Elke de Boer

Project title: *Integrative analysis of NGS and transcriptome data to elucidate the genetic etiology of unexplained cases of ERN ITHACA*

Duration	2 weeks
Short Bio	Elke is a medical doctor from training. She performed her PhD studies in the international context of the European consortium study Solve-RD. Solve-RD is based on the ERN structure and aims to genetically explain phenotypes of individuals with unexplained rare disease by systematic re-analysis, beyond-the-exome approaches and functional follow-up. During her PhD, Elke studied individuals with unexplained neurodevelopmental disorders and congenital anomalies.
Home Institution	Radboudumc, Nijmegen, the Netherlands
Host institution	Centre Hospitalier Universitaire, Dijon, France
Project description	My training stay in Dijon focused on the integrated analysis of whole genome sequencing (WGS) en RNA sequencing (RNAseq) data for individuals with rare genetic disorders, comprising neurodevelopmental disorders and congenital anomalies. I gained knowledge about RNAseq technology, analysis pipelines and interpretation steps, about integrating RNAseq and WGS analysis and interpretation, about RNAseq data visualization, and I expanded my knowledge about WGS interpretation.
Personal statement	In the long term, I aim to become a clinical geneticists with a research profile. Given that it can be expected that transcriptomics as well as WGS will enter the diagnostic process of unexplained rare disease cases, knowledge about this technology and its interpretation and integration with other datasets (NGS/phenotype) is highly beneficial for future patient care. This research mobility fellowship helped me in understanding the diseases of my future RD patients and in explaining this comprehensively to the families. For my ongoing

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	and future research studies, independently being able to analyse and interpret these types of datasets is an invaluable skill. One of my currently ongoing research studies involves WGS of undiagnosed individuals with a certain congenital anomaly syndrome. In this project, I can apply my learned skills on WGS interpretation. Also my experiences in Dijon as well as in Nijmegen can be applied with implementation of WGS in my new job (Erasmus MC), where WGS is not yet established. Additionally, in my new home institution Erasmus MC, RNAseq as integrated in both diagnostics and research. Because of my training stay, I can better understand the benefits and limitations of RNAseq in clinical practice as well as come up with research or diagnostic questions where RNAseq can be of use. Furthermore, this exchange has strengthened the collaboration of the Radboudumc with ERN ITHACA and Solve-RD partner Centre Hospitalier Universitaire de Dijon, leading to many more fruitful projects in the future.

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