EP_P014

CHALLENGES OF INITIATING GROWTH HORMONE THERAPY IN TURNER SYNDROME WITH CHIARI MALFORMATION: A FOLLOW UP REPORT

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INTRODUCTION/BACKGROUND

Turner syndrome (TS) associated with congenital central nervous system abnormalities are uncommon. Study on outcome of growth hormone therapy in this group are limited. We present a follow up report on challenges of growth hormone (GH) therapy in our patient with Turner Syndrome (45, XO) and Type I Arnold Chiari Malformation.

CASE

NB is a 17-year-old female who was first referred to us for short stature and dysmorphism. Karyotype confirmed 45, XO, hence, TS diagnosis was made. She was pre-pubertal at presentation, with height of 132 cm (-4.98 SDS), and mean parental height of 153 cm. Her bone age was delayed at 12.5 years.

She was planned for GH therapy. Polysomnography (PSG) revealed an incidental finding of central apnoea with Apnoea-Hypopnea Index (AHI) of 22.5/H. This led to a brain MRI which revealed cerebellar tonsils descended 7 mm below the foramen magnum, consistent with Type I Arnold Chiari malformation. A repeat PSG showed significant improvement of her AHI index. Following multidisciplinary discussion, a decision was made for GH therapy.

She was started on GH therapy and the dosage was gradually titrated. After 3 months of GH therapy, her height velocity improved from 2.7 cm/year to 5.2 cm/year. She remained well, however a PSG post-GH therapy revealed worsening apnoea. Family counselling was done, and her parents were not keen to continue with GH therapy.

CONCLUSION

TS with Chiari malformation is uncommon. Commencement of GH therapy in this group requires multidisciplinary management. Close surveillance of potential side effects is crucial to avoid potential adverse events.

EP_P015

A CASE OF NON-CLASSICAL CONGENITAL ADRENAL HYPERPLASIA (NCCAH) PRESENTING WITH CENTRAL PRECOCIOUS PUBERTY

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INTRODUCTION/BACKGROUND

Non-classical congenital adrenal hyperplasia (NCCAH) usually has a late presentation as compared to classical CAH as the presentation is often atypical. Presentation in boys can be asymptomatic through family screening or with clinical presentation such as signs of premature adrenarche and growth spurt.

CASE

MFI, a 4 year-4-month-old male was referred to the paediatric endocrine clinic with complaints of body odour for 2 months, associated with pubic hair and acne. He seemed to be taller compared to his peers with a recent height spurt. His parents are not consanguineous with no significant family history. He has facial acne with signs of precocious puberty. He has Tanner II genitalia with stretched penile length of 7 cm, presence of pubic hair, and bilateral testicular volume of 4 ml.

Baseline investigations revealed low cortisol level, elevated 17-hydroxyprogesterone (17OHP) and detectable testosterone levels. Short synacthen test showed low cortisol level, with significantly elevated 17-OHP with a peak of 931 nmol/L. Both sodium and potassium were normal, however, renin was elevated at 166.5 mU/L (normal range 4.2 – 59.7) with aldosterone at a lower range of normal at 155 pmol/L (normal range 102 – 859). LHRH test showed pubertal respond with peak LH at 11.4 IU/L. Bone age was 7 years more advanced than his age, at 11.4 years. Genetic test for CAH panel was not performed due to financial constraint. Diagnosis of NCCAH with central precocious puberty was made and hydrocortisone, fludrocortisone and GnRH analogue were started.

CONCLUSION

This case illustrates an unusual presentation of a male patient with NCCAH complicated by central precocious puberty due to androgen excess that has activated the

central pubertal response. Treatment aims to control the CAH as well as suppress puberty with GnRH analogue, with the hope of preserving the patient's final adult height.

EP_P016

A CASE OF AUTOSOMAL DOMINANT OSTEOPETROSIS TYPE 2 WITH A CLCN7 GENE MUTATION

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INTRODUCTION/BACKGROUND
Osteopetrosis encompasses a group of rare metabolic bone diseases characterized by impaired osteoclast activity or development, resulting in high bone mineral density making affected individuals paradoxically prone to fracture. The autosomal dominant adult (benign) form is associated with milder symptoms often appearing in later childhood and adulthood whereas the autosomal recessive infantile (malignant) form has severe presentations appearing in early childhood, and typically fatal if untreated.

The CLCN7 gene plays a pivotal role in the pathogenesis of various forms of osteopetrosis. Disruption of CLCN7 expression results in severe lysosomal storage disorders.

CASE
We present a 14-year-old female who was referred to the Paediatric Endocrine clinic, Hospital Putrajaya (Malaysia) for history of recurrent fractures after trivial insult. She had sustained a closed fracture of the left distal end of the tibia/fibula at age 7 years and bilateral distal third tibia/fibula at age 13 years. She also complained of lower back and lower limb pain after exertion. He had normal dentition and mild bowing of both lower limbs, with no organomegaly. Ophthalmology and ENT assessments were normal. There was no remarkable abnormality in serum biochemistry, besides Vitamin D deficiency and elevated aspartate aminotransferase (AST). Plain radiographs showed a generalized increase in bone density. Whole exome sequencing confirmed a heterozygous likely pathogenic variant of the CLCN7 gene (CLCN7 856C>T). There is a history of fracture following trivial insult in a sibling. Genetic testing for the family will be sent in the future. The patient was started on vitamin D supplementation and advised adequate dietary calcium intake.

CONCLUSION
In conclusion, we report a patient with osteopetrosis due to a novel mutation of the CLCN7 gene. Management of the less severe forms of osteopetrosis is not so clear as expert guidelines have focused on treatment of the severe infantile forms which require hematopoietic cell transplantation. Scarcity of published studies on osteopetrosis precludes the development of evidence-based guidelines for the management of these patients.