Curriculum Vitae(senior)

Joris Vermeesch, Laboratory of cytogenetics and genome research)

EDUCATION AND PROFESSIONAL EXPERIENCE

2016-2020	Chair Department of Human Genetics, KU Leuven
2013-	Full Professor, Department of Human Genetics, KU Leuven
2009-	Part time full Professor, Department of Human Genetics KU Leuven
2008-	Coordinator Genomics Core, UZ-KU Leuven
2007-2009	Associate Professor, Department of Human Genetics, KU Leuven
2004-2007	Assistant Professor, Department of Human Genetics, KU Leuven
2001	Director Cytogenetics unit, Center of Human Genetics, UZ Leuven
1999-2001	Groupleader genomics in Aventis CropScience, Ghent, Belgium
1993-1999	Postdoctoral fellow, KU Leuven
1988-1993	Ph.D in Chemistry, Nebraska, USA
1988	Ir. Bioengineer University of Gent, Belgium

EDUCATIONAL ACTIVITIES

Supervised 23 PhD students and 9 postdoctoral fellows, four of which became independent professors. In addition, over 15 international trainees have visited the laboratory has over the last five years, of which several senior scientists. He is teaching several genetics and genomic courses at KU Leuven & was faculty member of European School of Genetic Medicine, 26thCourse in Medical Genetics ; Faculty member of ECA training courses in cytogenetics & Faculty member of European Advanced Postgraduate Course in Classical and Molecular Cytogenetics. In addition, he organized national master classes in genomics and is leading the interuniversity course on human genetics.

SELECTED RECENT MAJOR GRANTS

2019-2023 H2020-ES-MSCA-ITN: Innovative training network in reproduction (512K€)
2019-2021 4KANKER- PROSTK 2018-134: Genomic and epigenomic profiling of circulating tumor-DNA for non-invasive breast cancer detection (225K€)
2019-2023 TBM FWO T003819N: Long Read Sequencing for the detection of cryptic rearrangements in patients with developmental disorders (473K€; 2PIs)
2018-2022 DEFIS-DEFPR C14/18/092: SymBioSys: Computationally disentangling cellular heterogeneity (1,566K€; 5 PIs)
2017-2020 FWO G0E1117N: Structural variation as cause of phenotypic variability in 22q11DS patiënts (311K€)
2017-2019 IMINDDs: GAP; Human genome sequencing (377K€).
2016-2018 H2020-OTHER-WIDESPRE: Expanding scientific excellence for research to maternal and fetal health (252K€)

Infrastructure grants

2021-2024 IRI-FWO: EU-Genomics: EU Infrastructure for Genome Analysis (647K€)
2021-2023 Genomics Core, Core facility (150k€)
2019-2023 H2020-ES-RI: Europese geavanceerde infrastructuur voor innovatieve genomica; (777K€; Total 12M€)
2017-2021 COREFAC GENOMIC CORE -Core facility Genomics Core (150K€)

RESEARCH STATEMENT

Professor Joris Vermeesch heads the Laboratory for Cytogenetics and Genome Research, pioneering genomic technologies to map the causes and mechanisms underlying rare developmental disorders with a focus on structural variation and mosaicism detection. Technologies developed and implemented by the laboratory are arrayCGH, single cell haplotyping, liquid biopsy and whole genome sequencing to enable the study of early embryonic development and early placentation, non-invasive prenatal and cancer screening and monitoring and detection of novel causes of developmental

disorders. By combining those methods, we aim to unravel the mutational mechanisms which shape the human genome and are causing diseases from the early developmental stages to birth. In collaboration with clinical geneticists and clinical/educational psychologists and psychiatrists, the laboratory actively seeks to define the molecular causes of developmental, mental, and behavioral disturbances. The laboratory has been studying 22q11DS as a paradigm for other genomic disorders.

MAIN ACHIEVEMENTS

Genomic instability in human embryos Contrary to dogma, we demonstrated the human embryo is extremely prone to chromosomal segregation errors (Nature Medicine, > 50 publications). Subsequently, by developing single cell haplotyping methods we showed more recently non-canonical zygotic cleavage divisions causing whole genome segregation errors resulting in mixoploidy and chimerism (4 papers, Genome Biology). We obtained 3 patents on single cell sequencing and haplotyping, have worked with companies to create commercial products for the IVF market (OnePGT sold by Agilent) and have introduced the technologies in products used by preimplantation genetic testing laboratories.

Neurodevelopmental disorders caused by structural variation. We pioneered array technology and proved that CNVs are a major cause of developmental disorders (> 30 papers). We have either led or participated in the identification of novel syndromic rare disease entities caused by structural variation (> 100 papers). We succeeded in changing the diagnostic workflows in prenatal and postnatal diagnosis in Belgium, developed best practice guidelines as well as clinical guidelines and leveraged healthcare reimbursement. Internationally, we partnered in over 20 policy papers and guidelines papers. This work led to a spin-off company Cartagenia which developed genomic interpretation software solutions. Recent work focuses on implementing whole genome sequencing enabling genome wide mapping of genetic lesions, mapping the clinical consequences of genomic disorders, understanding the genetic causes leading to the variability seen in genomic disorders (COST,FWO action) and a FWO-TBM project to implement Long read sequencing.

22q11Deletion Syndrome: We focus on 22q11DS (1/2000 births). I was the European anchor in the international brain behavior Consortium (IBBC), an NIMH funded genomics project (10M$) in which WGS was obtained from 1500 22q11DS patients. We uncovered hypervariability in the low copy repeats and variation in the remaining allele which we hypothesize to underlie the high incidence of neurodevelopmental anomalies in this population. With the aim of understanding the causes for the phenotypic variability, we have been sequencing and mapping genetic lesions in 22q11DS patients leading to 25 publications, 10 last author. We discovered novel causes of rare phenotypic variation and uncovered unprecedented structural variation in the low copy repeats, likely drivers of the rearrangements (Nature Medicine, Mol.Psychiatry, ..)

Liquid Biopsy: Our group developed and implement low pass genome wide cfDNA sequencing methods leveraging non-invasive detection of fetal aneuploidies and cancers (20 first and last author publications, JAMA oncology, Lancet,…). Most recently, we have developed a method mapping the epigenomic changes (patent pending) and are combining multi-omics technologies to deconvolute cfDNA sequencing patterns and map disease states. We also developed methods to map rare variants. In 2021, we acquired an SBO-FWO grant (1.5M€) to further develop cfDNA analyses.

Founder and coordinator of KULeuven ***Genomics Core*** (www.genomicscore.be) facility, a knowledge hub for novel genomic technologies enabling access to both fundamental and clinical research as well as diagnostic activities. It concentrates all clinical and research activities, and bioinformatics necessary for providing expert single-cell and NGS services. The team exist of 23 persons. We acquired two large Hercules grant enabling us to acquire the PacBio Sequel I (1.2M€;2013-2016) and SequelII (500k€;2021-2025), became official KULeuven core facility and participate in *EASIGenomics (Horizon2020-infraia)* a network of 14 University cores.

Other scientific output and impact

Keynote/invited speaker on over 25 international meetings/year during the last 5 years including all main international meetings concerning reproductive genetics (2019, Beijng, International Society Reproductive Genetics; ESHRE 2017,2019; ASHRE 2018;, 2019; ISPD 2016, 2017, organizer 2018; PGDInternational Society (2018 Bangkok, Berlin,.., 2013-2020; Montreal 2018), gynaecology (Moscow 2019; Naples 2019; Utrecht 2019;…) and genetics (European Society of Human Genetics, 2017, 2018, 2019; European cytogenetics Society, 2015, 2017, 2019(keynote)as well as national genetic societies Berlin, 2019; Porto, 2020, Greece 2021, Neurenberg, 2022).

EXTENDED PUBLICATION LIST

Journal articles

Lannoo, L., van Straaten, K., Breckpot, J., Brison, N., De Catte, L., Dimitriadou, E., Legius, E., Peeters, H., Parijs, I., Tsuiko, O., Vancoillie, L., Vermeesch, J.R., Van Buggenhout, G., Van den Bogaert, K., Van Calsteren, K., Devriendt, K. with Devriendt, K. (corresp. author) (2022). Rare autosomal trisomies detected by non-invasive prenatal testing: an overview of current knowledge. *EUROPEAN JOURNAL OF HUMAN GENETICS*. doi: 10.1038/s41431-022-01147-1

Tuveri, S., Debackere, K., Marcelis, L., Dierckxsens, N., Demeulemeester, J., Dimitriadou, E., Dierickx, D., Lefesvre, P., Deraedt, K., Graux, C., Michaux, L., Cools, J., Tousseyn, T., Vermeesch, J.R., Wlodarska, I. with Wlodarska, I. (corresp. author) (2022). Primary mediastinal large B-cell lymphoma is characterized by large-scale copy-neutral loss of heterozygosity. *GENES CHROMOSOMES & CANCER*, *61* (10), 603-615. doi: 10.1002/gcc.23069

Che, H., Jatsenko, T., Lenaerts, L., Dehaspe, L., Vancoillie, L., Brison, N., Parijs, I., Van den Bogaert, K., Fischerova, D., Heremans, R., Landolfo, C., Testa, A.C., Vanderstichele, A., Liekens, L., Pomella, V., Wozniak, A., Dooms, C., Wauters, E., Hatse, S., Punie, K., Neven, P., Wildiers, H., Tejpar, S., Lambrechts, D., Coosemans, A., Timmerman, D., Vandenberghe, P., Amant, F., Vermeesch, J.R. with Vermeesch, J.R. (corresp. author) (2022). Pan-Cancer Detection and Typing by Mining Patterns in Large Genome-Wide Cell-Free DNA Sequencing Datasets. *CLINICAL CHEMISTRY*. doi: 10.1093/clinchem/hvac095 Open Access

Siermann, M., Tsuiko, O., Vermeesch, J.R., Raivio, T., Borry, P. with Siermann, M. (corresp. author) (2022). A review of normative documents on preimplantation genetic testing: Recommendations for PGT-P. *GENETICS IN MEDICINE*, *24* (6), 1165-1175. doi: 10.1016/j.gim.2022.03.001

Petsophonsakul, P., Pirmani, A., De Brouwer, E., Akand, M., Botermans, W., Van Der Aa, F., Vermeesch, J.R., Offner, F., Wuyts, R., Moreau, Y., Maes, I., Blockx, I., Van Rompuy, P., Lewi, M., Vannieuwenhuyse, B. (2022). Augmenting THerapeutic Effectiveness Through Novel Analytics (ATHENA) - A Public and Private Partnership Project Funded by the Flemish Government (VLAIO). *Stud Health Technol Inform*, *294*, 829-833. doi: 10.3233/SHTI220601 Open Access

Vermeesch, J.R., Lenaerts, L. with Vermeesch, J.R. (corresp. author) (2022). Commentary on Multiple Copy Number Variants Detected by Noninvasive Prenatal Testing Comment. *CLINICAL CHEMISTRY*, *68* (5), 634-634. doi: 10.1093/clinchem/hvac042

Vermeesch, J.R. (2022). The Hunt for the Chromosome 22q11.2 Deletion Syndrome Schizophrenia Genes. *Biol Psychiatry*, *91* (8), 692-693. doi: 10.1016/j.biopsych.2022.02.002

Rack, K., De Bie, J., Ameye, G., Gielen, O., Demeyer, S., Cools, J., De Keersmaecker, K., Vermeesch, J.R., Maertens, J., Segers, H., Michaux, L., Dewaele, B. with Dewaele, B. (corresp. author) (2022). Optimizing the diagnostic workflow for acute lymphoblastic leukemia by optical genome mapping. *AMERICAN JOURNAL OF HEMATOLOGY*, *97* (5), 548-561. doi: 10.1002/ajh.26487 Open Access

Masset, H., Ding, J., Dimitriadou, E., Debrock, S., Tsuiko, O., Smits, K., Peeraer, K., Voet, T., Esteki, M.Z., Vermeesch, J.R. (2022). Single-cell genome-wide concurrent haplotyping and copy-number profiling through genotyping-by-sequencing. *NUCLEIC ACIDS RESEARCH*. doi: 10.1093/nar/gkac134 Open Access

Lejeune, C., Dierickx, D., Wildiers, H., Lannoo, L., Van Calsteren, K., Vandecaveye, V., Menten, B., Vermeesch, J., Amant, F. with Amant, F. (corresp. author) (2022). Pushing the boundaries. Concurrent Hodgkin lymphoma and breast cancer treatment with preservation of pregnancy: A case report. *Gynecologic Oncology Reports*, *39*, Art.No. 100937. doi: 10.1016/j.gore.2022.100937 Open Access

Siermann, M., Claesen, Z., Pasquier, L., Raivio, T., Tsuiko, O., Vermeesch, J.R., Borry, P. with Siermann, M. (corresp. author) (2022). A systematic review of the views of healthcare professionals on the scope of preimplantation genetic testing. *JOURNAL OF COMMUNITY GENETICS*, *13* (1), 1-11. doi: 10.1007/s12687-021-00573-w Open Access

Dierckxsens, N., Li, T., Vermeesch, J.R., Xie, Z. with Dierckxsens, N. (corresp. author), Xie, Z. (corresp. author) (2021). A benchmark of structural variation detection by long reads through a realistic simulated model. *GENOME BIOLOGY*, *22* (1), Art.No. ARTN 342. doi: 10.1186/s13059-021-02551-4 Open Access

Paluoja, P., Teder, H., Ardeshirdavani, A., Bayindir, B., Vermeesch, J., Salumets, A., Krjutskov, K., Palta, P. with Palta, P. (corresp. author) (2021). Systematic evaluation of NIPT aneuploidy detection software tools with clinically validated NIPT samples. *PLOS COMPUTATIONAL BIOLOGY*, *17* (12), Art.No. ARTN e1009684. doi: 10.1371/journal.pcbi.1009684 Open Access

De Coster, T., Masset, H., Tsuiko, O., Smits, K., Van Soom, A., Vermeesch, J. (2021). 51 Genome-wide abnormalities resulting from heterogoneic cell division persist in the blastocyst-stage bovine embryo. *Reprod Fertil Dev*, *34* (2), 260-261. doi: 10.1071/RDv34n2Ab51

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Tsuiko, O., Vanneste, M., Melotte, C., Ding, J., Debrock, S., Masset, H., Peters, M., Salumets, A., De Leener, A., Pirard, C., Kluyskens, C., Hostens, K., van de Vijver, A., Peeraer, K., Denayer, E., Vermeesch, J.R., Dimitriadou, E. with Vermeesch, J.R. (corresp. author), Dimitriadou, E. (corresp. author) (2021). Haplotyping-based preimplantation genetic testing reveals parent-of-origin specific mechanisms of aneuploidy formation. *NPJ GENOMIC MEDICINE*, *6* (1), Art.No. ARTN 81. doi: 10.1038/s41525-021-00246-0 Open Access

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Paluoja, P., Teder, H., Ardeshirdavani, A., Bayindir, B., Vermeesch, J., Salumets, A., Krjutškov, K., Palta, P. (2021). Systematic evaluation of NIPT aneuploidy detection software tools with clinically validated NIPT samples. doi: 10.1101/2021.05.27.445941 Open Access

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A single-center retrospective analysis of over 85,000 pregnancies. *ECLINICALMEDICINE*, *35*, Art.No. ARTN 100856. doi: 10.1016/j.eclinm.2021.100856 Open Access

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Fiksinski, A.M., Bearden, C.E., Bassett, A.S., Kahn, R.S., Zinkstok, J.R., Hooper, S.R., Tempelaar, W., Vorstman, J.A S., Breetvelt, E.J. with Fiksinski, A.M. (corresp. author) (2021). A normative chart for cognitive development in a genetically selected population. *NEUROPSYCHOPHARMACOLOGY*, *47* (7), 1379-1386. doi: 10.1038/s41386-021-00988-6

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patients with ectrodactyly. *EUROPEAN JOURNAL OF MEDICAL GENETICS*, *63* (11), Art.No. ARTN 104009. doi: 10.1016/j.ejmg.2020.104009 Open Access

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