Severe Neonatal Hypercalcemia; a challenging case

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.

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Clinical Presentation

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.

Holistic Care Management Strategy

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 lacksquarematernal gestational diabetes.
- 30% weight loss and severe dehydration at presentation, \bullet lethargic and hypotonic.
- No subcutaneous fat necrosis was felt. \bullet

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)
- 250H 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



Periosteal Erosions in Chest radiograph

dose) • Discharged with O. furosemide 2mg/kg/d in 2 divided doses

• IV furosemide 1mg/kg

• IV pamidronate (1/2

 \bullet

bid

- Medical Follow up Therapeutic Agents Resuscitation Surgical and Genetic Immediate Intervention Counselling Treatment At 9 months At 6 weeks Cinacalcet 5mg • Ca. daily (a At presentation At 3 months Pending supplements calcimimetic) and tailed off and titrated to a omitted • Pre-op optimization IV 0.9% NaCl 150 - 175 • Pending genetic maximum of 20mg of Ca - 3rd dose of IV ml/kg/d bid (=11mg/kg/d),
 - 2 nd dose of IV pamidronate
- Total parathyroidectomy with implantation of ½ of the 4th parathyroid gland under the L/ biceps muscle

pamidronate and

hyperhydration

- Periop IV Ca. infusion, supplemented with oral calcitriol and calcium carbonate
- confirmation

Genetic confirmation pending.

discharged on o turosent de 2mg/kgdd

cinacalcet smo daily staned

Serum corrected Calcium levels since presentation (mmol/L)



Serum PTH Levels since presentation (pg/ml)

2nd partidionate

NSHPT is known to result in poor neurodevelopmental outcomes. It can be fatal if not promptly recognized and treated



Bibliography

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appropriately.

Prompt diagnosis and appropriate therapy with the involvement of specialists where the keys to this successful story

58--P3 **57ESPE**

Bone, growth plate and mineral metabolism

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Poster presented at: 57th ESPE 2018 Meeting ATHENS GRE 7-29 September 2018



