

Severe Neonatal Hypercalcemia; a challenging case

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Clinical Diagnosis

A neonate born to consanguineous parents with a typical clinical presentation and having evidence of hyperparathyroidism lead to a most probable diagnosis of neonatal severe hyperparathyroidism.

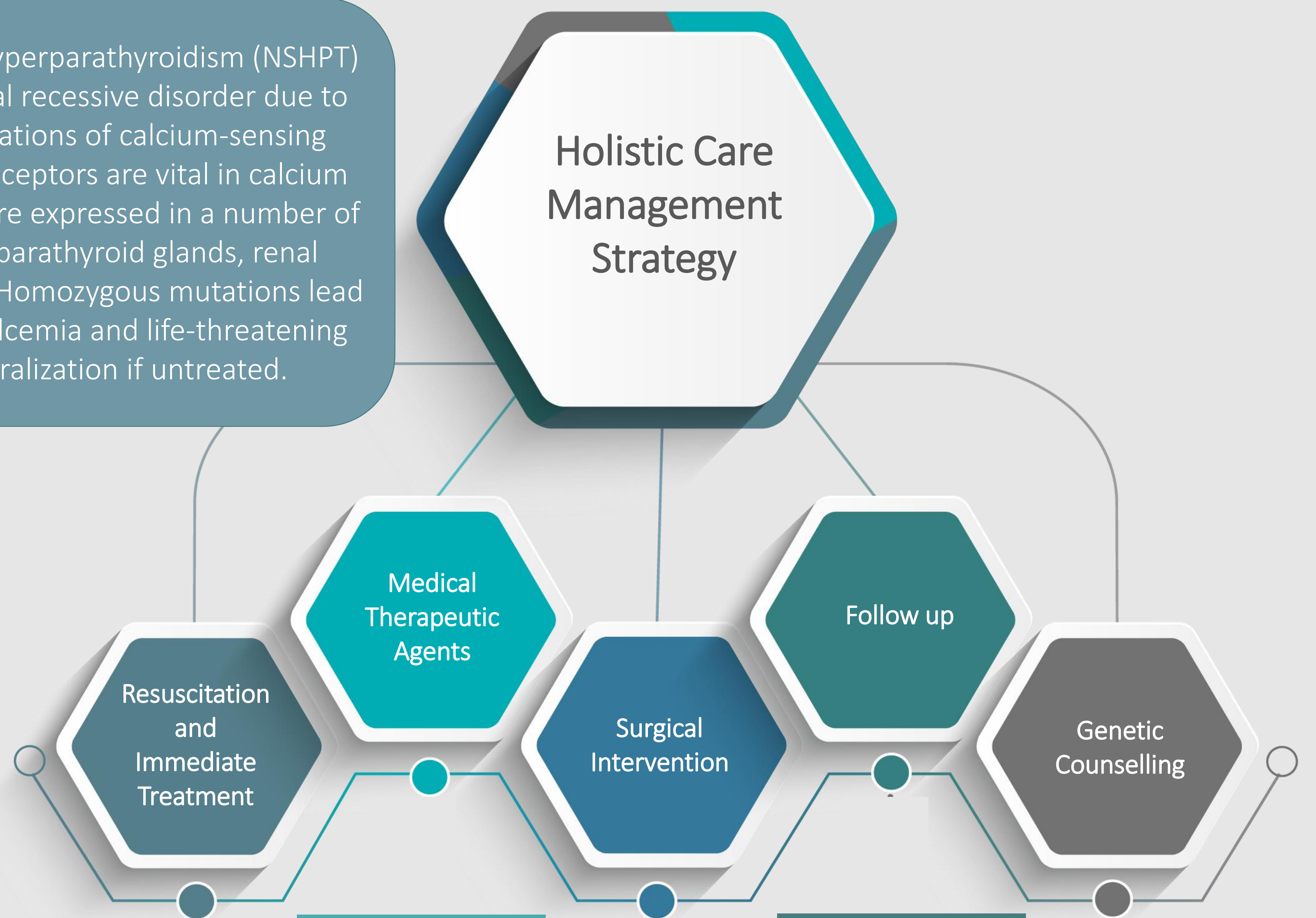
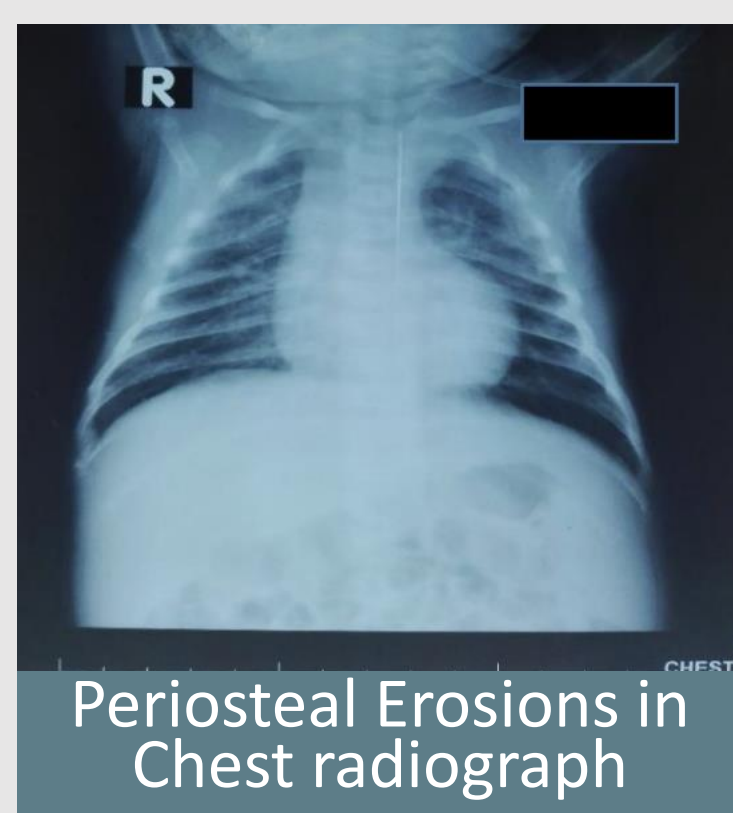
Neonatal severe hyperparathyroidism (NSHPT) is a rare autosomal recessive disorder due to inactivating mutations of calcium-sensing receptor. These receptors are vital in calcium homeostasis and are expressed in a number of tissues such as parathyroid glands, renal tubules and bone. Homozygous mutations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

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.

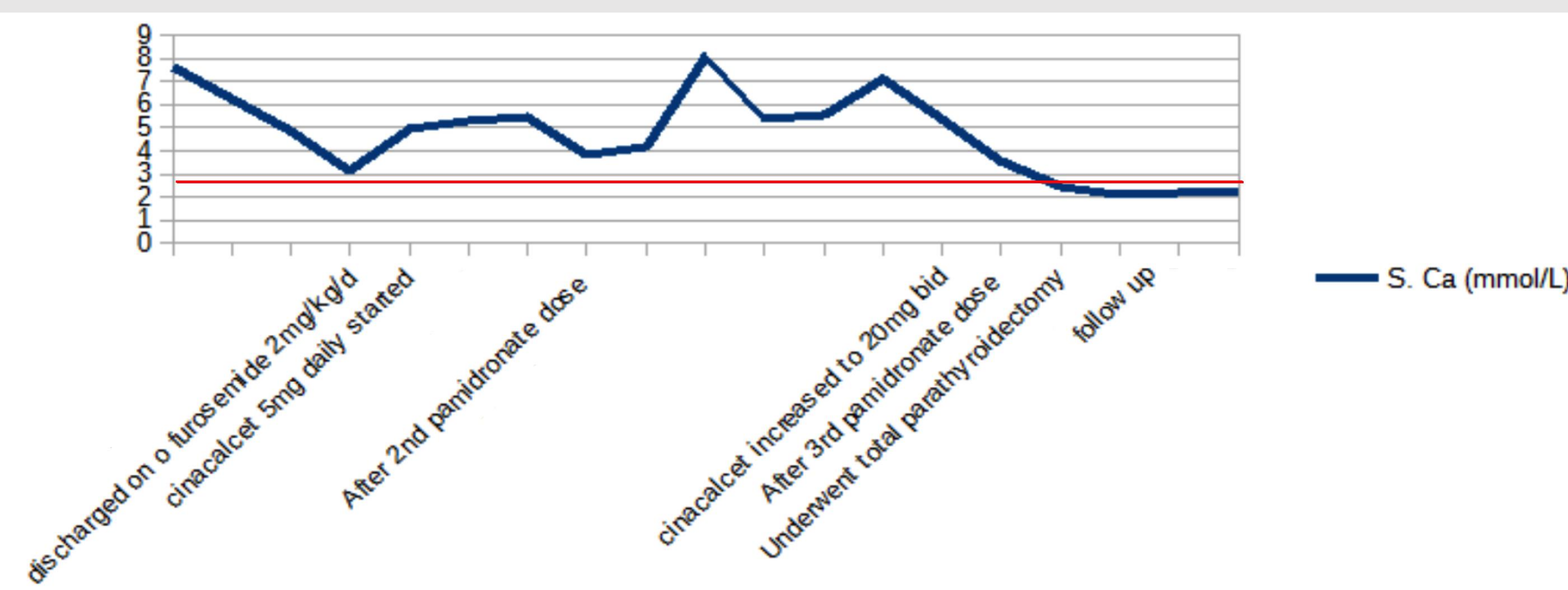
Investigation

- 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. phosphate – LOW (0.75 mmol/L; 0.87-1.45)
- ALP – NORMAL (246 IU/L; 60-425)
- S. PTH – SKY HIGH (403.2 pg/ml; 4-72)
- 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.

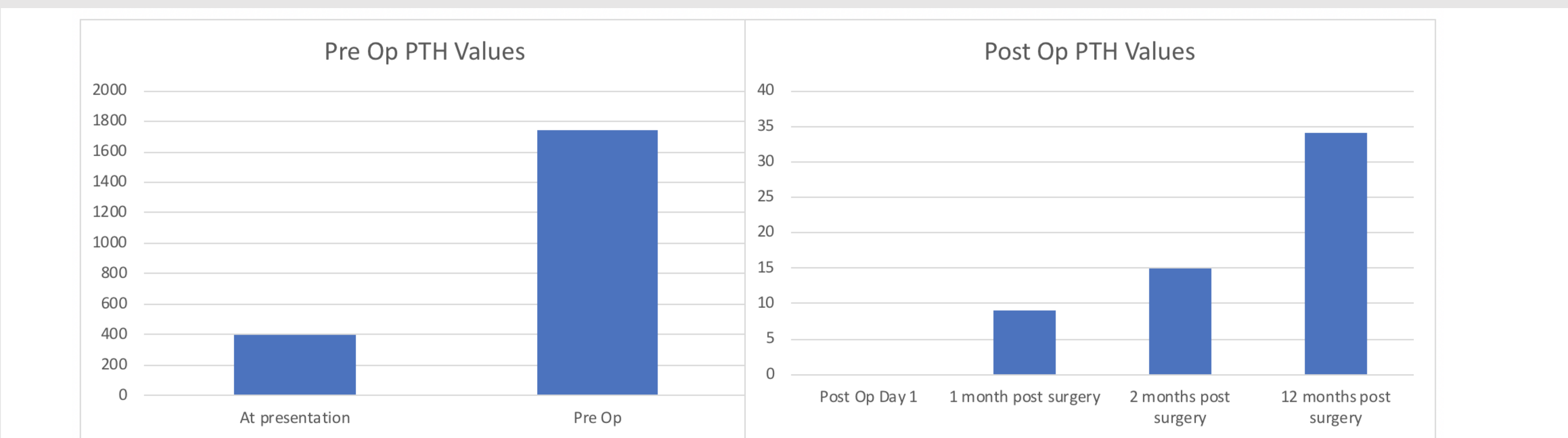


At presentation	At 6 weeks	At 3 months	At 9 months	Pending
<ul style="list-style-type: none"> IV 0.9% NaCl 150 - 175 ml/kg/d IV furosemide 1mg/kg bid IV pamidronate (1/2 dose) Discharged with O. furosemide 2mg/kg/d in 2 divided doses 	<ul style="list-style-type: none"> Cinacalcet 5mg daily (a calcimimetic) and titrated to a maximum of 20mg bid (=11mg/kg/d), 2nd dose of IV pamidronate 	<ul style="list-style-type: none"> Pre-op optimization of Ca - 3rd dose of IV pamidronate and hyperhydration Total parathyroidectomy with implantation of 1/2 of the 4th parathyroid gland under the L/ biceps muscle Periop - IV Ca. infusion, supplemented with oral calcitriol and calcium carbonate 	<ul style="list-style-type: none"> Ca. supplements tailed off and omitted 	<ul style="list-style-type: none"> Pending genetic confirmation

Serum corrected Calcium levels since presentation (mmol/L)



Serum PTH Levels since presentation (pg/ml)



Bibliography

- <https://rarediseases.info.nih.gov/diseases/2838/neonatal-severe-hyperparathyroidism>
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- Block G. Calcium, calcimimetics and clinical outcomes. *Clin J Am Soc Nephrol.* 2006;1(2):170-171