

Núria Queralt Rosinach

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Research interests

Biomedical Informatics; Knowledge Representation; **Ontologies**; Linked Data; **Hypothesis Generation**; **Artificial Intelligence (AI)**, **Explainable AI**; BioNLP; **Genotype-Phenotype-Environment**; Rare, Complex and Infectious Diseases; Data/Knowledge Integration; Network Medicine; Translational Informatics; Precision Medicine; Distributed Analysis; **Federated Machine Learning**; FAIR data, FAIR services, and **FAIR analytics**

Terms highlighted in bold are my main research interests.

Education

- 2006–2008 Pompeu Fabra University / Barcelona University, *Barcelona*, Spain
- M.Sc. in Bioinformatics for Health Sciences
 - Thesis title: *The role of a cation- π interaction in the agonist conformation of estrogen-like nuclear receptors Super-family*
 - Advisor: Dr. Jordi Mestres
- 2002–2006 Rovira i Virgili University, *Tarragona*, Spain
- PhD in Quantum Chemistry
 - Dissertation title (Defense May 2010): *Study of the magnetic coupling in heterometallic complexes with oxamido, oxamato, tiooxalato and analog bridged ligands*
 - Advisors: Prof. Dr. Rosa Caballol Lorenzo and Dr. Coen de Graaf
- 1995–2001 Rovira i Virgili University, *Tarragona*, Spain
- Diplom (B.Sc.) in Chemistry
 - Thesis title: *Ab initio CASSCF and CASPT2 study of the photochemistry of bicyclo[3.1.0]hex-3-en-2-one*
 - Advisor: Dr. Mar Reguero

Professional experience

- 2020–present **Postdoctoral researcher** *Department of Human Genetics, Leiden University Medical Centre (LUMC), Leiden (NL). Biosemantics Group.*
- ✓ Led grant proposal for explainable AI between Twente University and LUMC, co-led the ELIXIR Rare Diseases 2022/23 Implementation Study proposal, help with other grant proposals in the group
 - ✓ Led and achieved EHDEN Data Partner funding for the European Rare Kidney Disease Registry

- ✓ Supervised three MSc students on: 'Knowledge graphs and explainable AI for Duchenne Muscular Dystrophy drug repurposing', 'Automated drug repurposing workflow for rare diseases', and 'Huntington disease structured review for hypothesis generation', and help the supervision of one MSc student on 'Federated Learning on personal health data' and one PhD candidate on 'Conceptual goal modelling for FAIRification'
 - ✓ Published research in peer-reviewed and pre-print journals as first author, and contributed in several scientific publications and in project deliverables
 - ✓ Gave talks in six international conferences, MSc Murcia University and Turin University programmes, the Swiss Personalized Health Network initiative seminar series, and LUMC FAIRification seminars
 - ✓ Led the ELIXIR Machine Learning Synthetic Data focus group and generated clinical synthetic data for COVID-19 studies in TWOC
 - ✓ Developed the COVID-19 epidemiological and monitoring ontology, and the LUMC Clinical Ontology
 - ✓ Led interoperability project between OMOP CDM and CDE semantic models for rare disease research analytics
 - ✓ Led the semantic modelling of multi-modal research data for Duchenne Muscular Dystrophy and for COVID-19, and involved in GA4GH Phenopackets, Semantic Phenopackets, rare disease CDE semantic and consent modelling
 - ✓ Led FAIRification of multimodal research data and graph-based analytics for rare diseases BIND and EJP RD projects
 - ✓ Led the development of the Rare Disease specific FAIR Maturity Indicators
 - ✓ Led FAIRification of COVID-19 clinical research data at the LUMC for federated analytics
 - ✓ Led six hackathon projects: 1. 'Infrastructure for Synthetic Health Data' 2022 BioHackathon Europe; 2. 'RD-specific FAIR Maturity Indicators' 2022 Swatathon; 3. 'Mapping GA4GH Phenopackets and OHDSI OMOP for COVID-19 disease epidemics and federated analytics' 2021 BioHackathon Europe; 4. 'Exploring grlc and Salad to align Web APIs with Linked Data' and 'Adding logical structure to the COVID-19 epidemiology ontology' 2021 Swatathon; 5. 'The COVID-19 epidemiology ontology' 2020 BioHackathon Europe; 6. 'Ontology-based approaches for the COVID-19 pandemic' 2020 COVID-19 BioHackathon
 - ✓ Organized two workshops: 'The GA4GH Phenopackets' for EJP RD 2021 sessions; 'FAIR Data Analytics' for ELIXIR All hands 2022
- 2019–2020 **Research associate** *Department of Biomedical Informatics, Harvard Medical School, Boston (USA). Avillach Lab.*
- ✓ Implemented data access Jupyter notebook examples in Python and R using the Patient-centered Information Commons-Standardized Unification of Research Elements (PIC-SURE) API for projects such as the NIH Undiagnosed Diseases Network (UDN) and the NHLBI BioData Catalyst
 - ✓ Developed a data characterization of the UDN data resource, a patient registry of phenotypic and genomic information
 - ✓ Participated in the evaluation of a new serverless service in the cloud for biomedical research on large-scale data sets
- 2019–2019 **Visiting researcher** *Computational Bioscience Research Center, King Abdullah University of Science and Technology (KAUST), Thuwal (KSA). The Bio-Ontology Research Group.*
- ✓ Implemented a knowledge graph and a symbolic AI method for drug repositioning on metabolic rare diseases

- ✓Developed ontology-based text mined metabolite-phenotype data sets for human and plants
- ✓Attended logic club seminars on the Homotopy type theory
- 2016–2019 **Research associate** *Department of Integrative Structural and Computational Biology, Scripps Research (SR), La Jolla (USA). The Su Lab.*
 - ✓Developed a drug repositioning method for rare diseases
 - ✓Implemented the NGLY1 Deficiency knowledge graph for evidence-based biomedical hypothesis discovery and a Python library for its creation, use and management
 - ✓Implemented a Wikibase instance for community contribution to the NGLY1 Deficiency knowledge graph
 - ✓Created a provenance model for biomedical information in Wikidata
- 2012–2016 **Research fellow** *Research Programme on Biomedical Informatics (GRIB), Hospital del Mar Medical Research Institute (IMIM), Pompeu Fabra University (UPF), Barcelona (Spain). Integrative Biomedical Informatics Group.*
 - ✓Implemented the DisGeNET-RDF linked dataset and its accessibility and exploration through a SPARQL endpoint, Faceted Browser, and for download as a dump via a well-detailed Web interface
 - ✓Developed the conversion of DisGeNET to RDF and nanopublication formats for the Open PHACTS project
 - ✓Developed a Disease Biomarker database and its Web interface for the eTOX project
- 2010–2016 **Biostatistics consultant** *Open University of Catalonia, Barcelona (Spain). Biostatistics: fundamentals and applications Post-graduate.*
 - ✓Final project consultant-manager
- 2010–2012 **Bioinformatics technician fellow** *Institut d'Investigacions Biomèdiques August Pi i Sunyer-IDIBAPS, Barcelona (Spain). Bioinformatics Platform.*
 - ✓Developed high-level analysis, interpretation and report of metabolomics and transcriptomics data
 - ✓Developed new bioinformatic tools such as a database powered by the BioMart Technology, and set up a mirror of the UCSC Genome Browser
 - ✓Executed the maintenance and update of in-house web-based tools and external licenses
 - ✓Developed informatic support administrating the Linux Bioinformatics servers of the platform
- 2009–2010 **Bioinformatics technician** *Rovira i Virgili University, Tarragona (Spain). Bioinformatics Group.*
 - ✓Assisted in the study of the possible epigenetic regulation and imprinting of human genes by means of diet elements
 - ✓Implemented a pipeline of manifold software to treat and analyse genomic data
- 2007–2008 **MSc researcher** *Pompeu Fabra University, Barcelona (Spain). Systems Pharmacology Group.*
 - ✓Managed the *Masters thesis on 3D structure conservation in the LBD domain of Nuclear Receptor Super-family*
 - ✓Organized and developed a research study using bioinformatic resources
- 2002–2006 **PhD researcher** *Rovira i Virgili University, Tarragona (Spain). Quantum Chemistry Group.*
 - ✓Involved in a leading research group and trained in doing research

- ✓Taught undergraduated in Fortran programming and wet lab training in chemical-physics experiments
- 2001–2002 **Teacher Assistant** *Rovira i Virgili University*, Tarragona (Spain). Physical and Inorganic Chemistry Department.

Honors and awards

- 2010 ISCIH/MICINN Fellowship, *ISCIH/MICINN Instituto de Salud Carlos III/Ministerio de Ciencia e Innovación*, Madrid, Spain
- 2006 Tarraco Empresa Joven Entrepreneurship Prize, *2nd Prize*, AIJEC Associació Independent de Joves Empresaris de Catalunya, Tarragona, Spain
- 2004 Reus Creació Empreses Entrepreneurship Prize, *3rd Prize*, Ajuntament de Reus-REDESSA Reus Desenvolupament Econòmic SA/Rovira i Virgili University, Reus, Spain
- 2002-2006 URV Graduate Fellowship, *Rovira i Virgili University*, Tarragona, Spain

Researcher identifiers

- ORCID: 0000-0003-0169-8159
- ResearchID: B-7613-2016

Publications

Under review

- [1] C. H. Bernarbé, **N. Queralt-Rosinach**, V. Souza, L. Santos, A. Jacobsen, B. Mons, M. Roos, *The use of Foundational Ontologies in Bioinformatics*, *Research Square*, 2022

Peer-reviewed journal articles

- [1] N. Matentzoglou, J. P. Balhoff, S. M. Bello, C. Bizon, M. Brush, T. J. Callahan, C. G. Chute, W. D. Duncan, C. T. Evelo, D. Gabriel, J. Graybeal, A. Gray, B. M. Gyori, M. Haendel, H. Harmse, N. L. Harris, I. Harrow, H. B. Hegde, A. L Hoyt, C. T. Hoyt, D. Jiao, E. Jiménez-Ruiz, S. Jupp, H. Kim, S. Koehler, T. Liener, Q. Long, J. Malone, J. A. McLaughlin, J. A. McMurry, S. Moxon, M. C. Munoz-Torres, D. Osumi-Sutherland, J. A. Overton, B. Peters, T. Putman, **N. Queralt-Rosinach**, K. Shefchek, H. Solbrig, A. Thessen, T. Tudorache, N. Vasilevsky, A. H. Wagner, C. J. Mungall, *A Simple Standard for Sharing Ontological Mappings (SSSOM)*, *Database*, 2022, Volume 2022, baac035
- [2] J. O. B. Jacobsen, M. Baudis, G. S. Baynam, J. S. Beckmann, S. Beltran, O. J. Buske, T. J. Callahan, C. G. Chute, M. Courtot, D. Danis, O. Elemento, A. Essenwanger, R. R. Freimuth, M. A. Gargano, T. Groza, A. Hamosh, N. L. Harris, R. Kaliyaperumal, K. C. Kent Lloyd, A. Khalifa, P. M. Krawitz, S. Köhler, B. J. Laraway, H. Lehvälaiho, L. Matalonga, J. A. McMurry, A. Metke-Jimenez, C. J. Mungall, M. C. Munoz-Torres, S. Ogishima, A. Papakonstantinou, Davide Piscia, Nikolas Pontikos, **N. Queralt-Rosinach**, Marco Roos, Julian Sass, Paul N. Schofield, D. Seelow, A. Siapos, D. Smedley, L. D. Smith, R. Steinhaus, J. Chandrabose Sundaramurthi, E. M. Swietlik, S. Thun, N. A. Vasilevsky, A. H. Wagner, J. L. Warner, C. Weiland, The GAGH Phenopacket Modeling Consortium, M. A. Haendel, P. N. Robinson, *The GA4GH Phenopacket schema defines a computable representation of clinical data*, *Nat Biotechnol*, 2022, 40, 817-820
- [3] M. Mayers, R. Tu, D. Steinecke, T. S. Li, **N. Queralt-Rosinach**, A. I. Su, *Design and application of a knowledge network for automatic prioritization of drug mechanisms*, *Bioinformatics*, 2022, 38, 10, 2880–2891

- [4] **N. Queralt-Rosinach**, R. Kaliyaperumal, C. H. Bernabé, Q. Long, S. A. Joosten, H. J. van der Wijk, E. L. A. Flikkenschild, K. Burger, A. Jacobsen, B. Mons, M. Roos, BEAT-COVID Group, COVID-19 LUMC Group, *Applying the FAIR principles to data in a hospital: challenges and opportunities in a pandemic*, *J Biomed Semant*, 2022, 13, 12
- [5] R. Kaliyaperumal, M. Wilkinson, P. Alarcón Moreno, N. Benis, R. Cornet, B. dos Santos Vieira, M. Dumontier, C. H. Bernabé, A. Jacobsen, C. M. A. Le Cornec, M. Prieto Godoy, **N. Queralt-Rosinach**, L. J. Schultze Kool, M. A. Swertz, P. van Damme, J. K. van der Velde, N. van Lin, S. Zhang, M. Roos, *Semantic modelling of common data elements for rare disease registries, and a prototype workflow for their deployment over registry data*, *J Biomed Semant*, 2022, 13, 9
- [6] A. Yamaguchi and **N. Queralt-Rosinach**, *A proof-of-concept study of extracting patient histories for rare/intractable diseases from social media*, *Genomics & Informatics*, 2020, 18(2):e17
- [7] A. Waagmeester, G. Stupp, S. Burgstaller-Muehlbacher, B. M. Good, M. Griffith, O. Griffith, K. Hanspers, H. Hermjakob, K. Hybiske, S. M. Keating, M. Manske, M. Mayers, E. Mitraka, A. R. Pico, T. Putman, A. Riutta, **N. Queralt-Rosinach**, L. M. Schriml, D. Slenter, G. Tsueng, R. Tu, E. Willighagen, C. Wu, A. I. Su, *Wikidata as a FAIR knowledge graph for the life sciences*, *eLife*, 2020, 9:e52614
- [8] **N. Queralt-Rosinach**, G.S. Stupp, T.S. Li, M.D. Mayers, M. E. Hoatlin, M. Might, B.M. Good, A.I. Su, *Structured Reviews for Data and Knowledge Driven Research*, *Database*, 2020, Volume 2020, baaa015
- [9] R. A. Vos, T. Katayama, H. Mishima, S. Kawano, S. Kawashima, J.-D. Kim, Y. Moriya, T. Tokimatsu, A. Yamaguchi, Y. Yamamoto, H. Wu, P. Amstutz, E. Antezana, N. Aoki, K. Arakawa, J. Bolleman, E. Bolton, R. J. P. Bonnal, H. Bono, K. Burger, H. Chiba, K. Cohen, E. Deutsch, J. Fernández-Breis, G. Fu, T. Fujisawa, A. Fukushima, A. García, N. Goto, T. Groza, C. Hercus, R. Hoehndorf, K. Itaya, N. Juty, T. Kawashima, J.-H. Kim, A. Kinjo, M. Kotera, K. Kozaki, S. Kumagai, T. Kushida, T. Lütteke, M. Matsubara, J. Miyamoto, A. Mohsen, H. Mori, Y. Naito, T. Nakazato, J. Nguyen-Xuan, K. Nishida, N. Nishida, H. Nishide, S. Ogishima, T. Ohta, S. Okuda, B. Paten, J.-L. Perret, P. Prathipati, P. Prins, **N. Queralt-Rosinach**, D. Shinmachi, S. Suzuki, T. Tabata, T. Takatsuki, K. Taylor, M. Thompson, I. Uchiyama, B. Vieira, C.-H. Wei, M. Wilkinson, I. Yamada, R. Yamanaka, K. Yoshitake, A. C. Yoshizawa, M. Dumontier, K. Kosaki, T. Takagi, *BioHackathon 2015: Semantics of data for life sciences and reproducible research*, *F1000Research*, 2020, 9, 136
- [10] M.D. Mayers, T.S. Li, **N. Queralt-Rosinach**, A.I. Su, *Time-resolved evaluation of compound repositioning predictions on a text-mined knowledge network*, *BMC Bioinformatics*, 2019, 20, 653
- [11] J.L. Oliveira, P. Sernadela, L. González-Castro, C. Carta, E.v.d. Horst, P. Lopes, R. Kaliyaperumal, R. Thompson, **N. Queralt-Rosinach**, E. Lopez, L. Wood, C. Lamanna, M. Gilling, M. Orth, R. Martinez, M. Posada, D. Taruscio, P. Robinson, M. Roos, A. Robertson, *Linked Registries: connecting rare diseases patient registries through a semantic web layer*, *BioMed Research International*, 2017, vol. 2017, 8327980
- [12] R. Hoehndorf and **N. Queralt-Rosinach**, *Data Science and Symbolic AI: Synergies, challenges and opportunities*, *Data Science*, 2017, 1, 27–38
- [13] M. Alshahrani, M. A. Khan, O. Maddouri, A. R. Kinjo, **N. Queralt-Rosinach**, R. Hoehndorf, *Neuro-symbolic representation learning on biological knowledge graphs*, *Bioinformatics*, 2017, 33, 2723–2730

- [14] J. Piñero, À. Bravo, **N. Queralt-Rosinach**, A. Gutiérrez-Sacristán, J. Deu-Pons, E. Centeno, J. García-García, F. Sanz, L. I. Furlong, *DisGeNET: a comprehensive platform integrating information on human disease-associated genes and variants*, *Nucleic Acids Research*, 2017, 45, D833–D839
- [15] T. Kuhn, C. Chichester, M. Krauthammer, **N. Queralt-Rosinach**, R. Verborgh, G. Giannakopoulos, A-C. N. Ngomo, R. Viglianti, M. Dumontier, *Decentralized Provenance-Aware Publishing with Nanopublications*, *PeerJ Computer Science*, 2016, 2, e78
- [16] D. Digles, B. Zdrzil, J.-M. Neefs, H. Van Vlijmen, C. Herhaus, A. Caracoti, J. Brea, B. Roibás, M. Loza, **N. Queralt-Rosinach**, L. I. Furlong, A. Gaulton, L. Bartek, S. Senger, C. Chichester, O. Engkvist, C. Evelo, G. F. Ecker and E. Jacoby, *Open PHACTS Computational Protocols for in silico Target Validation of Cellular Phenotypic Screens: Knowing the Knowns*, *Med. Chem. Commun*, 2016, 7, 1237–1244
- [17] **N. Queralt-Rosinach**, J. Piñero, À. Bravo, F. Sanz, L.I. Furlong, *DisGeNET-RDF: Harnessing the Innovative Power of the Semantic Web to Explore the Genetic Basis of Diseases*, *Bioinformatics*, 2016, 32, 2236–2238
- [18] **N. Queralt-Rosinach**, T. Kuhn, C. Chichester, M. Dumontier, F. Sanz, L.I. Furlong, *Publishing DisGeNET as Nanopublications*, *Semantic Web*, 2016, 7, 519–528
- [19] J. Piñero, **N. Queralt-Rosinach**, À. Bravo, J. Deu-Pons, A. Bauer-Mehren, M. Baron, F. Sanz, L.I. Furlong, *DisGeNET: a discovery platform for the dynamical exploration of human diseases and their genes*, *Database (Oxford)*, 2015, 2015, bav028
- [20] À. Bravo, J. Piñero, **N. Queralt-Rosinach**, M. Rautschka, L.I. Furlong, *Extraction of relations between genes and diseases from text and large-scale data analysis: implications for translational research*, *BMC Bioinformatics*, 2015, 16, 55
- [21] À. Bravo, M. Cases, **N. Queralt-Rosinach**, F. Sanz, L.I. Furlong, *A knowledge-driven approach to extract disease-related biomarkers from the literature*, *BioMed Research International*, 2014, 2014, 253128
- [22] M. Dumontier, C.J.O. Baker, J. Baran, A. Callahan, L. Chepelev, J. Cruz-Toledo, N.R. del Rio, G. Duck, L.I. Furlong, N. Keath, D. Klassen, J.P. McCusker, **N. Queralt-Rosinach**, M. Samwald, N. Villanueva-Rosales, M.D. Wilkinson, R. Hoehndorf, *The SemanticScience Integrated Ontology (SIO) for biomedical research and knowledge discovery*, *Journal of Biomedical Semantics*, 2014, 5, 1–14
- [23] I. Negodaev, **N. Queralt-Rosinach**, R. Caballol, C. de Graaf, *Theoretical study of the magnetic exchange interaction in catena- μ -Tris[oxalato⁽²⁻⁾-O¹,O²;O³,O⁴]-dicopper complex with interlocked helical chains*, *Chemical Physics*, 2011, 379, 109–115
- [24] **N. Queralt-Rosinach**, J. Mestres, *A canonical cation- π interaction stabilizes the agonist conformation of estrogen-like nuclear receptors*, *European Biophysics Journal*, 2010, 39, 1471–5
- [25] **N. Queralt**, D. Taratiel, C. de Graaf, R. Caballol, R. Cimraglia, C. Angeli, *On the applicability of multireference second-order perturbation theory to study weak magnetic coupling in molecular complexes*, *Journal of Computational Chemistry*, 2008, 29, 994–1003
- [26] **N. Queralt**, C. de Graaf, J. Cabrero, R. Caballol, *Ferrimagnetic coupling in oxamido-bridged Mn(II)Cu(II) compounds: a combined CASPT2 and DDCI study*, *Molecular Physics*, 2003, 101, 2095–2102

Conference presentations

- [1] P. Perdomo-Quinteiro, K. Wolstencroft, M. Roos, **N. Queralt-Rosinach**, *Knowledge graphs and Explainable AI for Drug Repurposing on Rare Diseases, 13th International Conference on Biomedical Ontology (ICBO) Role of Ontologies in Biomedical AI workshop. Ann Arbor MI USA, 25-28 September 2022, 2022*
Talk
- [2] **N. Queralt-Rosinach**, R. Kaliyaperumal, V. Emonet, M. Wilkinson, M. Hanauer, M. Roos, *Rare disease specific FAIR Maturity Indicators, 21st European Conference on Computational Biology (ECCB). Sitges Barcelona, 12-21 September 2022, 2022*
Talk
- [3] N. Benis, R. de Groot, P. Alarcón, R. Kaliyaperumal, M. Roos, R. Cornet, **N. Queralt-Rosinach**, *EJP RD meets OHDSI: enabling interoperability for rare disease research, 30th Conference on Intelligence Systems for Molecular Biology (ISMB) Bio-Ontologies COSI. Madison WI USA, 10-14 July 2022, 2022*
Talk
- [4] R. de Groot, N. Benis, P. Alarcón, R. Kaliyaperumal, M. Roos, R. Cornet, **N. Queralt-Rosinach**, *OMOP CDM for European rare disease registries, 2022 OHDSI European Symposium. Rotterdam The Netherlands, 24-26 June 2022, 2022*
Poster
- [5] R. Kaliyaperumal, G. Singh, **N. Queralt-Rosinach**, J. Bayjanov, P. B. T. Hoen and M. Roos, *Phenopackets for the Semantic Web, 13th International Conference on Semantic Web Applications and Tools for Health Care and Life Sciences (SWAT4HCLS). Virtual, Leiden The Netherlands, 10-14 January 2022, 2022*
Talk
- [6] C. H. Bernabé, **N. Queralt-Rosinach**, V. E. Silva Souza, L. O. Bonino da Silva Santos, A. Jacobsen, B. Mons, M. Roos, *The use of Foundational Ontologies in Bioinformatics, 13th International Conference on Semantic Web Applications and Tools for Health Care and Life Sciences (SWAT4HCLS). Virtual, Leiden The Netherlands, 10-14 January 2022, 2022*
Talk
- [7] **N. Queralt-Rosinach**, C. H. Bernabé, Q. Long, R. Kaliyaperumal, M. Roos, *LUMC Clinical Ontology for Biomedical Research, 12th International Conference on Biomedical Ontology (ICBO). Bozen-Bolzano Italy, 15-18 September 2021, 2021*
Talk
- [8] **N. Queralt-Rosinach**, M. Wilkinson, R. Kaliyaperumal, C. H. Bernabé, Q. Long, M. Dumontier, P. N. Schofield, M. Roos, *Reuse of Design Pattern Measurements for Health Data, 12th International Conference on Formal Ontology in Information Systems (FOIS). Bozen-Bolzano Italy, 13-17 September 2021, 2021*
Demonstration
- [9] C. H. Bernabé, A. Jacobsen, **N. Queralt-Rosinach**, L. O. Bonino da Silva Santos, V. E. Silva Souza, B. Mons, M. Roos, *Goal-models to support communication, planning and guiding of FAIRification, 29th Conference on Intelligence Systems for Molecular Biology (ISMB) Bio-Ontologies COSI/20th European Conference on Computational Biology (ECCB). Virtual, 25-30 July 2021, 2021*
Poster

- [10] **N. Queralt-Rosinach**, P. N. Schofield, R. Hoehndorf, C. Weiland, E. Schultes, C. H. Bernabé, M. Roos, *COVID-19 epidemiology and monitoring ontology, 29th Conference on Intelligence Systems for Molecular Biology (ISMB) Bio-Ontologies COSI/20th European Conference on Computational Biology (ECCB). Virtual, 25-30 July 2021, 2021*
Talk
- [11] C. H. Bernabé, A. Jacobsen, **N. Queralt-Rosinach**, L. O. Bonino da Silva Santos, V. E. Silva Souza, B. Mons, M. Roos, *Goal-models to support communication, planning and guiding of FAIRification, Bioinformatics & Systems Biology Conference (BioSB). Virtual, 15-16 June 2021, 2021*
Talk
- [12] R. Kaliyaperumal, **N. Queralt-Rosinach**, K. Burger, L. O. Bonino da Silva Santos, M. Hanauer, M. Roos, *Enabling FAIR discovery of Rare Disease digital resources, dHealth Conference. Virtual, 11 -12 May, 2021, 2021*
Talk
- [13] **N. Queralt-Rosinach**, R. Kaliyaperumal, C. Bernabé, Q. Long, H. J. van der Wijk, B. Mons, M. Roos, *FAIR Data Management to Access Patient Data, The 19th International Semantic Web Conference (ISWC) DaMaLOS workshop. Virtual, 1-6 November, 2020, 2020*
Talk
- [14] **N. Queralt-Rosinach**, S. M. Bello, R. Hoehndorf, C. Weiland, P. Rocca-Serra, P. N. Schofield, *Modeling quantitative traits for COVID-19 case reports, Intelligent Systems for Molecular Biology (ISMB) Bio-Ontologies COSI. Virtual, 15 -16 July, 2020, 2020*
Talk and Poster
- [15] T. Kuhn, A. Meroño-Peñuela, A. Malic, J. H. Poelen, A. H. Hurlbert, E. Centeno Ortiz, L. I. Furlong, **N. Queralt-Rosinach**, C. Chichester, J. M. Banda, E. Willighagen, F. Ehrhart, C. Evelo, T. B. Malas, and M. Dumontier, *Nanopublications: A Growing Resource of Heterogeneous Provenance-Centric Linked Data, eScience IEEE International Conference. Amsterdam, The Netherlands, 29 October - 1 November, 2018, 2018*
Talk
- [16] T. Kuhn, E. Willighagen, C. Evelo, **N. Queralt-Rosinach**, E. Centeno, L. I. Furlong, *Reliable Granular References to Changing Linked Data, 16th International Semantic Web Conference, Vienna, Austria, October 21–25, 2017 (ISWC2017), 2017*
Talk
- [17] A. Waagmeester, E. Willighagen, **N. Queralt-Rosinach**, E. Mitraka, S. Burgstaller-Muehlbacher, T. E. Putman, J. Turner, L. M. Schriml, P. Pavlidis, A. I. Su, B. M. Good, *Linking Wikidata to the rest of the Semantic Web, M. Scott Marshall, A. Waagmeester, F. Francissen, 9th International SWAT4LS Conference. Semantic Web Applications and Tools for Life Sciences. Amsterdam, NL, December 5-8th, 2016, 2016*
Talk
- [18] G. Fu, E. Bolton, **N. Queralt-Rosinach**, L.I. Furlong, V. Nguyen, A. Sheth, O. Bodenreider, M. Dumontier, *Exposing Provenance Metadata Using Different RDF Models, A. Splendiani, SWAT4LS International Conference. Semantic Web Applications and Tools for Life Sciences. Cambridge, UK, December 7-10th, 2015, 2015*
Talk

- [19] **N. Queralt-Rosinach** and L.I. Furlong, *DisGeNET RDF: A Gene-Disease Association Linked Open Data Resource*, Adrian Paschke, Albert Burger, Paolo Romano, M. Scott Marshall, Andrea Splendiani, *SWAT4LS 2013. Semantic Web Applications and Tools for Life Sciences*. Edinburgh, UK, December 10, CEUR Workshop Proceedings. Volume 1114, 2013, 2013

Poster

- [20] **N. Queralt-Rosinach** and L.I. Furlong, *DisGeNET: from MySQL to Nanopublication, modelling Gene-Disease Associations for the Semantic Web*, Adrian Paschke, Albert Burger, Paolo Romano, M. Scott Marshall, Andrea Splendiani, *SWAT4LS 2012. Semantic Web Applications and Tools for Life Sciences*. Paris, France, November 28, CEUR Workshop Proceedings. Volume 952, 2012, 2012

Poster

Tutorials

- 2015 DisGeNET: a discovery platform for translational bioinformatics, *Semantic Web Applications and Tools for Life Sciences International Conference (SWAT4LS)*
- 2015 DisGeNET: a discovery platform for translational bioinformatics, *Swiss Institute of Bioinformatics*, Bioinformatics course

PHD THESIS

- 2010 **N. Queralt-Rosinach**, *Study of the magnetic coupling in hetero-metallic complexes with oxamido, oxamato, tiooxalato and analog bridged ligands*, Rovira i Virgili University, May 2010

Software and data resources

- 2022 **The Duchenne and Becker Muscular Dystrophy Structured Review**, *Duchenne Muscular Dystrophy and research question specific knowledge graph and Web interface for hypothesis generation*, <http://167.71.4.209:7475/browser/>
- 2022 **Rare Diseases specific FAIR Maturity Indicators**, *Rare diseases domain specific FAIR Maturity Indicators to guide FAIRification*, <https://github.com/NuriaQueralt/RD-FAIRmetrics>
- 2021 **COVID-19 Epidemiology and Monitoring Ontology**, *OBO ontology for epidemiology and monitoring the COVID-19 outbreak*, <https://github.com/NuriaQueralt/covid19-epidemiology-ontology>
- 2021 **LUMC Clinical Ontology**, *Ontology for biomedical research in the LUMC academic hospital*, <https://fairsharing.org/bsg-s001616/>
- 2020 **COVID-19 synthetic patient cytokine Knowledge Graph**, *COVID-19 synthetic patient data RDF graph for biomedical research*, <http://178.63.49.197:8081/>
- 2020 **Quantitative trait model**, *RDF model for measurements collected in health data research*, <https://github.com/NuriaQueralt/BioHackathon/tree/master/bh20-ontology-qt>
- 2019 **NGLY1 Deficiency Wikibase**, *Platform for NGLY1 Deficiency research community curation*, <http://ngly1graph.org/contribute/>
- 2018 **BioKnowledge reviewer Python Library**, *Software for knowledge graph generation and analysis*, <https://github.com/NuriaQueralt/bioknowledge-reviewer>
- 2018 **NGLY1 Deficiency Structured Review**, *NGLY1 Deficiency and research question specific knowledge graph and Web interface for hypothesis generation*, <http://ngly1graph.org/>

- 2017 **NGLY1 Deficiency Knowledge Graph**, *Disease Knowledge Network*, <https://github.com/NuriaQueralt/ngly1-graph>
- 2017 **NGLY1 Deficiency Structured Deep Phenotyping**, *curated HPO disease-phenotype association dataset*, https://github.com/NuriaQueralt/ngly1-graph/blob/master/curation/kylo/neo4j/networks/v20180118/lam_2016_curated_network-lam_2016_edges.tsv
- 2017 **Wikidata**, *CIViC model*
- 2016 **Wikidata**, *Provenance model*
- 2016 **disgenet2r**, *R package*, <https://bitbucket.org/albags/disgenet2r>
- 2015 **DisGeNET RDF**, *Linked Dataset*, <http://www.disgenet.org/rdf>
- 2015 **DisGeNET Nanopublications**, *Linked Dataset*, <http://www.disgenet.org/rdf#nanopubs>
- 2015 **DisGeNET Discovery platform**, *Platform with a Web interface, Cytoscape plugin, Linked Data analysis, and programmatic access*, <http://www.disgenet.org>
- 2015 **The BeFree System**, *Text-mining tool composed by a BioNER and RE modules*, <http://ibi.imim.es/befree/>
- 2014 **The SemanticScience Integrated Ontology**, *OWL ontology*, <https://github.com/MaastrichtU-IDS/semanticscience>
- 2014 **Disease-related biomarker database**, *Database and Web interface*, <http://ibi.imim.es/disease-related-biomarker-database/>

Computer skills

<i>Programming</i>	Python, RDF, OWL, SPARQL, CYPHER, MySQL, PHP, HTML, R, C, Perl, Fortran, UNIX shell scripting, Apache, Virtuoso, Neo4j and Wikibase server management.	<i>Development</i>	Git/GitHub, Jupyter Notebook, Anaconda, PyCharm, RStudio, Docker.
<i>Bioinformatics</i>	OWLAPI, Protegé, R&Bioconductor, IPA, Cytoscape, EMBOSS, RepeatMasker, ClustalW, MAMMOTH-mult, Pymol.	<i>Databases</i>	WikiData, Monarch, EBI-RDF, UMLS, UCSC, GEO, GO, Swissprot, PDB, NucleaRDB.
<i>Data science</i>	AWS SageMaker, PyKEEN, PyTorch, sklearn, Weka.	<i>Computational chemistry</i>	Software for quantum computation: MOLCAS and DDCI, and for molecular modelling: CERIUS and MOL-DEN.
<i>GitHub</i>	https://github.com/NuriaQueralt		

Invited conferences|workshops|hackathons

- 2022 BioHackathon-Europe, 7-11 November, Paris (France), <https://biohackathon-europe.org/>, <https://github.com/elixir-europe/biohackathon-projects-2022/tree/main/15>
- 2022 ICBO 2022 ROBI workshop, 25-28 September 2022, Ann Arbor (MI, US), <https://icbo-conference.github.io/icbo2022/>, <https://bio-ontology-research-group.github.io/icbo-2022-ml-workshop/>
- 2022 GA4GH 10th Plenary, 22-23 September 2022, <https://www.ga4gh.org/event/ga4gh-10th-plenary/>
- 2022 ECCB 2022, 12-21 September 2022, Sitges (Catalonia, Spain), <https://eccb2022.org/>

- 2022 The ISMB 2022 Bio-Ontologies COSI, 10-14 July 2022, Madison (WI, US), <https://www.iscb.org/ismb2022-program/abstracts/bio-ontologies>
- 2022 2022 OHDSI European Symposium, 24-26 June 2022, Rotterdam (NL), <https://www.ohdsi.org/2022-european-symposium/>
- 2022 Invited talk, FAIR Training Workshop, Murcia University, 15 June 2022, virtual
- 2022 ELIXIR All hands 2022, 7-10 June 2022, Amsterdam (NL), <https://elixir-europe.org/events/elixir-all-hands-2022>
- 2022 ELIXIR All hands 2022 FAIR Data Analytics workshop, 7-10 June 2022, Amsterdam (The Netherlands), <https://elixir-europe.org/events/elixir-all-hands-2022>
- 2022 Invited talk, MSc Bioinformatics programme, Murcia University, 24 March 2022, virtual
- 2022 SWAT4HCLS 2022 hackathon, 13 January 2022, virtual, https://hackmd.io/hPchZveQTcar9_wkuSKi_w?view#Welcome-to-the-SWAT4HCLS-2022-hackathon
- 2022 SWAT4HCLS 2022, 10-13 January 2022, virtual, shorturl.at/BJSY1
- 2021 SPHN Webinar, 8 December 2021, virtual, <https://www.linkedin.com/company/swiss-personalized-health-network-sphn/>
- 2021 BioHackathon-Europe, 8-12 November 2021, Barcelona (Catalonia, Spain), <https://biohackathon-europe.org/>, <https://github.com/elixir-europe/biohackathon-projects-2021/tree/main/projects/36>
- 2021 MyScienceWork 2021, September 2021, shorturl.at/bdkq9, <https://youtu.be/bdM3wcMhnfA>
- 2021 Open Science FAIR 2021, 20-23 September 2021, virtual, <https://www.opensciencefair.eu/>
- 2021 ICBO 2021, 15-18 September 2021, Bozen-Bolzano (Italy), <https://icbo2021.inf.unibz.it/program/>
- 2021 FOIS 2021, 13-17 September 2021, Bozen-Bolzano (Italy), <https://fois2021.inf.unibz.it/>
- 2021 The ISMB/ECCB 2021 Bio-Ontologies COSI, 25-30 July 2021, virtual, https://www.iscb.org/cms_addon/conferences/ismbeccb2021/tracks/bio-ontologies
- 2021 SWAT4HCLS virtual hackathon, 15 and 19 January, virtual, https://swat4hcls.wiki.opencura.com/wiki/Main_Page
- 2020 BioHackathon-Europe, 9-13 November, virtual, <https://biohackathon-europe.org/>, <https://github.com/elixir-europe/BioHackathon-projects-2020/tree/master/projects/30>
- 2020 ISWC 2020 DaMaLOS workshop, 1-6 November, virtual, <https://iswc2020.semanticweb.org/>, <https://zbmed.github.io/damalos/>
- 2020 The ELIXIR::GA4GH: Advancing genomics via expedited Data Access enabled by standards and ontologies workshop co-located with ECCB2020, 2 September, virtual, <https://eccb2020.info/ntbew02-elixirga4gh-advancing-genomics-through-expedited-data-access-enabled-by-standards-and-ontologies/>
- 2020 The ISMB 2020 Bio-Ontologies COSI, 15-16 July, virtual, https://www.iscb.org/cms_addon/conferences/ismb2020/tracks/bio-ontcosi
- 2020 The 10th European Conference on Rare Diseases & Orphan Products (ECRD), 14-15 May, virtual, <https://www.rare-diseases.eu/>
- 2020 BioHackathon 2020 COVID-19, 5-11 April, virtual, <https://github.com/virtual-biohackathons/covid-19-bh20/>

- 2020 Biomedical Linked Annotation Hackathon 6 (BLAH6), 4-7 February Tokyo Japan, <https://blah6.linkedannotation.org/>
- 2019 BioHackathon, 1-7 September Fukuoka, Japan, <http://2019.biohackathon.org/>
- 2018 BioHackathon, 9-15 December Matsue, Japan, <http://2018.biohackathon.org/>
- 2018 Cancer Variant Interpretation Hackathon and Curation Jamboree, 15-16 October San Diego, USA, <https://github.com/griffithlab/civic-meeting>
- 2018 NCATS Biomedical Data Translator Hackathon, 8-10 January San Diego, USA
- 2017 BioHackathon, 9-16 September Morioka, Japan, <http://2017.biohackathon.org/>
- 2017 NIH HeartBD2K meeting, 20-21 April La Jolla, USA, <http://www.heartbd2k.org/>
- 2017 10th International Biocuration Conference, 26-29 March Palo Alto, USA, <https://med.stanford.edu/biocuration.html>
- 2016 BioHackathon, 12-18 June Tsuruoka, Japan, <http://2016.biohackathon.org/>
- 2016 Linking Life Science Data: Design to Implement, 18-19 February Vienna, Austria, <http://www.openphactsfoundation.org/event-announcement-linking-life-science-data-design-to-implementation-and-beyond/>
- 2015 International SWAT4LS Conference, 7-10 December Cambridge, UK, <http://www.swat4ls.org/workshops/cambridge2015/>
- 2015 BioMedBridges Symposium: Open bridges for life science data, 17-18 November Hinxton, UK, <http://www.biomedbridges.eu/news/symposium-open-bridges-life-science-data>
- 2015 BioHackathon, 13-19 September Nagasaki, Japan, <http://2015.biohackathon.org/>
- 2015 Open PHACTS Workshop, 15-18 March Santiago de Compostela, Spain, <http://www.openphacts.org/news-and-events/conferences/2015>
- 2014 BYOD on Rare Disease Registries and Biobanks, 26-27 November Rome, Italy, <http://www.dtls.nl/fair-data/byod/byods/byod-on-rare-disease-registries-and-biobanks/>
- 2014 Network of BioThings 1st Hackathon, 26-27 April Maastricht, Netherlands, <http://projects.bigcat.unimaas.nl/hackaton-biothings/>
- 2013 International SWAT4LS Conference, 9-12 December Edinburgh, UK, <http://www.swat4ls.org/workshops/edinburgh2013/>
- 2012 International Semantic Web Applications and Tools for Life Sciences (SWAT4LS) Conference, 28-30 November Paris, France, <http://www.swat4ls.org/workshops/paris2012/>

Research projects involved

- 2021–2023 Brain Involvement in Dystrophinopathies (BIND), N° 847826
- 2020–2022 Health Holland Trusted World of Corona (TWOC), *The goal of this trusted guide to the world of COVID-19 is to help clinicians, the scientific community, policy makers and politicians and the public at large to get near real time accurate, expert-annotated and specific information in a modern, user friendly and easily accessible format*, <https://www.health-holland.com/project/2020/trusted-world-of-corona>
- 2019–2023 European Joint Programme Rare Diseases (EJP RD), N° 825575
- 2019–2021 ELIXIR Rare Disease (RD) Infrastructure, *This project will provide foundational infrastructure building blocks for the RD community, building on ELIXIR-supporting infrastructure and objectives: Data analysis; ensuring data is FAIR; training*, <https://elixir-europe.org/about-us/commissioned-services/rare-disease-infrastructure>

- 2016–2018 National Center for Advancing Translational Sciences Biomedical Data Translator program, *OT3TR002019*
- 2010–2018 National Institute of General Medical Sciences, *R01GM089820*
- 2014–2020 MedBioinformatics, *Grant agreement ID: 634143*
- 2011–2016 Open PHACTS, *The Open PHACTS consortium is building the Open PHACTS Discovery Platform. This will be freely available, integrating pharmacological data from a variety of information resources using a semantic web approach and providing tools and services to question this integrated data to support pharmacological research, <http://www.openphacts.org/>*
- 2010–2017 eTOX, *The eTOX project aims to develop a drug safety database from the pharmaceutical industry legacy toxicology reports and public toxicology data, and innovative in silico strategies and novel software tools to better predict the toxicological profiles of small molecules in early stages of the drug development pipeline, <http://www.etoxproject.eu/>*
- 2007–2009 CANCERGRID, *Grid aided computer system for rapid anti-cancer drug design, www.cancergrid.eu/*

Research funds

- 2022–2023 ELIXIR Rare Diseases 2022/23 Implementation Study
- Funding body: ELIXIR
 - Funded body: ELIXIR Rare Diseases Community nodes
- 2021 EHDEN 10/2021 - 5th Data Partner
- Funding body: The European Health Data & Evidence Network (EHDEN)
 - Funded body: The European Rare Kidney Disease Registry (ERKReg)

Research supervised

SUPERVISION AT LUMC

PhD advisor

- 2020–present Mr César H. Bernabé, *Biomedicine*, Conceptual goal modelling for FAIRification, Primary mentors: Dr. Annika Jacobsen and Dr. Marco Roos

MSc advisor

- 2020–present Mr Karolis Cremers, *Bioinformatics*, Huntington disease structured review for hypothesis generation, Primary mentors: Dr. Eleni Mina, Dr. **Núria Queralt Rosinach** and Dr. Katy Wolstencroft

Graduated

- 2021–2022 Mr Pablo Perdomo Quinteiro, *MSc Bioinformatics*, Knowledge graphs and explainable AI for Duchenne Muscular Dystrophy drug repurposing, Primary mentors: Dr. **Núria Queralt Rosinach**, Dr. Katy Wolstencroft
- 2021–2022 Ms Carmen Reep, *MSc Bioinformatics*, Automated drug repurposing workflow for rare diseases, Primary mentors: Dr. Eleni Mina, Dr. **Núria Queralt Rosinach**, Dr. Katy Wolstencroft
- 2020–2021 Mr Casper van Aarle, *MSc Data Science, Radboud University (NL)*, Federated Learning on personal health data, Primary mentors: Prof. Dr. Ir. Djoerd Hiemstra and Prof. Dr. Peter-Bram A.C. 't Hoen

SUPERVISION AT SR

- 2016 Ms Siyue Wu, *STSI Summer Student Research Internship Data Science*, Drug repurposing, Primary mentors: Prof. Dr. Andrew Su and Dr. Benjamin Good

Teaching

Open University of Catalonia (UOC)

- 2010–2016 Bioinformatics Final Project, *MSc Bioinformatics*

Rovira i Virgili University (URV)

- 2002–2005 FORTRAN 77 PROGRAMMING LANGUAGE, *BSc Chemistry*
2001–2006 Chemical-physics wet lab, *BSc Chemistry*

Professional affiliations

- 2021–present Research Data Alliance (RDA)
2012–present International Society for Computational Biology (ISCB)

Professional service

Editorial work

- 2016–present Member of Editorial Board, *Data Science: Methods, Infrastructure, and Applications*

Reviewer for journals

Journal of Biomedical Semantics, *Data Science: Methods, Infrastructure, and Applications*, F1000Research, BMC Bioinformatics, PLOS ONE, Bioinformatics, Scientific Data, Applied Ontology, Journal of Web Semantics

Conference organization

- 2022 Organizing committee for Intelligent Systems for Molecular Biology (ISMB) Bio-Ontologies COSI 2022
2022 PC Member for Semantic Web Applications and Tools for Health Care and Life Sciences (SWAT4HCLS) International Conference 2022
2021 PC Member for International Conference on Biomedical Ontologies (ICBO) 2021
2021 PC Member for Formal Ontology in Information Systems (FOIS) 2021
2021 PC Member and chair for Intelligent Systems for Molecular Biology (ISMB) Bio-Ontologies COSI 2021
2021 Organizing committee for Intelligent Systems for Molecular Biology (ISMB) Bio-Ontologies COSI 2021
2020 PC Member and chair for Intelligent Systems for Molecular Biology (ISMB) Bio-Ontologies COSI 2020
2018 PC Member for Semantic Web Applications and Tools for Health Care and Life Sciences (SWAT4HCLS) International Conference 2018
2017 PC Member for Semantic Web Applications and Tools for Health Care and Life Sciences (SWAT4HCLS) International Conference 2017
2016 PC Member for Semantic Web Applications and Tools for Health Care and Life Sciences (SWAT4HCLS) International Conference 2016
2015 PC Member for Semantic Web Applications and Tools for Health Care and Life Sciences (SWAT4HCLS) International Conference 2015

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