

RaDaR Rare Disease Group Annual Report

April 2015 – March 2016

Rare Disease Group	aHUS												
Lead Clinician	David Kavanagh												
Summary of RDG meetings	<p>aHUS Rare Diseases Working Group Meeting 27th November 2015</p> <p>Agenda</p> <ol style="list-style-type: none"> 1. Minutes of last meeting (20th October 2014) 2. aHUS database (Amy Osborne) 3. National aHUS service <ol style="list-style-type: none"> a. Adults (David Kavanagh) b. Paediatrics (Sally Johnson) 4. Autoantibody associated aHUS (Vicky Brocklebank) 5. aHUSUK and aHUS Alliance developments (Len Woodward) 6. 100K project and aHUS (Danny Gale) <p>Attendees</p> <table style="width: 100%; border: none;"> <tr> <td>David Kavanagh (Chair)</td> <td>Tim Goodship</td> </tr> <tr> <td>Vicky Brocklebank (Secretary)</td> <td>Sally Johnson</td> </tr> <tr> <td>Kevin Marchbank</td> <td>Len Woodward</td> </tr> <tr> <td>Amy Osborne</td> <td>Danny Gale</td> </tr> <tr> <td>Steve Perkins</td> <td>Claire Harris</td> </tr> <tr> <td>Anna Richards</td> <td>Terry Cook</td> </tr> </table>	David Kavanagh (Chair)	Tim Goodship	Vicky Brocklebank (Secretary)	Sally Johnson	Kevin Marchbank	Len Woodward	Amy Osborne	Danny Gale	Steve Perkins	Claire Harris	Anna Richards	Terry Cook
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Summary of patient events	<p>The Second aHUS Patient & Families' Conference, London, June 2015.</p> <p>Chairs - Emma Woodward & David Kavanagh</p> <p>Programme</p> <p>Complement and aHUS - Dr Kevin Marchbank aHUS penetrance in Families - Prof Tim Goodship The World of aHUS - Len Woodward aHUS -Many Variants Many Triggers - Dr David Kavanagh The Pregnancy Trigger -Dr Edwin Wong The Transplant Trigger - Prof Neil Sheerin The NICE Research Update - Prof Neil Sheerin The Logistics of Research and Registries - Dr Detlef Bockenhauer Funding Research - Sandra Currie</p>												
Grant applications submitted	Nil												
Grants awarded	Nil												

Publications and presentations

- 1 Challis, R.C., Araujo, G.S., Wong, E.K., Anderson, H.E., Awan, A., Dorman, A.M., Waldron, M., Wilson, V., Brocklebank, V., Strain, L., Morgan, B.P., Harris, C.L., Marchbank, K.J., Goodship, T.H. and Kavanagh, D. (2016) 'A De Novo Deletion in the Regulators of Complement Activation Cluster Producing a Hybrid Complement Factor H/Complement Factor H-Related 3 Gene in Atypical Hemolytic Uremic Syndrome', *J Am Soc Nephrol.* 2016 Jun;27(6):1617-24
- 2 Chromosomal rearrangement—A rare cause of *complement factor 1* associated atypical haemolytic uraemic syndrome. Gleeson PJ, Wilson V, Cox TE, Sharma SD, Smith-Jackson K, Strain L, Lappin D, McHale T, Kavanagh D*, Goodship TH*. *Immunobiology* (2016) in press *These authors made equal contributions to this work
- 3 Sheerin N*, Kavanagh D*, Goodship TH*, Johnson S*. A national specialised service in England for atypical haemolytic uraemic syndrome - the first year's experience. *QJM* 2015 109 (1) 27-33 *These authors made equal contributions to this work
- 4 Blaum BS, Hannan JP, Herbert AP, Kavanagh D, Uhrin D, Stehle T. Structural basis for sialic acid-mediated self-recognition by complement factor H. *Nat Chem Biol* 2015;11(1):77-82.
- 5 Phillips, E.H., Westwood, J.P., Brocklebank, V., Wong, E.K.S., Tellez, J.O., Marchbank, K.J., McGuckin, S., Gale, D.P., Connolly, J., Goodship, T.H.J., Kavanagh, D., Scully, M.A. The role of ADAMTS 13 activity and complement mutational analysis in differentiating acute thrombotic microangiopathies *J Thromb Haemost.* 2015. 14(1):175-85.
- 6 Hunt D, Kavanagh D, Drummond I, Bellamy C, Günther C, Overell J, Evans S, Jackson A, Chandran S. Thrombotic Microangiopathy Associated with Interferon- β (2014) *N Eng J Med* 370(13):1270-71.
- 7 Wong, E., Challis, R., Sheerin, N., Johnson, S., Kavanagh, D. and Goodship, T.H. (2015) 'Patient stratification and therapy in atypical haemolytic uraemic syndrome (aHUS)', *Immunobiology.* S0171-2985(15)00070-4
- 8 Nester CM, Barbour T, de Cordoba SR, Dragon-Durey MA, Fremeaux-Bacchi V, Goodship TH, Kavanagh D, Noris M, Pickering M, Sanchez-Corral P, Skerka C, Zipfel P, Smith RJ. Atypical aHUS: State of the art. *Mol Immunol* 2015 ;67(1):31-42.
- 9 Kavanagh D, Smith-Jackson K. Eculizumab in children with hemolytic uremic syndrome. *Kidney Int.* 2016; 89(3):537-8.

Highlights	Successful Second aHUS Patient Families' Conference
Problems	Delayed award of national centre for aHUS