

Nicole de Leeuw, PhD

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Nicole de Leeuw has a Master of Science degree in Medical Biology (University of Utrecht, the Netherlands), a Master's in Nutritional Sciences (University of Illinois, USA) and a PhD in Molecular Biology from the University of Nijmegen, the Netherlands. She completed her four-year-training in clinical cytogenetics at the Nijmegen department of Human Genetics in 2003 and was subsequently appointed there as Clinical Laboratory Geneticist. Since 2012, she is leader of the theme Intellectual Disability & Congenital Anomalies within the Division of Genome Diagnostics in Nijmegen. Her main activities involve the application of genome wide SNP-based array analysis and Whole Exome Sequence (WES) data analysis of both nucleotide and copy number variants in constitutional genome diagnostics. Together with her colleagues she aims to efficiently identify the genetic cause of a patient's disorder and to unravel the underlying mechanism in order to provide adequate information for appropriate genetic counselling.

She is actively involved in external quality assessment, she participates in national and international educational activities; she is supervisor of the Clinical Laboratory Geneticist training programme in Nijmegen, member of the European Cytogeneticists Association (ECA) board of directors and coordinator of the ECARUCA database (www.ecaruca.net).