Supplementary Material 2

Two patients (cases 2 and 4) had no pathological variants. Case 1 was diagnosed with thyroid agenesis and had previously undergone comprehensive genetic evaluation with no specific etiology. The NGS panel identified a heterozygous deletion of the cadherin-15 gene (*PCDH15*) known as the cause for Usher syndrome (OMIM 601067), and a heterozygous missense variant of the ER transmembrane glycoprotein wolframin gene (*WFS1*) causing Wolfram syndrome (OMIM 606201), both of uncertain significance. Case 3 with known *TPO* mutation (c.1618 C>T; p.R540X) had a novel missense variant of 1 amino acid in position 97 in the calcium channel, voltage-dependent, L-type, alpha-1D subunit gene (*CACNA1D*) of uncertain significance. Mutations in this gene have been described in patients with hyperaldosteronism and neurological abnormalities (OMIM 114206). In case 5, with the same *TPO* mutation as in case 3, a heterozygous deletion causing replacement of 1 amino acid with no frameshift was identified in the collagen of the basement membrane, alpha-3 chain gene (*COL4A3*). Mutations in *COL4A3* have been reported in Alport syndrome (OMIM 120070). This patient had normal kidney function. Mutations in these four genes have only been reported with a homozygous manner of inheritance in patients with deafness.