University of Birmingham

Publications

Please see below lists of publications resulting from the GAPP project:

Open all sections

2015

Jones ML, Norman JE, Morgan NV, Mundell SJ, Lordkipanidzé M, Lowe GC, Daly ME, Simpson MA, Drake S, Watson SP, Mumford AD; on behalf of the UK GAPP study group(2015) **Diversity and impact of rare variants in genes encoding the platelet G protein-coupled receptors (http://www.ncbi.nlm.nih.gov/pubmed/?term=Diversity+and+impact+of+rare+variants+in+genes+encoding+the+platelet+G+protein-coupled+receptors)** . *Thromb Haemost*113(3) [Epub ahead of print]

2014

Leo V, Morgan N, Bem D, Jones M, Lowe G, Lordkipanidzé M, Drake S, Simpson M, Gissen P, Mumford A, Watson S, Daly M; The UK GAPP Study Group (2014) **Use of next generation sequencing and candidate gene analysis to identify underlying defects in patients with inherited platelet function disorders (http://www.ncbi.nlm.nih.gov/pubmed/?term=10.1111%2Fjth.12836)** . *J Thromb Haemost*[Epub ahead of print]

Nisar S, Lordkipanidze M, Jones ML, Dawood BB, Murden S, Cunningham MR, Mumford AD, Wilde JT, Watson SP, Mundell SJ and Lowe GC (2014) A novel thromboxane A_2receptor N42S variant results in reduced surface expression and platelet dysfunction. *Thromb Haemost* 111(5):923-32 **http://www.ncbi.nlm.nih.gov/pubmed/24452735 (http://www.ncbi.nlm.nih.gov/pubmed/24452735)**

Lordkipanidze M, Lowe G, Kirkby NS, Chan M, Lundberg M, Morgan NV, Bem D, Nisar S, Leo V, Jones LJ, Mundell SJ, Daly ME, Mumford A, Warner TD and Watson SP (2014) Characterization of multiple platelet activation pathways in patients with bleeding as a high-throughput screening option: use of 96-well Optimulassay. *e-Blood* 123:e11-e22 **http://www.ncbi.nlm.nih.gov/pubmed/24408324 (http://www.ncbi.nlm.nih.gov/pubmed/24408324)**

Patel YM, Lowe GC, Lordkipanidzé M, Nisar SP, Garner K, Stockley J, Daly M, Watson SP, Austin SK and Mundell S (2014) A novel mutation in the $P2Y_{12}$ receptor and a function-reducing polymorphism in PAR-1 results in chronic bleeding in a patient. *J Thromb Haemost* 12:716-25 **http://www.ncbi.nlm.nih.gov/pubmed/24612435 (http://www.ncbi.nlm.nih.gov/pubmed/24612435)**

Dovlatova N, Lordkipanidzé M, Lowe GC, Dawood B, May J, Heptinstall S, Watson SP and Fox SC (2014) Evaluation of a whole blood remote platelet function test for the diagnosis of mild bleeding disorders. *J Thromb Haemost* 12(5):660-5 **http://www.ncbi.nlm.nih.gov/pubmed/24618131 (http://www.ncbi.nlm.nih.gov/pubmed/24618131)**

Daly ME, Leo VC, Lowe GC, Watson SP and Morgan NV (2014) What is the role of genetic testing in the investigation of patients with suspected platelet function disorders? *Br J Haematol* 165:193-203 **http://www.ncbi.nlm.nih.gov/pubmed/24479992 (http://www.ncbi.nlm.nih.gov/pubmed/24479992)**

2013

Lowe GC, Sánchez Guiu L, Chapman O, Rivera J, Lordkipanidzé M, Dovlatova N, Wilde J, Watson SP and Morgan NV (2013) The use of microsatellite markers provides a rapid approach for autozygosity mapping in Hermansky-Pudlak syndrome: identification of the second HPS7 mutation in a patient presenting late in life. *Thromb Haemost* 109:766-8 **http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3641626/ (http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3641626/)**

Lowe G, Lordkipanidze M and Watson SP (2013) Utility of the ISTH Bleeding Assessment Tool in predicting platelet defects in participants with suspected inherited platelet function disorders. *J Thromb Haemost* 11(9):1663–1668 **http://www.ncbi.nlm.nih.gov/pubmed/23809206 (http://www.ncbi.nlm.nih.gov/pubmed/23809206)**

Stockley J, Morgan NV, Bem D, Lowe GC, Lordkipanidzé M, Dawood B, Simpson MA, Macfarlane K, Horner K, Leo VC, Talks K, Motwani J, Wilde JT, Collins PW, Makris M, Watson SP and Daly ME (2013) Enrichment of FLI1 and RUNX1 mutations in families with excessive bleeding and platelet dense granule secretion defects. ***Blood* 122(25):4090-3 (http://www.bloodjournal.org/content/122/25/4090.long)**

Lordkipanidzé M, Lowe GC and Watson SP (2013) Simultaneous measurement of ATP release and LTA does not potentiate platelet aggregation to epinephrine. ***Thromb Haemost* 110(1):199-201 (http://th.schattauer.de/en/contents/archive/issue/1763/manuscript/19670.html)**

Mumford AD, Nisar S, Darnige L, Jones ML, Bachelot-Loza C, Gandrille S, Zinzindohoue F, Fischer AM, Mundell SJ and Gaussem P (2013) Platelet dysfunction associated with the novel Trp29Cys thromboxane A receptor variant. ***J Thromb Haemost* 11(3):547-54 (http://onlinelibrary.wiley.com/doi/10.1111/jth.12117/abstract)**

Watson SP, Lowe GC, Lordkipanidzé M, Morgan NV(2013) Genotyping and phenotyping of platelet function disorders. ***J Thromb Haemost* 11(1):351-63 (http://onlinelibrary.wiley.com/doi/10.1111/jth.12199/abstract)**

2012

Dawood BB, Lowe GC, Lordkipanidzé M, Bem D, Daly ME, Makris M, Mumford A, Wilde JT and Watson SP (2012) Evaluation of participants with suspected heritable platelet function disorders including recommendation and validation of a streamlined agonist panel. *Blood* 120(25):5041-9 **Download document [pdf] (/Documents/college-mds/research/plategappubs/Blood-paper-2012.pdf)**

Jones ML, Murden SL, Bem D, Mundell SJ, Gissen P, Daly ME, Watson SP and Mumford AD(2012) Rapid genetic diagnosis of heritable platelet function disorders using next generation sequencing: proof-of-principle with Hermansky-Pudlak syndrome. *J Thromb Haemost* 10:306-9 **Download document [pdf] (/Documents/college-mds/research/plategappubs/jonesml2012.pdf)**

Pre 2012

Nisar S, Daly ME, Federici AB, Artoni A, Mumford A, Watson SP and Mundell S (2011) An intact PDZ-motif is essential for correct P2Y12 purinoceptor traffic in human platelets. *Blood* 118:5641-51 **Download document [pdf] (/Documents/college-mds/research/plategappubs/nisars2011.pdf)**

Nash C, Severin S, Makris M, Mumford A, Wilde J, Senis Y and Watson SP (2010) Src kinases are essential for primary aggregation by Gi-coupled receptors. *J Thromb Haemost* 8:2294-304 **Download document [pdf] (/Documents/college-mds/research/plategappubs/nash-c-2010.pdf)**

Mumford AD, Dawood BB, Daly ME, Murden SL, Williams MD, Protty MB, Spalton JC, Wheatley M, Mundell SJ and Watson SP (2010) A novel thromboxane A2

receptor D304N variant that abrogates ligand binding in a patient with a bleeding diathesis. *Blood* 115(2):363-9 **Download document [pdf] (/Documents/college-mds/research/plategappubs/mumford2010.pdf)**

Watson S, Daly M, Dawood B, Gissen P, Makris M, Mundell S, Wilde J and Mumford A (2010) Phenotypic approaches to gene mapping in platelet function disorders - identification of new variant of P2Y12, TxA2 and GPVI receptors. *Hamostaseologie* 30(1):29-38 Review **Download document [pdf] (/Documents/college-mds/research/plategappubs/watsons2010.pdf)**

Daly ME, Dawood BB, Lester WA, Peake IR, Rodeghiero F, Goodeve AC, Makris M, Wilde JT, Mumford AD, Watson SP and Mundell SJ (2009) Identification and characterisation of a novel P2Y12 variant in a patient diagnosed with type 1 von Willebrand disease in the European MCMDM-1VWD study. *Blood* 113:4110-3 **Download document [pdf] (/Documents/college-mds/research/plategappubs/dalyme2009.pdf)**

Dawood BB, Wilde J and Watson SP (2007) Reference curves for aggregation and ATP secretion to aid diagnose of platelet-based bleeding disorders: effect of inhibition of ADP and thromboxane A(2) pathways. *Platelets* 18(5):329-45 **Download document [pdf] (/Documents/college-mds/research/plategappubs/dawoodbb2007.pdf)**

Morgan NV, Pasha S, Johnson CA, Ainsh JR, Eady RAJ, Dawood BB, McKeown C, Trembath RC, Wilde J, Watson SP and Maher ER (2006) A germline mutation in BLOC1S3/reduced pigmentation causes a novel variant of Hermansky-Pudlak syndrome (HPS8). *Am J Human Gen* 78:160-6 **Download document [pdf] (/Documents/college-mds/research/plategappubs/morgan-nv-2006.pdf)**