PA-P-08

A CASE OF A RENIN-SECRETING TUMOUR IN AN ADOLESCENT: A RARE YET CURABLE CAUSE OF HYPERTENSION

https://doi.org/10.15605/jafes.037.S2.88

Meenal Mavinkurve,[1,2] Cheng Hooi Peng,[2,3] Man Kein Seong Mun Kein Seong,[4,5] Norshazriman Sulaiman,[5,6] Nurshadia Samingan,[2,5] Muhammad Yazid Jalaludin,[2,5] Azriyanti Anuar Zaini[2,5]

[1]*School of Medicine, International Medical University, Jalan Rasah, Seremban Negeri Sembilan, Malaysia*
[2]*Department of Paediatrics, University Malaya Medical Centre, Kuala Lumpur, Malaysia*
[3]*Department of Paediatrics, Hospital Putrajaya, Malaysia*
[4]*Department of Pathology, University Malaya Medical Centre, Kuala Lumpur, Malaysia*
[5]*Faculty of Medicine, University Malaya, Kuala Lumpur, Malaysia*
[6]*Department of Biomedical Imaging, University Malaya Medical Centre, Kuala Lumpur, Malaysia*

INTRODUCTION

Reninomas are benign tumours of the juxtaglomerular apparatus that autonomously secrete renin. Only 100 cases have been reported in the literature. Adolescents with reninomas typically present with refractory hypertension that requires treatment with multiple anti-hypertensives. However, hypertension secondary to reninomas are curable with surgery. Another, minimally invasive procedure, known as cryoablation has been successfully used to cure hypertension in an adult with reninoma, but this has not been reported in an adolescent.

CASE

We conducted a retrospective chart review of the pertinent clinical, biochemical, radiological and histopathological details. We report on a 14-year-old male with a hypertensive emergency; blood pressure 180/100 mmHg and Bell's palsy. His initial investigations showed hypokalaemia 2.2 mmol/L, metabolic alkalosis, raised plasma renin activity 2235 mU/L and aldosterone 8056 pmol/L, suggesting a high-renin mineralocorticoid excess syndrome. A right-sided renal cortical cyst was seen on abdominal computed tomography, measuring 0.9 x 1.6 cm. In order to accurately establish lateralisation of the autonomous renin secretion, renal vein sampling (RVS) was conducted to determine renin ratios, which confirmed lateralisation to the right renal vein (ratio 2.72). His hypertension was difficult to control despite amlodipine, prazosin and verapamil and captopril. There were no complications. Following adequate optimisation of his hypertension, he underwent cryoablation of the lesion. The histopathology was conclusive for a juxtaglomerular tumour. One week post ablation, he had resolution of his hypertension and normalisation of the plasma renin activity to 13.4 mU/L after 1 month.

CONCLUSION

Reninoma, though rare, should be considered in adolescents who present with a triad of refractory hypertension, hypokalaemia, and metabolic alkalosis. It is a curable with surgery, but cryoablation should be given due consideration. This case report illustrates that cryoablation can be used successfully for the management of reninoma in adolescents.

PA-P-09

A CASE OF RECURRENT CRANIOPHARYNGIOMA POST-OPERATIVE WITH RESIDUAL DISEASE AND GH DEFICIENCY

https://doi.org/10.15605/jafes.037.S2.89

Ahmad Fahmi Bin Abdullah Asuhaimi, Mazidah Binti Noordin, Noor Shafina Binti Md Noor

Hospital UiTM, Puncak Alam, Selangor, Malaysia

INTRODUCTION

Craniopharyngioma is an uncommon intracranial tumour in childhood. Even though it is a benign tumour, recurrence of disease may occur which is commonly complicated with endocrinopathy. We present a case of recurrent craniopharyngioma post-resection with residual tumour complicated with multiple pituitary hormone deficiencies, including growth hormone (GH) deficiency.

CASE

A 7-year-8-month-old male presented with persistent headache and symptoms of increased intracranial pressure. MRI Brain showed suprasellar cystic mass. He underwent total resection of the tumour. HPE confirmed craniopharyngioma. He developed central diabetes insipidus, central hypothyroidism and ACTH deficiency post-operatively. Eleven months later, he presented with blurring of vision and increased sleepiness. Brain MRI confirmed recurrence of the tumour. Near-total-excision of the tumour was done as the tumour was adhered to the optic nerve and chiasma. After the surgery, he was under close surveillance for recurrence of disease. Annual MRI Brain surveillance showed stable residual disease.

The patient is currently 12-years-old. Apart from the endocrinopathies mentioned, he is now showing signs of growth hormone deficiency such as hypothalamic obesity with weight BMI at +3.35SDS. He has poor height velocity at 3 cm/year. He has metabolic syndrome including dyslipidaemia, and fatty liver. He also has delayed bone age and poor IGF-1 level. Family counselling was done to explain the role of GH therapy for him, including the risks and benefits.

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

https://doi.org/10.15605/jafes.037.S2.90

L Alexis Anand, Nalini M Selveindran, Jeanne Sze Lyn Wong, Janet Yeow Hua Hong
Paediatric Endocrine Unit, Hospital Putrajaya, Malaysia

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

https://doi.org/10.15605/jafes.037.S2.91

Pamela Lee Ling Tan and Song Hai Lim
Paediatrics Department, Sabah Women And Children's Hospital Likas, Malaysia

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.