Dear Patients, Sponsors and Supporters of the NEOCYST Research Consortium,

A lot has happened since our last update. The work of the consortium has contributed significantly to advance knowledge and scientific progress in the field of cystic kidney diseases. During the second funding period (2020–2023), 39 publications were published in high-impact journals. Continuing this successful path, we are delighted that the Federal Ministry of Education and Research (BMBF) is supporting a third funding period (2023-2026).

The NEOCYST registry continues to grow: currently, clinical data on over 650 patients with cystic kidney diseases is being documented. Over 5000 biological patient samples have been collected and made available for research projects.

Over the past year, many NEOCYST projects have focused on developing risk profiles for various disease entities. The Münster team led by Dr. König identified and published differences in kidney survival among nephronophthisis patients. In an international cooperative approach from NEOCYST with the European Reference Network (ERKNet), Prof. Joost Schanstra from Lyon was able to identify differences in kidney survival in the HNF1B cohort depending on the type and location of the genetic defect. In collaboration with the Polish working group led by Prof. Marcin Zaniew, an online tool was developed to predict the likelihood of a positive genetic test result for HNF1B: www.hnf1bproject.com. A NEOCYST internal project focused on a systematic investigation of the urine biomarker DKK3. The group was able to establish uDKK3 as a biomarker to assess disease severity and estimate short-term kidney function decline in the setting of renal ciliopathies. The working group led by Prof. Max Liebau has made significant contributions to understanding autosomal recessive polycystic kidney disease (ARPKD). Based on clinical data, a prediction tool has been developed to estimate the likelihood of kidney failure in early childhood.

Another highlight was the production of videos with information on Bardet-Biedl Syndrome and HNF1B nephropathy by the working group led by Dr. Cetiner. In these videos, affected families share their experiences, and experts provide information on the rare diseases in

patient friendly language. The videos are available on our homepage (www.neocyst.de) and can be accessed on the NEOCYST YouTube channel (https://www.youtube.com/@NEOCYST).

One of the biggest and most exciting developments was the establishment of the European TheRaCil Consortium (TheRaCil: Therapies for Renal Ciliopathies; www.theracil.eu). This research initiative brings together European experts in ciliopathies from various disciplines to pool knowledge and data to develop therapies for hereditary cystic kidney diseases in childhood. The TheRaCil Consortium is supported by the European Union for four years with nearly 8 million euros and was selected as one of 9 successful proposals out of over 200 competing initiatives. These are truly great news for the field of rare cystic kidney disease research. NEOCYST partners actively contribute to various working groups within this consortium. Promising drug candidates have been tested successfully in various model organisms and are now being developed further in collaboration with the pharmaceutical industry. Another central task involves the development of patient-centered therapy goals (so-called patient-reported outcome measures) that prioritize the perspectives and quality-of-life concerns of affected patients and their families in addition to medical objectives.

Our heartfelt appreciation goes to all partners who are dedicated to advancing research on renal ciliopathies in childhood. We look forward to continuing to work together to find solutions for rare cystic kidney diseases.

Best wishes,

Your NEOCYST Team

NEOCYST
Network for Early Onset Cystic Kidney Diseases