



RESULTS

A total of 1381 participants were included in the study, with the majority being female. There was strong correlation between FRAX+BMD and FRAX-BMD for both MOF ($r = 0.889$, $p < 0.001$) and HF ($r = 0.796$, $p < 0.001$). Concordance of 80.1% ($p < 0.001$) was seen in treatment recommendation between FRAX+BMD and FRAX-BMD (treatment recommended $n = 505$; no treatment recommended, $n = 601$). Concordance was highest in the youngest and eldest age groups with 91.1% and 85.8%, respectively. Among the discordant, FRAX-BMD underestimated treatment recommendation in 147 (10.6%) and overestimated in 128 (9.2%) participants. Age was the sole important predictor of discordance in treatment recommendations comparing both groups. FRAX-BMD had the least underestimation of treatment among the 80 to 90-year-old group (0.9%) and least overestimation in the 40 to 69-year-old group (1.2%).

CONCLUSION

FRAX-BMD had a good correlation with FRAX+BMD in a Malaysian population and is an acceptable alternative for treatment decision-making in situations where BMD services are not readily available.

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DILEMMAS IN THE DIAGNOSIS AND MANAGEMENT OF OSTEOPOROSIS IN A PATIENT WITH ALKAPTONURIA: SUCCESSFUL TREATMENT WITH TERIPARATIDE

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BACKGROUND

Management of osteoporosis in patients with alkaptonuria can be challenging. We report a patient with alkaptonuria who was successfully treated with teriparatide.

CASE

Laboratory and DXA were completed at our hospital.

A 69-year-old female with a diagnosis of alkaptonuria came in for osteoporosis follow-up. Following the diagnosis of multiple joint arthritis, she underwent several joint replacement surgeries. She also sustained fragility fractures in the foot. Physical exam revealed bluish discoloration of the conjunctiva, normal S1, split S2 and IV/VI systolic murmur over the right parasternal border. She also had limited mobility of the thoracic and lumbar spines, wrists, ankles, knees and hip joints. Laboratory examination revealed the following results: serum PTH 33 pg/mL, 25-OH vitamin D 28 ng/mL, osteocalcin 12 mg/mL, C-telopeptide 318 pg/mL, tyrosine 79.1 umol/L, 24-hour urine homogentisic acid 4.2 gms. Genetic testing showed compound heterozygous mutation for the HGD C. 496T2T>C and HGD C.1102A < G (p.mev368Va) variants, consistent with a diagnosis of alkaptonuria. DXA scan done at the age of 56 years showed osteoporosis (T score of -2.7 over femoral neck, -2.5 over total hip). She was treated with alendronate for 5 years in addition to nitisinone. While on alendronate, she sustained fragility fractures of the right radius and left ankle. After 5 years of alendronate, the patient was transitioned to teriparatide 20 mcg subcutaneously daily for 2 years, followed by annual intravenous zoledronic acid. For the subsequent seven years, the patient led an active life with no fractures. Follow-up DXA showed improvement to osteopenia at the radius. The presence of degenerative arthritis made the other sites difficult to interpret.

CONCLUSION

In this subset of patients, bisphosphonates are not as effective in preventing fragility fractures. However, teriparatide has shown some promise as an alternative treatment.