# Shanice Beerepoot

Course of peripheral neuropathy over time in patients with metachromatic leukodystrophy

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| **Short Bio** | I did my Bachelor study "Health and Life sciences" at the Vrije Universiteit in Amsterdam and graduated with honors in August 2014. As part of this study, I did a minor in Medicine which made me realize that I wanted to combine my research interests with clinical work. Therefore, I applied for the four-year Selective Utrecht Medical Master (SUMMA) at the Utrecht University, a special Master program in which students are trained in the field of both medicine and clinical research. I was fascinated by the anatomy and functionality of the human nervous system, so I did my general advanced clinical and research internships in Neurology. I graduated in July 2018 with a 4.0 GPA, and soon after, in August 2018, I started my Ph.D. studies regarding peripheral neuropathy in metachromatic leukodystrophy patients at the Amsterdam Leukodystrophy Center, Amsterdam University Medical Centre in Amsterdam and the Center for Translational Immunology, University Medical Centre Utrecht in Utrecht. Meanwhile, I am doing a post-graduate Master in Epidemiology and Medical Statistics at the Utrecht University. |
| **Home Institution** | Amsterdam Leukodystrophy Center, Department of Paediatric Neurology at Amsterdam University Medical Centre, Vrije Universiteit Amsterdam, The Netherlands |
| **Host Institution** | Department of Paediatric Neurology and Developmental Medicine, University Children's Hospital Tübingen, Germany |
| **Project description** | Metachromatic leukodystrophy (MLD) is an autosomal recessively inherited metabolic disorder in which especially the white matter of the brain and peripheral nerves are affected. Since clinical observations indicated that the severity and time course of peripheral neuropathy differ among MLD patients, and the impact of current treatments remains unclear, we aimed to study nerve conduction and ultrasound findings in a large MLD patient cohort with multiple measurements over time. This project allowed us to combine the data of German and Dutch MLD patients, resulting in a total of 285 |

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| | nerve conduction studies (from 97 patients) and 58 nerve ultrasounds (from 36 patients) to be analyzed. The initial findings indicate that peripheral neuropathy is most often severe, even in MLD patients without symptoms or with only cognitive symptoms. Interestingly, the nerve conduction velocity seems to remain quite stable over time without obvious influences of patient and treatment characteristics. Remarkably, patients with absent or only mild peripheral neuropathy over time shared a few exclusive genetic variants in the MLD gene ARSA. We are still working on this project. |
| **Personal statement** | My aim is to become a pediatric neurologist in an academic center. I have a special interest in rare leukodystrophies and I would like to combine patient care with clinical research in my future career. The EJP fellowship helps me to combine data from different centers to tackle the limited patient numbers in these rare diseases, and to develop my analytic skills with irregular, longitudinal data. In addition, collaborating with international colleagues in this field stimulates my clinical, academic and personal development. Finally, both the home institution and the host institution are international expert centers for MLD care and research. The insights from this collaborative study will allow the physicians to better inform not (yet) treated patients and their parents about future perspectives, and might also help to predict treatment outcomes and select patients for treatment modalities. |