CONCLUSION
Genetic mutations in children with NDM are common. Insulin remains the mainstay of therapy in INS-gene mutation. Genetic testing should be done to facilitate management.

EP_P003

ZOLEDRONIC ACID THERAPY FOR MONO-OSTOTIC LANGERHANS CELL HISTIOCYTOSIS: A CASE REPORT

https://doi.org/10.15605/jafes.038.S2.132

Pian Pian Tee[1,2] and Wei Li Cindy Ho[1]
[1]Khoo Teck Puat – National University Children's Medical Institute, National University Health System, Singapore
[2]Hospital Putrajaya, Putrajaya, Malaysia

INTRODUCTION/BACKGROUND
Langerhans cell histiocytosis (LCH) is a rare disease. It can affect any organ in the body but is primarily characterized by osteolytic bony lesions. Skeletal LCH may range from a unifocal, self-limiting, asymptomatic lesion to severe, painful, destructive lesions that are prone to pathological fractures. Treatment decisions are individualized according to location, size, surgical accessibility, and functional impairment. Hence, there is no standard of care at the moment. Zoledronic acid (ZA) has been used in some neoplastic bone conditions to slow down the progression and reduce the bone pain.

CASE
We report a 6-year-old male with unifocal bony LCH at the left tibia who responded well to ZA. He presented at 4 years old with limping and was subsequently not ambulatory due to severe pain. He had a tender swollen left shin without skin changes.

X-ray showed a poorly defined 4.7 x 1.1 cm permeative lytic lesion in the medullary cavity of the midshaft of the left tibia with endosteal thinning. Subsequent MRI and isotope bone scans (Tc-99m MDP) confirmed a suspicious primary bone malignant lesion. A bone biopsy showed a neoplastic proliferation of histiocytoid cells with strong diffuse positivity for CD 1a, which was in keeping with LCH.

This confirmed a symptomatic mono-ostotic LCH with significant cortical destruction that was at risk of fracture. ZA was initiated after careful evaluation. His pain completely resolved with a return of function six weeks after the first dose of ZA. He received 4 doses of ZA in total and demonstrated radiographic evidence of regression and remained in remission.

CONCLUSION
This case demonstrates the potential role of ZA therapy as the first line treatment for mono-ostotic LCH stabilisation and symptomatic control.

EP_P004

CASE REPORT: MUCOLIPIDOSIS BONE DISEASE RESEMBLING NEONATAL RICKETS

https://doi.org/10.15605/jafes.038.S2.133

Jian Ann Boo, Sze Teik Teoh, Eric Boon Kuang Ang
Hospital Sultanah Bahiyah, Malaysia

INTRODUCTION/BACKGROUND
Mucolipidosis II (I-cell disease) is a rare genetic metabolic disorder of lysosomal metabolism with a combined frequency of 1:422,000, that is characterized by coarse facial features, disproportionate short stature, hyperplastic gums, organomegaly, and retarded psychomotor development. These physical changes however require time to develop and are not apparent at birth. Opportunistically, neonates with I-cell disease are diagnosed after revealing typical spine changes in skeletal survey. Unlike nutritional rickets, their underlying cause and course of the bone disease are different.

CASE
We report a premature 34-week-old male with a birth weight of 1.6 kg. He was born in an ambulance while his mother was being transferred from a district hospital. He required cardiopulmonary resuscitation at birth and was admitted to the neonatal intensive care unit where he stayed for another 6 months due to respiratory reason. During his 1st to 4th months of life, he had markedly raised ALP level (>1000 IU/L), together with severe hyperparathyroidism that gradually resolved with time. He had radiographic changes resembling rickets which persisted and progressed to "chronic osteitis fibrosa cystica". His serum calcium, phosphate, and 25(OH)D_3 levels had always been normal. He never had a fracture. His diagnosis was later confirmed by marked elevation of plasma b-hexosaminidase, b-mannosidase and a-mannosidase. He was discharged home with tracheostomy and CPAP. He continues to survive at 11 months old by the time of this report.

CONCLUSION
Mucolipidosis osteodystrophy could resemble nutritional rickets during the neonatal period but it is not related to either deficient vitamin D or minerals. Failure to respond to conventional treatment (vitamin D and calcium or phosphate), should prompt for a search for other causes of increased bone turnover. In I-cell disease,

hyperparathyroidism often remits but bone disease progresses. Bisphosphonate therapy such as pamidronic acid has been described in those with recurrent fractures in later life.

EP_P005

CASE REPORT: VAN WYK-GRUMBACH SYNDROME: HYPOTHYROIDISM PRESENTING AS PRECOCIOUS PUBERTY

https://doi.org/10.15605/jafes.038.S2.134

Hazirah Ahmad
Hospital Sultanah Bahiyah, Malaysia

INTRODUCTION/BACKGROUND
Acquired causes of hypothyroidism such as Hashimoto's thyroiditis is mostly insidious and often goes undetected, unless there is concomitant thyroid gland enlargement or profound hypothyroid symptoms. Precocious puberty in girls is a rare cause of acquired hypothyroidism.

CASE
We report an 8 year-3-month-old female of Chinese-Indian descent, who presented with precocious puberty (bilateral breast budding, axillary hair, and pubic hair) about 6 months prior, with the mother's concern of poor learning in school. There were no other symptoms of puberty such as vaginal discharge or growth acceleration. She was adopted at 6 months old, thus, there is uncertainty about her biological family history. Her height was at the 25th centile and weight at 75th centile. She had a single café-au-lait spot at the right thigh, with Tanner stage 2 breasts, axillary hair, and pubic hair. She also had mild scoliosis with no other skeletal deformities. She had no thyroid gland enlargement but her facial expression was dull. Her blood investigations revealed pre-pubertal levels of gonadotrophins with undetectable estradiol, normal prolactin and negative b-HCG screen. Her thyroid function revealed markedly increased TSH (>100mIU/L) with severely low fT4 (<5.4 pmol/L). Her thyroid peroxidase antibody (anti-TPO) level was 131 IU/ml (<35). Ultrasound of the thyroid showed features in keeping with autoimmune thyroiditis with incidental thyroglossal duct cyst. Upon further questioning, the mother did recall prominent neck swelling since the past 2 years. Following L-thyroxine initiation, her thyroid function normalized and she showed significant improvement in height (she grew 10 cm/year) with progression of puberty. Her last bone age was 10 years old (CA: 9 year and 6 months).

CONCLUSION
Van Wyk-Grumbach syndrome is a relatively uncommon cause of pseudo-precocious puberty that often skips detection. Thyroid assessment is recommended in a girl presenting with precocious puberty, even in the absence of goiter. Timely diagnosis and treatment with L-thyroxine normalizes thyroid function and significantly improves linear growth.

EP_P006

CASE REPORT: EXOGENOUS CUSHING SYNDROME IN A GROWING CHILD FOLLOWING CHRONIC TOPICAL STEROIDS FOR FAMILIAL PSORIASIS

https://doi.org/10.15605/jafes.038.S2.135

Tsae Yun Tan
Hospital Sultanah Bahiyah, Malaysia

INTRODUCTION/BACKGROUND
Cushing syndrome is relatively uncommon in young children but exogenous Cushing syndrome is increasingly seen due to both prescribed and surreptitious intake of steroids.

CASE
We report a 6 year-10-month-old female, referred from Dermatology for obesity. She had itchy and scaly red plaques about 1 year prior. Both her mother and elder sister had been diagnosed with psoriasis and treated elsewhere. Even without a doctor's advice, she was given over-the-counter topical betamethasone dipropionate by her mother 1 year before consult. The mother applied the steroid generously over the child's whole body including the face and inguinal region twice a day for every flare, which usually occurred around 2 episodes every month. Since then, her daughter gained weight and stopped growing. She had florid Cushingoid features with emotional lability (easily tearing), truncal obesity, thick violaceous purple striae over the trunk, neck, upper, and lower limbs, and extensive erythematous scaly psoriatic plaques (BSA~80%). She was hypertensive with blood pressures ranging from 130-150/88-100 mmHg. She did not have proximal muscle weakness. Her eye assessment was negative for glaucoma or cataract, or hypertensive retinopathy changes. She had a pre-pubertal Tanner stage with no virilisation or hirsutism. She had persistently suppressed 8am cortisol level (<27.6 nmol/L) and low ACTH level (1.10 pmol/L, reference range 1.6-13.9) with normal 17OHP and DHEAS screen. She had borderline HbA1c (5.7%), dyslipidaemia, and non-alcoholic fatty liver disease based on an abdominal ultrasound. Her topical steroids for psoriasis were stepped down and she was initiated on steroid-sparing UV