## Awarded Grants 2018

## The following grants have been awarded in 2018

- Final common pathways in complement and non-complement dependent aHUS Dr P Walsh.

- Novel antibodies against complement factor B for diagnostics and functional assays.
- Professor C Harris.

- Understanding aHUS using in silica, ex vivo and in vivo modelling.
- Dr K Marchbank.

- Gene editing for cystic kidney disease. Professor J Sayer.

- The STOK (Self Testing Own Kidneys) Study - Dr Jonathan Murray

This study will assess whether a group of fifteen selected participants are able to use hand-held devices to safely and accurately self-test their own kidney blood and urine tests, in their own homes.

Each participant will self-measure these tests at home once per week, for a total of four weeks. On the same days that participants perform such tests at home, they will also attend the hospital for blood and urine samples to be tested conventionally by NHS staff and laboratory analysis. The study team will compare results of participant self-tests at home with results of tests performed conventionally by NHS staff in hospital, to determine if participant self-test results are accurate and reliable when compared to NHS standard testing.

The study will also examine how participants found the experience of using hand held devices at home to test their own kidney function. Such participant feedback aims to help determine how self-testing kidney function at home may benefit selected patients in real life. Potential study participants, including the study patient advisor, indicate how inconvenient it can be to attend medical facilities for routine kidney tests, when they feel well and have no other need to see a healthcare professional. Their experiences of having to plan activities and employment around such clinical visits, together with costs associated with travel and car parking, are often negative.

Finally, the study will explore how patient self-testing may benefit NHS expenditure and capacity in busy clinical areas.

If this study concludes participant self-testing of blood and urine at home is feasible, safe and accurate, associated with positive participant experiences and cost-effective, then development and application of such procedures could potentially benefit patients and NHS systems in a wide variety of clinical settings.

- Mechanisms and Treatment of a Novel Ciliopathy Syndrome - Prof John Sayer

Joubert Syndrome (JS) is an incurable multisystem inherited condition and often leads to childhood renal failure. More than 30 genes have been associated with JS. We have recently identified in a family from our renal genetics clinic a novel genetic cause of JS.

The purpose of this investigation is to (1) undertake a detailed characterisation of this novel genetic cause of JS, to reveal the underlying disease mechanisms and identify potential therapeutic targets, (2) develop and validate an integrated and personalised platform to model JS and renal ciliopathies in general.

To achieve these goals, we plan to:

1. generate a model by surgically editing genome with CRISPR/Cas9 system, to recapitulate the exact mutation found in human patients
2. derive kidney organoids from patient-derived induced pluripotent stem cells

This generous award will allow us to contribute to the advancement of our understanding of JS, whilst also providing an important evaluation of different disease models and the potential of stem cell technologies that could serve as a paradigm for many other aspects of kidney disease research.