

ESPN Research Grant Annual Report 2014/2015

European CAKUT Registry and biobanking

Background

Congenital abnormalities of the kidney and urinary tract (CAKUT) account for the most frequent congenital organ malformations that cause chronic kidney disease, leading to excessive morbidity and mortality in humans. While many CAKUT cases are sporadic, familial clustering is common, suggesting that the pathogenesis is influenced by genetic factors. Variable CAKUT phenotypes in different family members with the same gene mutation ranging from asymptomatic structural abnormalities to severe end-stage renal disease (ESRD), have been reported. The European Society for Paediatric Nephrology (ESPN) Working Group CAKUT/Urinary Tract Infection (UTI)/Bladder Dysfunction aims at establishing a web-based registry of European cases of familial CAKUT with familial CAKUT being defined as more than one sibling or one generation affected in the family and/or consanguinity within the family.



EURECA – European Registry for Familial CAKUT Cases

a) The EURECA Registry (European Registry for Familial CAKUT Cases) has been implemented as a web-based registry open to all members of the society (<http://www.nefrokid.it/EURECA/v0/>). This registry involves the most recent web technologies (AngularJS framework, responsive design, REST API pattern) and will be accessible by mobile devices. We expect to generate a comprehensive collection of familial CAKUT cases throughout Europe by the means of this Registry initiative. As CAKUT (in its overall presentation) is rather frequent we believe that the rate of success of identification of new causative genes will be much higher when restricting the study patients to familial cases. A multinational approach of experts in the field will further strengthen the outcome of the study. The web application will integrate a pedigree draw in order to better understand the genetic implications of the disease. The registry will be supported by DNA collections and biobanking. The approval by the ethics committee of the University Duisburg-Essen is awaited. Informed consent and Information material has been created in English language. A translation in different important languages is planned after approval of the local ethics committee.

b) Financial Report

1. A defined amount of the ESPN Research Grant (520,42 €) has been spent on the **EURECA Kick-off-Meeting** in Bologna on Feb 6th, 2015 at the Ospedale Sant'Orsola including travel and accommodation of S. Weber. Participant of the Meeting were G. Montini (Bologna, now Milano, Italy), N. Crisafi (Torino, Italy) and S. Weber (Essen, Germany). Technical and financial issues of the EURECA Registry were discussed and afterwards decided on by Skype conferences.

2. Approximately 5.000 € will be spent on the establishment of the web-interface, including the internet domain and fees for external software. Recipient of the fee: Ninni Crisafi, Software Engineering

3. Remaining amount (on Aug 22nd, 2015): 14.479,58 €

Stefanie Weber