Table SI. Different forms of congenital hypothyroidism and associated causative genes selected in the present study.

Thyroid alteration		Gene symbol	Full name	RefSeq (genome) accession number	RefSeq (mRNA) accession number	CDS length	Number of exons	Protein function
Primary congenital hypothyroidism	Thyroid dysgenesis	*PAX8*	Paired box gene eight	NG_012384.1	NM_003466	1353	12	A thyroid enriched transcription factor, involved in thyroid follicular cell development and expression of thyroid-specific genes.
		FOXE1（TTF-2）	Thyroid transcription factor 2	NG_011979.1	NM_004473.3	1122	1	A transcription factor functions in thyroid development
		NKX2-5	NK2 transcription factor related, locus 5	NG_013340.1	NM_004387.3	975	2	A transcription factor functions in heart formation and development
		TSHR	Thyroid stimulating hormone receptor	NG_009206.1	NM_000369.2	2295	10	A receptor for thyrothropin and thyrostimulin, a major controller of thyroid cell metabolism
		NKX2-1 (TTF-1)	NK2 homeobox 1	NG_013365.1	NM_001079668.2	1206	3	A thyroid-specific transcription factor，regulates the expression of thyroid-specific genes and genes involved in morphogenesis
	Dysormonogenesis	*DUOX2（THOX2)*	Dual oxidase 2	NG_009447.1	NM_014080.4	4647	34	H_2O_2 generation in the follicle
		DUOXA2	Dual oxidase maturation factor 2	NG_016992.1	NM_207581.3	963	6	Necessary for proper cellular localization and maturation of functional dual oxidase 2.
		TPO	Thyroid peroxidase	NG_011581.1	NM_000547.5	2802	17	Catalyses the oxidation, organification, and coupling reactions
		SLC26A4(PDS)	Solute carrier family 26, member 4 (Pendred syndrome,	NG_008489.1	NM_000441.1	2343	21	Transports iodine across apical membrane

			PDS					
		TG	Thyroglobulin	NG_015832.1	NM_003235.4	8307	48	Supports for thyroid hormone synthesis
		SLC5A5(NIS)	Solute carrier family 5 (sodium iodide symporter), member 5	NG_012930.1	NM_000453.2	1932	15	Transports iodine across basal membrane
		IYD (DEHAL1)	Iodotyrosine deiodinase	: NG_016007.1	NM_203395.2	870	5	Nitroreductase-related enzyme capable of deiodinating iodotyrosines
		SECISBP2	SECIS binding protein 2	NG_012177.1	NM_024077.3	2565	17	Essential bound protein mediating the correct binding of selenium with thyroxine deiodinated enzyme
Secondary or Central hypothyroidism	Isolated central congenital hypothyroidism(isolated TSH deficiency)	*TSHB*	(Aliases: TSH-B, TSH-BETA) Thyroid stimulating hormone beta	NG_015891.1	NM_000549.3	417	3	The beta subunit of thyroid stimulating hormone, functions in the control of thyroid structure and metabolism
		IGSF1	Immunoglobulin superfamily, member 1	NG_021190.1	NM_001170961.1	4026	20	May be involved in the expression of normal TRHR expression, causative gene of X-linked central hypothyroidism
		TRHR	Thyrotropin-releasing hormone receptor	NG_017161.1	NM_003301.5	1197	2	AG protein-coupled receptor for thyrotropin-releasing hormone
	Combined pituitary hormone deficiencies(Deficiency in pituitary transcription factors)	*HESX1*	Homeobox gene expressed in ES cells	NG_008242.1	NM_003865.2	558	4	A transcriptional repressor in the developing forebrain and pituitary gland
		LHX3	LIM homeobox protein 3	NG_008097.1	NM_014564.3	1209	6	A transcription factor that is required for pituitary development and motor neuron specification
		LHX4	LIM homeobox protein 4	NG_008081.1	NM_033343.3	1173	6	A transcription factor involved in the control of differentiation and

								development of the pituitary gland
		POU1F1（PIT1）	POU domain, class 1, transcription factor 1	NG_008225.2	NM_000306.2	876	6	Regulates the expression of several genes involved in pituitary development and hormone expression.
		PROP1	Prophet of Pit1, paired-like homeodomain transcription factor	NG_015889.1	NM_006261.4	681	3	A transcription factor responsible for pituitary development and hormone expression
		SOX3	SRY (sex determining region Y)-box 3	NG_009387.1	NM_005634.2	1341	1	A transcription factor involved in the regulation of embryonic development and in the determination of the cell fate.
Peripheral hypothyroidism	thyroid hormone resistance	*THRB*	Thyroid hormone receptor, beta	NG_009159.1	NM_000461.4	1387	10	A nuclear hormone receptor for triiodothyronine, mediating the biological activities of thyroid hormone.
		THRA	Thyroid hormone receptor, alpha	NG_023345.1	NM_199334.3	1473	9	A nuclear hormone receptor for triiodothyronine, mediating the biological activities of thyroid hormone.
	thyroid hormone transprotation abnormality	*SLC16A2 (MCT8)*	Solute carrier family 16 (monocarboxylic acid transporters), member 2 (putative transport)	NG_011641.1	NM_006517.4	1620	6	Mediating the uptake of thyroid hormones into the brain
Syndromic hypothyroidism	Pendred syndrome	*SLC26A4(PDS)*	Solute carrier family 26, member 4 (Pendred syndrome, PDS	NG_008489.1	NM_000441.1	2343	21	Functions as both a repressor and activator of transcription and is specifically involved in the development of pancreatic beta cells, the thyroid, eye, liver and kidney.

		FOXI1	Forkhead box i1	NG_012068.1	NM_012188.4	1137	2	May play an important role in the development of the cochlea and vestibulum, as well as in embryogenesis.
	Neonatal diabetes & congenital hypothyroidism	*GLIS3(ZNF515)*	Glis family zinc finger 3	NG_011782.1	NM_152629.3	2328	10	Functions as both a repressor and activator of transcription and is specifically involved in the development of pancreatic beta cells, the thyroid, eye, liver and kidney.
	Bamforth-Lazarus syndrome	*FOXE1*（*TTF-2*）	Thyroid transcription factor 2	NG_011979.1	NM_004473.3	374	1	A transcription factor functions in thyroid development
	Choreoathetosis	*NKX2-1 (TTF-1)*	NK2 homeobox 1	NG_013365.1	:NM_001079668.2	402	3	A thyroid-specific transcription factor，regulates the expression of thyroid-specific genes and genes involved in morphogenesis
	Johanson–Blizzard syndrome	*UBR1*	Ubiquitin protein ligase E3 component n-recognin 1	NG_012182.1	NM_174916	1750	47	Responsible for facilitating the direct ubiquitination of substrate proteins
Transient congenital hypothyroidism		*ZNF252P*	Zinc finger protein 252, pseudogene	NG_12047.2	NM_000465.2	5210	5	A candidate gene for a patient's transient neonatal thyroid dysfunction
		DUOXA2	Dual oxidase maturation factor 2	NG_016992.1	NM_207581.3	321	6	Necessary for proper cellular localization and maturation of functional dual oxidase 2.

RefSeq, reference sequence; CDS, coding DNA sequence.