European Joint Programme on Rare Diseases

Results

Project Information

EJP RD

Grant agreement ID: 825575

Project website

DOI
10.3030/825575

Project closed

EC signature date
13 December 2018

Start date
1 January 2019

End date
31 August 2024

Funded under
SOCIETAL CHALLENGES - Health, demographic change and well-being

Total cost
€ 100 655 230,97

EU contribution
€ 55 073 831,17

Coordinated by
INSTITUT NATIONAL DE LA SANTE ET DE LA RECHERCHE MEDICALE
France

CORDIS provides links to public deliverables and publications of HORIZON projects.

Links to deliverables and publications from FP7 projects, as well as links to some specific result types such as dataset and software, are dynamically retrieved from OpenAIRE.

Deliverables

Documents, reports (74)

Fourth Public Call documents JTC2019-2022: call text, guidelines for applicants, proposal templates

Other lead beneficiaries ANR 11 FFRD 12 and ISCIII 44Public Call documents JTC20192022 call text guidelines for applicants proposal templates due at month 35

First Report on processed genome-phenome datasets and multi-omics use cases analysed, including description of new cloud and online analysis functionalities and tools

Other lead beneficiary UMCG (35) First Report on processed genome-phenome datasets and multi-omics use cases analysed, including description of new cloud and online analysis functionalities and tools due at month 12

Third Report on processed genome-phenome datasets and multi-omics use cases analysed, including description of new cloud and online analysis functionalities and tools

Other lead beneficiary UMCG 35Third Report on processed genomephenome datasets and multiomics use cases analysed including description of new cloud and online analysis functionalities and tools due at month 36

First Report on course on interpretation of genetic variants and quality standards

First Report on course on interpretation of genetic variants and quality standards due at month 12

Fourth Analysis of national state of play and alignment process with EJP RD

Other lead beneficiary MUW 38Analysis of national state of play and alignment process with EJP RD due at month 55

Second Public Call documents JTC2019-2022: call text, guidelines for applicants, proposal templates

Other lead beneficiaries: ANR (11), FFRD (12) and ISCIII (44) Public Call documents JTC2019-2022: call text, guidelines for applicants, proposal templates due at month 11

First Report on ExPRESS

First Report on ExPRESS EURORDIS Summer School due at month 12

Fifth report from the face-to-face ExCom and Policy Board meeting

Report from the facetoface ExCom and Policy Board meeting due at month 55

Public Call document for Networking scheme

Other lead beneficiary: CSO/MOH (78) Public Call document for Networking scheme

First Report on International course on undiagnosed diseases

First Report on International course on undiagnosed diseases due at month 24

Final list of prioritization criteria
Other lead beneficiary FTELE 25Final list of prioritization criteria

Second List of research and innovation needs requiring medium- or long-term approach and related Task Forces
List of research and innovation needs requiring medium- or long-term approach and related Task Forces due at month 18

Second Report on processed genome-phenome datasets and multi-omics use cases analysed, including description of new cloud and online analysis functionalities and tools
Other lead beneficiary UMCG (35) Second Report on processed genome-phenome datasets and multi-omics use cases analysed, including description of new cloud and online analysis functionalities and tools due at month 24

Third report from the face-to-face ExCom and Policy Board meeting
Report from the face-to-face ExCom and Policy Board meeting due at Month 31

Second List of funded networks in Networking scheme
List of funded networks in Networking scheme due at month 54

Fourth Annual strategic report and Action plan for Pillar 2, including: Systematic surveys reports, QMS of Pillar 2 description, GDPR compliance report and sustainability planning reporting
Annual strategic report and Action plan for Pillar 2 including Systematic surveys reports QMS of Pillar 2 description GDPR compliance report and sustainability planning reporting update at month 45

Second Report on training of patient representatives on scientific innovation and translational research in RD
Second Report on training of patient representatives on scientific innovation and translational research in RD due at month 60

Second Report on ExPRESS
Second Report on ExPRESS EURORDIS Summer School due at month 60

Collection of 20 curated (sub)Portal on WikiPathways
Collection of 20 curated subPortal on WikiPathways

Third List of research and innovation needs requiring medium- or long-term approach and related Task Forces
List of research and innovation needs requiring medium or longterm approach and related Task Forces due at month 30

Report on core set of unified FAIR data standards

Report from the Kick-off meeting

Dynamic catalogue of EJP-RD sustainable resources including Service Roadmap Database
Dynamic catalogue of EJPRD sustainable resources including Service Roadmap Database Lead beneficiary INSERM RADICO

First RE(ACT) Congress report
First RE(ACT) Congress report due at M16

Second Annual reports on implementation of training programs
Second Annual reports on implementation of training programs due at month 60

List of first phase projects financed

First List of research and innovation needs requiring medium- or long-term approach and related Task Forces
List of research and innovation needs requiring medium or longterm approach and related Task Forces due at month 6

Fourth Scoping paper
Other lead beneficiaries ISCIII 44 and INSERM 1Scoping paper due at month 43

Draft content of the online academic course

Set of template agreements for EJP RD website

Second Report on EURORDIS' Leadership Programme
Second Report on EURORDIS’ Leadership Programme due at month 48

Third Analysis of national state of play and alignment process with EJP RD
Other lead beneficiary MUW 38Analysis of national state of play and alignment process with EJP RD due at month 32

Third Public Call documents JTC2019-2022: call text, guidelines for applicants, proposal templates
Other lead beneficiaries: ANR (11), FFRD (12) and ISCIII (44) Public Call documents JTC2019-2022: call text, guidelines for applicants, proposal templates due at mont 23

Training plan for paediatric patients' in the EJP
Other beneficiary FSJDTraining plan for paediatric patients in the EJP

First Report on sample data management training workshops

First Report on sample data management training workshops due at month 12

EJP RD ERN training programmes

First Public Call documents JTC2019-2022: call text, guidelines for applicants, proposal templates
Other lead beneficiaries: ANR (11), FFRD (12) and ISCIII (44) Public Call documents JTC2019-2022: call text, guidelines for applicants, proposal templates due at month 1

Call documents for validation

Second Analysis of national state of play and alignment process with EJP RD
Other lead beneficiary MUW (38) Analysis of national state of play and alignment process with EJP RD due at month 21

First Scoping paper
Other lead beneficiaries: ISCIII (44) and INSERM (1) Scoping paper due at Month 7

Fourth Report on processed genome-phenome datasets and multi-omics use cases analysed, including description of new cloud and online analysis functionalities and tools
Other lead beneficiary UMCG 35Fourth Report on processed genomephenome datasets and multiomics use cases analysed including description of new cloud and online analysis functionalities and tools due at month 48

Second Annual strategic report and Action plan for Pillar 2, including: Systematic surveys reports, QMS of Pillar 2 description, GDPR compliance report and sustainability planning reporting
Annual strategic report and Action plan for Pillar 2, including: Systematic surveys reports, QMS of Pillar 2 description, GDPR compliance report and sustainability planning reporting update at month 21

Completed pathway analysis workflow (data analysis including both types of genetic variant linking and network creation)
Completed pathway analysis workflow data analysis including both types of genetic variant linking and network creation

Fourth report from the face-to-face ExCom and Policy Board meeting
Report from the facetoface ExCom and Policy Board meeting due at Month 43

Results of survey on preferences, needs and resources from the ERNs ecosystem

Report on paediatric patients' experts training courses
Other beneficiary: FSJDReport on paediatric patients’ experts training courses

First Report on core set of FAIR software tools and on extended set of unified FAIR data standards, applied in EJP RD

First Report on core set of FAIR software tools and on extended set of unified FAIR data standards applied in EJP RD due at month 30

Second Report on sample data management training workshops

Second Report on sample data management training workshops due at month 60

First Report of Orphanet nomenclature training for trainers and national trainings

First Report of Orphanet nomenclature training for trainers and national trainings due at month 14

First Report from strategic workshop with national policy makers

Other lead beneficiary MUW 38Report from strategic workshop with national policy makers due at month 34

Third Annual strategic report and Action plan for Pillar 2, including: Systematic surveys reports, QMS of Pillar 2 description, GDPR compliance report and sustainability planning reporting

Annual strategic report and Action plan for Pillar 2 including Systematic surveys reports QMS of Pillar 2 description GDPR compliance report and sustainability planning reporting update at month 33

Prioritization scheme including decision-making process

Other lead beneficiary FTELE 25Prioritization scheme including decisionmaking process

Second RE(ACT) Congress report

Second REACT Congress report due at Month 30

Report on the State of the Art of existing resources

First List of funded networks in Networking scheme

List of funded networks in Networking scheme due at month 21

First Analysis of national state of play and alignment process with EJP RD

Other lead beneficiary MUW (38) Analysis of national state of play and alignment process with EJP RD due at Month 9

First Report on International course on Rare Disease Registries and FAIRification of data at source

First Report on International course on Rare Disease Registries and FAIRification of data at source due at month 12

Second Report of Orphanet nomenclature training for trainers and national trainings

Second Report of Orphanet nomenclature training for trainers and national trainings due at month 60

Fifth Scoping paper
Other lead beneficiaries ISCIII (44) and INSERM (1)Scoping paper due at month 55

Second Scoping paper
Other lead beneficiaries ISCIII (44) and INSERM (1) Scoping paper due at month 19

Second Report on course on interpretation of genetic variants and quality standards
Second Report on course on interpretation of genetic variants and quality standards due at month 60

Second Report on International course on Rare Disease Registries and FAIRification of data at source
Second Report on International course on Rare Disease Registries and FAIRification of data at source due at month 60

First Annual strategic report and Action plan for Pillar 2, including: Systematic surveys reports, QMS of Pillar 2 description, GDPR compliance report and sustainability planning reporting
Annual strategic report and Action plan for Pillar 2, including: Systematic surveys reports, QMS of Pillar 2 description, GDPR compliance report and sustainability planning reporting due at M9

First Annual report on implementation of training programs
First Annual reports on implementation of training programs due at month 13

Second Report from strategic workshop with national policy makers
Other lead beneficiary MUW 38Report from strategic workshop with national policy makers due at month 58

Second Report on core set of FAIR software tools and on extended set of unified FAIR data standards, applied in EJP RD
Second Report on core set of FAIR software tools and on extended set of unified FAIR data standards applied in EJP RD due at month 48

Third Scoping paper
Other lead beneficiaries ISCIII (44) and INSERM (1) Scoping paper due at month 31

Completed network analysis workflow (active node detection, lifestyle factor network evaluation and extended network analysis for drugs and toxic compounds)

Completed network analysis workflow active node detection lifestyle factor network evaluation and extended network analysis for drugs and toxic compounds

Fourth List of research and innovation needs requiring medium- or long-term approach and related Task Forces
List of research and innovation needs requiring medium or longterm approach and related Task Forces due at month 42

First report from the face-to-face ExCom and Policy Board meeting
Report from the face-to-face ExCom and Policy Board meeting due at M7

First Report on EURORDIS' Leadership Programme
First Report on EURORDIS' Leadership Programme due at month 12

Final Report of funded demonstration projects
Other lead beneficiaries: AP-HP (54), IOR (61) and ERN EpiCAREFinal Report of funded demonstration projects

First Report on training of patient representatives on scientific innovation and translational research in RD
First Report on training of patient representatives on scientific innovation and translational research in RD due at month 24

Second report from the face-to-face ExCom and Policy Board meeting
Report from the face-to-face ExCom and Policy Board meeting due at M19

Websites, patent fillings, videos etc. (6)

Third update Virtual platform of RD resources annotated with EJP ontological model
Third update of the Virtual platform of RD resources annotated with EJP ontological model due at month 48

Second update Virtual platform of RD resources annotated with EJP ontological model
Second update of the Virtual platform of RD resources annotated with EJP ontological model due at month 36

EJP RD website

EJP RD Newsletter

Virtual platform of RD resources annotated with EJP ontological model

First version of the Virtual platform of RD resources annotated with EJP ontological model due at month 12

First update Virtual platform of RD resources annotated with EJP ontological model

First update of the Virtual platform of RD resources annotated with EJP ontological model due at month 24

Other (12)

Fourth version Additional facilities integrated to resources regarding data deposition and access, including user guidelines and documentation

Other lead beneficiary: ELIXIR (76)Fourth version of the Additional facilities integrated to resources regarding data deposition and access, including user guidelines and documentation due at month 48

First Ontological model of resources metadata

First Ontological model of resources metadata due at mont 12

First version of Additional facilities integrated to resources regarding data deposition and access, including user guidelines and documentation

Other lead beneficiary ELIXIR (76) First version of Additional facilities integrated to resources regarding data deposition and access, including user guidelines and documentation due at month 12

Fifth version Additional facilities integrated to resources regarding data deposition and access, including user guidelines and documentation

Other lead beneficiary: ELIXIR (76)Fifth version of the Additional facilities integrated to resources regarding data deposition and access, including user guidelines and documentation due at month 60

Third version Additional facilities integrated to resources regarding data deposition and access, including user guidelines and documentation

Other lead beneficiary ELIXIR 76Third version of Additional facilities integrated to resources regarding data deposition and access including user guidelines and documentation due at month 36

Content of the first 5 online modules

Content of the full online course

Fifth Ontological model of resources metadata

Fifth Ontological model of resources metadata due at month 60

Second version Additional facilities integrated to resources regarding data deposition and access, including user guidelines and documentation

Other lead beneficiary: ELIXIR (76) Second version of Additional facilities integrated to resources regarding data deposition and access, including user guidelines and documentation due at month 24

Fourth Ontological model of resources metadata

Fourth Ontological model of resources metadata due at month 48

Second Ontological model of resources metadata

Second Ontological model of resources metadata due at month 24

Third Ontological model of resources metadata

Third Ontological model of resources metadata due at month 36

Publications

Peer reviewed articles (100)

CHARON: An Imaging-Based Diagnostic Algorithm to Navigate Through the Sea of Hereditary Degenerative Ataxias

Author(s): Alessandra Scaravilli, Mario Tranfa, Giuseppe Pontillo, Bernard Brais, Giovanna De Michele, Roberta La Piana, Francesco Saccà, Filippo Maria Santorelli, Matthis Synofzik, Arturo Brunetti, Sirio Cocozza
Published in: The Cerebellum, 2024, ISSN 1473-4230
Publisher: Springer Nature
DOI: 10.1007/s12311-024-01677-y

Common conditions of use elements. Atomic concepts for consistent and effective information governance

Author(s): Maria del Carmen Sanchez Gonzalez, Pim Kamerling, Mariapia Iermito, Sara Casati, Umar Riaz, Colin D. Veal, Monika Maini, Francis Jeanson, Oussama Mohammed Benhamed, Esther van Enckevort, Annalisa Landi, Yanis Mimouni, Clèmence Le Cornec, Domenico A. Coviello, Tiziana Franchin, Francesca Fusco, Jose Antonio Ramírez García, Loes F. M. van der Zanden, Alexander Bernier, Mark D. Wilkinson, Heimo Mu
Published in: Scientific Data, Issue 11, 2024, ISSN 2052-4463
Publisher: Springer Nature
DOI: 10.1038/s41597-024-03279-z

A FAIR catalog of ontology-driven conceptual models

Author(s): Tiago Prince Sales, Pedro Paulo F. Barcelos, Claudenir M. Fonseca, Isadora Valle Souza, Elena Romanenko, César Henrique Bernabé, Luiz Olavo Bonino da Silva Santos, Mattia Fumagalli, Joshua Kritz, João Paulo A. Almeida, Giancarlo Guizzardi
Published in: Data & Knowledge Engineering, Issue 147, 2023, Page(s) 102210, ISSN 0169-023X
Publisher: Elsevier BV
DOI: 10.1016/j.datak.2023.102210

Phenotypic features of RETREG1-related hereditary sensory autonomic neuropathy

Author(s): Arman Çakar; Gulandam Bagirova; Hacer Durmuş; Oya Uyguner; Yeşim Parman
Published in: Journal of the peripheral nervous system., 2023, ISSN 1529-8027
Publisher: -
DOI: 10.1111/jns.12581

Variant recurrence confirms the existence of a FBXO31 -related spastic-dystonic cerebral palsy syndrome

Author(s): Ivana Dzinovic, Matej Škorvánek, Petra Pavelekova, Chen Zhao, Boris Keren, Sandra Whalen, Somayeh Bakhtiari, Sheng Chih Jin, Michael C. Kruer, Robert Jech, Juliane Winkelmann, Michael Zech
Published in: Annals of Clinical and Translational Neurology, 2021, ISSN 2328-9503
Publisher: Wiley
DOI: 10.1002/acn3.51335

Consensus recommendations on mental health issues in Phelan-McDermid syndrome

Author(s): Ingrid D.C. van Balkom, Monica Burdeus-Olavarrieta, Jennifer Cooke, A. Graciela de Cuba, Alison Turner, Annick Vogels, Anna Maruani
Published in: European Journal of Medical Genetics, Issue 66, 2024, Page(s) 104770, ISSN 1769-7212
Publisher: Elsevier BV
DOI: 10.1016/j.ejmg.2023.104770

Innovative methodologies for rare diseases clinical trials

Author(s): Rima Nabbout, Ralf-Dieter Hilgers
Published in: Orphanet Journal of Rare Diseases, Issue 19, 2024, ISSN 1750-1172
Publisher: BioMed Central
DOI: 10.1186/s13023-024-03189-8

Generation of an induced pluripotent stem cell line carrying biallelic deletions (SCTCi019-B) in ALDH7A1 using CRISPR/Cas9

Author(s): Imke M.E. Schuurmans, Ka M. Wu, Clara D.M. van Karnebeek, Nael Nadif Kasri, Alejandro Garanto
Published in: Stem Cell Research, Issue 71, 2024, Page(s) 103173, ISSN 1873-5061
Publisher: Elsevier
DOI: 10.1016/j.scr.2023.103173

Lissencephaly: Update on diagnostics and clinical management
Author(s): Matti Koenig, William B. Dobyns, Nataliya Di Donato
Published in: European Journal of Paediatric Neurology, Issue 35, 2022, Page(s) 147-152, ISSN 1090-3798
Publisher: W. B. Saunders Co., Ltd.
DOI: 10.1016/j.ejpn.2021.09.013

Extending inherited metabolic disorder diagnostics with biomarker interaction visualizations
Author(s): Denise N. Slenter, Irene M. G. M. Hemel, Chris T. Evelo, Jörgen Bierau, Egon L. Willighagen, Laura K. M. Steinbusch
Published in: Orphanet Journal of Rare Diseases, Issue 18, 2023, ISSN 1750-1172
Publisher: BioMed Central
DOI: 10.1186/s13023-023-02683-9

Ensembl 2024
Author(s): Peter W Harrison, M Ridwan Amode, Olanrewaju Austine-Orimoloye, Andrey G Azov, Matthieu Barba, If Barnes, Arne Becker, Ruth Bennett, Andrew Berry, Jyothish Bhai, Simarpreet Kaur Bhurji, Sanjay Boddu, Paulo R Branco Lins, Lucy Brooks, Shashank Budhanuru Ramaraju, Lahcen I Campbell, Manuel Carbajo Martinez, Mehrnaz Charkhchi, Kapeel Chougule, Alexander Cockburn, Claire Davidson, Nishadi H De S
Published in: Nucleic Acids Research, Issue 52, 2024, Page(s) D891-D899, ISSN 0305-1048
Publisher: Oxford University Press
DOI: 10.1093/nar/gkad1049

Remote visualization of large-scale genomic alignments for collaborative clinical research and diagnosis of rare diseases
Author(s): Alberto Corvò, Leslie Matalonga, Dylan Spalding, Alexander Senf, Steven Laurie, Daniel Picó-Amador, Marcos Fernandez-Callejo, Ida Paramonov, Anna Foix Romero, Emilio Garcia-Rios, Jorge Izquierdo Ciges, Anand Mohan, Coline Thomas, Andres Felipe Silva Valencia, Csaba Halmagyi, Mallory Ann Freeberg, Ana Töpf, Rita Horvath, Gary Saunders, Ivo Gut, Thomas Keane, Davide Piscia, Sergi Beltran
Published in: Cell Genomics, Issue 3, 2023, Page(s) 100246, ISSN 2666-979X

Publisher: Elsevier Inc.
DOI: 10.1016/j.xgen.2022.100246

Genome Aggregation Database Version 4—New Challenges of Variant Analysis in Movement Disorders

Author(s): Elisabetta Indelicato, Luigi Michele Romito, Philip Harrer, Nico Golfrè Andreasi, Isabel Colangelo, Robert Kopajtich, Juliane Winkelmann, Holger Prokisch, Barbara Garavaglia, Michael Zech
Published in: Movement Disorders, Issue 39, 2024, Page(s) 1237-1238, ISSN 0885-3185
Publisher: John Wiley & Sons Inc.
DOI: 10.1002/mds.29797

The phenotypic and genetic spectrum of patients with heterozygous mutations in cyclin M2 (CNNM2)

Author(s): Gijs A. C. Franken, Dominik Müller, Cyril Mignot, Boris Keren, Jonathan Lévy, Anne-Claude Tabet, David Germanaud, María-Isabel Tejada, Hester Y. Kroes, Rutger A. J. Nievelstein, Elise Brimble, Maria Ruzhnikov, Felix Claverie-Martin, Maria Szczepańska, Martin Ćuk, Femke Latta, Martin Konrad, Luis A. Martínez-Cruz, René J. M. Bindels, Joost G. J. Hoenderop, Karl-Peter Schlingmann, J
Published in: Human Mutation, 2021, ISSN 1059-7794
Publisher: John Wiley & Sons Inc.
DOI: 10.1002/humu.24182

Definition and clinical variability of SHANK3-related Phelan-McDermid syndrome

Author(s): Michael Schön, Pablo Lapunzina, Julián Nevado, Teresa Mattina, Cecilia Gunnarsson, Kinga Hadzsiev, Chiara Verpelli, Thomas Bourgeron, Sarah Jesse, Conny M.A. van Ravenswaaij-Arts, Raoul C. Hennekam
Published in: European Journal of Medical Genetics, Issue 66, 2023, Page(s) 104754, ISSN 1769-7212
Publisher: Elsevier BV
DOI: 10.1016/j.ejmg.2023.104754

Evaluating FAIR maturity through a scalable, automated, community-governed framework

Author(s): Mark D. Wilkinson, Michel Dumontier, Susanna-Assunta Sansone, Luiz Olavo Bonino da Silva Santos, Mario Prieto, Dominique Batista, Peter McQuilton, Tobias Kuhn, Philippe Rocca-Serra, Mercè Crosas, Erik Schultes
Published in: Scientific Data, Issue 6/1, 2019, ISSN 2052-4463
Publisher: Nature
DOI: 10.1038/s41597-019-0184-5

Combined Single Gene Testing and Genome Sequencing as an Effective Diagnostic Approach for Anophthalmia and Microphthalmia Patients

Author(s): Rabia Basharat, Kim Rodenburg, María Rodríguez-Hidalgo, Afeefa Jarral, Ehsan Ullah, Jordi Corominas, Christian Gilissen, Syeda Tatheer Zehra, Usman Hameed, Muhammad Ansar, Suzanne E. de Bruijn
Published in: Genes, Issue 14, 2023, Page(s) 1573, ISSN 2073-4425
Publisher: Multidisciplinary Digital Publishing Institute (MDPI)
DOI: 10.3390/genes14081573

Ensembl 2021
Author(s): Kevin L Howe, Premanand Achuthan, James Allen, Jamie Allen, Jorge Alvarez-Jarreta, M Ridwan Amode, Irina M Armean, Andrey G Azov, Ruth Bennett, Jyothish Bhai, Konstantinos Billis, Sanjay Boddu, Mehrnaz Charkhchi, Carla Cummins, Luca Da Rin Fioretto, Claire Davidson, Kamalkumar Dodiya, Bilal El Houdaigui, Reham Fatima, Astrid Gall, Carlos Garcia Giron, Tiago Grego, Cristina Guijarro-Clarke, Lea
Published in: Nucleic Acids Research, Issue 49/D1, 2020, Page(s) D884-D891, ISSN 0305-1048
Publisher: Oxford University Press
DOI: 10.1093/nar/gkaa942

Ensembl 2022
Author(s): Fiona Cunningham, James E Allen, Jamie Allen, Jorge Alvarez-Jarreta, M Ridwan Amode, Irina M Armean, Olanrewaju Austine-Orimoloye, Andrey G Azov, If Barnes, Ruth Bennett, Andrew Berry, Jyothish Bhai, Alexandra Bignell, Konstantinos Billis, Sanjay Boddu, Lucy Brooks, Mehrnaz Charkhchi, Carla Cummins, Luca Da Rin Fioretto, Claire Davidson, Kamalkumar Dodiya, Sarah Donaldson, Bilal El Houdaigui
Published in: Nucleic Acids Research, Issue 50, 2023, Page(s) D988-D995, ISSN 0305-1048
Publisher: Oxford University Press
DOI: 10.1093/nar/gkab1049

A neutral comparison of statistical methods for analyzing longitudinally measured ordinal outcomes in rare diseases
Author(s): Martin Geroldinger, Johan Verbeeck, Konstantin E. Thiel, Geert Molenberghs, Arne C. Bathke, Martin Laimer, Georg Zimmermann
Published in: Biometrical Journal, Issue 66, 2024, ISSN 0323-3847
Publisher: John Wiley & Sons Ltd.
DOI: 10.1002/bimj.202200236

Sustainable approaches for drug repurposing in rare diseases: recommendations from the IRDiRC Task Force
Author(s): Galliano Zanello, Diego Ardigò, Florence Guillot, Anneliene H. Jonker, Oxana Iliach, Hervé Nabarette, Daniel O'Connor, Virginie Hivert
Published in: Rare Disease and Orphan Drugs Journal, Issue 2, 2024, Page(s)

9, ISSN 2771-2893
Publisher: OAE Publishing Inc.
DOI: 10.20517/rdodj.2023.04

Exploring the Therapeutic Potential of Ectoine in Duchenne Muscular Dystrophy: Comparison with Taurine, a Supplement with Known Beneficial Effects in the mdx Mouse
Author(s): Caroline Merckx, Jana Zschüntzsch, Stefanie Meyer, Robrecht Raedt, Hanne Verschuere, Jens Schmidt, Boel De Paepe, Jan L. De Bleecker
Published in: International Journal of Molecular Sciences, Issue 23, 2024, Page(s) 9567, ISSN 1422-0067
Publisher: Multidisciplinary Digital Publishing Institute (MDPI)
DOI: 10.3390/ijms23179567

Levels of Neurofilament Light at the Preataxic and Ataxic Stages of Spinocerebellar Ataxia Type 1
Author(s): Carlo Wilke, David Mengel, Ludger Schöls, Holger Hengel, Maria Rakowicz, Thomas Klockgether, Alexandra Durr, Alessandro Filla, Bela Melegh, Rebecca Schüle, Kathrin Reetz, Heike Jacobi, Matthis Synofzik
Published in: Neurology, Issue 98, 2024, ISSN 0028-3878
Publisher: Lippincott Williams & Wilkins Ltd.
DOI: 10.1212/wnl.0000000000200257

Additional file 2 of Clinically relevant combined effect of polygenic background, rare pathogenic germline variants, and family history on colorectal cancer incidence
Author(s): Hassanin, Emadeldin; Spier, Isabel; Bobbili, Dheeraj R.; Aldisi, Rana; Klinkhammer, Hannah; David, Friederike; Dueñas, Nuria; Hüneburg, Robert; Perne, Claudia; Brunet, Joan; Capella, Gabriel; Nöthen, Markus M.; Forstner, Andreas J.; Mayr, Andreas; Krawitz, Peter; May, Patrick; Aretz, Stefan; Maj, Carlo
Published in: BMC Medical Genomics, Issue 1, 2023, ISSN 1755-8794
Publisher: BioMed Central
DOI: 10.6084/m9.figshare.22620470.v1

Additional file 1 of The de novo FAIRification process of a registry for vascular anomalies
Author(s): Groenen, Karlijn H. J.; Jacobsen, Annika; Kersloot, Martijn G.; dos Santos Vieira, Bruna; van Enckevort, Esther; Kaliyaperumal, Rajaram; Arts, Derk L.; ‘t Hoen, Peter A. C.; Cornet, Ronald; Roos, Marco; Kool, Leo Schultze
Published in: Orphanet Journal of Rare Diseases, Issue 1, 2021, ISSN 1750-1172
Publisher: BioMed Central
DOI: 10.6084/m9.figshare.16570885

Biallelic variants in the SORD gene are one of the most common causes of hereditary neuropathy among Czech patients

Author(s): P. Laššuthová, R. Mazanec, D. Staněk, L. Sedláčková, B. Plevová, J. Haberlová, P. Seeman
Published in: Scientific Reports, Issue 11, 2022, ISSN 2045-2322
Publisher: Nature Publishing Group
DOI: 10.1038/s41598-021-86857-0

Detection of Telomeric DNA:RNA Hybrids Using TeloDRIP-qPCR
Author(s): Ilaria Rosso, Fabrizio d'Adda di Fagagna
Published in: International Journal of Molecular Sciences, Issue 21/24, 2020, Page(s) 9774, ISSN 1422-0067
Publisher: Multidisciplinary Digital Publishing Institute (MDPI)
DOI: 10.3390/ijms21249774

Integrative analysis of multi-omics data reveals importance of collagen and the PI3K AKT signalling pathway in CAKUT
Author(s): Jumamurat R. Bayjanov, Cenna Doornbos, Ozan Ozisik, Woosub Shin, Núria Queralt-Rosinach, Daphne Wijnbergen, Jean-Sébastien Saulnier-Blache, Joost P. Schanstra, Bénédicte Buffin-Meyer, Julie Klein, José M. Fernández, Rajaram Kaliyaperumal, Anaïs Baudot, Peter A. C. 't Hoen, Friederike Ehrhart
Published in: Scientific Reports, Issue 14, 2024, ISSN 2045-2322
Publisher: Nature Publishing Group
DOI: 10.1038/s41598-024-71721-8

orsum: A Python package for filtering and comparing enrichment analyses using comprehensible rules
Author(s): Ozan Ozisik, Morgane Térézol, Anaïs Baudot
Published in: BMC Bioinformatics, Issue 23(1), 2022, Page(s) 293, ISSN 1471-2105
Publisher: BioMed Central
DOI: 10.1186/s12859-022-04828-2

Towards FAIRification of sensitive and fragmented rare disease patient data: challenges and solutions in European reference network registries.
Author(s): Bruna dos Santos Vieira, César H. Bernabé, Shuxin Zhang, Haitham Abaza, Nirupama Benis, Alberto Cámara, Ronald Cornet, Clémence M. A. Le Cornec, Peter A. C. 't Hoen, Franz Schaefer, K. Joeri van der Velde, Morris A. Swertz, Mark D. Wilkinson, Annika Jacobsen & Marco Roos
Published in: Orphanet Journal of Rare Diseases, 2022, ISSN 1750-1172
Publisher: BioMed Central
DOI: 10.1186/s13023-022-02558-5

Assessing resolvability, parsability, and consistency of RDF resources: a use case in rare diseases

Author(s): Shuxin Zhang, Nirupama Benis, Ronald Cornet
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Building Expertise on FAIR Through Evolving Bring Your Own Data (BYOD) Workshops: Describing the Data, Software, and Management-focused Approaches and Their Evolution
Author(s): César H. Bernabé, Lieze Thielemans, Rajaram Kaliyaperumal, Claudio Carta, Shuxin Zhang, Celia W.G. van Gelder, Nirupama Benis, Luiz Olavo Bonino da Silva Santos, Ronald Cornet, Bruna dos Santos Vieira, Nawel Lalout, Ines Henriques, Alberto Cámara Ballesteros, Kees Burger, Martijn G. Kersloot, Friederike Ehrhart, Esther van Enckevort, Chris T. Evelo, Alasdair J. G. Gray, Marc Hanauer, Kristina H
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The case for open science: rare diseases
Author(s): Yaffa R Rubinstein, Peter N Robinson, William A Gahl, Paul Avillach, Gareth Baynam, Helene Cederroth, Rebecca M Goodwin, Stephen C Groft, Mats G Hansson, Nomi L Harris, Vojtech Huser, Deborah Mascalzoni, Julie A McMurry, Matthew Might, Christoffer Nellaker, Barend Mons, Dina N Paltoo, Jonathan Pevsner, Manuel Posada, Alison P Rockett-Frase, Marco Roos, Tamar B Rubinstein, Domenica Taruscio, Esther
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Publisher: Oxford Academic
DOI: 10.1093/jamiaopen/ooaa030

Dystonia in <scp>ATP</scp> Synthase Defects: Reconnecting Mitochondria and Dopamine
Author(s): Elisabetta Indelicato, Sylvia Boesch, Niccolo' E. Mencacci, Daniele Ghezzi, Holger Prokisch, Juliane Winkelmann, Michael Zech
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Ten quick tips for building FAIR workflows
Author(s): Casper de Visser, Lennart F. Johansson, Purva Kulkarni, Hailiang Mei, Pieter Neerincx, K. Joeri van der Velde, Péter Horvatovich, Alain J. van Gool, Morris A. Swertz, Peter A. C. ‘t Hoen, Anna Niehues
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Heterogeneous Phenotypic Evolution in <scp>ANO3</scp>-Related Dystonia Due to the Recurrent <scp>p.Glu510Lys</scp> Variant
Author(s): Elisabetta Indelicato, Sylvia Boesch, Michael Zech
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Extreme phenotypic heterogeneity in non-expansion spinocerebellar ataxias
Author(s): Paulina Cunha, Emilien Petit, Marie Coutelier, Giulia Coarelli, Caterina Mariotti, Jennifer Faber, Judith Van Gaalen, Joana Damasio, Zofia Fleszar, Michele Tosi, Clarissa Rocca, Giovanna De Michele, Martina Minnerop, Claire Ewenczyk, Filippo M. Santorelli, Anna Heinzmann, Thomas Bird, Matthias Amprosi, Elisabetta Indelicato, Alberto Benussi, Perrine Charles, Claudia Stendel, Silvia Romano, Marina
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Publisher: University of Chicago Press
DOI: 10.1016/j.ajhg.2023.05.009

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Author(s): Martin FJ et al
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Diagnostic yield from cardiac gene testing for inherited cardiac conditions and re-evaluation of pre-ACMG variants of uncertain significance
Author(s): Jane Murphy, Claire W. Kirk, Deborah M. Lambert, Catherine McGorrian, Roddy Walsh, Terri P. McVeigh, Terence Prendiville, Deirdre Ward, Joseph Galvin, Sally Ann Lynch
Published in: Irish Journal of Medical Science (1971 -), Issue 193, 2024, Page(s) 1775-1785, ISSN 0021-1265
Publisher: Royal Academy of Medicine in Ireland
DOI: 10.1007/s11845-024-03650-4

Drug repurposing in Rett and Rett-like syndromes: a promising yet underrated opportunity?
Author(s): Claudia Fuchs, Peter A. C. ‘t Hoen, Annelieke R. Müller, Friederike Ehrhart, Clara D. M. Van Karnebeek
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Recommendations from the IRDiRC Working Group on methodologies to assess the impact of diagnoses and therapies on rare disease patients
Author(s): Galliano Zanello; Chun-Hung Chan; David A. Pearce; IRDiRC Working Group
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Towards the international interoperability of clinical research networks for rare diseases: recommendations from the IRDiRC Task Force
Author(s): Rima Nabbout, Galliano Zanello, Dixie Baker, Lora Black, Isabella Brambilla, Orion J. Buske, Laurie S. Conklin, Elin Haf Davies, Daria Julkowska, Yeonju Kim, Thomas Klopstock, Harumasa Nakamura, Kim G. Nielsen, Anne R. Pariser, Jose Carlos Pastor, Maurizio Scarpa, Maureen Smith, Domenica Taruscio, Stephen Groft
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Consensus recommendations on counselling in Phelan-McDermid syndrome, with special attention to recurrence risk and to ring chromosome 22
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Age-dependent neurodegeneration and neuroinflammation in a genetic A30P/A53T double-mutated α-synuclein mouse model of Parkinson's disease
Author(s): Lisa Rauschenberger, Jennifer Behnke, Alexander Grotemeyer, Susanne Knorr, Jens Volkmann, Chi Wang Ip
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Synthetic datasets for open software development in rare disease research

Author(s): Ibraheem Al-Dhamari, Hammam Abu Attieh, Fabian Prasser
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The European joint programme on rare diseases: building the rare diseases research ecosystem
Author(s): Yanis Mimouni, Juliane Halftermeyer, Yanna Petton, Pauline Adam, Clément Moreau, Ana Rath, Roseline Favresse, Birute Tumiene, Daria Julkowska
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Head movement dynamics in dystonia: a multi-centre retrospective study using visual perceptive deep learning
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The use of foundational ontologies in biomedical research
Author(s): César H. Bernabé; Núria Queralt-Rosinach; Vítor E. Silva Souza; Luiz Olavo Bonino da Silva Santos; Barend Mons; Annika Jacobsen; Marco Roos
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Artificial intelligence-enhanced electrocardiography derived body mass index as a predictor of future cardiometabolic disease
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The FAIR Data Point: Interfaces and Tooling

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De novo variants in neurodevelopmental disorders—experiences from a tertiary care center

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Clinically relevant combined effect of polygenic background, rare pathogenic germline variants, and family history on colorectal cancer incidence

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WikiPathways: connecting communities

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Exploring pathway interactions to detect molecular mechanisms of disease: 22q11.2 deletion syndrome

Author(s): Woosub Shin, Martina Kutmon, Eleni Mina, Therese van Amelsvoort, Chris T Evelo, Friederike Ehrhart

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Peripheral temperature dysregulation associated with functionally altered NaV1.8 channels
Author(s): Simon Loose, Annette Lischka, Samuel Kuehs, Carla Nau, Stefan H. Heinemann, Ingo Kurth, Enrico Leipold
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Description of Osmolyte Pathways in Maturing Mdx Mice Reveals Altered Levels of Taurine and Sodium/Myo-Inositol Co-Transporters
Author(s): Caroline Merckx, Gwenny Cosemans, Jana Zschüntzsch, Robrecht Raedt, Jens Schmidt, Boel De Paepe, Jan L. De Bleecker
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Publisher: Multidisciplinary Digital Publishing Institute (MDPI)
DOI: 10.3390/ijms23063251

SUsPECT: a pipeline for variant effect prediction based on custom long-read transcriptomes for improved clinical variant annotation
Author(s): Renee Salz, Nuno Saraiva-Agostinho, Emil Vorsteveld, Caspar I. van der Made, Simone Kersten, Merel Stemerdink, Jamie Allen, Pieter-Jan Volders, Sarah E. Hunt, Alexander Hoischen, Peter A.C. ’t Hoen
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Overlap of vitamin A and vitamin D target genes with CAKUT-related processes [version 2; peer review: 2 approved, 2 approved with reservations]
Author(s): Chris T. Evelo; Alberto Mantovani; Anaïs Baudot; Friederike Ehrhart; Ozan Ozisik
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ABCA4 Variant c.5714+5G>A in Trans With Null Alleles Results in Primary RPE Damage
Author(s): Jana Sajovic, Andrej Meglič, Zelia Corradi, Mubeen Khan, Aleš Maver, Martina Jarc Vidmar, Marko Hawlina, Frans P. M. Cremers, Ana Fakin
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FAIR Data Point: A FAIR-Oriented Approach for Metadata Publication
Author(s): Luiz Olavo Bonino da Silva Santos, Kees Burger, Rajaram Kaliyaperumal, Mark D. Wilkinson
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Use Cases Requiring Privacy-Preserving Record Linkage in Paediatric Oncology
Author(s): Dieter Hayn, Karl Kreiner, Emanuel Sandner, Martin Baumgartner, Bernhard Jammerbund, Markus Falgenhauer, Vanessa Düster, Priyanka Devi-Marulkar, Gudrun Schleiermacher, Ruth Ladenstein, Guenter Schreier
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Establishing the first pan-European Registry for rare bone and mineral disorders: EuRR-Bone
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The RD-Connect Genome-Phenome Analysis Platform: Accelerating diagnosis, research, and gene discovery for rare diseases
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DOI: 10.1002/humu.24353

Getting your DUCs in a row - standardising the representation of Digital Use Conditions
Author(s): Francis Jeanson, Spencer J. Gibson, Pinar Alper, Alexander Bernier, J. Patrick Woolley, Daniel Mietchen, Andrzej Strug, Regina Becker, Pim Kamerling, Maria del Carmen Sanchez Gonzalez, Nancy Mah, Ann Novakowski,

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A catalogue of 863 Rett-syndrome-causing MECP2 mutations and lessons learned from data integration
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Targeting shared molecular etiologies to accelerate drug development for rare diseases
Author(s): Galliano Zanello; Macarena Garrido-Estepa; Ana Crespo; Daniel O'Connor; Rima Nabbout; Christina Waters; Anthony Hall; Maurizio Taglialatela; Chun-Hung Chan; David A Pearce; Marc Dooms; Philip John Brooks
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A Resource for Guiding Data Stewards to Make European Rare Disease Patient Registries FAIR
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DOI: 10.5334/dsj-2023-012

Consensus recommendations on chewing, swallowing and gastrointestinal problems in Phelan-McDermid syndrome
Author(s): Ausra Matuleviciene; Kamile Siauryte; Els Kuiper; Andreas M. Grabrucker
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A Generic Workflow for the Data FAIRification Process
Author(s): Annika Jacobsen, Rajaram Kaliyaperumal, Luiz Olavo Bonino da Silva Santos, Barend Mons, Erik Schultes, Marco Roos, Mark Thompson
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FAIR Principles: Interpretations and Implementation Considerations
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A pipeline-friendly software tool for genome diagnostics to prioritize genes by matching patient symptoms to literature
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System-level analysis of genes mutated in muscular dystrophies reveals a functional pattern associated with muscle weakness distribution
Author(s): Ozan Ozisik, Svetlana Gorokhova, Mathieu Cerino, Marc Bartoli, Anaïs Baudot
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DOI: 10.1038/s41598-024-60761-9

Leveraging Biolink as a “Rosetta Stone” Between C-Path and EJP-RD Semantic Models Provides Emergent Interoperability

Author(s): Pablo Alarcon, Ian Braun, Emily Hartley, Daniel Olson, Nirupama Benis, Ronald Cornet, Mark Wilkinson, Ramona L. Walls
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IgLON5 deficiency produces behavioral alterations in a knockout mouse model
Author(s): Jon Landa, Ana Beatriz Serafim, Mercedes Alba, Estibaliz Maudes, Laura Molina-Porcel, Anna Garcia-Serra, Francesco Mannara, Josep Dalmau, Francesc Graus, Lidia Sabater
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Scale for the Assessment and Rating of Ataxia (SARA): Development of a Training Tool and Certification Program
Author(s): Marcus Grobe-Einsler, Arian Taheri Amin, Jennifer Faber, Hartmut Völkel, Matthis Synofzik, Thomas Klockgether
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Composite endpoints, including patient reported outcomes, in rare diseases
Author(s): Johan Verbeeck; Maya Dirani; Johann W. Bauer; Ralf-Dieter Hilgers; Geert Molenberghs; Rima Nabbout
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Genetics meets function in sodium channel-related pain disorders
Author(s): Jannis Körner, Natja Haag, Ingo Kurth, Angelika Lampert
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Additional file 1 of Towards the international interoperability of clinical research networks for rare diseases: recommendations from the IRDiRC Task Force
Author(s): Nabbout, Rima; Zanello, Galliano; Baker, Dixie; Black, Lora; Brambilla, Isabella; Buske, Orion J.; Conklin, Laurie S.; Davies, Elin Haf; Julkowska, Daria; Kim, Yeonju; Klopstock, Thomas; Nakamura, Harumasa; Nielsen, Kim G.; Pariser, Anne R.; Pastor, Jose Carlos; Scarpa, Maurizio; Smith,

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CHD8-related disorders redefined: an expanding spectrum of dystonic phenotypes
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Genetic and drug-induced hypomagnesemia: different cause, same mechanism
Author(s): Willem Bosman, Joost G. J. Hoenderop, Jeroen H. F. de Baaij
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Incidence and Prevalence of Fibrous Dysplasia/McCune-Albright Syndrome: A Nationwide Registry-Based Study in Denmark
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Unraveling dystonia circuitry in rodent models using novel neuromodulation techniques
Author(s): Lisa Rauschenberger, Chi Wang Ip
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Consensus recommendations on altered sensory functioning in Phelan-McDermid syndrome
Author(s): Margreet Walinga, Sarah Jesse, Norma Alhambra, Griet Van Buggenhout
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A Simple Standard for Sharing Ontological Mappings (SSSOM)

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Multifactorial Assessment of Motor Behavior in Rats after Unilateral Sciatic Nerve Crush Injury

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Corrigendum: Reactive astrogliosis in the era of single-cell transcriptomics

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Published in: Frontiers in Cellular Neuroscience, Vol 17 (2023), Issue 2, 2023, ISSN 1662-5102
Publisher: Frontiers Research Foundation
DOI: 10.3389/fncel.2023.1212975

Consensus recommendations on organization of care for individuals with Phelan-McDermid syndrome

Author(s): A.M. van Eeghen, D. Stemkens, José Ramón Fernández-Fructuoso, A. Maruani, K. Hadzsiev, I.D.C. van Balkom, C.M.W. Gaasterland, M.J. Klein Haneveld, Klea Vyshka, A. Hugon, A.M. van Eeghen, Norma Alhambra, Britt-Marie Anderlid, Stephanie Andres, Emmelien Aten, Rui Barbosa Guedes, Maria C. Bonaglia, Thomas Bourgeron, Monica Burdeus-Olavarrieta, Maya J. Carbin, Jennifer Cooke, Robert J. Damstra, Ire
Published in: European Journal of Medical Genetics, Issue 66, 2023, Page(s) 104747, ISSN 1769-7212
Publisher: Elsevier BV
DOI: 10.1016/j.ejmg.2023.104747

Automated approach for quality assessment of RDF resources

Author(s): Shuxin Zhang, Nirupama Benis, Ronald Cornet
Published in: BMC Medical Informatics and Decision Making, Issue 23, 2024, ISSN 1472-6947

Publisher: BioMed Central
DOI: 10.1186/s12911-023-02182-8

Machine learning in Huntington's disease: exploring the Enroll-HD dataset for prognosis and driving capability prediction

Author(s): Jasper Ouwerkerk, Stephanie Feleus, Kasper F. van der Zwaan, Yunlei Li, Marco Roos, Willeke M. C. van Roon-Mom, Susanne T. de Bot, Katherine J. Wolstencroft, Eleni Mina
Published in: Orphanet Journal of Rare Diseases, Issue 18, 2023, ISSN 1750-1172
Publisher: BioMed Central
DOI: 10.1186/s13023-023-02785-4

Interactive neuroinflammation pathways and transcriptomics-based identification of drugs and chemical compounds for schizophrenia

Author(s): Lisa Koole, Pilar Martinez-Martinez, Therese van Amelsvoort, Chris T. Evelo, Friederike Ehrhart
Published in: The World Journal of Biological Psychiatry, Issue 25, 2024, Page(s) 116-129, ISSN 1562-2975
Publisher: WFSBP
DOI: 10.1080/15622975.2023.2281514

Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity

Author(s): Leslie Matalonga, Steven Laurie, Anastasios Papakonstantinou, Davide Piscia, Elisabetta Mereu, Gemma Bullich, Rachel Thompson, Rita Horvath, Luis Pérez-Jurado, Olaf Riess, Ivo Gut, Gert-Jan van Ommen, Hanns Lochmüller, Sergi Beltran, Alessandra Renieri, Ali Dursun, Antoni Matilla-Duenas, Bru Cormand, Carlo Rivolta, Carmen Ayuso, Carmen Espinós, Christian Scerri, Dilek Yalnizoglu, Doriette Soler
Published in: The Journal of Molecular Diagnostics, Issue 22/9, 2020, Page(s) 1205-1215, ISSN 1525-1578
Publisher: American Society for Investigative Pathology
DOI: 10.1016/j.jmoldx.2020.06.008

ABCA4 c.6480-35A>G, a novel branchpoint variant associated with Stargardt disease

Author(s): María Rodríguez-Hidalgo, Suzanne E. de Bruijn, Zelia Corradi, Kim Rodenburg, Araceli Lara-López, Alicia Valverde-Megías, Almudena Ávila-Fernández, Lidia Fernandez-Caballero, Marta Del Pozo-Valero, Jordi Corominas, Christian Gilissen, Cristina Irigoyen, Frans P. M. Cremers, Carmen Ayuso, Javier Ruiz-Ederra, Susanne Roosing
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Publisher: Frontiers Media
DOI: 10.3389/fgene.2023.1234032

Appraising the Role of Astrocytes as Suppliers of Neuronal Glutathione Precursors
Author(s): Dolores Pérez-Sala, María A. Pajares
Published in: International Journal of Molecular Sciences, Issue 24, 2023, Page(s) 8059, ISSN 1422-0067
Publisher: Multidisciplinary Digital Publishing Institute (MDPI)
DOI: 10.3390/ijms24098059

Gene-environment interaction elicits dystonia-like features and impaired translational regulation in a DYT-TOR1A mouse model
Author(s): Colette Reinhold, Susanne Knorr, Rhonda L. McFleder, Lisa Rauschenberger, Muthuraman Muthuraman, Panagiota Arampatzi, Tom Gräfenhan, Andreas Schlosser, Michael Sendtner, Jens Volkmann, Chi Wang Ip
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Publisher: Academic Press
DOI: 10.1016/j.nbd.2024.106453

Optimizing designs in clinical trials with an application in treatment of Epidermolysis bullosa simplex, a rare genetic skin disease
Author(s): Joakim Nyberg, Andrew C. Hooker, Georg Zimmermann, Johan Verbeeck, Martin Geroldinger, Konstantin Emil Thiel, Geert Molenberghs, Martin Laimer, Verena Wally
Published in: Computational Statistics & Data Analysis, Issue 199, 2024, Page(s) 108015, ISSN 0167-9473
Publisher: Elsevier BV
DOI: 10.1016/j.csda.2024.108015

Beyond Pathway Analysis: Identification of Active Subnetworks in Rett Syndrome
Author(s): Ryan A. Miller, Friederike Ehrhart, Lars M. T. Eijssen, Denise N. Slenter, Leopold M. G. Curfs, Chris T. Evelo, Egon L. Willighagen, Martina Kutmon
Published in: Frontiers in Genetics, Issue 10, 2019, ISSN 1664-8021
Publisher: Frontiers Media
DOI: 10.3389/fgene.2019.00059

Statistical recommendations for count, binary, and ordinal data in rare disease cross-over trials
Author(s): Martin Geroldinger; Johan Verbeeck; Andrew C. Hooker; Konstantin E. Thiel; Geert Molenberghs; Joakim Nyberg; Johann Bauer; Martin Laimer; Verena Wally; Arne C. Bathke; Georg Zimmermann
Published in: Orphanet J Rare Dis, Issue 18(1):391, 2023, ISSN 1750-1172
Publisher: BioMed Central
DOI: 10.1186/s13023-023-02990-1

Semantic modelling of Common Data Elements for Rare Disease registries, and a prototype workflow for their deployment over registry data

Author(s): Rajaram Kaliyaperumal; Mark D Wilkinson; Pablo Alarcón Moreno; Nirupama Benis; Ronald Cornet; Bruna dos Santos Vieira; Michel Dumontier; César Henrique Bernabé; Annika Jacobsen; Clémence M. A. Le Cornec; Mario Prieto Godoy; Núria Queralt-Rosinach; Leo J Schultze Kool; Morris A Swertz; Philip van Damme; Joeri K van der Velde; Nawel van Lin; Shuxin Zhang; Marco Roos
Published in: Journal of Biomedical Semantics, 2022, ISSN 2041-1480
Publisher: BioMed Central
DOI: 10.1186/s13326-022-00264-6

Protein domains provide a new layer of information for classifying human variations in rare diseases

Author(s): Mélanie Corcuff; Marc Garibal; Jean-Pierre Desvignes; Céline Guien; Coralie Grattepanche; Gwenaëlle Collod-Béroud; Estelle Ménoret; David Salgado; Christophe Béroud; Christophe Béroud
Published in: Front Bioinform, 2023, ISSN 2673-7647
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Copy number variant risk loci for schizophrenia converge on the BDNF pathway

Author(s): Friederike Ehrhart, Ana Silva, Therese van Amelsvoort, Emma von Scheibler, Chris Evelo, David E.J Linden
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Publisher: WFSBP
DOI: 10.1080/15622975.2024.2327027

Other (27)

SUsPECT: A pipeline for variant effect prediction based on custom long-read transcriptomes for improved clinical variant

Author(s): Renee Salz, Nuno Saraiva-Agostinho, Emil Vorsteveld, Caspar I. van der Made, Simone Kersten, Merel Stemerdink, Jamie Allen, Pieter-Jan Volders, Sarah E. Hunt, Alexander Hoischen, Peter A.C. ’t Hoen
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DOI: 10.1101/2022.10.23.513417

A Systems Biology Workflow to Support the Diagnosis of Inherited Metabolic Disorders: a study on Pyrimidine and Urea Cycle disorders

Author(s): Denise N. Slenter, Irene M.G.M. Hemel, Chris T. Evelo, Jörgen Bierau, Egon L. Willighagen, Laura K.M. Steinbusch
Published in: medRxiv, 2022
Publisher: Cold Spring Harbour Laboratory
DOI: 10.1101/2022.01.31.21265847

Lightweight Distributed Provenance Model for Complex Real–world Environments

Author(s): Rudolf Wittner; Cecilia Mascia; Matej Gallo; Francesca Frexia; Heimo Müller; Markus Plass; Jörg Geiger; Petr Holub
Published in: Crossref, Issue 1, 2022
Publisher: Sci Data
DOI: 10.1038/s41597-022-01537-6

A polymorphic AT-repeat causes frequent allele dropout for an MME mutational hotspot exon

Author(s): Helle Høyer; Hilde T Hilmarsen; Raute Sunder-Plassmann; Geir J Braathen; Peter M Andersen; Christian Beetz; Sandra Hacker; Øystein L Holla; Ingo Kurth; Wolfgang N Löscher; Simone B C F Reiter; Sabine Rudnik-Schöneborn; Linda Strand; Reinhard Windhager; Martina Witsch-Baumgartner; Jan Senderek; Michaela Auer-Grumbach
Published in: Crossref, Issue Oct;59(10):1024-1026, 2022
Publisher: J Med Genet
DOI: 10.1136/jmedgenet-2021-108281

Alpha-Synuclein is Involved in <scp>DYT1</scp> Dystonia Striatal Synaptic Dysfunction

Author(s): Giulia Ponterio; Gaia Faustini; Ilham El Atiallah; Giuseppe Sciamanna; Maria Meringolo; Annalisa Tassone; Paola Imbriani; Silvia Cerri; Giuseppina Martella; Paola Bonsi; Arianna Bellucci; Antonio Pisani
Published in: Crossref, Issue 37(5):949-961, 2022
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DOI: 10.1002/mds.29024

The European Genome-phenome Archive in 2021.

Author(s): Mallory A. Freeberg; Lauren A Fromont; Teresa D'Altri; Anna Foix Romero; Jorge Izquierdo Ciges; Aina Jene; Giselle Kerry; Mauricio Moldes; Roberto Ariosa; Silvia Bahena; Daniel Barrowdale; Marcos Casado Barbero; Dietmar Fernandez-Orth; Carles Garcia-Linares; Emilio Garcia-Rios; Frédéric Haziza; Bela Juhasz; Oscar Martinez Llobet; Gemma Milla; Anand Mohan; Manuel Rueda; Aravind Sankar; Dona Sha
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Publisher: Oxford Academic
DOI: 10.1093/nar/gkab1059

A mosaic PIK3CA variant in a young adult with diffuse gastric cancer: case report.

Author(s): te Paske, Iris B. A. W.; Garcia-Pelaez, José; Sommer, Anna K.; Matalonga, Leslie; Starzynska, Teresa; Jakubowska, Anna; Valle, Laura; Capella, Gabriel; Aretz, Stefan; Holinski-Feder, Elke; Steinke-Lange, Verena; Laner, Andreas; Schröck, Evelin; Rump, Andreas; Ligtenberg, Marjolijn; Hoischen, Alexander; Geverink, Nicoline; Evans, D. Gareth; Tischkowitz, Marc; Laurie, Steven; van der Post, Rachel
Published in: European Journal of Human Genetics ,, Issue vol. 29 , no. 9, 2021, Page(s) pp. 1354-1358
Publisher: European Journal of Genetics
DOI: 10.1038/s41431-021-00853-6

Isolation and transfection of myenteric neurons from mice for patch-clamp applications

Author(s): Samuel Kuehs; Laura Teege; Ann-Katrin Hellberg; Christina Stanke; Natja Haag; Ingo Kurth; Robert Blum; Carla Nau; Enrico Leipold
Published in: Crossref, Issue 1, 2022
Publisher: Front. Mol. Neurosci
DOI: 10.3389/fnmol.2022.1076187

De-novo FAIRification via an Electronic Data Capture system by automated transformation of filled electronic Case Report Forms into machine-readable data.

Author(s): Martijn G. Kersloot; Annika Jacobsen; Karlijn H. J. Groenen; Bruna dos Santos Vieira; Rajaram Kaliyaperumal; Ameen Abu-Hanna; Ronald Cornet; Peter A C 't Hoen; Marco Roos; Leo J. Schultze Kool; Derk L. Arts
Published in: VOLUME=122;TITLE=Journal of Biomedical Informatics, Issue 1, 2021
Publisher: Science direct
DOI: 10.1016/j.jbi.2021.103897

Variant recurrence confirms the existence of a FBXO31 -related spastic-dystonic cerebral palsy syndrome

Author(s): Dzinovic, I.; Škorvánek, M.; Pavelekova, P.; Zhao, C.; Keren, B.; Whalen, S.; Bakhtiari, S.; Chih Jin, S.; Kruer, M.C.; Jech, R.; Winkelmann, J.; Zech, M.
Published in: Annals of Clinical and Translational Neurology, Vol 8, Iss 4, Pp 951-955 (2021), Issue 1, 2021
Publisher: Ann Clin Transl Neurol

Vesicular Acetylcholine Transporter Alters Cholinergic Tone and Synaptic Plasticity in DYT1 Dystonia.

Author(s): Annalisa Tassone; Giuseppina Martella; Maria Meringolo; Valentina Vanni; Giuseppe Sciamanna; Giulia Ponterio; Paola Imbriani; Paola Bonsi; Antonio Pisani
Published in: Movement disorders, Issue 1, 2021

Publisher: Movement disorders
DOI: 10.1002/mds.28698

FAIR Genomes metadata schema promoting Next Generation Sequencing data reuse in Dutch healthcare and research

Author(s): K. Joeri van der Velde; Gurnoor Singh; Rajaram Kaliyaperumal; XiaoFeng Liao; Sander de Ridder; Susanne Rebers; Hindrik H. D. Kerstens; Fernanda de Andrade; Jeroen van Reeuwijk; Fini E. De Gruyter; Saskia Hiltemann; Maarten Ligtvoet; Marjan M. Weiss; Hanneke W. M. van Deutekom; Anne M. L. Jansen; Andrew P. Stubbs; Lisenka E. L. M. Vissers; Jeroen F. J. Laros; Esther van Enckevort; Daphne Stemkens;
Published in: van der Velde , K J , Singh , G , Kaliyaperumal , R , Liao , X , de Ridder , S , Rebers , S , Kerstens , H H D , de Andrade , F , van Reeuwijk , J , de Gruyter , F E , Hiltemann , S , Ligtvoet , M , Weiss , M M , van Deutekom , H W M , Jansen , A M L , Stubbs , A P , Vissers , L E L M , Laros , J F J , van Enckevort , E , Stemkens , D , 't Hoen , P A C , Beliën , J A M , van Gijn , M E & Swertz, Issue 1, 2022
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DOI: 10.1038/s41597-022-01265-x

A formalization of one of the main claims of "Overlap of vitamin A and vitamin D target genes with CAKUT-related processes" by Ozisik et al. 2021.

Author(s): Friederike Ehrhart, Chris T. Evelo
Published in: Data Science, Issue 5(1), 2022, Page(s) 25-27
Publisher: IOS Press
DOI: 10.3233/ds-210041

Long-read technologies identify a hidden inverted duplication in a family with choroideremia

Author(s): Zeinab Fadaie; Kornelia Neveling; Tuomo Mantere; Ronny Derks; Lonneke Haer-Wigman; Amber den Ouden; Michael Kwint; Luke O'Gorman; Dyon Valkenburg; Carel B. Hoyng; Christian Gilissen; Lisenka E.L.M. Vissers; Marcel R. Nelen; Frans P.M. Cremers; Alexander Hoischen; Susanne Roosing
Published in: HGG Advances, 2, 1 - 11, Issue 1, 2022
Publisher: Science direct
DOI: 10.1016/j.xhgg.2021.100046

A Community-Driven, Openly Accessible Molecular Pathway Integrating Knowledge on Malignant Pleural Mesothelioma

Author(s): Marvin Martens; Franziska Kreidl; Friederike Ehrhart; Didier Jean; Merlin Mei; Holly M. Mortensen; Alistair Nash; Penny Nymark; Chris T. Evelo; Ferdinando Cerciello
Published in: "Frontiers in Oncology, Frontiers, 2022, 12, pp.849640. ⟨10.3389/fonc.2022.849640⟩", Issue 1, 2022

Publisher: Frontiers in oncology
DOI: 10.3389/fonc.2022.849640

Beacon v2 and Beacon networks: A “lingua franca” for federated data discovery in biomedical genomics, and beyond

Author(s): Jordi Rambla; Michael Baudis; Roberto Ariosa; Tim Beck; Lauren A. Fromont; Arcadi Navarro; Rahel Paloots; Manuel Rueda; Gary Saunders; Babita Singh; John D. Spalding; Juha Törnroos; Claudia Vasallo; Colin D. Veal; Anthony J. Brookes
Published in: instname:, Issue 1, 2022
Publisher: Zurich Open Repository and Archive, University of Zurich
DOI: 10.1002/humu.24369

Gitelman-Like Syndrome Caused by Pathogenic Variants in mtDNA.

Author(s): Daan H H M Viering; Karl-Peter Schlingmann; Marguerite Hureaux; Tom Nijenhuis; Andrew Mallett; Melanie Chan; André P van Beek; Albertien M. van Eerde; Jean-Marie Coulibaly; Marion Vallet; Stéphane Decramer; Solenne Pelletier; Günter Klaus; Martin Kömhoff; Rolf Beetz; Chirag Patel; Mohan Shenoy; Eric J. Steenbergen; Glenn Anderson; Ernie M.H.F. Bongers; Carsten Bergmann; Daan M. Panneman; Richa
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Publisher: JASN
DOI: 10.1681/asn.2021050596

Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases

Author(s): Gemma Bullich; Leslie Matalonga; Montserrat Pujadas; Anastasios Papakonstantinou; Davide Piscia; Raúl Tonda; Rafael Artuch; Pia Gallano; Glòria Garrabou; Juan R. González; Daniel Grinberg; Míriam Guitart; Steven Laurie; Conxi Lázaro; Cristina Luengo; Ramon Martí; Montserrat Milà; David Ovelleiro; Genís Parra; Aurora Pujol; Eduardo Tizzano; Alfons Macaya; Francesc Palau; Antònia Ribes; Lui
Published in: reponame:r-IIB SANT PAU. Repositorio Institucional de Producción Científica del Instituto de Investigación Biomédica Sant Pau, Issue 1, 2022
Publisher: The journal of molecular diagnostics
DOI: 10.1016/j.jmoldx.2022.02.003

Frameshift mutation S368fs in the gene encoding cytoskeletal β-actin leads to ACTB-associated syndromic thrombocytopenia by impairing actin dynamics

Author(s): Johannes N. Greve; Frederic V. Schwäbe; Thomas Pokrant; Jan Faix; Nataliya Di Donato; Manuel H. Taft; Dietmar J. Manstein
Published in: Crossref, Issue 1, 2022

Publisher: Elsevier GmbH
DOI: 10.1016/j.ejcb.2022.151216

Goal-models to support communication, planning and guiding of FAIRification
Author(s): Bernab��, C��sar Henrique; Jacobsen, Annika; Queralt Rosinach, N��ria; Bonino da Silva Santos, Luiz Olavo; Silva Souza, V��tor E.; Mons, Barend; Roos, Marco
Published in: Zenodo, Issue 1, 2021
Publisher: Zenodo
DOI: 10.5281/zenodo.5784628

Financing models for sustainable data reuse infrastructure
Author(s): Hooft, Rob; Roos, Marco
Published in: Issue 5, 2023
Publisher: Zenodo
DOI: 10.5281/zenodo.7949903

Applying the FAIR Data Principles to the Registry of Vascular Anomalies (VASCA)
Author(s): Bruna dos Santos Vieira, Karlijn Groenen, P.A.C. ‘t Hoen, Annika Jacobsen, Marco Roos, Rajaram Kaliyaperumal, Martijn Kersloot, Ronald Cornet, Leo Schultze Kool
Published in: Studies in Health Technology and Informatics, Issue Volume 271: dHealth 2020 – Biomedical Informatics for Health and Care, 2020, Page(s) 115 - 116, ISSN 0926-9630
Publisher: IOS Press
DOI: 10.3233/shti200085

A FAIR data model for PRISMA (Personalised RISk-based MAmmascreening) Study
Author(s): Liao, Xiaofeng; de Jong, Milou; van Damme, Philip; Cornet, Ronald; Dos Santos Vieira, Bruna; Lutomski, Jennifer; Brullemans-Spansier, Mirjam; 't Hoen, Peter
Published in: Zenodo, Issue 1, 2022
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Converging pathways found in copy number variation syndromes with high schizophrenia risk
Author(s): Friederike Ehrhart, Ana Silva, Therese van Amelsvoort, Emma von Scheibler, Chris Evelo, David E.J. Linden
Published in: bioRxiv, 2022, Page(s) United States
Publisher: Cold Spring Harbour Laboratory
DOI: 10.1101/2022.02.07.479370

Rare disease education in Europe and beyond: time to act

Author(s): Birute Tumiene 1, Harm Peters 2 3, Bela Melegh 4, Borut Peterlin 5, Algirdas Utkus 1 3, Natalja Fatkulina 6, György Pfliegler 7, Holm Graessner 8 9, Sanja Hermanns 8 9, Maurizio Scarpa 10, Jean-Yves Blay 11, Sharon Ashton 12, Lucy McKay 13, Gareth Baynam 14 15 16
Published in: Orphanet Journal of Rare disease, Issue 19;17(1):441, 2022
Publisher: BMC
DOI: 10.1186/s13023-022-02527-y

The impact of dedicated FAIRification stewardship guiding European Reference Networks towards making rare disease resources FAIR
Author(s): Inês Henriques; Bruna dos Santos Vieira; César Henriques Bernabé; Shuxin Zhang; Alberto Cámara Ballesteros; Jose Antonio Ramírez García; Joeri van der Velde; Nirupama Benis; Peter 't Hoen; Marco Roos; Annika Jacobsen; Ronald Cornet; Mark Wilkinson; Franz Schaefer; Morris Swertz
Published in: Issue 9, 2023
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DOI: 10.5281/zenodo.7915232

Solving patients with rare diseases through programmatic reanalysis of genome-phenome data
Author(s): Matalonga, Leslie; Hernández-Ferrer, Carles; Piscia, Davide; Schüle, Rebecca; Synofzik, Matthis; Töpf, Ana; Vissers, Lisenka E. L. M.; de Voer, Richarda; Tonda, Raul; Laurie, Steven; Fernandez-Callejo, Marcos; Picó, Daniel; Garcia-Linares, Carles; Papakonstantinou, Anastasios; Corvó, Alberto; Joshi, Ricky; Diez, Hector; Gut, Ivo; Hoischen, Alexander; Graessner, Holm; Beltran, Sergi; Cohen, En
Published in: European journal of human genetics, Issue 1, 2021
Publisher: Eur J Hum Genet
DOI: 10.1038/s41431-021-00852-7

Book chapters (2)

Overview of treatment strategies in paraneoplastic neurological syndromes
Author(s): Jeroen Kerstens, Maarten J. Titulaer
Published in: Handbook of Clinical Neurology, Paraneoplastic Neurologic Disorders, 2024, Page(s) 97-112, ISSN 0072-9752
Publisher: Elsevier
DOI: 10.1016/b978-0-12-823912-4.00015-3

Privacy-Preserving Linkage of Distributed Pseudonymised Datasets in a Virtual European Rare Disease Platform
Author(s): Dieter Hayn, Emanuel Sandner, Abishaa Vengadeswaran, Elena-Alexandra Tãtaru, Mark Wilkinson, Marc Hanauer, Karl Kreiner, Guenter

Schreier
Published in: Studies in Health Technology and Informatics, Digital Health and Informatics Innovations for Sustainable Health Care Systems, 2024
Publisher: IOS Press
DOI: 10.3233/shti240683

Non-peer reviewed articles (15)

Performance assessment of ontology matching systems for FAIR data
Author(s): Philip van Damme; Jesualdo Tomás Fernández-Breis; Nirupama Benis; Jose Antonio Miñarro-Gimenez; Nicolette F. de Keizer; Ronald Cornet
Published in: Journal of Biomedical Semantics, 13(1):19. BioMed Central Ltd., Issue 1, 2022, ISSN 2041-1480
Publisher: Journal of biomedical semantics
DOI: 10.1186/s13326-022-00273-5

Solving unsolved rare neurological diseases—a Solve-RD viewpoint
Author(s): Schüle, Rebecca; Timmann, Dagmar; Erasmus, Corrie E.; Reichbauer, Jennifer; Wayand, Melanie; van de Warrenburg, Bart; Schöls, Ludger; Wilke, Carlo; Bevot, Andrea; Zuchner, Stephan; Beltran, Sergi; Laurie, Steven; Matalonga, Leslie; Graessner, Holm; Synofzik, Matthis; Baets, Jonathan; Balicza, Peter; Chinnery, Patrick; Dürr, Alexandra; Haack, Tobias; Hengel, Holger; Horvath, Rita; Houlden, Henry
Published in: European journal of human genetics, Issue 1, 2021, ISSN 1476-5438
Publisher: European Journal of Human Genetics
DOI: 10.1038/s41431-021-00901-1

Possible role for rare TRPM7 variants in patients with hypomagnesaemia with secondary hypocalcaemia
Author(s): Rosa Vargas-Poussou; Felix Claverie-Martin; Caroline Prot-Bertoye; Valentina Carotti; Jenny van der Wijst; Ana Perdomo-Ramirez; Gloria M Fraga-Rodriguez; Marguerite Hureaux; Caro Bos; Femke Latta; Pascal Houillier; Joost G J Hoenderop; Jeroen H F de Baaij
Published in: reponame:r-IIB SANT PAU. Repositorio Institucional de Producción Científica del Instituto de Investigación Biomédica Sant Pau, Issue 1, 2022, ISSN 0931-0509
Publisher: Oxford University Press
DOI: 10.1093/ndt/gfac182

Applying the FAIR principles to data in a hospital: challenges and opportunities in a pandemic

Author(s): Queralt Rosinach, N.; Kaliyaperumal, R.; Bernabe, C.H.; Long, Q.Q.; Joosten, S.A.; Wijk, H.J. van der; Flikkenschild, E.L.A.; Burger, K.; Jacobsen, A.; Mons, B.; Roos, M.; BEAT COVID Grp; COVID-19 LUMC Grp
Published in: Journal of Biomedical Semantics, 13(1). BMC, Issue 1, 2022, ISSN 2041-1480
Publisher: BMC
DOI: 10.1186/s13326-022-00263-7

DMD – BIOMARKERS & OUTCOME MEASURES
Author(s): I. Verhaart, P. 'tHoen, M. Roos, E. Vroom
Published in: Neuromuscular Disorders, Issue 30, 2020, Page(s) S86, ISSN 0960-8966
Publisher: Elsevier BV
DOI: 10.1016/j.nmd.2020.08.137

The de novo FAIRification process of a registry for vascular anomalies
Author(s): Karlijn H.J. Groenen, Annika Jacobsen, Martijn G. Kersloot, Bruna Vieira, Esther van Enckevort, Rajaram Kaliyaperumal, Derk L. Arts, Peter A.C. ‘t Hoen, Ronald Cornet, Marco Roos, Leo Schultze Kool
Published in: Orphanet Journal of Rare Diseases, Issue 16(1), 2021, Page(s) 376, ISSN 1750-1172
Publisher: BioMed Central
DOI: 10.1186/s13023-021-02004-y

Understanding signaling and metabolic paths using semantified and harmonized information about biological interactions
Author(s): Ryan A. Miller; Martina Kutmon; Anwesha Bohler; Andra Waagmeester; Chris T. Evelo; Egon Willighagen
Published in: Plos one, Issue 1, 2022, ISSN 1932-6203
Publisher: Public Library of Science
DOI: 10.1101/2020.07.31.230599

The “A” of FAIR – As Open as Possible, as Closed as Necessary
Author(s): Annalisa Landi, Mark Thompson, Viviana Giannuzzi, Fedele Bonifazi, Ignasi Labastida, Luiz Olavo Bonino da Silva Santos, Marco Roos
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Author(s): Marco Roos, Mark D. Wilkinson, Ronald Cornet, Deborah Mascalzoni, Veronica Popa, Ian Harrow,Claudio Carta, Yaffa R. Rubinstein, Dipak Kalra, Ana Rath, Victoria Hedley, Gülçin Gümüş

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Author(s): Erika Souche Sergi Beltran, Erwin Brosens, John W Belmont, Magdalena Fossum, Olaf Riess, Christian Gilissen, Amin Ardeshirdavani, Gunnar Houge, Marielle van Gijn , Jill Clayton-Smith , Matthis Synofzik , Nicole de Leeuw, Zandra C Deans, Yasemin Dincer, Sebastian H Eck, Saskia van der Crabben, Meena Balasubramanian, Holm Graessner , Marc Sturm, Helen Firth, Alessandra Ferlini, Rima Nabbout, Elfrid
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Author(s): Jonathan Lawson; Moran N. Cabili; Giselle Kerry; Tiffany Boughtwood; Adrian Thorogood; Adrian Thorogood; Pinar Alper; Sarion R. Bowers; Rebecca R. Boyles; Anthony J. Brookes; Matthew Brush; Tony Burdett; Hayley L. Clissold; Stacey Donnelly; Stephanie O. M. Dyke; Mallory A. Freeberg; Melissa Haendel; Chihiro Hata; Petr Holub; Francis Jeanson; Aina Jene; Minae Kawashima; Shuichi Kawashima; Melissa A
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Author(s): Federica Invernizzi; Andrea Legati; Alessia Nasca; Eleonora Lamantea; Barbara Garavaglia; Mirjana Gusic; Robert Kopajtich; Holger Prokisch; Massimo Zeviani; Costanza Lamperti; Daniele Ghezzi

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Adenine base editing efficiently restores the function of Fanconi anemia hematopoietic stem and progenitor cells

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Identifying obstacles hindering the conduct of academic-sponsored trials for drug repurposing on rare-diseases: an analysis of six use cases

Author(s): Marta del Álamo; Christoph Bührer; Dirk Fisher; Matthias Griese; Paul Lingor; Giovanni Palladini; Nicolas Sireau; Virginie Hivert; Luca Sangiorgi; Florence Guillot; Juliane Halftermeyer; Lenka Soucková; Kristýna Nosková; Regina Demlová
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Conference proceedings (4)

The COVID-19 epidemiology and monitoring ontology

Author(s): Queralt-Rosinach, Núria; Schofield, Paul N.; Hoehndorf, Robert; Weiland, Claus; Schultes, Erik; Bernabé, César H.; Roos, Marco
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Author(s): Núria Queralt-Rosinach1, Mark Wilkinson2, Rajaram Kaliyaperumal1,César H. Bernabé1, Qinqin Long1, Michel Dumontier3, Paul N. Schofield4andMarco Roos
Published in: CEUR Workshop Proceedings, 2021, ISSN 1613-0073
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Author(s): Michael Clarkson; Allyson Egan; Mark A Little
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Thesis and dissertations (1)

FAIR metadata, models, and interfaces to support machine-readable data access and use conditions
Author(s): null null, Oussama Mohammed Benhamed
Published in: 2024
Publisher: Universidad Politecnica de Madrid - University Library
DOI: 10.20868/upm.thesis.81819

Datasets

Datasets via OpenAIRE (46)

Rare disease resources
Author(s): Shuxin Zhang
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Additional file 3 of Automated approach for quality assessment of RDF resources
Author(s): Zhang, Shuxin; Benis, Nirupama; Cornet, Ronald
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Research Data related to article "Myopathic mitochondrial DNA depletion syndrome associated with biallelic variants in LIG3"
Author(s): Ghezzi, Daniele; Legati, Andrea
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Author(s): Bernabé, César H.; Queralt-Rosinach, Núria; Silva Souza, Vítor E.; Bonino da Silva Santos, Luiz Olavo; Jacobsen, Annika; Mons, Barend; Roos, Marco
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Author(s): Ozisik, Ozan; Ehrhart, Friederike; Evelo, Chris T; Mantovani, Alberto; Baudot, Anaïs
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Author(s): Shuxin Zhang
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Author(s): Bernabé, César H.; Queralt-Rosinach, Núria; Silva Souza, Vítor E.; Bonino da Silva Santos, Luiz Olavo; Jacobsen, Annika; Mons, Barend; Roos, Marco
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Author(s): dos Santos Vieira, Bruna; Bernabé, César H.; Zhang, Shuxin; Abaza, Haitham; Benis, Nirupama; Cámara, Alberto; Cornet, Ronald; Le Cornec, Clémence M. A.; 't Hoen, Peter A. C.; Schaefer, Franz; van der Velde, K. Joeri; Swertz, Morris A.; Wilkinson, Mark D.; Jacobsen, Annika; Roos, Marco
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Author(s): Shuxin Zhang
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Author(s): Queralt-Rosinach, N; Wilkinson, M; Kaliyaperumal, R; Bernabé, CH; Long, Q; Dumontier, M; Schofield, PN; Roos, M
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Software

Software via OpenAIRE (3)

orsum

Author(s): Ozisik, Ozan; Térézol, Morgane; Baudot, Anaïs
Publisher: Zenodo
DOI: 10.5281/zenodo.6036031; 10.5281/zenodo.6036032; 10.5281/zenodo.6519133

rWikiPathways

Author(s): Willighagen, Egon; Pico, Alexander
Publisher: bio.tools

RD-Connect Genome-Phenome Analysis Platform (GPAP)

Author(s): Laurie, Steven; Beltran, Sergi
Publisher: bio.tools

Other Research Products

Other Research Products via OpenAire (7)

The FAIR Game

Author(s): Roos, Marco; Carta, Claudio; dos Santos Vieira, Bruna; Kaliyaperumal, Rajaram; Kersloot, Martijn; Jacobsen, Annika; Cornet, Ronald; Wilkinson, Mark; Hanauer, Marc; Benis, Nirupama; Lee Cellai, Laura; Bernabé, César Henrique; Taruscio, Domenica; 't Hoen, P.A.C.; Cámara Ballesteros, Alberto; de Oliveira Coelho Henriques, Ines; Ramírez García, Jose Antonio; Zhang, Shuxin; van der Velde, Joeri; Swertz, Morris A.
Published in: Zenodo

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Author(s): Bernabé, César; Jacobsen, Annika; Santos, Luiz; Roos, Marco
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Author(s): Cámara Ballesteros, Alberto; dos Santos Vieira, Bruna; Roos, Marco; Carta, Claudio; Bernabe, Cesar Henrique; Zhang, Shuxin; Benis, Nirupama; van der Velde, Joeri; Kaliyaperumal, Rajaram; Coelho de Oliveira Henriques, Ines
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Author(s): Ozisik, Ozan; Kara, Nazli Sila; Abbassi-Daloii, Tooba; Térézol, Morgane; Queralt-Rosinach, Núria; Jacobsen, Annika; Sezerman, Osman Ugur; Roos, Marco; Evelo, Chris T.; Baudot, Anaïs; Ehrhart, Friederike; Mina, Eleni
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Author(s): Liao, Xiaofeng; de Jong, Milou; van Damme, Philip; Cornet, Ronald; Dos Santos Vieira, Bruna; Lutomski, Jennifer; Brullemans-Spansier, Mirjam; 't Hoen, Peter
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Author(s): Clarkson, Michael; Egan, Allyson; Little, Mark A
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DNA damage response at telomeres boosts the transcription of SARS-CoV-2 receptor ACE2 during aging
Author(s): Sara Sepe; Francesca Rossiello; Valeria Cancila; Fabio Iannelli; Valentina Matti; Giada Cicio; Matteo Cabrini; Eugenia Marinelli; Busola Alabi; Alessia di Lillo; Arianna Di Napoli; Jerry Shay; Claudio Tripodo; and Fabrizio d'Adda di Fagagna

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