



## The Chinese University of Hong Kong Non-confidential Abstract of Technology Disclosure

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**Title:**

### **A Rapid Test for Detection of Specific HLA-genotype**

**CUHK Ref. No.:**

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**Inventor(s):**

Professor Margaret HL NG, Department of Anatomical and Cellular Pathology

**Non-confidential abstract:**

This technology serves as a prototype for rapid and inexpensive detection of specific HLA genotype using HLA-B\*1502 as an example. The rapid test for B\*1502 allele by simple set-up and at low costs has far reaching pharmacogenetic implications in health care quality and resource allocation.

It has been demonstrated by us and others that carriers of HLA-B\*1502 allele, a high prevalence (~20%) allele in Asia including Hong Kong, have a dramatically elevated risk for developing the potentially fatal carbamazepine induced Steven Johnson Syndrome/Toxic Epidermal Necrolysis (C-SJS/TEN). SJS/TEN are severe adverse reactions characterized by a rapidly developing blistering exanthema of macules and target-like lesions accompanied by extensive and severe mucosal detachments with potential life threatening consequences. Morbidities are significant and fatality has been reported in 10-30% of the cases. The high positive (93.6%) and negative (100%) predictive values of B\*1502 carrier status for C-SJS/TEN reported by a previous study in Taiwan confirmed the value of B\*1502 as a good predictive genetic marker for C-SJS/TEN. Carbamazepine is a first-line drug for patients with epilepsy, neuropathic pain and bipolar affective disorder. These diseases account for up to 10% of the general population. Thus, testing for HLA-B\*1502 before prescription of carbamazepine is essential to help avoid the severe adverse drug reactions in susceptible individuals.

However, the existing technologies of HLA testing are expensive and time-consuming. To meet the health care needs, we have developed a new simple test which can rapidly (within 1 hour) and accurately detect HLA-B\*1502 based on loop-mediated isothermal amplification principles. The test involves specially designed primer sets which can efficiently detect HLA B\*1502 accurately at an isothermal condition. No prior DNA extraction is necessary, which allows more rapid production of results and can be performed easily by clinicians or nurses untrained for genetic testing. Huge costs would be saved from the cheaper test, faster results, shorter hospital stays, earlier disease control and no more need of managing C-SJS/TEN. More importantly, our study on B\*1502 may serve as a prototype in the approach on HLA genotyping. The provision of simple and cheap HLA testing may also imply that a statistically powerfully large sample size could be achieved in HLA association research, particularly in the validation study of rare target HLA allele. The success of this approach as proved in our study will certainly revolutionize HLA testing in both clinical and research arenas.

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