SCREEN4CARE EU IMI Project: WP3 Detecting Treatable Rare Diseases through Genetic Newborn Screening

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FACTS & FIGURES

Start date:	01/10/2021
End date:	30/09/2027
IMI funding:	11 938 569 €
EFPIA in kind:	14 308 605 €
Total Cost:	26 247 174 €
Project website:	screen4care.eu
Social media:	

CHALLENGE

Rare Diseases: Bridging the Gap Through Newborn Screening

Over **7,000** rare disease

affecting up to **36 million** people across EU

8 YEARS The Diagnostic Gap

70%+ Genetic Foundation

Loss of treatment opportunity due to late diagnosis

A Key solution:

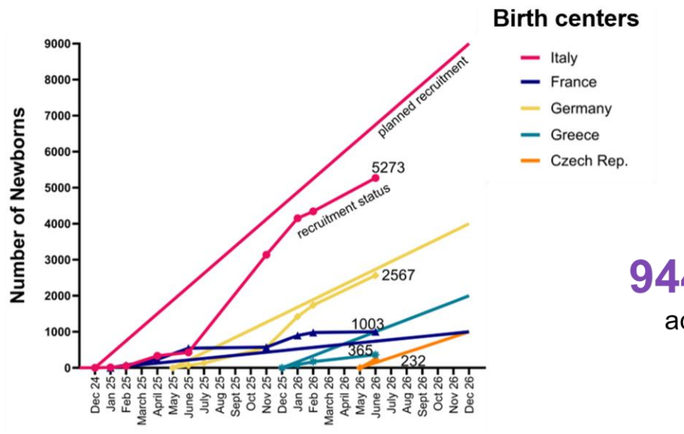
Genetic Newborn Screening

transforms the diagnostic process, moving away from waiting for symptoms to appear towards identifying treatable conditions from birth

Accelerating **Care and Treatment**

Early screening leads directly to a more timely diagnosis and immediate access to life-changing treatments

RESULTS

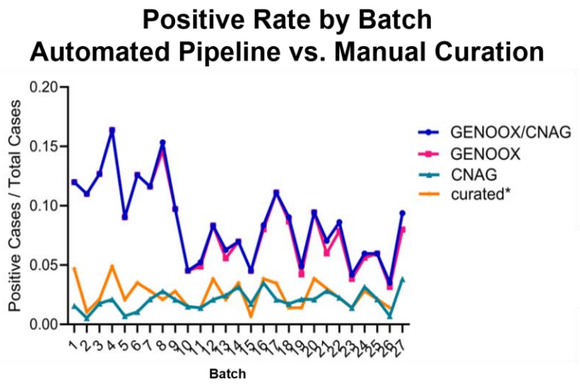


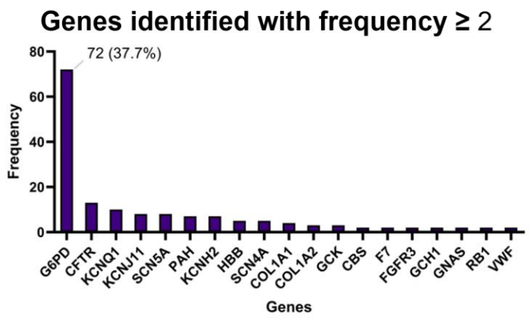
9440 babies recruited (Dec. 2024 – Jun. 2026) across participating birth centers

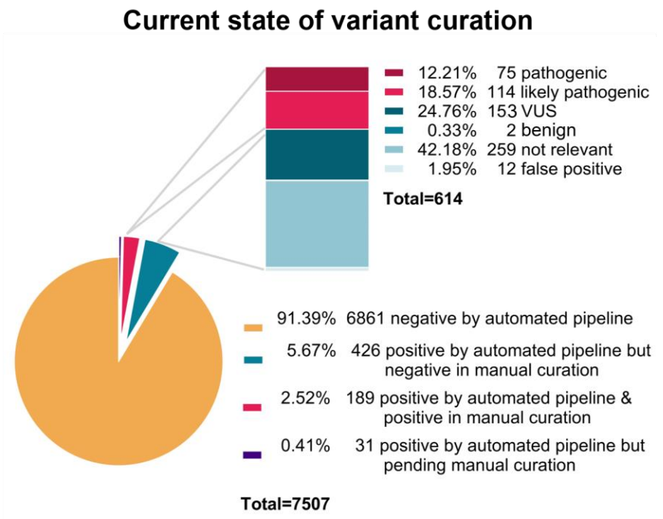
Initial results from the first babies enrolled in screening

As of May 2026 (Batch 1-27)

7,507	8,6%	2,5%	78*
Sequenced	**Presumptive positivity before manual curation**	**Validated true positive**	**Reported**
Panel sequencing successfully finalised	614 referred for expert curation at reference center	189 validated/total	Findings communicated to families
			* Others in progress

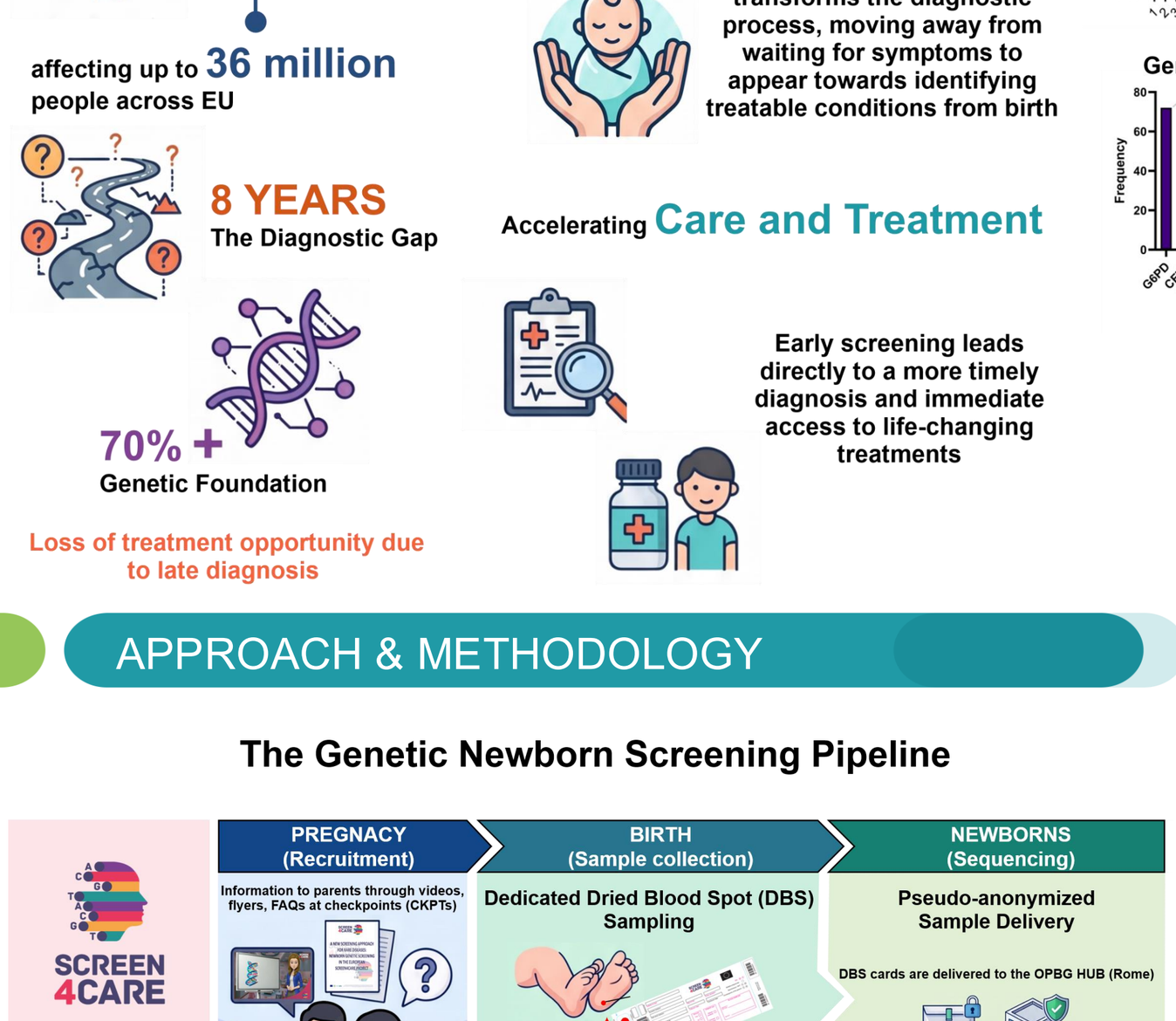






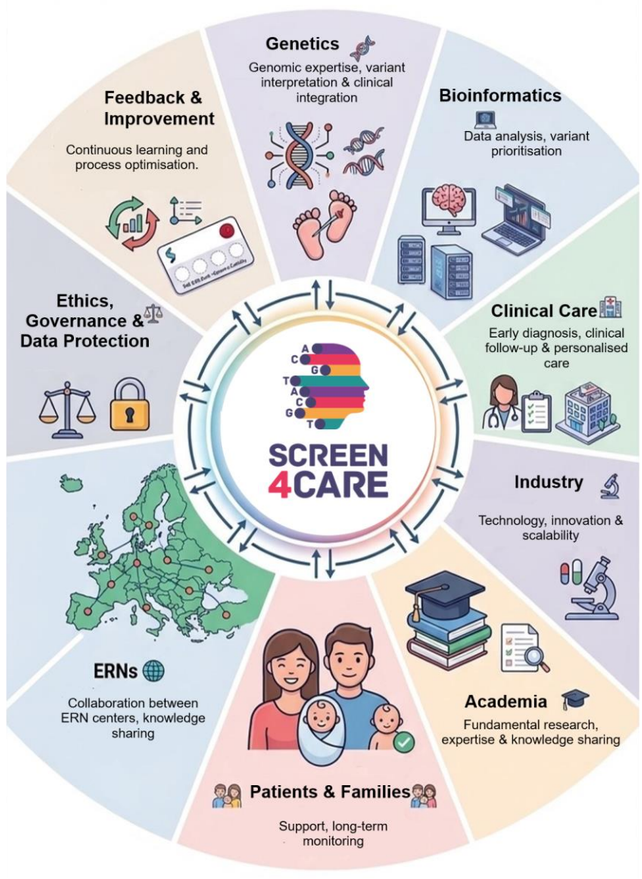
APPROACH & METHODOLOGY

The Genetic Newborn Screening Pipeline



VALUE OF IMI COLLABORATION

Connecting Research, Care & Innovation



CAREER IMPACT

SKILLS DEVELOPED

ADVANCED GENOMIC SKILLS

Variants identification; Pathogenicity assessment using multiple sources; ACMG classification

CLINICAL DATA INTEGRATION

Genotype-phenotype correlation; Clinical interpretation

MULTIDISCIPLINARY COLLABORATION

Knowledge exchange across disciplines; Clinical–bioinformatics collaboration; Harmonisation of approaches across Europe; Public-Private collaboration

CAREER IMPACT

- Expanded genomic expertise
- International professional network & exposure to public-private partnership models
- Experience in translational research

From genomic data to clinical impact: developing expertise at the intersection of research, healthcare and innovation

Screen4Care genetic newborn screening: the key to unlocking the gateway to genomic medicine

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