Webinar on IMI2 - Call 23 webinar: Rare Disease diagnosis by using new born genetic screening and digital technologies

Participant list

29.06.2020

Name	Surname	Organisation	Country	E-mail address	Potential contribution to the project in terms of expertise, resources and activities.
Ramiro	Agraz	IIC	Spain	ramiro.agraz@iic.uam.es	We have experience in Artificial Intelligence, e-health
Ilaria	Allora	Politecnico di Torino	Italy	ilaria.allora@polito.it	
Zuzanna	Andrzejewska	Institut Imagine	France	zuzanna.andrzejewska@institutimagine.org	genetic diseases models, pipelines for drug/biomarkers discovery/validation, genetic screening, single-cell multiOMICs, AI
Agustin	Arasanz Duque	VHIR	Spain	agustin.arasanz@vhir.org	Project Management
Dimitrios	Athanasiou	WORLD DUCHENNE ORGANIZATION / UPPMD	Greece	DATHAX@GMAIL.COM	Rare Diseases
Segolene	Ayme	INSERM	France	segolene.ayme@inserm.fr	extensive experience in policy setting of newborn screening and large experience in ethics of genetic screening
Sevgi	Bagislar Bremang	Izmir Biomedicine and Genome Center	Turkey	sevgi.bagislar@ibg.edu.tr	IBG would like to contribute its expertise, infrastructure, and national network to rare disease research
Cécile	Bernard	Aix Marseille university Institute MarMaRa	France	cecile.bernard.1@univ-amu.fr	

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Sarah	Berrocoso	IDIVAL	Spain	proyectoseuropeos@idival.org	
Florence	Bietrix	EATRIS	Netherlands	florencebietrix@eatris.eu	
Olivier	Blin	AMU	France	olivier.blin@ap-hm.fr	Orphan drug dev Orpgandev
François	Boemer	CHU Liege	Belgium	F.Boemer@chuliege.be	We are already working in our laboratory on the implementation of NGS technologies into newborn screening.
Jan-Willem	Boiten	Lygature	Netherlands	janwillem.boiten@lygature.org	Program Management, Communication, data infrastructure support
Virginie	Bros-Facer	EURORDIS	France	virginie.bros-facer@eurordis.org	Patient engagement in diagnostic research, genomics, newborn screening and ethical issues related to these topics
Ana Cristina	Calvo Royo	University of Zaragoza	Spain	accalvo@unizar.es	Identificationof biomarkers of rare and neurodegenerative diseases, traslational studies, gene and protein analysis
Jessica	Cibrian	FI Group	Spain	jessica.cibrian@fi-group.com	Coordination & Consortium Management. Communication, Dissemination & Exploitation Partner
Kristl	Claeys	University Hospitals Leuven	Belgium	Kristl.Claeys@uzleuven.be	specialist in neuromuscular diseases, which all are rare diseases

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Martina	Cornel	Amsterdam University Medical Centers	Netherlands	mc.cornel@amsterdamumc.nl	experience in policy making in the domain of neonatal screening, see e.g. doi: 10.1038/ejhg.2013.90.
Ronald	Cornet	Amsterdam UMC, dept of Medical Informatics	Netherlands	r.cornet@amsterdamumc.nl	FAIR data in the context of rare diseases, focusing on modeling and representation of clinical data
Cristina	Cuscó	Vall d'Hebron Institute of Research	Spain	cristina.cusco@vhir.org	
Ilse	Custers	Lygature	Netherlands	ilse.custers@gmail.com	HEOR on heel prick screening programs, diagnostics (Personalised Medicine). Rare diseases network
Marta	de Diego	CDTI	Spain	marta.dediego@cdti.es	
Pieter	de Koning	Leiden University Medical Center	Netherlands	p.j.a.de_koning@lumc.nl	
Jordi	Diaz-Manera	Newcastle University	United Kingdom	jordi.diaz-manera@newcastle.ac.uk	Knowledge in AI applied to the diagnosis of neuromuscular diseases
Dimitar	Dimitrov	Micar Innovation (Micar21)	Bulgaria	info@micar21.com	Micar Innovation (Micar21) is a drug discovery factory. AI, ML. NLP
Dermot	Doyle	Doyle SARL-S	Germany	doyle.dermot@gmail.com	Long experience in EU level projects, covering both regulatory and market dynamics. Consortia management expertise.
Anna	Dziubczynska-Pytko	IPPT PAN	Poland	anna.dziubczynska-pytko@kpk.gov.pl	
Kristina	Eskenazi	Micar21	Bulgaria	kritina.eskenazi@biocluster.bg	drug discovery, in silico, AI pipelines & models

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Berta	Español	TEAM-IT RESEARCH, S.L.	Spain	bespanol@teamitresearch.com	Management
Roseline	Favresse	French Foundation for Rare Diseases	France	roseline.favresse@fondation-maladiesrares.com	RD research support. Coordinating an e-learning course on RD research (1st topic is on diagnosis) within EJP-RD.
Judit	Garcia Villoria	Hospital Clinic	Spain	jugarcia@clinic.cat	
José Manuel	González de Aledo Castillo	Hospital Clinic de Barcelona	Spain	gonzalezde@clinic.cat	I work in the newborn screening programme of Catalonia
Geraldine	Gouzer	Paris Brain Institute - ICM	France	geraldine.gouzer@icm-institute.org	Clinical research experts in rare neurological diseases (Ataxia,Huntington, rare dementia, ALS,Tourette, ec)
Claus	Gravholt	Aarhus University Hospital	Denmark	ch.gravholt@dadlnet.dk	Expert in rare genetic endocrine diseases, working with bioinformatics and diagnostics
Katarina	Gvozdanovic	HALMED	Croatia	katarina.gvozdanovic@gmail.com	
Victoria	Hedley	Newcastle University	United Kingdom	victoria.hedley@ncl.ac.uk	Rare Disease Policy expertise, incl. on the status quo of NBS; my Centre brings expertise (in NBS and AI) in neuromuscular dis.
B.	Heinemann	MCS Data Labs	Germany	b.heinemann@mcs-datalabs.com	Hardware-/ software ecosystem, smart digital devices (wearables), AI/ ML, customizable to specific health conditions.

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Naveed	Ishaque	Charité	Germany	naveed.ishaque@charite.de	Genomics, NGS, workflows, data management, cloud, AI, machine learning, cancer, multi-omics integration, epigenetics,single cell
Daria	Julkowska	INSERM - EJP RD	France	daria.julkowska@ejprarediseases.org	European Joint Programme on Rare Diseases
Rita	Juneja	Pharmaceutical Research and Manufacturers Association	Thailand	rita@prema.or.th	To find Thai researchers and form a partnership for research projects in IMI platform leading to new discoveries
Joanna	Kaldrack	MDC	Germany	joanna.kaldrack@mdc-berlin.de	
Donal	Killackey	SSPC - Pharmaceutical Research Centre Ireland	Ireland	donal.killackey@ul.ie	Developing highly luminescent and in vivo applicable long-wavelength sensory/probes/imaging agent systems for use in screening
Alex	Knight	Holmusk Europe	United Kingdom	alex.knight@holmusk.com	Expertise in analytics of EHR, clinical trial data using AI/machine learning and semi-mechanistic modelling; consumer app design
Jens	Kofod	Greater Copenhagen	Denmark	jens.erik.kofod@regionh.dk	
Tess	Korthout	The Hyve	Netherlands	tess@thehyve.nl	FAIR data management, remote clinical trials
Satu	Kuure	Univ. of Helsinki	Finland	satu.kuure@helsinki.fi	mutation-specific disease modelling in mouse and rat, embryologic and fetal development, GM-models

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Gergana	Kyosovska-Peshtenska	BAPEMED	Bulgaria	gergana.kyosovska@pdc-eu.com	Personalized Medicine, Patient Advocacy
Natalia	Loete	INSAVALOR	France	natalia.loete@insavalor.fr	AI, Data mining; Modelling and data analysis; Imaging and modelling biological systems; E-health; Biomolecules, biomaterials
Rosa Maria	Lopez Galera	SERVICIO DE BIOQUIMICA Y GENETICA MOLECULAR	Spain	rmlopez@clinic.cat	
Mariangela	Lupo	Consorzio per Valutazioni Biologiche e Farmacologiche	Italy	mlupo@cvbf.net	
Mariangela	Lupo	Consorzio per Valutazioni Biologiche e Farmacologiche	Italy	mlupo@teddynetwork.net	
Gregory	Maes	University of Leuven	Belgium	gregory.maes@kuleuven.be	Valorisation Manager
Sylvie	Maiella	ORPHANET_INSERM_US14	France	sylvie.maiella@inserm.fr	Orphanet is a knowledge base on RD and Orphanet drugs. Orphanet also maintains the Orphanet rare disease nomenclature
Lucia	Malaguarnera	University of Catania	Italy	lucmal@unict.it	
Myroslava	Malets	University	Ukraine	muroslavamalets@gmail.com	New research in the field of epilepsy and early hydrocephalus. Genetic

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					causes of epilepsy in children with early hydrocephalus
laura	martinelli	IN SRL	Italy	l.martinelli@insrl.eu	currently working with a top level consortium applying to this call
Sara	Mas Assens	VHIR	Spain	sara.mas@vhir.org	
Eleonore	Mathe	Spark Tx	United Kingdom	eleonore.mathe@gmail.com	
Laurène	Mathey	Hospices Civils de Lyon	France	laurene.mathey@chu-lyon.fr	
Gert	Matthijs	University of Leuven	Belgium	gert.matthijs@uzleuven.be	Genetics, EuroGentest quality issues, International rare disease policy (e.g. G2MC), Population screening policy, WGS technology
Giiovanni	Migliaccio	CVBF	Italy	gmigliaccio@cvbf.net	Regulatory expertise in translational medicine, cell therapies and advanced product including diagnostics
Eva	Molero	TEAM-IT Research	Spain	emolero@teamitresearch.com	Project management, communication, sustainability
Marjorie	Monleau	Ad'Occ	France	marjorie.monleau@agence-adocc.com	
kejla	musaraj	Consorzio per Valutazioni Biologiche e Farmacologiche	Italy	kmusaraj@cvbf.net	Project Coordination, Project Management and Project Monitoring
Christoffer	Nellaker	University of Oxford	United Kingdom	christoffer.nellaker@wrh.ox.ac.uk	Image based phenotype analysis, lead on consortium building framework for sharing of identifiable patient data.

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Ugur	Özbek	Acibadem University School of Medicine	Turkey	ugur.ozbek@acibadem.edu.tr	ACURARE (Center of Rare Disease and Orphan Drugs); Expertise on clinical and molecular genetics of Rare and Undiagnosed diseases
Sonia	Pajares	Inborn Errors of Metabolism Division. Biochemistry and Molecular Genetics Department. Hospital Clinic of Barcelona	Spain	spajares@clinic.cat	
Hannes	Perko	AIT Austrian Institute of Technology	Austria	hannes.perko@ait.ac.at	digital health information systems
Valeria	Pignataro	CVBF	Italy	vpignataro@cvbf.net	
Cristina	Pintucci	Ghent university	Belgium	cristina.pintucci@ugent.be	Project management at setting up and following up (large) projects
Diana	Pirjol	HDS	Romania	dpirjol@human-datascience.org	Data science, clinical experience, expertise in epidemiology
Irene	Pitsillidou	CYPRUS LEAGUE AGAINST RHEUMATISM	Cyprus	roulla810@live.com	I will bring my knowledge and expertise working voluntarily for 13 years for my organisation and as a EULAR PRP since 2010
Haritz	Plazaola	IDIBAPS	Spain	PLAZAOLA@clinic.cat	
Natalya	Prilipko	University of Haifa	Israel	natalyap@univ.haifa.ac.il	We have researchers interested in joining a consortium

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Lena	Prochnow	EURICE – European Research and Project Office GmbH	Germany	l.prochnow@eurice.eu	Project management, Innovation management (Communication, Dissemination, Exploitation activities)
Ana	Rath	Inserm-Orphanet	France	ana.rath@inserm.fr	Orphanet reference knowledge base on RD, computable reference nomenclature (ORPHAcodes) & ontologies incl. curated HPO links.
Santi	Rello-Varona	IdiPAZ institute	Spain	international.projects@idipaz.es	We are a Hospital, we can bring patients and clinical research expertise.
Antonia	Ribes	Hospital Clinic de Barcelona	Spain	aribes@clinic.cat	Expertise in neonatal screening, expertise in the discovery of new genes trough NGS. Functional studies and biomarkers
Ettore	Rizzo	enGenome srl	Italy	erizzo@engenome.com	We can support the project with eVai, our CE IVD software that enables a faster and more accurate diagnosis of RD from NGS data.
Ester	Rodriguez	Bellvitge Biomedical Research Institute - IDIBELL	Spain	ester.rodriguez@idibell.cat	stem cells, IPS, organoids
Patrick	Ruch	SIB	Switzerland	patrick.ruch@hesge.ch	Variant interpretation tools; Text Mining
Philippe	Ryvlin	Human Brain Project	Switzerland	philipperyvlin@gmail.com	The Medical Informatic Platform (Human Brain Project) offers unique AI

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					capacity accross 30 hospitals to identify rare diseases
Elvina	Sakellariou	Duchenne Data Foundation	Greece	sakellariou.elvina@duchennedatafoundation.org	Data management, Data FAIR Principles, AI
Doron	Samuell	Behaviour/Deloitte	Cyprus	director@behaviour.ai	Established expertise in behavioural research, analytics, project management, clinical research, technology development
Luca	Sangiorgi	Rizzoli Orthopedic Institute - BOND ERN	Italy	luca.sangiorgi@ior.it	registries collecting data (clinical, genetic, imaging.) on bone rare diseases with long follow-up. AI algorithms tested on data
Travis	Satnarine	Port of Spain General Hospital	Trinidad and Tobago	travissatnarine@gmail.com	Dedication and love for research.
Maurizio	Scarpa	MetabERN, Udine University Hospital	Italy	maurizio.scarpa@metab.ern-net.eu	Coordinator, European Reference Network for Metabolic diseases, MetabERN, expert in diagnosis and newborn genetic screening
Christian	Schaaf	University Hospital Heidelberg	Germany	christian.schaaf@med.uni-heidelberg.de	Human genetics
M.	Schulte	LifeGlimmerGmbH	Germany	schulte@lifeglimmer.com	
Nir	Shaked	ISERD Israel Innovation Authority	Israel	nir.s@iserd.org.il	
Montserrat	Sole Castellvi	IBB_UAB	Spain	montserrat.sole.castellvi@uab.cat	Genetic analysis; diagnostic devices. Expertise in rare diseases &

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					replacement strategies and nanoprotein approaches.
Marie	Stas	NCP Wallonie	Belgium	marie.stas@ncpwallonie.be	
Carl	Steinbeisser	collaborate.eu	Germany	carl@collaborate.eu	Good track record in project management and proposal writing. Setup of collaboration platforms for your team.
Volker	Straub	Newcastle University	United Kingdom	volker.straub@ncl.ac.uk	Expertise in translational research in rare diseases, especially genetic neuromuscular diseases, sequencing and networking
Peter	Szegner	Semmelweis University - Health Services Management Training Centre	Hungary	szegner@emk.sote.hu	
Mireia	Tomàs	FISABIO	Spain	tomas_mirgin@gva.es	
Roser	Torra	IIB SANT PAU	Spain	rtorra@fundacio-puigvert.es	Extensive knowledge on genetic kidney diseases
Birute	Tumiene	Vilnius University	Lithuania	tumbir@gmail.com	expertise in coordination of H2020projects/programs, more than 40 years expertise in nationwide neonatal screening

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Dagmar	Václavíková	Masaryk University	Czech Republic	dagmar.vaclavikova@med.muni.cz	Research facilities, experts
Daniel	Weibel	Research Development Office, Erasmus Medical Center, Rotterdam, NL	Netherlands	d.weibel@erasmusmc.nl	www.erasmusmc.nl
Ingunn	Westerheim	Osteogenesis Imperfecta Federation Europe (OIFE)	Norway	president@oife.org	Patient perspective on rare bone diseases
Michael	Wilbur	EURORDIS	France	michael.wilbur@eurordis.org	
Gerhard	Wingender	Izmir Biomedicine and Genome Center (IBG, www.ibg.edu.tr)	Turkey	gerhard.wingender@ibg.edu.tr	I am the coordinator of IBG's ERA Chair project, which will foster IBG's aim to become the national node for rare disease RDI.
Nicole	Wyss	Euresearch	Switzerland	nicole.wyss@euresearch.ch	
Raquel	Yahyaoui	Málaga Regional University Hospital. Insitute of Biomedical Research in Málaga (IBIMA)	Spain	raquelyahyaoui@gmail.com	Research in metabolic disorders and newborn screening

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Lidiia	Zhytnik	Osteogenesis Imperfecta Federation Europe (OIFE)	Estonia	lida.zhytnik@gmail.com	patient perspective in rare bone diseases