

## Rare Disorders Working Party

### Membership

Andrew Mumford                      Chairman  
Sam Ackroyd  
Louise Bowles  
Pratima Chowdary  
John Grainger  
Jason Mainwaring  
Mary Mathias  
Niamh O'Connell  
Raza Alikhan

The group met face to face in November 2012.

### Remit

The Rare Disorders Working Party has a track record of publication of highly cited management guidelines but has not been active since 2009. A newly convened group was reformed in August 2012 and comprises UK Haemophilia clinicians with personal expertise in diagnosis and management of rare disorders.

### Activities

The main activities of the WP have been to update the obsolete 2004 'rare coagulation disorders' guideline. The revised guideline will have greater focus by omitting Ehlers-Danlos syndrome but will include the fibrinolysis pathway disorders PAI-1 deficiency and antiplasmin deficiency. Descriptions of individual disorders have now been written using standardised sub-headings to improve clarity and brevity and have been assembled into a first draft manuscript for review by WP members at the 2013 annual meeting.

The Rare Disorders Working Party has collaborated with the Paediatric Working Party to design a UK survey of intracranial haemorrhage in haemophilia and rare coagulation and platelet disorders launched in September 2013. The WP has contributed to a UKHCDO consultation on adoption of recombinant FXIII concentrate for prophylaxis in FXIII deficiency and facilitated launch of the EU Prospective registry of FXIII and fibrinogen deficiency at UK centres within the EUPHANET consortium framework.

Dr Andrew Mumford  
Chairman, Rare Disorders Working Party  
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