Update on Rare Diseases
From Christoph Wanner, ERA-EDTA Registry Chairman

In May 2009 we reported in our Newsletter that the Registry will aim to focus on rare diseases that are going to be dissected from the large database. We now provide the full spectrum of diseases that are documented as leading causes for dialysis initiation throughout Europe (see figure). To move in that direction appeared to be timely since WGIKD - Working Group on Inherited Kidney Disorders has been created and officially recognised as ERA-EDTA Working Group. This working group takes into account the wide diversity of the inherited kidney disorders area, including the adult and paediatric nephrology, and the clinical, research, genetics, physiology and pathophysiology aspects.

Newsleter 16, June 2010 reported that research has successfully initiated with Oliver Gross and his team from Göttingen, Germany in respect to a “European Alport Registry joint analysis”. Substantial analyses are going to be published this year. Another project in the environment of rare diseases was started with Jerome Harambat from France on “Treatment strategies and outcomes of paediatric renal transplantation in Europe”.

Dr. Harambat has moved to Amsterdam on a research grant to explore the database. In this respect, the most successful collaboration is currently being done with the European Society for Paediatric Nephrology (ESPN)/ERA-EDTA Registry. Ongoing exploration of the database is with cystinosis, oxalosis (i.e. primary hyperoxaluria type 1) and autosomal recessive polycystic kidney disease. We are optimistic that our spectrum of research and data analysis from rare diseases to large cohort studies (focussed data collection) will further attract young researchers and be successful in the future.
Renal prognosis in cystinosis patients – Results from the ESPN/ERA-EDTA Registry
From Karlijn van Stralen and Elena Levetchenko (Belgium)

Cystinosis is a rare autosomal recessive disorder of lysosomal cystine transporter cystinosin encoded by the CTNS gene (17p13). The disease affects one to two per 100,000 newborns with higher incidence in some North-European regions such as Nord Pas de Calais and Bretagne in France. Cystinosis has been subdivided in an infantile form, also termed nephropathic infantile cystinosis (NC), a juvenile form that is characterized by less severe renal disease, and a late-onset form with primarily ocular symptoms. During early childhood, kidneys represent the primarily affected organs in NC and nearly all patients with NC develop renal Fanconi syndrome during the first year of life. Without treatment, most children progress to end-stage renal disease before the age of 10 years. Cystine-depleting drug cysteamine was first introduced as a potential treatment for NC in 1976, but was only used on a broader scale at the end of 1980s. Few excellent single-centre trials demonstrated improved renal function survival in cystinosis patients adequately treated with cysteamine, however, broad-scale outcome data are lacking. The ESPN/ERA-EDTA registry has collected, in conjunction with the ERA-EDTA registry, data from 31 European countries on paediatric patients entering renal replacement therapy. During the ERA-EDTA conference, we will show long-term outcome of cystinosis patients between 1979 and 2008, a period that encompasses the introduction of treatment with cysteamine, and compare them to a cohort of patients entering renal replacement therapy for other renal diseases. During the ERA-EDTA Registry symposium (Saturday June 25, 8.00-9.30 a.m.) results from this study will be presented.

The DIAlysis and Pregnanacies: a European Registry (DIAPER) study
From Marlies Noordzij, Graham Lipkin (UK), and Enrico Imbasciati (Italy)

Women with chronic kidney disease often have amenorrhea but may still occasionally ovulate and thus conceive. Although unusual, pregnancy even occurs in women treated with dialysis. It is widely stated and believed that the frequency of pregnancy in women of childbearing age (14–44 years) on dialysis is increasing, possibly due to improved and intensified dialysis regimens. Also the survival of infants born to women treated with dialysis seems to be increasing. However, these developments are not well documented.

At the ERA-EDTA Registry we are currently performing a systematic review of the literature on this topic in the last 30 years (1980-2010) to get a complete overview of what has already been published on this topic. This systematic review includes 35 case series and surveys comprising a total of 451 pregnancies in 430 women treated with chronic haemodialysis or peritoneal dialysis. The number of pregnancies reported in the international literature over the last 3 decades is limited, because pregnancies in women treated with dialysis are difficult to study. This is because they remain uncommon and occur in widely separated dialysis centers. For the same reason, most nephrologists will encounter only one or two pregnant dialysis patients during their time in practice. As a result, most of the published reports on pregnancies in women with end-stage renal disease are case reports or small case series. There some reports of larger groups of women treated with dialysis by the time of conception from the United States (registry of 184 cases) and Brazil (52 cases from one center). However, there are, to our knowledge, no European reports on larger groups of cases available.

Therefore, we initiated the “DIAlysis and Pregnancies: a European Registry (DIAPER) study”. With this study we aim to estimate the incidence of pregnancies in chronic dialysis patients between 2005 and 2010 in different European countries, by means of data collection using an electronic survey. In addition, we aim to assess which treatment regimens are used for pregnant dialysis patients and which outcomes of pregnancy are most common in mother and child. The results of this study may be used to optimize fetal and maternal outcomes.

The data collection for the DIAPER study is expected to start in the autumn of 2011 and we hope that many of you will participate so that we can get a complete overview of Europe. For more information please contact Marlies Noordzij at m.noordzij@amc.uva.nl.


ERA-EDTA Registry Activities during the ERA-EDTA Congress
Prague, June 23-26, 2011

• Thursday, June 23, 10.00 - 12.00 Meeting for national and regional Registries
• Saturday, June 25, 08.00 - 09.30 ERA-EDTA Registry Symposium Programme:
  1) Introduction on the Registry 2011 Christoph Wanner, Würzburg, Germany
  2) Is survival after transplantation associated with pre-transplant dialysis modalities?
    Anneke Kramers, Amsterdam, Netherlands
  3) In RRT patients non-cardiovascular mortality is as important as cardiovascular mortality
    Judith Vogelzang, Amsterdam, Netherlands
  4) Reasons to start dialysis
    Kitty Jager, Amsterdam, Netherlands
  5) Competing risk models: registry case studies
    Marlies Noordzij, Amsterdam, Netherlands
  6) Long-term outcome in cystinosis: data from ESPN/ERA-EDTA Registry
    Elena Levetchenko, Leuwen, Belgium

Registry publications
In the first half of 2011, almost 20 papers with Registry authorship have been published. A complete list of these publications can be found on our website: www.era-edta-reg.org.