone doses optimised
- Long term management discussed at specially convened multidisciplinary team meeting involving paediatric endocrinology, paediatric surgery and child psychology/ psychiatry teams and the family

LONG TERM PLAN

- Delay any **irreversible surgical interventions** until;
  - –Patient at least 18 years of age
  - –Capable of making an independent informed decision regarding future gender identity
  - –Legally of age to give informed consent for oophorectomy (if male gender decided upon)
- **Suppress female puberty** with GnRH analogs till such time
- Discuss further on possibility of initiating **male hormone replacement therapy**, and its timing, with the family, when peers enter puberty
- Provide long term **counselling and psychological support**
- Arrange appropriate **transition of care** to adult services in the future, utilising a multidisciplinary care approach

CONCLUSIONS

- Management of virilised 46 XX children with CAH, with a male gender of rearing is **complex and challenging**
- Establishment of **multidisciplinary care teams** and **consensus guidelines** on management would be beneficial to clinicians managing such complex patients and the families

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