output a powerpoint file with the following outline and change all of the references to use Vancouver referencing format: Presentation Layout and Script Slide 1: Title Slide

Visual: A clean, professional title slide. Use the Queen's University Belfast logo in a corner. The main visual could be an abstract graphic representing the intersection of digital data and human health (e.g., a DNA helix merging with binary code).

Text on Slide:

Title: Challenges and Opportunities in Designing, Delivering, and Evaluating Digital Innovations for Rare Conditions

Your Name: Joshua

Event: PhD Interview

Date: 22nd October 2025

What to Say (Script):

"Good morning. Thank you for the opportunity to be here today. My name is Joshua, and this morning I'd like to discuss the challenges and opportunities I anticipate in the complex but vital field of digital innovation for rare conditions."

Slide 2: The Rare Disease Paradox

Visual: A split-screen visual. On one side, a single, silhouetted person representing individual rarity. On the other side, a collage of many diverse faces, representing the large collective population. An arrow connecting them labelled "The Diagnostic Odyssey".

Text on Slide:

Headline: The Rare Disease Paradox: Individually Rare, Collectively Common

Over 7,000 rare conditions affect millions in the UK.

The "Diagnostic Odyssey": A protracted journey to an answer.

UK Framework Priorities: Faster Diagnosis, Increased Awareness, Coordinated Care.

Digital innovation is positioned as the critical enabler.

What to Say (Script):

"The landscape of rare conditions is defined by a fundamental paradox. While each of the 7,000-plus conditions is, by definition, rare, together they affect millions of people across the UK. For these individuals, the path to a diagnosis is often a long and frustrating journey, aptly termed the ‘diagnostic odyssey’.

"Recognising this, the UK Rare Diseases Framework has set clear national priorities: to speed up diagnosis, improve awareness among healthcare professionals, and better coordinate care. In this context, digital innovation isn't just an interesting development; it's positioned as the critical enabler to turn these strategic goals into a reality for patients."

Slide 3: The Core Opportunity: A Paradigm Shift to Integrated Intelligence

Visual: A diagram showing fragmented data silos (GP records, hospital notes, genomic data, lab results) transforming into a single, unified circle labelled "Integrated Patient Record". From this circle, arrows point outwards to "Faster Diagnosis," "Personalised Medicine," and "Accelerated Research."

Text on Slide:

Headline: From Data Silos to Integrated Intelligence

The Foundation: Unified digital infrastructures like Northern Ireland's encompass programme.

The Power: Integrating clinical data with multi-omic research (genomics, epigenetics).

The Vision: Creating a 'Learning Health System' where every clinical interaction informs research and improves care.

This aligns directly with national strategies like Genome UK.

What to Say (Script):

"The single greatest opportunity before us is a paradigm shift away from fragmented data towards integrated intelligence. For too long, a patient's story has been scattered across disconnected systems.

"Here in Northern Ireland, the rollout of the encompass programme is a landmark step, creating a single digital care record for every citizen. This provides a foundational clinical data backbone.

"But the true potential is unlocked when we integrate this clinical data with the rich multi-omic data streams being pioneered here at Queen's—combining genomics, epigenetics, and clinical information to build a complete picture.

"This creates the blueprint for a true 'learning health system'—one that learns from every patient interaction to accelerate research and deliver more personalised medicine, fulfilling the vision set out in national strategies like Genome UK."

Slide 4: A Framework for Progress: The Tripartite Challenge

Visual: A Venn diagram with three overlapping circles. Each circle is labelled with one of the core challenges. The overlapping centre is labelled "Sustainable & Equitable Innovation."

Text on Slide:

Headline: Navigating a Tripartite Challenge

Realising the potential of digital innovation requires balancing three core, interconnected tensions:

Scale vs. Specificity: Reconciling 'big data' needs with individual patient complexity.

Potential vs. Reality: Bridging technological promise with clinical and patient adoption.

Collaboration vs. Confidentiality: Balancing open data sharing with the ethical imperative of privacy.

What to Say (Script):

"However, this technological promise is not self-fulfilling. Its realisation depends on navigating what I call a tripartite challenge. Progress requires us to constantly balance three core, interconnected tensions.

"First, the tension between the immense scale required by data-driven technologies and the profound specificity of an individual patient.

"Second, the gap between the theoretical potential of a new tool and the complex reality of its adoption by clinicians and patients.

"And third, the ethical tightrope we must walk between the scientific need for open collaboration and the non-negotiable need for patient confidentiality. These are not separate hurdles; they are interwoven facets of a single problem."

Slide 5: Challenge I: Reconciling Algorithmic Scale with Human Specificity

Visual: An icon of a large database with an arrow pointing to a single human figure, with a "gap" or "mismatch" symbol between them.

Text on Slide:

Headline: The 'Big Data' Problem in a 'Small Data' World

The Mismatch: AI and Machine Learning are trained on vast datasets; rare disease is inherently 'small data'.

The Interoperability Gap: Genomic data is complex, vast, and probabilistic. EHRs like encompass are designed for structured, deterministic clinical data.

The Missing Piece: Structured data often fails to capture the patient's crucial 'lived experience'.

What to Say (Script):

"The first challenge is technical. Technologies like machine learning thrive on 'big data', yet the world of rare disease is one of 'small data'—characterised by limited patient numbers and significant heterogeneity. This makes traditional clinical trial models difficult and demands innovative analytical approaches.

"Furthermore, there's a fundamental interoperability gap. Genomic data is massive, complex, and probabilistic. Our electronic health records, by contrast, are built to handle structured, deterministic information like diagnoses and prescriptions. Forcing one into the other is a formidable challenge that current systems are ill-equipped for.

"Crucially, even the most complete dataset often misses the most important information: the patient's lived experience, which is vital for creating solutions that are truly meaningful."

Slide 6: Challenge II: Bridging Technological Potential with Clinical Reality

Visual: A simple two-panel cartoon. Panel 1: A developer proudly holds a complex, shiny app. Panel 2: A doctor looks overwhelmed by paperwork, and a patient looks confused at their phone. An arrow between them is labelled "The Adoption Gap".

Text on Slide:

Headline: A Socio-Technical Problem, Not Just a Technical One

The Patient Divide: Barriers include digital literacy, access, and lack of user involvement in design.

Solution: Community co-design and human-centred principles.

The Clinician Divide: Barriers include workflow disruption, administrative burden, and lack of training.

Solution: Focus on change management and seamless integration.

What to Say (Script):

"The second challenge is socio-technical. A perfect algorithm is useless if it's not used. For patients, a 'digital divide' exists, created by barriers like low digital literacy or tools that aren't designed with their input. The solution here is a move towards community co-design, involving users from the very beginning.

"For clinicians, the barriers are just as significant. They operate in high-pressure environments, and will resist tools that disrupt workflows or add to their administrative burden. Successful implementation is therefore a change management problem, not just a technology deployment problem. We must ask not only 'does it work?', but 'how does it fit into the work of the clinic and the life of the patient?'"

Slide 7: Challenge III: Balancing Open Collaboration with Patient Confidentiality

Visual: A graphic of a globe made of connected data points, with a padlock and shield symbol at its centre.

Text on Slide:

Headline: The Ethical Tightrope of Genomic Data

The Need: Global data sharing is essential to achieve the statistical power needed for rare disease research.

The Risk: Genomic data is uniquely and permanently identifiable. It can never be truly anonymised.

The Governance: Moving beyond technical fixes to new social contracts.

Trusted Research Environments (TREs): Analyse data in-situ.

Dynamic Consent: An ongoing process, not a one-time event.

What to Say (Script):

"The final challenge is profoundly ethical. Progress in rare disease research is uniquely dependent on pooling data globally. Yet, the very data we need to share—a person's genome—is also uniquely identifiable. Unlike other medical data, it can never be truly anonymised, and it contains sensitive information about a patient's entire family, often without their consent.

"This creates a deep tension. The path forward lies in robust governance. This includes using Trusted Research Environments, where data is analysed securely in one place without being moved. It also means evolving our model of consent from a one-time event to a dynamic, ongoing process, empowering patients as active partners in research. Ultimately, earning and maintaining public trust is the bedrock of all future progress."

Slide 8: Conclusion & A Trajectory for Future Research

Visual: A simple roadmap graphic with a starting point ("Today's Challenges") and an endpoint ("Integrated, Equitable Systems"). Key milestones along the road are labelled "Novel Analytics," "Co-Design," and "Ethical Governance."

Text on Slide:

Headline: The Path Forward: From Tools to Systems

The goal is not to build better tools, but to architect better integrated socio-technical systems.

This requires an interdisciplinary approach, combining:

Novel Analytics for small, complex data.

Socio-technical Design principles like co-creation.

Robust Governance to build and maintain trust.

A PhD project could make a significant contribution by tackling a piece of this puzzle, for example, by developing and validating a framework for co-designing tools to capture the patient's lived experience.

What to Say (Script):

"In conclusion, the challenge is to move beyond creating isolated digital tools and instead focus on architecting integrated socio-technical systems.

"This requires a truly interdisciplinary approach, one that develops novel analytical methods for small data, embeds the principles of co-design at its core, and is built upon a robust foundation of ethical governance.

"A prospective PhD project, such as the one on offer here, is perfectly positioned to make a significant contribution by tackling a critical piece of this complex puzzle. For instance, focusing on how we can develop and validate a framework for co-designing digital tools that effectively capture and integrate the patient's lived experience, thereby bridging that critical gap between algorithmic potential and clinical reality.

"By addressing these fundamental challenges, we can help translate the immense promise of digital innovation into tangible, equitable, and lasting benefits for the entire rare disease community."

Slide 9: Thank You & Questions

Visual: A clean, simple slide.

Text on Slide:

Thank You

Questions?

What to Say (Script):

"Thank you for your time and attention. I would be delighted to answer any questions you may have."

Slide 10: References

Visual: A standard, text-based reference list. Use a small font size to fit everything on one slide.

Text on Slide:

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