

Rare Diseases Operational Management Board, January 2015

RDG End of Year Report 2014

Dear RDG Coordinator,

Please file a report to the OMB for your activity in the calendar year 2014, and return it to Chrissie Jacobs at UKRR. Ongoing recognition of the RDG by the Renal Association is dependent on receipt of a report.

This report is deliberately minimal and should not take long to complete, not least because some of the information is obtainable centrally by the OMB. Emphasis is placed on the early benefits for patients, which focuses on patient support meetings and patient engagement within the RDG.

Please fill out the table below and return the entire word document with any supplements that you wish to be filed, specifically minutes of RDG or patient information meetings.

Name of RDG	ARPKD Rare Disease Working Group
Name of Coordinator	Dr Larissa Kerecuk
Name of Administrator Email address Telephone number	Melanie Dillon
Name of person/person's cleared to work with patient identification Email address Telephone number	Melanie Dillon
RDG meetings: Summary <i>(append written minutes if that is easier)</i>	Minutes of RDG meetings from June and December 2014 are attached.
Patient Information meetings: Summary <i>(append written minutes if that is easier)</i>	The 3 rd ARPKD Patient Information Day was held in London Saturday 28 th June 2014. Presentations are available online .
Grant applications submitted	None
Grants awarded	None

Publications and presentations	<p>Guay-Woodford, L.M., Bissler J.J., Braun, M.C, Bockenbauer, D., Cadnapaphornchai, M.A., Dell, K.M., Kerecuk, L., Liebau, M.C., Alonso-Pecllet, M.H., Shneider, B., Emre, S., Heller, T., Kamath, B.M., Murray, K.F., Moise, K., Eichenwald, E.E., Evans, J., Keller, R.L., Wilkins-Haug, L., Bergmann, C., Gunay-Aygun, M., Hooper, S.R., Hardy, K.K., Hartung, E.A., Streisand, R., Perrone, R., Moxey-Mims, M. (2014) Consensus Expert Recommendations for the Diagnosis and Management of Autosomal Recessive Polycystic Kidney Disease: Report of an International Conference. Journal of Pediatrics. Sep;165(3):611-7</p>
Highlights (free text)	<p>The third ARPKD Patient Information Day was held at the Institute for Child Health at Great Ormond Street Hospital, London on Saturday 28th June 2014.</p> <p>The day included talks from expert doctors, nurses and scientists about ARPKD as well as opportunities for families to share their own experiences. Workshops were held on a number of topics including benefits and coping skills.</p> <p>The 2015 Patient Information Day will be held in Leeds in July.</p> <p>International Consensus Guidelines for the diagnosis and management of ARPKD have now been published and are available online.</p>
Problems (free text)	<p>There are on-going delays with RaDaR and concerns about its future financial security. Granular data fields are still not available, limiting data entry.</p> <p>Regular updates as to how many patients have been recruited UK wide would be beneficial to help plan research strategies.</p>