



## Dr Melissa Nel

### Senior Lecturer, Department of Medicine Neuroscience Institute

Dr Melissa Nel completed her MBChB in 2008 and her PhD in 2016, both at the University of Cape Town (UCT). A senior lecturer in the Department of Medicine at UCT's Neuroscience Institute, she leads the Neurogenomics Lab. Dr Nel's current research focuses on elucidating the genetic basis of amyotrophic lateral sclerosis and other inherited neuromuscular disorders in African populations, using cutting-edge long-read whole genome sequencing technology in collaboration with the UCT-based Neurology Research Group and the ALS Africa Network. Dr Nel is passionate about African-led genomics research and local bioinformatics capacity building to drive the real-world implementation of genomic medicine in resource-limited settings. Her Neurogenomics Lab partners with DNA sequencing service providers in South Africa. This enables her team to deliver cost-effective and rapid analysis of genomic data for rare disease diagnosis according to international best practice guidelines.

Beyond research Nel contributes significantly to the scientific community as an expert member of ClinGen's ALS [Amyotrophic lateral sclerosis] Gene and Variant Curation Expert Panels, guiding clinical genetic testing practices globally. She also advocates for increased African representation in genetic research through her participation in the H3Africa's Rare Disease Working Group, addressing disparities in genomic data generation and sharing and proposing solutions for improved diagnosis of rare diseases on the continent.

As a registered ClinVar submitter, her lab promotes diversity and inclusion in genomic medicine by sharing curated genetic variant data globally. Their commitment to advancing equitable access to genomic medicine underscores their role as an emerging local leader in neurogenomics research, bridging critical gaps in understanding and treating ALS and neuromuscular disorders in African populations.