ERA PerMed funded projects on:

Multidisciplinary research projects on personalised medicine - development of clinical support tools for personalised medicine implementation

JTC2021 Midterm Symposium

February 12-13, 2025

Berlin, Germany

EP PerMed
European Partnership
for **Personalised Medicine**

ERA PerMed

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Content

Introduction

Personalised medicine (PM) represents a paradigm shift in healthcare, emphasizing tailored medical approaches that consider individual variability in genes, other biological factors, environment, and lifestyle. As one of the most promising advancements in modern healthcare, PM has the potential to transform the prevention, diagnosis, and treatment of a wide range of diseases, improving patient outcomes and quality of life. The field's complexity and interdisciplinary nature call for coordinated research efforts and collaboration across borders, disciplines, and sectors.

The ERA PerMed network, funded under the EU Horizon 2020 framework, was at the forefront of fostering international collaboration in PM research. ERA PerMed played a critical role in aligning research priorities, pooling resources, and advancing the implementation of PM across Europe and beyond. The network's commitment to fostering global partnerships is continued through its successor, EP PerMed, which will further strengthen the PM research, innovation and implementation ecosystem.

The JTC2021 call, titled "Development of Clinical Support Tools for Personalised Medicine Implementation," supported 22 exceptional consortia comprising interdisciplinary teams from diverse research domains. These projects address critical healthcare challenges, including cancer, cardiovascular diseases, neurological disorders, autoimmune conditions, and more. By developing clinical tools, integrating omics technologies, and exploring ethical, legal, and social implications, these initiatives are paving the way for the integration of PM into everyday clinical practice.

This symposium offers a platform for researchers, clinicians and other stakeholders to exchange knowledge, showcase advancements, and explore strategies to address challenges encountered in the JTC2021-funded projects. Through presentations, panel discussions, and networking sessions, the symposium fosters scientific dialogue and encourages the formation of new collaborations that will further advance PM research and implementation. The programme also features a workshop on Responsible Research and Innovation (RRI) conducted by the QUEST Centre in Berlin and a panel discussion dedicated to patient engagement in PM research, highlighting the importance of inclusive and ethical practices in advancing healthcare innovations.

On behalf of ERA and EP PerMed, we extend our gratitude to all participants for their dedication and collaboration. We wish you an inspiring and productive symposium that will spark new ideas and partnerships, accelerating the translation of PM research into tangible health benefits for patients everywhere.

JTC2021 Midterm Symposium Agenda

12-13 February 2025

Hotel Palace Berlin, Budapester Str. 45, 10787 Berlin, Germany

Day 1 | Wednesday, February 12th

12:40 – 13:40	Registration and refreshments
13:40 – 14:00	Opening Session Welcome note and Introduction of EP PerMed Introduction of JTC2021 | JTC2021 secretariat
14:00 – 15:20	Session 1: Research Presentations | Chairs: William Duddy & Delia Sánchez AGORA, Sophie Brouard ArtiPro, Julia Stingl BRAVA, Matteo Cesari CORSAI, Marzia Bedoni
15:20 – 16:00	Coffee-Break & Poster session
16:00 – 17:20	Session 2: Research Presentations | Chairs: Åsa Andersson & Stefano Comai ECLAI, Gema Moreno-Bueno EDAP-AD, Rik Ossenkoppele IMAGene, Serena Oliveri WODIA, Stefan Rahr Wagner
17:25-19:30	RRI Workshop Quest Centre Berlin
20:00	Networking Dinner Spreegold BIKINI Berlin, Restaurant 1.OG Budapester Straße 50, 10787 Berlin

Day 2 | Thursday, February 13th

9:00 – 9:10	Short welcome
9:10 – 10:50	Session 3: Research Presentations | Chairs: William Duddy & Delia Sánchez MIRACLE, Paola Ulivi Pattern-Cog, Jussi Tohka PER-NEPH, Paola Romagnani PerCard, Mark van Gils TECH-TOYS, Giuseppina Sgandurra
10:50 – 11:30	Coffee Break & Poster session
11:30 – 12:40	Patient engagement in personalised medicine research – challenges & advantages
12:40 – 13:40	Lunch
13:40 – 15:20	Session 4: Research Presentations | Chairs: Åsa Andersson & Stefano Comai PerFluid, Thomas Desaive PerSAIDs, Isabella Ceccherini PETictCAC, Thomas Beyer PRE-CARE ML, Stefan Kalabakov CytoMARK, Irene de la Calle Fuentes
15:20 – 15:50	Coffee Break (voting for best poster)
15:50 – 17:10	Session 5: Research Presentations | Chairs: William Duddy & Åsa Andersson i-RECORDS, Djillali Annane REDESIGN, Franziska Baenke ScandRA, Johan Askling SYMMETRY, Ekaterina Popova
17:10 – 17:30	Video and Poster Award Ceremony
17:30 – 17:50	Feedback, summary and farewell

Session Chairs

We are honoured to have a panel of esteemed experts as session chairs for the JTC2021 Midterm Symposium. Their leadership and insights will guide the discussions, ensuring a dynamic and enriching exchange of ideas throughout the event.

Åsa Andersson

Dr. Andersson is a professor in biomedicine at Halmstad University, Sweden. Åsa Andersson has a PhD in Molecular Biotechnology from University of Umeå, became an associate professor (docent) in Medical Inflammation Research at Lund University, and is at present professor in Biomedicine at Halmstad University. She takes a special research interest in chronic inflammation, including immunology, molecular biology, and the interaction between chronic inflammation and physical exercise. Research projects include (-) the role of high intensity interval training in axial spondyloarthritis, (-) physical exercise and inflammatory and other biomarkers in children with cerebral palsy (CP), and (-) the role for a transcriptional repressor in chronic inflammatory disease.

She has also researched genetics for development of autoimmune disease, like multiple sclerosis and rheumatoid arthrtis in experimental models, and investigated the immunoglobulin gene repertoire in type 1 diabetes.

Key research interests

- Molecular aspects of chronic inflammation
- Development of biomarkers in chronic inflammatory diseases and CP
- The role for physical exercise in chronic inflammatory, musculoskeletal

Stefano Comai

Stefano Comai, PharmD, PhD, is an Associate Professor of Pharmacology at the University of Padua, Italy, and an Adjunct Professor at McGill University, Canada. In 2024, he became President of the International Society for Tryptophan Research (ISTRY). His research focuses on the role of tryptophan metabolism—including serotonin, melatonin, and kynurenine pathways—in brain function and their contribution to the etiology and treatment of mental disorders and related comorbidities. His work aligns with personalised medicine principles, aiming to identify biomarkers for disease progression and treatment response in neuropsychiatric disorders. He also explores innovative psychopharmacological targets, such as the endocannabinoid system and psychedelics.

William Duddy

Dr. Duddy is Senior Lecturer in Stratified Medicine (Bioinformatics) at the Personalised Medicine Centre in Ulster University's School of Medicine. As co-lead of the neuromuscular research team, he builds on a diverse background in molecular & cellular biology, biochemistry, and bioinformatics, to integrate systems biology data analytics into the team's cell and molecular studies. This integrative approach has led the team to investigate the role of secretory vesicles in neuromuscular and neurodegenerative disorders, and to their identification of vesicles as a source of neuronal toxicity. Dr. Duddy's commitment to neuromuscular research and wider interest in Personalised Medicine derive from losing his brother to Duchenne muscular dystrophy.

Delia M. Sánchez Varela

Dr. Delia M. Sánchez Varela is a distinguished medical doctor and bioethics expert with a career spanning over three decades in public health, bioethics, and international health. She holds a Doctorate in Medicine from the Universidad de la República (UdelaR), Uruguay, a Master's in Public Health and Community Medicine from the Hebrew University of Jerusalem, Israel, and a Master's in Bioethics from the Universidad Libre Internacional de las Américas. She is also a Specialist in International Health (PAHO) and holds a diploma in economics for non-economists from UdelaR.

Dr. Sánchez Varela served as Associate Professor and Coordinator of the Academic Unit of Bioethics at UdelaR and has been an influential member of various ethics and bioethics committees, including UNESCO's International Bioethics Committee and the National Commission on Health Research Ethics. She has held leadership roles in the Ministry of Health, the MERCOSUR Health Systems Observatory, and international organizations such as COHRED and the Alliance for Health Policy and Systems Research. A prolific author, she has contributed to over 50 publications in public health and bioethics, shaping policy and ethical frameworks in Latin America and beyond.

Presentation Abstracts

AGORA: Algorithm of Graft Outcome in Renal Allotransplantation

Presented by: Sophie Brouard
Nantes University Hospital, France

In renal transplantation, monitoring the risk of subclinical rejection and graft failure remains challenging. AGORA partners developed and validated non-invasive biomarkers allowing the prediction of patients at high/low risk of subclinical rejection and graft failure. The objectives are to build a European non-invasive clinical decision-making tool for immunological risk stratification of graft failure through a retrospective study on an existing biocollection of 300 patients and a multicenter randomized open label trial of immunosuppression minimization AGORAC. Economic efficiency will be evaluated during the trial. We will integrate users' perspectives by performing sociological interviews to promote the involvement of patients in their clinical care and to help clinicians in decision-making. Ancillary functional studies will reinforce AGORA to assess immunological events following immunosuppression minimization. AGORA could impact health care pathway for kidney transplant recipients by incorporating a medical decision tool for personalising immunosuppressive therapy.

Coordinator:
Sophie Brouard, Institute of Transplantation Urology and Nephrology (ITUN), Nantes University Hospital, France

Partners:
Oriol Bestard, Vall d'Hebron University Hospital, Spain
Anders Åsberg, Rikshospitalet, Oslo University Hospital, Norway
Nicolas Degauque, UMR 1064 Centre de Recherche en Transplantation et Immunologie (CRTI), University of Nantes, France

ArtiPro: Artificial intelligence for personalised medicine in depression - analysis and harmonization of clinical research data for robust multimodal patient profiling for the prediction of therapy outcome

Presented by: Julia C Stingl
University Hospital of RWTH Aachen, Germany

Personalised medicine aims to predict therapeutic response according to a personal profile that includes clinical, biological, and genetic data. This project focuses on depression. It aims to establish an artificial intelligence platform that brings together data from clinical research on the components of these profiles with the purpose of identifying predictors for response to depression treatment. The results will be combined into a single data platform that enables the use of large multimodal datasets to develop predictive models of symptoms and outcome data, thus enhancing the impact of these data. Artificial intelligence approaches will be investigated to identify novel biomarkers that can predict response to treatment. This will help to develop of a decision support system for personalized therapy while identifying the specific ethical and legal requirements that need to be fulfilled.

Coordinator:
Julia C Stingl, University Hospital of RWTH Aachen and University Hospital Heidelberg, Germany

Partners:
Maria Giulia Bacalini, IRCCS Istituto delle Scienze Neurologiche di Bologna, Italy
Roberto Viviani, Institute of Psychology, University of Innsbruck, Austria
Noam Shomron, Tel Aviv University, Israel
Espen Molden, Center for Psychopharmacology, Diakonhjemmet Hospital, University of Oslo, Norway
Catharina Scholl, Federal Institute for Drugs and Medical Devices, BfArM, Bonn, Germany
Nada Bozina, Dept of Pharmacology, School of Medicine, University of Zagreb, Croatia

BRAVA: Behaviours in REM Sleep: Personalised Automatic 3D Video Analysis as Novel Tool to Detect Alphasynucleinopathies

Presented by: Matteo Cesari
Medical University of Innsbruck, Innsbruck, Austria

Rapid eye movement (REM) sleep behaviour disorder (RBD) is characterised by abnormal muscular activity and dream enactment in REM sleep. In its isolated form (iRBD), it is recognised as an early stage of alpha-synucleinopathies (i.e. Parkinson's disease, dementia with Lewy bodies and multiple system atrophy). However, iRBD is often not recognised. An early, accurate, automated and population-extended recognition of iRBD, would be essential to recognise patients in early stages of alpha-synucleinopathies, enabling a timely initiation of disease modifying treatments. Furthermore, objective and automated methods would improve follow up of iRBD patients and allow personalised treatments. We aim to develop and validate a novel small, light and portable 3D video-based technology employing artificial intelligence as powerful, automatic, stand-alone instrument to identify and follow-up iRBD patients. We believe that this novel tool can revolutionise the way in which iRBD patients are identified and followed-up.

Coordinator:
Birgit Högl, Medical University of Innsbruck, Innsbruck, Austria

Co-Coordinator:
Matteo Cesari, Medical University of Innsbruck, Innsbruck, Austria

Partners:
Federica Provini, UOC Clinica Neurologica Rete Neurologica Metropolitana – NeuroMet Bellaria Hospital, IRCCS Istituto delle Scienze Neurologiche di Bologna, Bologna, Italy

Alex Iranzo, Consorci Institut d'Investigacions Biomèdiques August Pi i Sunyer (IDIBAPS), Barcelona, Spain

Claudia Trenkwalder, Center for Parkinsonism and Movement Disorders, Paracelsus-Elena-Klinik, Kassel, Germany

Isabelle Arnulf, Sleep Disorders Unit, Assistance Publique Hôpitaux de Paris-Sorbonne, Paris, France

CORSAI: Raman analysis of saliva from COPD patients as new biomarker: AI-based point-of-care for the disease monitoring and management

Presented by: Marzia Bedoni
IRCCS Fondazione Don Carlo Gnocchi ONLUS, Milan, Italy

Chronic obstructive pulmonary disease (COPD) is a debilitating and chronic pulmonary syndrome that causes a rapid decline in lung function. Nowadays, there is not specific biomarker that allows its immediate identification and the phenotyping of COPD patients is based on very long standard procedures, exposing them to the high risk of exacerbation and hospitalisation. Therefore, it is of primary importance to search for a unique biomarker that can help clinicians in the differential diagnosis of COPD patients from those with asthma, the evaluation of their exacerbation risk and the identification of non-adherence to therapy. On these bases, the main goal of the CORSAI project is to validate a new method based on the Raman spectroscopy (RS) analysis of saliva (ideal biofluid for diagnostics and monitoring purposes, as the collection procedure is minimally invasive) for the optimised and personalised management of COPD patients. The Raman spectrum of saliva (Raman fingerprint) will represent a single biomarker for COPD, obtained in a sensitive, fast and label-free manner. By the combination of RS-based method with artificial intelligence (AI), the project will lead to the COPD patients' management in a personalised medicine dimension, with a particular focus on stratification of patients, prediction of the risk of exacerbation and adherence to therapy. Finally, the ultimate goal is to transfer the RS directly to the hospital, thanks to the use of a portable Raman spectroscope: it will be possible to test the effectiveness of a point of care method able to investigate different aspects of COPD in a single analysis.

Coordinator:
Paolo Banfi, IRCCS Fondazione Don Carlo Gnocchi ONLUS, Milan, Italy

Co-coordinator:
Marzia Bedoni, IRCCS Fondazione Don Carlo Gnocchi ONLUS, Milan, Italy

Partners:
Messina Vincenzina, Università degli studi di Milano Bicocca (UniMIB), Milan, Italy

Heinz Manuel, Geratherm Respiratory GmbH (GERA), Germany

Soler Nestor, Institut d'Investigacions Biomèdiques August Pi I Sunyer (IDIBAPS), Barcelona, Spain

Tirzīte Madara, Riga Stradins University (RSU), Riga, Latvia

CytoMARK: Development of a personalised non-invasive diagnosis of endometrial cancer using proteomic markers in cervical fluids and clinical data

Presented by: Irene de la Calle Fuentes
Vall d'Hebron Research Institute, Barcelona, Spain

Endometrial cancer is the fourth most common cancer in women, and its incidence is increasing. Early detection is fundamental to patients' survival. Currently, no screening methods are available and diagnosis is a multistep process that includes minimally-invasive and invasive tests. This inaccurate diagnostic process is a burden on our healthcare system.

This funded project aims is to advance the development of a non-invasive, objective, and personalised diagnostic tool of endometrial cancer using cervical fluid protein biomarkers and clinical data. In this proposal, we will validate protein biomarkers in a retrospective clinical study coordinated by VHIR (Spain) and with the top-edge technology on targeted proteomics, led by the LIH partner (Luxembourg). Molecular markers will be combined with clinical data by the USC partner (Spain) and the most promising biomarkers will be transferred to an antibody technology by ICOSAGEN (Estonian SME partner) and SolarBiotec (Türkiye SME partner). Throughout the project, the HU partner (Türkiye) and VHIR will ensure the clinical validation of the developed non-invasive tool and the valorisation of the asset to meet the stakeholders' requirements. The resulting tool is a change in the paradigm on how endometrial cancer patients are managed and will benefit patients, doctors and the health system.

Coordinator:
Eva Colas, Vall Hebron Institute of Research (VHIR), Barcelona, Spain

Partners:
Gunnar Dittmar, Luxembourg institute of health, Strassen, Luxembourg
María Ángeles Casares de Cal, Faculty of Mathematics, Universidad de Santiago de Compostela, Spain
Andres Tover, Icosagen Cell Factory, Tartu, Estonia
Murat Gultekin, Hacettepe University Hospitals, Ankara, Türkiye
Yalın Kılıç, SOLAR BİYOTEKNOLOJI LTD, Izmir, Türkiye

ECLAI: Personalised Clinical Management of Endometrial Cancer using Liquid Biopsy, Genomics and Artificial Intelligence

Presented by: Gema Moreno Bueno
Consorcio Centro de Investigación Biomédica en Red M.P (CIBER), Spain

Precision oncology represents a challenge when it comes to Endometrial Cancer (EC). The incidence of this cancer has increased in the last years and the prognostic and therapeutic options for advanced disease stages are still poor. Thus, to improve the treatment of EC patients with poor prognosis, it is necessary to gain more knowledge in the cancer molecular biology and also in the new approaches to capture the innate intra-tumour heterogeneity (ITH), which is highly present in EC. The objective of the ECLAI consortium is to reach personalised EC management by developing new tools that recapitulate the heterogeneous molecular composition of tumors and finally establish new and effective therapeutic regimens. With this aim, our consortium will combine: a) the use of non-invasive biopsies, which capture ITH, and the genomic characterisation of the tumor within the disease evolution, b) the generation of preclinical models to test personalised alternative targeted therapies and c) the use of machine learning strategies to decipher a recurrence and therapeutic response rate algorithm, named ECLAI, with clinical application to improve EC management. This workflow will be applied with the advise and support of patients' associations and ENITEC, the European research network on EC. Altogether, this pioneering strategy has the final goal to improve the management and life quality of EC patients who currently have limited clinical opportunities.

Coordinator:
Gema Moreno Bueno, Consorcio Centro de Investigación Biomédica en Red M.P (CIBER), Spain
Project Manager: Laura Muinelo Romay, Consorcio Centro de Investigación Biomédica en Red M.P (CIBER), SPAIN

Partners:
Marcin Stanisław Bobiński, Medical University of Lublin, Poland
Camilla Krakstad, University of Bergen, Norway
Andres Salumets, Tervisetehnoloogiate Arenduskeskus AS (Competence Centre on Health Technologies (CCHT), Estonia

EDAP-AD: Early and accurate diagnostic and prognostic markers for Alzheimer's disease

Presented by: Rik Ossenkoppele
Clinical Memory Research Unit, Lund University, Sweden

Alzheimer's disease (AD) has a major impact on daily functioning and quality of life and is characterised by a huge socio-economic burden, with at least 50 million people affected worldwide. For clinical practice, we need to develop algorithms based on easily accessible and time- and cost-effective tests like blood-based biomarkers and digital cognitive tests for personalised diagnosis, prognosis and correct symptomatic treatment. For clinical trial development, we need to optimise the screening procedure to accurately identify individuals with AD in pre-symptomatic stage or prodromal disease stages and predict progression rates at an individual level. The methods need to be robust and generalizable, and the test results need to be optimally communicated to patients. This initiative will leverage demographically and ethnically diverse population-based, primary care and memory clinical cohorts that are deeply phenotyped and have long-term follow-up data available. Using state-of-the-art machine learning approaches, we will define, validate and implement accessible and cost-effective AD biomarkers for personalised diagnostic and prognostic work-up, and to facilitate development of disease modifying treatments in AD. By revolutionising the diagnostic work-up and improving participant selection and monitoring for clinical trials, the personalised medicine approach developed in EDAP-AD will meet the challenges posited by AD.

Coordinator:
Rik Ossenkoppele, Clinical Memory Research Unit, Lund University, Sweden

Partners:
Gunhild Waldemar, Danish Dementia Research Centre, Rigshospitalet, Denmark

Marc Suárez-Calvet, IIS Institut Hospital del Mar d'Investigacions Mèdiques, Spain

Bruno Vellas, Toulouse University Hospital, France

Michael Ewers, University Hospital, Ludwig-Maximilian-University Munich, Germany

Julia Anne Schnabel, Helmholtz Center Munich, Germany

IMAGene: Epigenomic and machine learning models to predict pancreatic cancer: development of a new algorithm to integrate clinical, omics, DNA methylation biomarkers and environmental data for early detection of pancreatic cancer in high-risk individuals

Presented by: Serena Oliveri
Istituto Europeo di Oncologia (IEO) IRCCS, Italy

Pancreatic cancer (PC) has the lowest survival rate of all cancers in Europe, with no early detection strategies available. First-degree relatives of patients with PC have at least a 2-fold increased risk of developing the disease, although it is well-known that besides family history, also older age, tobacco, alcohol abuse and other epidemiological risk factors predispose to PC onset. Even in families with high genetic predisposition to PC, existing cancer predictive Machine Learning models are of very limited use, since their predictive accuracy is generally low. Epigenetic biomarkers are not a part of existing risk indexes yet, although strong evidence shows that methylation patterns in blood can efficiently predict cancer mortality and that liquid biopsy has a potential to revolutionise early cancer diagnostics. The IMAGene project will develop, calibrate and test a comprehensive Cancer Risk Prediction Algorithm (CRPA) to predict PC in high-risk (HR) asymptomatic subjects, by including omics, imaging, epidemiologic, lifestyle and psychological data records. IMAGene will also investigate the potential for DNA methylation biomarkers to improve currently available risk indexes, and validate the feasibility of using liquid biopsies for early detection of cancer in HR individuals. A detailed ethical and cost-utility analysis will respectively guide a responsible application of the procedures and will balance benefits and impact for the health care system in four EU countries.

Coordinator:
Serena Oliveri, Istituto Europeo di Oncologia (IEO) IRCCS, Italy

Partners:
Victor Moreno, Bellvitge Biomedical Research Institute (IDIBELL), Catalan Institute of Oncology (ICO), University of Barcelona (UB) and CIBER of Epidemiology and Public Health, Spain

Tomasz K Wojdacz, Pomeranian Medical University in Szczecin (PMU), Poland

Ovidiu Balacescu, The Oncology Institute "Prof Dr. Ion Chiricuta" (IOCN), Romania

Louis Buscail, Centre Hospitalier Universitaire de Toulouse (CHUT), France

i-RECORDS: International – Rapid rEcognition of CORticosteroiDs sensitivity or resistance in Sepsis

Presented by: Djillali Annane
Assistance Publique – Hôpitaux de Paris, France

Sepsis and COVID-19 are a major burden for populations worldwide. In sepsis/COVID-19, a dysregulated host response to infection is the hallmark supporting the routine use of corticosteroids (CS), a low-cost and highly efficient class of immunomodulators. Stratifying patients based on individual immune response may improve the balance of benefit-to-risk of CS treatment.
iRECORDS will generate signatures of CS sensitivity/resistance of individual septic/COVID-19 patients. These signatures will be based on characterisations of biological systems by using targeted approaches at levels of DNA, RNA, proteins (i.e. cytokines), hormones, and metabolites. Artificial Intelligence methods will be used to develop efficient signatures by integrating high dimensional multi-level data from previous studies and newly generated data. The resulting signatures will define personalised treatment rules for patients, and thereby improve their chance to survive.

Coordinator :
Djillali Annane, Assistance Publique – Hôpitaux de Paris, France

Partners :
Karine Zeitouni, Université de Versailles Saint-Quentin-en-Yvelines, France
Josef Briegel, Ludwig-Maximilians-University, Germany
Michael Bauer, Jena University Hospital, Germany
Jesús Villar, Fundación Canaria Instituto de Investigación Sanitaria de Canarias - CIBER de Enfermedades Respiratorias, Instituto de Salud Carlos III, Spain
Yasser Nassar, Cairo University, Egypt

MIRACLE: A machine learning approach to identify patients with resected non-small-cell lung cancer with high risk of relapse

Presented by: Paola Ulivi
IRST-IRCCS, Italy

Early-stage non-small-cell lung cancer (ES-NSCLC) represents 20-30% of all NSCLC and is characterised by a high survival rate after surgery, with variability in clinical outcome among patients sharing the same disease stage, suggesting that other factors could determine the risk of relapse. We hypothesize that multiple factors could influence the prognosis of resected ES-NSCLC patients, such as tumour tissue and tumour microenvironment (TME) characteristics, liquid biopsy, radiomics features and clinical-pathological factors. MIRACLE aims to develop and validate a machine learning algorithm acting as a clinical decision support tool for disease free survival prediction based on joint analysis of biological, clinical and radiologic features. A previously prospectively collected cohort of ES-NSCLC patients will be considered as a training set. Tumour tissue and TME characteristics will be analysed using DNA and RNA sequencing; liquid biopsy will be used to assess free circulating DNA and extracellular vesicles; radiomics parameters will be retrieved from computed tomography images. All these features, together with clinico-pathological factors, will be integrated in a model that will enable personalised patient treatment. The developed algorithm will be validated in a prospective cohort enrolled during the timeframe of project MIRACLE.

Coordinator:
Paola Ulivi, IRST-IRCCS, Italy

Partners:
Enriqueta Felip, Fundacio Hospital Universitari Vall d'Hebron (HUVH) – Institut de Recerca (VHIR)/ Fundacio Privada Institut d'Investigacio Oncologica de Vall d'Hebron (VHIO), Spain
Julien Mazieres, Centre Hospitalier Universitaire Toulouse, France
Erhard Rahm, University of Leipzig, Germany
Vanessa Nurock, Université de Côte d'azur (CRHI), France

Pattern-Cog: Personalised aging pattern for early risk detection and prevention of cognitive impairment and dementia in cognitively healthy individuals

Presented by: Jussi Tohka
University of Eastern Finland, Kuopio, Finland

Pattern-Cog aims to improve dementia prevention strategies by developing and validating a machine learning-based personalised medicine framework for detecting the earliest signs of impending cognitive decline, enabling early and personalised multi-domain interventions. Findings from multi-domain lifestyle trials have emphasized that intervention effectiveness may be dependent on a methodology that does not yet exist, i.e., the accurate identification of at-risk individuals who are most likely to benefit. Pattern-Cog will address this methodological gap by (1) developing methods for predicting future cognitive decline based on clinical data and distinguishing between healthy individuals at higher risk for mild cognitive impairment and dementia and those who remain healthy; and (2) testing the methodology in ongoing dementia prevention trials. Instead of a standard machine learning approach, we propose an innovative concept of personalised aging pattern rooted in data from healthy individuals.

Coordinator:
Jussi Tohka, University of Eastern Finland, Kuopio, Finland

Partners:
Christian Gaser, Jena University Hospital, Jena, Germany
Francesca Mangialasche, Karolinska Institutet, Solna, Sweden
Alberto Rabano, Fundación Centro de Investigación de Enfermedades Neurológicas (FCIEN), Madrid, Spain
Petra Ritter, Charité Universitätsmedizin Berlin, Berlin, Germany
Jean Georges, Alzheimer Europe, Luxembourg

PerCard: Personalised Prognostics and Diagnostics for Improved Decision Support in Cardiovascular Diseases

Presented by: Mark van Gils
Tampere University, Finland

Cardiovascular Diseases (CVD) account for 45% of all deaths in Europe. Eighty percent of premature heart diseases are preventable, if personal risks can be identified early. However, currently used risk models 1) do not reflect true populations, especially with regard to gender, 2) do not give sufficient consideration to the genetic backgrounds of individuals, and 3) do not use all the information available in different data sources.
PerCard combines different data with novel analysis methods (AI, machine learning, signal processing) to deliver an improved risk modelling tool. The developed methods are explainable, practical, accessible, and affordable. Development combines existing Finnish and Italian data and new-to-be-collected data in Italy. Ethical and societal aspects, including gender and accessibility to all, receive special attention.
PerCard's international consortium is formed by Tampere University (Finland, coordinator), Polytechnic University of Milan (Italy), Centro Cardiologico Monzino (Italy), and Protestant University of Applied Sciences Ludwigsburg (Germany).

Coordinator:
Mark van Gils, Faculty of Medicine and Health Technology, Tampere University, Finland

Partners:
Luca Mainardi, Information and Bioengineering, Politecnico di Milano, Italy
Claudio Tondo, Centro Cardiologico Monzino, Italy
Kirsten Brukamp, Health Sciences, Protestant University of Applied Sciences Ludwigsburg, Germany

PerFluid: Personalised perfusion guided fluid therapy

Presented by: Thomas Desaive
GIGA – In Silico Medicine, University of Liège, Belgium

Acute circulatory failure (ACF) and shock affect ~30% of intensive care unit (ICU) patients and occurs when the heart and circulation cannot perfuse critical organs to support their function. Adding fluid is the primary treatment to restore perfusion. However, only 50% of fluid interventions are effective. The rest have deleterious or no effect.

There is a critical need to develop a clinically feasible, low-cost means to determine which patients will respond to fluid, and which will not.

PerFluid will develop a novel model-based method to continuously measure stressed-blood volume to assess perfusion, and thus personalise fluid therapy treatment of ACF patients in the ICU. We will combine physiological models and currently available measurements to capture currently unmeasurable key physiological parameters in real-time – in particular stressed blood volume and perfusion. This metric will be combined with a novel, yet simple clinical protocol for validation in clinical proof-of-concept tests.

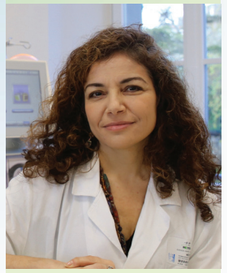
Using only currently available measurements makes PerFluid far more clinically feasible. The protocol is designed to integrate seamlessly with typical care practice, so no work is added. Digitally driven, it is potentially very low cost, with significant potential to address the high mortality and cost of ACF by personalising and optimising care.

Coordinator:
Thomas Desaive, GIGA – In Silico Medicine, University of Liège, Belgium

Partners:
Balázs Benyó, Budapest University of Technology and Economics, Hungary
Knut Möller, Institute of Technical Medicine, Furtwangen University, Germany
Endre Zima, Heart and Vascular Center, Semmelweis University, Hungary
J. Geoffrey Chase, University of Canterbury, New Zealand

PER-NEPH: Implementation of PERsonalised management in NEPHrotic syndrome

Presented by: Paola Romagnani
Meyer Children's Hospital, Florence, Italy



Nephrotic syndrome in children and young adults is a medical problem of diverse pathophysiology and prognosis. Current diagnostic algorithms fail to avoid under- and overtreatment with toxic drugs as defining the underlying cause is difficult. We developed a diagnostic algorithm to stratify patients through advanced genetic testing, reverse phenotyping, and personalised disease models. This can double the current diagnostic rate in patients not responding to therapy and to predict disease relapse in those that progress to end stage kidney disease and undergo kidney transplant.

The aims of this project are:

1. Implementation of this diagnostic algorithm in selected European sites by a) selecting patients for a genetic testing, b) whole exome sequencing, c) validating genotype-phenotype correlation, and d) assessment of variants of unknown significance by functional studies with patient urine-derived renal progenitors disease models.
2. Personalising the assessment of non-genetic forms and of relapse after transplant by identifying patients negative to the genetic testing and with proteinuria relapse after kidney transplant, as well as personalising the detection of immunologic factors by super resolution microscopy and circulatory permeability factors by 3D organ-on-a-chip model system.
3. Assessing the cost-effectiveness as well as clinical, ethical, and legal consequences of this algorithm.

Coordinator:
Paola Romagnani, Meyer Children's Hospital, Florence, Italy

Partners:
Elena Lazzeri, University of Florence, Italy
Josep M. Cruzado, Hospital of Bellvitge, Bellvitge Biomedical Research Institute (IDIBELL), L'Hospitalet de Llobregat, Catalonia, Spain
Hans-Joachim Anders, University Hospital of Ludwig-Maximilians University Munich (LMU Klinikum), Germany

PerSAIDs: PERsonalised medicine for SAIDs

Presented by: Isabella Ceccherini
IRCCS Istituto Giannina Gaslini (IGG), Italy

Systemic Auto-Inflammatory Diseases (SAIDs) are a growing number of rare conditions with monogenic or multifactorial genetic etiology, causing deregulation of the mechanisms that control innate immune responses. For specific monogenic SAID, personalised medicine (PM) is already an established reality. However, there is evidence that personalised approaches could be beneficial also for other SAIDs, 70-80%, known as “undefined” (uSAIDs) due to the fact that molecular testing cannot provide diagnostic confirmations.
The present proposal will link some of the largest registries and bio-sample repositories on SAIDs in Europe to i) analyse available data and produce new omics, ii) generate standardised protocols and common bioinformatics pipelines for data management and analysis, iii) simplify reuse of data in compliance with FAIR principles and GDPR. We will also develop appropriate tools to decode the disease complexity, thus improving SAIDs classification, diagnosis and prognosis, and supporting the discovery of novel therapies.

Coordinator:
Isabella Ceccherini, IRCCS Istituto Giannina Gaslini (IGG), Italy

Partners:
Sergio Decherchi, Fondazione Istituto Italiano di Tecnologia (IIT), Italy
Dirk Foell, University of Muenster (WWU), Germany
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PETictCAC: Predicting risk of cachexia in cancer patients

Presented by: Thomas Beyer
Medical University Vienna (MUV), Austria

Unintentional weight loss is common in patients suffering from advanced cancer. This condition has long been recognised as a frequent and life-threatening complication of many malignancies, but research has only recently begun to uncover its molecular basis. Clinicians refer to cancer associated cachexia (CAC) once weight loss exceeds 5% over 6 months. Therapeutic strategies to revert weight loss after it commenced are ineffective, possibly because they are initiated too late or tackle the wrong pathway. The project explores novel approaches to diagnose CAC before weight loss occurs to implement therapeutic interventions earlier.
We propose to develop a computational framework that uses artificial intelligence (AI) to support an automated analysis of interactions between organs that we can visualise and quantify non-invasively using positron emission tomography (PET). PET imaging of glucose uptake reveals the metabolic connectivity between organs and can conceivably detect the mobilisation of resources in fat and muscle tissue before weight loss weakens the patient. If successful, our computational framework will be developed into a clinical decision-making tool integrating ethical and legal considerations. This tool will determine individual patient risk to develop cachexia and, thus, support personalised therapeutic interventions.

Coordinator:
Thomas Beyer, Medical University Vienna (MUV), Austria

Partners:
Roberto Sciagrà, Azienda Ospedaliero Universitaria di Careggi (AOUC), Italy
Osama Sabri, University Leipzig (ULEI), Germany
Peter Sandøe, University of Copenhagen (UCPH), Denmark

PRE-CARE ML: Predicting Cardiovascular Events Using Machine Learning

Presented by: Stefan Kalabakov
Hasso-Plattner-Institut, Potsdam, Germany

Cardiovascular disease is the leading cause of death and causes tremendous suffering, socioeconomic loss, and burden to health systems. Atherosclerosis, a condition that is clinically silent, i.e. it often remains undetected until it is too late, underlies major cardiovascular events such as heart attacks and strokes.
Early identification of people at high risk for such clinical events enables preventive actions. However, conventional risk prediction scores are often not widely adopted in otherwise healthy and symptom-free people. At the same time, medical information is increasingly digitalised. This leads to huge amounts of electronic health data amenable to risk prediction. Yet, conventional approaches fail to handle this data in its entirety and harness it for medical decision-making.
Here, we use artificial intelligence (AI) methods to develop modern risk prediction tools for early identification of people at high risk for major cardiovascular events. This endeavor builds on our previous experience in using machine-learning algorithms for risk prediction. In our multidisciplinary consortium, we aim to validate and improve our models across different hospital networks and populations. Second, we will integrate our models in hospital information systems and assess their impact on daily hospital routine. Lastly, we will address effective risk communication strategies in order to effect behavioral changes in patients.
Our ultimate aim is to develop easy-to-use, accessible, and reliable risk prediction tools that allow early identification of people at high risk in order to set actions to prevent major cardiovascular events and thereby reduce the global impact of cardiovascular disease.

Coordinator:
Peter P. Rainer, Medical University of Graz, Graz, Austria

Partners:
Erwin Böttinger, Hasso Plattner Institute for Digital Engineering, Potsdam, Germany and Icahn School of Medicine at Mount Sinai, New York City, USA
Max Gordon, Karolinska Institutet, Stockholm, Sweden
Paulo Mazzoncini de Azevedo Marques, Ribeirão Preto Medical School at University of São Paulo, São Paulo, Brazil
Ekart Robert, University Clinical Centre Maribor, Maribor, Slovenia

REDESIGN: TREatment DEcision baSed on organoIds in Gastric cancer

Presented by: Franziska Baenke
University Hospital Carl Gustav Carus Dresden, Dresden, Germany

The REDESIGN consortium proposes a multidisciplinary programme based on patient-derived organoids (PDOs) to guide personalised treatment in gastric cancer (GC) patients. Standard-of-care for locally advanced GC in Europe is neoadjuvant FLOT chemotherapy. Pathological response to neoadjuvant treatment is directly linked to overall survival. However, 63% of patients show resistance to the treatment and have no pathologic major response after treatment. Two different mechanisms are important in the aetiology of resistance: intra-tumoural heterogeneity and the accumulation of mutations that circumvent the point of action of a given drug. Deciphering intra-tumoural heterogeneity and predicting mutations that emerge under selective pressure of treatment will enable early identification of upcoming drug resistance, allowing tailored treatment strategies before the start of treatment.
The consortium aims to unravel treatment resistance mechanisms and design treatment strategies that consider the identified causes of resistance. By using multiple biopsies derived from gastric cancer patients at the time of diagnosis, exposing them to the standard of care FLOT treatment as well as pharmacotyping with other small agents, differential responses were observed. Integrating the functional drug response data with the genomics of these PDOs, we investigate the selective treatment pressure caused by FLOT to the multiple PDOs to dissect clonal evolution within a tumor. The trajectories of the PDOs can lead to the identification of potential sensitivities to other agents. Additionally, the patient's perspective of their patient care was assessed during this study to reveal the patient's understanding of PDOs and personalised treatment decisions. The overarching aim is to improve personalised treatment by combining the gained knowledge of evolutionary trajectories and mechanisms of resistance to therapy as well as addressing ethical and social challenges when using PDOs to aid clinical decision making in the era of precision medicine.

Coordinator:
Daniel E. Stange, University Hospital Carl Gustav Carus Dresden, Dresden, Germany

Partners:
Mette N. Svendsen, University of Copenhagen, Copenhagen, Denmark
Bon-Kyoung Koo, Institute of Molecular Biotechnology of Austrian Academy of Sciences, Vienna, Austria
Steffen Rulands, Max Planck Institute for the Physics of Complex Systems, Dresden, Germany

ScandRA: Personalised medicine in RA by combining genomics, biomarkers, clinical and patient-data from the Scandinavian countries, and by integrating the knowledge generated into routine care

Presented by: Johan Askling
Karolinska Institutet, T2 Karolinska University Hospital, Stockholm, Sweden

Rheumatoid Arthritis (RA) is a chronic disease that displays a significant variation in clinical picture, response to therapy, and longer-term outcomes. With few and crude predictors available, our means for an individualised strategy are limited. A personalised medicine (PM) approach to RA requires new biomarkers, and algorithms, to support diagnosis and choice of effective treatment. For PM to transform clinical rheumatology practice, new tools to bring such algorithms to patients and to clinical practice are needed. ScandRA builds on our successful collaboration between academia, healthcare, patients, and the private sector in biomarker technologies, data interoperability and e-health. In the Scandinavian countries, we have collected large amounts of information on patients with RA in our longitudinal prospective registers and biobanks, the largest and most detailed world-wide. We are now uniquely positioned i) to make detailed clinical data on RA disease activity, treatment, and life-style available for joint analyses with novel genomic- and biomarker-data from blood samples from these cohorts, ii) to take the next step, towards integration of the emergent results into clinical practice. Planned work include i) knowledge generation from novel analyses of our RA data, ii) value creation through development of decision support tools based on these insights, and iii) work with the ethical, legal and social challenges that are necessary for successful implementation.

Coordinator:
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Hilde Berner Hammer, Diakonhjemmet Hospital, Oslo, Norway
Johan Rönnelid, Uppsala University, Uppsala, Sweden
Sascha Swiniarski, Phadia GmbH, Freiburg, Germany
Niels Steen Krogh, Zitelab ApS, Copenhagen, Denmark

SYMMETRY: Subpopulation Heterogeneity and Microenvironmental Engagement as Predictors for Treatment Resistance in Lymphoma

Presented by: Ekaterina Popova
Universitätsklinikum Düsseldorf (UKD), Germany

Lymphomas, like most other cancers, consist of multiple distinct cancer cell populations within each patient. These tumour subpopulations have distinct genetic and biological characteristics leading to different drug response profiles. We will characterise these tumour subpopulations in aggressive lymphoma on multiple biological levels including gene mutations, gene expression, protein abundance, surface protein profiles, and drug response. Using the in-depth characterisation of subpopulations in the discovery cohort we will investigate large clinical patient cohorts with multiparametric immunofluorescence and describe the association of subclonal histology and response to chemotherapy. The improved understanding of tumour subpopulations and their impact on therapy efficacy combined with the ability to detect those subpopulations in diagnostic biopsies using multiparametric immunofluorescence has the potential to improve treatment stratification and thereby patient survival while reducing side-effects and treatment costs. We will perform in-depth interviews with patients treated in our centers for personalised medicine to optimise the communication of increasingly complex diagnostic and therapeutic procedures in personalised medicine. These data will be complemented by quantitative surveys to create communication guidelines for personalised medicine.

Coordinator:
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Peter Horvath, Single-Cell Technologies Ltd., Szeged, Hungary
Sabina Sangaletti, Fondazione IRCCS Istituto Nazionale Tumori, Milan, Italy
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Julia Inthorn, Evang. Landeskirche Hannovers, Center for Health Care Ethics, Hanover, Germany
Ilze Štrumfa, Rīga Stradiņš University, Rīga, Latvia

TECH-TOYS: Acquire digiTal biomarkErs in infanCy witH sensorized TOYS for early detection and monitoring of neurodevelopmental disorders

Presented by: Giuseppina Sgandurra
IRCCS Fondazione Stella Maris, Pisa, Italy

Neurodevelopmental disorders are a group of frequent (1/10 children) sensori-motor, cognitive, communication, learning, behavioural disorders of multifactorial aetiology, with onset early in life but with life-long consequences. Despite advances in our understanding of aetiology, diagnosis and start of intervention are often late (many months after onset of first clinical signs) and not based on quantitative data. TECH-TOYS aims to develop a new technological home interactive play setting (i.e. a gym equipped with a sensorised mat, a set of sensorised toys, wearable inertial movement units and cameras) to provide easy-to-handle quantitative digital biomarkers of infant's neurodevelopment and infant-caregiver interaction. Previously big data acquired, and new ones collected prospectively on motor behaviours, together with gaze activities and social competence in infant-caregiver interaction, will provide an Explainable Artificial Intelligent Precision Model for early detection of atypical features. Ethical, Legal, Social aspects (ELSA) and Health Technology Assessment (HTA) will provide key factors in decision-making process and cost effectiveness analysis. Moreover, parents' organizations will have a strong involvement in the project activities and in the Ethics Monitoring Board and will contribute to the design of platform and of the Personalised Precision Model. The results will open new frontiers for early, timely, personalised, home based, quantitative detection of neurodevelopment in the first months of life.

Coordinator:
Giovanni Cioni, IRCCS Fondazione Stella Maris, Pisa, Italy

Partners:
David Cohen, Salpétrière Hospital, Paris, France
Mohamed Chetouani, Institut des Systèmes Intelligents et de Robotique, Sorbonne University, Paris, France
George Marckman, Ludwig Maximilians University, Munich, Germany
Marco Pirini, Khymeia Srl, Padua, Italy
Hatice Kose, Istanbul Technical University, Istanbul, Türkiye

The following patient associations are supporting the TECH-TOYS project: Coordinamento Etico dei Caregivers, Italy: caregivers.pisa.it | ANDIAM, Italy: andiam.it | EPPURsiMUOVE, Italy: facebook.comASDeppursimuove | ENVOLUDIA, France: www.envoludia.org | SERCEV, Türkiye: sercev.org.tr

WODIA: Personalised Medicine Screening and Monitoring Programme for Pregnant Women Suffering from Preeclampsia and Gestational Hypertension

Presented by: Stefan Wagner
Aarhus University, Aarhus, Denmark

Half a million women die each year giving birth. The two pregnancy-related blood pressure complications, preeclampsia and gestational hypertension, are the cause of 76,000 mothers & ½ million infants dying each year. Infants who survive often experience long-term health problems, including cerebral palsy, chronic lung disease, blindness & hearing loss, and the resulting societal healthcare costs are high.

Existing work has proposed a range of screening procedures and algorithms for detecting those pregnancies that will later develop preeclampsia or gestational hypertension. However, these are resources intensive for the service providers to implement, and challenged with a high false positive rate of up to 10%.

With the WODIA project we aim to make it safer for women to give birth by identifying and tracking the early signs of preeclampsia and gestational hypertension while the complications are still preventable, by developing a personalised medicine screening, therapy, and home monitoring service.

WODIA will combine the maternal characteristics with a range of biomarkers. Together, these will allow for more effective targeted personalised medicine with individual medication dosing and fewer and more effective clinical visits.

Coordinator:
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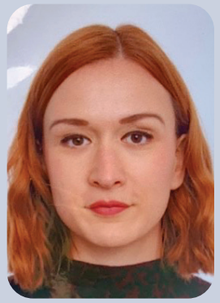
Preis Krzysztof, Gameta Gdynia Centrum Zdrowia Sp. z o.o, Medical University of Gdansk, Gdansk Poland
Lucian Andrei, Zitec Com SRL, Zitec Bucharest, Bucharest, Romania

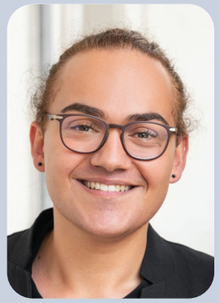
Poster Abstracts

Project ArtiPro

Patient-Reported Outcomes of Antidepressant Drug Therapy: A Comparative Analysis



Paula Marinovic



Jason-Christopher Radermacher

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3 University of Zagreb, School of medicine, Zagreb, Croatia
4 University Hospital Heidelberg, Department of Clinical Pharmacology, Heidelberg, Germany

Abstract:

Introduction: Patient-reported outcomes are becoming increasingly important in the evaluation of drug therapies. They capture the patient's perspective on their symptoms, treatment experiences, and overall well-being, unlike traditional clinical measures that focus only on objective assessments.

Objectives: To investigate patients' perceptions regarding the efficacy and safety of their antidepressant therapy.

Methods: 300 patients from three study centers were included - geriatric hospital, Germany (A), specialized psychiatric clinic, Germany (B), psychiatric outpatient clinic, Croatia (C). Patients were assessed with the same questionnaire that included questions about the patient's current state (possible symptoms) and questions about possible antidepressant side effects.

Results: Patients' knowledge towards the reason for their antidepressant therapy was significantly better in the specialized hospital cohorts (B and C) compared to the elderly patients in cohort A.

When asked openly, group C patients reported no side effects (99%) in comparison to 31% from group A and 71% from group C.

When asked specifically, patients from all three centers gave similar answers while with increasing age more side effects were stated.

Conclusions: Patient-reported outcomes are a great starting point for moving in the direction of patient-centered medicine and can help healthcare providers make more informed decisions regarding antidepressant drug therapy.

Project BRAVA

Automatic Identification of Nocturnal Movements using Depth Vide

Simon Feuerstein

Simon Feuerstein[1], Bernhard Kohn[2], Luca Baldelli3, Qi Tang[1], Merve Aktan Süzgün[1], Victoria Anselmi[1], Ambra Stefani[1], Alex Iranzo[4], Claudia Trenkwalder[5], Isabelle Arnulf[6], Birgit Högl[1], Federica Provini[3] and Matteo Cesari[1]

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2 Austrian Institute of Technology, Vienna, Austria
3 IRCCS Istituto delle Scienze Neurologiche di Bologna, Bologna, Italy AND Department of Biomedical and NeuroMotor Sciences, University of Bologna, Bologna, Italy
4 Sleep Unit, Neurology Service, Hospital Clínic Barcelona, Universitat de Barcelona, IDIBAPS, CIBERNED, Barcelona, Spain
5 Paracelsus-Elena Klinik, Kassel, Germany
6 AP-HP, Hôpital Pitié-Salpêtrière, Service des Pathologies du Sommeil, National Reference Centre for Narcolepsy and rare hypersomnias, Paris, France AND Sorbonne Université, Institut du Cerveau - Paris Brain Institute - ICM, Inserm, CNRS, APHP, Hôpital de la Pitié Salpêtrière, Paris, France

Abstract:

Visual identification of nocturnal patient's movements in sleep studies is time-consuming and subjective. We present an automated pipeline for identification of movements during rapid eye movement (REM) sleep using a depth camera with a bird's-eye view setup, offering a scalable and non-intrusive solution and compare it against movement annotations set by experts.

Data were collected from 25 participants across two European sleep labs, including individuals with isolated REM sleep behavior disorder (n=10), disorders of arousal (n=5), sleep-related hypermotor epilepsy (n=5), and controls (n=5).

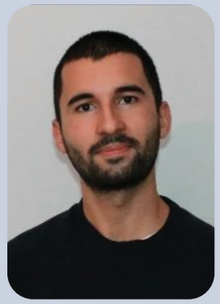
Depth videos were recorded using a PMD Flexx2 3D camera, recording in 224x172, 5 fps. To reduce noise in the depth videos, artifact removal addressing issues such low-confidence data and other outliers resulting from deep edges, reflective surfaces and airborne dust was applied.

Motion maps were generated using convolutional and optical flow methods, and movement signals derived from these maps were used to automatically infer movements. Evaluation was performed by comparing automatic annotations with expert annotations in REM sleep, calculating Intersection over Union, F1-score, precision, recall, and accuracy.

This approach supports broader accessibility for home-based studies as well as reducing expert workload in clinical and research settings.

Project IMAGene

A novel anomaly detection pipeline with Autoencoders for predicting PDAC from whole-blood DNA methylation data



Lois Riobó-Mayo

Lois Riobó-Mayo[1,2,3], Miguel Socolovsky[1,2,4] , Clizia Cincidda[5], Giulia Ongaro[5], Cindy Canivet[6], Louis Buscail[6], Ovidiu Balacescu[7], Tomasz Wojdacz[8], Serena Oliveri[5], Víctor Moreno[1,2,4]

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2 Doctoral Programme in Medicine. University of Barcelona (UB)
3 Consortium for Biomedical Research in Epidemiology and Public Health (IDIBELL)
4 Department of Clinical Sciences, Faculty of Medicine, University of Barcelona (UB),
5 IEO - Istituto Europeo di Oncologia,
6 Centre Hospitalier Universitaire de Toulouse (CHUT) ,
7 Oncology Institute Prof. Dr. I. Chiricuta Cluj-Napoca,
8 Clinical Epigenetics Laboratory, Pomeranian Medical University

Abstract:

Pancreatic cancer (PDAC) presents a significant global health challenge, mainly due to the absence of cost-effective screening methods. DNA methylation study, due to its known relationship with oncogènesis, represents a very promising approach. Moreover, Deep Learning (DL) algorithms are a particularly powerful tool for analysing patterns in high dimensional data.

For this analysis, whole-blood samples analyzed with Illumina microarrays were used for epigenome-wide association studies (EWAS). A DL based method called anomaly detection was employed for identifying genomic regions with abnormal methylation patterns.

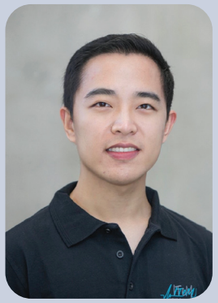
For this purpose, we used a dataset of 431 controls and 393 future PDAC cases. A subset of controls was used for training the model, a DL Autoencoder (AE). Once it is trained it can be used for identifying regions in which there are anomalies in its structure in comparison with these healthy patients.

In a validation of this model as a predictive tool we used a different independent dataset, coming from the Framingham Cohort study. The algorithm distinguished between groups with an AUC: 0.713 [0.574 – 0.840].

Our pipeline has yielded statistically significant results , but limitations in sample size must be solved in order to refine the algorithm and increase the robustness of the validation.

Project PerFluid

Harmonic Analysis for Separating Ventilation and Perfusion Signals in Electrical Impedance Tomograph



Rongqing Chen

Rongqing Chen[1], Alberto Battistel[1], András Lovas[2], Balázs Benyó[3], Stefan J. Rupitsch[4] and Knut Möller[1]

1 Institute of Technical Medicine, Hochschule Furtwangen, Germany
2 Department of Anaesthesiology and Intensive Therapy, Kiskunhalas Semmelweis Hospital, Hungary
3 Department of Control Engineering and Information Technology, Faculty of Electrical Engineering and Information Technology, Budapest University of Technology and Economics, Hungary
4 Department of Microsystems Engineering (IMTEK), Faculty of Engineering, University of Freiburg, Germany

Abstract:

Electrical Impedance Tomography (EIT) is primarily used to monitor respiratory activity. However, cardiac-related signals, though smaller in amplitude, are also present and can be distinguished by their frequency. We introduced a harmonic analysis method to separate respiration and perfusion signals in EIT data.

The method models EIT signals as amplitude-modulated components at distinct frequencies. This mathematical framework is applied to global impedance data to generate frequency-specific images that isolate respiration and perfusion dynamics. Cardiac activity, alongside pulmonary activity, is detected in both the heart and lung regions through EIT results. These impedance variations primarily result from localized shifts in blood volume, as blood's higher conductivity influences regional impedance during its flow.

The separation results reveal conflicting and delayed impedance behavior in the lung region, likely due to arterial pulse-wave propagation into the pulmonary vascular tree. While the precise origin and composition of these changes remain partially understood, factors such as mechanical deformation and organ movement are likely contributors.

To validate this approach, clinical measurements were conducted at Semmelweis Hospital in Hungary. Data collected during patient trials demonstrate the method's effectiveness in separating ventilation and perfusion signals, advancing EIT's potential for functional imaging and clinical application.

Project PerSAIDs

Revealing Unique Autoantibody Signatures in Systemic Autoinflammatory Diseases via Protein Array Profiling

Gabriel A. Vignolle

Gabriel A. Vignolle[1], Jasmin Huber[1], Yasmin Gillitschka[1], Sergio Decherchi[2], Yulia Rodina[2], Paolo Uva[3], Simona Gattulli[3], Isabella Ceccherini[4], Camilla Speziani[5], Dirk Foell[6], Marco Gattorno[5], Juan Arostegui I.[7], Sabrina Fühner[6], Seza Ozen[8], Christa Nöhammer[1], Klemens Vierlinger[1]

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6 Pädiatrische Rheumatologie und Immunologie, Universitätsklinikum Münster, Münster, Germany
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8 Department of Pediatric Rheumatology, Hacettepe University, Ankara, Türkiye

Abstract:

Systemic Autoinflammatory Diseases (SAIDs) are chronic, debilitating disorders driven by immune dysregulation involving various cell types, cytokines, and autoantibodies. Identifying autoantibody patterns linked to SAIDs is essential for improving diagnostics, understanding disease mechanisms, and developing personalized therapies. Our study aimed to define distinct autoantibody profiles, evaluate immune response heterogeneity, and analyze correlations between autoantibody reactivity and clinical parameters. We hypothesized that specific autoantibody signatures correlate with SAID phenotypes and disease progression, providing insights into pathogenesis and informing therapies. Using AIT's 16k protein microarray, capable of profiling antibodies against 7,390 human proteins, serum samples from 114 individuals were analyzed. Autoantibody signatures were identified using protein microarrays, with statistical thresholds of adjusted p-value < 0.05 and log2 fold-change >1 or <-1. Machine learning models, including Elastic-Net and Naïve Bayes, demonstrated performance criteria of AUC ≥ 0.7, and sensitivity/specificity ≥ 0.6. UMAP dimensionality reduction revealed distinct autoantibody profiles across disease groups. Antibodies targeting USP5, a deubiquitinating enzyme, emerged as a potential marker of immune dysregulation.

This high-throughput immunomics approach identified unique autoantibody profiles for various SAIDs, highlighting immune response heterogeneity. These findings improve diagnostic precision, identify therapeutic targets, and support personalized treatment strategies, advancing SAID management and patient outcomes.

Project REDESIGN

Patient-derived organoids as tool to decipher tumour heterogeneity in gastric cancers

Vivian Mittné

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2 Center for Genome Engineering, Institute for Basic Science (IBS), Daejeon, Republic of Korea
3 National Center for Tumor Diseases (NCT/UCC), Dresden, Germany; German Cancer Research Center (DKFZ), Heidelberg, Germany; Faculty of Medicine and University Hospital Carl Gustav Carus, Technische Universität Dresden, Dresden, Germany; Helmholtz-Zentrum Dresden-Rossendorf (HZDR), Dresden, Germany

Abstract:

Over the last decade, patient derived organoids (PDOs) have become an invaluable tool for disease modelling and a better understanding of the properties of individual cancers. Specifically, treatment response may depend on the cell trajectories that a given tumour will establish over time. In order to understand this heterogeneity in gastric cancer, multiple samples of the primary tumour were collected at the time of diagnosis and used to establish PDOs. To evaluate treatment response of individual patients, at least three PDOs from the same patient were assessed for their response to the standard of care chemotherapy regimen FLOT (5-Fluoroacil, Docetaxel, Oxaliplatin, Leucovorin). Interestingly, the response to FLOT did not significantly differ between PDOs derived from different locations from the same patient. However, when exposed to alternative therapies such as kinase inhibitors and small molecules, differential responses were observed among these PDOs. Sequencing data from PDOs suggest that distinct tumour locations harbour unique mutations, potentially explaining the variation in response. In conclusion, we show that by using different PDOs from one patient enhances our understanding of tumour heterogeneity and may facilitate the identification of alternative treatment options that target specific molecular pathways.

Project TECH-TOYS

From CareToy to TECH-TOYS: a co-design approach for the early detection of Neurodevelopmental Disorders

Silvia Filogna

Silvia Filogna[1], Elena Beani[1,2], Mattia Franchi De' Cavalieri[1,3], Giada Martini[1], Fabrizia Festante[1,2], Olena Chorna[1,2,4], Erhan Biçer[5], Giulio Bertamini[6], Nursena Boluk[5], Louis Simon[7], Şeyma Takir[5], Elif Toprak[8], Pinar Uluer[9], Mohamed Chetouani[7], David Cohen[6], Hatice Kose[5], Duygun Erol Barkana[9], Giuseppina Sgandurra[1,2], Giovanni Cioni[1]

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4 NEUROFARBA, University of Florence, Florence, Italy;
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6 Department of Child and Adolescent Psychiatry, Pitiè-Salpêtrière Hospital, Sorbonne, France;
7 Institut de Systemes Intelligents et de Robotique, Sorbonne Université, Paris, France;
8 Yeditepe University Galatasaray University Istanbul, Türkiye;
9 Galatasaray University Istanbul, Türkiye

Abstract:

Early detection of Neurodevelopmental Disorders (NDDs) is crucial for timely interventions on infants and improved clinical outcomes; however, capturing quantitative atypical development markers remains challenging. Building on insights from the previous biomechatronic gym, the CareToy, TECH-TOYS was developed as a high-tech ecological play environment for infant's assessment. The TECH-TOYS Environment is the result of an interdisciplinary co-design process involving clinicians, engineers, families, ethicists, and industry partners. This collaboration produced an optimal solution that aligns with clinical requirements, technological constraints, and stakeholder needs. Specifically, mat and wearable sensors were selected to avoid interfering with natural infants' movements while capturing data on motor behavior and emotional states during interactive play with caregivers. Sensorised toys were developed to maintain infants' affordance and encourage meaningful interactions. High-resolution cameras were chosen to capture accurate and occlusion-free images, ensuring optimal data quality for automatic analyses. Analysis of data collected from the CareToy system also demonstrated how a multimodal Machine Learning pipeline could support the identification of digital biomarkers indicative of atypical neurodevelopment.
The integration of past insights and collaborative expertise establishes a novel co-design approach for early NDD assessment, enabling digital biomarker identification and supporting personalised, validated predictive diagnoses.

Additional Speakers and Experts

RRI Workshop

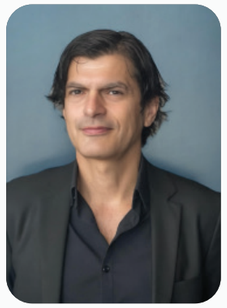
Ulf Tölch

Ulf Tölch obtained his PhD in biology at Ludwig Maximilans University in Munich. His research includes cognitive neuroscience, quantitative methods and statistical modelling of decision making. After his habilitation in psychology, he joined the BIH QUEST Center for responsible Research. There he currently serves as a research group leader where his research team explores robust methodological approaches in preclinical settings to inform and improve research decisions. Beyond this he is also responsible for educational formats and training at the QUEST.

Natascha Drude

Natascha Drude, PhD, specializes in drug development and laboratory animal science, holding her doctorate from RWTH Aachen University. She completed a second master's in 2018 in laboratory animal science focused on microsurgery and molecular imaging. At the Department of Experimental Molecular Imaging (ExMI), she concentrated her research on nanomedicine and severity assessment. A certified FELASA LAS specialist, she joined the BIH QUEST Center for Responsible Research in 2020 and now leads Responsible PrecliniX, enhancing preclinical-to-clinical translation. Natascha also advises Medizinisches Kompetenzzentrum c/o HCx Consulting GmbH (Medizin im Grünen), supporting the integration of medical devices into clinical practice.

Patient Engagement Panel



Juan-Jose Ventura

Juan-Jose Ventura of Cancer Patients Europe studied Pharmacy and obtained his PhD at the University Complutense of Madrid. He has conducted basic research in international institutions such as EMBL, HHMI, CNIO, University of Cambridge and KU Leuven. He has aimed to find target cells and molecular biomarkers that could be used for diagnosis and therapy of several cancers (Liver, Colon, Lung). After switching his career path, he moved to work as patient advocate. He has participated in more than 20 EU research projects and helped to bring the voice of patients to the latest research trends.

Lidewij Eva Vat

Lidewij Eva Vat is a leading expert in transforming health research and care systems through patient and public engagement. She holds a PhD focused on embedding patient engagement in decision-making processes within health research systems. Currently, she is Program Director at The Synergist, a non-profit organization that drives multi-stakeholder collaborations to tackle complex societal challenges and create sustainable solutions in health and other fields.

Dr. Vat has over a decade of experience in stakeholder engagement, monitoring and evaluation, and facilitating international, multi-stakeholder projects. She contributed to the PARADIGM initiative by developing tools to assess the value and impact of patient engagement and served as Training and Capacity Lead for NL SUPPORT, where she led the development of patient-centered research infrastructure and innovative training programs. Her work advances sustainable and equitable healthcare solutions through collaboration, innovation, and system improvement. Programs she is currently involved in include: From Testing to Targeted Treatments, the Patient-Focused Medicines Initiative and the IHI project GUIDE.MRD.

Resources on Patient Engagement:

- PFMD Tools for Patient Engagement: PFMD works to support systematic, efficient, and meaningful PE in research and beyond. The tools co-created by the community may be helpful for research projects within EP PerMed. You can explore them here.
- Recommendations for Research Funders: PFMD aims to share recommendations, examples, and mechanisms to support funders in enabling meaningful PE in research. You can view the latest insights from the Patient Engagement Open Forum session on this topic here. A publication summarizing these recommendations and good practices is also in progress.

Poster Prize Awardee

Lena Lorenzer

Lena Lorenzer is a Junior Researcher at the Medical University of Graz, Austria, working on the international PRE-CARE ML project since June 2024. The project focuses on predicting cardiovascular events using machine learning techniques. As part of this project, she plans to pursue a PhD later this year to further explore the integration of machine learning models in clinical decision-making. Lena recently completed her Master's degree in Biomedical Engineering at Graz University of Technology in October 2024, specializing in Computational Neuroscience and Bioinformatics. During her studies, she gained experience in the processing and analysis of biological data, particularly in addressing neuroscientific questions using computational methods.

JTC2021 FUNDERS

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EP PerMed has received funding from the European Union's Horizon Europe research and innovation programme under grant agreement No 101137129