PP-B-09

BRITTLE BONE DISEASE BECOMES UNBREAKABLE WITH BISPHOSPHONATE INFUSION

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CASE
Osteogenesis imperfecta is a lifelong inheritable disease and currently has no definitive cure. A 32-year-old female born to a non-consanguineous couple of Filipino descent consulted for chronic back pain. The patient had a history of recurrent fractures on low-intensity trauma starting from her toddler years. She had an unremarkable family history and prenatal and perinatal courses. Physical examination noted short stature with greyish blue scleral hue, triangular face, no bowing of upper and lower extremities, and positive Adam's forward bending test. Laboratory results showed normal serum levels of calcium, phosphorus, vitamin D, and PTH. Spine imaging showed thoracolumbar dextrolevoscoliosis. The patient was clinically diagnosed with Osteogenesis Imperfecta type I and was handled using a multidisciplinary approach composed of physical therapy, surgical interventions, genetic counseling, and bone-targeted therapy. Medical management was done using bisphosphonate therapy for 3 doses. Currently, the patient has minimal back pain with no recurrence of fracture and the latest bone densitometry values are within the expected range for age.

KEYWORDS
osteogenesis imperfecta, bisphosphonate, brittle bone disease

PP-B-10

TRANSIENT HYPERPHOSPHATASEMIA IN CHILDREN TREATED WITH GRISEOFULVIN

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CASE
The hallmark of transient hyperphosphatasemia (TH) is an elevation of serum alkaline phosphatase (ALP), which resolves within a few weeks or months without evidence of liver or bone disease. Despite its unknown etiology, it is frequently found in children under the age of five following an acute viral infection. We report a 7-year-old male who developed TH after receiving griseofulvin treatment for tinea capitis. He complained of fever and generalized erythematous rashes two weeks after treatment. Because of a possible drug allergy, blood tests were evaluated. Liver function showed normal transaminase and bilirubin levels, but the ALP was extremely high (2,657 IU/L). His serum calcium, phosphorus, PTH, and vitamin D were normal. Griseofulvin or viral exanthem were suspected of causing hyperphosphatasemia. Because his scalp lesion was worsening, itraconazole was substituted. After two months of monitoring, his ALP returned to normal. As a result, TH was diagnosed. Therefore, awareness of this event may prevent unnecessary investigations.

KEYWORDS
transient hyperphosphatasemia, hyperphosphatasemia

PP-B-11

SUN EXPOSURE AND VITAMIN D STATUS AND ITS ASSOCIATION WITH BONE TURNOVER MARKERS IN TRANSFUSION-DEPENDENT ADULT THALASSEMIA PATIENTS

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INTRODUCTION
Bone disease in patients with thalassemia, encompassing both osteopenia and osteoporosis, is on the rise, partly due to improvements in treatment and survival rates. Bone metabolism in thalassemia is influenced by many confounding factors including hormonal abnormalities, vitamin D deficiency, and iron load from multiple blood transfusions. Bone turnover markers are potentially useful as non-invasive tests to assess bone remodeling in this high-risk population. Due to the increasing frequency of thalassemia bone disease and limited data on vitamin D status among thalassemia patients in countries with adequate sun exposure, this study aimed to assess vitamin D status and its association with sun exposure, bone turnover markers (BTM) and ferritin in adult transfusion-dependent thalassemia patients in Malaysia.

METHODOLOGY
This cross-sectional study involved transfusion-dependent thalassemia patients (n = 40) from the adult hematology clinic. All participants had anthropometric measurements, sun exposure index (SI) measured by calculating body-surface-area x hours of exposure/week, as well as a complete biochemical profile including ferritin, calcium

and phosphate levels, 25-hydroxyvitamin(OH)D level, and bone turnover markers serum P1NP and serum CTX as bone formation and bone resorption markers respectively. Vitamin D deficiency was defined as 25-hydroxyvitamin D of less than 75 nmol/L. Vitamin D inadequacy was a combination of vitamin D deficiency and insufficiency.

RESULTS

The study population included 47.5% female and 52.5% male subjects with a mean age of 27.5 ± 5.2 years. Almost all (95%) had elevated serum ferritin (>1000 ug/l) and various endocrinopathies. Increased BTM was detected in 27.5% of patients (n = 11). A high prevalence of vitamin D inadequacy (95%) was observed, with vitamin D insufficiency of 32.5% and deficiency of 62.5%, correlating with ferritin levels (r = -0.444, p = 0.005) and serum P1NP (r = -0.364, p = 0.024). A majority (72.5%) had inadequate sun exposure, particularly among the females (p = 0.021) and Malays (p = 0.003). There was no significant correlation between SI and vitamin D status (r = 0.037, p = 0.824) or BTM.

CONCLUSION

This study revealed a high prevalence of vitamin D inadequacy among adult transfusion-dependent thalassemia patients and low sun exposure among females and Malays. Vitamin D inadequacy was associated with high ferritin and bone formation markers reflecting increased bone remodeling which can lead to higher fracture risk due to bone fragility. Hence, it is important to recognize and treat vitamin D deficiency early in these patients to prevent its deleterious effects on bone health.

KEYWORDS

bone disease, thalassemia, bone turnover markers, vitamin D, sun exposure

PP-B-12

A CASE OF MALABSORPTION PRESENTING WITH OSTEOMALACIA, COAGULOPATHY AND DELAYED PUBERTY

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CASE

A 26-year-old Filipino female presented initially with hematochezia. At the age of 2, following blunt abdominal trauma, the patient underwent intestinal bypass surgery (gastrojejunostomy). Thereafter, she experienced recurrent abdominal pain and diarrhea, with poor weight gain and short stature. Later, she would report delayed puberty (breast budding at 16; menarche at 18). She also reported bone and joint pain coupled with facial and extremity paresthesias, with X-rays revealing signs of osteopenia. Development of hematochezia resulting in severe anemia, in association with recurring gastrointestinal symptoms, prompted admission. Workup revealed elevations in prothrombin time, which improved following Vitamin K administration. GI endoscopy revealed no structural lesions. Skeletal X-rays revealed generalized decreases in mineralization, with lateral views showing concaving fish-mouth deformities in the L1 to L5 vertebral bodies. Vitamin D levels were found to be deficient; this improved only following large doses of daily Vitamin D3 administration. Hormonal studies revealed hypogonadotropic hypogonadotropism, likely stemming from malnutrition.

KEYWORDS

osteomalcia, coagulopathy, vitamin D, delayed puberty

PP-B-13

NAVIGATING THE DIAGNOSTIC CHALLENGES OF CALCIPENIC RICKETS COMPLICATED BY HYPERCOAGULATION, HYPOKALEMIA, AND SECONDARY AMENORRHEA IN A 21-YEAR-OLD FEMALE

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CASE

Rickets, a metabolic bone disorder primarily attributed to insufficient mineralization of the epiphyseal plate or osteoid calcification failure due to vitamin D or calcium deficiency, encompasses a spectrum of etiological factors including genetic anomalies and drug-induced manifestations. This report details a case of calcipenic rickets in a 21-year-old Indonesian female who presented with growth retardation and leg deformities since age 13. Extensive evaluation revealed hypocalcemia, hyperparathyroidism, elevated alkaline phosphatase, reduced phosphate levels, and decreased vitamin 25(OH)D concentrations. Alongside calcipenic rickets, the patient exhibited hypercoagulability, hypokalemia, and secondary amenorrhea. Treatment comprised Vitamin D supplementation (5000 IU) and calcium supplementation. Early diagnosis, guided by history, physical examination, and laboratory and radiological assessments, is essential. The profound impact of rickets on stature and bone structure underscores the urgency of timely diagnosis and appropriate intervention to ensure optimal outcomes for affected individuals.

KEYWORDS

rickets, vitamin D, diagnosis