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Screening newborns to help fight rare diseases

Early genetic checks being developed by researchers with EU and industry funding will accelerate treatments for illnesses that affect millions of people in Europe.

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Dr Alessandra Ferlini wants medical help for millions of people in Europe to begin before they even realise it.

Ferlini, associate professor in medical genetics at the University of Ferrara in Italy, aims for every child born in Europe to be genetically screened at birth for a range of rare diseases. Such conditions each affect no more than five in 10 000 people.

Not so rare

While they are considered rare, these diseases as a whole number more than 7 000, affect as many as 36 million people in the EU and will touch one in 17 Europeans during their lifetimes. Around 80% of rare diseases have a genetic origin and, of those, 70% start in childhood.

‘Genetic screening offers the significant benefit of providing more accurate data for more rare disease conditions,’ said Ferlini.

She leads a research project that receives EU funding and industry support to fast-track the diagnosis and treatment of rare diseases in Europe through genetic checks on newborns and data analysis aided by artificial intelligence.

Called SCREEN4CARE, the project runs for five years through 2026.

It will begin a trial in Italy in the coming weeks and then expand the experiment to other EU countries, with doctors genetically screening about 25 000 newborns for 245 treatable rare diseases.

Rare diseases as a whole range from cardiovascular and gastrointestinal to skin and bone illnesses. Particular ones include cystic fibrosis, haemophilia and sickle cell anaemia.

Sooner, better

Genetic newborn screening, or gNBS, means children with such diseases would be diagnosed earlier. This in turn would result in more effective treatments.

As a secondary benefit, gNBS would produce valuable data for researchers studying these illnesses.

‘It would enable families to understand what is happening with their child, find the necessary support and care, and receive financial assistance,’ said Gulcin Gumus, senior manager of research and policy at EURORDIS, an alliance of more than 1 000 organisations that represent patients with rare diseases.

EURORDIS is part of SCREEN4CARE, which has more than 30 participants, including universities, research institutes and companies from 14 countries across Europe. Participant diversity is a required feature of EU-funded research to help ensure that it translates into concrete benefits for people.

SCREEN4CARE receives industry support under the Innovative Medicines Initiative, an EU public-private partnership.

Diagnosing rare diseases in Europe currently takes an average of almost five years, according to a May 2024 EURORDIS survey. Screening would reduce this timescale significantly, meaning more timely treatments and more efficient use of healthcare resources.

The genetic screening method involves taking five drops of blood two days after birth and performing next-generation sequencing.

This is faster than current national programmes, which screen for a smaller number of rare diseases using blood tests that look for metabolic or proteomic markers of disease.

Fairer together

The organisation of health care being a national responsibility, existing screening programmes also vary widely from country to country. For example, Italy screens for over 40 conditions, while Romania does so for just two.

The SCREEN4CARE team aims to align practices across the EU, ensuring better and fairer healthcare in Europe.

‘Adopting a uniform approach to newborn screening across EU Member States is an important way to attempt to ensure equitable and timely access to rare disease diagnosis for European citizens,’ said Aldona Zygmunt, director of policy and public affairs at pharmaceutical manufacturer Pfizer, the industry partner of SCREEN4CARE.

Participation in the project’s pilot programme will be voluntary; only parents who want their children to be screened will opt in.

SCREEN4CARE is still tackling questions, including where the data will be stored, who will have access to the information and who will own it until the children reach adulthood, according to Ferlini.

These involve data privacy, a tricky subject because health and genetic information is sensitive. The EU has generally been keen to ensure strong data privacy safeguards.

Database drive

Once privacy matters are addressed, widespread data collection in this health field presents a major opportunity. Rare diseases offer researchers relatively few data points to work with, particularly if these are fragmented among EU countries.

As a result, researchers are often unable to study such illnesses in depth and can miss opportunities to improve diagnosis or develop better treatments.

At first, samples under SCREEN4CARE will be stored in a project data repository and then possibly in secure biobanks. The researchers hope to establish a partnership with EU initiatives, including the future European Partnership on Rare Diseases and the recently agreed European Health Data Space.

The information gathered by screening newborns under SCREEN4CARE would feed into EU efforts to strengthen research on rare diseases.

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More info

- SCREEN4CARE
- EU action on rare diseases
- EU action on research for rare diseases
- EU health research and innovation
- Innovative Health Initiative