Supplementary Data

Supplementary Table S1. Prediction of Functional Effects of the Variants of *DUOX2* by *In Silico* Assays

Variants	*Coding DNA reference sequence*	*RS number*	*PolyPhen-2*[a]	*SIFT score*[b]	*CADD*[c]	*AAF*[d]	*VCF*[e]
p.A649E	c.1946 C>A	rs748793969	0.736	0	17.13	0.04%	15:45398525 G>T
p.R885L	c.2654 G>T	rs181461079	0.631	0	26.4	0.4%	15:45396158 C>A
p.P1391A	c.4171 C>G	rs771198569	0.542	0	29	0.01%	15:45387703 G>C
p.G488R	c.1462 G>A	rs191759494	1	0	33	0.2%	15:45400357 C>T
p.SF965-6SfsX29	c.2895_2898 delGTTC	rs530719719	NA	NA	NA	0.6%	15:45393426_45393 429delGAAC

[a]Polymorphism Phenotyping v2 HumVar (Polyphen2) predicts the variants to be benign, possibly damaging, or probably damaging corresponding to probability interval (0–0.2), (0.2–0.85) and (0.85–1), respectively.

[b]SIFT sequence homology-based tool sorts intolerant from tolerant amino acid substitutions and predicts a phenotypic effect, scores <0.05 are deleterious.

[c]Combined annotation dependent depletion (CADD) scores ≥ 12 are predicted to be damaging, and scores ~ 25 are consistent with a dominant phenotype.

[d]Alternative allele frequency (AAF) is the maximum frequency of the alternative allele (relative to the human reference genome) in any population in The Exome Aggregation Consortium, 1000 Genomes, or Exome Varient Server.

[e]VCF; variant name in VCF notation for human genome reference hg19 in antisense orientation.

NA; not available; RS number, reference single nucleotide variants number.