

Novel SLC34A1 mutation in a girl infant with infantile hypercalcemia

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Case

Age/Sex : 28-day-old/a girl infant

Chief complaint : hypercalcemia, nephrocalcinosis

Previous illness Hx : increased renal echogenesity detected at 28 weeks' gestation

Symptom : constipation without poor feeding, hypotonia, and lethargy

Family history : the father of the neonate with a history of nephrolithiasis

Laboratory findings

Calcium/Phosphorus **12.8** mg/dL (8.8-10.8 mg/dL) / **4.3** mg/dL (4.8-8.2 mg/dL)

25-OH-vitamin D/1-25-OH Vitamin D : 13.01 ng/mL / **71.27** pg/mL

Intact Parathyroid hormone : **3.6** pg/mL (15-65 pg/mL)

24-hour urinary calcium excretion : 20.4 mg/kg (normal range < 4 mg/kg)

Targeted exome sequencing : SLC34A1 missense mutation

(A) c.1337G>A (B) c.1483C>T

Treatment : hydration, formula feeding (a low calcium, vitamin D free)

Medullary nephrocalcinosis of the left kidney



SLC34A1_exon12

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TGTGATCAGCATIGAGAGGGCCCTACCCGCTCACACTGGTTCCCAACATCGGCACCCACCACCACGGCCATCCTGGCT
TGTGATCAGCATIGAGAGGGCCCTACCCGCTCACACTGGTTCCCAACATCGGCACCCACCACCACGGCCATCCTGGCT
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TGTGATCAGCATIGAGAGGGCCCTACCCGCTCACACTGGTTCCCAACATCGGCACCCACCACCACGGCCATCCTGGCT
TGTGATCAGCATIGAGAGGGCCCTACCCGCTCACACTGGTTCCCAACATCGGCACCCACCACCACGGCCATCCTGGCT
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Patient

Mother

Father

(A)

SLC34A1_exon13

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AACATCTCGGGTATCCTTCTGTGGTACCCGGTCCCTGCACAGCCTGCCATCCGCATGCCCCAAGGGCCCTGGGAAACGCACGG
AACATCTCGGGTATCCTTCTGTGGTACCCGGTCCCTGCACAGCCTGCCATCCGCATGCCCCAAGGGCCCTGGGAAACGCACGG
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AACATCTCGGGTATCCTTCTGTGGTACCCGGTCCCTGCACAGCCTGCCATCCGCATGCCCCAAGGGCCCTGGGAAACGCACGG
AACATCTCGGGTATCCTTCTGTGGTACCCGGTCCCTGCACAGCCTGCCATCCGCATGCCCCAAGGGCCCTGGGAAACGCACGG
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Patient

Mother

Father

(B)

Conclusions

✓ Clinicians need to consider testing of SLC34A1 in patients with hypercalcemia and hypercalciuria, and nephrocalcinosis. Further studies regarding the clinical course, long-term prognosis, and efficacy of a supplement with phosphate in similar patients will be required.

