APPENDIX 2

MUTATIONAL ANALYSIS FOR PLATELET DISORDERS (CLINICAL TESTING)

N.B.: It is recommended, whenever possible, that test results be communicated to patient(s) in a setting that includes appropriate (genetic) counselling.

GLYCOPROTEIN DEFICIENCIES	
Glanzmann thrombasthenia (genes: *ITGA2B*, *ITGB3*)	• Center for Nephrology and Metabolic Disorders (sequencing)
Bernard-Soulier syndrome (genes: *GP1BA*, *GP1BB*, *GP9)*	• Prevention Genetics* • Blood Center of Wisconsin (sequencing)
STORAGE GRANULE DISORDERS	
Hermansky-Pudlak syndrome (genes: *HPS1-8 – HPS1*, *AP3B1*, *HPS3*, *HPS4*, *HPS5*, *HPS6*, *DTNBP1*, *BLOC1S3*)	• Prevention Genetics* • Casey Eye Institute
Chediak-Higashi syndrome (gene: *LYST*)	• Prevention Genetics* • Casey Eye Institute
FAMILIAL THROMBOCYTOPENIAS	
MYH9-related disease (gene: *MYH9)*	• Prevention Genetics* • Blood Center of Wisconsin (sequencing)
Platelet-type VWD (gene: *GP1BA*)	• Prevention Genetics* • Blood Center of Wisconsin (sequencing)
WAS/XLT (gene: *WAS)*	• Prevention Genetics* • Blood Center of Wisconsin (sequencing)
CAMT (gene: *MPL*)	• Prevention Genetics* • Blood Center of Wisconsin (sequencing)
FPD/AML (gene: *RUNX1*/*CBFA2)*	• Prevention Genetics* • Blood Center of Wisconsin (sequencing)
TAR (1q21.1 Deletion syndrome)	• North Bristol NHS Trust, Bristol Genetics Lab (targeted analysis for common mutation as well as MLPA**)

OTHER	
Dyserythropoietic anemia with thrombocytopenia (gene: *GATA1)*	• Prevention Genetics*
Ehlers-Danlos syndrome (genes: *COL5A1/2, COL3A1, COL1A1/2 PLOD1)*	• Connective Tissue Gene Tests (sequencing and deletion/duplication analysis via high density targeted array)
Noonan syndrome (genes: *PTPN11, SOS1, RAF1, KRAS, NRAS, SHOC2, BRAF, MEK1, MEK2, HRAS, CBL, SPRED1)*	• Harvard Partners Center for Genetics and Genomics (sequencing)
Gray platelet syndrome (gene: *NBEAL2*)	• Prevention Genetics*

***For all tests listed, both sequence analysis and deletion/duplication analysis by Array CGH is available upon request.**
****Multiplex Ligation-dependent Probe Amplification (deletion analysis)**

- **The following is a list of laboratory websites for updated information and required laboratory forms:**
  - **Blood Center of Wisconsin, Milwaukee, WI, USA: http://www.bcw.edu/bcw**
  - **Casey Eye Institute, Portland, OR, USA: http://www.ohsu.edu/xd/health/services/casey-eye/diagnostic-services/cei-diagnostics/index.cfm**
  - **Center for Nephrology and Metabolic Disorders, Weisswasser, Germany: http://www.moldiag.com/**
  - **Connect ive Tissue Gene Tests, Allentown, PA, USA: http://www.ctgt.net/**
  - **Harvard Partners, Boston, MA, USA: http://pcpgm.partners.org/**
  - **Prevention Genetics, Marshfield, WI, USA: http://www.preventiongenetics.com/**
  - **North Bristol, NHS Trust, Bristol Genetics Laboratory: http://www.nbt.nhs.uk/clinicians/services-referral/bristol-genetics-laboratory/genetics-a-z-services and http://www.nbt.nhs.uk/sites/default/files/filedepot/incoming/Thrombocytopenia-Absent_Radius_%28TAR%29_Syndrome_Service_at_BGL.pdf**