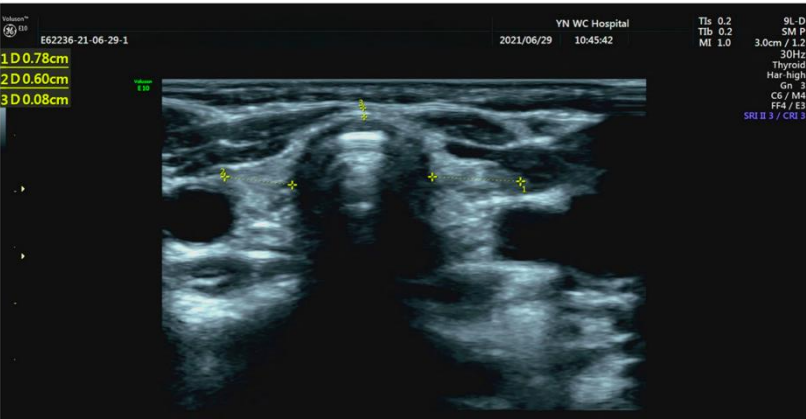
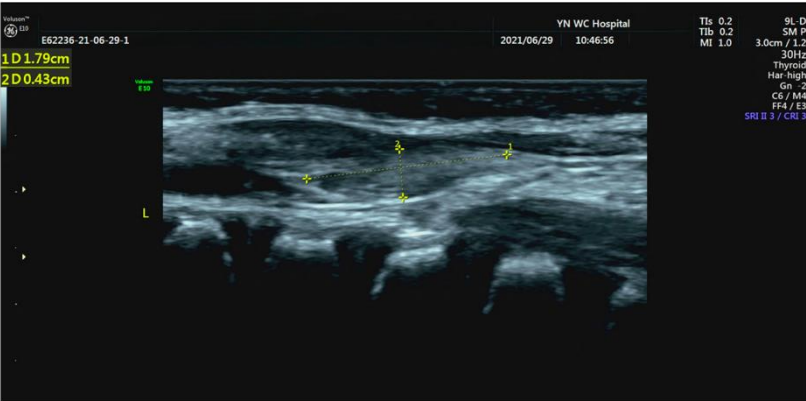
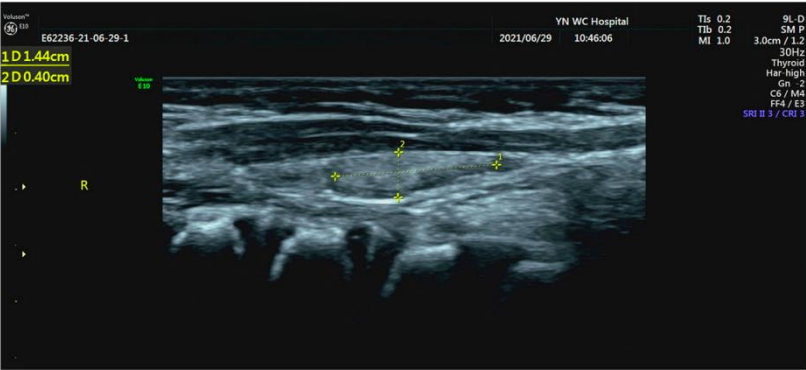


**Table S1.** Function and conservative prediction of variants.

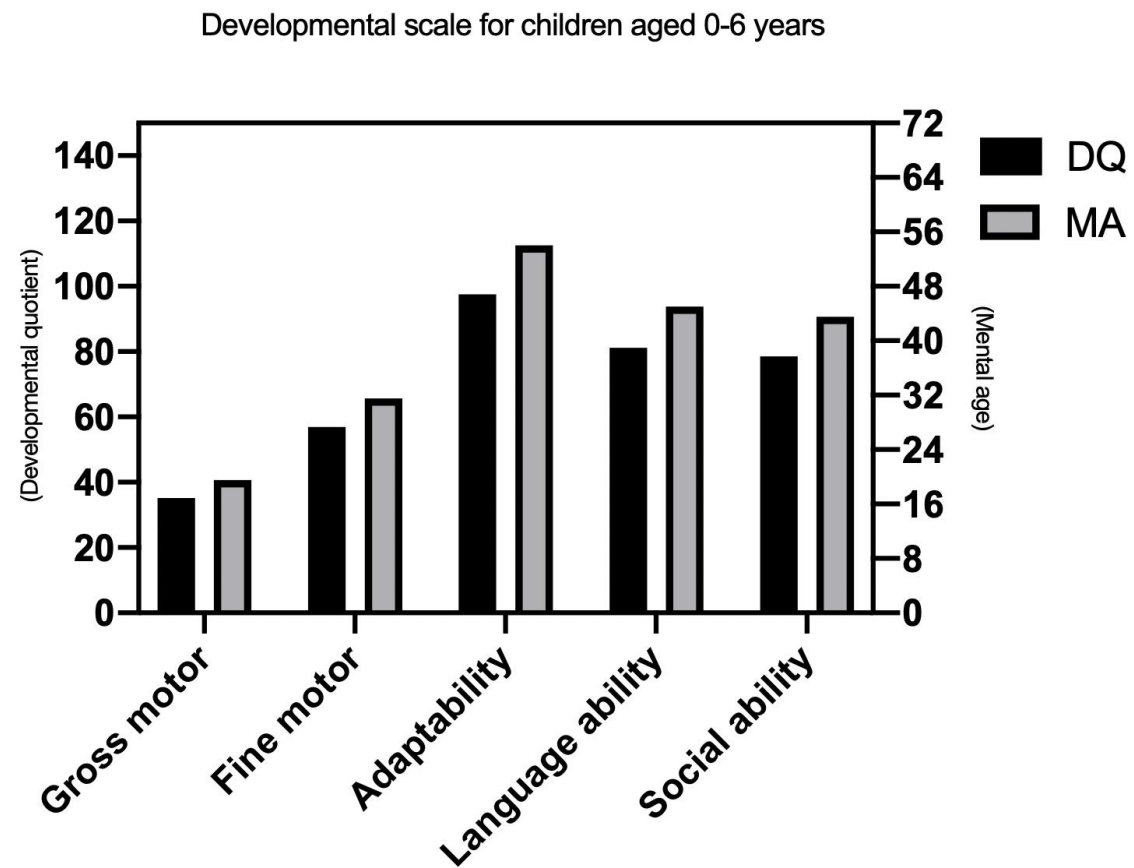
Variants	Polyphen-2_HVAR	SIFT	MutationTaster	PROVEAN	ClinPred	REVEL	GERP++	MutationAssessor
PAX8: c.149A>C	Probably_ damaging	Damaging	Disease_ causing	Damaging	pathogenic	Damaging	Conserved	High
PAX8: c.329G>A	Probably_ damaging	Damaging	Disease_ causing	Damaging	pathogenic	Damaging	Conserved	Low
NKX2-1: c.706A>G	Probably_ damaging	Damaging	Disease_ causing	Damaging	pathogenic	Damaging	Conserved	Medium

**Table S2.** Primers used for the expression vectors and thyroglobulin (TG)-promoter cloning.

Vector	Forward primer	Reverse primer	Variant/Insert
TG-pGL3	GATCTGGATCTAAGTAAGCTTCAGGCAG AGTGGATCTGAGC	CAGTACCGGAATGCCAAGCTTTTCCTG GGAGGAAGGAGGAG	TG promoter sequence (-411/+89)
PAX8-c149- pCMV3	ACATCTCTCGCCCGCTCCGCGTCAGCCA TGGC	GAGCGGGCGAGAGATGTCGCAGGGCC TTACAC	c.149A>C (p.Q50P)
PAX8-c329- pCMV3	ATCCGAGACCAGCTCCTGGCTGAGGGCG TCTG	AGGAGCTGGTCTCGGATCTCCCAGGCA AACAT	c.329G>A (p.R110Q)
NKX2-1-c706-pCMV3	CAGGTCGAGATCTGGTTCCAGAACCACC GCTA	AACCAGATCTCGACCTGCGTGGGCGTC AGGTGGAT	c.706A>G (p.K236E)
NKX2-1-GFP-pcDNA3.1	TAACGGCCGCCAGTGTGCTGGAATTCAT GTGGTCCGGAGGC	TTGCTCACCATGGTGGGCCAGGTCCGA CCGTATA	<i>NKX2-1</i> cDNA sequence



**Figure S1.** The thyroid ultrasound of P3. The sectional size of bilateral thyroid lobes: 1.44 \* 0.78 \* 0.40cm on the right (reference values for 4 years old, 2.79\*0.86\*0.69 cm), 0.08cm thick at the isthmus (reference value 0.14 cm), 1.79 \* 0.60 \* 0.43 cm on the left (reference values 2.66\*0.94\*0.60 cm).



**Figure S2.** The developmental evaluation of P3 at 55 months. The average developmental quotient is 69.9 (The reference values of score for 0-3-year-old children, 90-115), and the average intellectual age is 38.7 months.