Scientists identify gene responsible for severe skin condition

March 23 2011

The drug, called carbamazepine, is commonly used to treat patients with epilepsy and other diseases such as depression and trigeminal neuralgia. Although successful in treating the majority of patients, carbamazepine can cause side-effects that range from a mild skin irritation to severe blistering of the whole body.

The team, in collaboration with the Wellcome Trust Sanger Institute, screened more than a million variants in DNA across the human genome to understand why some patients are more prone to the drug's side-effects than others. Research in Taiwan has already identified a gene that predisposes Asian patients to the skin condition, but Liverpool scientists discovered that this gene could not be used to predict the reaction in Caucasian people.

Researchers have now identified a gene, called HLA-A*3101, in Caucasian patients that increases the risk of developing a reaction to the drug from 5% to 26%. Researchers are now working with clinicians and drug regulators to investigate how these new findings can translate into clinical practice.

Professor Munir Pirmohamed, NHS Chair in Pharmacogenetics from the University's Wolfson Centre for Personalised Medicines, said: "Adverse drug reactions are a major cause of hospital admissions. Carbamazepine is widely used and the majority of patients respond well to the treatment, but a small percentage develop skin conditions that in severe circumstances can result in painful blistering all over the body.

"For the first time we have found a significant link between the drug and the skin condition in Caucasian people that also complements the findings in Asian patients. We can now begin to work with clinicians and regulators to maximise the benefits of the drug and minimise the side-effects."

Dr Ana Alfirevic, from the University's Wolfson Centre for Personalised Medicine, said: "This is a significant finding that highlights the importance and advancement of new genetic technologies. We aim to support the development of medicines based on a patient's unique genetic make-up to allow clinicians to prescribe the most effective and safe treatments."

Dr Gianpiero Cavalleri, from the Royal College of Surgeons, said: "Rapid advances in genetic technology, together with a strong collaborative effort, have allowed us to make this important advance, which should make the treatment of epilepsy safer. Working with colleagues in Europe and the US, we have been able to access large numbers of patients to allow us to investigate common genetic trends and the mechanisms that result in this potentially serious condition."

More information: Research conducted at the University was funded by the Department of Health and is published in the *New England Journal of Medicine.*

Provided by University of Liverpool