



# A NEW SCREENING APPROACH FOR RARE DISEASES: NEWBORN GENETIC SCREENING IN THE EUROPEAN SCREEN4CARE PROJECT



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. Neither the JU nor the EU is however participating as a contracting authority in this procurement. Any dissemination of results must indicate that it reflects only the author's view and that the JU is not responsible for any use that may be made of the information it contains.



## WHAT ARE RARE GENETIC DISEASES?

Genetic diseases are caused by the presence of **alterations** (“errors”) in **DNA**. The genetic heritage of each of us is written in our DNA sequence, which is contained in the cell nucleus. In the DNA sequence there are **genes** which are specific regions that function to produce the proteins of our body.

**Rare diseases** affect less than **1 in 2.000 people**. To date, more than **7.000 rare diseases** are known. Most rare diseases have a **pediatric onset** and have a **genetic origin**. These are often serious conditions, especially if not diagnosed and treated.

## WHAT IS NEWBORN SCREENING?

Newborn screening is used to identify many pathologies within the first 48-72 hours of life by taking few drops of blood (dried bloodspot, **DBS**) from the heel of the newborn.

There are **two types** of newborn screenings. **Newborn metabolic screening** at birth is currently carried out in Europe. **Newborn laboratory screening** is currently available in the Czech Republic: it checks for 20 diseases (genetic, endocrine, metabolic, ...). **Newborn genetic screening (gNBS)** offers an **added value** to metabolic screening by **directly analyzing DNA**. In fact, a genetic disorder is not always associated with a specific metabolite. Genetic screening is possible thanks to the use of new generation techniques (NGS) that allow the analysis of gene panels (multiple genes at the same time).

## WHAT IS THE SCREEN4CARE PROJECT?

The **SCREEN4CARE Project (S4C)** is a five-year European research project with a total budget of 25 million euro and funded by the Innovative Medicines Initiative (IMI 2 JU). **Screen4Care aims to meet the urgent need to accelerate the diagnosis time for rare disease.**

The project is coordinated by the **Medical Genetics Unit of the University-Hospital of Ferrara, directed by Professor Alessandra Ferlini**. The project will offer newborn genetic screening in Italy (Ferrara, Modena, Rome, Siena) and in some provinces in Germany, France, for **12 months**, starting from mid-2024, up to a total of 25 thousand newborns in Europe.

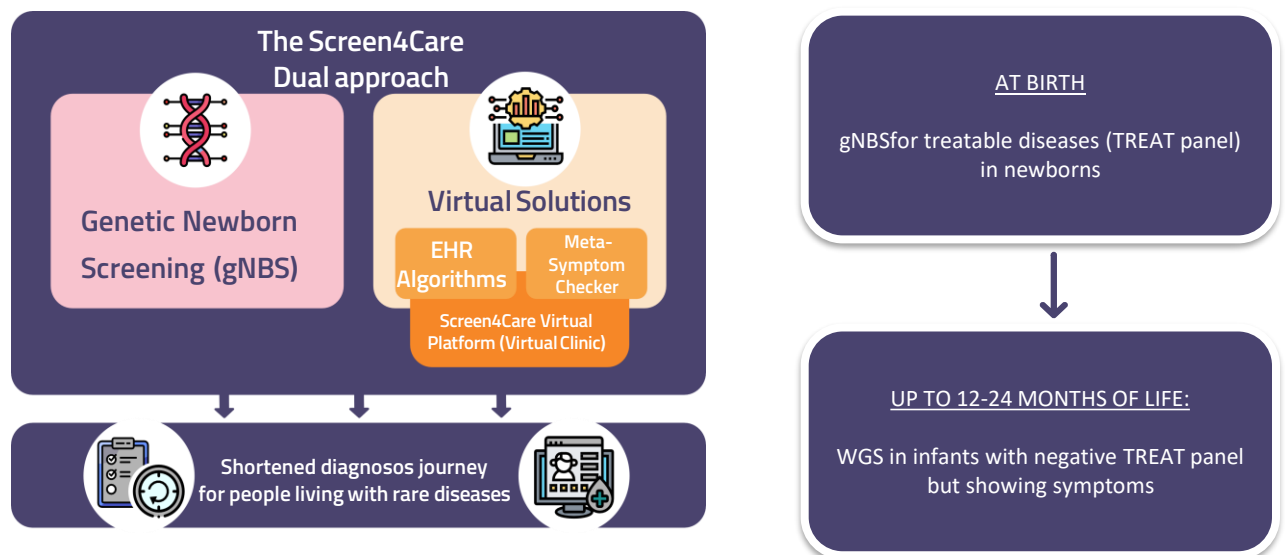
# SCREEN 4CARE



## WHAT DOES THE PROJECT INCLUDE?

If you join the project, a **newborn genetic screening (gNBS)** for **treatable diseases** will be offered to your child, in addition to the mandatory newborn metabolic screening. This screening will include some genetic diseases (**TREAT panel**) for which there is an **approved therapy**.

In addition, for infants tested negative on newborn genetic screening, but who will manifest **early symptoms** in the first two years of life, It will be offered the **Whole Genome Sequencing (WGS)**, the most extensive genetic analysis currently available.



## WHAT IS THE TREAT-PANEL?

It is a new generation sequencing (NGS) technique that allows us to "read" many genes associated with multiple diseases in a single analysis. A molecular analysis of 245 genes **associated with rare diseases** that are treatable with an available and approved therapy for patients (pharmacological/genetic/dietary) will be performed. The goal is to start therapy as soon as possible to treat the disease.

## WHAT IS THE WHOLE GENOME SEQUENCING?

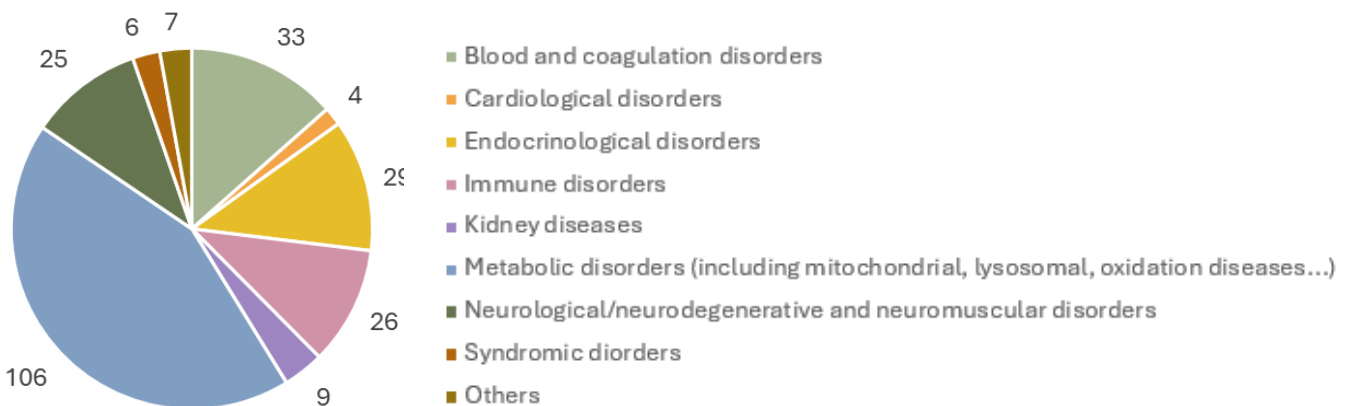
Whole Genome Sequencing (or WGS) is the most extensive genetic analysis currently available. This analysis makes it possible to "read" ALL the "letters" (nitrogen bases) that compose the DNA. **S4C will offer WGS to newborns with negative results from the newborn genetic screening but show early symptoms in the first one to two years of life.**

# SCREEN 4CARE



## WHICH DISEASES ARE INCLUDED IN THE SCREENING?

The screening tests for diseases of various types: metabolic, neuromuscular, endocrinological, immunological and many others! If you ask, you will be provided with the whole list.



## WHAT ARE THE ADVANTAGES AND RISKS?

The benefits of the TREAT-panel are the possibility of screening for **early diagnosis**, which allows access to reference centres (European Reference Networks, ERNs) for **available therapies and treatments**, as well as the possibility of making informed decisions for **future pregnancies**.

WGS allows for the **timely** diagnosis of genetic disease in the event of symptoms and, in the case of the TREAT-panel, access to reference centers and the ability to consciously choose for future pregnancies.

With regard to the risks, as for any blood collection, there are minimal risks associated with **taking blood** from the heel (already required by law for metabolic screening) for the TREAT-panel, or intravenously for WGS (same risks as for all blood draws).

**Incidental** (unexpected), or **secondary** (not related to the specific disease) **results** may emerge for TREAT-panel and to a greater extent for WGS. These findings will be reported to you according to the guidelines of the scientific societies (SIGU, ESHG, ACMG).

# SCREEN 4CARE



## DON'T FORGET THAT...

For all those born in the time span established by the project, it is possible to participate in newborn genetic screening by:

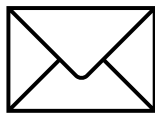
TREAT-panel

TREAT and WGS panel

- Participation is voluntary, **not mandatory**, and must be expressed through **informed written** consent for each of the options chosen.
- Participation is free of charge.
- Joining **the Screen4Care Project** DOES NOT REPLACE prenatal visits and newborn laboratory screening.



FOR FURTHER INFORMATION  
Contact us by email or get to know  
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on our social networks:



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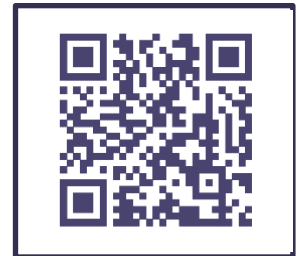
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