

Supplementary Table 1. Clinical and laboratory findings of the patients with a clinical prediagnosis of RTH $\beta$  and without THRB gene variant

| Patient no | Sex | Age years | Initial presentation | Previous treatment  | Tachycardia | Height cm (SDS)<br>BMI (SDS) | FT3 ng/L<br>(adult: 2.3-4.2)<br>(children: 3-4.7) | FT4 ng/dL<br>(adult: 0.89-1.76)<br>(children: 0.83-1.43) | TSH mU/L<br>(0.55-4.78) | Autoimmunity            | Thyroid volume<br>(SDS or mL) | Thyroid nodule                    | Pituitary MRI | Other clinical features |
|------------|-----|-----------|----------------------|---------------------|-------------|------------------------------|---------------------------------------------------|----------------------------------------------------------|-------------------------|-------------------------|-------------------------------|-----------------------------------|---------------|-------------------------|
| 1          | M   | 9.5       | Asymptomatic         | None                | No          | 151 (2.8)<br>18.2 (0.59)     | 8.29                                              | 1.95                                                     | 2.9                     | Negative                | 3.17 SDS                      | No                                | No            | CNV normal              |
| 2          | M   | 13.4      | Asymptomatic         | None                | No          | 157.7 (-1.45)<br>18 (-1.1)   | 6.59                                              | 1.72                                                     | 1.9                     | Negative                | -0.97 SDS                     | No                                | No            | (-)                     |
| 3          | F   | 39        | Palpitation          | Propranolol         | Yes         | NA                           | 4.5                                               | 1.88                                                     | 3.57                    | Negative                | 12.8 mL                       | Isohypoechoic nodule at follow-up | Normal        | CNV normal              |
| 4          | M   | 28        | Palpitation          | Bisoprolol fumarate | Yes         | NA                           | 5.92                                              | 2.17                                                     | 1.96                    | Negative                | 25.9 mL                       | No                                | Tshoma        | (-)                     |
| 5          | M   | 18.4      | Palpitation          | Mmz                 | Yes         | NA                           | 4.79                                              | 1.67                                                     | 1.34                    | Negative                | 11.4 mL                       | No                                | Normal        | (-)                     |
| 6          | F   | 25        | Asymptomatic         | None                | No          | NA                           | 3.09                                              | 2.21                                                     | 2.07                    | Anti-TPO +<br>Anti-Tg + | 6.3 mL                        | No                                | Normal        | (-)                     |
| 7          | F   | 8.5       | Asymptomatic         | None                | No          | 129.5 (0.14)<br>20.2 (1.5)   | 6.5                                               | 2.1                                                      | 2.6                     | Negative                | -0.27 SDS                     | No                                | No            | (-)                     |
| 8          | M   | 44        | Asymptomatic         | None                | No          | NA                           | 6.4                                               | 2.1                                                      | 1.5                     | NA                      | NA                            | NA                                | No            | (-)                     |
| 9          | M   | 11.9      | Asymptomatic         | None                | No          | 150.7 (0.11)<br>15.9 (-1.24) | 4.09                                              | 2.08                                                     | 1.9                     | Negative                | 0.01 SDS                      | No                                | No            | (-)                     |
| 10         | M   | 18        | Asympotomatic        | None                | No          | NA                           | 5.08                                              | 2.1                                                      | 0.78                    | Negative                | 2.2 SDS                       | No                                | No            | (-)                     |
| 11         | M   | 24        | Asympotomatic        | None                | No          | 171<br>19.8                  | 5.3                                               | 2.5                                                      | 1.6                     | Negative                | 7.4 mL                        | No                                | Normal        | (-)                     |
| 12         | F   | 3.2       | Palpitation          | Propranolol         | Yes         | 114.8 (0.24)<br>15.7 (0.13)  | 6.1                                               | 1.7                                                      | 2.7                     | Negative                | -0.13 SDS                     | No                                | No            | (-)                     |
| 13         | M   | 26        | Asymptomatic         | None                | No          | NA                           | 6.1                                               | 2.3                                                      | 1.6                     | Negative                | 19.2 mL                       | No                                | Normal        | (-)                     |
| 14         | M   | 8.3       | Asymptomatic         | None                | No          | 132 (0.71)<br>18 (0.9)       | 5.9                                               | 1.9                                                      | 1.6                     | Negative                | 1.27 SDS                      | No                                | No            | (-)                     |
| 15         | F   | 11.3      | Asymptomatic         | None                | No          | 145.5 (-0.26)<br>18.3 (0.03) | 5.7                                               | 1.6                                                      | 1.8                     | Negative                | 0.6 SDS                       | No                                | No            | (-)                     |
| 16         | M   | 50        | Asymptomatic         | None                | No          | NA                           | 7.2                                               | 2.4                                                      | 2.2                     | Negative                | 19.8 mL                       | Isoechoic nodule FNAB: benign     | Microadenoma  | (-)                     |
| 17         | M   | 3         | Asymptomatic         | None                | No          | 90 (-0.86)<br>17.9 (1.1)     | 7.2                                               | 1.9                                                      | 1.9                     | Negative                | 3.7 SDS                       | No                                | No            | (-)                     |

SDS: standard deviation score, NA: not available, BMI: body mass index, FNAB: fine-needle aspiration biopsy, FT3: free triiodothyronine, FT4: free tetraiodothyronine, TSH: thyroid-stimulating hormone, Mmz: methimazole, Anti-TPO: antithyroid peroxidase, Anti-Tg: anti-thyroglobulin, MRI: magnetic resonance imaging, CNV: copy number variant, RTH $\beta$ : resistance to thyroid hormone  $\beta$ , F: female, M: male