

Working together to find answers: Nephrotic Syndrome Rare Diseases Clinical Research Consortium (NEPTUNE)

Minimal change disease (MCD) and focal segmental glomerulosclerosis (FSGS) generate an enormous individual and societal financial burden, accounting for approximately 12% of prevalent end stage renal disease (ESRD) cases (2005) at an annual cost in the US of more than \$3 billion. However, the clinical classification of these diseases is widely believed to be inadequate by the scientific community and we still do not have a comprehensive molecular understanding. Given our poor understanding of MCD and FSGS biology, it is not surprising that the available therapies are imperfect. The therapies lack a clear biological basis, and as many families have experienced, they are often not beneficial, and in fact may be significantly toxic. Given these observations, it is essential that research be conducted that address these serious obstacles to effectively caring for patients.

In response to a request for applications by the National Institutes of Health, Office of Rare Diseases (NIH, ORD) for the creation of Rare Disease Clinical Research Consortia, the University of Michigan in association with a number of affiliated universities joined together with The NephCure Foundation in collaboration towards the establishment of a Nephrotic Syndrome (NS) Rare Diseases Clinical Research Consortium. A grant application was submitted for a multidisciplinary research and educational platform, to bring together clinical and translational scientists, and increase our knowledge about Glomerular diseases such as FSGS, and MCD. Through the efforts of Drs. Larry Holzman and Matthias Kretzler and the NephCure constituency, many of whom contacted congressional leadership for support; in September we received the stunning news that NEPTUNE was awarded a \$6.25million grant from the Office of Rare Disease. This combined with a \$2 million commitment from NephCure and a \$2 million in-kind commitment from the University of Michigan combines for a \$10 million five year study.

The importance of this program lies in the fact that it allows several major barriers in Glomerular research to be overcome to effectively conduct research and interventional studies. Most importantly this provides the infrastructure for continued collaboration of researchers in this field which will advance the care of MCD and FSGS patients internationally. The specific aims of the Consortium are as follows:

1. Establish an infrastructure to efficiently conduct clinical and translational research in nephrotic syndrome (NS).



2. Identify and characterize new biomarkers (used for the diagnosis of Glomerular diseases) and/or potential therapeutic targets for NS.
3. Conduct two clinical studies in NS.
4. Develop and implement a postdoctoral training program that is suitable and appropriate for training of individuals with MD/PhD to become skilled in conducting research in rare kidney disease.
5. Develop multimedia lay and physician educational resources on NS in collaboration with Nephcure (<http://www.nephcure.org>), and Halpin (<http://www.halpinfoundation.org>)
6. Develop and maintain a high quality, secured and user friendly repository of clinical data and biospecimens (a biobank) of NS patients for sharing among researchers nationally and worldwide through the web thus further stimulating research into these diseases internationally.

