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)-appropriately 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 alfalcacidol (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 alfalcacidol 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)
- Alfalcacidol 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)

- 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.

Table 1: Summary of effects

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<thead>
<tr>
<th>Pre-Teriparatide</th>
<th>Post Teriparatide</th>
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<tbody>
<tr>
<td>Hypercalciuria</td>
<td>No Hypercalciuria</td>
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<td>Hypocalcemia worsening during intercurrent illness</td>
<td>Sustained maintenance of normocalcemia</td>
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<td>Difficulty in motor activities and easy fatigueability</td>
<td>Noticeable improvement in motor function &amp; quality of Life</td>
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Reference