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Pillar 2
2021 Annual Retreat

Open session 3 May 2021

Online

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03 May 2021

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(Ana Rath, Franz Schaefer)

10:00 Resource finder Demonstration
(Sergi Beltran, Alberto Corvo)

10:30 Metadata Models & Standards
(Marc Hanauer, Marc Wilkinson, Rajaram Kaliyaperumal)

11:00 Break

11:30 Query Builder Resource & Record-Level pilot demonstration
(Anthony Brookes, Heimo Müller, Marc Hanauer, Haitham Abaza, Rajaram Kaliyaperumal, David Reinert, Alessandro Sulis/Vittorio Meloni, Collin Veal)

12:30 Open discussion

13:00 Lunch

Afternoon session (CEST)

14:00 FAIRification with ERN registries: stewards' work and R&D of practical guidance
(Marco Roos, Bruna Dos Santos Vieira, Nirupama Benis)

14:30 System biology output example: CAKUT work
(Chris Evelo, Friederike Ehrhart, Franz Schaefer, Peter-Bram t'Hoen)

15:00 Break

15:30 Distributed Federated Consent Control
(Anthony Brookes, Annalisa Landi, Yanis Mimouni, Spencer Gibson, Esther van Enckevort, Mark Wilkinson)

16:00 Experimental data archiving and analysis resource
(Morris Swertz, Giselle Kerry, Sergi Beltran, Carles Garcia)

16:30 Open Discussion

17:00 End of the Meeting

Session description

Resource finder Demonstration *10:00-10:30 CEST*

Chair/co-chair, Speaker, Rapporteur

Chair: Ana Rath
Speakers: Sergi Beltran & Alberto Corvo
Rapporteur: Carles Garcia

Description

One of the aims of EJP-RD is to increase the discoverability of resources that can be useful to the Rare Disease community.

To achieve this goal EJP-RD has developed the Resource Finder, a new tool to easily find important rare disease resources based on different categories. It is based on a simple interface that provides information on IRDiRC recognized and EJP-RD funded resources and tools.

Objectives

- To learn where to find and how to use the Resource Finder.
- To learn which type of information it can provide and how is it organized.

Metadata Model & Standards *10:30-11:00 CEST*

Chair/co-chair, Speaker, Rapporteur

Co-Chair, Speaker Marc Hanauer, Mark Wilkinson, Rajaram Kaliyaperumal
Rapporteur Rajaram Kaliyaperumal

Description

While people often speak of FAIR Data, the fact is that FAIRness is only 10% about data, and 90% about metadata - the majority of the FAIR Principles speak about (meta)data, indicating that they refer to both metadata and data. The objective of the FAIR Principles is to achieve a world where data can be discovered, accessed, and reused in an accurate manner by both humans and machines. This necessitates that data be richly described, to ensure that it is correctly contextually interpreted, and that the data components can be understood.

In the EJP-RD, the FAIR team has made several choices regarding how to build rich metadata records to describe rare disease resources such as registries and biobanks. These pivot around the selection of DCAT2 as our primary metadata model, which allows us to extend that model into RD-specific metadata elements, and the use of the Linked Data Platform as an

underpinning architecture, ensuring that the relationships between these new metadata elements can be reliably described for fully mechanized exploration. In this session, we will describe these features, and show examples of how they are used for exploration of RD resources.

Objectives

- To learn about the EJP metadata model, based on DCAT 2.0 standard
  - DCAT2.0 general concept and EJP metadata model extension
- To learn about how Linked Data Platform is underpinning the FAIRification process from Resources Metadata to Records Metadata
- To learn how the metadata model could be implemented with a methodology to produce a FAIRdatapoint

QB Resource- & Record-Level pilot demonstration *11:30-12:30 CEST*

Chair/co-chair, Speaker, Rapporteur

Co-Chairs: Anthony Brookes, Heimo Müller, Marc Hanauer, Haitham Abaza, David Reinert

Speakers: Anthony Brookes, David Reinert, Marc Hanauer, Heimo Müller, Rajaram Kaliyaperumal, Alessandro Sulis/Vittorio Meloni, Collin Veal

Rapporteur: Haitham Abaza

Description

Towards federated discoverability of RD resources and content, the Query Builder Work Focus (QBWF) is proposing two parallel, synergistic approaches: (i) a federated network for resource discovery (by inter-connecting resource catalogues); and (ii) a federated network for record-level discovery (for safe and ethico-legally appropriate discovery of patients, samples, publications, experts/researchers and datasets).

After having built these pilot projects a number of learnings occurred. Many possibilities of interoperability to other EJP RD work foci opened up. Many new ideas for further development and pilot projects appeared.
During this presentation the Query Builder work focus would like to share the achievements, the learnings, new ideas and possibilities with the audience and discuss how they can be further explored and incorporated into the EJP RD discovery ecosystem.

The presentation will cover:

- the overall vision of the discovery ecosystem and its' limitations, current status and next steps
- explanations on the underlying technologies of both the resource- and record-level pilot, as well as live demonstrations.

- specifications of an early LDP prototype and ontology mapping services and plans on how to incorporate them
- an outlook on how to integrate the resource-, record-level, fair datapoint and other pilot projects
- as well as an outlook and discussion on what pilot projects derived from the learnings of the now completed pilots

Schedule

11:30 – 11:35	Welcoming and Overall Vision
11:35 – 11:55	Resource-Level projects
11:55 – 12:15	Record-Level projects
12:15 – 12:30	Integration work

Objectives

- Update the audience on the achievements, ideas and possibilities of the Query Builder work.
- Discuss on how these achievements, ideas and possibilities can be further explored and incorporated into the EJP RD discovery ecosystem.
- Discuss about the next steps of the query builder work focus, current challenges, practicalities and opportunities.

FAIRification with ERN registries: stewards' work and R&D of practical guidance *14:00-14:30 CEST*

Chair/co-chair, Speaker, Rapporteur

Co-Chair and Speakers: Marco Roos, Bruna Dos Santos Vieira, Nirupama Benis

Rapporteur: Nirupama Benis

Description

The EJP RD Virtual Platform will use the FAIR principles for its operation: by agreeing on how resources can make themselves findable, accessible, interoperable, and reusable, for humans, but especially for machines, we can generate a robust virtual platform that is efficient by automation, and ready for AI and federated analysis. Implementing FAIR principles is far from trivial. There are many standards to choose from, resources are heterogeneous, and FAIR is new to many stakeholders. Therefore, we have a dedicated task on the process of FAIRification. It pertains to identifying and assessing standards and tools for implementing FAIR principles, guidelines and metrics for FAIRness evaluation, organising expertise, aligning with the VP infrastructure, and supporting training.

One important aspect of FAIRification is *organising* the FAIRification process. For ERNs we have developed an organisational model to guide the process based on previous experiences: a three-party setup of ERN registry stewards, ERN registry software providers, EJP RD FAIRification stewards. They collaborate to guide and develop customized procedures from the basic FAIRification steps. The EJP RD stewards also form a team to discuss their experiences and pass on needs to EJP RD researchers and developers. In a few months the stewards have become a critical element in the development of FAIRification and guidance on FAIRification.

Objectives

- To update on the work of the FAIRification stewards.
- To discuss the research and development of FAIRification guidance, encompassing workshops and coffee sessions, engagement with external experts, and a use case on 'smart guidance'.
- To discuss the role and next steps of EJP RD supported stewardship.

Systems biology output example: CAKUT work *14:30-15:00 CEST*

Chair/co-chair, Speaker, Rapporteur

Co-chair, Speaker: Chris Evelo, Franz Schaefer, Peter-Bram t'Hoen

Rapporteur: Friederike Ehrhart

Description

The systems biology work package is all about (multi) omics data analysis. For this we build resources and work on integration of other data and information, e.g., from nutrition, drug and toxicology. One of our use cases is a dataset about Congenital Anomalies of Kidney and Urinary Tract disorders (CAKUT) provided by University of Toulouse (ERN: ERKnet). In this session we show what we did with the data in terms of data analysis and providing workflows, and the resources we build with the data (FAIRification) and the combined knowledge of clinical, biomedical and data integration experts (building pathways and disease networks).

Objectives

- Demonstrate data analysis workflows
- Demonstrate resource building

Distributed Federated Consent Control work *15:30-16:00 CEST*

Chair/co-chair, Speaker, Rapporteur

Chair: Anthony Brookes

Speakers: Annalisa Landi, Yanis Mimouni, Spencer Gibson, Esther van Enckevort, Mark Wilkinson

Rapporteur: Spencer Gibson, Maria del Carmen Sanchez Gonzalez

Description

Consent and use conditions pervade all aspects of Pillar 2 and the VP. There are various sources of restriction and permission that apply to research assets (individual consent, institution/research requirements, GDPR-related considerations, ethical constraints), various domains to which they relate (biobanking, registries, clinical research), and many application areas (informing and communicating rights/restrictions, gathering consent, recording applicable conditions, discovery applications, assets sharing, data analysis, etc). The Distributed/Federated Consent Control Work Focus seeks to identify, develop, test and provide a range of common tools and approaches to help the RD field converge and unify around its handling of consent and use condition information.

This session will summarise a series of theoretic and practical undertakings that will provide cornerstones for tool development and optimal handling of consent and use conditions within EJP RD, maximally aligned to ideas and systems emerging elsewhere in other RD projects and settings.

Schedule

- **Consent and Use conditions in EJP RD & IRDiRC**
  Anthony Brookes - 6 minutes
- **Standardised consent forms for RD registries**
  Annalisa Landi / Yanis Mimouni - 6 minutes
- **Practical use of ontologies, Common Consent Elements (CCE)**
  Spencer Gibson- 6 minutes
- **Framework semantic model for automated sharing decisions**
  Esther van Enckevort / Mark Wilkinson - 6 minutes
- **Questions/Discussion** - 6 minutes

Objectives

- Establish a Pillar-2 wide understanding of the challenges and opportunities
- Gather feedback on current areas of development
- Discuss applications and tools to implement Work Foci advances

Experimental data archiving and analysis resources *16:00-16:30 CEST*

Chair/co-chair, Speaker, Rapporteur

Chair: Morris Swertz

Speakers: Giselle Kerry, Sergi Beltran

Rapporteur: Carles Garcia

Description

Several key data and bio-material resources are included in EJP-RD Pillar 2 and will be part of the Virtual Platform. Some of these resources were originally developed specifically for Rare Diseases, but others had a more general scope. The resources included focus on different types of data: registries (JRC-ERDRI, RD-Connect Registry and Biobank Finder, known also as IDcards), patient cohorts (RadiCo, BBMRI-ERIC Directory), biobanks (RD-Connect Sample Catalogue, RD-Connect Registry and Biobank Finder, BBMRI-ERIC Directory), cell lines (hPSCreg, Cellosaurus), mouse models (INFRAFRONTIER), raw omics data (EGA, Pride, MetaboLights), genome-phenome analysis platforms (DECIPHER, RD-Connect GPAP) and cloud computing. In this session we will hear the latest developments from these resources as part of EJP-RD and their ongoing work to connect to the Virtual Platform.

Objectives

- To explain the latest developments and adaptations from key EJP-RD data and bio-material resources
- To explain the ongoing work to connect these resources to the Virtual Platform

Schedule

- 20 minutes presentations
- 10 minutes Q&A

Speakers' and Chairs' Biosketches

Alberto Corvo

Fundació Centre de Regulació Genòmica (CNAG-CRG) – Spain

Alberto Corvo is a Front-End Engineer at the National Center of Genomic Analysis currently involved in the conceptual design and development of the Genome-Phenome Analysis Platform (GPAP). The platform aims at supporting clinicians and researchers in the diagnosis and investigation of genome variants in rare diseases and cancer. He designs, plans and develops the GPAP platform in all its crucial aspects, from APIs requirements to UX to address end-users needs.

He is also involved in several European Projects as the European Joint Programme on Rare Diseases, RD-Connect, GenoMed4All.

In December 2019, he obtained his PhD at Eindhoven University of Technology. His research focus was on Visual Analytics applications for Philps Digital & Computational Pathology. He worked on empowering pathologists with image analysis support for diagnostics in highly interactive software. At the same time, he designed and implemented visual analytics applications to help researchers in the analysis of 2D spatial data in computational pathology.

Ana Rath

Institut National de la Santé et de la Recherche Médicale (INSERM)_Orphanet – France

- Co-Leader of EJP RD Pillar 2 "innovative coordinated access to data and services for transformative RD research"
- Co-Leader of EJP RD Work Package 10 "User-driven strategic planning and transversal activities for Pillar 2 data ecosystem"
- Co-Leader of EJP RD Work Package 11 "Common virtual platform for discoverable data and resources for RD research"

Ana Rath is a medical doctor with a background in general surgery and a master's degree in Philosophy. She oriented her career to medical information in 1997 and joined Orphanet (www.orpha.net) in 2005. She acts as managing editor of the Topic Advisory Group on rare diseases at the World Health Organization. She chairs the Orphanet rare diseases ontology (ORDO, http://bioportal.bioontology.org/ontologies/ORDO). Ana is the current Director of Orphanet and the coordinator of RD-ACTION, the EU Joint Action for rare diseases (2015-2018) and of the HIPBI-RD project. She chairs the Orphanet Rare Disease Ontology (ORDO).

Annalisa Landi

Gianni Benzi Pharmacological Research Foundation, Bari – Italy.

- Co-Contributor of the EJP RD Work Package 4 "Ethical, regulatory, legal and IPR framework of the EJP RD"

Annalisa is a PharmD researcher with a II level post-graduate Master in Regulatory Sciences at University of Pavia. She carried out an internship at Leiden University Medical Center in the FAIR team of Marco Roos. Member of the WG3 of the European Network of Centres for Pharmacoepidemiology and Pharmacovigilance (ENCePP). She collaborates in several national and international projects dealing with ethics in biomedical research. She is involved in scientific, ethical and regulatory activities particularly related to clinical studies, data protection and confidentiality, plan and management of patient registries and medicine databases, health data and accessibilities issues

Anthony J. Brookes

University of Leicester (ULEIC) – Great Britain

- Co-Leader of EJP RD Work Package 10 "User-driven strategic planning and transversal activities for Pillar 2 data ecosystem"
- Co-Leader of EJP RD Work Package 12 "Enabling sustainable FAIRness and Federation at the record level for RD data, patients and samples"
- EJP-RD co-representative in GA4GH

Director BINERI, Director HDR-UK Leicester, Research Group leader.

Qualifications in preclinical Medicine (Honours) and Medical Biochemistry (1st Class), creator of various technology patents, Founder of two biodata related companies, 30 years' experience as academic researcher in leading institutions. Published >180 peer reviewed articles and reviews, served two 3-year terms on the HUGO Council, co-founded the Human Variome Project and the Human Genome Variation Society (now Vice-President)

Bruna Dos Santos Vieira

Radboudumc (RUMC) – The Netherlands

- Leader of the EJP-RD FAIRification Stewards

Bruna has >5 years' experience in public hospitals and a background in hospital management and public health. During 2011-2014, she studied Hospital Management at University Feevale (Brazil) developing practical solutions through integrated projects for strategic planning, processes, people, logistics, and financial management in different Brazilian public health institutions. In 2018 she obtained her Public Health Master's degree from the University of Porto (Portugal), focusing on data quality of hospital-based cancer registries. Since 2019, she works as a data steward at the Center for Molecular and Biomolecular Informatics (CMBI) and Radiology departments at Radboudumc (Netherlands). She currently collaborates with

the Registry for Rare Vascular Anomalies (VASCA) and EJP-RD Metadata and FAIRification WFs. Her most recent contributions include the de novo FAIRification process of a registry for vascular anomalies, ontological representations of the WHO COVID-19 Rapid Version CRF, ISSVA Classification, PRISMA database, and providing data stewardship for Radboudumc COVID-19 studies.

Chris Evelo

Maastricht University (UM) – The Netherlands

- Co-Leader of EJP RD Work Package 13 "Enabling multidisciplinary, holistic approaches for rare disease diagnostics and therapeutics"

Prof. Evelo is the founder and head of the department of Bioinformatics - BiGCaT and is a PI in the Maastricht Center for Systems Biology (MaCSBio). His research focus is on bioinformatics for integrative systems biology; aiming at a better interpretation of experimental data through integration in data models that build on structuring existing knowledge. Integrative approaches are multi-faceted, and as a generalist he is involved in many projects related to capturing and processing, integrating and understanding experimental data. This includes the interoperability approaches underlying such efforts: standards, ontologies, mapping tools and documentation of the origin of the data and methods used (the provenance). He is a co-lead of the interoperability platform of ELIXIR which he represents in the rare disease use case. WikiPathways and the accompanying mapping, analysis and network biology tools form his core project. WikiPathways is a resource for community curation of biological pathways and originates from collaboration with Alex Pico's group at UCSF. He is a board member of the Open PHACTS foundation for large-scale semantic web-based knowledge structuring of relations between chemicals, gene-products and diseases. Chris is scientific advisor for the IMI project for translational quantitative systems biology (TransQST) and for two SME's (Edgeleapand Micelio). He is a.o. governing council member of the nutrigenomics organization NuGO, a member of the micronutrient genomics organization for which he lead the IRSES project Microgennet, involved in H2020 projects OpenRiskNet and EUToxRisk and a work package leader in the COST CHARME for harmonization of standards in biology and a member of the JPI ENPADASI.

David Reinert

Goethe-University Frankfurt am Main (GUF) – Germany

- Co-Contributor of the EJP RD Work Package 11 "Common virtual platform for discoverable data and resources for RD research"
- Co-Contributor of the EJP RD Work Package 12 "Enabling sustainable FAIRness and Federation at the record level for RD data, patients and samples"

David Reinert has a master's degree in computer science from the University of Applied Sciences Darmstadt, Germany (h-da).

Since 2015 he has been working in the field of medical informatics at Fraunhofer IGD, Darmstadt, covering several visual healthcare and medical image analysis projects.
Since 2020 he is part of the Medical Informatics Group at Goethe University Frankfurt am Main, Germany and the EJP-RD Query Builder team.

Friederike Ehrhart

Maastricht University (UM) – The Netherlands

- Responsible of the EJP RD Work Focus "Pathway creation and curation"
- Deputy lead of EJP RD Work Package 13 "Enabling multidisciplinary, holistic approaches for rare disease diagnostics and therapeutics"

Dr. Friederike Ehrhart is currently assistant professor at the Department of Bioinformatics (head of department: Prof. Dr. Chris Evelo). Educated and trained as cell and molecular biologist, working on tissue engineering of neurological, pancreatic and liver tissue, and high throughput and omics data approaches. Joining the Department of Bioinformatics at Maastricht University in 2015 she works on systems biology and bioinformatics methods to analyse (single or multi) omics data, especially within pathway and network analysis. Together with the Dutch Rett Expertise Centre in Maastricht she is working since 2016 on the systems biology of Rett syndrome, a rare neurological disorder with a genetic cause and on several other neurological and metabolic disorders. Friederike has coordinated and co-coordinated 7 public and 3 industry funded projects and published 31 papers. Within ELIXIR (EU bioinformatics infrastructure project) she coordinated an implementation study which analyses and improves molecular data interoperability of rare diseases. She is a member of the SSBP (Society for the Study of Behavioural Phenotypes) to investigate (rare) genetic syndromes and their phenotypic outcome including foetal alcohol syndrome, Prader-Willi and Angelman syndrome.

Franz Schaefer

Universitätsklinikum Heidelberg (UKL-HD) – Germany

- Co-Leader of EJP RD Pillar 2 "innovative coordinated access to data and services for transformative RD research"
- Co-Leader of EJP RD Work Package 13 "Enabling multidisciplinary, holistic approaches for rare disease diagnostics and therapeutics"

Franz Schaefer is a Professor of Paediatrics and Chief of the Paediatric Nephrology Division at Heidelberg University Hospital.
He leads the work package "Data sharing and integration" in ERICA, the ERN Research Coordination and Support Action and is a member of the c4c paediatric clinical research network. Moreover, he coordinates several international clinical networks and research consortia such as ERKNet, the European Rare Kidney Disease Reference Network (since 2017), the IPNA Global Kidney Replacement Therapy Registry (since 2016), the EURenOmics Consortium for High-Throughput Research in Rare Kidney Diseases (2013-2018),

the PodoNet Consortium for podocyte diseases (since 2009), and the ESCAPE Clinical Research Network for Chronic Kidney Disease in Children (since 2004). Dr. Schaefer has published more than 650 scientific articles and book chapters, co-edits the standard textbooks 'Paediatric Kidney Disease' and 'Paediatric Dialysis' and is President-Elect of the International Paediatric Nephrology Association (IPNA).

Haitham Abaza

Goethe University Frankfurt (GUF) – Germany

- Co-Contributor of the EJP RD Work Package 11 "Common virtual platform for discoverable data and resources for RD research"
- Co-Contributor of the EJP RD Work Package 12 "Enabling sustainable FAIRness and Federation at the record level for RD data, patients and samples"

Dr. Haitham Abaza is a Research Associate affiliated with the Medical Informatics Group at Frankfurt University Hospital. He received his Doctoral degree in Medical Informatics from the Hanover Medical School and his MSc. degree in Biomedical Engineering from Cairo University. He is currently involved in EJP RD's Query Builder activities focused on the discoverability of RD resources. He is also involved in the registry design and interoperability activities, co-leading the initiative to extend the EU RD Platform's Set of Common Data Elements with Domain-specific Common Data Elements (DCDEs). His former areas of focus include mHealth applications, robotic surgical systems and healthcare technology management.

Marc Hanauer

Institut National de la Santé et de la Recherche Médicale (INSERM)_Orphanet – France

- Co-Contributor of the EJP RD Work Package 11 "Common virtual platform for discoverable data and resources for RD research"
- Co-Responsible of the EJP RD Work Focus "Metadata model & alignment service"

Marc Hanauer is an engineer at INSERM and Chief Technology Officer at Orphanet since 2007 and Orphanet Deputy director since 2017. He is also in charge of managing Orphanet's innovation strategy. Previous to his current position, Marc worked for different internet start-ups from 2000 on-wards. He has an academic background in information and communication sciences.

Marco Roos

Leiden University Medical Centre (LUMC) – The Netherlands

- Co-Leader of EJP RD Work Package 12 "Enabling sustainable FAIRness and Federation at the record level for RD data, patients and samples"

Dr. Marco Roos is group leader of the Biosemantics group that is one of the co-founders of the FAIR principles, following the initiative of the group's prof. Barend Mons in 2014. Marco's career is characterized by research on enhancing biomedical research by interdisciplinary collaboration and methods emerging from computer science ('e-Science'), with particular interest in the contribution of Semantic Web, ontologies, and computational workflows. His current primary focus is on FAIR data stewardship, federated infrastructure, and FAIR-based data analytics to enhance translational research in the domain of rare diseases. He co-leads the ELIXIR rare disease user community, and the EJP RD work package 12 on enabling sustainable FAIRness and Federation at record level for RD data, patients and samples. He also initiated the Rare Disease GO FAIR implementation network to foster the implementation of FAIR principles in the wider rare disease community. Other application areas are oncology and Covid-19, and genomic data in personal lockers.

Mark Wilkinson

Isaac Peral Senior Researcher, Centre for Plant Biotechnology and Genomics, Universidad Politécnica de Madrid, Spain

- Co-Contributor of the EJP RD Work Focus "(Meta)data model & alignment service"

Mark has a B.Sc.(Hons) in Genetics from the University of Alberta, and a Ph.D. in Botany from the University of British Columbia. He spent four years at the Max Planck Institut für Züchtungsforschung in Köln, Germany, pursuing studies in a mix of plant molecular and developmental biology and bioinformatics. He then did a research associateship at the Plant Biotechnology Institute of the National Research Council Canada, focusing on the problem of biological data representation and integration for the purposes of automated data mining. In the subsequent 20 years, his laboratory has focused on designing biomedical data/tool representation, discovery, and automated reuse infrastructures - what are now called "FAIR Data" infrastructures. He is the lead author of the primary FAIR Data Principles paper, and lead author on the first paper describing a complete implementation of those principles over legacy data. He is a founding member of the FAIR Metrics working group, tasked with defining the precise, measurable behaviours that FAIR resources should exhibit, and the author of the first software application capable of a fully-automated and objective evaluation of "FAIRness". He continues to pursue research in the domain of automated data discovery and analytics.

Morris Swertz

Academisch Ziekenhuis Groningen (UMCG) – The Netherlands

- Co-Responsible of the EJP RD Work Foci "Metadata model & alignment service" & "FAIRification"

Prof. Dr. Morris Swertz coordinates large scale bioinformatics research and support to the Dept. of Genetics, other UMCG departments, and several (inter)national consortia. His research lines focus on: (1) methods for DNA, RNA and phenotype data analysis, and (2) user-friendly systems for large scale data management, analysis, interpretation and sharing. Swertz developed a novel data framework (Bioinformatics 2012) and computational framework (Molgenis/Compute) to enable large genome analyses, e.g. used in Genome of the Netherlands (EJHG 2013, Nature Genetics 2014), discovery of novel human gene-disease associations based on eQTL studies (NAR 2014), inconsistencies in the metabolic pathway knowledge (BMC Systems Biology 2013), and to inform genetic variant interpretation in various diseases (Human Mutation 2011-2015). Morris also developed analysis methods to call genotypes from RNA-sequencing that enables identification of (rare) variants affecting gene- expression levels (Genome Med 2015) and a framework to estimate RD variant pathogenicity classification for clinical application (Human Mutation 2015, Genome Biology 2017)

Finally, Morris is Co-Lead of BBMRI-NL 2.0 'IT', genetics data sharing in VKGL & VKGN, Co-lead of UMCG Personalized Genomics Medicine programme Lead of UMCG Research IT program, focus on big data

Nirupama Benis

- Co-Responsible of the EJP RD Work Foci "Metadata model & alignment service" &"FAIRification "

Nirupama Benis is a post-doctoral researcher at AMC, Amsterdam, working on methodology related to FAIRification with Interoperability as a strong focus. She has a background in Biomedical engineering and received her PhD in multi-omics data analysis from Wageningen University, The Netherlands. Her current research topics include general FAIRification methodology and Interoperability in –omics data

Peter-Bram t'Hoen

- Co-Contributor of the EJP RD Work Package 11 "Common virtual platform for discoverable data and resources for RD research"
- Co-Contributor of the EJP RD Work Package 12 "Enabling sustainable FAIRness and Federation at the record level for RD data, patients and samples"
- Co-Contributor of the EJP RD Work Package 13 "Enabling multidisciplinary, holistic approaches for rare disease diagnostics and therapeutics"

Peter-Bram 't Hoen obtained his MSc degrees in Biochemistry and Pharmacochemistry from the Vrije Universiteit Amsterdam with distinction 'cum laude' and his PhD in biopharmaceutical sciences from Leiden University. From 2002-2018, he worked as postdoctoral researcher, assistant and associate professor in the department of Human Genetics at Leiden University Medical Center. In 2018, he was appointed as professor of Bioinformatics at Radboudumc Nijmegen, where he is leading the Centre for Molecular and Biomolecular Informatics (CMBI). He develops new approaches for the analysis of -omics data, their integration with clinical data, and the discovery of molecular signatures that can be used in disease diagnostics, disease progression and the response to therapy ('personalised medicine').

Rajaram Kaliyaperumal

Leiden University Medical Centre (LUMC) – The Netherlands

- Co-Contributor of the EJP RD Work Package 11 "Common virtual platform for discoverable data and resources for RD research"
- Co-Contributor of the EJP RD Work Package 12 "Enabling sustainable FAIRness and Federation at the record level for RD data, patients and samples"

Rajaram Kaliyaperumal was born in Pondicherry, India. He received a B.Tech degree in Biomedical Engineering from Pondicherry University, India in 2008 and an M.Sc degree in Biomedical Engineering from Linköping University, Sweden in 2011. In 2012 he joined the department of Computer and Information Science, Linköping University as a software engineer. During this time, he developed methods and tools to align and repair ontologies. In 2013 he joined the Biosemantics group, Leiden, in the Netherlands as a software developer. His current research activities include investigating the use of semantic web technology in the context of FAIR data and developing prototypes to demonstrate generating and the use of FAIR data.

Sergi Beltran

Fundació Centre de Regulació Genòmica (CNAG-CRG) – Spain

- Co-Leader of EJP RD Work Package 11 "Common virtual platform for discoverable data and resources for RD research"
- EJP-RD co-representative in GA4GH

Sergi Beltran has over 20 years of experience with genomics and bioinformatics and has participated in over 60 international publications. He leads the development of the RD-Connect platform (platform.rd-connect.eu) and is a partner in EU projects Solve-RD and Genomed4All. His group participates in the IRDiRC/GA4GH MatchMaker Exchange (MME) and GA4GH Beacon projects. Within ELIXIR-EXCELERATE, Dr. Beltran participates in the RD use case co-led from the CNAG-CRG and in benchmarking activities.

Spencer Gibson

University of Leicester (ULEIC) – Great Britain

- Co-Contributor of the EJP RD Work Foci "Distributed and Federated Consent Control"

Dr Spencer Gibson is a Molecular Biologist specialising in SNP genotyping assay development and platform adaptation. He currently works at the Department of Genetics, University of Leicester. He is involved in research in Genetics, Molecular Biology and Biotechnology. His current work involves the use of genomic data in clinical and medical research. Part of his work involves the investigation of novel methods for sharing of clinical data as a part of E.U. funded European initiative within the Brookes lab bioinformatics group (currently investigating novel uses for block chains).

Yanis Mimouni

Institut National de la Santé et de la Recherche Médicale (INSERM) – France

- Senior Project Manager – Coordination and Transversal Activities and Pillar 2 – Innovative coordinated access to data and services for transformative rare diseases research

Yanis is a Doctor of Pharmacy with a MSc in Pharmaceutical Medicine (Clinical Development of Health Products). He participated in the set-up and coordinated more than forty European and international research projects. These latter included rare diseases therapeutic area and comprised phase I, II, III and IV clinical trials; registries; translational; diagnostic, pharmaco-epidemiological and pharmacovigilance projects. Yanis initiated the exploitation of medical Information Systems and Databases to optimize project execution and inform Medical Decision. He joined the European Joint Program on Rare Diseases in July 2019 as Senior Project Manager. His role is to participate in the management of the coordination and transversal activities as well as all activities related to the Pillar 2 – Virtual Platform for Data and Services.