

Teriparatide (rhPTH) therapy in a boy with Hypoparathyroidism-Deafness-Renal dysplasia (HDR) syndrome due to GATA3 mutation

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Background

Hypoparathyroidism is usually treated with calcium and vitamin D analogues, the long term use of which could lead to side effects like nephrocalcinosis. Subcutaneous recombinant human parathormone [rhPTH] can potentially avoid these side effects. We report a 3-year-old boy with HDR syndrome who has been successfully treated with Teriparatide (1-34 rhPTH).

Case

- Term male infant born to non-consanguineous Asian parents
- Hypocalcemic seizures at 1 month of age needing intensive care support with ventilation.
- Parathyroid hormone (PTH)-inappropriately low at the time of hypocalcaemia suggesting hypoparathyroidism.
- Bilateral sensorineural hearing loss from an early age requiring hearing aids.
- Delayed motor milestones
- Hypocalcaemia-managed with calcium supplements and calcitriol.
- At 3 years of age, the family moved to UK from India.
- Commenced on alfacalcidol (40ng/kg/day) and calcium supplements (2.5mmol/kg/day).
- 25-hydroxy-vitamin D level and ECHO were normal. FISH for 22q11 deletion-negative.
- Genetic evaluation revealed a heterozygous GATA3 mutation.
- High doses of alfacalcidol and oral calcium only just maintained normocalcemia, but resulted in hypercalciuria, with hypocalcemic episodes happening during intercurrent illnesses.
- Renal function and renal ultrasound-normal with no evidence of nephrocalcinosis
- Commenced on Teriparatide subcutaneously at a dose of 4 microgram twice daily resulting in normalisation of plasma calcium and phosphate levels and improvement in hypercalciuria(Figure 1, 2 & Table 1)
- Alfacalcidol and calcium supplements were gradually weaned and stopped.
- Teriparatide is maintaining calcium within normal range with noticeable improvement in motor function.

Response to Teriparatide (1-34 rhPTH)

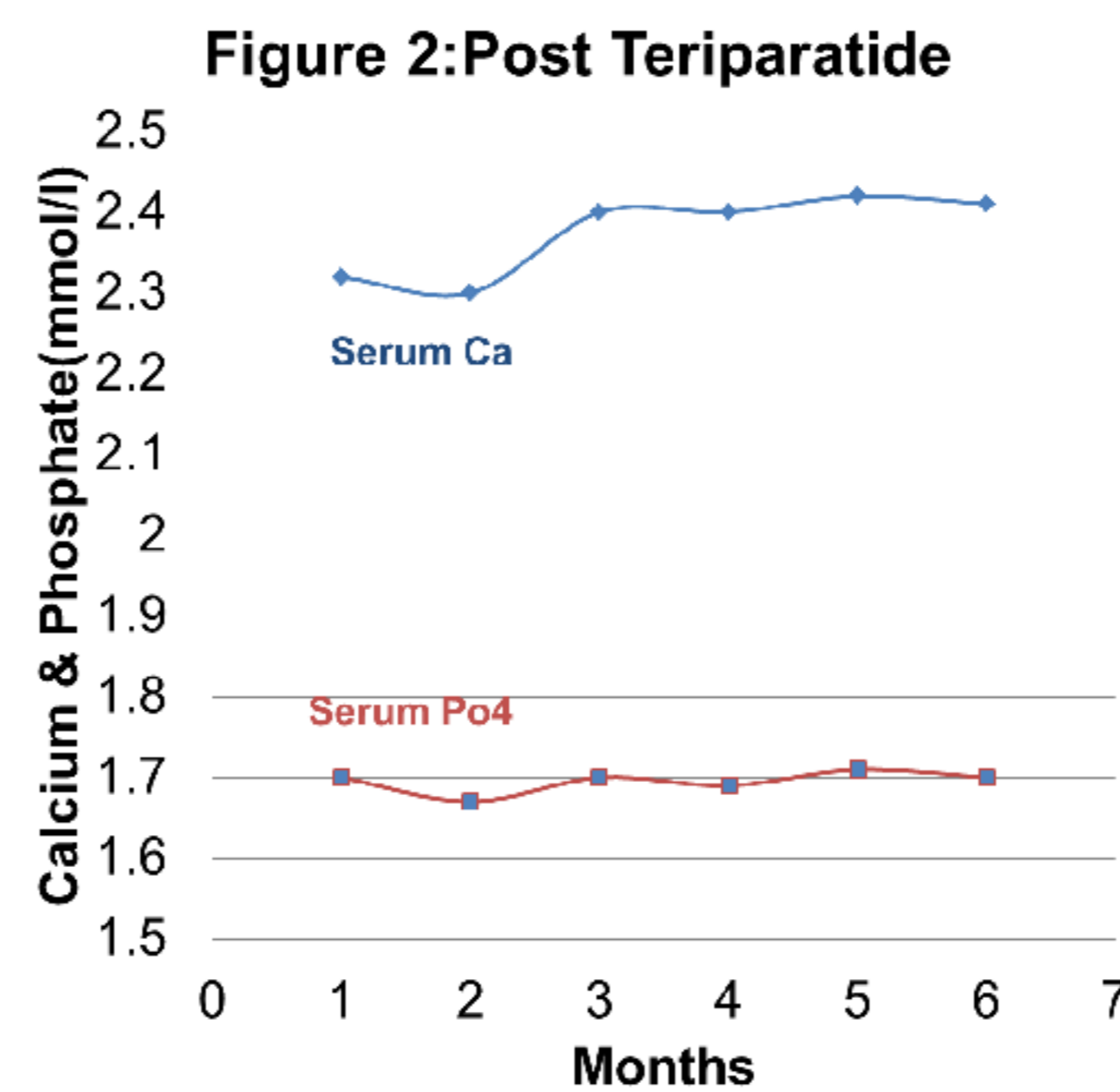
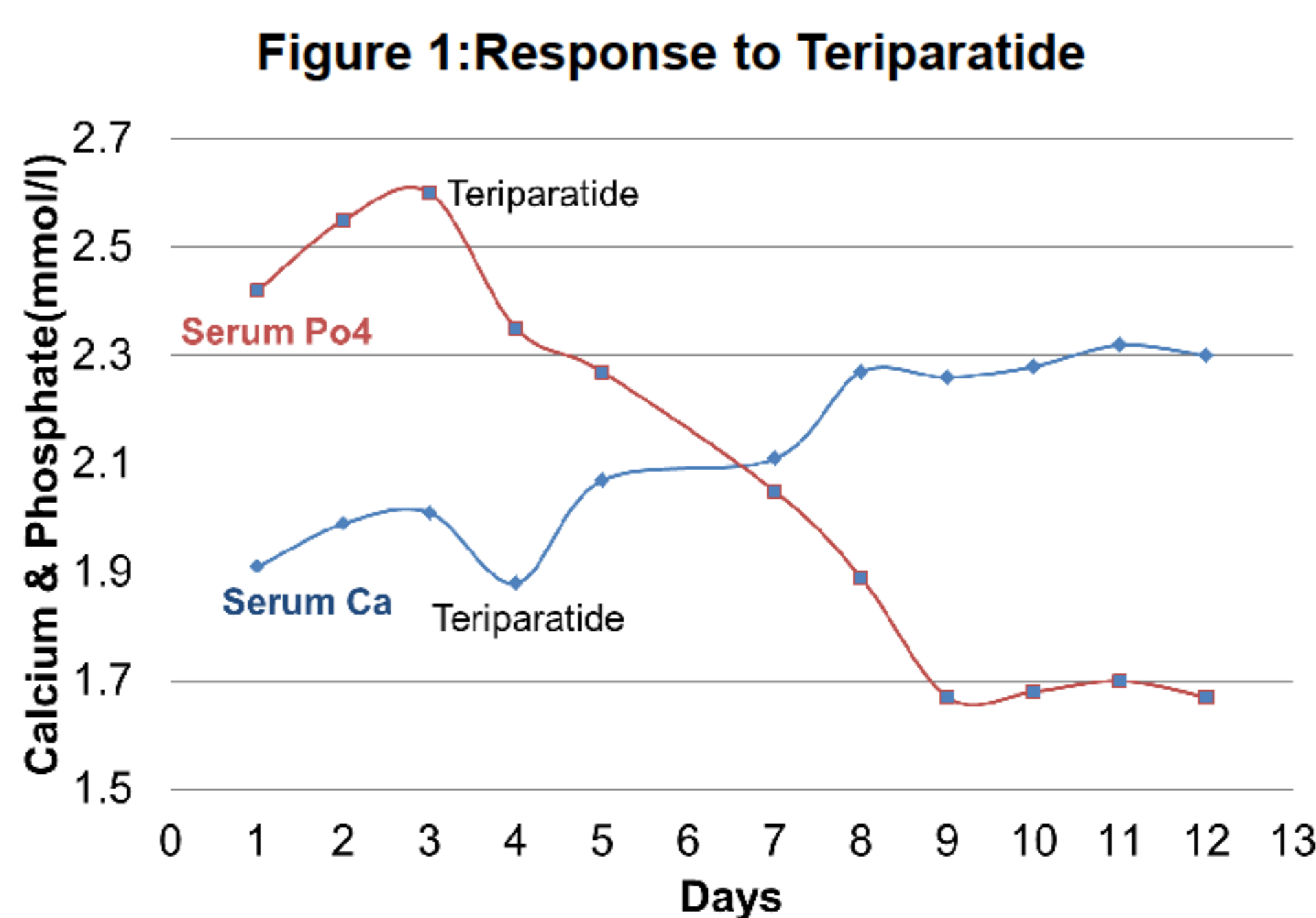


Table 1: Summary of effects

Pre-Teriparatide	Post Teriparatide
Hypercalciuria	No Hypercalciuria
Hypocalcemia worsening during intercurrent illness	Sustained maintenance of normocalcemia
Difficulty in motor activities and easy fatiguability	Noticeable improvement in motor function & quality of Life.

Conclusion

- Teriparatide is an effective therapeutic option for patients with hypoparathyroidism.
- Helps to normalise serum calcium and phosphate levels, minimising side-effects like nephrocalcinosis
- May aid improvement in motor function.

Reference

Patrizia Matarazzo, Gerdi Tuli, Ludovica Fiore, Alessandro Mussa, Francesca Feyles, Valentina Peiretti, Roberto Lala. Teriparatide (rhPTH) treatment in children with syndromic hypoparathyroidism. Journal of Pediatric Endocrinology and Metabolism. Volume 27, Issue 1-2, Pages 53–59.

