## Postdoctoral scientist – computational biology in clinical genetics

## Towards an understanding of the phenotypic variability of 22q11DS

SymbioSys is a consortium of computational and molecular biologists and (cyto)geneticists at the University of Leuven (Belgium) focusing on the detection of individual genomic variation, how it arises and leads to specific types of constitutional disorders and cancer. The candidate will be a key player managing an international project co-funded by NIMH to unravel the phenotypic variability of the 22q11 Deletion Syndrome. The 22q11.2 deletion syndrome (22q11.2DS), also known as Velo-Cardio-Facial syndrome, Shprintzen syndrome or DiGeorge syndrome, is a genomic disorder characterized by a recurrent microdeletion with an estimated prevalence of 1 in 2000 - 4000 live births. The project aims to unravel the genetic factors underlying the phenotypic variation in those patients. The international consortium will generate genome sequences from hundreds of patients.

The candidate will coordinate those efforts in Leuven and develop will develop novel tools to (1) mine the variation, (2) associate the variation with the phenotypic observations in the patient. This involves day-to-day collaboration with other postdoctoral and doctoral researchers who develop bioinformatic strategies for genetic variant detection and visualization based on massive parallel sequence analysis of genomic DNA extracted from blood. It also will require a close collaboration with clinical geneticists and other health workers to update and understand the phenotypic features of this patient population.

We offer a competitive package and a fun, dynamic environment with a top-notch consortium of young leading scientists in bioinformatics, human genetics, and cancer. Our consortium offers a rare level of interdisciplinarity, from machine learning algorithms to fundamental advances in molecular biology to direct access to the clinic. This research will be performed at the intersection of the Department of Human Genetics, the Department of Electrical Engineering and the Genomics Core Facility. You will be part of the Department of Human Genetics which is a vivid international research centre that studies key issues relevant to human health. The University of Leuven is one of Europe's leading research universities, with English as the working language for research. Leuven is one of Europe's most beautiful university towns, just outside Brussels, at the heart of Europe.

**Profile**

The ideal candidate holds a PhD degree in bioinformatics-genomics with a good understanding of genetics and DNA sequencing technology and has experience in molecular biology. The position necessitates analytical, algorithmic and mathematical skills. Programming and (statistical) data analysis experience is an asset. Prior experience working with (Illumina) sequencing data and/or assembly / alignment of next-generation data as well as a PhD relating to the

development and application of methods for genome-wide detection of genetic variation would be a distinct advantage, but is not required. Good communication skills are important for this role.

The candidate will collaborate closely with researchers across the consortium and contribute to the reporting of the project. Qualified candidates will be offered the opportunity to work semi-independently under the supervision of a senior investigator, mentor PhD students, and contribute to the acquisition of new funding. A three-year commitment is expected from the candidate.

### Related SymBioSys publications


Van der Aa N, Cheng J, Mateiu L, Esteki MZ, Kumar P, Dimitriadou E, Vanneste E, Moreau Y, Vermeesch JR, Voet T. Genome-wide copy number profiling of single cells in S-phase reveals DNA-replication domains. Nucleic Acids Res. 2013 Apr 1;41(6):e66.

Sifrim, A., Van Houdt, J., Tranchevent, L., Nowakowska, B., Sakai, R., Pavlopoulos GA, G., Devriendt, K., Vermeesch, J., Moreau, Y., Aerts, J. (2012). Interpretation of single nucleotide variation in human disease : a Swiss-knife approach to annotation and analysis. *Genome Medicine, 4*(9), art.nr. 73.

Van Houdt, J., Nowakowska, B., Sousa, S., van Schaik, B., Seuntjens, E., Avonce, N., Sifrim, A., Abdul-Rahman,O., van den Boogaard, M., Bottani, A., Castori, M., Cormier-Daire, V., Deardorff, M., Filges, I., Fryer, A., Fryns, J., Gana, S., Garavelli, L., Gillessen-Kaesbach, G., Hall, B., Horn, D., Huylebroeck, D., Klapecki, J., Krajewska-Walasek, M., Kuechler, A., Lines, M., Maas, S., Macdermot, K., McKee, S., Magee, A., de Man, S., Moreau, Y., Morice-Picard, F., Obersztyn, E., Pilch, J., Rosser, E., Shannon, N., Stolte-Dijkstra, I., Van Dijck, P., Vilain, C., Vogels, A., Wakeling, E., Wieczorek, D., Wilson, L., Zuffardi, O., van Kampen, A., Devriendt, K., Hennekam, R., **Vermeesch, J**. (2012). Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. ***Nature Genetics***2012 Feb 26;44(4):445-9

Vanneste, E., Voet, T., Le Caignec, C., Ampe, M., Konings, P., Melotte, C., Debrock, S., Amyere, M., Vikkula,M., Schuit, F., Fryns, J., Verbeke, G., D'Hooghe, T., Moreau, Y., **Vermeesch, J.** (2009). Chromosome instability is common in human cleavage-stage embryos. ***Nature Medicine****, 15*(5), 577-583


### Application

Please send in PDF: (1) a CV including education (with Grade Point Average, class rank, honors, etc.), research experience, and bibliography, (2) a one-page research statement, and (3) three references (with phone and email) to Joris.Vermeesch@med.kuleuven.be, Cc Yves. Moreau@med.kuleuven.be and Ann.Swillen@uzleuven.be -. Start date is as early as practical.