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