OBJECTIVES

Introduction: Genital abnormalities are a source of concern and anxiety to parents and patients and in some cases, for the physicians who may have difficulty making pathological and eventually genetic diagnosis. They range from simple small penis and labial adhesions to the complex genital ambiguity and disorders of sex development.

Objectives: To determine the types of genital abnormalities presenting in the Paediatric endocrinology unit of the University of Port Harcourt Teaching Hospital and discuss the management and challenges faced during this period.

RESULTS

There were 31 children presenting with genital abnormalities of various kinds. Sex assigned to these children was 8 females and 23 males irrespective of complete pathological diagnosis. The median age of presentation was 13 months with a range of 0.1 – 168 months. The commonest diagnosis was micropenis (32.2%) with various forms of DSD being the second commonest (29%) and most females had labial fusion (16.1%). Females with labial fusion had complete resolution following oestrogen cream application, and 4 of the 7 children with DSD died during the period under review. hCG stimulation increased penile length of males with constitutional delay in puberty, who were reassured and only marginally in those with hypogonadotropic hypogonadism and Klinefelters syndrome.

DISCUSSION AND CONCLUSIONS

Making diagnosis and managing complex genital abnormalities like DSD in UPTH remain challenging because of lack of diagnostic equipment and drugs. Collaborating with European and other national laboratories help our team reach some pathologic diagnosis however genetic diagnosis is still difficult. Though some conditions seem simple to diagnose, managing the psychological consequences may not be as straightforward. Parents of children with DSD always ask for surgical intervention and with counselling, many reluctantly agree to postpone the surgeries. Hydrocortisone tablets for children with CAH is usually difficult to obtain so many have to use prednisolone. All parents were concerned about their children’s chances of fertility and some who were counselled on fertility preservation, were willing to assist their children undergo this process if needed.

References

