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Hypogonadotrophic Hypogonadism – A Case Report

https://doi.org/10.15605/jafes.034.S114

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INTRODUCTION

Hypogonadotrophic hypogonadism (HH) is a rare condition usually affecting the males. The estimated incidence is 1-10:100000 live births. Common presentations are infertility and delayed puberty.

CASE

We report a case of HH who has been followed up for the past 17 years since birth. The clinical courses including presenting features, diagnosis and management will be described.

The patient was referred for micropenis with left retractile gonad at birth. Initial investigations showed a normal male karyotype of 46XY, low testosterone level and normal renal ultrasonography. Gonadotrophins were not available. There was history of micropenis in the maternal family member. He was subsequently followed up as outpatient for under-virilised male. At the age of 4, left orchidopexy was done. MRI pituitary was done to rule out central defect and noted to be normal. Human chorionic gonadotrophin stimulation test was done and showed poor testicular response. His growth parameter has always been within the normal centile. Other pituitary hormones were normal. At 12 years old, he was noted still pre-pubertal. Gonadotrophin releasing hormone test revealed a poor gonadotrophin response and diagnosis was made. Retrospectively, he did report poor sense of smell. He was started on intramuscular testosterone therapy at 12.5 years. Puberty progressed with induction and he achieved stage five of Tanner staging at 15 years old. However, his testes remained small and penile length is less than 2.5 standard deviation below the adult mean.

CONCLUSION

This is likely a congenital HH which involves lifelong treatment and monitoring. Early diagnosis allows better counselling and preparation for the family and patient throughout the course of disease.

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Case Report: Pamidronate Infusion in a 3-Month Old Infant with Osteogenesis Imperfecta

https://doi.org/10.15605/jafes.034.S115

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INTRODUCTION

Bisphosphate is a well-recognised treatment for children with osteogenesis imperfecta (OI). However, for neonatal-onset or young infants, there are no large groups reported and no clear guideline on the safest dosing regimen.

CASE

Our patient presented with a left femur fracture at the age of 1.5 months. A week later, she was diagnosed with severe OI when she suffered four more fractures despite minimal handling; bilateral humerus, left clavicle and right femur. Her sclerae had a tinge of blue at first review. She was started on pamidronate two-monthly cycle (regimen as below) at 3 months old after the fractures had healed. Prior to pamidronate, she was started on cholecalciferol with optimisation of vitamin D level>75 nmol/L. 1st cycle 0.25 mg/kg for 1 day, 2nd cycle 0.25 mg/kg for 3 days, Subsequent cycles 0.5 mg/kg for 3 days every 2 months (total dose 9 mg/kg/year). For the 1st cycle of pamidronate, she had transient hypophosphatemia and mild hypocalcemia 2.02 mmol/L post-infusion, easily corrected with oral calcium carbonate and calcitriol. For the 2nd cycle, she was also given a short course of oral calcium carbonate and calcitriol as post-infusion her calcium level was borderline at 2.1 mmol/L. Subsequently for her 3rd and 4th cycle, her calcium levels were stable post-infusion with no need for additional supplements.

In total, she received 4 cycles 2 monthly apart (total cumulative dose 3.8 mg/kg) and did not had any recurrent fractures since treatment. There had also been a marked improvement in her gross motor development and mobilisation.

CONCLUSION

We report a young infant with OI who showed a good short-term outcome with pamidronate treatment and tolerated well our dosing regimen.