“Detection of low level mosaicism using High Resolution Melt, droplet digital PCR and single cell RNA sequencing: prognostic, diagnostic and screening applications.”

Dr David Godler

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Dr Godler leads the Cyto-Molecular Diagnostics Research group at the Murdoch Children’s Research Institute, regularly publishing in high quality specialty journals such as Clinical Chemistry, Neurology, JAMA Neurology, Genetics in Medicine and Human Molecular Genetics. Using the clinical resources of the Victorian Clinical Genetics Services and those of national and international collaborators, his work focuses on understanding disorder aetiology and on test development for: improved diagnosis of chromosomal abnormalities, non-invasive prenatal testing and epigenetic disorders associated with intellectual disability and autism. Dr Godler also heads the world’s largest fragile X syndrome prevalence study in 100,000 newborns (NHMRC funded), and a Prader-Willi Syndrome newborn screening pilot to provide evidence regarding expanding current newborn screening in Australia and internationally. He is also the PI on DNA methylation studies, funded through national and international philanthropy, utilising droplet digital PCR to detect low level mosaicism missed by standard testing in developmental delay referrals of unknown cause.