forehead, wide wide-spaced nipple, and no blue sclera. His respiratory assessment was normal. His skeletal survey reported generalised osteopenia with bowed limbs and plastic fracture over the right femur consistent with osteogenesis imperfecta. He had normal serum calcium, persistently low ALP (18-25 IU/L), iPTH: 69.3 pg/ml (14.9-56.9) and 25 (OH) vit D3: 51.70 nmol/L (insufficient). There was no family history of bone diseases and parents were not consanguineous. His genetic results showed heterozygous, autosomal recessive, likely pathogenic, ALPL (NM_000478.6), Exon 2, c.29T>C, p. (Ile10Thr), and ALPL (NM_000478.6), Exon 9, c.991G>A, p. (Val331Met). He continued to improve without treatment and was last reviewed at 5 months old, with appropriate development and no fracture.

CONCLUSION

There are very few cases of hypophosphatasia reported locally. It is important to highlight the differential as compared to osteogenesis imperfecta. Hypophosphatasia could present differently, with our case reflecting the benign perinatal type, whereas others would require enzyme replacement, or asfotase-alfa as the first medical treatment for perinatal, infantile and juvenile onset of the disease.

EP_P017

CONGENITAL HYPOTHYROIDISM WITH DIFFERING PHENOTYPE IN TWO SIBLINGS WITH THE SAME DUOX2 MUTATION

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INTRODUCTION/BACKGROUND

Congenital hypothyroidism (CH) is caused by thyroid gland structural or functional defects. Thyroid dysgenesis is typically sporadic but dyshormogenesis is frequently inherited as autosomal recessive. Dual oxidase 2 (DUOX2) gene mutation causes abnormal iodide organification in thyroid hormone synthesis.

We report 2 siblings with CH sharing the same mutation in DUOX2, however only one had permanent CH.

CASE

A 14-year-old female had CH detected at birth by high cord TSH (>100 m IU/L), and thyroid function test (TFT) on day 3 of life (TSH >100 m IU/L, free T4 8.57 pmol/L). Thyroid replacement was started at a dose of 8 mcg/kg/day. Ultrasound at age 3-year-old showed a hypoplastic thyroid gland. She had permanent CH as TFT was deranged when thyroxine was withheld at age 3. Her younger brother, now aged 9 years, had CH diagnosed at age 1 month when he presented with prolonged jaundice (TSH 56.4 m IU/L, free T4 8.89 pmol/L). His cord TSH was normal (7.18 m IU/L). Thyroxine replacement was started at a dose of 7 mcg/kg/day. Ultrasound at age 3 showed a normal-sized thyroid gland. Thyroxine was stopped at age 3, and subsequent TFT remained normal, indicating transient CH. Both had no goiter, nor comorbidity. No other family members had a thyroid disorder. Parents were non-consanguineous. Whole exome sequencing (3Billion, South Korea) revealed both siblings had the same pathogenic heterozygous DUOX2 mutation (c.3329G>A). It was inconclusive as the second abnormal allele was not detected.

CONCLUSION

The same DUOX2 mutation can have different phenotypes within the same family. Genetic testing has a role in evaluating CH etiology, especially when at least two family members are affected. Newborn siblings of a child with CH need timely monitoring of TFT if cord TSH is normal. Other genetic methods are needed to detect the second variant of DUOX2 in these siblings.

EP_P018

BONE HEALTH SURVEILLANCE AMONG AT-RISK CHILDREN AND ADOLESCENTS IN KUCHING, SARAWAK, MALAYSIA

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INTRODUCTION/BACKGROUND

Chronic health conditions impose poor bone health due to underlying inflammatory conditions, reduced weight-bearing activity and pubertal delay.

METHODS

This prospective study was conducted as the pilot project for bone health surveillance among at-risk children and adolescents followed up by a multidisciplinary team from Paediatrics Department, Sarawak General Hospital from January to February 2024. A bone health screening questionnaire was administered, followed by a physical examination, and biochemical and radiological investigation.

CASE

A total of sixteen subjects (6 males, 10 females) with a mean age of 10.7 ± 2.74 years were recruited. Four subjects had

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.

EP_P019

"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.