

How health system structure shapes Rare Tumour Risk Syndrome (RTRS) care: lessons from three European models for EU policy



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INTRODUCTION

Rare Tumour Risk Syndromes (RTRS) are inherited conditions that increase lifetime cancer risk. Effective care requires **early detection, multidisciplinary coordination, and long-term follow-up**. Yet across Europe, care remains fragmented, with disparities in funding and access to genetic services. Healthcare system design critically shapes RTRS pathways, influencing delivery and sustainability.

As part of the Horizon Europe PREVENTABLE project, we explored how financial and structural features of three national health systems affect RTRS care—offering lessons for rare disease policy more broadly.



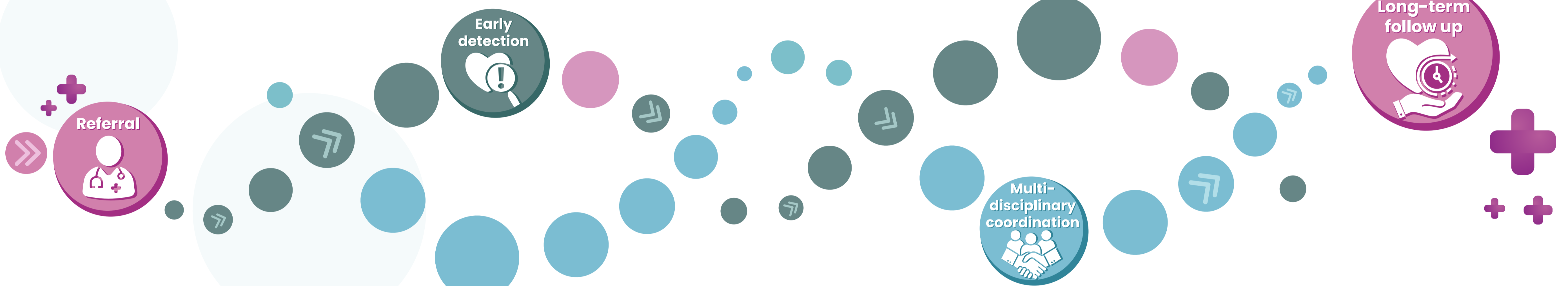
Adapt health system structures to ensure equitable access, early detection, and sustainable RTRS care across Europe.



EU-wide guidelines and reimbursement models are needed to guarantee equal RTRS care in all health systems.

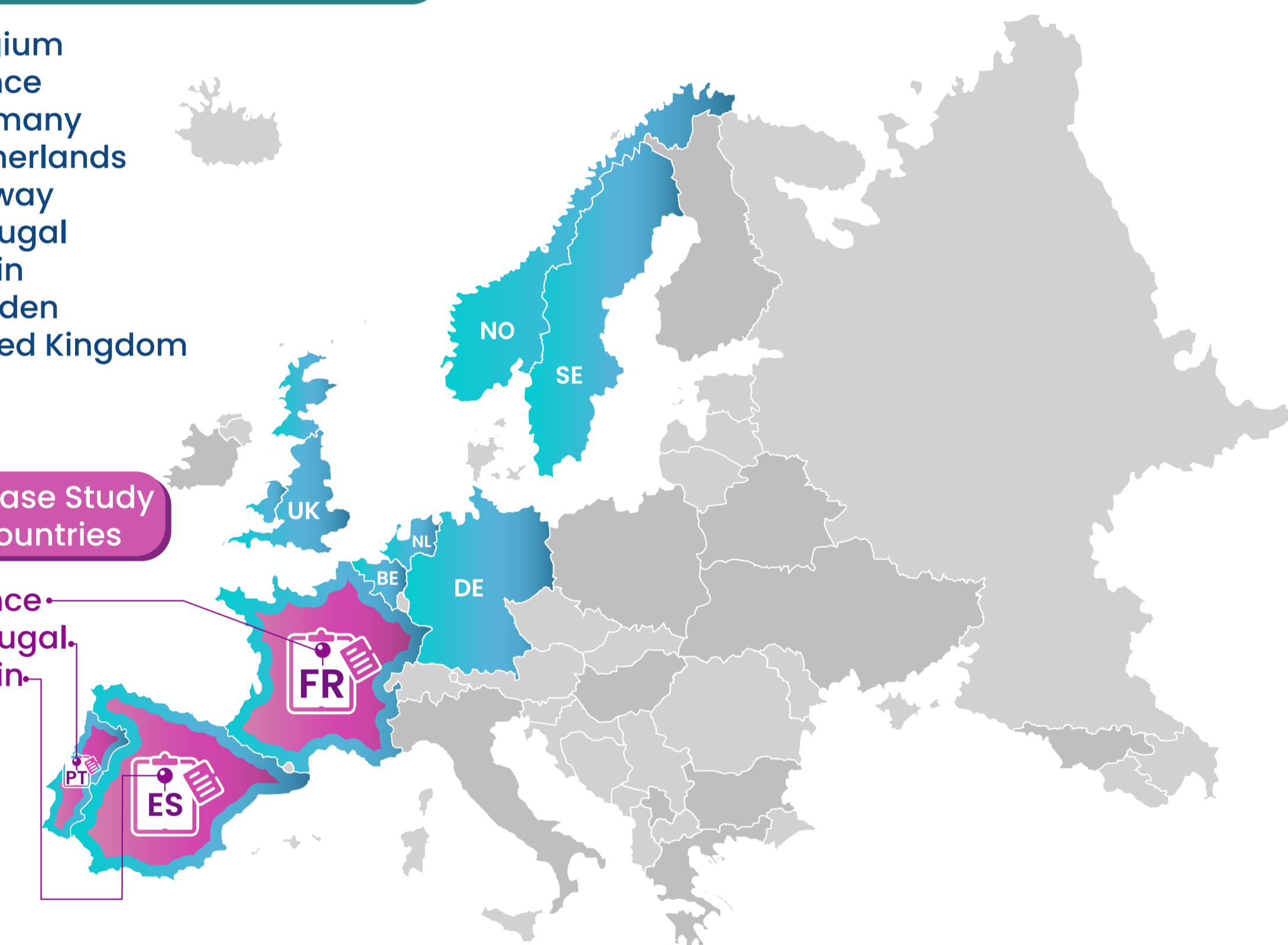


Prevention is not an expense. It is a cost-saving public health strategy. Funding RTRS today prevents tomorrow's cancer and care inequalities.



Countries that belong to the PREVENTABLE consortium

- BE Belgium
- FR France
- DE Germany
- NL Netherlands
- NO Norway
- PT Portugal
- ES Spain
- SE Sweden
- UK United Kingdom



METHODS

Design: SWOT analysis with three case studies – France (centralised), Spain (decentralised), and Portugal (mixed).

Aim: Assess how system structures influence RTRS care access and financing.

Data sources: Desk research, semi-structured interviews, qualitative analysis.

Focus questions: How is RTRS care implemented? What systemic challenges persist?

Sample: 6 interviews with specialists from France, Spain, and Portugal.

Context: Part of the PREVENTABLE project's broader SWOT analysis covering 13 European countries and 19 specialists on RTRS care pathways.



Policy Recommendations



Adapt health system structures
Integrate genetic services across centralised, decentralised, and mixed systems to reduce fragmentation.



Ensure equity in access
Guarantee coverage for genetic testing, prevention, and follow-up, regardless of geography or income.



Secure sustainable funding
Support outcome-based reimbursement and stable hospital financing to strengthen prevention and innovation.



Foster European collaboration
Invest in interoperable IT, expand the workforce, and reinforce RTRS networks and ERN-GENTURIS.

RESULTS FINDINGS

of the SWOT Analysis of Case Studies

STRENGTHS

Internal aspects of RTRS care pathways that are functioning well and contribute positively to patient outcomes.

WEAKNESSES

Internal challenges within RTRS care pathways that hinder the quality, effectiveness, and accessibility of care.

OPPORTUNITIES

Potential areas for growth to optimise care pathways for RTRS patients.

THREATS

External risk and systemic challenges that hinder the effective management of RTRS care.

Centralised Healthcare Systems

- Universal coverage, full reimbursement
- National guidelines & tariffs
- Multidisciplinary oncology boards
- Reimbursed cancer screening

- DRG too rigid for RTRS
- Limited prevention coverage
- Fragmented IT systems
- Specialist shortages & regional gaps

- Prevention-linked reimbursement models
- New coordination roles (case managers, advanced nurses)
- Digital monitoring for genetics
- EU support for RTRS networks

- Specialists shift to private care
- Rising patient costs
- Slow adoption of innovation
- System under strain from budget cuts

Decentralised Healthcare Systems

- Universal cancer care coverage
- Shared professional guidelines
- Regional innovation
- New specialists: clinical genetics

- Unequal access by region
- Fragmented health records
- Genetics speciality only recent
- Slow inter-regional authorisations
- Low RTRS awareness in primary care

- EU-level guidelines reduce regional variation
- European funding for rare diseases
- Cascade testing for families
- Mapping expertise to improve referrals

- Ongoing regional disparities
- Slow integration of new specialists (clinical geneticists)
- Administrative delays across regions
- Gaps in non-medical support (psychology, nutrition, etc.)

Mixed Healthcare Systems

- Universal oncology coverage
- Social security support (esp. palliative care)
- Private care: shorter waits & choice

- Chronic underfunding
- Risk consultations only in three centres (Lisbon, Porto & Coimbra)
- Long waits for exams & surgery
- No public/private consultation
- Opaque private pricing, cost variation

- Online waiting list transparency
- EU collaboration & joint projects
- Public-private partnerships in testing & follow-up
- Regulation of private pricing

- Competition between private providers
- Interrupted treatments when insurance ends
- High out-of-pocket costs (tests, fertility, drugs)
- Negative media image of public care
- Innovation costs exceed budgets

Shared Findings

Cross-cutting lessons for RTRS

- Universal coverage for oncology care
- Specialist expertise & cancer centres
- Participation in ERN-GENTURIS

- Underfunded hospitals & staff shortages
- Limited prevention reimbursement
- Fragmented IT / records systems

- EU-level guidelines & funding
- Digital tools for monitoring & follow-up
- Strengthened RTRS networks & collaboration

- Inequities in patient access
- Slow adoption of innovation
- Rising out-of-pocket costs
- Sustainability pressures on public systems



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