New EU Research Project “Screen4Care”: Accelerating Diagnosis for Rare Disease Patients Through Genetic Newborn Screening and Artificial Intelligence

The international consortium aims to tackle the major hurdle for rare disease patients – the lengthy and convoluted diagnosis journey – via an innovative research approach based on two central pillars: genetic newborn screening and artificial intelligence (AI)-based tools.

Ferrara, Italy, 1 October 2021 – Today an international public-private consortium of 35 partners announced the launch of Screen4Care – a research project that aims to significantly shorten the time required for rare disease diagnosis and efficient intervention by utilising genetic newborn screening and advanced analysis methods such as machine learning. The project will run for a period of five years with a total budget of EUR 25 million provided by the Innovative Medicines Initiative (IMI 2 JU), a joint undertaking of the European Union and the European Federation of Pharmaceutical Industries and Associations (EFPIA).

There are more than 7,000 known rare diseases, conditions that affect one, or less than one, person in 2,000. These conditions, which collectively impact an estimated 27-36 million people across the EU and will affect one in 17 people during their lifetime, are often severe, multisystemic chronic diseases that put patients at risk of permanent organ damage and degeneration. Patients typically face an arduous journey to proper diagnosis, enduring on average eight years of countless doctor’s consultations, misdiagnoses and ineffective treatments. Lengthy diagnosis journeys place a heavy burden on patients, their families and society. They also hinder swift intervention – such as appropriate treatments or enrolment in clinical trials – and patient empowerment, realised through strategies such as lifestyle adjustments, family planning, genetic counselling and coping with the psychosocial and/or financial consequences of the condition.

Screen4Care will use a multi-pronged strategy to shorten the time to diagnosis and treatment for patients with rare diseases:

1) Genetic newborn screening: The project will drive newborn screening (using genetic testing and related advanced genomic technologies), which is anticipated to be an effective tool for early diagnosis given that approximately 72% of rare diseases have a genetic cause and 70% of rare disease patients are children.

2) AI-based tools: The project will design and develop new AI algorithms to identify patients at early disease onset via electronic health records and develop a repository of AI ‘symptom checkers’ to help patients who are in the midst of their diagnosis journeys – both supporting symptom-based diagnosis later in life.

This project has received funding from the Innovative Medicines Initiative 2 Joint Undertaking (JU) under grant agreement No. 101034427. The JU receives support from the European Union’s Horizon 2020 research and innovation programme and EFPIA. www.imi.europa.eu
“Screen4Care has the potential to help patients benefit from precision diagnostics that lead to better care, meaningful hospital visits and a better overall quality of life – and to help strengthen healthcare systems through novel diagnostic tools and a resource-efficient technological infrastructure”, said Alessandra Ferlini, Associate Professor of the Department of Medical Sciences, and Head of the Medical Genetics Unit at the University of Ferrara, Italy, and Scientific Coordinator of the project. “Of particular importance to the project is the active involvement of stakeholders to help shape the design and decisions for value-based healthcare.”

“Because of the nature of their conditions, rare disease patients and their families commonly experience delays to diagnosis, which can lead to serious consequences for their health as well as their ability to plan for their future”, said Nicolas Garnier, Doctor in Experimental Medicine, Director of Patient Advocacy, Rare Disease Global Product Development at Pfizer, and Project Lead on behalf of EFPIA. “We therefore challenged ourselves to address this most pressing issue: accelerating patients’ path to diagnosis.”

Building a digital infrastructure to foster exchange among physicians, patients, relatives and caretakers

In addition to its goal of developing the core early-diagnosis system, Screen4Care aims to establish a digital infrastructure and ecosystem to engage patients, parents of newborns and caregivers as equal decision-makers in the diagnosis process. The ecosystem will provide an open innovation platform, which allows for continuous data collection and information exchange, aiding the development of next-generation diagnostics and enabling physicians, patients and relatives to make informed decisions at an earlier stage. Screen4Care proposes that this will contribute to minimised disease progression, improved patient health and quality of life and an optimised use of healthcare resources.

The Screen4Care team comprises 21 academic partners led by the University of Ferrara, nine industrial project partners led by Pfizer, and four small and medium-sized enterprises. It brings together experts in genetics, bioinformatics, data management and standards, imaging for phenotyping, ethics and health preference research, decision-analytic modelling, and cybersecurity. Furthermore, the consortium is complemented by EURORDIS, representing rare disease patients in Europe to promote robust dialogue and ensure the needs and preferences of the rare disease community guide progression of the project. “Screen4Care is dedicated to giving everybody representation at the table”, explained Dr Garnier. “The project’s focus on equity sets it apart, and it is a unique opportunity to effect meaningful, longstanding change for the diverse rare disease community.”

***

This project has received funding from the Innovative Medicines Initiative 2 Joint Undertaking (JU) under grant agreement No. 101034427. The JU receives support from the European Union’s Horizon 2020 research and innovation programme and EFPIA. www.imi.europa.eu
Press Release

**Project Key Facts**

*Full Name:* Screen4Care – Shortening the path to rare disease diagnosis by using newborn genetic screening and digital technologies  
*Start Date:* 1 October 2021  
*Duration:* 5 years  
*Budget:* 25 Mio €  
*Coordinator:* University of Ferrara, Italy  
*Project Lead:* Pfizer Ltd, UK  
*Website:* [www.screen4care.eu](http://www.screen4care.eu)  
*Social Media:* [LinkedIn](http://www.linkedin.com) | [Twitter](http://twitter.com)

**Project Partners**

**Austria**  
Ludwig Boltzmann Gesellschaft GmbH  
Research Institute AG & Co KG  
SBA Research gemeinnützige GmbH

**Bulgaria**  
Bulgarian Association for Personalized Medicine  
Bulgarian Association for the Promotion of Education and Science

**Czech Republic**  
University Karlova- 2nd Faculty of Medicine and Motol University Hospital

**Denmark**  
Copenhagen Business School  
FindZebra ApS  
Novo Nordisk A/S  
Syddansk University

**France**  
Eurodis-Rare Diseases Europe  
Lysogene S.A.  
Sanofi-Genzyme

**Germany**  
Charité - Universitätsmedizin Berlin  
Eurice - European Research and Project Office GmbH  
Max-Planck-Gesellschaft zur Förderung der Wissenschaften e.V.  
Universitätsmedizin Göttingen – Georg-August-Universität Göttingen  
University Hospital Bonn  
University Hospital Erlangen

**Ireland**  
University College Dublin, National University of Ireland (Inclusive Design Research Centre-SMARTlab)

This project has received funding from the Innovative Medicines Initiative 2 Joint Undertaking (JU) under grant agreement No. 101034427. The JU receives support from the European Union’s Horizon 2020 research and innovation programme and EFPIA. [www.imi.europa.eu](http://www.imi.europa.eu)
Press Release

Israel
Genoox

Italy
Consorzio Futuro in Ricerca
Pediatric Hospital Bambino Gesù
University of Ferrara
University of Siena

Spain
Centro Nacional de Análisis Genómico (CNAG-CRG)

Sweden
Uppsala University

Switzerland
F. Hoffmann-La Roche AG
sitem-insel AG
Novartis Pharma AG
Takeda Pharmaceuticals International AG
University Bern

The Netherlands
ProQR Therapeutics NV

United Kingdom
Illumina Cambridge Ltd
Pfizer Ltd

Contact

Project Coordinator
University of Ferrara
Prof. Alessandra Ferlini
Phone: +39 532974439; mobile +39 3405493446
Mail: fla@unife.it

Project Management
EURICE GmbH
Dr. Cora Meyer
Phone: +49 30 374415-834
Mail: c.meyer@eurice.eu

This project has received funding from the Innovative Medicines Initiative 2 Joint Undertaking (JU) under grant agreement No. 101034427. The JU receives support from the European Union’s Horizon 2020 research and innovation programme and EFPIA. www.imi.europa.eu