CONCLUSION

This case highlights the challenge of initiation of GH therapy, in a patient with a background history of recurrent craniopharyngioma and residual disease. Proper counselling with the patient and family is crucial to explain the clinical indications, risks and benefits of the GH therapy. A multidisciplinary approach of the management involving the paediatric endocrinologists, oncologists, neurosurgeons, radiologists, rehab physicians and dietitians together with close surveillance of primary disease are extremely important.

PA-P-10

MIXED GONADAL DYSGENESIS WITH ISODICENTRIC Y CHROMOSOMES: A CASE SERIES

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INTRODUCTION

Isodicentric Y chromosomes are formed by intrachromosomal recombination or fusion of sister chromatids following Y chromosome breakage.

CASES

CASE 1

A four-month-old male with ambiguous genitalia had a stretched penile length (SPL) of 2.4 cm, glandular hypospadias, palpable right gonad and empty left scrotum. External genitalia score (EGS) was 7 and external masculinization score (EMS) was 6.5. Investigation showed normal 17-OHP [33.04 nmol/L, reference value (RV) 12-36], ACTH (2.1 pmol/L, RV 1.6-13.9) and s erum cortisol (239.15 nmol/L, RV 145-619); and elevated renin (>550 mU/L, RV 4-89). He was in mini-puberty at three months, with LH 1.7 mU/mL, FSH 5.4 mU/mL, testosterone 3.14 nmol/L and anti-Müllerian hormone (AMH) 350.3 pmol/L (RV 235.5-1125.9). Ultrasonography showed a right testis with empty left scrotal sac and no Müllerian structures. Karyotype revealed 73% (45,X) and 27% (46,X idic{Y}) p11.2 with isodicentric chromosome Yq.

CASE 2

A four -month-old male presented with ambiguous genitalia, SPL 2.5 cm, perineal hypospadias, palpable right testis at the inguinal region, impalpable left testis, EGS 5.5 and EMS 5.5. Work-up showed normal 17-OHP (19.9 nmol/L) and serum cortisol (255 nmol/L); and elevated aldosterone (>3656 pmol/L) and renin (128.9 mU/L). Investigations post-delivery revealed mini-puberty with LH 6.59 IU/L, FSH 4.84 IU/L, testosterone 5.86 nmol/L and estradiol 43 pmol/L. AMH at 4 months was 435.8 pmol/L. Abdominal ultrasonography showed embedded penis with bilateral inguinal testes and no Müllerian structures. FISH with SRY gene probe revealed the first cell line (74.5%) of isodicentric chromosome Y and the second cell line (25.5%) of 45,X.

CONCLUSION

Patients with isodicentric Y chromosomes have various presentations necessitating follow-up to monitor growth, puberty, fertility, gonadal dysgenesis and short stature.

PA-P-11

CLINICAL FEATURES AND SHORT-TERM OUTCOMES OF CHILDREN WITH TURNER SYNDROME IN A CHILDREN'S HOSPITAL

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INTRODUCTION

Turner syndrome (TS) is the most common sex chromosome abnormality in females. This syndrome is usually diagnosed in females with characteristic features and a partial or complete absence of one X chromosome. We aimed to describe the clinical features and short-term outcomes of the children with TS being seen at our hospital.

METHODOLOGY

This is a descriptive study. Children with TS who attended the endocrine clinic in Sabah Women and Children's Hospital were enrolled. We obtained their pertinent data through a review of their case folders. Diagnosis of TS was confirmed via chromosomal study postnatally. Their clinical features and short-term outcomes were described.