Newsletter Issue #2

Welcome…

to the 2nd issue of our **Screen4Care** newsletter! This newsletter series will take you on our exciting journey to accelerate rare disease diagnosis through genetic newborn screening and digital technologies. We will update you on our latest progress and numerous links for more information about our project. **Have fun reading!**

Consortium Meeting

2nd Screen4Care Consortium Meeting

On April 25th and 26th, 2024, the Screen4Care consortium held their 2nd Consortium Meeting (hybrid), which our project partner, Novo Nordisk, hosted in Copenhagen, Denmark. This meeting allowed project partners and stakeholders to engage in fruitful discussions about the project's progress and preparation for

upcoming activities. Additionally, the partners enjoyed a guided tour of the former Viking Fishing Village known today as Copenhagen and basked in a scenic joint dinner at the Copenhagen Opera House.

Summary of the 2nd Consortium meeting

New Partners Joining the Consortium

Incoming Partners to the Consortium

The Screen4Care consortium would like to take this opportunity to welcome our new partners to the consortium. As a result of the recent amendment to the project plan, the Screen4Care consortium welcomed three new Centre Hospital-Universitaire Dijon Bourgogne, UCB Biopharma SRL and Wallac Oy (Revvity).

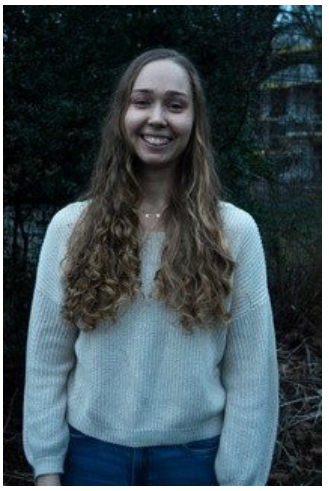
These new partners will add value to the project by contributing their experience, expertise, and resources to support tasks in various work packages.

Click here for more information about incoming partners

In the Spotlight

Rare Disease Interview

In celebration of the International Day of Women and Girls in Science 2024, the Screen4Care project interviewed Ina Baumann, a medical student living with a rare disease. Ina discussed various topics, such as her passion for medicine, gender-based biases, the importance of healthy habits and the challenges of pursuing a STEM career while living with a rare disease.

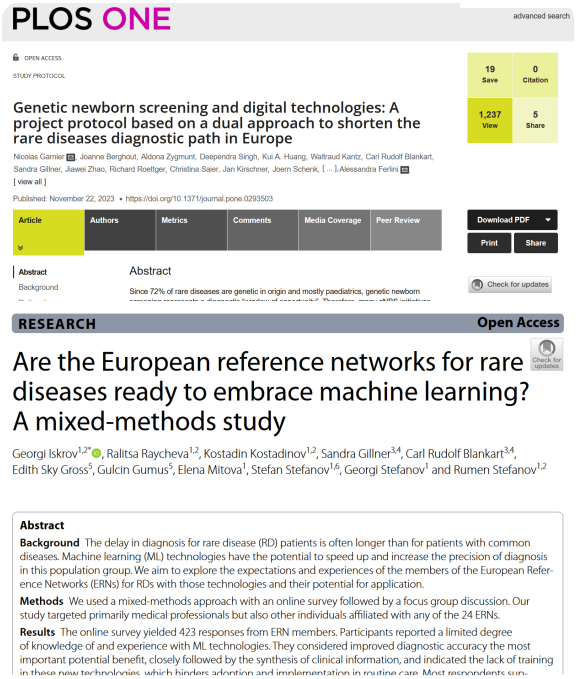


Click here for more information about the Rare Disease Interview

Publications

Recent Publications

As the Screen4Care project develops, the consortium members have disseminated various research publications as they continue contributing to the rare disease research community.


PLOS ONE

advanced search

OPEN ACCESS

STUDY PROTOCOL

Genetic newborn screening and digital technologies: A project protocol based on a dual approach to shorten the rare diseases diagnostic path in Europe

Nicolas Garnier, Joanne Berghout, Aldona Zygmunt, Deependra Singh, Kui A. Huang, Waltraud Kantz, Carl Rudolf Blankart, Sandra Gillner, Jiawei Zhao, Richard Roettger, Christina Saier, Jan Kirschner, Joern Schenk, [...] Alessandra Ferlini

[view all]

Published: November 22, 2023 • https://doi.org/10.1371/journal.pone.0293503

19 Save | 0 Citation | 1,237 View | 5 Share

Article | Authors | Metrics | Comments | Media Coverage | Peer Review

Download PDF | Print | Share

Abstract

Background

Since 72% of rare diseases are genetic in origin and mostly paediatrics, genetic newborn

Check for updates

RESEARCH Open Access

Are the European reference networks for rare diseases ready to embrace machine learning? A mixed-methods study

Georgi Iskrov[1,2*], Ralitsa Raycheva[1,2], Kostadin Kostadinov[1,2], Sandra Gillner[3,4], Carl Rudolf Blankart[3,4], Edith Sky Gross[5], Gulcin Gumus[5], Elena Mitova[1], Stefan Stefanov[1,6], Georgi Stefanov[1] and Rumen Stefanov[1,2]

Abstract

Background The delay in diagnosis for rare disease (RD) patients is often longer than for patients with common diseases. Machine learning (ML) technologies have the potential to speed up and increase the precision of diagnosis in this population group. We aim to explore the expectations and experiences of the members of the European Reference Networks (ERNs) for RDs with those technologies and their potential for application.

Methods We used a mixed-methods approach with an online survey followed by a focus group discussion. Our study targeted primarily medical professionals but also other individuals affiliated with any of the 24 ERNs.

Results The online survey yielded 423 responses from ERN members. Participants reported a limited degree of knowledge of and experience with ML technologies. They considered improved diagnostic accuracy the most important potential benefit, closely followed by the synthesis of clinical information, and indicated the lack of training in these new technologies, which hinders adoption and implementation in routine care. Most respondents sup-


Social Science & Medicine 340 (2024) 116442

Contents lists available at ScienceDirect

Social Science & Medicine

journal homepage: www.elsevier.com/locate/socscimed

ELSEVIER

We're implementing AI now, so why not ask us what to do? – How AI providers perceive and navigate the spread of diagnostic AI in complex healthcare systems

Sandra Gillner[a,b,*]

[a] KPM Center for Public Management, University of Bern, Freiburgstr. 3, 3010, Bern, Switzerland

[b] Swiss Institute for Translational and Entrepreneurial Medicine (sitem-insel), Freiburgstr. 3, 3010, Bern, Switzerland

ARTICLE INFO

Handling Editor: Medical Sociology Office

ABSTRACT

Despite high expectations of artificial intelligence (AI) in medical diagnostics, predictions of its extensive and rapid adoption have so far not been matched by reality. AI providers seeking to promote and perpetuate the use of this technology are faced with the complex reality of embedding AI-enabled diagnostics across variable implementation contexts. In this study, we draw upon a complexity science approach and qualitative methodology to understand how AI providers perceive and navigate the spread of AI in complex healthcare systems. Using semi-structured, one-to-one interviews, we collected qualitative data from 14 providers of AI-enabled

frontiers | Frontiers in Public Health

TYPE Systematic Review

PUBLISHED 15 September 2023

DOI 10.3389/fpubh.2023.1214766

Check for updates

OPEN ACCESS

EDITED BY
Chiuhui Mary Wang,
Rare Diseases International, France

REVIEWED BY
Sunyang Fu,
Mayo Clinic, United States
Tim Hulsen,
Philips Research, Netherlands

*CORRESPONDENCE
Ralitsa Raycheva
raycheva@raredis.org

RECEIVED 05 May 2023
ACCEPTED 30 August 2023
PUBLISHED 15 September 2023

CITATION
Raycheva R, Kostadinov K, Mitova E, Bogoeva N, Iskrov G, Stefanov G and Stefanov R (2023) Challenges in mapping

Challenges in mapping European rare disease databases, relevant for ML-based screening technologies in terms of organizational, FAIR and legal principles: scoping review

Ralitsa Raycheva[1,2*], Kostadin Kostadinov[1,2], Elena Mitova[2], Nataliya Bogoeva[2], Georgi Iskrov[1,2], Georgi Stefanov[2] and Rumen Stefanov[1,2]

[1]Department of Social Medicine and Public Health, Faculty of Public Health, Medical University of Plovdiv, Plovdiv, Bulgaria, [2]Bulgarian Association for Promotion of Education and Science, Institute for Rare Disease, Plovdiv, Bulgaria

Click here to find the Screen4Care publications

Collaboration Initiatives

Screen4Care at ICONS 2023

The Screen4Care consortium actively engages with networks and initiatives within the rare disease community. On October 5th-6th, 2023, the Screen4Care consortium participated in the second Annual Conference of the International Consortium on Newborn Sequencing (ICoNS). The Scientific Project Coordinator, Prof. Alessandra Ferlini, and the former EFPIA lead, Dr. Nicolas

Garnier, presented the Screen4Care project to global experts from various newborn screening research projects.

Click here for more information on Screen4Care at ICONS Conference 2023

Screen4Care was an associate partner in 2024 at the 12th European Conference on Rare Diseases & Orphan Products (ECRD 2024)

The Screen4Care project was represented at the 12th European Conference on Rare Diseases & Orphan Products in Brussels, Belgium, on May 15th-16th, 2024. The conference, themed "Action Within Reach: Pioneering Solutions for Rare Diseases," provided a valuable platform for addressing the challenges faced by individuals with rare diseases. In the capacity of an associate partner, Prof. Alessandra Ferlini (virtually), the Scientific Project Coordinator, and Dr. Aldona Zygmunt, the EFPIA lead, represented the Screen4Care project to a diverse audience, including patient advocates, academics, healthcare professionals, industry stakeholders, payers, regulators, Member State representatives, and policymakers.

Click here for more information about the 12th European Conference on Rare Diseases &

Events and Conferences

Rare Disease Awareness Day Lunch

On February 29th, 2024, project partner Sitem-Insel (SI) hosted a Rare Disease Awareness Day Lunch at the Swiss Institute for Translational and Entrepreneurial Medicine! This year's event focused on uniting patients' knowledge, healthcare professionals' expertise, and researchers' scientific excellence. The activities led by SI researchers on genetic newborn screening and digital technologies were groundbreaking. They discussed the immense value these advancements bring to the rare disease community as part of the Screen4Care project. Additionally, the event showcased how CSL Behring's research and development is making a real difference in improving patients' lives on the Insel Campus Bern. It was an inspiring and impactful day!

The 10th International Symposium on Health Technology Assessment: A Decade of Advancements in Newborn Screening

On April 5, 2024, the Institute for Rare Diseases hosted its 10th International Symposium on Health Technology Assessment in Sofia, Bulgaria. This year’s symposium theme was "Health Technology Assessment and Newborn Screening." The symposium featured renowned speakers and highlighted the progress made through initiatives like Screen4Care. Discussions focused on the impact of such initiatives on national healthcare strategies and the need for comprehensive screening processes. The commitment to advancing newborn screening and addressing challenges faced by those living with rare diseases remains steadfast. The symposium is part of a series of initiatives to improve healthcare and enhance the quality of life for individuals with rare diseases.

European Human Genetics Conference 2024 (ESHG 2024)

From June 1-4, 2024, Prof. Alessandra Ferlini, the Scientific Project Coordinator of Screen4Care, and Sergi Beltran, a representative of the project partner Centro Nacional de Análisis Genómico (CNAG), attended the 57th European Human Genetics Conference in Berlin, Germany. The ESHG conference serves as a platform for showcasing the latest scientific and technological advancements in human genetics, aiming to educate and inspire the next generation of human geneticists. The Screen4Care project poster was presented at the event.

Rare Disease Day 2024

Members of Screen4Care participated in the #LightUpForRare campaign to celebrate Rare Disease 2024!

Connect with us:

Eurice - European Research and Project Office GmbH

Heinrich-Hertz-Allee 1, 66386, St.Ingbert