Information on Doctoral thesis of Fellows Truong Thi Hue

1. Full name: Truong Thi Hue

2. Sex: Female

3. Date of birth: 20/05/1976

4. Place of birth: Khanh Hoa

5. Admission decision number: 5429/QĐ-SĐH dated 30/10/2008 by the President of Vietnam National University, Hanoi.

6. Changes in academic process: Decision 737/QĐ-SĐH-TN, Dated 14/06/2010 on modification of the thesis title by the Rector of VNU University of Science.

7. Official thesis title: Investigation of some mitochondrial genome mutations in Vietnamese encephalomyopathy patients.

8. Major: Biochemistry

9. Code: 62.42.30.15

10. Supervisors:

1. Assoc. Prof. Dr. Phan Tuan Nghia

2. Assoc. Prof. Dr. Nguyen Thi Van Anh

11. Summary of the new findings of the thesis:

- The procedures for detection of 15 different point mutations including T3271C and T3291C (MELAS syndrome); A8344G and T8356C (MERRF syndrome); G11778A, G3460A and T14484C (LHON syndrome); T8993G/C and T9176G (Leigh syndrome); A1555G (deafness syndrome); G4298A, T10010C, T14727C, T14728C and T14709C (encephalomyopathy syndrome) in the human mitochondrial genome were established by using PCR-RFLP methods in combination with nucleotide sequencing. 106 patients with encephalomyopathy symptoms were screened for the mutations and it was found that six patients (5.7%) carried A3243G mutation, 2 patients (3.9%) carried T14727C mutation and one patient (0.9%) carried G8251A mutation.

- 72 patients with the encephalomyopathy symptoms were screened for the presence of the 9 bp-deletion and 23 patients were found to carry the 9 bp-deletion.

The correlation between the 9 bp-deletion and 15 screened pathogenic mutations was studied but no reliable correlation was found.

- A procedure for the detection and determination of the percentage of the A3243G mutation (MELAS) by real-time PCR using Taqman LNA (locked nucleic acid) fluorescent probes was established and a positively proportional correlation between the mutation percentage and disease severity was found.

- The T14727C mutation in the MTTE gene encoding for mitochondrial transfer RNA of glutamic acid ($tRNA^{Glu}$) in encephalomyopathy patients was reported for the first time in this study and it was maternally inherited.

12. Practical applicability:

The procedure for detection mitochondrial gene mutations established in this study can be easily developed into protocols for examination of mitochondrial genome mutations-related diseases in hospitals.

13. Further research directions:

- Further screening of various pathogenic mitochondrial gene mutations in patients with mitochondrial disorder diseases.

- Further development of the procedure for quantitation of the heteroplasmy of different mitochondrial gene mutations to find the correlation between the percentage of the mutation and disease status.

14. Thesis-related publications:

[1]. Truong Thi Hue, Pham Minh Hue, Le Ngoc Yen, Pham Thi Van Anh, Ngo Diem Ngoc, Phan Tuan Nghia (2012), “Detection of 9-bp deletion in mitochondrial genome in Vietnamese patients with suspected mitochondrial encephalomyopathy”, *Journal of Biology* 34 (2), pp. 246-252.

[2]. Truong Thi Hue, Nguyen Van Lieu, Pham Van Anh, Nguyen Thi Van Anh, Phan Tuan Nghia (2012), “Detetion and quantitation of A3243G mutation in the human mitochondrial genome using PCR-RFLP and real-time PCR with LNA Probes”, *Journal of Biotechnology* 10 (3), pp. 421-427.

[3]. Truong Thi Hue, Phan Tuan Nghia, Nguyen Thi Van Anh, Pham Le Anh Tuan, Pham Van Anh (2012), “A new mutation in the gene encoding for mitochondrial transfer RNA of glutamic acid

(tRNA^{Glu}) in two Vietnamese encephalomyopathy patients", *VNU Journal of Science* 28 (2S), pp. 123-128.