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.

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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.