Personal statement

I have always found Congenital Disorders of Glycosylation (CDG) the most intriguing metabolic disease. For fifteen years, since I finished my fellowship in genetics and inborn errors of metabolism, I have been involved in the diagnosis and care for CDG patients. CDGs have an extremely variable clinical presentation; abnormal glycosylation can affect any organ or organ system, and can mimic many other disorders. The diagnosis of CDG is challenging, even for a metabolic specialist.

Working since 2002 in the umcRadboud as a metabolic pediatrician in Nijmegen, NL, I established NijmegenCDG; a center with the goal of improving quality of life for patients with CDG. Parallel with developing better diagnostics and care for CDG patients I also established a clinical Center of Excellence. CDG research has always stemmed from my clinical work. With our team in Nijmegen, and international collaboration, we discovered twelve different novel types of CDG (ATP6V0A2-CDG, DPM3-CDG, SRD5A3-CDG, DK1-CDG, DPM2-CDG, MAN1B1-CDG, SLC35A1-CDG; ATP6AP1-CDG, CCDC115-CDG, TMEM119-CDG, ATP6V1A-CDG; ATP6V1E1-CDG). We established a validated progression assessment score and developed international treatment guidelines in Dutch and in English. We published several studies on the clinical outcome of different CDGs. During the Nijmegen period our team was awarded with more then 300 000 euros. In those 10 years I mentored five graduate students and several master students in the topics of glycosylation.

From my early involvement in the collaborative European grant focusing on diagnostics, studying the pathomechanism and diagnosing novel types of CDGs (EUROGLYCAN and EUROGLYCA*NET*), up till today, in 2017 (EURO-CDG2) I work together with a excellent international team of glycobiologist, biochemists and physicians, all devoted to CDG, and building knowledge to improve the care for CDG patients.

My major personal goal has been always focussing on the translational aspects of glycosylation research, and treatment development. When we defined PGM1-CDG, a metabolic disease, affecting both glycosylation and glycogen metabolism and potentially treatable with galactose supplements, this unique disorder became the basis of my current research. My research goal in the past 4 years, working at Tulane University Medical School, shifted significantly from diagnostics to therapy. Understanding how galactose affects and regulates glycosylation offers new avenues for therapy not just in PGM1 deficiency, but in many other CDGs. This was the start of establishing clinical trials in CDG.

In the last years I continued giving invited lectures and education on CDG (Mayo Clinics, Rochester, SERGG, Florida, Barcelona, Porto, Sanford Burnham Institute, La Jolla, Australian Society of Human Genetics, Tanzania, etc).

One of my proudest moment was when during the 2nd World CDG Conference I was awarded with the “CDG Hope and Dream Award”.

Since 2015 I became member of the board for CDG-CARE, which is the international CDG association, with board members from the US and associates from Europe. This makes it possible to keep contact with collegues working with CDG patients, and the patients as well.

My most motivating challenges in my professional life are that in 2016 I became Editor in Chief of the Journal of Inherited Metabolic Disease, our professional scientific journal, and that just this year I became vice-coordinator of the MetabERN, the European Reference Network for metabolic disorders for 68 academic centers from more then 20 countries.

I also became member of the SSIEM Council for the Society for Study of Inborn Errors of Metabolism (SSIEM). These positions allow me to collaborate within a large network, gain information, share my knowledge and disseminate my research at the broadest level.