Marco Roos

Group leader and senior scientist biosemantics

Summary

Specialties: Interdisciplinary research, e-Science, molecular cell biology, Linked Data and ontologies, computational research workflows

My research focus is on making state-of-the-art computer science applicable to enhance biomedical research, particularly techniques for knowledge discovery and linked data to enhance research on rare human genetic diseases. My biological interests are the role of the genome and chromatin in the functioning of the cell and translational research, especially in the domain of rare diseases. After including computer science in my MSc in molecular biology, I have worked as a multidisciplinary researcher in research groups in life science and computer science. Now, as group leader of the Biosemantics research group of prof. Dr. Barend Mons, LUMC, I lead the research, development and application of knowledge discovery methods for human genetics research. As co-lead of the rare disease use case of the European life science data infrastructure Elixir, an important aspect of my work is advocating principles of FAIR and Linked Data to create a powerful substrate and world-wide robust infrastructure for knowledge discovery across distributed heterogeneous data resources.

Experience

Group leader and Senior research scientist LUMC BioSemantics group at Leiden University Medical Center

2009 - Present (7 years)

The LUMC biosemantics group develops and applies knowledge discovery methods to elucidate the mechanisms governing disease. The group endorses a multidisciplinary e-Science approach, involving workflows for reproducible experiments and Linked Data and text mining to create substrates for knowledge discovery.

Co-lead work package rare disease use case at ELIXIR

September 2015 - Present (1 year 2 months)

Co-lead together with Ivo Gut on the rare disease use case in Elixir, the pan-European infrastructure for life science data. Task leader for standards for data stewardship of rare disease data.

Chair rare disease linked data and ontology task force at multiple projects

November 2013 - Present (3 years)

The rare disease linked data and ontology task force is a knowledge hub for linked data and ontology experts with a special interest in rare diseases and rare disease experts with special interest in interoperability. It fosters training and software development activities and supports knowledge exchange between technical

experts and knowledge transfer to rare disease experts. It has members from institutes in the EU, USA and Australia and is supported by several projects such as RD-Connect, Elixir, and BBMRI. It was born out of the EU project RD-Connect that aims to connect rare disease data resources (omics sources, biobanks, registries).

Active member Netherlands BioInformatics Centre (NBIC) at NBIC

2007 - 2014 (7 years)

Topics of interest: e-Science, computational workflows, life science information management, data integration, chromatin research / epigenetics

Bioinformatician (PhD) at Adaptive Information Disclosure (AID), Institute of Informatics, University of Amsterdam

January 2006 - 2009 (3 years)

Co-develop new methods for computational biology based on emerging e-science technology.

Bioinformatician (post-doc and sub-project leader) at University of Amsterdam, Integrative Bioinformatics Unit

August 2003 - December 2006 (3 years 5 months)

Involved in setting up the Integrative Bioinformatics Unit (IBU), and in establishing interdisciplinary research for bioinformatics based on new approaches emerging from e-science.

Bioinformatician (post-doc) at Human Genetics Department, Academic Medical Hospital, University of Amsterdam

December 2001 - July 2003 (1 year 8 months)

Part of team responsible for the development of the Human Transcriptome Map, statistical methods for its analysis, especially for the discovery of 'Regions of IncreaseD Gene Expression' (RIDGEs), and correlations with other genome features (Versteeg et al, Genome Research, 2003).

PhD student Molecular Cytology at Molecular Cytology Department, University of Amsterdam

September 1996 - July 2001 (4 years 11 months)

Investigate the architecture and movement of chromatin (DNA+associated proteins) during the cell cycle by fluorescent (confocal) microscopy. First in eukaryotic nuclei of Indian Muntjac cells (unsuccesful), then in the bacterium Escherichia coli (thesis).

Replacement military service at Radiobiology, Academic Medical Hospital, Unitversity of Amsterdam

October 1995 - October 1996 (1 year 1 month)

Contribute to research on discriminating female and male producing bull sperm cells. Develop image processing method to measure DNA content from images of fluorescently labelled DNA in bull sperm cells.

Education

University of Amsterdam

Master's Degree, Cell/Cellular and Molecular Biology, 1988 - 1995

Interests

Molecular Cytology, Chromatin research, Biology, computational biology, bioinformatics, e-science, Semantic Web, Linked Data, Interdisciplinary research

Skills & Expertise

Linked Data
Semantic Web
Data Mining
Data Analysis
Data Modeling
Data Management
Workflow Management
Epigenetics
Chromatin
Molecular & Cellular Biology
Rare Diseases
Semantic Interoperability
Bioinformatics
Molecular Biology
Computational Biology
Fluorescence
Cell
DNA
Life Sciences
Genetics
Biology
Biomarker Discovery
Genomics
Image Processing
Confocal Microscopy
Science

Languages

English	(Full professional proficiency)
Dutch	(Native or bilingual proficiency)
German	(Limited working proficiency)

Projects

RD-Connect
Members:Marco Roos

Chair of the cross-project rare disease linked data and ontology task force.

Elixir

Members:Marco Roos

EpiGeniusHD

Members:Marco Roos

Supervisor PhD student Eleni Mina and main applicant of this project, originally supported by NBIC. The project is to investigate the role of epigenetics in HD using computational workflows and a multidisciplinary approach. A key element is the close collaboration with the HD research group of Willeke van Roon-Mom. With this work we contribute to the CHDI Genetic Modifier group, and the Rare Disease Connect project (http://www.rd-connect.eu). Eleni's project is also a paradigm for related projects that I co-supervise, such as on Polycystic Kidney Disease and Metabolic Syndrome.

OpenPHACTS

Members:Marco Roos

Publications

List of publications

Authors: Marco Roos
