Supplementary Table S-2. Indices about PTH secretion and parathyroid gland size in cases with neonatal severe primary hyperparathyroidism.$

<table>
<thead>
<tr>
<th>Reference for case(s)</th>
<th>FHH gene</th>
<th>Age while indexing parathyroid size (weeks)</th>
<th>Highest serum Ca++ (mg/dl)</th>
<th>Highest serum PTH (x upper nl)</th>
<th>Parathyroid size indices (various units)</th>
<th>Parathyroid mass estimate# (x upper nl)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hillman DA, Scriver CR, et al 1964</td>
<td>DNA</td>
<td>12</td>
<td>22</td>
<td>-----</td>
<td>3 gl.@ Each 10x nl</td>
<td>10</td>
</tr>
<tr>
<td>Hillman DA, Scriver CR, et al 1964</td>
<td>DNA</td>
<td>2</td>
<td>29</td>
<td>-----</td>
<td>2 gl. Each 0.5 cm</td>
<td>4</td>
</tr>
<tr>
<td>Goldbloom RB, Gillis DDA, et al 1972</td>
<td>NA</td>
<td>12</td>
<td>27</td>
<td>-----</td>
<td>4 gl. Each &gt; 10x nl</td>
<td>10+</td>
</tr>
<tr>
<td>Grantmyre EB 1973</td>
<td>FH</td>
<td>12</td>
<td>27</td>
<td>-----</td>
<td>2.5 gl. 100 mg</td>
<td>10</td>
</tr>
<tr>
<td>Rhone DP 1975</td>
<td>NA</td>
<td>4</td>
<td>31</td>
<td>-----</td>
<td>3.5 gl. 163 mg</td>
<td>15</td>
</tr>
<tr>
<td>Thompson NW, Carpenter LC, et al 1984</td>
<td>FH</td>
<td>12</td>
<td>20</td>
<td>-----</td>
<td>3 gl. 4-fold</td>
<td>4</td>
</tr>
<tr>
<td>Thompson NW, Carpenter LC, et al 1984</td>
<td>FH</td>
<td>3</td>
<td>24</td>
<td>-----</td>
<td>3 gl. 1.5 cm~</td>
<td>high~</td>
</tr>
<tr>
<td><strong>Steinmann B, Gnehm HE, et al 1984</strong></td>
<td>DNA</td>
<td>8</td>
<td>15</td>
<td>20</td>
<td>4 gl. Each 0.5 cm</td>
<td>4</td>
</tr>
<tr>
<td>Lutz P, Kane O, et al 1986</td>
<td>FH</td>
<td>2</td>
<td>20</td>
<td>-----</td>
<td>2 gl. Each 0.5 cm</td>
<td>4</td>
</tr>
<tr>
<td>Cooper L, Wertheimer J, et al 1986</td>
<td>FH</td>
<td>3</td>
<td>26</td>
<td>-----</td>
<td>4 gl. Each 0.4-0.5 cm</td>
<td>4</td>
</tr>
</tbody>
</table>
Key L, Thorne M, et al 1990  NA  2  34  70  4 gl. Each 0.5 cm  4
Blair JW, Carachi R 1991  NA  2  32  -----  3 gl. Each 0.3-0.5 cm  2

$ Gland sizes from autopsy in 1 unoperated case (Hillman DA, Scriver CR, et al 1964) and from first parathyroidectomy of all others. Several had persistence or recurrence of hypercalcemia after parathyroidectomy. Later tissues were not included for those.

* DNA  Homozygous mutation of CASR reported separately and later.

  NA  Not available. This implies no family history suggestive of heredity disorder, and no CASR test result available.

  FH  Family history suggests hereditary disorder; i.e. parental consanguinity and/or other family member(s) with mild or severe hypercalcemia.

@ Gl  parathyroid glands

# Fold x upper normal estimated from normal weight of 6 mg (Gilmour JR, Martin WJ 1937); thus, estimate as (weight in mg)/10 mg  or  for volume  \((4/3 \pi [r \text{ in mm}]^3)/10\). Normal diameter in neonate is 0.1-0.3 cm (Key L, Thorne M, et al 1990)

~ Diameter of 1.5 cm is a very high outlier value, and gives estimate as a high outlier for gland volume.

Agarwal SK, Mateo C, Marx SJ 2009 Rare germline mutations in cyclin-dependent kinase inhibitor genes in MEN1 and related states. J Clin Endocrinol Metab/94 1826-34.


Beckers A, Aaltonen LA, Daly AF, Karhu A 2013 Familial isolated pituitary adenomas (FIPA) and the pituitary adenoma predisposition due to mutations in the aryl hydrocarbon receptor interacting protein (AIP) gene. Endocr Rev/34 239-77.


Scholl UI, Nelson-Williams C, Yue P 2012 et al Hypertension with or without adrenal hyperplasia due to different inherited mutations in the potassium channel KCNJ5. Proc Nat Acad Sci (USA)/109 2533-2538.


