



Edwin Cuppen (born Boxmeer, The Netherlands in 1970) , obtained his masters in molecular sciences at the Agricultural University Wageningen in 1994 (cum laude) and his PhD degree in 1999 at the Radboud University in Nijmegen in the group of Prof. Be Wieringa. During his education, he spent half a year in het Jaenish lab at the Whitehead Institute in Boston. From 1999 to 2002 he performed postdoctoral research at the Netherlands Cancer Institute in Amsterdam and the Hubrecht Institute in Utrecht, The Netherlands, in the group of Prof. Ronald Plasterk. In 2002 he became a principle investigator at the Hubrecht Institute and in 2007 he was appointed professor of Genome Biology at the Biology department of the Utrecht University. In 2009 he was appointed professor of Human Genetics and head of the research section of the Medical Genetics department of the University Medical Center Utrecht, while continuing his work at the Hubrecht Institute.

His area of expertise is in genomics and genetics and his scientific interests are in functional and personal genomics. In 2005, Edwin Cuppen was awarded a prestigious European Young Investigators Award for his work on naturally occurring and induced genetic variation in the laboratory rat. He was one of the first to generate gene knockout models in the rat and recently showed the widespread effect of copy number variation on gene expression levels.

In his current work he combines experimental methods, including next-generation sequencing technology, with bioinformatic approaches to identify functional elements in genomes and to understand the effects of genetic variation under normal and disease conditions. His work at the Hubrecht Institute focusses on the use animal model systems to elucidate general mechanisms underlying the (molecular) phenotypic effects of genetic variation in genomes, with special focus on the effects of structural variation. At the University Medical Center research is driven by patient phenotypes, including cancer and focussed on the identification of the genetic basis. His research group has a long track record in high-throughput DNA analysis and was the first in Europe to implement AB's next-generation sequencing technology SOLiD. This platform is now used for genome sequencing and mutation detection/discovery, but also for (small) RNA sequencing, ChIP-Seq, and structural variation detection. Currently, programs are set up to implement next-generation sequencing approaches for diagnostic purposes in clinical genetics and personalized cancer treatment. The group has a variety of academic and non-academic collaborations for the development and scaling of next-generation sequencing applications as well as future single molecule sequencing.

Edwin Cuppen is inventor on various patents and cofounder and CSO of the biotech startup company InterNA Technologies that focuses on miRNA-based diagnostics and therapeutics. He is also cofounder of InterNA Genomics that provides bioinformatic services for the analysis of next-generation sequencing data, including small RNA discovery and profiling.

Key publications

Nijman I, Mokry M, van Boxtel R, Toonen P, de Bruijn E, and Cuppen E. (2010). Mutation discovery by targeted genomic enrichment of multiplexed barcoded samples. *Nature Methods* 7:913-5

Linsen SE, de Wit E, Janssens G, Heater S, Chapman L, Parkin RK, Fritz B, Wyman SK, de Bruijn E, Voest EE, Kuersten S, Tewari M, Cuppen E. (2009). Limitations and possibilities of small RNA digital gene expression profiling. *Nature Methods* 6:474-476.

Guryev V, Saar K, Adamovic T, Verheul M, van Heesch S, Cook S, Pravenec M, Aitman T, Jacob H, Shull JD, Hubner N, and Cuppen E. (2008) Distribution and functional impact of DNA copy number variation in the rat. *Nature Genetics* 40: 538-545.

Guryev V, Smits BMG, van de Belt J, Verheul M, Hubner N, and Cuppen E. (2006) Haplotype block structure is conserved across mammals. *PLoS Genetics* 2: e121

Berezikov E, Guryev V, van de Belt J, Wienholds E, Plasterk RHA, and Cuppen E. (2005) Phylogenetic shadowing and computational identification of human microRNA genes. *Cell* 120: 21-24.

Berezikov, E., Plasterk, R.H.A., and Cuppen, E. (2002). GENOTRACE: cDNA-based local GENOME assembly from TRACE archives. *Bioinformatics* 18:1396-7. (IF= 4.9)