

## Abstract (บทคัดย่อ)

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**Project Code** : MRG60800172

**Project Title** : การศึกษาการกลายพันธุ์ของยีน STAT6 ต่อการเกิดโรคภูมิแพ้

**Investigator** : ผู้ช่วยศาสตราจารย์แพทย์หญิง นริศรา สุรทนต์นันท์  
หน่วยภูมิแพ้และภูมิคุ้มกัน ภาควิชากุมารเวชศาสตร์  
คณะแพทยศาสตร์ จุฬาลงกรณ์มหาวิทยาลัย

**E-mail Address**: mayzped@gmail.com

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**Objective** : To investigate the functional consequences of the STAT6 mutation

**Methods** : We described a child who had idiopathic anaphylaxis in infancy, atopic dermatitis and allergic eosinophilic gastroenteritis with protein-losing enteropathy. His father had atopic dermatitis and food allergy. Both were heterozygous mutations in *STAT6* DNA binding domain. Flow cytometric analysis, cytokine measurements by ELISA/ Luminex assays from the patient samples, luciferase assay and immunofluorescence in *STAT6* mutants compared to wild type transfectants were performed to prove that the mutation resulting in activation of STAT6.

**Results** : Various experiments confirmed that the mutants are pathogenic. 3-D Structural modelling of STAT6 revealed that the *STAT6* mutation is located at the surface of the STAT6 DNA binding region, potentially having a stronger binding affinity. Immunofluorescence study confirmed that mutant *STAT6* preferentially localized in the nucleus. Constitutively DNA binding activation of STAT6 mutants was also detected through HEK293T cell luciferase assays. Flow cytometric-based analysis of T helper cell populations and intracellular cytokine measurements from patient cells showed that immune responses of the patient directed toward a