Association between thyroid specific genotypic variation and phenotypic expression of dyshormonogenetic goiter and Hashimoto’s thyroiditis in children and adolescents

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OBJECTIVES

**Background:** Though very common, thyroid specific genetic studies on hypothyroidism in children is lacking from South India

In children and adolescents - with hypothyroidism due to dyshormonogenetic goiter (DH) and Hashimoto’s thyroiditis (HT)

- To screen for NIS, DUOX2 and TPO gene mutations
- To correlate between mutations and hypothyroid phenotype

METHODS

- Genomic DNA was extracted from peripheral blood leucocytes
- PCR and direct sequencing were used to analyse for NIS, DUOX2 and TPO genes
- Detailed clinical, biochemical and follow-up data were recorded in a structured proforma
- Subjects with hypothyroidism were treated with thyroxine replacement
- Detailed genetic analysis with 142 SNP (single nucleotide peptides) and 8 sets of primers were done.

RESULTS

- Age of the cohort was 11 ± 4.5 (5 – 17) years
- F: M ratio was 17:3
- Hypothyroidism was overt and subclinical in 14 and 6 patients respectively
- Family history of hypothyroidism was present in 7 patients (35%)
- Genetic analysis shows that heterozygous NIS mutations were seen in 5 children with HT and in 3 children with DH
- A homozygous mutation was picked up in a child with HT
- Heterozygous TPO mutations found in 2 cases of HT
- No mutation was found in DUOX gene

A variation from C>T(homozygous mutation) was found at 17983561 position( rs:740695),06(HT)

CONCLUSIONS

- NIS gene mutations appears to be most prevalent mutations in HT and DH amongst South Indian children in this study
- The iodine deficiency and ethnic factors may be responsible for this pattern. Further studies are needed to characterize hypothyroid phenotypes in children.

(Key words: Dyshormonogenetic goiter, Hashimoto’s thyroiditis, NIS mutation, Genotype, Phenotype)