

## RaDaR Rare Disease Group Annual Report

April 2016 – March 2017

Rare Disease Group	aHUS
Lead Clinician	David Kavanagh
Summary of RDG meetings	RDG meeting postponed until National aHUS centre established. Planned June 2017
Summary of patient events	Patient leads input to design of national aHUS centre
Grant applications submitted	2x MRC clinical fellowships
Grants awarded	Nil
Publications and presentations	<p>1: Bruel A, Kavanagh D, Noris M, Delmas Y, Wong EKS, Bresin E, Provôt F, Brocklebank V, Mele C, Remuzzi G, Loirat C, Frémeaux-Bacchi V, Fakhouri F. Hemolytic Uremic Syndrome in Pregnancy and Post-Partum. Clin J Am Soc Nephrol. 2017 Jun 8. pii: CJN.00280117. doi: 10.2215/CJN.00280117. [Epub ahead of print] PubMed PMID: 28596415.</p> <p>2: Goodship TH, Cook HT, Fakhouri F, Fervenza FC, Frémeaux-Bacchi V, Kavanagh D, Nester CM, Noris M, Pickering MC, Rodríguez de Córdoba S, Roumenina LT, Sethi S, Smith RJ; Conference Participants. Atypical hemolytic uremic syndrome and C3 glomerulopathy: conclusions from a "Kidney Disease: Improving Global Outcomes"(KDIGO) Controversies Conference. Kidney Int. 2017 Mar;91(3):539-551. doi:10.1016/j.kint.2016.10.005. Epub 2016 Dec 16. PubMed PMID: 27989322.</p> <p>3: Challis RC, Ring T, Xu Y, Wong EK, Flossmann O, Roberts IS, Ahmed S, Wetherall M, Salkus G, Brocklebank V, Fester J, Strain L, Wilson V, Wood KM, Marchbank KJ, Santibanez-Koref M, Goodship TH,</p>

	<p>Kavanagh D. Thrombotic Microangiopathy in Inverted Formin 2-Mediated Renal Disease. J Am Soc Nephrol. 2017 Apr;28(4):1084-1091. doi: 10.1681/ASN.2015101189. Epub 2016 Dec 14. PubMed PMID: 27974406; PubMed Central PMCID: PMC 5373440.</p> <p>4: Kavanagh D, McGlasson S, Jury A, Williams J, Scolding N, Bellamy C, Gunther C, Ritchie D, Gale DP, Kanwar YS, Challis R, Buist H, Overell J, Weller B, Flossmann O, Blunden M, Meyer EP, Krucker T, Evans SJ, Campbell IL, Jackson AP, Chandran S, Hunt DP. Type I interferon causes thrombotic microangiopathy by a dose-dependent toxic effect on the microvasculature. Blood. 2016 Dec 15;128(24):2824-2833. Epub 2016 Sep 23. PubMed PMID: 27663672; PubMed Central PMCID: PMC 5159705.</p>
<p><b>Highlights</b></p>	<p>NHS England awarded a contract to run a National Specialist Service for aHUS to The National Renal Complement Therapeutics Service Newcastle</p>
<p><b>Problems</b></p>	<p>Delayed aHUS national centre - now awarded</p>