

Patients predisposed to blood clots present genetic markers

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UCD researchers jointly led by Conway Fellow, Dr. Patricia Maguire and Professor Des Fitzgerald, Professor of Molecular Medicine & Vice-President for Research, have identified new genes that contribute to platelet function and promote blood clot formation. Their findings are described recently in a leading haematology journal, *Blood*.

This collaborative research is a significant advance for the Bloodomics consortium, a European wide effort by scientists to uncover genetic markers that can identify those people most at risk of clot formation and heart attacks.

Platelets are small blood cells that circulate in the bloodstream until they are called into action. When activated, platelets trigger a sequence of events that cause a blood clot to form. This is a normal response when a blood vessel is damaged but, when a blood clot forms as part of a stroke or heart attack, it can deprive the heart muscle of vital oxygen.

Unravelling the genetic roots of atherothrombosis and cardiovascular disease will enable better screening for those most at risk of these diseases and offer targeted therapeutic interventions.

The research carried out in UCD Conway Institute initially identified 63 genes linked to a variation in platelet response. The team carried out further investigation on six of these genes and were able to show that two resulting proteins in particular, *commd7* and *lrrfip1*, have a significant role in regulating thrombus formation.

Commenting on the work, Dr. Maguire said, “By tracing the cellular network and understanding the function of the proteins involved in the platelet response, we want to provide new insight into the causes of [heart disease](#). This work is a substantial step towards our goal of developing novel platelet inhibitors that can lower the morbidity and mortality associated with these prevalent diseases.”

Maguire and her team also published significant work last year in the *Proceedings of the National Academy of Sciences* describing a novel platelet signalling network that dampens [platelet](#) function. They believe that this signalling pathway may not be correctly regulated in heart disease formation and its discovery is an important development to find new drugs in the fight against heart disease. Dr Brian Steele, the first author on that manuscript, won a prestigious young investigator award at the 2009 International Society of Thrombosis and Haemostasis meeting in Boston for his work on this subject.

More information: Maguire and her team also published significant work last year in the *Proceedings of the National Academy of Sciences* describing a novel platelet signalling network that dampens platelet function.

Provided by University of Dublin

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