Severe Neonatal Hypercalcemia; a challenging case

Hashim R., Prematilake D., Gunasekara B., S. Na / K / Cl / Mg – normal

Clinical Presentation

- D-10 neonate, fourth-born to 2nd degree consanguineous parents, presented with poor feeding and low-grade fever of 3 days duration.
- Birth weight of 3.9 kg with no perinatal complications or maternal gestational diabetes.
- 30% weight loss and severe dehydration at presentation, lethargic and hypotonic.
- No subcutaneous fat necrosis was felt.

Investigation

- Negative septic screen.
- CBS – 140 mg/dl; S. Na / K / Cl / Mg – normal
- S. Ca (corrected) – HIGH (7.61 mmol/L; 2.2-2.7)
- S. phosphat – LOW (0.75 mmol/L; 0.87-1.45)
- ALP – NORMAL (246 IU/L; 60-425)
- S. PTH – SKY HIGH (403.2 pg/ml; 4 – 73)
- 25OH vitamin D – NORMAL (50.2 nmol/L; sufficient >50)
- Urine Ca/Cr ratio – not performed (was on IV furosemide)
- Parents’ – normal calcium and urine Ca/Cr ratio
- Genetic confirmation pending.

Serum corrected Calcium levels since presentation (mmol/L)

Serum PTH Levels since presentation (pg/ml)

Clinical Diagnosis

Neonatal severe hyperparathyroidism (NSHPT) is a rare autosomal recessive disorder due to inactivating mutations of calcium-sensing receptor. These receptors are vital in calcium homeostasis and are expressed in a number of tissues such as parathyroid glands, renal tubules and bone. Homozygous mutations lead to severe hypercalcemia and life-threatening bone demineralization if untreated.

Bibliography