# A novel varient of *SLC34A1* gene in an infant with Idiopathic infantile hypercalcemia

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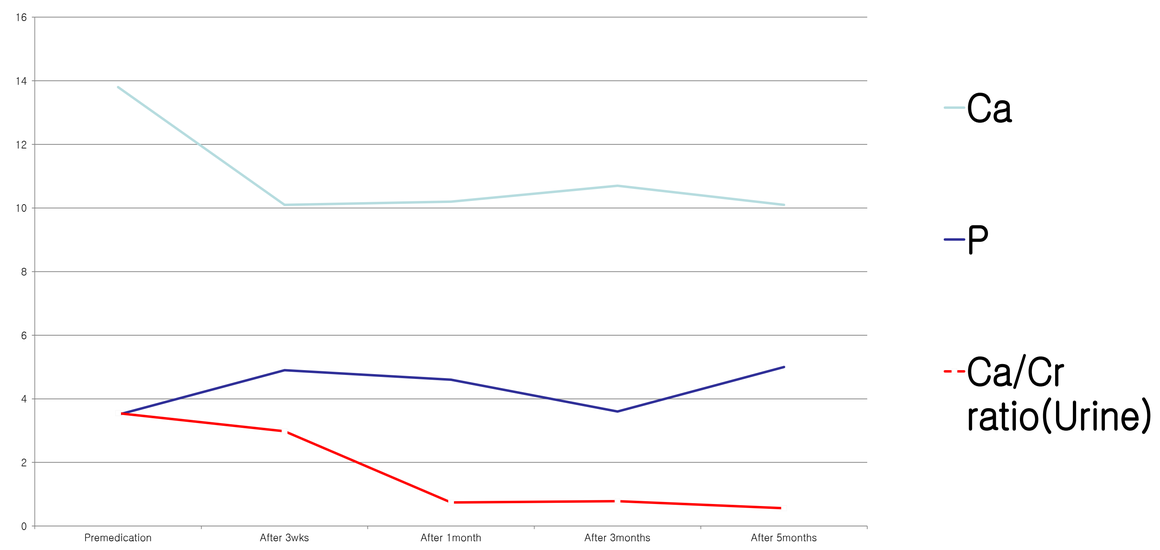
**Background and Aims:** Idiopathic infantile hypercalcemia is one of rare diseases characterizing hypercalcemia in infancy. Renal phosphate absorption in proximal tubules plays a very important role in the phosphate and calcium homeostasis. SLC34A1 is known a key regulator of renal phosphate reabsorption. SLC34A1 gene mutation is one of very uncommon causes of idiopathic infantile hypercalcemia. We have experienced a case of idiopathic infantile hypercalcemia caused by a homozygous novel variant c.1483C>T(p.Arg495Cys) of SLC34A1 gene.

**Table.1 Biochemical characteristics of the patient.**

| Laboratory Findings | |
|---|---|
| Ca | 13.8 mg/dL |
| P | 3.5 mg/dL |
| Mg | 2.0 mEq/L |
| ALP | 572 U/L (ref : 60~360U/L) |
| i-PTH | 0.6 pg/mL |
| PTHrP | <1.1 pmol/L |
| 25(OH)vitamin D3 | 65 ng/mL |
| 1,25(OH) vitamin D3 | 79 pg/mL |
| Ca/Cr Ratio(Urine) | 1.4 (<0.8) |
| P (Urine) | 9.6 mg/dl |

**Table.2. Changes in blood Calcium after supplyment of PO phosphorus.**

| | Premedication | After 3wks | After 1month | After 3month | After 5month |
|---|---|---|---|---|---|
| Ca(mg/dL) | 13.8 | 10.1 | 10.2 | 10.7 | 10.1 |
| P | 3.5 | 4.9 | 4.6 | 3.6 | 5.0 |
| Ca/Cr ratio (Urine) | 3.55 | 2.98 | 0.74 | 0.78 | 0.56 |




**sanger sequencing result**
**NM_003052.4 (SLC34A1):c.1483C>T, p.Arg495Cys**

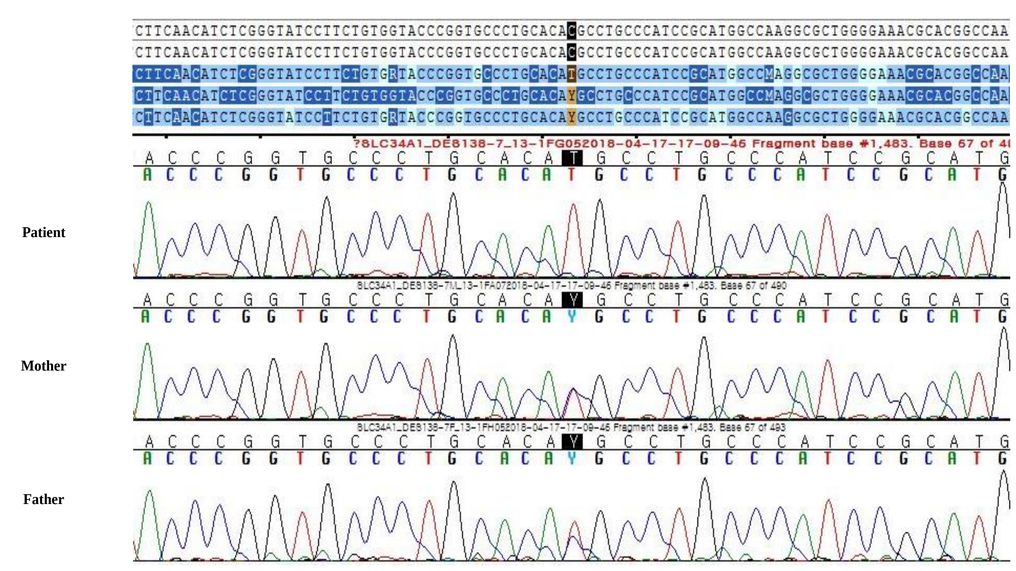



**Fig.1. Sanger sequencing result of Patient & Parents.**

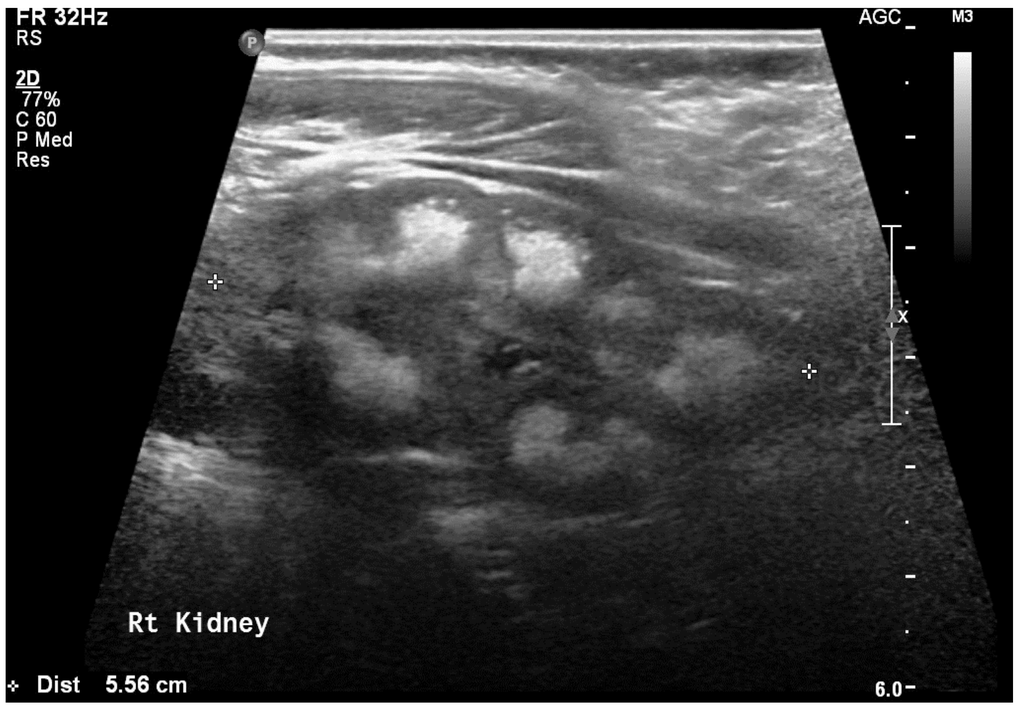



**Fig.2. This figure shows that Medullarynephrocalcinosis of Patient .**

**Results**: A 5-month-old boy was transferred to Kyungpook National University Children's Hospital because of sustained hypercalcemia with hypercalciuria. Laboratory investigations revealed a serum calcium level of 12.6 mg/dL (normal range: 9.0–10.6), phosphate level of 3.7 mg/dL(normal range: 4.8–8.2), serum magnesium level of 2.0mEq/L (normal range: 1.44–3.12), intact PTH level of 0.6 pg/mL (normal range: 10–65),PTHrP<1.1pmol/L (normal range:<2.0), 25(OH)vitamin D3 level of 65 ng/mL (normal range: 8.0–51.9) and 1,25(OH) vitamin D3 level of 79 pg/mL(normal range: 25-65). Spot urine calcium/urinary creatinine ratio (mg%: mg%)was elevated 1.4 (normal level: <0.8 for infant).Targeted exome sequencing in the patient was performed, resulting in a homozygous novel variant c.1483C>T(p.Arg495Cys) of *SLC34A1* gene confirmed by Sanger sequencing

**Conclusion**: We report a case with idiopathic infantile hypercalcemia caused by a novel variant of *SLC34A1* gene mutation