Mystery bleeding disorders could be unraveled by new research efforts

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(Medical Xpress)—Platelet disorders are heavily underdiagnosed, little understood and cannot be cured. University of Birmingham researchers and the Birmingham Platelet Group are running a UK-wide clinical trial 'Genotyping and platelet phenotyping' (GAPP) funded by the British Heart Foundation (BHF). The trial includes lab research by BHF-funded scientist Dr Yotis Senis in an effort to better identify patients and potentially to develop new treatments.

A breakthrough from Dr Senis – published in the journal *Science Signalling* – has revealed the lack of a protein called G6b-B could be behind some platelet disorders. Clinicians running the GAPP trial – led by BHF Professor Steve Watson and co-ordinated by Dr Gill Lowe – can now screen patients by testing the gene related to their G6b-B protein. Studying those identified could then help Dr Senis and his team to develop targeted treatments for sufferers with this mutation.

Dr Senis said: "We pass information back and forth between basic scientists and clinicians– much like a game of ping-pong. Discoveries we make in the lab provide information for screening patients. The characteristics of the screened patients then feed back into our basic science research. By collaborating in this way we are quickly starting to build a picture of the genetics and associated characteristics of a whole range of the platelet disorders out there."

The estimated 2,000 people in the UK with inherited platelet disorders can experience bruising from the slightest pressure - by wearing a watch

or rucksack for example - and can suffer spontaneous nose bleeds. Noah Edwards was diagnosed with platelet function disorder with thrombocytopenia at the age of one, and has been involved in the GAPP clinical study to improve knowledge on problems with platelet function.

Ruby Edwards, Noah's mother and founder of platelet disorder charity Funny Blood, is hugely encouraged by the work at Birmingham:

"It's very difficult when your child is diagnosed with a condition which is so little understood. I am delighted that Noah is among the patients who have been studied in GAPP as it brings us closer to understanding these rare yet important conditions."

Dr Hélène Wilson, BHF Research Advisor, said "In this paper the authors show that the protein G6b-B appears to play a key role, previously unknown, in platelet function, in mice. Indeed, mice lacking this protein show signs of platelet-based bleeding disorders. Further experiments are needed in humans to investigate the role of this protein in disease.

"As well as potentially benefiting patients with rare clotting disorders, we hope this research will also open up new therapeutic avenues to develop treatments which prevent the deadly blood clots that can cause heart attacks and strokes."

More information: stke.sciencemag.org/cgi/conten … /abstract/5/248/ra78

Provided by University of Birmingham