

2. **Survey of patients with rare disorders in the UK.** We have agreed that each disorder will be surveyed separately and sequentially because the work involved will be too great for a single project. The first disorder to be examined will be FXIII deficiency. The number of patients on the UK register is about 50 and we hope to recruit any available families via haemophilia centre directors (who will be written to individually). The project is expected to take 2 years, examining in detail the clinical features including bleeding symptoms in heterozygotes, and where not already done, the molecular genetics. There is a need to validate the currently available assay methods which are only performed in a small number of laboratories, and the assays for this study will be performed centrally in Sheffield by Steve Kitchen. A grant application is underway and the project will be suitable for a SpR or Clinical Nurse Specialist. The working party is producing a generic bleeding history proforma based on those recently published which will be computerised and web-based with the help of the National Haemophilia Database group. This proforma will be used for all the disorders with minor adjustments as necessary.

3. **Future work.** We plan to work with the NHD to clarify what platelet disorders are registered. There is a large group of 'severe platelet disorders' other than Glanzmann's thrombasthenia and Bernard-Soulier syndrome; further analysis of this group of disorders is desirable initially by changing the database to capture better information.

Publications

'A review of inherited platelet disorders with guidelines for their management on behalf of the UKHCDO'. Paula HB Bolton-Maggs, Elizabeth A Chalmers, Peter L Collins, Paul Harrison, Steve Kitchen, Ri J Liesner, Adrian Minford, Andrew D Mumford, Liakat A Parapia, David J Perry, Steve P Watson, Jonathan T Wilde, Michael D Williams.
Accepted for publication in British Journal of Haematology (August 2006).

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