Central congenital hypothyroidism caused by a novel mutation, C47W, in the cysteine knot region of TSHβ

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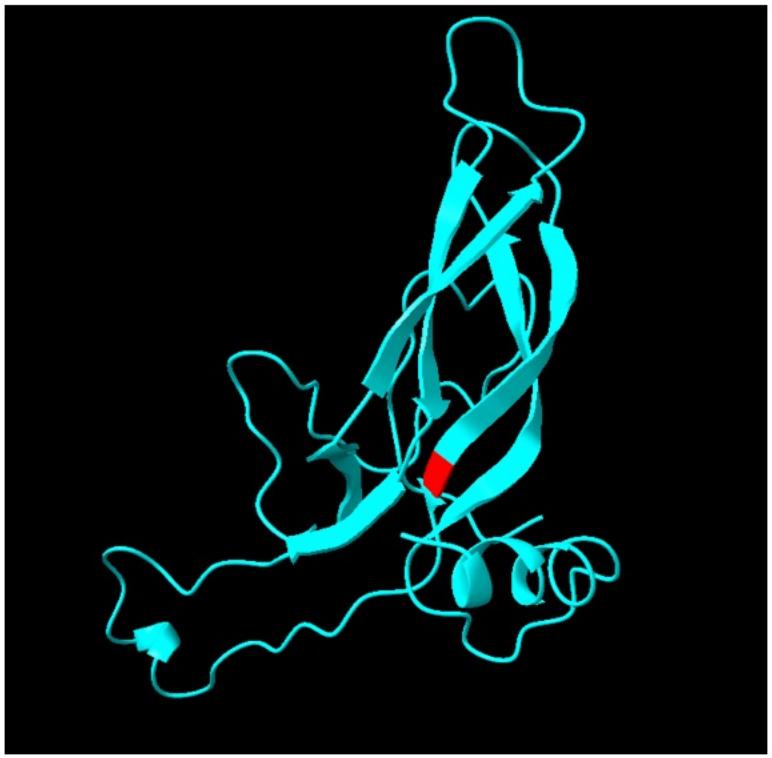
Supplemental Table 1. List of known genes related to thyroid disorders.

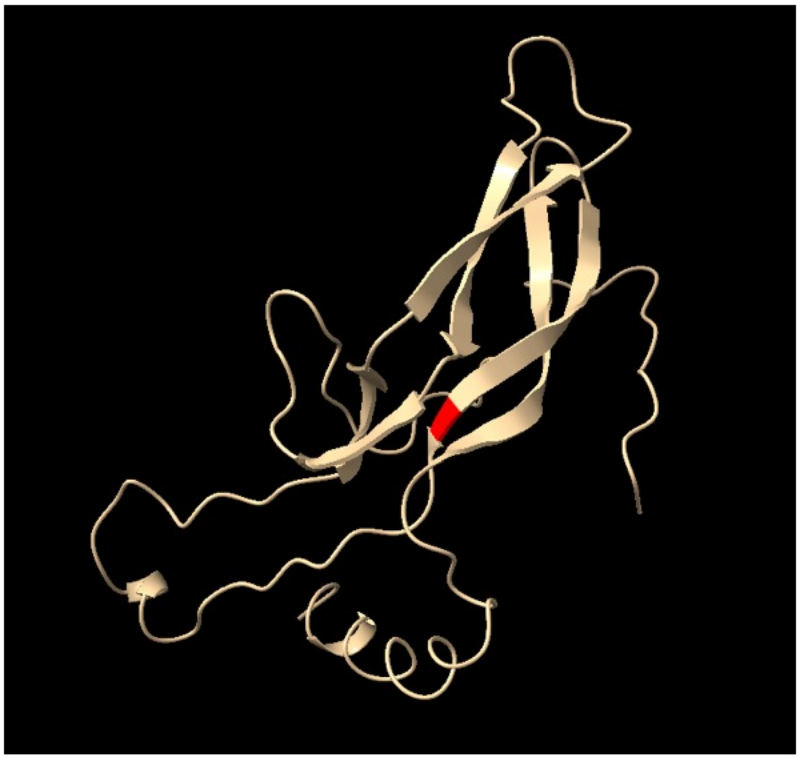
Supplemental Figure 1. Protein modeling in silico was performed for TSHβ, wild type (left) and mutant (right). Mutation site (in red) causes significant change in the three-dimensional predicted structure as shown. Methodology for modeling: Kallberg M, Wang H, Wang S, Peng J, Wang Z, Lu H, Xu. Template-based protein structure modeling using the RaptorX web server. Nat Protoc 2012;7:1511-1522. Ma J, Peng J, Wang S, Xu J. A conditional neural fields model for protein threading. Bioinformatics 2012;28:i59-i66. Ma J, Wang S, Zhao F, Xu J. Protein threading using context-specific alignment potential. Bioinformatics 2013;29:i257-i265.

Genes Related to thyroid disorder								
AADAT	*DUOX1*	*GNAS*	*KMT2D*	*POU1F1*	*RYR2*	*SLC30A10*	*THRB*	*TTR*
ALB	*DUOXA1*	*HHEX*	*NCOR2*	*PROP1*	*SALL1*	*SLC5A5*	*TPO*	*TUBB1*
ALMS1	*DUOX2*	*HOXA3*	*NKX2-1*	*PSMA1*	*SECISBP2*	*SLCO1C1*	*TRH*	*UBR1*
ATXN2	*DUOXA2*	*IGSF1*	*NKX2-5*	*PSMA3*	*SERPINA7*	*STAMBP*	*TRHR*	*VAV3*
CDCA8	*EXOSC2*	*IRS4*	*NKX2-6*	*PSMD2*	*SLC16A2*	*TBL1X*	*TRIP11*	
DIO1	*FGF8*	*IYD*	*NTN1*	*PSMD3*	*SLC17A4*	*TBX1*	*TRIP12*	
DIO2	*FOXE1*	*JAG1*	*P4HB*	*PTH1R*	*SLC26A4*	*TG*	*TSHB*	
DIO3	*GLIS3*	*KDM6A*	*PAX8*	*PTRH2*	*SLC26A7*	*THRA*	*TSHR*	

TSHB Protein Modeling



Wild Type



c.T141G p.C47W