INTRODUCTION
Thyroid hormone is known as greatly influence on growth and development in fetuses and newborns. If the detection of the disease is delayed, hypothyroidism can cause irreversible damage, so early detection and treatment is very important. Hypothyroidism can be divided into permanent and temporary cases depending on the duration of treatment, but there is no predictor that can completely differentiate those two. However, as genes related to hypothyroidism are revealed, genetic analysis can help predicting whether hypothyroidism will be transient.

AIM
Analyzing causative genetic variations and clinical characteristics with congenital hypothyroidism to predict disease persistence.

METHOD
Subjects: 147 congenital hypothyroidism patients who want to implement the hypothyroidism NGS panel were enrolled (Male n=84, 57%).
Hypothyroidism NGS panel covers 30 genes: DUOX2, DUOX1, FOXE1, GNAS1, HESX1, IGF1, JAG2, KCNJ18, Lhx8, MC2R, MCT8, MECT1, MTC1, MTHFR, NIP1, NIS, NR5A1, OTX2, PAX3, PAX6, PAX8, PTH1R, PROP1, RASAL1, RAX, RET, SLCO1B1, SUPT5H, TG, THRA, THRB, TPO, TRH, TRHRI, TSH, TSHRI, LHX4, SOX2, GLIS3, OTX2, DUOX1, ISGF1, SOX3.

RESULTS

CONCLUSIONS
It is expected that causative genetic analysis of congenital hypothyroidism will be helpful in actively stopping the treatment of congenital hypothyroidism according to the clinical condition of the patient, except when the disease is estimated due to pathogenic or likely pathogenic genes.

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ANALYSIS OF HYPOTHYROIDISM NGS TEST IN KOREAN PATIENTS WITH CONGENITAL HYPOTHYROIDISM IN A SINGLE CENTER

Table 1. Genes and the frequency which were detected as VOUS. 121 VOUS variants were identified in 90 patients.

Among 147 patients, 50 patients (34.0%) had known pathogenic or likely pathogenic genes and 33 patients (22.4%) had none of related mutations. There were 9 eutopic thyroid, 4 agenesis, 1 hemiplasia patient and 33 patients could stop treatment until now.

Table 2. Clinical and the related genetic information of 15 transient congenital hypothyroidism patients who could stop medication at 1 year old.

Table 3. Clinical and the related genetic information of 18 transient congenital hypothyroidism patients who could discontinue the treatment after 3 years old.

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