

WILSON'S DISEASE SPECIAL INTEREST GROUP (SIG): ANNUAL REPORT 2017-18

This annual report covers the period 1 September 2017 to 31 August 2018.

Membership

The BASL Committee appointed an initial SIG Lead for 2017-18, Dr Bill Griffiths, Consultant Hepatologist, Addenbrookes Hospital, Cambridge. An election for the position of SIG lead will be held subsequently.

BASL members were given the opportunity to sign up for the SIG via the members' section of the BASL website. Non-members may also join the group. At the time of writing this report, there were 85 individuals included in the member database.

Meetings

Two meetings of the SIG were held during 2017-18:

- 14 December 2017, London, 30 participants
- 14 June 2018, London, 34 participants.

Key achievements

BASL has enabled the first real coming together of the many specialist areas involved in Wilson's disease across the UK, with a view to improving health outcomes for this rare disorder. The SIG comprises input from a number of disciplines including Public Health, Biochemistry, Neurology, Hepatology and Metabolic Medicine. It includes both paediatric and adult representation and an active contribution from the Wilson's Disease Support Group.

The SIG has covered four main areas and reports back as follows:

- 1) Service delivery – in accordance with the UK Strategy for Rare Disease, a preliminary proposal to NHSE for defined Wilson's disease Specialist Centres has been submitted and accepted for further evaluation by the HPB CRG. A more detailed SIG-endorsed service specification will be presented to the CRG in October. This is in parallel with a policy document currently going through NHSE for the commissioning of trientine.
- 2) National Registry – with the support of the SIG, Public Health England, specifically NCARDRS, are establishing a Rare Disease registry, which will capture relevant clinical data on all Wilson's disease patients in England moving forward.
- 3) Diagnosis and monitoring – the SIG has brought together the clinical and laboratory expertise in monitoring Wilson's disease. A guideline document will be produced for agreed best practice, for ratification at the next meeting.
- 4) Research – the SIG has discussed all research on Wilson's disease occurring in the UK and is facilitating national recruitment, e.g. the UCL/Sheffield neurological cohort study (CROWD). New collaborative links exploring novel DNA/RNA therapy have been established. Pharmaceutical trials are in process and in development.

Further information available at: <https://www.basl.org.uk/index.cfm/content/page/cid/31>.