short CV Dr. Albertien M. van Eerde, MD, PhD, clinical geneticist

Date of birth	02-03-1978	
Nationality	Dutch	
Education	2017-2018	UMC Utrecht Steyn Parve Programme (leadership programme for female researchers aspiring a professorship)
	2016	UMC Utrecht Research Talent Programme
	2011-2013 (March)	Last phase of Clinical Genetics specialty training, UMC Utrecht with subspecialisation in nephro-/uro-genetics
	2004-2011	specialty training Clinical Genetics, UMC Utrecht, in combination with PhD project
	2003-2004	physician-researcher Clinical Genetics UMC Utrecht / Genetics research section UMC Utrecht
	2001-2003	medical school UMC Utrecht: clinical internships (with elective internship in clinical genetics at the Erasmus MC Rotterdam)
	2000-2001	visiting student-investigator: UC San Francisco, Dr. Laura Bull's laboratory: Genetics of Aagenaes syndrome
	2000	IFMSA summer school Lund
	1996-2000	medical school UMC Utrecht
	1990-1996	Leiden Preparatory High School (Stedelijk Gymnasium): cum laude
PhD	Thesis	Genetics of Congenital Anomalies of the Kidney and Urinary Tract Towards Elucidation of Genetic Factors in the Etiology of Vesico-Ureteral Reflux
	University	Utrecht University
	Supervisor	Prof.dr. V.V.A.M. Knoers (promotor) Prof.dr. C. Wijmenga (promotor) Dr. J.C. Giltay (co-promotor) Prof.dr. T.P.V.M. de Jong (co-promotor)
	Date of graduation	June 8th 2011
MD (Medical Doctor degree)	Subject	n/a
	University	Utrecht University
	Date of graduation	February 25th, 2003
	Subject	Clinical Genetics

Medical specialism	Planned registration date	Registered: 13-3-2013 Re-registered: 13-3-2018 (for the standard 5 year period)
	Training abroad	2000-2001: visiting student-investigator: UC San Francisco, Dr. Laura Bull's laboratory: Genetics of Aagenaes syndrome
Current position	2013 onwards Clinical geneticist at the Department of Genetics, UMC Utrecht	

Research Grants

"The RENome and beyond"
Dutch Kidney Foundation, PhD Student Grant (15OP14): €260.000

"CKD – Y: Improving genetic diagnostics in Young CKD/ESRD patients
Dutch Kidney Foundation, Kolff Postdoc Startup Grant (KSTP12-010): €150.000
NutsOhra Fund (1303-70): €90.000

Summary of Previous Research

"To improve the outcome on all levels for patients (and their family members) with severe renal disease of yet unknown and/or potentially hereditary etiology, by providing them with optimal genetic diagnostics and counseling. Actuating intelligently designed, patient-specific, tailored counseling and therapy is the goal."

Since my doctoral research with Prof. Knoers on genetics of congenital anomalies of the kidney and urinary tract (2011), I have been determined to bridge the gap between bench and bedside nephrogenetics research. Up to date, my publication record comprises publications in high impact journals such as Nature Reviews Nephrology, JASN, Kidney International and the American Journal of Human Genetics. I was awarded various grants, including a Dutch Kidney Foundation Kolff stipend (€150k) and follow-up grant (€260k) and a grant from the NutsOhra Fund (€90k). I was the first to show that a significant number (~15%) of young adult end stage renal disease patients has a known monogenic disease. Half of these had a wrong or no clinical diagnosis. This was the first nephrogenetic study in a cohort of renal disease patients regardless of their (previous) clinical diagnosis. This insight, and the potential impact of a genetic diagnosis, is what is at the core of most of my activities. My research focusses on optimizing diagnostic strategies in kidney disease by applying amongst others, gene panel-based genetic testing. For this work, I was awarded the best abstract prize at ERA-EDTA (European Nephrology Association) 2015, and won the WCN (International Society of Nephrology) 2015 Young Nephrologist award. In 2017 I was honoured to win the ERA-EDTA Stanley Shaldon award for Young Investigators (10.000). Subsequent abstracts by my PhD student, were selected for oral presentations at both the European and American Societies for Nephrology annual meetings, and have won a best abstract prize at the European Society and NFN Autumn meeting. With this work, I established my own line: Translational Nephrogenetics; Causes of End Stage Renal Disease.

Moving forward, I am currently expanding on the collaboration with the UMCG Transplantlines group and the IgeneTRAIN consortium. This consortium consists of a variety of cohorts with patients with end stage renal disease.

My research is not only be cohort based, but with my unique position as chair of the Expert Center and the multidisciplinary outpatient clinic, I can perform extended diagnostics in unique cases (pediatric and adult), resulting in the discovery of new genes or mechanisms. Currently I have several of these projects going on.

Research Output

https://www.ncbi.nlm.nih.gov/pubmed/?term=van+eerde+am

Other Activities

Since I became a certified clinical geneticist, I have improved nephrogenetic care in direct and indirect ways:

a. I have set-up the multidisciplinary outpatient clinics for adult and pediatric nephrogenetics
b. I have designed the genepanels for our nephrogenetic diagnostic tests. Based on my scientific data, I have added a genepanel for early onset renal failure of all causes. With these genepanels, our diagnostic laboratory is one of the leading laboratories for diagnostic nephrogenetics.
c. I am the consulting geneticist for the multidisciplinary outpatient clinic for preconception and pregnancy care for patients with renal disease
d. I have coordinated both the NFU and the EU approval of our UMC Utrecht Center of Expertise for Hereditary and Congenital Nephrologic and Urologic Disorders. Now, as our chair, I am our representative in the newly formed European Reference Network: ERKNET (in which I am also working group co-chair).

I am in the ERA-EDTA Young Nephrologists Platform Board and I am in both the ERA-EDTA and the ESPN Working groups for inherited kidney disease. Recently I joined the Dutch Medical Specialist Council board for Innovation and Science on behalf of the Dutch clinical geneticists' association.

I am invited and principal author of the NFN guideline "Genetics for Nephrologists". I am on the Dutch multidisciplinary (SKMS) guideline committee for the guideline Renal Disease and Pregancy.

I enthusiastically teach about nephrogenetics to diverse audiences: Dutch Kidney Foundation PhD students, nephrologists, paediatric nephrologists, paediatricians, residents (both in genetics and nephrology) and patients. I co--organized the 2018 ERA-EDTA Young Nephrologists Platform/ Working Group Inherited Kidney Diseases CME on "How to become your local nephrogenetics expert" (Belgrade Oct 2018). I reach patients via contributions to activities, brochures and information movies of e.g. the Dutch Kidney Patient Foundation and the Dutch Kidney Foundation.