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Hors classe (Section 65)
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Research area

Theme: Genetics and genomics of autoimmune and inflammatory diseases
Methods: multifactorial genetics, integrative genetics, multiomics, biostatistics, bioinformatics

Prior professional experience

- *2022 - :* Associate Professor in Biostatistics, Hors Classe, Medical School **Université Paris Cité, Institut Necker Enfants Malades (INEM)**, team IMMEDIAB of Nicolas Venteclef; project: multiomics of diabetes
- *2019-2021:* Associate Professor in Biostatistics, **Hors Classe**, Medical School Université de Paris, **Centre de Recherche des Cordeliers**, **team IMMEDIAB** of Nicolas Venteclef; project: multiomics of type 2 diabetes and cardiovascular complications
- *2011-2019:* **Associate Professor in Biostatistics**, classe normale, Medical School **Université Paris Diderot**, INSERM UMRS-958 Genetics of diabetes of Cécile Julier; project: genetics and genomics of type I diabetes
- *2009-2011:* **Advanced Postdoctoral Scientist**, **INSERM UMRS-958** Genetics of diabetes of Cécile Julier; project: genetical genomics of type I diabetes
- *2005-2009:* **Post-Doctoral Scientist**, **Wellcome Trust Centre for Human Genetics, Oxford**, lab of Pr JC Knight; project: genetic variability of gene expression in the human MHC
- *2000-2004:* **PhD**, Necker hospital, **INSERM U580**, headed by Pr HJ Garchon; project: genetic susceptibility of auto-immune acquired myasthenia gravis
- *1999-2000:* Master 2 (DEA) 6-months **internship**, Necker hospital, **INSERM U25** of JF Bach; project: genetic susceptibility of auto-immune acquired myasthenia gravis
- *1998:* 2-months **internship**, **Pasteur Institute of Lille**, **CNRS EP10** headed by Pr P Froguel ; project: genetics of type 2 diabetes and cardio-vascular complication in Mauritius
- *1998:* one-year **internship**, Saint-Antoine hospital, Université Pierre et Marie Curie, **INSERM U 402** headed by Pr J Capeau ; project: candidate gene analysis in lipoatrophic diabetes
- *1997:* 2-months **internship**, **Généthon II, CNRS URA 1922**, headed by Pr J Weissenbach; project: genetic homozygoty mapping in a skin disease

Education and degrees

***2004*: Ph.D**. in Genetics/Immunology, University Paris Descartes
***2000*: Advanced Diploma** (DEA/M2) of Molecular Genetics, University Paris Descartes
***1999*: M. Sc.** in Cell Biology and Physiology, University Pierre & Marie Curie
***1998*: B.Sc.** in Cell Biology and Physiology, University Pierre & Marie Curie

Further Education

2020: Certifiens, **certificate in pedagogy**, SAPIENS, Alliance Sorbonne Paris Cité
2018: MOOC on **reproducible Science**, France Université Numérique and INRIA
2018: MOOC on **metabolomics**, France Université Numérique and CNRS
2018: MOOC on **Ethics in Research**, France Université Numérique and Université de Lyon
2016: **Bayesian statistics**, Inserm course
2012: EBI Advanced **RNA-Seq and ChIP-Seq** data analysis course
2010: **Databases** for biology, Inserm course
2008: Introduction to **SQL**, IT Learning Program, Oxford University

2008: **Statistics for Biosciences**, Online R course for Medical Science division, supported by the Dept. of Statistics, Oxford University. Since 2008, this module is one of the four core certificates of the Oxford University MSc in Bioinformatics.
2007: **Affymetrix training** on expression exon arrays
2006: **Statistics with R** Level 3: Computing and Graphics, IT Learning Program, Oxford University.
2006: Wellcome Trust Centre Advanced Course: **Microarrays and transcriptome**

Teaching activities

- **Teaching:** 192 hours per year in biostatistics, bioinformatics, genomics and genetics at Medical school and in the Life Sciences department of Université Paris Cité and two professional courses at the French Institute of Bioinformatics (IFB) and at the Institut François Jacob at the CEA.

I am in charge of several courses in the different departments:

- "population genetics" and "data analysis" at Medical School
- "Massive genomics data analysis", "RNASeq analyses", "Human genetics: methodology", "bioinformatics-genomics" for master students of European Master of Genetics
- "R/statistics" module of the professional training in integrative bioinformatics at the IFB
- "RNASeq analyses" professional week-course for the Institute François Jacob at the CEA

- **Pedagogy:**
  - 2020 : CertifiENS certificate in pedagogy from SAPIENS, Alliance Sorbonne Paris Cité
  - PLASMA: development of an e-Learning platform to teach analysis of massive data (plasmabio.org) and its documented and open-source software solution (https://github.com/plasmabio/plasma); 3 grants; 2 national (JOBIM 2020, JRES 2022) and 2 international conferences (JupyetrCon 2020, ESHG 2021) communications and **one peer-reviewed publication** in 2022: https://hal.archives-ouvertes.fr/hal-03563658

Scientific expertise

- **Local:** elected member of the research board of Paris Diderot University (2018-2020), elected member of the scientific board of the Medical school of Université Paris Cité since 2014, jury panel of the USPC doctoral school in 2016, jury member of 3 PhDs, 2 thesis committees
- **National**: Member of the Scientific Advisory Board of TAGC lab (Marseille) since 2022, organizing committee of JOBIM 2021, HCERES reviewer since 2017, elected member of the INSERM Specialized Scientific Committee 1 (2016-2021, vice-president in 2017 for the competitive recruitment of researchers), external expert of the INSERM CSS1 (2022-2026), committee panels for the recruitment of 8 associate professors, 1 thesis committee in Lille
- **International**: ESHG 2022, 2023 abstract reviewer; Diabetes UK; Peer-reviewing: Nature Communications, Diabetologia, Diabetes, Clinical Genetics and 7 other journals

Edition

Guest-editor of a special issue in "Genes" [IF = 4.1] on genetics of diabetes in 2015 with 8 papers

Research and pedagogy grants and awards

2024: Société francophone du diabète (SFD) – Association des Jeunes Diabétiques (AJD), Glygene project, 25 k€, PI
2022-2023 : ANR-18-IDEX-0001, Université de Paris – National University of Singapore, call for innovative projects in Higher Education, Meet UP with NUS : PLASMA project, 46 k€, co-PI
2019-2022: IDEX Université de Paris, axe stratégique Formation d'excellence, PLASMA project, 62 k€, co-PI
2019-2021: Région Ile-de-France, Trophées EdTech 2018, PLASMA project, 75 k€, co-PI
2013-2014: Juvenile Diabetes Research Foundation Innovative grant, $110,000, PI
2009-2013: NIH-DP3, $299,520, leader of workpackage 2
2011-2012: JDRF Transition Award, $100,000, PI
2010-2011: PHC Alliance between France and UK, 7800 euros and £4400, co-PI
2009-2011: JDRF Advanced Postdoctoral Fellowship: $91,000, PI
2007: Oxford University, Wellcome Trust Centre for Human Genetics, Individual Exceptional Merit Award

Scientific direction

2 postdoc scientists, 1 PhD, 1 EPHE diploma, 7 M2 (5 French, 2 foreign), 1 Master in UK, 1 technician

Scientific publications

*29 = 24 [author] + 4 [investigator] as part of the Type 1 Diabetes Genetics Consortium marked by **
3 corresponding author[@] and 7 1st author*; h-index=17, 2693 citations (Scopus); supervised; blue: selected publications*

1. Julla JB, Girard D, Diedisheim M, Saulnier PJ, Tran Vuong B, Blériot C, Carcarino E, De Keizer J, Orliaguet L, Nemazanyy I, Potier C, Khider K, Tonui DC, Ejlalmanesh T, Ballaire R, Mambu Mambueni H, Germain S, Gaborit B, Vidal-Trécan T, Riveline JP, Garchon HJ, Fenaille F, Lemoine S, Potier L, Castelli F, Carlier A, Masson D, Roussel R, **Vandiedonck C**, Hadjadj S, Alzaid F, Gautier JF, Venteclef N. Blood Monocyte Phenotype Is A Marker of Cardiovascular Risk in Type 2 Diabetes. Circ Res. 2023 Dec 28. *Online ahead of print*. [IF: 20.1] [doi: 10.1161/CIRCRESAHA.123.322757]

   ↳ First main paper by the PhD student I co-supervised showing an alteration of monocyte phenotypes associated to an increase risk of cardiobascular events in patients with type 2 diabetes using immunophenotypic, metabolomic and transcriptomic data.

2. Fumeron F, Velho G, Alzaid F, El Boustany R, **Vandiedonck C**, Bonnefond A, Froguel P, Potier L, Marre M, Balkau B, Roussel R, Venteclef N. Genetic variants of interferon-response factor 5 are associated with the incidence of chronic kidney disease: the D.E.S.I.R. study. Genes Immun. 2023 Nov 17 [IF: 4.25] [doi: 10.1038/s41435-023-00229-4 ; hal-04342437].

3. Laredo M, **Vandiedonck C**, Miró Ò, González Del Castillo J, Alquézar-Arbé A, Jacob J, Piñera P, Mégarbane B. Are there differences in the relationship between respiratory rate and oxygen saturation between patients with COVID-19 and those without COVID-19? Insights from a cohort-based correlational study. Emerg Med J, 2023 Oct3 ; emermed-2022-212882 [IF : 3.81] [doi: 10.1136/emermed-2022-212882 ; hal-04245035v1]

4. Girard D and **Vandiedonck C[@] (2022)** How dysregulation of the immune system promotes diabetes mellitus and cardiovascular risk complications. ***Front Cardiovasc Med,*** 2022 Sept 29; 9:991716 [IF:6.05; 5 citations] @corresponding author. Review [doi: 10.3389/fcvm.2022.991716; hal-03814151v1]

   ↳ Review co-written with Diane Girard I co-supervised for her PhD. It describbes mechanisms of immune system dysregulation, including genetic factors either specific or shared between type 1 diabetes, type 2 diabetes and atherosclerosis.

5. Kevorkian JP, **Vandiedonck C**, Laganier J, Lopes A, Burlacu R, Féron F, Chaix ML, Sène D, Riveline JP, Gautier JF, Mégarbane B **(2022)**. High-dose corticosteroids adjusted to oxygen requirement and monitoring of serum C-reactive protein to improve outcome of non-critically ill Covid-19 patients - The CocAA-CoLa Plus Study. ***Minerva Medica***, 2023 May 11 [IF: 5.58] [doi: 10.23736/S0026-4806.22.08326-4 ; hal-04245021v1]

6. Kevorkian JP, Lopes A, Sène D, Riveline JP, **Vandiedonck C**, Féron F, Nassarmadji K, Mouly S, Mauvais-Jarvis F, Gautier JF, Mégarbane B **(2021)** Oral corticoid, aspirin, anticoagulant, colchicine, and furosemide to improve the outcome of hospitalized COVID-19 patients - the COCAA-COLA cohort study. ***J Infect.*** 2021 Feb. [IF:38.6; 11 citations] [doi: 10.1016/j.jinf.2021.02.008; hal-03858469]

7. Kevorkian JP, Riveline JP, **Vandiedonck C**, Girard D, Galland J, Féron F, Gautier JF, Mégarbane B. **(2021)** Early short-course corticosteroids and furosemide combination to treat non-critically ill COVID-19 patients: An observational cohort study. ***J Infect.*** 2021 Jan;82(1):e22-e24. [IF:38.6; 10 citations] [doi: 10.1016/j.jinf.2020.08.045; hal-03858561].

8. Diedisheim M, Carcarino E, **Vandiedonck C**, Roussel R, Gautier JF, Venteclef N. **(2020)**, Regulation of inflammation in diabetes: From genetics to epigenomics evidence. ***Mol Metab***. 2020 Nov;41:101041. Review [IF:8.6; 21 citations] [doi: 10.1016/j.molmet.2020.101041; hal-02989301v1]

9. **Vandiedonck C[@] (2018).** Genetic association of molecular traits: a help to identify causative variants in complex diseases. ***Clinical Genetics***, 2018 Mar, 93(3) :520-532. [IF= 4.4; 33 citations] *@corresponding author*, Review [doi: 10.1111/cge.13187; hal-03814278v1]

↳ This reference presents an exhaustive review of variants regulating gene expression (eQTLs) and other molecular traits and their usefulness in deciphering causative variants in complex diseases.

10. Lenfant C, Baz P, Degavre A, Philippi A, Senée V, **Vandiedonck C**, Derbois C, Nicolino M, Zalloua P, Julier C. **(2017)** Juveline-onset diabetes and congenital cataract: « double-gene » mutations mimicking a syndromic diabetes presentation. ***Genes***, Nov 2017, 8(11) [IF = 4.1; 8 citations] [doi: 10.3390/genes8110309; hal-01848294v1]

11. Gwinner F, Boulday G, **Vandiedonck C**, Arnould M, Cardoso C, Nikolayeva I, Guitart-Pla O, Denis CV, Christophe OD, Beghain J, Tournier-Lasserve E, Schwikowski B. **(2016)** Network-based analysis of omics data: The LEAN method. ***Bioinformatics***, Oct 2016 [IF=6.9; 22 citations] [doi :10.1093/bioinformatics/btw676]

12. Holt RJ, **Vandiedonck C**, Willis-Owen SA, Knight JC, Cookson WO, Moffatt MF, Zhang Y **(2015)** A functional AT/G polymorphism in the 5'-untranslated region of SETDB2 in the IgE locus on human chromosome 13q14. ***Genes and Immunity*** , Oct 2015, 16, 488-494 [IF=2.7; 5 citations] [doi:10.1038/gene.2015.36]

13. Onengut-Gumuscu S, Chen WM, Burren O, Cooper NJ, Quinlan AR, Mychaleckyj JC, Farber E, Bonnie JK, Szpak M, Schofield E, Achuthan P, Guo H, Fortune MD, Stevens H, Walker NM, Ward LD, Kundaje A, Kellis M, Daly MJ, Barrett JC, Cooper JD, Deloukas P; **Type 1 Diabetes Genetics Consortium***, Todd JA, Wallace C, Concannon P, Rich SS. **(2015)** Fine mapping of type 1 diabetes susceptibility loci and evidence for colocalization of causal variants with lymphoid gene enhancers. ***Nature Genetics***, 2015 April; 47 (4):381-386 [IF= 41.4; 445 citations] **among the 426 collaborators.*[doi:10.1038/ng.3245]

↳ This study of reference is the final step of the GWAS performed on Type 1 Diabetes led by the T1DGC, with a fine mapping of the associated regions thanks to the use of a high-resolution microarray. This paper is among the top 1% of highly cited papers (source: Web of Science)

14. Plant K, Fairfax BP, Makino S, **Vandiedonck C**, Radhakrishnan et Knight JC **(2014)** Fine mapping genetic determinants of the highly variably expressed MHC gene ZFP57. ***European Journal of Human Genetics***, 2014 Apr; 22(4):568-71 [IF=5.3; 13 citations] [doi:10.1038/ejhg.2013.244]

15. Hunt KA, Smyth DJ, …, **Type 1 Diabetes Genetics Consortium***, et al. **(2012)** Rare and functional SIAE variants are not associated with autoimmune disease risk in up to 66,924 individuals of European ancestry. ***Nature Genetics***, 2012 January; 44(1):3-5 [IF= 41.4; 35 citations] **among the 473 collaborators.*[doi: 10.1038/ng.1037]

16. **Vandiedonck C**[@], Taylor MS, Lockstone HE, Plant K, Taylor JM, Durrant C, Broxholme J, Fairfax BP and Knight JC **(2011)** Pervasive haplotypic variation in the spliceo-transcriptome of the human MHC. ***Genome Research,*** 2011 Jul ; 21(7) :1042-54. [Impact Factor, IF= 14.6; 56 citations] *@corresponding author* [doi: 10.1101/gr.116681.110; hal-03814273v1]

↳ This study presents the first transcription map of the human Major Histocompatibility Complex (MHC) accounting for the genetic diversity of this most diverse genomic region of the human genome. It reveals a differential expression depending on the genetic background and that the alternative splicing mechanism is more frequent in the MHC than in the rest of the genome. This paper made the cover of the Genome Research journal issue and was commented in Nature Review Genetics : "Complex disease : ups and downs at the MHC". Muers M. ***Nature Review Genetics*** 2011, 12(7) :456-7 [doi: 10.1038/nrg3021].

17. Holt RJ, Zhang Y, Binia Aristea, Dixon AL, **Vandiedonck C**, Cookson WO, Knight JC and Moffatt MF **(2011)** Allele-specific transcription of the asthma associated gene PHF11 modulated by Oct-1. ***J Allergy Clin Immun,*** 2011 Apr; 127(4):1054-62. [IF= 13.1; 13 citations][doi: 10.1016/j.jaci.2010.12.015]

18. Hilner JE, Perdue LH, Sides EG, Pierce JJ, Wägner AM, Aldrich A, Loth A, Albret L, Wagenknecht LE, Nierras C, Akolkar B; **T1DGC* (2010).** Designing and implementing sample and data collection for an international genetics study: the Type 1 Diabetes Genetics Consortium (T1DGC). ***Clin Trials,*** 2010; 7(1 Suppl):S5-S32. [IF=2.7; 15 citations] *******among the 425 collaborators* [doi: 10.1177/1740774510373497]

19. **Vandiedonck C** and Knight JC **(2009).** The human Major Histocompatibility Complex as a paradigm in genomics research. ***Brief Funct Genomics***, 2009 Sep;8(5):379-94. [IF= 4.2; 81 citations], Review [doi: 10.1093/bfgp/elp010]

20. Barrett JC, Clayton DG, Concannon P, Akolkar B, Cooper JD, Erlich HA, Julier C, Morahan G, Nerup J, Nierras C, Plagnol V, Pociot F, Schuilenburg H, Smyth DJ, Stevens H, Todd JA, Walker NM, Rich SS; **Type 1 Diabetes Genetics Consortium* (2009)**. Genome-wide association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes. ***Nature Genetics***, 2009 Jun;41(6):703-7. [IF= 41.4; 1352 citations] *among the 382 collaborators [doi: 10.1038/ng.381]

21. **Vandiedonck C**, Raffoux C, Eymard B, Tranchant C, Dulmet E, Krumeich S, Gajdos P, Garchon HJ **(2009)** Association of HLA-A in autoimmune myasthenia gravis with thymoma. ***J Neuroimmunol***, 2009 May 29;210(1-2):120-3. [IF=3.4; 28 citations][doi: 10.1016/j.jneuroim.2009.02.004]

22. Campino S, Forton J , Raj S, Mohr A , Auburn S , Fry A, Mangano V, **Vandiedonck C**, Richardson A, Rockett K, Clark T, Kwiatkowski D. **(2008)** Validating discovered cis-acting regulatory genetic variants: application of an allele specific expression approach to HapMap populations. ***Plos One***, 3(12):e4105. [IF= 3.2; 20 citations] [doi: 10.1371/journal.pone.0004105]

23. Taylor JM, Wicks K, **Vandiedonck C**, Knight JC (**2008**) Chromatin profiling across the human tumour necrosis factor gene locus reveals a complex, cell type-specific landscape with novel regulatory elements. ***Nucleic Acids Res***, 36(15):4845-62. [IF= 19.1; 22 citations] [doi: 10.1093/nar/gkn444]

24. Giraud M, **Vandiedonck C**, Garchon HJ **(2008)** Genetic factors in autoimmune myasthenia gravis. ***Annals of New York Academy of Sciences***, 1132:180-92. [IF=4.7; 74 citations][doi: 10.1196/annals.1405.027]

25. Giraud M, Taubert R, **Vandiedonck C**, Ke X, Levi-Strauss M, Pagani F, Baralle FE, Eymard B, Tranchant C, Gajdos P, Vincent A, Willcox N, Beeson D, Kyewski B, Garchon HJ **(2007)** An IRF8-binding promoter variant and AIRE control CHRNA1 promiscuous expression in thymus. ***Nature,*** 448:934-937. [IF= 69.5; 169 citations][doi: 10.1038/nature06066]

↳ This study unveils the genetic and molecular mechanisms controlling the central immune tolerance to the muscle acetylcholine receptor, the target autoantigen of an autoimmune disease, acquired myasthenia gravis.

26. **Vandiedonck C**, Capdevielle C, Giraud M, Krumeich S, Jais JP, Eymard B, Tranchant C, Gajdos P, Garchon HJ **(2006)** Association of the PTPN22*R620W Polymorphism with Autoimmune Myasthenia Gravis. ***Annals of Neurology,*** 59:404-407. [IF=10.4; 107 citations][doi: 10.1002/ana.20751]

↳ This study showed for the first time the important predisposing role of this genetic variant in autoimmune myasthenia gravis. Since, this variant with pleiotropic effects in numerous autoimmune diseases has been replicated in several GWAS.

27. **Vandiedonck C**, Giraud M, Garchon HJ **(2005)** Genetics of autoimmune myasthenia gravis: the multifacted contribution of the HLA complex. ***J Autoimmunity***, vol 25, Supp1:6-11. [IF= 14.5; 34 citations], Review [doi: 10.1016/j.jaut.2005.09.010]

28. **Vandiedonck C**, Beaurain G, Giraud M, Hue-Beauvais C, Eymard B, Tranchant C, Gajdos P, Dausset J, Garchon HJ **(2004)** Pleiotropic effects of the 8.1 HLA haplotype in patients with autoimmune myasthenia gravis and thymus hyperplasia. ***Proc Natl Acad Sci U S A,*** 101:15464-15469. [IF= 12.5; 76 citations] [doi: 10.1073/pnas.0406756101]

↳ This work is my major PhD result. It reveals the complexity of genetic associations of the MHC in autoimmune myasthenia gravis. At least three distinct effects were demonstrated.

29. Fischer J, Urtizberea JA, Pavek S, **Vandiedonck C**, Bruls T, Saker S, Alkatip Y, Prud'homme JF, Weissenbach J **(1998)** Genetic linkage of progressive pseudorheumatoid dysplasia to a 3-cM interval of chromosome 6q22. ***Hum Genet***, 103:60-64. [IF= 4.1; 18 citations] [doi: 10.1007/s004390050784]

Software and tools

Plasma (https://github.com/plasmabio/plasma), 2 Jupyter applications: ipycytoscape (https://github.com/cytoscape/ipycytoscape) and ipyigv (https://github.com/QuantStack/ipyigv) and the upgrade of nbgrader0.8 (github.com/jupyter/nbgrader/) were funded by the PlasmaBio project

Oral communications

7 after peer-reviewing and 2 upon invitation at international conferences, 6 in research lab institutes

- Sélection :

- European Society of Human Genetics (ESHG), Virtual Conference, ePoster 28/08/2021
- American Society of Human Genetics (ASHG) 60th annual meeting, Washington DC, 04/11/2010
- Nature conference, 4th Genomics of Common Diseases meeting, Houston, 08/10/2010
- Federation of Clinical Immunology Societies (FOCIS) 10th Annual Meeting, Boston, 25/06/2010
- Seminar at the Sanger Institute during the Wellcome Trust Centre Advanced Course : Microarrays and transcriptome, 09/04/2006
- Autoimmunity and Immunopathologie club (SFI), Paris, 25/06/2003
- Federation of Clinical Immunology Societies (FOCIS) 3rd Annual Meeting, Paris, 18/05/2003

- Invitation :

- Jupyter workshop for education, CNAM, Paris, 22-24/01/2023
- Institut de Recherche Saint-Louis (IRSL), Paris, Inserm UMR-S 718, unit seminar, 05/04/2019
- Institut Cochin, Paris, seminar in the team of Agnès Lehuen, 28/01/2019
- Institut des Cordeliers (UMR_S 1138), Paris, seminar in the team "Immunity and Metabolism in Diabetes – IMMEDIAB" of Nicolas Venteclef, 16/01/2019
- USPC – Universidade de Sao Paulo Meeting – Hematology/bioinformatics conference, Paris, 08/03/2016
- Alternative Splicing day for bioinformatics, Statistical Department of Oxford, 22/04/2008
- Association Française contre la myopathie (AFM), Paris, 12/05/2002
- INSERM U 383, Paris, unit seminar 14/05/2001

Science outreach

Invited speaker at the Pasteur Institute for the first "Researchers and research volunteers colloquium" on 10/06/2016 *https://www.youtube.com/watch?v=sinJqj4kpUU&feature=youtu.be*