Supplemental FIG. 1.

Thyroid ultrasonography of the patient. A, An image obtained at age 28 years (before right hemithyroidectomy). She had a 15 x 25 mm hypoechogetic solid nodule in the right lobe (indicated by an arrow). B, An image obtained at age 40 years, showing a small nodule in the left lobe (indicated by an arrow). She had two additional nodules in her thyroid. Note that small hypoechogetic loci were observed in the thyroid parenchyma (inset, an enlarged image). Bars = 1 cm.
Supplemental FIG. 2.

Schematic diagrams showing the development of the somatic mosaic PAX8 mutation. Mutation-carrying cells are indicated in orange, while cells without the mutation are indicated in skyblue. In the patient, mutation was detected in tissues derived from all three germ layers: the thyroid (endoderm), lymphocytes (mesoderm), and keratinocytes (nail; ectoderm). Furthermore, transmission of the mutation from the patient to her offspring indicates that the mutation was also present in her oocytes. Her thyroid and lymphocytes were mosaic for the somatic mutation, as shown by histology (FIG. 2) and GeneScan analyses (FIG. 1), respectively. Presence of a somatic mosaic mutation in the three germ layers and germ cells suggests that the mutation was acquired between the two-cell stage and the blastocyst stage, when the cells were totipotent.