ERN | EuroBloodNet

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Equitable Access to Diagnostics and Early Treatment

A strategic contribution to **HLM4RARE Working Group 3**

Equitable access to diagnosis and care

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SESSION 1: EU ACTION PLAN FOR RARE DISEASES

PRIORITY ACTION 3: EQUITABLE ACCESS TO DIAGNOSTICS AND EARLY TREATMENT

EUROPEAN PRIORITY FOR RARE DISEASES

HLM4RARE WG 3: Accelerate equitable access to diagnostics and enable early treatment onset with innovative orphan drugs and digital innovation for unmet Medical needs

HLM 4RARE

CHALLENGES

- Access to timely and accurate diagnosis remains unequal across Member States and regions.
- Prenatal and newborn screening pathways remain fragmented, with limited harmonization of quality, timeliness and referral standards.
- Centers of expertise, ERNs, genomic technologies and data infrastructures are not yet fully integrated into diagnostic and care pathways.

ERN-EUROBLOODNET RESPONSE

Cross-border health

Telemedicine

KEY INITIATIVES

Screen4Care Europe, uRADAR, RWD, ERDERA

EU ACTION PLAN FOR RARE DISEASES

- WG1 Governance & Action Plan
- WG2 Workforce Development
- WG3 Diagnostics & Early treatment
- WG4 Clinical Trials
- WG5 National Infrastructure
- WG6 Real-World Evidence
- WG7 Equitable access to therapies
- WG8 Funding & Sustainability

Two intertwined gaps: getting diagnosed — and getting treated

For people with rare diseases, equity breaks down at two points along the pathway. Where a patient lives still decides whether — and how fast — each is overcome.

THE DIAGNOSTIC GAP

— **Specialised tests are not universal.** Confirmatory assays sit in a handful of reference labs; many centres must send samples away.

— **Time and expertise are unevenly shared.** Long turnaround and scarce expertise delay or miss the diagnosis.

— **Result:** diagnostic delay concentrated in less-resourced regions and non-expert centres.

THE TREATMENT GAP

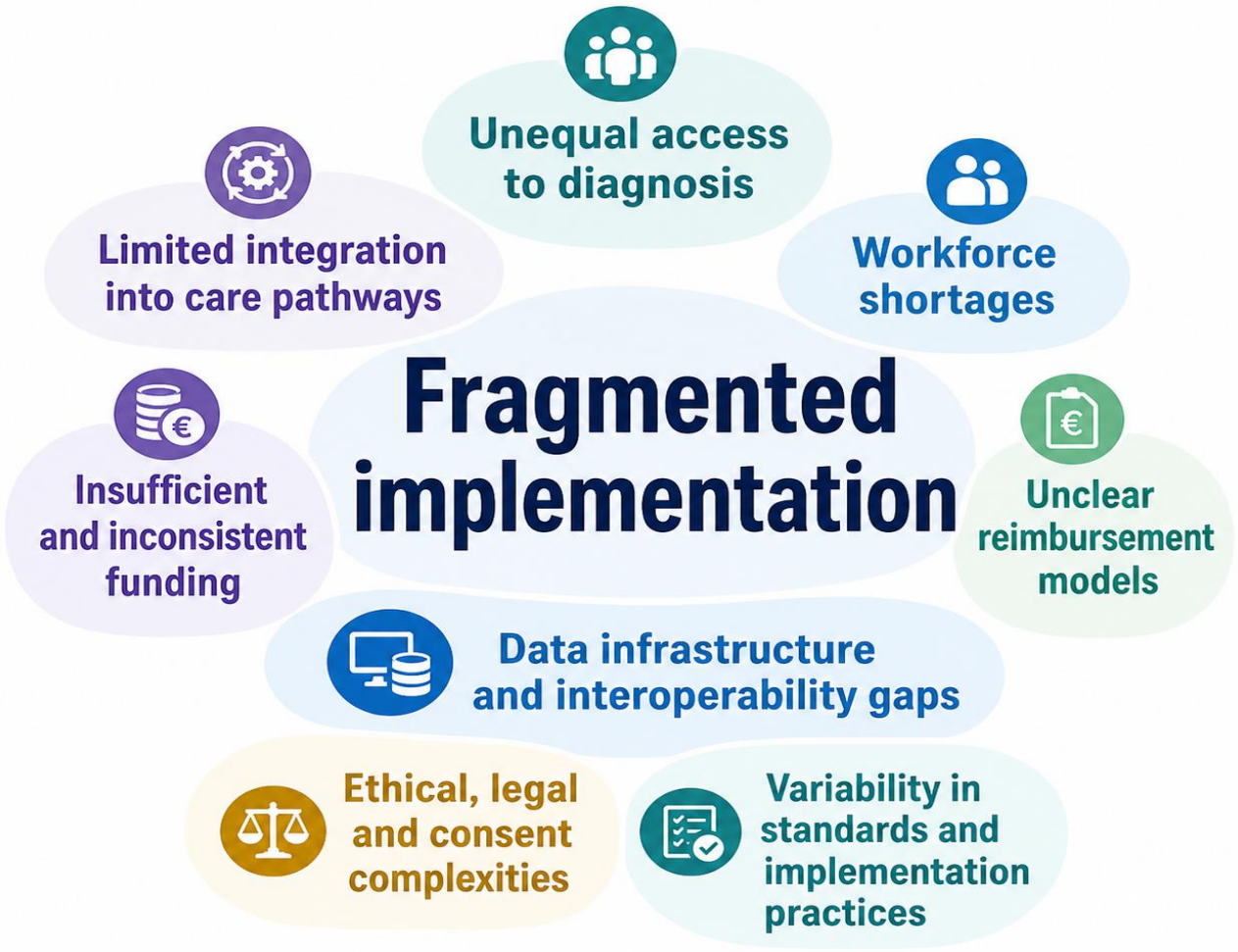
— **Approved therapies are not on every market.** Orphan medicines launch and are reimbursed at very different speeds across Member States.

— **Fiscal capacity and cohort size matter.** Small countries and small patient numbers weaken access and bargaining power.

— **Result:** a “postcode lottery” — the same diagnosis, a different outcome by geography.

Equity means a guaranteed minimum: timely diagnosis and access to the best available care — everywhere in the EU.

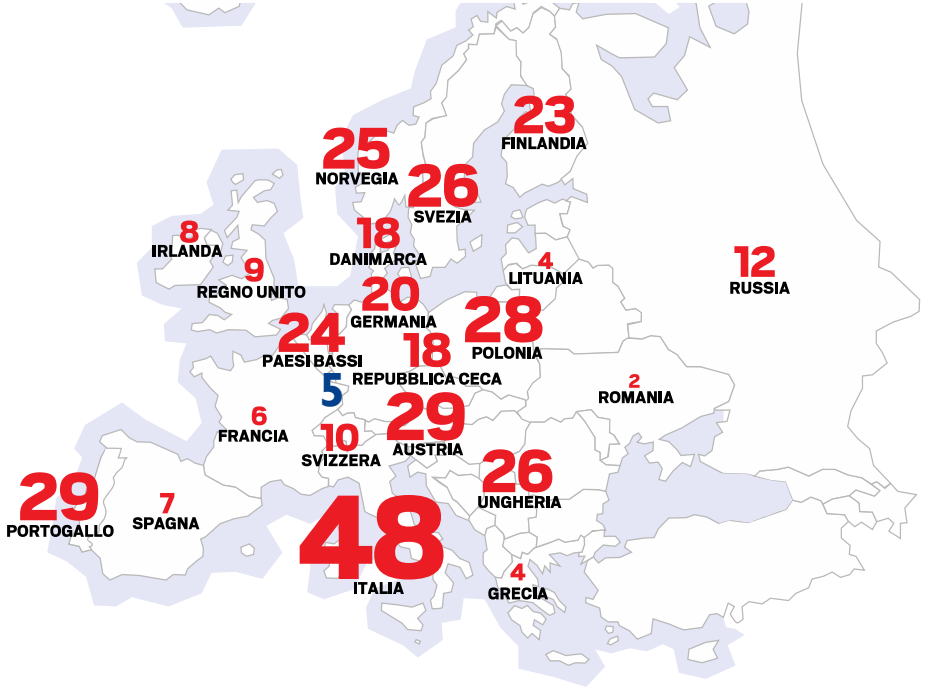
SHARED STRUCTURAL CHALLENGES ACROSS THREE KEY AREAS: NEWBORN SCREENING, GENOMIC DIAGNOSIS & ANTENATAL DIAGNOSIS



DIFFERENCES IN NEWBORN SCREENING PROGRAMMES BY MEMBER STATE

·The conditions that we have chosen to screen

- *Neonatal Screening in Europe Revisited: An ISNS Perspective on the Current State and Developments Since 2010 J. Gerard Loeber et al* (Int. J. Neonatal Screen. 2021, 7, 15. https://doi.org/10.3390/ijns7010015
- Still shows considerable variation in practice both in the way that newborn screening is conducted and the number of conditions screened.
- *Number of conditions* 2 - 48 conditions screened in different countries in Europe in 2025



·The way in which we conduct screening

• Day of sampling	24h – 120h
• Sampling to analysis	1-2d, to 30d
• Screening is optional or compulsory	30 optional, 17 compulsory
• Informed of the outcome of screening	30 No, 10 Yes
• Consent to storage	34 – No, 10 - Yes

Difference
Difference
Difference

NEWBORN SCREENING: HIGHLY FRAGMENTED AND UNEVENLY IMPLEMENTED

01 Newborn screening in Europe remains fragmented, with no common EU framework guiding implementation across Member States.

02 The scope of screening programmes varies significantly, with major differences in the number of conditions covered.

03 Variation in implementation practices affects the quality and comparability of screening programmes.

04 Financial constraints and workforce shortages continue to limit the expansion and quality of screening programmes.

05 Governance and coordination of newborn screening systems remain inconsistent across countries.

06 Screening is not systematically integrated with care pathways, leading to delays in diagnosis, referral and treatment.

07 Data collection, monitoring and performance evaluation are not harmonised at EU level, limiting system improvement.

08 The uptake of innovation, including genomic technologies, remains uneven due to regulatory, ethical and resource barriers.

Screen4Care – gNBS Pilot Overview

- EU IMI2 project (2021–2026) focused on early diagnosis of rare diseases
- Genetic & Genomic Newborn Screening (gNBS) using dried blood spots (DBS)
- Objective: detect treatable rare diseases before symptom onset

Timeline

- 2021–2023: Project setup, ethics, pipeline & panel development
- 2024: Ethics approvals and preparation of pilot screening
- Late 2024 / Early 2025: Start of newborn recruitment
- 2025: Active recruitment and sequencing (~18–25k newborns)
- 2026: Data consolidation and policy recommendations

Strategic Impact

- Evidence generation for EU-wide gNBS implementation
- Reduction of diagnostic delay in rare diseases
- Support for policy decisions and health system integration

TREAT Panel

Purpose

- Focus on treatable and actionable rare diseases
- Designed to complement traditional metabolic NBS

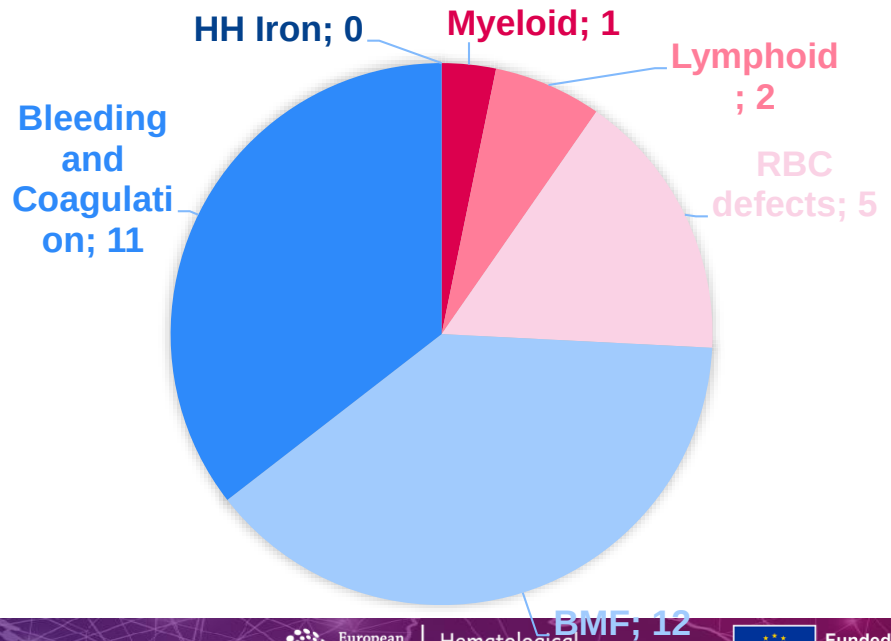
Gene selection criteria

- Strong gene–disease association
- Early childhood onset and high disease severity
- High penetrance and technical feasibility by NGS

Composition

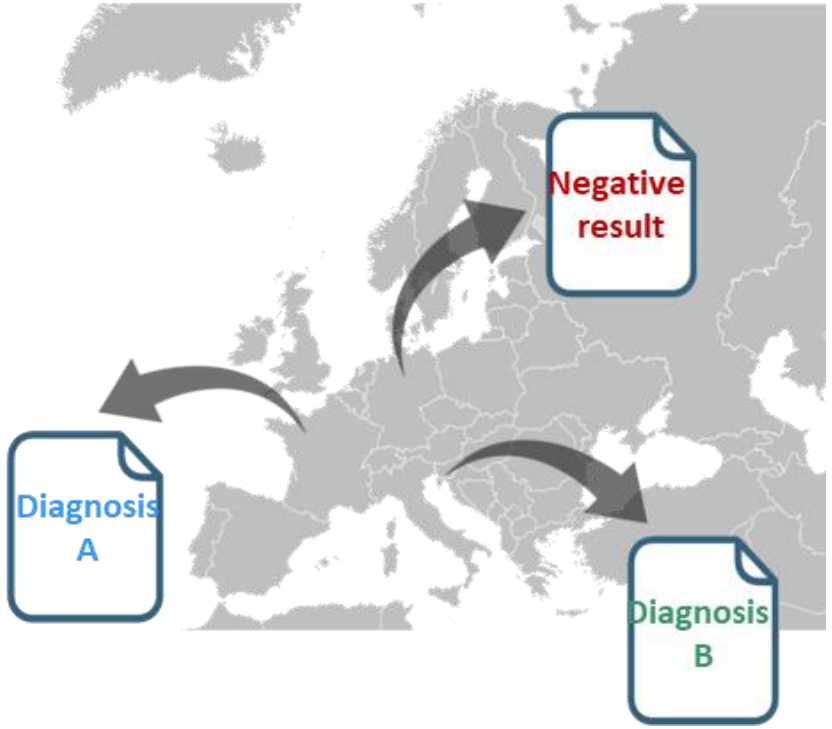
- 245 genes selected from an initial list of ~484 gene–disease pairs
- Includes metabolic, neurological, immunological, endocrine disorders
- Only pathogenic / likely pathogenic variants are reported

Category	ERN	Total number of genes (245)
Blood and coagulation disorders	EuroBloodNet	33
Cardiological disorders	GuardHeart	4
Endocrinological disorders	ENDO-ERN	29
Immunological disorders	ERN RITA	26
Kidney diseases	ERKNet	9
Metabolic (including mithocondrial disorders, oxidation disorders, lysosomial disorders, etc...)	MetabERN	106
Neurolologic/neurodegenerative and neuromuscular disorders	RND, Euro-NMD,ITHACA	25
Syndromic	ITHACA	6
Others	ERN-BOND, ERN-LUNG, ERN-EYE	7



GENOMIC DIAGNOSIS: QUALITY AND TRANSLATION

Quality of NGS based genetic diagnostic testing across Europe significantly differs

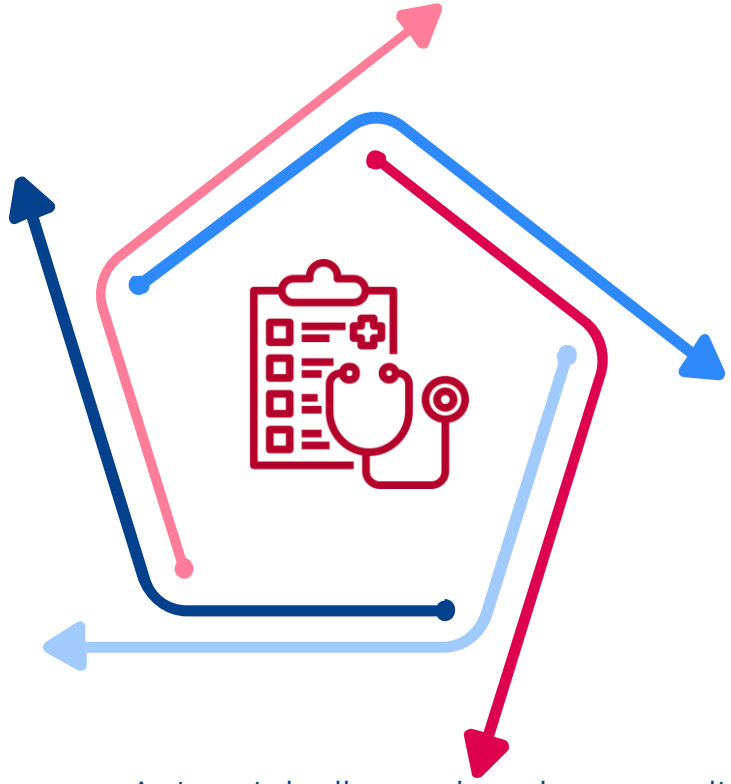


No framework in place for translation of diagnosis into care, treatments and therapy

ANTENATAL DIAGNOSIS: SIMILAR UNEVEN ACCESS AND COMPLEX IMPLEMENTATION

Antenatal diagnosis is advancing rapidly, but access to high-quality prenatal ultrasound, maternal–fetal medicine and confirmatory testing remains uneven across health systems.

Antenatal diagnosis is most effective when it is fully integrated into a continuum of care that includes counselling, confirmatory testing, clinical management and follow-up.



There is significant variation in how antenatal diagnostic services are implemented across countries, including differences in testing protocols, timing, consent procedures and regulatory frameworks.

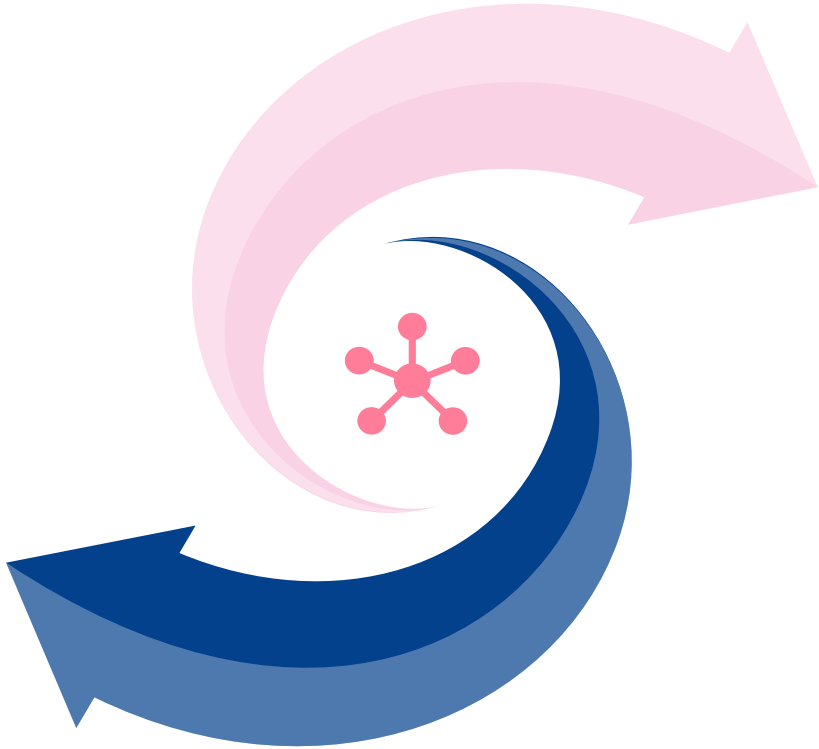
Antenatal diagnosis still involves significant overall costs, including high-quality imaging, laboratory infrastructure, data analysis and specialist personnel.

Antenatal diagnosis raises sensitive ethical questions. This includes decisions about pregnancy and uncertain outcomes.

WG 3 EARLY DIAGNOSIS: STRATEGIC PRIORITIES UNDER DISCUSSION

Pillar I: Newborn Screening

1. EU-wide harmonisation of standards and scope
2. Integrated data and digital Infrastructure
3. End-to-end care pathways (Screening → Treatment)
4. Strong EU governance and coordination mechanisms
5. Multi-stakeholder collaboration and ecosystem integration



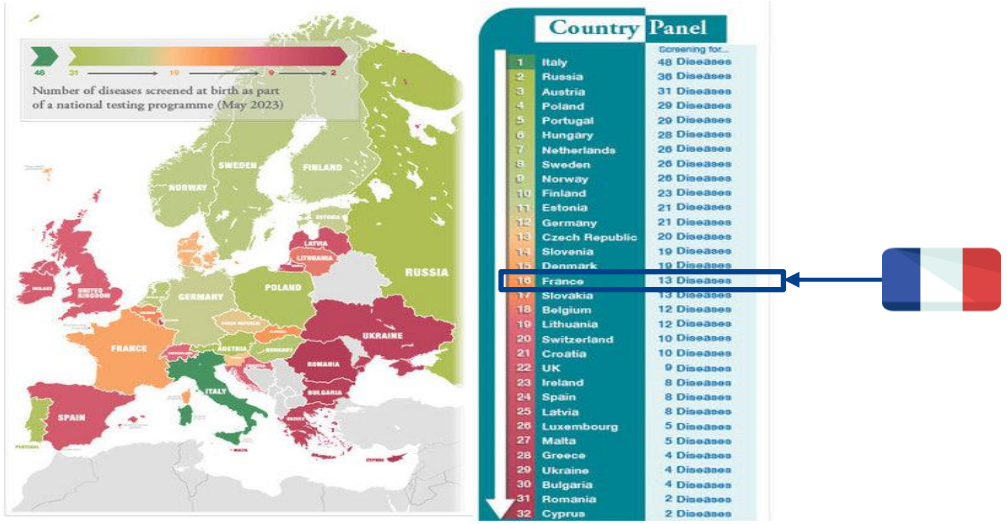
Pillar II: Rapid genetic diagnosis

1. European minimum access standard for rare disease diagnosis
2. ERN- and Centre of Expertise-based pathways for complex cases
3. Interoperable coding, registry and genomic data infrastructure
4. Routine reanalysis and unresolved-case solving
5. Equitable implementation across all Member States

French NBS Program Timeline

The French newborn screening (NBS) program, a pioneer since 1972, is behind several European countries that screen for 20 or more rare diseases.

1972	1978	1985	1995	2002	2018	2020	2021	2023	2025
Phenylketonuria	Congenital hypothyroidism	Sickle cell disease (overseas)	Sickle cell disease in mainland France Congenital adrenal hyperplasia	Cystic fibrosis	Full reorganization of NBS	MCAD deficiency	Authorization of first-line genetic testing	MSUD, HT-1, IVA, DGCDH, LCHAD, HCU, CUD	SMA, immune deficiencies, VLCAD



	Country	Panel (Screening for...)
1	Italy	48 Diseases
2	Russia	36 Diseases
3	Austria	31 Diseases
4	Poland	29 Diseases
5	Portugal	29 Diseases
6	Hungary	28 Diseases
7	Netherlands	26 Diseases
8	Sweden	26 Diseases
9	Norway	26 Diseases
10	Finland	23 Diseases
11	Estonia	21 Diseases
12	Germany	21 Diseases
13	Czech Republic	20 Diseases
14	Slovenia	19 Diseases
15	Denmark	19 Diseases
16	France	13 Diseases
17	Slovakia	13 Diseases
18	Belgium	12 Diseases
19	Lithuania	12 Diseases
20	Switzerland	10 Diseases
21	Croatia	10 Diseases
22	UK	9 Diseases
23	Ireland	8 Diseases
24	Spain	8 Diseases
25	Latvia	8 Diseases
26	Luxembourg	5 Diseases
27	Malta	5 Diseases
28	Greece	4 Diseases
29	Ukraine	4 Diseases
30	Bulgaria	4 Diseases
31	Romania	2 Diseases
32	Cyprus	2 Diseases

New genomics technologies thus present an opportunity to catch up and turn the French NBS program into a tool for the early identification of children with treatable rare diseases.

Sources: sante.gouv.fr, National Neonatal Screening Program, 2021 Activity Report, Charles River Associates & Novartis, 2023

Perigenomed in Clinic: Selection of the gene list

The candidate genes selected for the first phase are drawn from a predetermined gene list, based on lists from other international pilot projects and reviewed by French experts.

Collaboration with Other International Pilot Projects

GUARDIAN STUDY

BeginNGS NEWBORN GENOMIC SEQUENCING

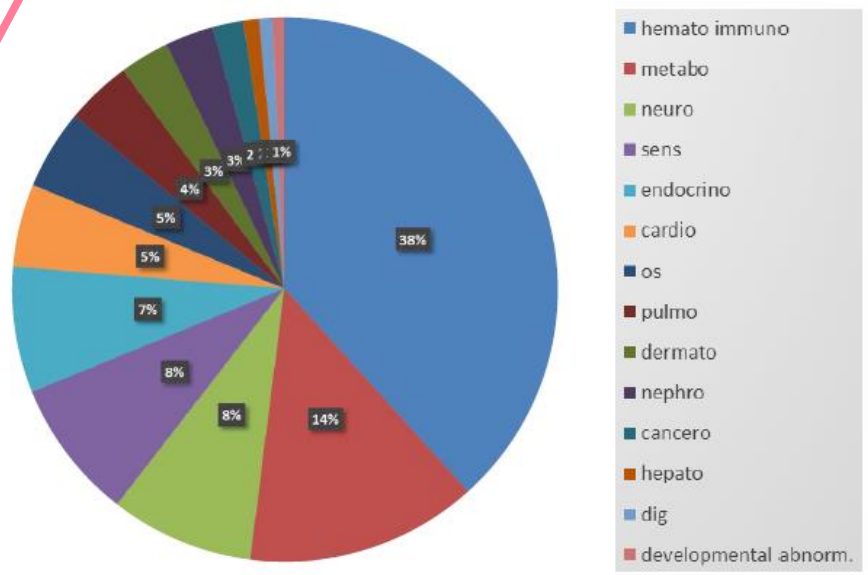
Genomics england

Study of Transferability in France (actionability, management, etc.) in collaboration with the FSMR Rare Diseases Network.

filières de santé maladies rares

PERIGENOMED

PERINATAL GENOMIC MEDICINE



List 1 : « treatable »

400 genes responsible for 171 treatable rare diseases or disease groups, for which early diagnosis would confer substantial benefit to the child before the age of 5

List 2 : « actionnable »

407 genes for 218 diseases or disease groups, with expanded considerations regarding types of actionability, age of symptom onset, and other factors

Perigenomed in Clinic – The biological workflow and performance

Preliminary results from the Dijon experience

Newborn Genomic Screening

Total cases

1.5%

Genomic Screening Positivity Rate

Represents the percentage of newborns with a positive genomic screening result.

Escalation & Interpretation Funnel

32%

Require Biologist Interpretation

A high volume of cases currently necessitates manual review by a laboratory biologist.

12%

Escalated to Multi-Disciplinary Review

Cases requiring complex analysis are discussed in RCP (Multidisciplinary Team) meetings.

Expert Intervention

1.3%

of All Cases Require External Experts

Only one-third of RCP-discussed cases eventually require specialized expert advice.

High Expert Reactivity

External specialists demonstrate rapid response times when providing diagnostic opinions.

Final Results & Timeliness

Inclusion — Results Delivered

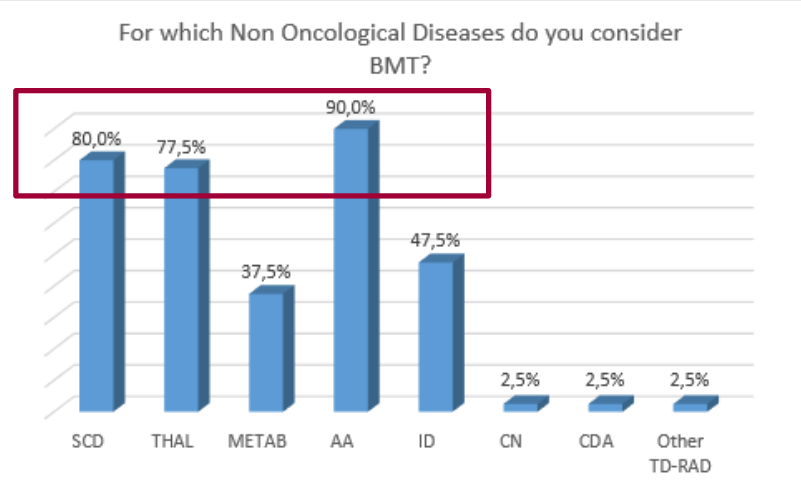
Results are delivered to clinicians or parents approximately two weeks after inclusion.

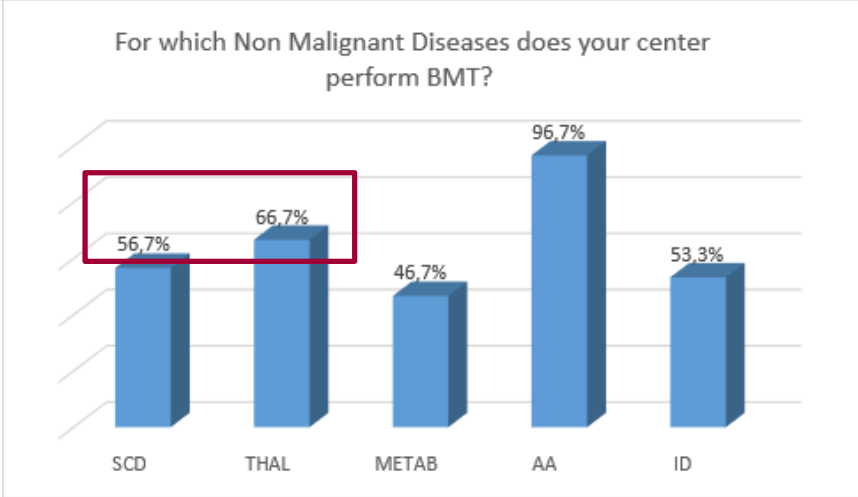
EU MS access to: Bone marrow transplant in non-Onco 2018

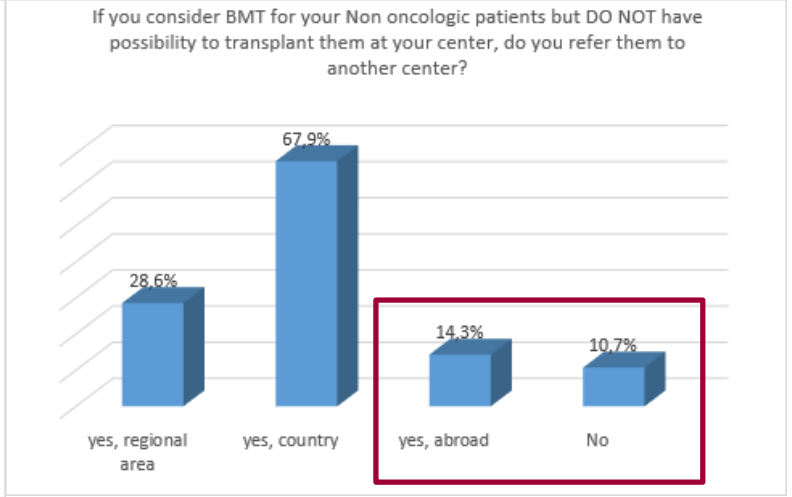
- Bone marrow transplant Units recognized for oncology, Expertise in non-Oncological diseases differs from Oncological diseases, Oncological patients are prioritized – waiting lists

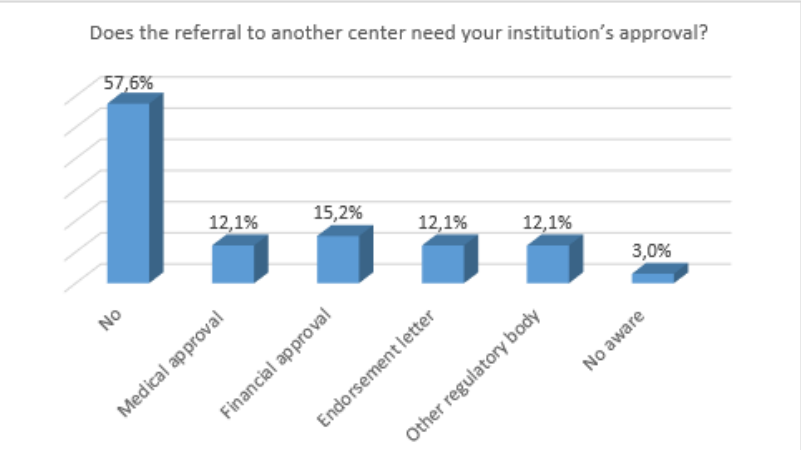
EBMT
European Society
for Blood and Marrow Transplantation

- 48 responders from 44 HCPs / 44 HCPs from 56 non-oncological









EuroBloodNet Pilot bilateral agreement for cross-border health on BMT in SCD

Ireland (Dublin) – Italy (Padova) 2018-2019

- Clinical departments - Medical Protocol
- Administrative - Financial and Logistics Protocols
- Health Ministries Legal Framework (EU Directive and Regulation)

- COVID19
- Ireland decided to create the Expert Unit in Dublin

Clinical Patient Management System version 2.0 (CPMS)

	Active Accounts	Patients in the system	Patients enrolled
May 2025	100	56	2
Sept 2025	103	58	2
May 2026	**132 (33 new in 2026)**	**60**	**2 (1 under discussion)**

- **New patient case in progress** (24/04/2026): VWD (NL) – under discussion – 1 Meeting via CPMS planned 20/05

Opportunities for Virtual Panels:

- Newborn screening results: what to report?
- Diagnosis in Rare Anemia Disorders
- Follow up of cross border patients

A case in point: thrombotic thrombocytopenic purpura (TTP)

TTP is a haematological emergency: untreated mortality exceeds 90%, yet with prompt, appropriate treatment most patients survive. It makes the two gaps tangible.

DIAGNOSTIC BARRIER — ADAMTS13 TESTING

— **Diagnosis hinges on one test.** Confirmation requires ADAMTS13 activity below 10%.

— **Rapid testing is not universal.** Many centres send samples out, losing days when hours matter.

— **So teams act on clinical scores alone,** creating a gap between expert and non-expert centres.

TREATMENT BARRIER — ACCESS TO THERAPY

— **Standard of care has evolved.** Plasma exchange + immunosuppression, now with caplacizumab.

— **Caplacizumab improves outcomes.** Faster platelet recovery; fewer recurrences, thrombo-embolic events and deaths.

— **But it is not available or reimbursed everywhere** — so access depends on the market, not the patient.

Same disease, same guidelines - but the time to diagnosis and the medicines on the shelf still differ across borders.

From barriers to a Plan of Action — Working Group 3

LEVERS WG3 CAN PULL

— **Minimum standards for diagnosis** — quality, timeliness and referral, including access to rapid confirmatory tests.

— **Embed ERNs & centres of expertise** into national diagnostic and care pathways.

— **Early access to innovation** — close the gap between approval and the patient.

— **Interoperable data & registries** to measure where inequity actually occurs.

TO SPARK OUR DISCUSSION

1. Which would be the main levers to **improve diagnosis performance?**

2. **What mechanism** closes the therapy-access gap for approved orphan drugs?

3. **Can ERNs / EuroBloodNet** act as guarantors of equitable referral and access?

4. **How do we measure equity** — which indicators belong on the Action-Plan dashboard?

THANK YOU!

European Reference Network | Hematological Diseases (ERN EuroBloodNet)

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