Conclusion: The main difficulty encountered with cancer developing from TGDC is that the diagnosis is usually made during surgery and from definitive pathological samples. The most common surgical procedure used is the Sistrunk procedure. Some studies have suggested that this procedure alone is an adequate therapy, but others advocate the need for total thyroidectomy. The Sistrunk procedure is considered to be appropriate for low-risk patients, but high-risk patients must undergo total thyroidectomy. The decision to perform a total thyroidectomy in this patient was based on her high-risk classification due to: age, sex, cyst size, and a positive FNA for malignancy. Follow-up includes an annual physical examination, thyroglobulin levels, and an US every 6 months during the first year and annually thereafter.

Pediatric Endocrinology

PEDIATRIC SEXUAL DIFFERENTIATION, PUBERTY, AND BONE BIOLOGY

Screening of Vitamin D and Calcium Concentrations in Neonates of Mothers at High Risk of Vitamin D Deficiency

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Objective: The aim of this study was to determine, retrospectively, the serum 25OHD and calcium concentrations of screened neonates of mothers at high risk of 25OHD deficiency (maternal 25OHD < 25 nmol/L or unknown vitamin D concentrations and risk factors for vitamin D deficiency) and critically analyse whether their measurements contributes to the management of these neonates.

Methods: Serum 25OHD and calcium concentrations from 600 samples of umbilical cord blood or venous blood collected from neonates over a 12-month period were analysed. 25OHD concentrations were reported for all while both the corrected calcium concentrations and vitamin D concentrations were available for 569 samples.

Results: There was little or no evidence of association between neonatal 25OHD concentrations and gender, gestational age or birth weight. There was a high prevalence of vitamin D insufficiency (27.6%, 30–50 nmol/L) and deficiency (21.3%, < 30 nmol/L) in neonates from high-risk maternal groups. There was a statistically positive but weak correlation ($\rho = 0.22$, $P < 0.0001$) between serum calcium and 25OHD concentrations. Only 7 neonates out of 569 (1.2%) had calcium levels in the hypocalcaemic range; however, a significant number (47.6%) were reported to be in the hypercalcaemic range. Nearly all of these were venous samples collected in first 24 hours after birth. We calculated the reference interval for corrected calcium from our data of venous samples in first 24 hours and the upper limit was significantly higher (2.38–3.04 mmol/L) than the standard reference range used.

Conclusion: Vitamin D deficiency is prevalent in neonates of high-risk mothers but the risk of hypocalcaemia due to vitamin D deficiency at birth is low. Screening neonates entails blood testing which can cause distress to neonates and their parents, substantial impost on staff and financial burden on the health care system. 25OHD deficiency is corrected relatively easily in neonates with supplementation and vitamin D supplementation of neonates from birth without routine screening appears to offer better value of care. Also, the data from this study suggest that the paediatric reference range for corrected calcium concentrations in neonates is higher and the paediatric reference range should be reconsidered.

Bone and Mineral Metabolism

BONE AND MINERAL CASE REPORTS II

Charge Syndrome: Unusual Cause of Hypogonadism Leading to Osteoporosis

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CHARGE syndrome is an unusual cause of hypogonadism; it is characterized by coloboma, heart defect, ateresia choanae, retarded growth and development, genital hypoplasia and ear anomalies. Two-thirds of affected patients have a mutation within the chromodomain helicase DNA-binding protein-7 gene, which is involved in embryonic development. The involvement of this gene in the pathogenesis of isolated idiopathic hypogonadotropic hypogonadism (HH) has been postulated. The reported incidence of this syndrome ranges from 0.1–0.2/10000 (1).

A 24 year old female presented to our facility for further management of her HH and osteoporosis in the setting of her CHARGE syndrome. She was born full-term and diagnosed with this condition at the age of 6. Formal genetic testing as an adult demonstrated mutation within the CHD7 gene (chr 8:61,757,970). She had delayed puberty secondary to her hypogonadism; she was not treated with HRT as benefits were not considered significantly sufficient. She subsequently developed osteoporosis at the age of 20 which was treated at an outside facility with pamidronate IV Q4 months along with calcium and Vitamin D supplementation. Her initial Dual-energy X-ray absorptiometry (DXA) showed scoliosis in the lumbar spine [bone mass density (BMD): 0.514,osteoporosis by Z-score] with total hip showing BMD: 0.738,osteopenia by Z-score. Follow-up DXA after 3 years showed statistically significant improvement in bone mineralization of her L-spine [BMD: 0.595, +16%] and total hip [BMD: 0.777, +13.5%]. She presented to our facility in 2018 with labs showing normal calcium and 25-OH D; treatment with pamidronate was continued. She had a repeat DXA in 2019 which showed Z-scores of 0 in the left and right femoral necks. She was given the option of continued treatment for her osteoporosis versus monitoring and chose the latter with follow-up DXA.

Hypogonadotropic hypogonadism is associated with delays in puberty or pubertal arrest. Luteinizing Hormone Releasing Hormone and HCG tests should be performed within the four months of life or at puberty in cases of hypogenitalism. GH deficiency should be investigated as a cause for growth retardation with GH stimulation levels. Hormone replacement is often required at puberty for