Mystery bleeding disorders could be unravelled by new research efforts at University of Birmingham

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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) (1) 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 (2) – 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 (3), 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."

Notes to editors

(1) The GAPP (genotyping and platelet phenotyping) study looks at patients with excessive bleeding to see if they have an underlying platelet defect and to classify the nature of the defect and uncover the genetic reason for the change in platelet function. The trial is led by Dr Gill Lowe at the University of Birmingham and is headed by BHF Chair Professor Stephen Watson.

(2) Paper available online at: **stke.sciencemag.org/cgi/content/abstract/5/248/ra78?rss=1 (http://stke.sciencemag.org/cgi/content/abstract/5/248/ra78?rss=1)** DOI 10.1126/scisignal.2002936

(3) Funny Blood was founded in 2009 and is currently applying to be incorporated as a charitable trust to fund research and raise awareness of platelet function disorders (PFDs). The aim of the not-for-profit organisation is to help fund research into PFDs and support projects such as GAPP. Over the last year Funny Blood has grown and received both local and national publicity, including meeting with David Cameron at 10 Downing Street and appearing on BBC Breakfast TV.

The British Heart Foundation (BHF) is the nation's heart charity, dedicated to saving lives through pioneering research, patient care, campaigning for change and by providing vital information. But we urgently need help. We rely on donations of time and money to continue our life-saving work. Because together we can beat heart disease.

For more information visit **bhf.org.uk/pressoffice (http://www.bhf.org.uk/pressoffice)**

Funny Blood is a not-for-profit organisation founded to raise awareness of rare blood conditions called platelet function disorders. Funny Blood raises funds for research and awareness of the condition. We do – because nobody else does.

For more information visit **funnyblood.co.uk (http://www.funnyblood.co.uk)**

For media enquiries, please contact Jenni Ameghino, Media Relations Manager, University of Birmingham, 0121 415 8134 / pressoffice@contacts.bham.ac.uk

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