

Adult E-Poster

CONCLUSION

Permanent central DI occurs when the hypothalamus and/or pituitary stalk is irreversibly injured. It is important to monitor DI in post pituitary surgery. One of the diagnostic tests used is WDT.

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PYCNODYSOSTOSIS IN A YOUNG ADULT PRESENTING WITH FRAGILITY FRACTURE AND HIGH BONE MINERAL DENSITY: A CASE REPORT

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INTRODUCTION/BACKGROUND

Pycnodysostosis is a rare skeletal dysplasia and a subtype of osteopetrosis caused by deficient activity of the lysosomal protease cathepsin K (CTSK) gene. We present a case of pycnodysostosis exhibiting characteristic clinical features alongside a fragility fracture—an association that has been rarely reported.

CASE

A 33-year-old Malay female, born of a consanguineous marriage, presented with a subtrochanteric fracture of the proximal left femur following a trivial fall and underwent open reduction with locking compression plate fixation. She was noted to have a short stature (height: 145 cm). Further examination revealed frontal bossing, maxillary hypoplasia, and brachydactyly of both hands and feet. Other systemic examinations including neurological assessment were unremarkable. A family history of similar physical traits was noted in her late paternal grandmother, though medical records were unavailable. Her elder sister exhibited similar abnormalities. Laboratory investigations showed a low insulin-like growth factor 1 (IGF-1) level, but her insulin tolerance test was normal, excluding growth hormone deficiency. Other blood parameters, including complete blood count, serum calcium, phosphate, 25-hydroxy vitamin D, alkaline phosphatase, thyroid function tests, and cortisol were within normal limits. Bone mineral density (BMD) analysis revealed an elevated Z-score of +5.5 at the total hip and +0.9 at L1-L4. Genetic analysis identified a homozygous variant of uncertain significance in the CTSK gene, which is associated with autosomal recessive pycnodysostosis. Given the lack of a specific treatment, symptomatic management was initiated, focusing on fracture prevention, oral hygiene, and psychological support.

CONCLUSION

This case underscores the need to consider rare genetic skeletal dysplasias like pycnodysostosis in individuals presenting with fragility fractures, distinct craniofacial and skeletal features, and unusually high bone mineral density. Prompt recognition, aided by genetic testing, is crucial for guiding patient counseling, monitoring, and coordinated multidisciplinary care. Although no curative treatment exists, early diagnosis can help reduce complications and support a better quality of life.

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A SURPRISING TWIST: HYPONATREMIA INDUCED BY ZOLEDRONIC ACID – A RARE CLINICAL PUZZLE

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INTRODUCTION/BACKGROUND

Zoledronic acid is a potent bisphosphonate commonly used in the management of severe hypercalcemia, particularly in cases related to malignancy or metabolic bone disease. While hypocalcemia is a well-documented side effect following zoledronic acid administration especially in patients with primary hyperparathyroidism, there are no widely reported instances of hyponatremia in this patient population. We present a unique case of significant hyponatremia associated with zoledronic acid in a patient with primary hyperparathyroidism.

CASE

A 58-year-old female with a background of severe hypercalcemia secondary to primary hyperparathyroidism complicated by nephrocalcinosis and severe osteoporosis presented with recurrent hospital admissions due to severe hypercalcemic episodes. Over a two-month period, she received three doses of intravenous zoledronic acid infusion, followed by a single subcutaneous injection of denosumab. During follow-up in the clinic, laboratory investigations revealed significant hyponatremia (sodium: 113 mmol/L) and hypophosphatemia (phosphate: 0.52 mmol/L). There was no glycosuria, no hypokalemia, no hypouricemia and venous blood gas was not acidotic which excludes Fanconi syndrome. She was asymptomatic and denied any episode of vomiting or diarrhea. Secondary causes of adrenal insufficiency, syndrome of inappropriate antidiuretic hormone secretion (SIADH), hypothyroidism and diuretic use were excluded. With