Genomics Education Programme

NHS
Health Education England

# Jana Vandrovcova

Jana is a geneticist and bioinformatician with a long-standing interest in molecular diagnostics and advancing genetic-driven personalised treatment. After obtaining her PhD (2006) in Biomedicine on the genetics of colorectal cancer (Karolinska Institutet, Stockholm and Charles University, Prague) she received postdoctoral training at the Institute of Neurology, UCL (2006-2010) and later at the MRC Clinical Sciences Centre, Imperial College London (2011-2014).

During this time Jana was involved in genomics and next generation sequencing projects in several clinical areas including neurodegeneration, familial hypercholesterolemia and connective tissue disorders.

Jana has always been drawn to large scale computational data analysis and its potential to revolutionise patient care. In 2010 Jana undertook a master's degree in bioinformatics at Birkbeck, London and a subsequent one-year employment as a user support officer at the European Bioinformatics Institute, Hinxton. In 2014, she returned to the Institute of Neurology to continue research into the genetics of neurological diseases and became a bioinformatics lead of several rare diseases and dementia focused projects. Jana's work incorporates genetic mapping, genomic analysis as well as gene expression profiling and plays an important role in novel gene discovery.

## Outline of Research (Part Time, Postdoctoral)

## A search for the missing: the utility of whole genome sequencing in clinical grade diagnostics of hereditary neurological disorders.

The implementation of whole genome sequencing into routine clinical practice through the pioneering work of The 100,000 Genomes Project has a great promise to dramatically change diagnoses and treatments of patients with rare disorders. During her research Jana is aiming to maximize the potential of whole genome sequencing by implementing additional bioinformatics pipelines to identify mutations not fully investigated using current protocols.
This will include:
1. The identification and validation of structural, copy number and repeat expansion variants which are a particularly common cause of many neurological disorders.
2. Using in-house generated transcriptomic data, to identify and validate variants located in intra and intergenic regions that are expressed in relevant tissues such as human brain and muscle but are not a part of the current genome annotation.
3. Making use of large patient datasets and deep phenotype information to identify novel disease genes and genetic modifying factors.