primary bone disorders, while another 8 subjects (50.0%) had exposure to glucocorticoids for the management of various condition including systemic lupus erythematous (2 subjects, 12.5%), juvenile dermatomyositis (2 subjects, 12.5%), juvenile idiopathic arthritis (1 subjects, 6.3%), ANCA vasculitis (1 subject, 6.3%), autoimmune hepatitis (1 subject, 6.3%) and acute lymphoblastic leukaemia (1 subject, 6.3%). Only half of the subjects elicited adequate dairy consumption and took vitamin D supplements in the form of cholecalciferol or alfacalcidol. Sedentary lifestyle was observed in two thirds of the subjects. The physical stigmata of bone fragility disorders were present in 4 patients, and they had genetic confirmation of osteogenesis imperfecta. Five subjects (31.3%) had fracture of long bones, as well as osteoporosis. Bone-active therapy with bisphosphonate had commenced in three patients. Serum 25-hydroxy vitamin D and parathyroid level were examined in 5 subjects and 2 subjects were detected to have vitamin D deficiency. Four subjects (25.0%) displayed vertebral fractures. Overall, the mean areal bone mineral density Z- scores were -2.78 ± 1.74 for hip, -1.87 ± 1.71 for lumbar spines and -3.07 ± 2.16 for total body less head.

CONCLUSION

Bone health screening among the children and adolescents vulnerable to osteoporosis should be imparted as the standard of care.

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"A GIRL, WITHOUT UTERUS OR VAGINA:" A CASE REPORT OF MAYER-ROKITANSKY-KUSTER-HAUSER SYNDROME

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INTRODUCTION/BACKGROUND

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare congenital disorder that affects female reproductive organs and is often only diagnosed during adolescence or early adulthood. It is estimated to affect at least 1 out of 4500 women and it remains poorly investigated and underreported.

CASE

A 4-year-old child, raised as a girl was referred to the clinic for atypical genitalia. She was born premature at 28 weeks of gestation with a birth weight of 1.22 kg (between 50th to 90th centile). Her parents were not consanguineous and there was no family history of disorder of sexual development. On examination, there was no hyperpigmentation of the genitalia. There was clitoromegaly, with the size of 26 x 12 mm, urethral meatus was seen at the base of the phallus, with no vaginal opening. The labioscrotal folds were not rugated. There were palpable masses at the bilateral inguinal region suggestive of gonads.

Chromosomal study done revealed 46, XX. Baseline hormonal workup including 17-OH progesterone and cortisol were normal. Testosterone was not detectable. Pelvic MRI pelvis was suspicious of MRKH syndrome, as the only visualized Mullerian structures present were rudimentary uterus and bilateral ovaries. Both cervix and upper vagina were not visualized. Apart from that, there were bilateral cystic lesions seen at the inguinal region most likely consistent with canal of Nuck cyst. Patient has also been referred to both genetic and surgical team for further management.

CONCLUSION

MRKH syndrome is usually diagnosed later in life. Early radiological imaging aids earlier diagnosis. This condition requires multidisciplinary management that can help both the patient and their family to cope with this uncommon condition, including the psychological and physiological consequences.