NEW CONTRIBUTIONS OF THE THESIS

"Detection of some gene mutations and genotype-phenotype correlation of thalassemia pediatric patients at hai phong children's hospital"

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SCIENTIFIC INSTRUCTORS:

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- Research to determine the rate of disease-causing gene mutations and genetic characteristics of the disease through studying a number of pedigrees of Thalassemia patients being treated at Hai Phong Children's Hospital.

- Applying molecular biology techniques to determine the type of mutation and other tests (total blood cell analysis, blood biochemical analysis, ...) to evaluate and contribute to the screening of Thalassemia disease.

- Develop and introduce a research of disease genealogies to manage genetic resources and provide genetic counseling

- Research is quite comprehensive on Thalassemia disease: Disease causes (mutant type), inheritance through generations, clinical and subclinical symptoms, treatment and monitoring of treatment results

- Detecting rare mutations in the world in particular and Vietnam in general.