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CHALLENGES TO IMPLEMENT EUROPEAN REFERENCE NETWORKS IN THE MALTESE SECONDARY HEALTHCARE SECTOR

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ABSTRACT: This study examines the barriers to integrating Malta's secondary healthcare sector into the European Reference Networks (ERNs), an EU initiative that aims to improve care for patients with rare and complex diseases through cross-border collaboration. Despite the benefits ERNs offer, Malta's participation remains constrained by regulatory fragmentation, limited resources, data governance concerns, and infrastructural shortcomings.

Adopting a qualitative interpretivist approach, the research involved five semi-structured interviews with healthcare professionals and policymakers selected through snowball sampling due to the specialised nature of the topic. Thematic analysis identified seven interconnected challenges, including weak governance structures, policy misalignment with EU expectations, limited digital interoperability, workforce capacity gaps, unclear institutional roles, and outdated infrastructure. A key issue highlighted was the absence of a dedicated national ERN coordination mechanism, which hampers strategic alignment and effective implementation.

The findings indicate that successful ERN integration requires more than regulatory compliance. It demands coordinated

national leadership, sustainable investment in digital systems and specialised training, and strengthened cross-sector governance. Although stakeholders expressed optimism regarding ERNs' potential benefits for small Member States like Malta, the study concludes that targeted health policy reform and long-term strategic planning are essential to enhance rare disease care delivery and ensure meaningful integration.

Keywords: *European Reference Networks; Rare Diseases; Health Governance*

1 Introduction

Imagine hearing, "There is no cure for you here". For over 30 million Europeans living with rare diseases, this is a devastating reality (European Commission, 2024). The European Reference Networks (ERNs), launched in 2017 (Talarico et al., 2022), were designed to address these gaps by fostering cross-border collaboration and pooling expertise to improve healthcare outcomes (Talarico et al., 2022). Despite their promise, systemic barriers, such as resource inequities (Talarico et al., 2020) and infrastructure limitations (Tumiene et al., 2021), have hampered their implementation across various Member States (MS).

The establishment of ERNs is part of the European Union's (EU) response to widespread health concerns and the need for better care for rare and complex conditions, which collectively affect millions of citizens (Moliner & Waligora, 2017). In 2011, the Directive on Patients' Rights in Cross-Border Healthcare provided the framework for establishing ERNs (Héon-Klin, 2017), with the first 24 networks launched in 2016, centralising expertise and improving access to specialised care across MS (Talarico et al., 2022). That same year, Malta launched its National Rare Disease Register to address the needs of approximately 30,000 citizens living with rare diseases (Directorate for Health Information and Research [DHIR], 2024). However, Malta faced challenges in participating in ERNs, with Health Minister Chris Fearne in 2017 attributing delays to the country's small size and limited resources (Farrugia, 2017). While implementation began in 2021, concerns about data-sharing practices and cybersecurity risks slowed progress (Ministry for Health and Active Ageing, 2024). Efforts to further integrate Malta into ERNs continued, as the country invested in telemedicine and aligned with EU data-sharing protocols to support cross-border collaboration (DHIR, 2024).

This study aimed to identify systemic, regulatory, and operational barriers to implementing the ERN system in the Maltese healthcare system and to assess the readiness of Maltese healthcare providers to meet ERN standards. Through this focused analysis, the research sought to generate insights that apply to other small nations facing similar constraints.

Guided by the research question, "What challenges do Maltese secondary healthcare providers face to become functional members of ERNs?" this dissertation sought to provide actionable strategies to address these barriers. The findings were intended to inform healthcare policy development and enhance care delivery for patients with rare diseases in Malta and similar contexts. In line with the study's overall research question, the research objectives were:

1. To identify the challenges policymakers face in implementing ERNs within the Maltese secondary healthcare sector.
2. To assess the views of different secondary healthcare providers in Malta regarding implementing ERNs.
3. To formulate a strategy identifying the essential attributes and capabilities required for healthcare providers to effectively participate as functional members of ERNs within Malta's secondary healthcare system.

2 Review of past studies

By collaborating through the framework of the ERNs, secondary health sector providers felt that with patient data now being shared on a larger scale than before, the chances of the private healthcare industry gaining access to information was more probable due to its efforts to gain a competitive edge in the market of medicinal products and medical equipment sales through its intertwining with the secondary healthcare sector of various EU MS. Research sufficient to implicate whether this has affected Malta's level of participation lacks; however, a cross-examination of national plans submitted by EU MS about the divulgement of their national strategies for treating rare diseases, published by Hedley et al. (2021), indirectly confirmed that the situation could have potentially existed. Hedley et al. (2021) stated that Malta was among the MS that had not yet submitted the national plan in 2020, even though it was requested in 2013, which was way before the commissioning of the ERNs.

2.1 Challenges Related to Data Protection and Privacy Concerns

Moreover, the fears of data exposure and conflicts of interest were not isolated to Malta. Studies have shown that other EU countries also experienced similar apprehensions. For instance, a 2019 report by the European Federation of Public Service Unions highlighted widespread unease about data security and the influence of private companies in public health sectors across Europe (Stevenson, 2019). These concerns underscored the need for stringent data protection measures and transparent policies to mitigate risks associated with ERN participation and to ensure that commercial motives did not compromise public health interests. Furthermore, according to von Ditfurth and Lienemann (2022), the new Data Governance Act (DGA) introduced data altruism frameworks, which raised concerns about the compatibility of these initiatives with existing GDPR provisions and the potential risks associated with data sharing and re-identification (von Ditfurth and Lienemann, 2022). Additionally, Verma and Gurtoo (2023) emphasised the importance of clearly defined objectives and robust monitoring mechanisms for data altruism organisations to mitigate privacy risks.

Additionally, recent academic research underscored the growing risk of cyberattacks on European healthcare institutions. Nifakos et al. (2021) discussed significant cyberattacks that disrupted services in hospitals across many countries, emphasising the urgent need for robust cybersecurity measures to protect sensitive health information from malicious actors. These authors, based on data from 70 peer-reviewed and conference articles, identified several shortcomings that contributed to the WannaCry ransomware attack of 2017, including the need for cybersecurity training among healthcare stakeholders. These authors highlighted vulnerabilities in the healthcare sector and the need for improved security protocols. Adil et al. (2024) emphasised the importance of multi-factor access control and ownership transfer frameworks to enhance security in future healthcare systems, illustrating the critical need for advanced security measures. Furthermore, the challenges of safeguarding health data in the digital age were exacerbated by the limitations of current EU regulations. According to Marelli et al. (2020), the General Data Protection Regulation (GDPR) faced issues like inconsistent enforcement across MS and inadequate protection against data inferences, posing significant threats to patient privacy and stated that strengthening the enforcement powers of national data protection authorities and enhancing transparency in data collection practices were critical steps to address these

vulnerabilities. This was because, in cross-border healthcare initiatives like ERNs, data security concerns extended beyond the borders of the patient's home MS, requiring each MS to ensure that its patients' data remained secure regardless of where it is accessed or processed.

2.2 Stakeholders' Views on the Implementation of ERN

The successful implementation of ERNs relied heavily on the engagement and cooperation of various stakeholders, including healthcare providers, patients, policymakers, and private industry representatives. Each group of stakeholders played a pivotal role in shaping the effectiveness and sustainability of ERNs (Tumiene et al., 2021). Therefore, this section was organised into four primary subsections to focus on the perspectives of healthcare providers, insights from patients and patient advocacy groups, policymakers' viewpoints, and private industry considerations.

Healthcare providers brought their clinical expertise and frontline experience to diagnose and treat rare diseases, making their input vital for the practical application of ERN initiatives. Patients and their advocacy groups offered invaluable insights into the lived experiences and specific needs of those directly affected by rare diseases, ensuring that ERN policies and practices remain patient centred. For example, Blay et al. (2021) emphasised the importance of healthcare providers in contributing their specialised knowledge and experiences to the ERN EURACAN. They noted that providers are essential in implementing best practices and standardising care across the network. Furthermore, various publications highlighted the significant role of patient advocacy groups in ensuring that the voices of those affected by rare diseases were heard (Blay et al., 2021; Nori et al., 2022). Their involvement helped tailor ERN policies to be more patient-centric, promoting better healthcare outcomes. In addition, the EC underscored the need for robust cooperation among all stakeholders, including policymakers, who created the necessary regulatory frameworks and funding mechanisms to support ERNs (Hedley et al., 2023). This collaboration was vital for overcoming administrative and legal barriers to cross-border healthcare, thereby enhancing the overall functionality of ERNs. The ability of policymakers to harmonise diverse national healthcare systems under a unified European strategy was essential for overcoming administrative and legal barriers to cross-border healthcare. Meanwhile, the private industry contributed through innovation in

medical products and technologies, driving the development of new treatments and therapies (Horgan et al., 2020). Their involvement also brought challenges, such as balancing commercial interests with ethical considerations and patient privacy.

Healthcare providers were at the forefront of patient care and generally viewed ERNs as a positive development. Iotova et al. (2021) highlighted the opportunities for enhanced collaboration and access to a broader pool of expertise and resources. Their work showed that clinicians and researchers particularly emphasised the importance of educational initiatives and collaborative efforts within ERNs. They identified significant knowledge gaps in treating rare endocrine conditions, especially during transition periods and neonatal care, and stressed the need for structured educational programs and resources tailored to these gaps. According to a survey by Talarico et al. (2020), many providers believed that ERNs facilitated better diagnostic and treatment options for rare diseases, which would have been challenging to achieve within a single national healthcare system. This network approach enabled the sharing of best practices, the standardisation of care, and the possibility of joint research initiatives, all of which contributed to improved patient outcomes. Additionally, Shishkov et al. (2023) underscored the significance of ERNs in improving clinical outcomes through enhanced interdisciplinary collaboration. Their study indicated that healthcare providers valued the collective expertise and resource pooling facilitated by ERNs, significantly enhancing the diagnostic and treatment capabilities for rare diseases. Moreover, Tumiene and Graessner (2021) highlighted the crucial role of ERNs in fostering a more integrated and patient-centred approach to care. They emphasised that healthcare providers appreciated the structured support and standardisation that ERNs bring, which was essential for managing complex rare diseases effectively.

One significant concern among healthcare providers was the adequacy of funding and resources allocated to ERNs. Many healthcare providers felt that despite the potential benefits of participation in ERNs, the funding provided by the EC and national governments was insufficient to cover the operational costs and necessary investments in technology and infrastructure (Talarico et al., 2020). This underfunding could have led to inequities in participation and access to ERNs, particularly for healthcare providers in less affluent MS. The uneven distribution of resources could have hindered the ability of these providers to fully engage in ERN activities, limiting the overall effectiveness of the

network. Many ERNs faced financial sustainability issues, with inadequate funding impacting their ability to deliver comprehensive care and conduct essential research (Vandevelde, 2019). This author emphasised the disparities in funding allocations across MS, argued that these inequities contributed to uneven access and participation in ERN activities and suggested that a more equitable distribution of resources is necessary to ensure all MS can benefit equally from the ERNs. Additionally, Adachi et al. (2023) pointed out that the disparities arose from variations in national healthcare budgets and the differing priorities set by individual governments, which further complicated the uniform implementation and success of ERNs across Europe.

Another significant concern of providers was that they must often balance the needs of ERNs with other critical healthcare services, leading to difficult decisions about where to allocate limited resources. This balancing act can result in tensions between departments and impacted the quality of care provided to patients with rare diseases (Hollak et al., 2016). Healthcare providers worried that the substantial resources required by ERNs and other cross-border initiatives, including advanced medical infrastructure, comprehensive data management systems, and cross-border collaborations essential for high-quality care, might have diverted funds and attention from other vital healthcare services (Tumiene et al., 2021). These authors indicated that, unlike other collaborations in healthcare, ERNs required significant overheads to manage their complex network of services and stakeholders, ensuring seamless integration and operational efficiency. Additionally, Horgan et al. (2019) highlighted that the dynamic resource allocation necessary for ERNs can strain existing healthcare systems, necessitating a more adaptive and responsive management approach to balance these needs effectively. These authors emphasised the healthcare providers' concern for the impact on other critical healthcare services, advocated for strategic planning and resource optimisation to ensure that ERNs and other cross-border healthcare initiatives do not compromise the broader healthcare objectives. This intricate balance of resources and priorities remained a pressing issue for healthcare providers engaged with ERNs.

Integrating advanced technologies was crucial for the effective functioning of ERNs, yet it posed a significant challenge for healthcare providers. Many institutions lacked the necessary technological infrastructure to support the data-sharing and communication requirements of ERNs, including electronic health record systems developed in a manner

compatible across different countries and regions, secure platforms for data sharing, and advanced diagnostic tools (dos Santos Vieira et al., 2022). According to Vassolo et al. (2021), the successful implementation of such technologies required substantial investments, which were often beyond the financial capabilities of many healthcare institutions. Additionally, Raycheva et al. (2024) emphasised the importance of interoperability between different technological systems, which was a significant hurdle. They argued that without standardised systems, the full potential of ERNs cannot be realised. Furthermore, Raycheva et al. (2024) emphasised the importance of ongoing training and support for healthcare providers to effectively utilise new technologies, suggesting that inadequate training can lead to inefficiencies and errors, ultimately compromising patient care.

3 Materials and Methods

This study employed a qualitative research design to examine the challenges and strategic requirements associated with implementing ERNs in Malta's secondary healthcare sector. A qualitative approach was selected, given the exploratory nature of the research and the limited contextual evidence regarding ERN integration in small healthcare systems. Semi-structured interviews were conducted with healthcare professionals and policymakers directly involved in ERN-related activities to capture in-depth, context-sensitive insights (Qu & Dumay, 2011; Hamilton & Finley, 2019).

3.1 Research Design and Philosophical Positioning

Primary data was collected to obtain first-hand perspectives from individuals directly engaged in ERN implementation. While ERN research has at times employed quantitative designs, such as surveys (e.g., Iotova et al., 2021), this study prioritised depth and interpretive understanding over statistical generalisability. Qualitative methods are particularly suitable for examining complex organisational and policy issues in healthcare because they capture nuance, meaning-making, and institutional context (Hamilton & Finley, 2019).

The research was guided by an interpretivist philosophy, which assumes that reality is socially constructed and shaped by individuals' experiences and context (Van Der Walt, 2020). An inductive approach was adopted, whereby theory and insights are developed

from empirical observation rather than used to test predetermined hypotheses (Morgan & Nica, 2020). This enabled the study to identify themes grounded in participant experiences and to generate practical, contextually relevant recommendations.

3.2 Data Collection

The primary data collection method comprised one-to-one semi-structured interviews. This method is widely used in qualitative research because it balances structure with flexibility, enabling consistent topic coverage while supporting probing and clarification (Qu & Dumay, 2011). One-to-one interviews were preferred over focus groups because they avoid group dynamics and can better support disclosure when discussing sensitive organisational or policy issues (Nyumba et al., 2018). Semi-structured interviews are also suitable for exploring professional and institutional experiences in healthcare settings (Hamilton & Finley, 2019; Chapman et al., 2015).

Interviews were conducted in English, a working language commonly used across Maltese health and policy sectors, to ensure clarity and avoid translation inconsistencies. Interviews were conducted either in private workplace settings or via secure videoconferencing when face-to-face meetings were not feasible. Participants received the interview questions in advance to increase transparency and allow preparation. With consent, interviews were audio-recorded and supplemented with brief notes to capture contextual observations.

The interview guide was structured into three sections aligned to the research objectives:

1. Section 1. Participant role and professional context (supporting Research Objective 2).
2. Section 2. Barriers and challenges affecting ERN implementation (supporting Research Objective 1).
3. Section 3. Organisational preparedness and strategies for ERN participation (supporting Research Objective 3).

The target participants comprised healthcare professionals and policymakers directly involved in ERN-related implementation, including clinicians specialising in rare diseases, hospital administrators, and policy advisors. Because no comprehensive directory exists for ERN-involved stakeholders in Malta, the population size could not be precisely determined, and a probabilistic sampling frame was therefore infeasible. A non-random

snowball sampling approach was therefore used. Initial participants were identified through professional networks and then asked to recommend additional relevant stakeholders. Snowball sampling is appropriate where the study population is specialised, difficult to identify, and where access depends partly on trust and professional referral (Etikan & Bala, 2017).

3.3 Data analysis

The analysis followed a structured approach that combined manual transcription with computer-assisted qualitative analysis using MAXQDA (Version 24.9.1). Manual transcription was chosen to preserve meaning, particularly where participants used Maltese expressions and context-specific phrasing. MAXQDA was selected for its capacity to organise qualitative datasets, support systematic coding, and facilitate thematic development (Rädiker & Kuckartz, 2020; Star et al., 2025). A thematic analysis approach was applied to identify patterns across the dataset. Thematic analysis is a flexible method for systematically identifying, analysing, and reporting themes within qualitative data (Christou, 2022; Naeem et al., 2023). It has also been applied in ERN-related research contexts to structure stakeholder perspectives and implementation considerations (Evangelista et al., 2016).

First-cycle coding comprised three complementary techniques: initial coding, process coding, and in vivo coding. Initial coding captured broad descriptive concepts; process coding focused on actions and implementation dynamics; and in-vivo coding preserved participants' language to maintain fidelity to stakeholder voice. Each interview generated approximately 120 first-cycle codes, which were iteratively refined and grouped.

Second-cycle coding synthesised first-cycle codes into higher-order categories and themes by clustering recurring patterns and refining overlaps. This process involved merging closely related codes, rewording codes into broader conceptual labels where appropriate, and removing outliers or duplicates. Category development was iterative, requiring repeated checks against the dataset to ensure coherence and representativeness (Saunders et al., 2023). MAXQDA supported this stage by enabling code retrieval, comparison across transcripts, and visual mapping of relationships among emerging themes.

3.4 Methodological Limitations

Several limitations should be considered. First, snowball sampling can introduce selection bias by overrepresenting participants who are more engaged or connected to ERN initiatives, potentially narrowing the range of views captured (Etikan & Bala, 2017). Second, semi-structured interviews carry risks of social desirability bias and interviewer influence; to mitigate this, questioning was kept neutral, and probing was used cautiously (Qu & Dumay, 2011). Third, interpretivist qualitative findings are context-dependent and not statistically generalisable (Van Der Walt, 2020). Finally, the cross-sectional design captured perspectives at one point in time and may not reflect future policy or organisational changes (Kozlowski et al., 2013).

4 Results

Seven themes emerged from the qualitative analysis, each representing a distinct cluster of constraints and enabling conditions shaping Malta's pathway toward full membership of ERNs. Although analytically separable, participants described these themes as mutually reinforcing.

4.1 Theme 1: Governance Architecture and Regulatory Frameworks Shaping the Integration of ERNs within Malta's Health System

Participants consistently described ERN integration as constrained by fragmented governance, overlapping mandates, and unclear accountability between policy formulation and service delivery. A ministry participant emphasised the thinness of administrative capacity in a micro-state setting: "many people wear many hats… but end up not being able to focus on anything." Although hierarchies were described as short, governance was experienced as lacking functional depth. Middle managers were perceived as able to enforce procedural compliance but unable to allocate financial resources, leading to risk-averse practices designed to minimise downstream liability (additional clearances, reviews, and approvals). In effect, compliance becomes the default managerial output because it is measurable and defensible, whereas enabling ERN participation requires discretionary resources and cross-unit coordination.

Clinicians described the lived consequences of this structure as "vertical" and "uphill." When ERN boards require timely submissions, delays arise from sequential approvals

(consent confirmation, data protection clearance, internal sign-off, then transfer). Participants reported that staff often rely on improvised coordination to chase signatures and locate documentation to avoid missing ERN deadlines—workarounds that signal high professional commitment but low system reliability. Data protection clearance emerged as a frequent bottleneck because imaging and clinical reports may contain embedded identifiers, and there are limited fast-track pathways for low-risk transfers.

Political incentives were presented as an additional layer shaping governance. Rare diseases were described as low salience relative to high-volume services and visible infrastructure. Participants noted that system investments essential to ERN participation, interoperability, registry harmonisation, and consent tracking, do not readily translate into immediate "public wins," whereas individual transfers abroad can generate visible narratives. Horizontal fragmentation further exacerbated governance problems: multiple consultant-led registries governed by different MoUs were described as creating inconsistent standards and ambiguity over what constitutes "the" national dataset when ERN hubs request information.

Strategic reforms proposed across interviews included legislative centralisation (e.g., a national ERN authority or commissioner), performance-based service-level agreements (SLAs) linking budgets to ERN outputs, and process re-engineering to reduce administrative friction (e.g., consolidating multiple signatures via electronic countersigning). One recurring "sweet spot" was a hybrid governance model: formal mandate and budgetary levers combined with operational redesign and structured representation of patient organisations.

4.2 Theme 2: Ethico-Legal and Data-Governance Imperatives for Cross-Border Rare-Disease Collaboration through ERNs

Participants framed GDPR compliance as foundational but not sufficient for ERN participation. Cross-border collaboration exposed a dense matrix of legal obligations and administrative expectations that translate into procedural load at the point of care. Clinicians described repeated and extensive documentation for data transfers (including DPIAs) as disproportionate in a low-volume, rare-disease setting, in which the same clinical team must manage both complex care and governance tasks. In practice, legal

caution becomes “work” that competes directly with clinical time, increasing the likelihood of delayed uploads and incomplete participation.

Dynamic consent models linked to Malta’s national eID infrastructure were proposed to enhance autonomy and to manage consent systematically over time. However, ministry and administrative participants cautioned that a more connected consent architecture increases the exposure surface for sensitive data (particularly genomic information) unless cybersecurity capacity is strengthened. This produced a consistent trade-off across accounts: patients want empowerment and efficiency, but institutions fear that technical weakness could lead to security incidents and loss of public trust.

Ethical concerns were not limited to privacy. Participants expressed concern about the commercial reuse of ERN datasets, particularly the risk that research agendas could shift toward profit-seeking products rather than broader patient benefit. Patient-led data governance mechanisms (e.g., licensing models tied to patient-benefit clauses) were suggested as safeguards to preserve legitimacy. At the same time, some participants noted that additional governance structures could add administrative weight unless standard templates and national-level coordination are provided.

Consent fragility emerged as a specific operational vulnerability. Participants highlighted that re-consenting patients at the age of majority is difficult when systems lack automated triggers or integrated records of consent status, thereby requiring manual tracing and resulting in silent attrition of longitudinal data. In a small population, this loss matters: rare-disease evidence depends on continuity. Most participants converged on a tiered consent architecture distinguishing (1) routine care, (2) pseudonymised registry participation, and (3) identifiable research/commercial partnerships, each with proportionate governance. Yet participants emphasised that such models cannot function without real-time consent tracking and tagging across clinical workflows and IT systems.

4.3 Theme 3: Organisational Capacity and Workforce Resilience as Determinants of ERN Operationalisation

Theme 3 positioned ERN engagement as an additional service layer that currently lacks explicit staffing and budget. Participants described ERN work—data entry, uploads, case coordination, board participation, follow-up documentation—as an “invisible department”

added onto already stretched clinical routines. This is amplified by Malta's reliance on single specialists in certain rare-disease areas, creating a single-point-of-failure dynamic: when one consultant is unavailable, ERN-related work slows, referrals are delayed, and institutional memory weakens. Participants linked these delays to real clinical consequences, including postponed funding approvals and longer time-to-treatment pathways.

All groups converged on the need for coordination roles, but diverged on design and sustainability. Ministry-oriented participants raised the practicality of fixed-term, EU-funded posts to work around headcount constraints. Clinicians cautioned that newly recruited coordinators without clinical context require supervision, potentially transferring workload rather than reducing it—especially if turnover is high. A second sustainability concern was raised: if coordination roles are funded temporarily, trained staff may be recruited elsewhere once contracts end.

Clinicians advocated formal recognition of ERN work through job plans and protected time. Without that protection, ERN duties are easily absorbed into generic "admin" and displaced during operational surges. Participants also discussed redistributing expertise without increasing permanent headcount through diaspora mentorship networks—leveraging Maltese specialists abroad for case input, second opinions, and periodic support. However, they stressed that this requires indemnity arrangements, credential verification, and structured governance to avoid informal dependence.

4.4 Theme 4: Health-Information Infrastructure and Interoperability Requirements for Seamless ERN Participation

Participants identified digital infrastructure as a core bottleneck because ERN participation depends on reliable movement of structured data, imaging, and documentation across systems and borders. Although Malta's scale creates the advantage that improvements in Mater Dei can transform most of the system, interviews repeatedly described a back-end reality of fragmented components with limited real-time interoperability. Clinicians described reliance on manual collation of reports, repeated data entry into different platforms, and non-real-time transfers—processes that are incompatible with ERN board timelines and create error risk.

A strategic debate emerged between proprietary solutions and open-source platforms. Some participants associated proprietary tools with vendor dependency and limited flexibility, while open-source was framed as potentially enabling consortium-based development and shared security patching with other states. Others expressed concern about long-term sustainability, support, and cyber risk, especially if systems evolve without adequate internal capacity.

Four prerequisites consistently emerged: (1) a unified patient identifier aligned with pseudonymisation; (2) HL7-FHIR compliant interoperability enabling near real-time exchange; (3) granular role-based access controls; and (4) expanded cybersecurity capacity. The absence of these prerequisites generated behavioural adaptations: clinicians described "self-rationing" uploads, prioritising cases with immediate therapeutic relevance and excluding borderline cases that are valuable for research and learning. This has consequences beyond workload: it skews the national evidence profile and reduces Malta's contribution to ERN knowledge generation.

4.5 Theme 5: Professional Culture, Continuing Education, and Change-Management Dynamics in the Adoption of ERNs

ERN adoption is shaped by socio-cultural dynamics as much as by systems and resources. Participants described hierarchical professional cultures, risk aversion, and "change fatigue" after successive waves of reform (digital tools, new reporting, pandemic-era changes). In this environment, ERNs are not rejected in principle; rather, they are interpreted through the lens of workload and perceived added bureaucracy. ERN literacy was uneven: junior staff with international exposure were described as more comfortable with cross-border consultation and secure sharing practices, while some senior clinicians were more cautious about external scrutiny, reputational risk, and the possibility of being judged against larger systems.

Continuing education priorities were contested. Ministry-oriented participants raised the opportunity cost of allocating training time to rare diseases versus high-incidence conditions; the NGO and clinical viewpoints emphasised the need for generalists to develop diagnostic suspicion early enough to trigger referral pathways. Micro-credentialing emerged as a compromise: short, stackable modules that yield CPD credit

without expanding lecture time, enabling broad-based competence without displacing core curricula.

Participants identified three levers for cultural change: experiential learning (e.g., secondments or sustained exposure to ERN hubs), institutional recognition (counting ERN board participation within career progression or scholarly contribution), and governance frameworks that permit appropriate cross-border communication while preserving accountability.

4.6 Theme 6: Patient-Centred Equity, Access, and Engagement Considerations in ERN Implementation

Participants highlighted that patients often remain peripheral to governance despite policy commitments to co-design. The NGO perspective emphasised that ERN-linked trials and specialist consultations frequently require travel and accommodation costs borne by families, creating inequity in participation. National reimbursement was described as more robust for approved treatments than for pre-approval trial phases, shifting risk and cost onto households and limiting Malta's inclusion in research pipelines.

Small-state procedures further shape equity. Participants described delays in exporting samples due to licensing, courier arrangements, and administrative steps that can exceed assay stability windows, excluding patients from time-sensitive diagnostics and sub-studies. Digital divides also appeared: older patients may rely on relatives for document handling and consent processes, raising privacy concerns and potentially reducing informed autonomy.

Policy participants proposed patient-facing portals leveraging Malta's high eID coverage to support referral tracking, secure messaging, and plain-language information. Clinicians stressed that digital tools must complement counselling, particularly for sensitive findings. Proposed interventions included travel bursaries, dynamic e-consent with readability-checked summaries, specialised translation resources fit for technical language, and patient navigation roles embedded in ERN coordination.

4.7 Theme 7: Consolidating External Partnerships in a Post-Brexit Landscape by Leveraging Malta's ERN Agreements toward Sustainable Integration and Operational Maturity

Participants described Malta's long-standing reliance on UK referral pathways as highly functional historically but increasingly frictional post-Brexit due to administrative complexity and compliance demands. This shift reframed ERNs as a strategic diversification mechanism rather than an optional enhancement. Policymakers described dependence on a single external partner as a systemic vulnerability; diversification across European centres was seen as a resilience strategy. Clinicians acknowledged the value of diversification but stressed that it increases administrative workload when multiple regulatory templates and contractual requirements must be reconciled.

Participants also pointed to knowledge-transfer opportunities: partnerships with ERN hubs could provide scalable learning through e-learning and structured exchange rather than costly sabbaticals. Procurement vulnerability—especially for ultra-orphan drugs—was discussed as another area where joint purchasing and ERN-linked mechanisms could restore bargaining power, though participants warned against exchanging one dependency for another without strengthening domestic diagnostic and pharmacovigilance capacity.

A dual strategy emerged: maximise ERN utilisation in the short term to buffer Brexit disruptions, while investing in domestic diagnostic capability (sequencing, bioinformatics, clinician-scientist development) to evolve Malta into a value-adding collaborator. Integrated governance was repeatedly identified as the missing mechanism to align these objectives with EU funding instruments.

4.8 Comparison with the Literature

The findings align with established literature identifying governance fragmentation and unclear accountability as persistent barriers to ERN participation and wider health-system integration (Hollak et al., 2016; Myhre et al., 2021). The digital readiness problems described by participants—fragmented systems, limited interoperability, and inconsistent data pathways—echo broader evidence on interoperability gaps and under-developed health information infrastructures across member states (dos Santos Vieira et al., 2022; Raycheva et al., 2024). Workforce strain and the lack of formal recognition for ERN activity map onto well-established healthcare burnout and organisational stress findings (Adriaenssens et al., 2015; Talarico et al., 2020; Rabin et al., 2023). Likewise, the operationalisation challenges of GDPR and patient privacy in cross-border collaboration

reflect ongoing debates in health data governance (Marelli et al., 2020; von Ditfurth & Lienemann, 2022).

Beyond convergence, this study contributes by offering micro-state-specific mechanisms rather than generic diagnoses. The empirical data foregrounds practical reform instruments—centralised authority for ERN governance, tiered consent models linked to enabling IT capabilities, workforce redesign through protected ERN roles, and equity measures such as navigation officers and travel support—extending normative calls for "better governance" into operational designs (De Santis et al., 2019; Panteli et al., 2023). The post-Brexit framing also adds a strategic dimension: ERNs function not only as clinical collaboration networks but as resilience infrastructure for small states recalibrating external dependency after geopolitical disruption.

5 Conclusion

The findings of this study underscore the need for targeted and strategic reforms to address the structural, regulatory, and cultural barriers obstructing Malta's full integration into the ERNs. Based on participant insights and international best practices, the following recommendations are proposed.

First, the Ministry for Health and Active Ageing should establish a national ERN Coordination Office led by an ERN Commissioner with consolidated authority over finance, data governance, and clinical oversight. This would address the fragmented governance model currently impeding progress. The model draws on the Nordic rare disease councils, such as in Finland, where patient organisations and public health officials share oversight authority to ensure both operational efficiency and participatory accountability (Helén et al., 2024). Such hybrid models have proven effective in aligning rare disease governance with EU objectives while maintaining local responsiveness.

Second, Mater Dei Hospital should formally embed ERN-related responsibilities into clinical job plans by allocating protected time for ERN engagement. In the UK, NHS consultants working in research-intensive or cross-border roles are allocated protected academic or service-improvement time within their contracts (Gideon, 2017). Participants in this study reported that ERN duties are currently treated as informal tasks. Adopting the

UK's protected-time model would professionalise ERN participation and reduce burnout risks by integrating these tasks into clinicians' official rosters.

Third, the Office of the Information and Data Protection Commissioner (IDPC), in collaboration with the DHIR, should implement a dynamic, tiered consent system integrated with Malta's national eID platform. A suitable model is presented by the eHealthBioR framework, developed in Cyprus, which enables citizens to provide, monitor, and withdraw consent for the use of their health and biobank data in real-time. This system empowers individuals by providing them with granular control over which research studies can access their data, in line with GDPR principles and EU ethical guidelines (Antoniades et al., 2021). The framework enables longitudinal clinical research while safeguarding privacy and improving administrative efficiency. Adopting a similar model would reduce the burden of repeated DPIAs while enhancing transparency and fostering citizen engagement in rare disease research linked to ERNs.

Fourth, the Ministry for Health and Active Ageing should establish a structured registrar secondment scheme in cooperation with EU-level initiatives under the ERNs. Rather than relying on informal mobility or isolated fellowships, Malta can participate in multilateral partnerships for cross-border workforce development, as recommended by Kroezen et al. (2016). Their study outlines how structured cooperation, including coordinated fellowships, mentorship networks, and multilateral staff exchanges, can enhance access to highly specialised training opportunities that small countries cannot provide alone. By aligning with EU-level frameworks and building on the experiences of countries with limited domestic sub-specialisation capacity, Malta can create a sustainable pathway for junior doctors to gain ERN-relevant expertise abroad and reintegrate into the local system. This approach strengthens national health system resilience and complements ERN participation by closing local skills gaps without permanent emigration.

Finally, the Ministry for Health and Active Ageing should adopt a national governance model for ERN engagement that mirrors Ireland's coordinated approach following Brexit. Following the UK's withdrawal from the EU, Ireland recognised the vulnerability of its historical dependence on UK-based specialist care, particularly for patients with rare diseases. In response, it strengthened its integration with ERNs through the establishment of the National Rare Diseases Office, which operates under the Health Service Executive

(Ward et al., 2022) as a central point of contact for ERN activities, clinical coordination, and policy development. This move allowed Ireland to shift from bilateral dependency to multilateral EU engagement, ensuring more sustainable and autonomous access to highly specialised healthcare (Ward et al., 2022). For Malta, the absence of a dedicated ERN office and a formal national plan to coordinate cross-border care for rare diseases creates a fragmented and reactive system vulnerable to external disruptions. Adopting a governance model similar to Ireland's would enhance Malta's capacity to navigate post-Brexit uncertainty, reduce administrative ambiguity in ERN referrals, and provide long-term policy continuity. This model would also position Malta to better align with EU health policy and funding frameworks, while improving patient access to coordinated, cross-border specialist care through ERNs.

References

1. Adachi, T., El-Hattab, A. W., Jain, R., Nogales Crespo, K. A., Quirland Lazo, C. I., Scarpa, M., Summar, M., & Wattanasirichaigoon, D. (2023). Enhancing Equitable Access to Rare Disease Diagnosis and Treatment around the World: A Review of Evidence, Policies, and Challenges. International Journal of Environmental Research and Public Health, 20(6), 4732. https://doi.org/10.3390/ijerph20064732

2. Adil, M., Khan, M. K., Kumar, N., Attique, M., Farouk, A., Guizani, M., & Jin, Z. (2024). Healthcare Internet of Things: Security Threats, Challenges, and Future Research Directions. IEEE Internet of Things Journal, 11(11), 19046–19069. https://doi.org/10.1109/jiot.2024.3360289

3. Adriaenssens, J., De Gucht, V., & Maes, S. (2015). Determinants and prevalence of burnout in emergency nurses: A systematic review of 25 years of research. International Journal of Nursing Studies, 52(2), 649–661. https://doi.org/10.1016/j.ijnurstu.2014.11.004

4. Antoniades, A., Papaioannou, M., Malatras, A., Papagregoriou, G., Müller, H., Holub, P., Deltas, C., & Schizas, C. N. (2021). Integration of Biobanks in National eHealth Ecosystems Facilitating Long-Term Longitudinal Clinical-Omics Studies and Citizens' Engagement in Research Through eHealthBioR. Frontiers in Digital Health, 3. https://doi.org/10.3389/fdgth.2021.628646

5. Blay, J.-Y., Casali, P., Bouvier, C., Dehais, C., Galloway, I., Gietema, J., Halámková, J., Hindi, N., Idbaih, A., Kinloch, E., Klümpen, H.-J., Kolarova, T., Kopeckova, K., Lovey, J., Magalhaes, M., Oselin, K., Piperno-Neumann, S., Ravnsbaek, A., Rogasik, M., & Safwat, A. (2021). European Reference Network for rare adult solid cancers, statement and integration to health care systems of member states: a position paper of the ERN EURACAN. ESMO Open, 6(4), 100174. https://doi.org/10.1016/j.esmoop.2021.100174

6. Chapman, A., Hadfield, M., & Chapman, C. (2015). Qualitative research in healthcare: An introduction to grounded theory using thematic analysis. Journal of the Royal College of Physicians of Edinburgh, 45(3), 201–205. https://doi.org/10.4997/jrcpe.2015.305

7. Christou, P. A. (2022). How to Use Thematic Analysis in Qualitative Research. Journal of Qualitative Research in Tourism, 3(2), 79–95. https://doi.org/10.4337/jqrt.2023.0006

8. De Santis, M., Hervas, C., Weinman, A., Bosi, G., & Bottarelli, V. (2019). Patient empowerment of people living with rare diseases. Its contribution to sustainable and resilient healthcare systems. Ann Ist Super Sanità, 55(3), 283–291. https://doi.org/10.4415/ANN_19_03_15

9. Directorate for Health Information and Research. (2024, September 27). Rare Diseases - The Department. Directorate for Health Information and Research. https://dhir.gov.mt/en/registries/rare-diseases/

10. dos Santos Vieira, B., Bernabé, C. H., Zhang, S., Abaza, H., Benis, N., Cámara, A., Cornet, R., Cornec, L., Peter, Schaefer, F., van, Swertz, M. A., Wilkinson, M. D., Jacobsen, A., & Roos, M. (2022). Towards FAIRification of sensitive and fragmented rare disease patient data: challenges and solutions in European reference network registries. Orphanet Journal of Rare Diseases, 17(1). https://doi.org/10.1186/s13023-022-02558-5

11. Etikan, I., & Bala, K. (2017). Sampling and Sampling Methods. Biometrics & Biostatistics International Journal, 5(6), 215–217. https://doi.org/10.15406/bbij.2017.05.00149

12. European Commission. (2024). European Reference Networks. Public Health. https://health.ec.europa.eu/rare-diseases-and-european-reference-networks/european-reference-networks_en#the-24-european-reference-networks

13. Evangelista, T., Hedley, V., Atalaia, A., Johnson, M., Lynn, S., Le Cam, Y., & Bushby, K. (2016). The context for the thematic grouping of rare diseases to facilitate the establishment of European Reference Networks. Orphanet Journal of Rare Diseases, 11(1). https://doi.org/10.1186/s13023-016-0398-y

14. Farrugia, C. (2017, March 3). Mater Dei to join European networks later on this year. Times of Malta. https://timesofmalta.com/article/mater-dei-to-join-european-networks-later-on-this-year.641294

15. Gideon, A. (2017). The Structure of Research Funding in Germany, the Netherlands and England (UK). In T.M.C. Asser Press eBooks (pp. 115–167). https://doi.org/10.1007/978-94-6265-168-5_4

16. Hamilton, A. B., & Finley, E. P. (2019). Reprint of: Qualitative Methods in Implementation research: an Introduction. Psychiatry Research, 283, 112629. https://doi.org/10.1016/j.psychres.2019.112629

17. Hedley, V., Bolz-Johnson, M., Hernando, I., Kenward, R., Nabbout, R., Romero, C., Schaefer, F., Upadhyaya, S., Arzimanoglou, A., Dollfus, H., Leroux, D., Scarpa, M., Schaefer, F., Verloes, A., Bolz-Johnson, M., Daria Julkowska, Rath, A., Hedley, V., Ussi, A., & Mimouni, Y. (2023). Together4RD position statement on collaboration between European reference networks and industry. Orphanet Journal of Rare Diseases, 18(1). https://doi.org/10.1186/s13023-023-02853-9

18. Hedley, V., Bottarelli, V., Weinman, A., & Taruscio, D. (2021). Shaping national plans and strategies for rare diseases in Europe: past, present, and future. Journal of Community Genetics, 12(2), 207–216. https://doi.org/10.1007/s12687-021-00525-4

19. Helén, I., Snell, K., Tarkkala, H., & Tupasela, A. (2024). Genome Finland: From rare diseases to data economy. Helsinki University Press.

20. Héon-Klin, V. (2017). European Reference networks for rare diseases: what is the conceptual framework? Orphanet Journal of Rare Diseases, 12(1). https://doi.org/10.1186/s13023-017-0676-3

21. Hollak, C. E. M., Biegstraaten, M., Baumgartner, M. R., Belmatoug, N., Bembi, B., Bosch, A., Brouwers, M., Dekker, H., Dobbelaere, D., Engelen, M., Groenendijk, M. C., Lachmann, R., Langendonk, J. G., Langeveld, M., Linthorst, G., Morava, E., Poll-The, B. T., Rahman, S., Rubio-Gozalbo, M. E., & Spiekerkoetter, U. (2016). Position statement on the role of healthcare professionals, patient organizations and industry in European Reference Networks. Orphanet Journal of Rare Diseases, 11(1). https://doi.org/10.1186/s13023-016-0383-5

22. Horgan, D., Bernini, C., Thomas, P. M., & Morre, S. A. (2019). Cooperating on Data: The Missing Element in Bringing Real Innovation to Europe's Healthcare Systems. Public Health Genomics, 22(3-4), 77–101. https://doi.org/10.1159/000503296

23. Horgan, D., Metspalu, A., Ouillade, M.-C., Athanasiou, D., Pasi, J., Adjali, O., Harrison, P., Hermans, C., Codacci-Pisanelli, G., Koeva, J., Szucs, T., Cursaru, V., Belina, I., Bernini, C., Zhuang, S., McMahon, S., Toncheva, D., & Thum, T. (2020). Propelling Healthcare with Advanced Therapy Medicinal Products: A Policy Discussion. Biomedicine Hub, 5(3), 1–23. https://doi.org/10.1159/000511678

24. Iotova, V., Schalin-Jäntti, C., Bruegmann, P., Broesamle, M., Bratina, N., Tillmann, V., Hiort, O., & Pereira, A. M. (2021). Educational and knowledge gaps within the European reference network on rare endocrine conditions. Endocrine Connections, 10(1), 37–44. https://doi.org/10.1530/ec-20-0480

25. Kozlowski, S. W. J., Chao, G. T., Grand, J. A., Braun, M. T., & Kuljanin, G. (2013). Advancing Multilevel Research Design. Organizational Research Methods, 16(4), 581–615. https://doi.org/10.1177/1094428113493119

26. Kroezen, M., Buchan, J., Dussault, G., Glinos, I., & Wismar, M. (2016). Policy Brief 20 - How can structured cooperation between countries address health workforce challenges related to highly specialized health care? Improving access to services through voluntary cooperation in the EU.

27. Marelli, L., Lievevrouw, E., & Van Hoyweghen, I. (2020). Fit for purpose? The GDPR and the governance of European digital health. Policy Studies, 41(5), 1–21. https://doi.org/10.1080/01442872.2020.1724929

28. Ministry for Health and Active Aging. (2024, April 29). European Reference Networks - crossborderhealthcaremalta. Crossborderhealthcaremalta. https://crossborderhealth.gov.mt/european-reference-networks/

29. Moliner, A. M., & Waligora, J. (2017). The European Union Policy in the Field of Rare Diseases. Advances in Experimental Medicine and Biology, Chapter 30, 561–587. https://doi.org/10.1007/978-3-319-67144-4_30

30. Morgan, D. L., & Nica, A. (2020). Iterative Thematic Inquiry: A New Method for Analyzing Qualitative Data. International Journal of Qualitative Methods, 19, 1–11. https://doi.org/10.1177/1609406920955118

31. Myhre, S. L., French, S. D., & Bergh, A. (2021). National public health institutes: A scoping review. Global Public Health, 17(6), 1–18. https://doi.org/10.1080/17441692.2021.1910966

32. Naeem, M., Ozuem, W., Howell, K. E., & Ranfagni, S. (2023). A Step-by-Step Process of Thematic Analysis to Develop a Conceptual Model in Qualitative Research. International Journal of Qualitative Methods, 22(1), 1–18. Sagepub. https://doi.org/10.1177/16094069231205789

33. Nifakos, S., Chandramouli, K., Nikolaou, C. K., Papachristou, P., Koch, S., Panaousis, E., & Bonacina, S. (2021). Influence of Human Factors on Cyber Security within Healthcare Organisations: A Systematic Review. Sensors, 21(15), 5119. https://doi.org/10.3390/s21155119

34. Nori, M., Fisher-Vance, D., Wuerth, L., Colenso, R., & Donovan, D. J. (2022). The global role of patients, advocates and caregivers in rare diseases. Future Rare Diseases, 2(2). https://doi.org/10.2217/frd-2022-0003

35. Nyumba, T., Wilson, K., Derrick, C. J., & Mukherjee, N. (2018). The Use of Focus Group Discussion methodology: Insights from Two Decades of Application in

Conservation. Methods in Ecology and Evolution, 9(1), 20–32. Wiley. https://doi.org/10.1111/2041-210X.12860

36. Panteli, D., Polin, K., Webb, E., Allin, S., Barnes, A., Degelsegger-Márquez, A., Ghafur, S., Jamieson, M., Kim, Y., Litvinova, Y., Nimptsch, U., Parkkinen, M., Rasmussen, A., Reichebner, C., Röttger, J., Rumball-Smith, J., Scarpetti, G., Seidler, A., Seppänen, J., & Smith, M. (2023). Health and Care Data. Health Systems in Transition, 25(2). ISSN 1817-6127.

37. Qu, S. Q., & Dumay, J. (2011). The qualitative research interview. Qualitative Research in Accounting & Management, 8(3), 238–264. https://doi.org/10.1108/11766091111162070

38. Rabin, S., Kika, N., Lamb, D., Murphy, D., Stevelink, S., Williamson, V., Wessely, S., & Greenberg, N. (2023). Moral Injuries in Healthcare Workers: What Causes Them and What to Do About Them? Journal of Healthcare Leadership, 2023(15), 153–160. https://doi.org/10.2147/jhl.s396659

39. Rädiker, S., & Kuckartz, U. (2020). MAXQDA PRESS Step by Step Focused Analysis of Qualitative Interviews with MAXQDA. https://kurs.uni-jena.de/pluginfile.php/36257/mod_folder/content/0/978-3-948768072-1.pdf

40. Raycheva, R., Kostadinov, K., Mitova, E., Iskrov, G., Stefanov, G., Vakevainen, M., Elomaa, K., Man, Y.-S., Gross, E., Zschüntzsch, J., Röttger, R., & Stefanov, R. (2024). Landscape analysis of available European data sources amenable for machine learning and recommendations on usability for rare diseases screening. Orphanet Journal of Rare Diseases, 19(1). https://doi.org/10.1186/s13023-024-03162-5

41. Saunders, M., Lewis, P., & Thornhill, A. (2023). Research Methods for Business students. (9th ed.). Pearson.

42. Shishkov, S. R., Tuccillo, L., Iotova, V. M., Pivonello, R., Pelsma, I. C., Pereira, A. M., & Biermasz, N. R. (2023). Mapping of the current transition of care practice for patients with pituitary disease at Endo-ERN reference centers. Endocrine Connections, 12(2). https://doi.org/10.1530/ec-22-0308

43. Star, J., Ringaert, L., & Larcombe, L. (2025). Qualitative Methods Case Study: Using MAXQDA in Indigenous HIV Journey Mapping Research. International Journal of Qualitative Methods, 24. https://doi.org/10.1177/16094069251356667

44. Stevenson, H. (2019). The European Semester: A literature review Published as part of the project: Public service trade unions -effective intervention in the European Semester Financed by the European Commission. https://www.epsu.org/sites/default/files/article/files/Literature%20review.pdf

45. Talarico, R., Aguilera, S., Alexander, T., Amoura, Z., Andersen, J., Arnaud, L., Avcin, T., Marsal Barril, S., Beretta, L., Bombardieri, S., Bortoluzzi, A., Bouillot, C., Bulina, I., Burmester, G. R., Cannizzo, S., Cavagna, L., Chaigne, B., Cornet, A., Corti, P., & Costedoat-Chalumeau, N. (2022). The added value of a European Reference Network on rare and complex connective tissue and musculoskeletal diseases: insights after the first 5 years of the ERN ReCONNET. Clinical and Experimental Rheumatology, 40(5), 3–11. https://doi.org/10.55563/clinexprheumatol/d2qz38

46. Talarico, R., Cannizzo, S., Lorenzoni, V., Marinello, D., Palla, I., Pirri, S., Ticciati, S., Trieste, L., Triulzi, I., Terol, E., Bucher, A., & Turchetti, G. (2020). RarERN Path: a methodology towards the optimisation of patients' care pathways in rare and complex diseases developed within the European Reference Networks. Orphanet Journal of Rare Diseases, 15(1). https://doi.org/10.1186/s13023-020-01631-1

47. Tumiene, B., & Graessner, H. (2021). Rare disease care pathways in the EU: from odysseys and labyrinths towards highways. Journal of Community Genetics, 12(2), 231–239. https://doi.org/10.1007/s12687-021-00520-9

48. Tumiene, B., Graessner, H., Mathijssen, I. M., Pereira, A. M., Schaefer, F., Scarpa, M., Blay, J.-Y., Dollfus, H., & Hoogerbrugge, N. (2021). European Reference Networks: challenges and opportunities. Journal of Community Genetics, 12(2), 217–229. https://doi.org/10.1007/s12687-021-00521-8

49. Van Der Walt, J. (2020). Interpretivism-Constructivism as a Research Method in the Humanities and Social Sciences -More to It Than Meets the Eye. International Journal of Philosophy, 8(1), 2333–5769. https://doi.org/10.15640/ijpt.v8n1a5

50. Vandevelde, N. M. (2019). Strategies to improve the quality of reference networks for rare diseases. Translational Science of Rare Diseases, 5(1-2), 1–22. https://doi.org/10.3233/trd-190032

51. Vassolo, R., Mac Cawley, A., Tortorella, G. L., Fogliatto, F. S., Tlapa Mendoza, D., & Narayanamurthy, G. (2021). Hospital Investments Decisions in Healthcare 4.0 Technologies: Challenges, Trends, and Research Directions (Preprint). Journal of Medical Internet Research, 23(8). https://doi.org/10.2196/27571

52. Verma, A., & Gurtoo, A. (2023). Evaluating global data policies around non-personal data on social and public good. Digital Policy, Regulation and Governance, 26(1), 72–94. https://doi.org/10.1108/dprg-03-2023-0044

53. von Ditfurth, L., & Lienemann, G. (2022). The Data Governance Act: – Promoting or Restricting Data Intermediaries? Competition and Regulation in Network Industries, 23(4), 270–295. https://doi.org/10.1177/17835917221141324

54. Ward, A. J., Murphy, D., Marron, R., McGrath, V., Bolz-Johnson, M., Cullen, W., Daly, A., Hardiman, O., Lawlor, A., Lynch, S. A., MacLachlan, M., McBrien, J., Ni Bhriain, S., O'Byrne, J. J., O'Connell, S. M., Turner, J., & Treacy, E. P. (2022). Designing rare disease care pathways in the Republic of Ireland: a co-operative model. Orphanet Journal of Rare Diseases, 17(1). https://doi.org/10.1186/s13023-022-02309-6