



A rare cause of hypercalcemia: Congenital Lactase Deficiency

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- A 2-month-old female was referred for hypercalcemia
- Birth weight of 3030 g from first cousin parents
- At presentation, Her height, weight and head circumference were
 - 55 cm (-1.25 SDS),
 - 3.4 kg (-2.83 SDS) and
 - 36 cm (-2.69 SDS), respectively
- Family history was unremarkable
- Hypercalcemia, hyponatremia and metabolic acidosis were detected (Ca:17.3 mg/dl, Na:127 mEq/L, pH:7.22, HCO₃:10.6, i.Ca:9.1 mg/dl)
- Additionally, the patient had hypercalciuria (Urine ca/cr:1.05 mg/mg), and medullary nephrocalcinosis on renal ultrasound.
- Physical examination was unremarkable
- Etiological evaluation for hypercalcemia was unrevealing except low 1.25(OH)₂D level (22 pg/ml)
- After an initial assessment, intravenous fluid and 1 mg/kg methylprednisolone was given once
- Further history revealed that she had watery-foamy diarrhea starting shortly after birth despite being fed exclusively with breast milk.
 - ✓ A malabsorption was considered and she was placed on lactose-free (LF) formula.
 - ✓ Serum calcium, decreased to 13.6 mg/dl on the second day after hydration and methyl prednisolone and returned to normal within a month of initiating LF formula.
 - ✓ No further hypercalcemia was detected on follow-up.
 - ✓ Diarrhea and vomiting also ceased and the patient started to thrive well.
- On her last examination, when she was 7.5-year-old, her height and weight were 116.6 cm (-1.3 SDS) and 27 kg (0.71 SDS), respectively with normal biochemistry.

Molecular analyses

- ✓ Absence of mutations related to the hypercalcemia
- ✓ Bi-allelic c.1364T>C (p.Ile455Thr) mutation in the *LCT* gene

Table. Laboratory follow-up of the patient

	First Evaluation (2 mos)	4. hour after intravenous fluid and methylprednisolone	24. hour with intravenous fluid	9. day with LF formula before discharge	4. Month with LF formula	Last Evaluation (7.5 yrs)
Ca (mg/dl)	17,3	15,9	13,6	10,5	9,3	9,3
P (mg/dl)	5,4	5	4,8	6	6,4	4,8
ALP (U/L)	156	131	119	130		251
PTH (pg/ml)	3,7				26,4	45,8
25OHD						18,2
Cr (mg/dl)	0,3	0,32		0,22		0,37
Alb (g/dl)	3,9			3,9		42
Mg (mg/dl)	3,6	3,3		1,9		2
Na (mEq/L)	127	130	146	140		
K (mEq/L)	3,5	3,1	3,2	5,1		
AST/ALT (U/L)	36/34			34/28		
Ph/PCO ₂ /HCO ₃	7,22/26,7/10,6	7,22/34/13,8	7,29/33,5/15,6	7,42/32,4/20,6		
ionize Ca (mg/dl)	9,1			5,54		
Laktat (mmol/L)	1,5					
1,25(OH) ₂ D (pg/ml)	22					
Spot idrar Ca/Cr (mg/mg)	1,05					0,02

Learning Points:

- ✓ Cause-specific treatment in hypercalcemia prevent unnecessary nonspecific treatments (Bisphosphonates, calcitonin etc) as LF diet provided and maintained normocalcemia in our patient. CLD should be kept in mind in the differential diagnosis of hypercalcemia and history of diarrhea should be carefully sought in these patients.

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