# What role does identification play to support Metabolic Dieticians and Scientists for care and research of patients with Phenylketonuria (PKU) - An information modelling perspective

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A dissertation submitted to the University of Dublin,

in partial fulfilment of the requirements for the degree of

Master of Science in Health Informatics

# Declaration

I declare that the work described in this dissertation is, except where otherwise stated, entirely my own work, and has not been submitted as an exercise for a degree at this or any other university.

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Frances Gibbons

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## Acknowledgements

This dissertation would not have been possible without the support of my family, friends, classmates, and supervisor. I will be forever indebted and grateful to the following people:

- My supervisor Prof Gaye Stephens – I appreciate the continued guidance and support during the dissertation process. Personally, and professionally your words made an impact that will never be forgotten. Your passion and work ethic is something to admire. Thank you for your teaching during the two years. I look forward to working with you in the future.
- Supporting supervisor Mr Damon Berry – Your passion, enthusiasm, and support helped me greatly. I look forward to our paths crossing and working with you in the future.
- Lucy Hederman – Thank you for your support. Your teaching and guidance are appreciated.
- Mr. Peter Gaffney – From the start of the MSc journey, your support and willingness to share your experience from years of service in the healthcare arena were greatly appreciated and encouraged me.
- Mrs. Deirdre Deverell – You are my mentor. Your support personally and professionally went above and beyond. I will be forever thankful to you. Our friendship will continue for many years.
- Ms. Ulfat Alramadhi – Thank you for your friendship, support, and guidance. Thank you for the spelling and grammar checks. The impact has been beyond what you might think. Your experience has been invaluable. I will be forever indebted to you.
- Mrs. Geraldine Elebert – Thank you for your friendship, support, and guidance. It has made a huge impact on my life, especially during the time of the dissertation.
- To my classmates – I learned so much from each of you. Your contributions during our two years together made my experience unforgettable. Thank you to everyone.
- My family – Thank you for your support. I love you all.

This MSc has been an important part of my journey in life. I will remember this time for the lows but more importantly the highs of completing the MSc. I have learned valuable skills that I will incorporate in future work. I hope to contribute some value to the future of Health Informatics in Ireland.

Thank you.

Frances Gibbons

## Abstract

The aim of this research is to identify the role that identification plays in supporting identified stakeholders in the care and management of patients with the disorder Phenylketonuria. The research idea has come from the challenges identified with interoperability of information in electronic health record systems as identified in the paper by Chen et al, 2015.

The motivation for this research is improved patient safety and data quality. To obtain the highest standards of patient safety, data quality needs to be improved. Interoperability of the data is integral and needs to play a major role in the eHealth strategy. Furthermore, it should be incorporated into the development plans of future integrated healthcare information systems (HIS).

The research question is 'What role does identification play to support Metabolic Dieticians and Scientists for care and research of patients with Phenylketonuria (PKU) – An information modelling perspective'. The answer will focus on three key areas. Firstly, the role of identifiers in integrated healthcare and research using an EHR. Secondly, identifier issues focusing on modelling and quality. Finally, identifying uses carried out by dieticians and scientists using information for care and research of patients with PKU. These use cases were identified by conducting focus groups using the 1-2-4-all liberating technique(McCandless, 2010). From researching these key areas, there is an anticipation that the results gathered could influence future standards for EHR communication.

Strategies were introduced which highlighted the current state and plans for health care in the future. For successful execution of an eHealth strategy, research showed it to be dependent on fundamental enablers being present. The key themes presented were integration, interoperability, and standardisation. The literature review identified a gap where the importance of identifiers should be included for future research and work.

The research process consisted of 11 stages. The research question leads the topic to be reviewed in the literature. Ethical approval was received from Temple Street Children's university hospital and Trinity College Dublin. Focus groups were held with scientists, dieticians, and health informaticians. Results from these were categorised, illustrated and tabulated to display the results. The synthesis of the literature and practical work led to the creation of personas, a UML activity diagram, and use cases.

EURO-CAS, European and international standard organisations like CEN and ISO along with national strategies like the EHR persona project from eHealth Ireland together create a standards accreditation process. The positive is that stakeholders from the National, European and International forums influence the decision making. The benefit of having a wide decision-making base is that the process is rendered more robust and complete. The practical work completed will hopefully contribute to the standards process for EHR communication.

For the successful implementation of a patient-centric eHealth system, there needs to be a shift from reactive to proactive healthcare. The research represented in this dissertation concerns itself with the role of identities in this shift, through the management of patients with Phenylketonuria (PKU).

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# Abbreviations

| | |
|---|---|
| AMET | Adult Metabolic Clinic |
| AAL | Ambient Assisted Living |
| BMS | Basic Medical Scientist |
| CIO | Chief Information Officer |
| CDW | Clinical Data Warehouse |
| CDA | Clinical Document Architecture |
| CSPD | Clinical Strategy and Programme Division |
| CAS | Conformity Assessment Scheme |
| DOH | Department of Health |
| DPLM | Department of Paediatric Laboratory Medicine |
| DBS | Dried Blood Spot |
| EHR | Electronic Health Record |
| EPI | Enterprise Person Index |
| EC | European Commission |
| ECHA | European Connected Health Alliance |
| ETHEL | European Health Telematics Association |
| EIF | European Interoperability Framework |
| EU | European Union |
| FHIR | Fast Healthcare Resources |
| FI | Fundamental Interoperability |
| GIRM | Generalised Identity Reference Model |
| HCP | Health Care Professional |
| HIQA | Health Information and Quality Exchanges |
| HIE | Health Information Exchanges |
| HL7 | Health Level Seven |
| HSE | Health Service Executive |
| HIMSS | The Health Information Systems Society |
| HIS | Healthcare Information System |
| HITCH | Healthcare Interoperability Testing and Conformance Harmonisation |
| IHI | Individual Health Identifier |
| ICT | Information and Communication Technologies |

| | |
|---|---|
| IPPOSI | Irish Platform for Patient Organisations, Science and Industry |
| LA | Laboratory Aide |
| LIS | Laboratory Information System |
| MPI | Master Patient Index |
| NCIMD | National Centre for Inherited Metabolic Disorders |
| OPD | Out Patient Department |
| PMS | Patient Management System |
| PHE | Phenylalanine |
| PKU | Phenylketonuria |
| PACS | Picture Archiving Communication System |
| RIS | Radiology Information Systems |
| SI | Semantic Interoperability |
| SOP | Standard Operating Procedure |
| TCD | Trinity College Dublin |
| TSCUH | Temple Street Children's University Hospital |

# 1 Introduction

Healthcare is a sector where the introduction of information and communication technologies (ICT) could improve efficiency, quality, and innovation. Throughout the European Union (EU) there has been a widespread commitment to eHealth, which is positive for future interoperability in eHealth systems (Health, 2012).

At the Irish Platform for Patient Organisations, Science and Industry (IPPOSI) Conference 2015, Health Service Executive (HSE) Chief Information Officer (CIO) at the time, Richard Corbridge introduced the strategy of eHealth Ireland. The main focus of the eHealth Ireland strategy is to deliver integrated care with the patient at the centre using digital solutions (IPPSOI, 2015). From a video created by IPPOSI, Yvonne Goff from HSE said "the eHealth Strategy, will allow for safer, better quality of care. Allowing silos of patient information to be brought together. Promoting health care versus sick care. Allowing care to be brought back to the community. eHealth will allow patients to stay at home and promote proactive healthcare" (IPPSOI, 2015). An example of this was discussed by Brian O'Mahony from the Irish Haemophilia Society. Brian O'Mahony described how patients were involved in the conception, for the development of the haemophilia patient mobile application. Patient interaction allows for future progression and updates based on patient needs. This process will aim for incorporating control shared decision making (IPPSOI, 2015).

Also at the IPPOSI conference, Dr. John Dinsmore from Trinity College Dublin (TCD), spoke of the primary benefit of patient centred care. "Instead of being a reactive healthcare system, we should adopt a proactive and preventative healthcare system. People should have ownership of their personal data, empowering them to look after their health. Benefits of a proactive and preventative system approach can offset the cost of health care to the healthcare system"(IPPSOI, 2015).

Derick Mitchell from IPPOSI spoke of the short-term benefits of eHealth in Ireland. "Improving efficiencies of health services and moving paper-based models to digital-based models. Long-term benefits are to improve the quality of care and patient safety.From patient perspective, eHealth will improve access to data, empower patients, allowing them to become more involved in their care"(IPPSOI, 2015).

Two key areas of the eHealth Ireland strategy that IPPOSI is focused on is the Individual Health Identifier (IHI) and Electronic Health Record (EHR). A shared vision for eHealth is required, utilising vast amounts of health data in a meaningful way.

The Health Information Management Systems Society (HIMSS) definition of EHR is "The Electronic Health Record (EHR) is a longitudinal electronic record of patient health information generated by one or more encounters in any care delivery setting. Included in this information are patient demographics, progress notes, problems, medications, vital signs, past medical history, immunisations, laboratory data and radiology reports. The EHR automates and streamlines the clinician's workflow. The EHR has the ability to generate a complete record of a clinical patient encounter – as well as supporting other care-related activities directly or indirectly via interface – including evidence-based decision support, quality management and outcomes reporting." This definition acknowledges the expansive data areas incorporated in patient health and the necessity for complete records to assist clinicians with the best possible outcomes for patients (Society, 2018).

eHealth Ireland describes a key component to the success of future healthcare delivery is a national electronic health record (EHR). The benefits of an EHR are seen to be patient-centric. eHealth Ireland lists the benefits of an EHR as improving waiting lists, having access to your own records, secure data, and healthcare professionals (HCP) working together in real-time, improving patient care. The primary focus of the EHR is for integrated care. The ability to share information across healthcare organisations is a vital part of the eHealth Ireland strategy. It will have a positive impact on patients, users, carers, social and healthcare professionals. Healthcare will be transformed to drive towards the delivery of safer personalised care (Ireland, 2018a).

In March 2018 the successor to CIO Richard Corbridge, Martin Curley commenced his term as CIO in the Health Service Executive (HSE). In May 2018, Martin launched the concept of "Stay Left, Shift Left"(Curley, 2018). From Martin's previous experience working in the semiconductor industry, the principle of Moore's law was used for innovation strategy. He aims to introduce a similar law for Irish healthcare. The 'Shift left' innovation strategy for Sláintecare is about moving treatment from acute hospitals to the community. The 'Stay Left, Shift Left' strategy will shift reactive care in hospitals to proactive and preventative care into the community and homes (Ireland, 2018b).

So, for the successful implementation of a patient-centric eHealth system, there needs to be a shift from reactive to proactive healthcare. The research represented in this dissertation concerns itself with the role of identities in this shift, through the management of patients with Phenylketonuria (PKU). Before I introduce the aims and research question, I am going to introduce the strategies and key themes of integration, interoperability, and standardisation to provide context.

## 1.1 Strategies

In Ireland, the department of health (DOH) statement of strategy 2016-2019 outlines the limitations in the current model of care. It describes the historic fragmented nature of the service. The DOH strategy reviews the development of a model that needs to be "more integrated and continuous, person-centered, and delivered at the lowest level of complexity consistent with patient safety". Integration is highlighted by the DOH as an integral objective of the eHealth Strategy (Health, 2016).

In 2013, the DOH launched its eHealth strategy for Ireland. The background information in the strategy outlined that, in order to efficiently and equitably facilitate the forecasted demand on healthcare and its systems the DOH need to realign the national healthcare budget. Increasing investment in eHealth systems and change management will aide the implementation of future eHealth plans (Health, 2016).

The eHealth strategy for Ireland document coincided with the European Union eHealth Action Plan 2012-2020 (Health, 2012). The main objectives of the EU action plan are to empower patients and healthcare providers (HCP), to integrate devices and technology and invest in research towards a personalised medicine future (Commission, 2012, ETHEL, 2017).

An important area in the HSE is integrated care. The long-term plan is to work with all stakeholders to integrate health services for all Irish citizens. The aim of integrated care is to join up health and social care services. To achieve this plan there needs to be a change in the current care process, ultimately improving quality and patient outcomes. The main organising principle of integrated care programme has patient perspective at the centre with involvement from the public, private and voluntary providers, clinicians and patient groups. It is based on patient empowerment, multi-disciplinary care, and illness prevention (HSE, 2016).

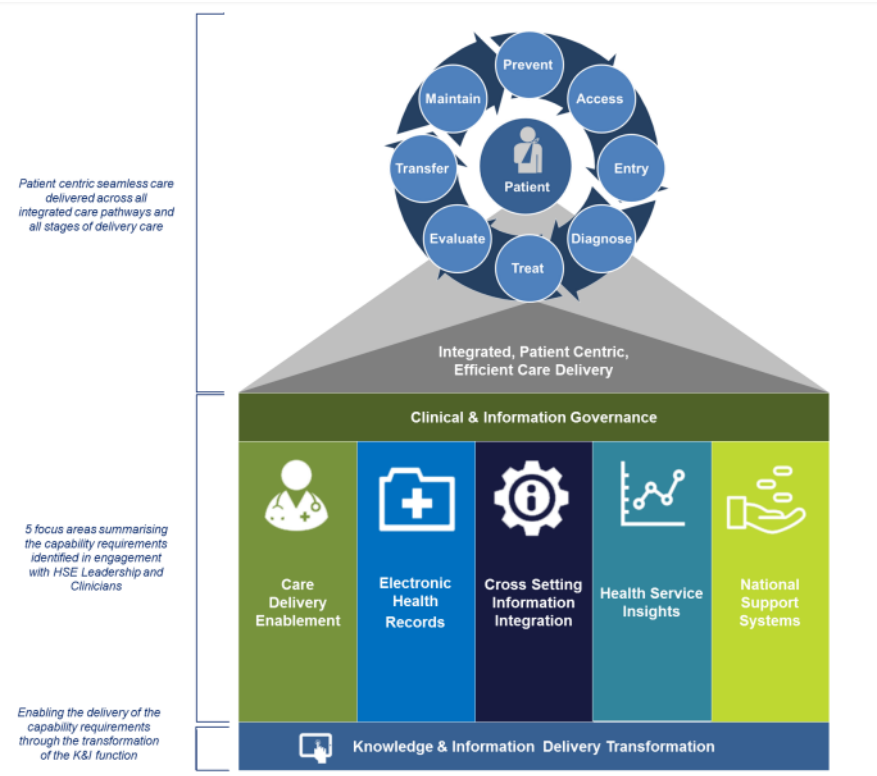



*Figure 1 Knowledge and Information plan from EHR Vision (Health, 2012)*

Figure one is extracted from the EHR vision and direction document. This document outlines the HSE's knowledge and information plan and how it will build onto the national eHealth strategy. As illustrated in figure one, the EHR is one of five focus areas identified to facilitate integrated care. Each of the five focus areas requires clinical and information governance for patient-centric care to be delivered across integrated care pathways at each stage of the delivery of care (Health, 2012).


*Figure 2 Patient-Centric Care from Knowledge Information Plan in EHR Vision (Health, 2012)*

Figure two depicts the integration of all information and knowledge sources involved in the delivery of healthcare via information technology-based systems. It includes patients at the centre. Also included are records, caregivers and their systems, monitoring devices and sensors, management and administrative functions. Figure two represents a fully integrated digital supply chain and involves high levels of automation and information sharing.

In June 2016, at the annual IPPOSI conference entitled 'Person-centred eHealth" Derick Mitchell discussed the theme of the 3 C's: Competencies, Care, and Community stating clinical and patient competencies will be key to driving true person-centred care and noting that there appeared to be a lack of shared understanding on what exactly constitutes integrated care. The roundtable and its broad range of participants and speakers served to bring together this community to highlight the key issues that must be addressed as Ireland moves towards an eHealth-enabled system of person-centred care (Ireland, 2016).

In 2016, the Clinical Strategy and Programmes Division (CSPD), in accordance with the Health Service Executive (HSE) initiated the Patient Narrative Project. The aim of the project is to enable the delivery of person centred care with improved experiences and outcomes. The HSE recognises integrated care requires the "voice of the user". The data and information

gained from patient narratives using Irish Health services will address the design and improvements of future eHealth Systems. There are three phases outlined in the patient narrative project. Phase one, led by the Irish Platform for Patient Organisation Science and Industry (IPPOSI), will find out what 'person-centred coordinated care' means to Irish patients, carers, and patient organisations. The CSPD created a process for phase two, which captured the experiences of a large number of people who require multiple health services. The aim is to influence future health services in this regard. Phase two was called YourVoiceMatters and was a short survey where users could provide information about making services better, influence decision making and improve services over time (HSE, 2017b). Finally, phase three included creating guidelines and developing tools for designers to create and improve integration between staff and systems in the healthcare arena (HSE, 2017a).

The Your Voice Matters project describes what the process involved is as the following "Person centred co-ordinated care provides me with access to and continuity in the services I need when and where I need them. It is underpinned by a comprehensive assessment of my life and my world combined with the information and support I need. It demonstrates respect for my preferences, building care around me and those involved in my care". This definition was created from 19 statements gathered from 4 regional workshops, 11 focus groups and 2 online surveys from people who used services, supported service users and patient/carer representative groups. The definition describes what people want and need from health care services in Ireland (HSE, 2017b).

The vision for healthcare in the EU is to utilise eHealth systems to improve cross-border healthcare, health security, development of eHealth products. Utilising the systems will increase sustainability and efficiency to enhance patient empowerment and quality of care. The vision will confront the barriers by improving interoperability and supporting research for development and innovation in eHealth (Commission, 2012).

The successful execution of an eHealth strategy has been shown to be dependent on a number of fundamental enablers being present.

- A standards based, multi-layered information and technical infrastructure is needed to provide a common platform for eHealth deployments.
- The Ecosystem will be an important mechanism for developing innovative solutions to classic eHealth proliferation issues such as procurement issues, technical interoperability and testing and legal enabling (Health, 2012).

European connected health alliance (ECHA) facilitates international multi-stakeholder connections around ecosystems. 40 countries across Europe, the USA, Canada, and China with 600 members are represented. ECHA work closely with the European Commission and national governments for the design of public policies and strategies for digital health (ECHA, 2018).

Each of the strategies discussed in this section mention integration, interoperability, and standardisation as areas which need to be improved. The next section will elaborate on those key themes and the requirement for the successful implementation of an eHealth strategy.

## 1.2 Integration

Healthcare is the only sector that doesn't ask about your digital identity. In an era where facial recognition and biometrics are constantly improving, the healthcare sector needs to catch up and take over. For this to be achieved Tony Heffernan from the Saoirse Foundation stated: "Unless all three sectors are aligned, we cannot get to where everyone wants to be". The three sectors are public, private and voluntary. Along with these sectors, the emphasis on integrated care needs to be motivated by a patient centred care.

Integration of all information and all knowledge sources involved in the delivery of healthcare via information technology-based systems. This includes patients and their records, caregivers and their systems, monitoring devices and sensors and management and administrative functions. Examples are, e-prescribing – repeat prescriptions, Telehealthcare – e.g. heart failure, manage health from home environment, Ambient Assisted Living and Body worn sensor devices.

Healthcare delivery sector lags behind other industries where powerful information-based systems have been embedded for over 30 years or more. Reasons for this include failure to adapt and change operational and management processes, lack of technical standards and reimbursement solutions. This dissertation will address the fact that not all stakeholders are included in the standards development process which leads to the lack of interoperability and poor data quality. The importance of stakeholder engagement in the standards development process has been highlighted in national, European and international forums. This research will demonstrate, using the example of the condition of PKU, how vital this engagement is in the creation of standards.

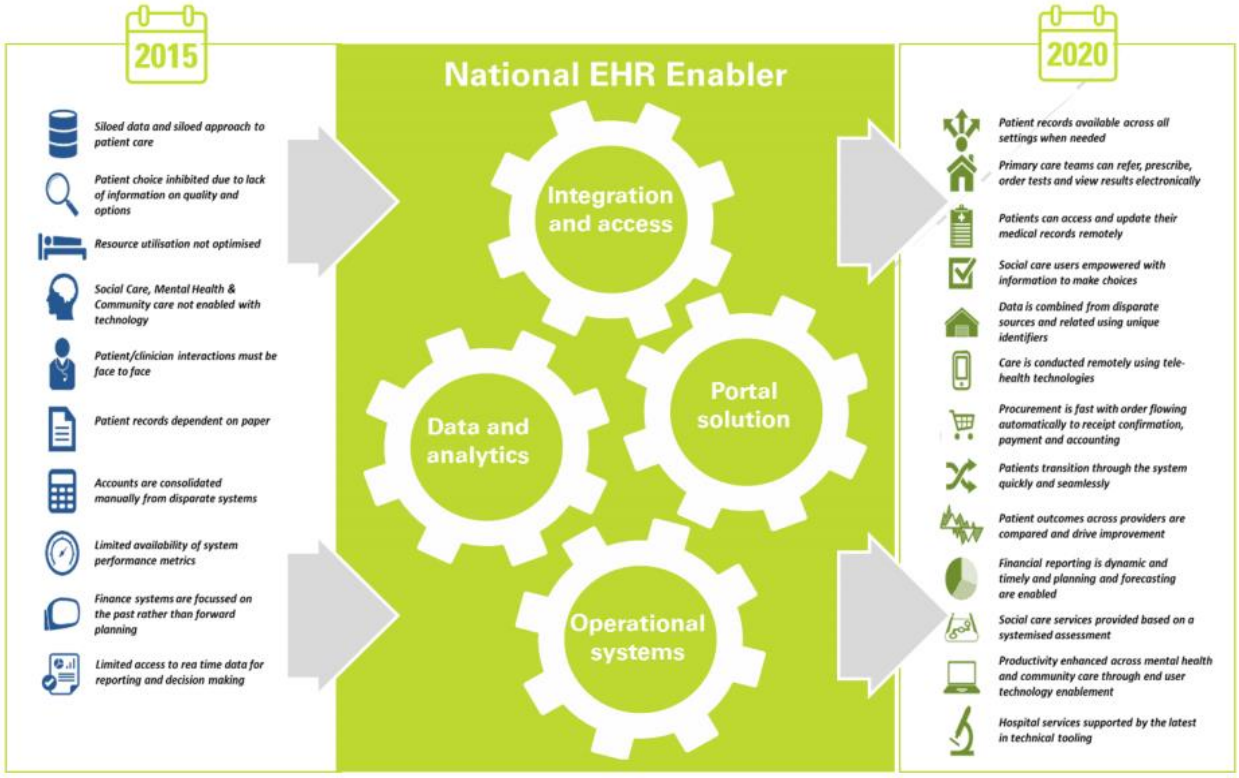



*Figure 3 National EHR vision from 2015 to 2020 with enablers required (Health, 2012)*

Figure three, depicts the state of healthcare systems in 2015, with the national EHR enablers like integration to have the successful implementation of an EHR. An example from figure three is the siloed data and siloed approach to patient care transitioned to patient records available across all healthcare settings when required (Health, 2012).

In addition to integration, we need interoperability. Recent research into eHealth Interoperability Conformity Assessment Scheme for Europe will compound the pathways and processes. The conformity scheme will promote the adoption and take-up of interoperability

testing of eHealth solutions against identified eHealth standards defined in the refined eHealth European Interoperability Framework (EIF) (Commission, 2017).

## 1.3 Interoperability

The EU action plan outlines limitations to the implementation of eHealth. One of the limitations listed is the lack of interoperability between eHealth Systems. The EU action plan describes how this limitation can contribute to the difficulty of health information exchanges (HIE). The European Commission recognises that a fundamental enabler to a successful eHealth strategy implementation is the formation of an "eHealth Ecosystem". The problem with eHealth Ecosystem systems is the lack of interoperability or essential interlinked elements (Health, 2012).

The Health Information and Quality Authority (HIQA) is an independent authority established to focus on safe quality care for people using health and social care services in Ireland. In HIQA's mandate, two key steps mentioned were setting standards for health and social services and health information. Following on from the HSE vision of patient-centric care, HIQA discusses developing person-centred standards based on best international practice. HIQA advise on the efficient and secure collection and sharing of health information, setting standards, evaluating and publishing information about the delivery and performance of Irish health services (HIQA, 2010).

HIQA defines interoperability as the ability of health information systems to share and understand data in a structured format. HIQA is developing health information technical standards to help with consistency in capturing and sharing health data. HIQA discusses the dimensions of data quality and a quality assessment tool which organisations can use to assess its data sources across all five dimensions. The five dimensions can be defined as, relevance, accuracy and reliability, timeliness and punctuality, coherence and comparability, accessibility and clarity (HIQA, 2010).

Healthcare Information and Management Systems Society (HIMSS) defines interoperability as the ability of computer systems, devices or software to exchange and interpret shared data (HIMSS, 2013). Interoperability is required for better coordination and integration across healthcare and health data exchange. HIMSS defines three levels of health information technology, foundational, structural and semantic. Foundational interoperability (FI) allows

data exchange from one information system to be received by another and does not require interpretation of the data. Structural Interoperability (SI) defines the format of the data exchange and ensures the data exchanges can be interpreted at the data field level. HIMSS defines Semantic Interoperability as the ability "of two or more systems or elements to exchange information and to use the information exchanged"(HIMSS, 2013).

## 1.4 Standardisation

The European Commission 2016 standardisation package, recognised the importance of global ICT Standards being implemented. Also recognised is the allowance of domain experts, national and regional authorities, health and social care professionals, patients and researchers to be directly involved in defining the semantics of the systems (Commisson, 2016).

Standardisation is one way to ensure the interoperability of systems. Often standards can be broad and not specific to requirements. Current standards for reference models do not currently recognise identities other than patients and healthcare professionals. There needs to be more inclusive and unified use of identity which would assist the use in health documentation (Ceusters and Smith, 2006).

ISO/EN13606 Health Informatics – Electronic health record communication, is the key standard involved in this research. ISO/EN13606 standard is currently under review. As it stands, ISO/EN13606 standard is not flexible. The current demographic model can only be of three main types: organisations, person or device (Chen et al., 2015). Other identities need to be considered, for example, laboratory. This research will address the role that identification plays to support stakeholders in the healthcare arena using an information modelling perspective.

Current two-level electronic health record (EHR) communication information models are physician and patient-centric. The restriction on types of entity makes it more difficult to manage information in an EHR which does not directly relate to the physician, for example, laboratory results, thus, limiting the use of two-level models in other healthcare information systems. In a paper entitled 'Using a generalised identity reference model with archetypes to support interoperability of demographics information in electronic health record systems', Chen, Berry, and Stephens promote the use of a generalised identity reference model (GIRM)

to facilitate the interoperability of identities between two-level model based EHR systems. They also demonstrate the strength and extensibility of using GIRM for the expression of other health-related identities (Chen et al., 2015). Current two-level models specifications make it more difficult to encourage the design of a two-level model for EHR feeder systems like a laboratory information system (LIS) and therefore putting a restriction on interoperability and improving data quality.

A key enabler identified for the eHealth strategy in Ireland is the provision of an individual health identifier (IHI). The IHI number will be used to identify a person and link their health records from different health systems. National registers for individual health identifiers and health service provider identifiers were established under the guidance of the health identifiers act, July 2014.

The IHI will uniquely and safely identify a person and in the absence of a unique national individual health identifier (IHI) in Ireland, data exchange will be an ineffective system (HSE). Poor data quality and specific identification systems contribute to the difficulty of sharing identifiable demographic information in a distributed EHR (Health, 2016). The IHI will aid the linking of health records from different health systems. For the IHI element to be established, in July 2014, the Health Identifiers Act was introduced (Book, 2014).

## 1.5 Importance of identifiers

In 1998, Arellano and Weber discussed that for the successful development of an integrated care system, it is necessary to create identities for patients. Accurate identity information will ensure relevant patient care information is truly available (Arellano and Weber, 1998). As recent as 2015, Chen et al., recognised that identities are required for other stakeholders in the healthcare system (Chen et al., 2015). Standards need to be changed to remove the rigidity. Flexibility is required for the introduction of new identifiers.

## 1.6 Vision for Healthcare

In figure four, my vision based on literature for healthcare is depicted. Patients should have input into every process leading into healthcare systems. Community and Primary care should be very closely linked which will help with proactive care. Starting with standards development process feeding into making standards which incorporate identifiers. The

standards developed will assist with data integration, which in turn will help with health care systems which are used in healthcare environments.

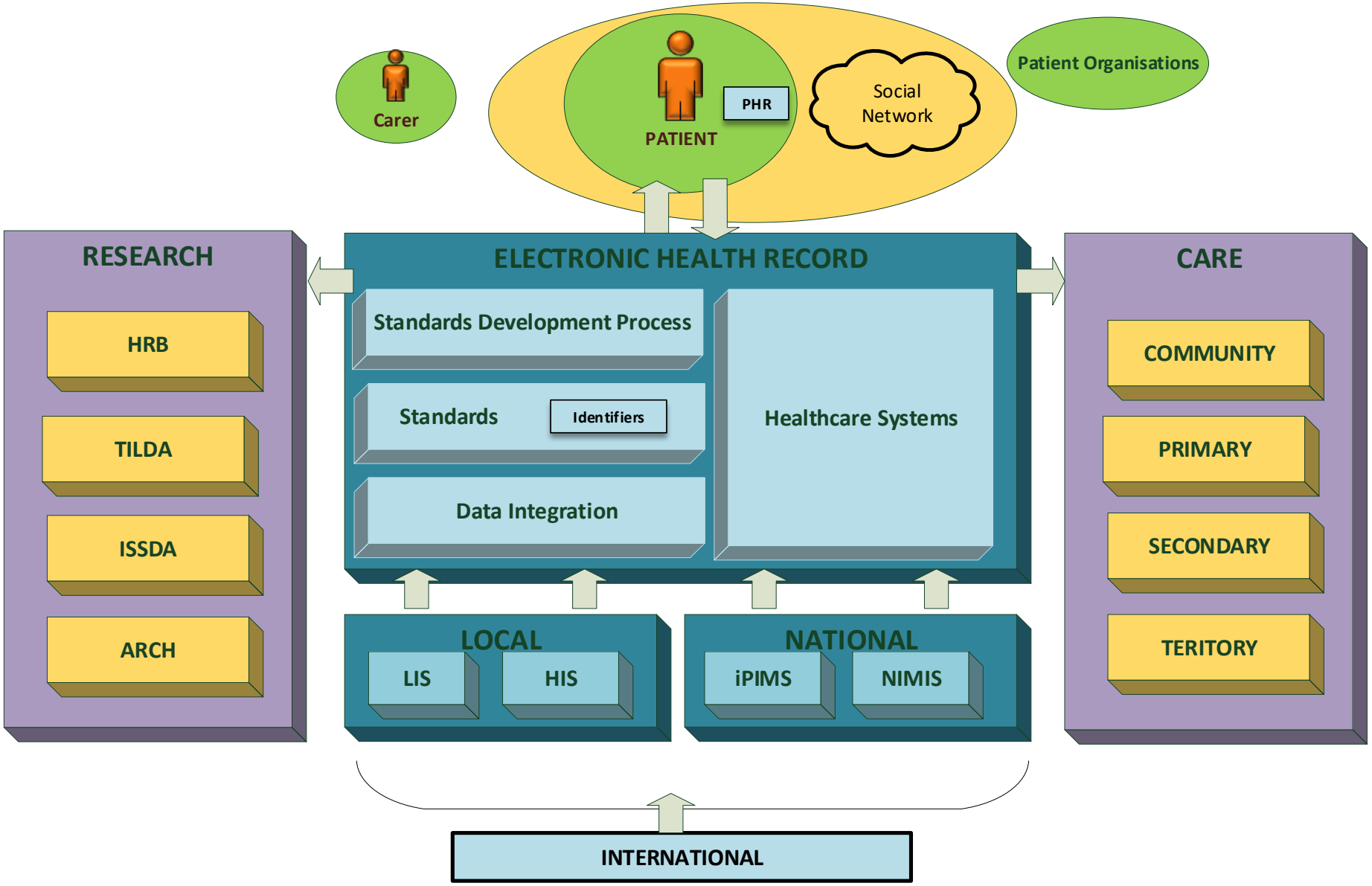



*Figure 4 Vision created from literature*

## 1.7 Aim and Motivation

The aim of this research is to identify the role that identification plays in supporting identified stakeholders in the care and management of patients with the disorder Phenylketonuria. The research idea has come from the challenges identified with the interoperability of information in electronic health record systems as identified in the paper by Chen et al, 2015.

The motivation for this research is improved patient safety and data qualiy. To obtain the highest standards of patient safety, data quality needs to be improved. Interoperability of the data is integral and needs to play a major role in the eHealth strategy. Furthermore, it should be incorporated into the development plans of future intergrated healthcare information systems (HIS).

## 1.8 Research Question

The research question is 'What role does identification play to support Metabolic Dieticians and Scientists for care and research of patients with Phenylketonuria (PKU) – An information modelling perspective'. The answer will focus on three key areas. Firstly, the role of identifiers in integrated healthcare and research using an EHR. Secondly, identifier issues focusing on modelling and quality. Finally, identifying uses carried out by dieticians and scientists using information for care and research of patients with PKU. These uses were identified by conducting 1-2-4-all liberating technique(McCandless, 2010). From researching these key areas, there is an anticipation that the results gathered could influence future standards for EHR communication.

## 1.9 Overview of the Research Process

This research sets out to answer the question "What role does identification play to support Metabolic Dieticians and Scientists for care and research of patients with Phenylketonuria (PKU) – An information modelling perspective". Figure five illustrates the research process. To achieve this the researcher needed to review the evolution of information modelling focusing on two level models representing the electronic health record (EHR) which have been employed by openEHR and the EN13603 standard for EHR communication. ISO EN13606 does not allow for flexibility when it comes to demographic entities, other entities, or resources. The demographic model of EN13606 can only be of three types, organisation,

person or device. To allow more flexibility to demographic models, identity trait blocks could be made available to create identity archetypes.

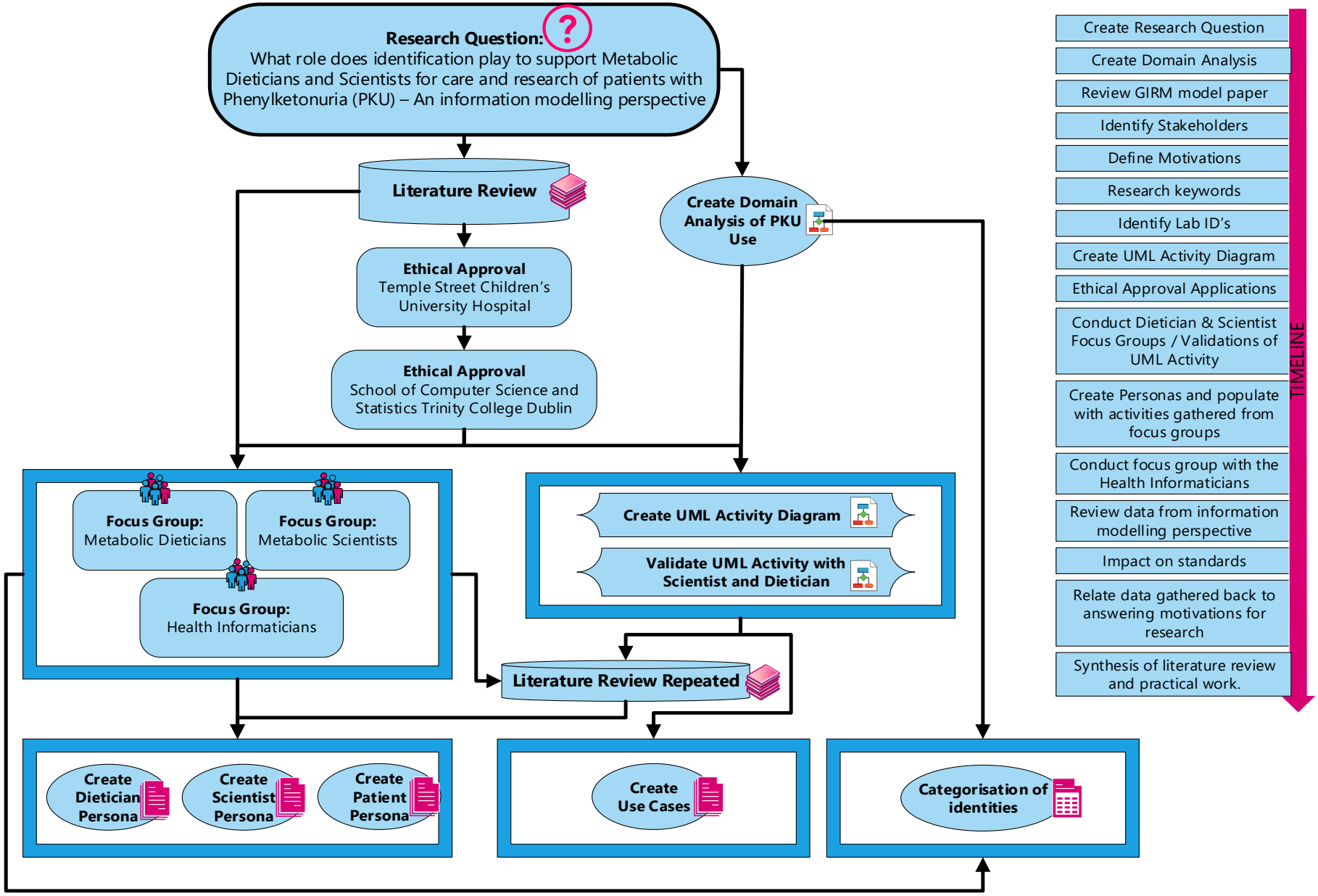



*Figure 5 Overview of the Research Process*

A literature review was conducted using the search terms in table 2. The limited amount of literature around identities made it difficult to conduct a review. In addition to the literature review the researcher used their knowledge and experience from working in a laboratory, understanding laboratory processes and laboratory information systems (LIS), data mining and data quality and previous assignments completed in the Health Informatics MSc to investigate, identification, integrated care and information modelling.

Table one shows the 11 different stages addressed by targeting identified stakeholders in the monitoring use case of a patient with PKU.

*Table 1* ***Stages of the Research Process***

| ***Stage*** | **Description** |
|---|---|
| *Stage 1* | Research question definition |
| *Stage 2* | Create Domain Analysis of PKU Use Case |
| *Stage 3* | Conduct Literature Review |
| *Stage 4* | Apply for Ethical Approval Temple Street Children's University Hospital |
| *Stage 5* | Apply for Ethical Approval Trinity College Dublin |
| *Stage 6* | Conduct Focus Groups |
| *Stage 7* | Validation of UML Activity Diagram |
| *Stage 8* | Literature Review Repeated |
| *Stage 9* | Create Personas |
| *Stage 10* | Create Use Cases |
| *Stage 11* | Review data, standards, and information modelling perspective. |

From the researcher's laboratory experience a UML activity diagram of a sample journey for a patient with the condition Phenylketonuria was created. The researcher listed the identities known to them, associated with the sample journey. The purpose of the focus groups is to determine the identities used by different stakeholders in the patient and sample journey. The metabolic scientists and dieticians were asked 'As Metabolic dieticians and Scientists you generate, collect and store data. From your experience, what are uses of data for care and research of patients with PKU?'. Once the uses for care and research were listed a secondary question was asked 'What are the identities you need to conduct those uses?'. The responses from each of the focus groups were accumulated and displayed in brainstorming diagrams and also tabulated to show the identities required for each use case. The results from the focus groups were used to create a patient, dietician and scientist persona.

## 1.10 Overview of the dissertation

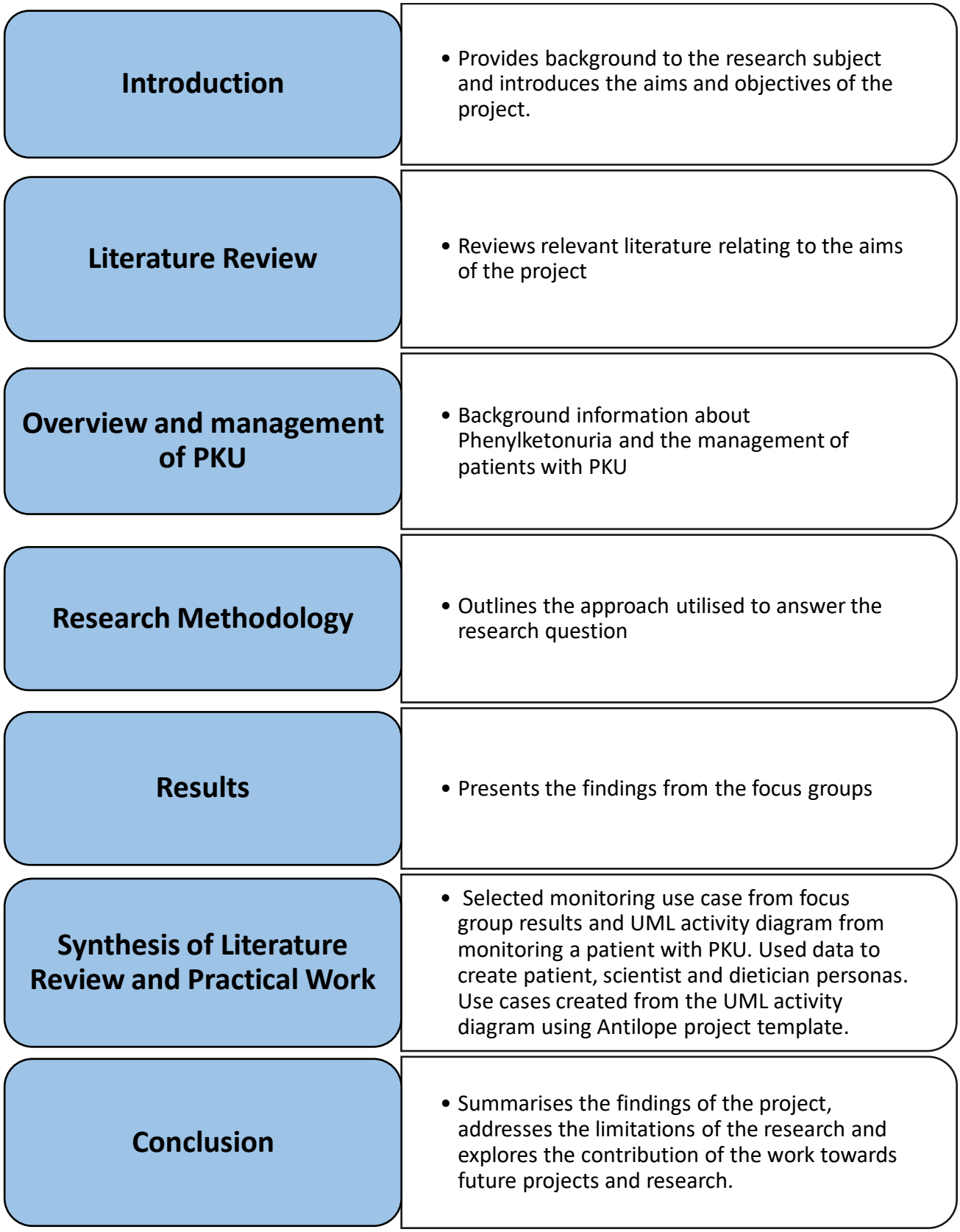



*Figure 6 Overview of the Dissertation*

# 2 Literature Review

This section is a review of a gap identified in the literature in the research area of identities, interoperability and reference modelling. This review contains data previously reported about Phenylketonuria and the process for management and treatment of PKU in Temple Street Children's University Hospital.

Before I introduce the ecosystems and interoperability projects, I am going to introduce the search terms and background to the research. Following this, the review will focus on the areas of interoperability, standardisation in association with reference modelling to provide context.

From conducting my literature review, the topic of identity reference modelling is quite sparse on literature. In section 2.3, the background of the research, the source paper is discussed. From this paper, my research question was created and motivations for the project were discussed. In my current working role as Laboratory IT Specialist and previous experience as a laboratory scientist in Temple Street Children's University Hospital (TSCUH), this gives me an interesting perspective and knowledge in the management of patients with Phenylketonuria (PKU). There is a unique aspect of patients living with PKU, monitoring of phenylalanine levels is performed by home blood sampling and sent to the laboratory directly. This differs from most conventional outpatient treatment management plans, where a patient may be self-monitoring as in the case of diabetes or patients having periodic blood tests taken in phlebotomy before out-patient appointments, for example, patients with HIV.

## 2.1 Search Terms

The Stella library, Science Direct and PubMed databases and Google search engine were used to conduct the literature review. Search terms identified from the literature have been categorised in table two. The categories are standards, identification, information modelling, patient, and legislation.

*Table 2 Search terms used for conducting a literature review*

| *Standards* | *Identification* | *Information Modelling* | *Patient* | *Legislation* |
|---|---|---|---|---|
| • Standards<br>• Standards Development Process (SDP) | • Trait<br>• Entity | • Information modelling<br>• Meta-modelling<br>• Reference Information Modelling | • Management of patients<br>• Access to information | • Assisted Decision Making (Capacity) Act 2015 |
| • Standards of care<br>• Care Standards | • Identity<br>• Identifiers | • Single level model<br>• fixed model<br>• Two-level modelling<br>• Reference model<br>• GIRM | • Phenylketonuria<br>• Phenylalanine | • Health Information and Patient Safety Bill |
| • Standards based information models | • Patient Master Index | • Use Cases<br>• Archetype | • Dieticians<br>• Scientists | |
| • OpenEHR<br>• ISO13606 | • Demographics | • EHR Communication<br>• Interoperability | • Laboratory | |

## 2.2 Background of the research

The paper which leads to the design of my research question is entitled 'Using a generalised identity reference model with archetypes to support interoperability of demographics information in electronic health record systems'(Chen et al., 2015). Current two-level EHR communication models are physician and patient-centric. This paper discusses the limitations for types of entities that are created in electronic health record (EHR) specifications. The limitations make it more difficult to manage information in an EHR which does not directly relate to the physician e.g. Laboratory results.

Chen, Berry, and Stephens introduce a generalised identity reference model (GIRM). The model is based on key characteristics identified from five surveyed demographic models. The writers evaluated GIRM by using it to express the EN13606 demographics model at the metadata level and showed how two-level modelling can support exchange of instances of demographic identities.

The writers evaluated the GIRM by using it to express the EN13606 demographic model in an extensible way at the meta level and show how two-level modelling can support the exchange of instances of demographic identities. This evaluation method was used to show the interoperability of identities between two-level models based EHR systems and validity and the extensibility of using GIRM for the expression of other health-related identities.

Two level models for representing the EHR have been employed by openEHR and the EN13606 standard for EHR communication. Two level models allow domain experts to configure, build and constrain the shape and content of EHRs. The first level is a reference model that comprises a set of reusable EHR building blocks. The second level comprises of archetypes. For interoperability of identity management between systems two-level models facilitate flexibility which can be constrained by EHR developers. Archetypes can be used to specialise in traits, trait parts or identified entities to allow representation of other demographics models using a single general RM. This shows the reference model is extendable into additional types of identifiable entity, e.g. sample.

An open domain-driven platform for developing flexible eHealth systems is openEHR. openEHR is a virtual community, with the aim of turning health data in the physical form into electronic form. This conversion will assist in ensuring universal interoperability of electronic data. openEHR is a multi-level single sourcing modelling approach with service-orientated software architecture. In each layer of the model, domains are built by experts following specifications published by the openEHR foundation. This approach targets the semantic level of health information, enabling functions like research querying. The openEHR approach enables a platform-based eHealth software market in which vendors and developers' solutions are interfaced via standardised information models. This avoids product and vendor lock-in, retains ownership of the data for secondary use and allows clinical input in solution development (openEHR, 2018)

## 2.3 Ecosystems and Interoperability Projects

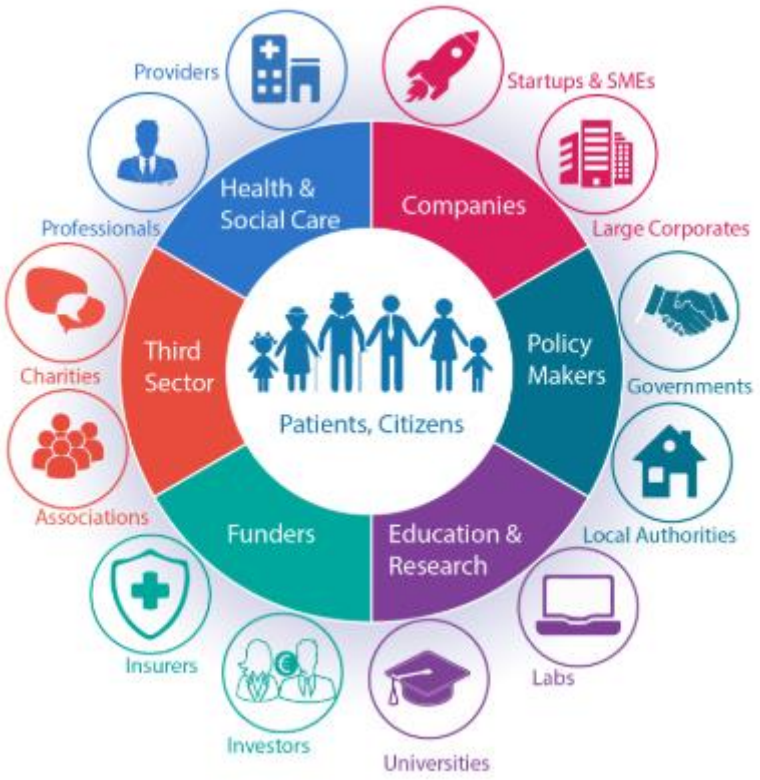



*Figure 7 ECHAlliance Ecosystem (ECHA, 2018)*

ECHAlliance ecosystems bring all key stakeholders together across 20 locations internationally. They facilitate the engagement between all relevant sectors for making change happen in connected health (ECHA, 2018). In figure five, the ECHAlliance ecosystem is depicted with the all stakeholders represented. In comparison to figure two, figure seven has more stakeholder inclusion. With increased stakeholder inclusion the ECHAlliance ecosystem will help improve interoperability, design and implement innovative solutions for different areas of healthcare from disease management to falls prevention (ECHA, 2018).

In January 2010, an EC-funded research project called HITCH started. Healthcare Interoperability Testing and Conformance Harmonisation (HITCH) was part of Europe's roadmap for developing Interoperability Conformance Testing for information systems in the healthcare arena. HITCH provided a vision for how interoperability of eHealth systems should be organised across Europe and further afield (HITCH, 2010). The three components of the vision are outlined as:

1) Analysis of existing eHealth testing tools and identification of tools still missing

2) Conception of a Quality Management System for cross-vendor and in-house interoperability testing
3) Comprehensive eHealth certification and quality labelling scenarios for Europe

The policy context from HITCH research noted the need for data to be ubiquitously available for optimum treatment of patient's (HITCH, 2010).

The HITCH project discussed the requirement of a standardised way for systems communication. 'If two systems can talk to each other, then there is "interoperability" between those systems' (HITCH, 2010). HITCH describes how standards together with medical protocols could be used to express health data in an electronic health record (EHR). The goal would be to share the information so that a computer system could understand and evaluate it to provide an understandable presentation to users.

Projects like HITCH have been built on previous work such as CALL for InterOPErability (CALLIOPE) which was launched in 2008. The main goal of the CALLIOPE project was to produce value for decision makers for national eHealth implementations. CALLIOPE was comprised of decision makers, implementers, professionals, patients, and other stakeholders who could share visions of how to establish interoperable eHealth services. The CALLIOPE network has established a successful collaborative platform for many actors in eHealth interoperability in Europe. 'The process has reached six main achievements:

1) Offering support to European decision-makers regarding EU level actions on eHealth.
2) Enlarging active representation of EU and European Free Trade Association (EFTA).
3) Enlarging active involvement of European eHealth stakeholder organisations.
4) Developing and validating an open working method among stakeholders
5) Creating a working collaboration method between the appropriate eHealth large scale pilots and wider range of member states and stakeholders CALLepSO collaboration.
6) Building an eHealth Interoperability road mapping process.

## 2.4 Interoperability

European Health Telematics Association (ETHEL) is the one multi-stakeholder organisation within Europe that brings together organisations and individuals in eHealth. ETHEL provides a platform for groups and organisations to interact, exchange ideas and information, leading to innovation and improvement in the delivery of eHealth solutions (ETHEL, 2017).

The European Commission (EC) funded a study about eHealth Interoperability Framework (EIF) which was published in June 2013. The study defined the vision of an EU eHealth Interoperability Framework with four levels including technical, semantic, organisational and legal. From the perspective of the levels mentioned the aim of the study is to apply the EIF to the domain of eHealth. As discussed in chapter one, interoperable health records are needed to help make care safer, health to be more patient-centric and inclusive (Consulting, 2013).

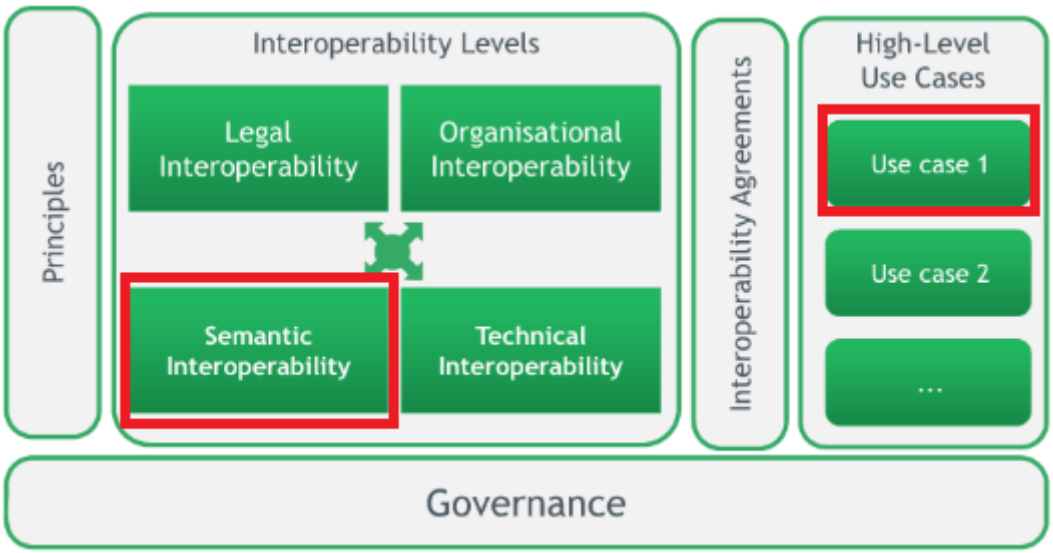



*Figure 8 Extract from the EIF Study of eHealth EIF Structure highlighting areas in red this research will contribute (Deloitte, 2013)*

As highlighted in figure eight, the focus of this research is to contribute a change to the semantic level of interoperability using a use case template. Extracted from the EIF study document, "semantic interoperability aims to precise "meaning of exchanged information which is preserved and understood by all parties""(Deloitte, 2013).

Between 2016 and 2018, the eHealth conformity Assessment Scheme for Europe (EURO-CAS) will develop a sustainable Conformity Assessment Scheme (CAS) for Europe. The CAS scheme will promote the adoption of interoperability testing eHealth solutions against identified

eHealth standards defined in refined eHealth European Interoperability Framework (eEIF). This project is funded by the EU Horizon 2020 research mentioned in the introduction. As mentioned previously, Karima Bourquard the director of Interoperability for IHE-Europe writes in a paper entitled "Contribution of standards and profiles to the interoperability in eHealth" the use of standards-based integration profiles will promote the use of interoperability harmonisation among Europe countries. This will accelerate the delivery of quality and efficiency in care processes (Bourquard, 2011).

Between 2013 and 2015, the Antilope project was focused on the dissemination and adoption of the eEIF. The project created, validated and disseminated a common approach for testing and certification of eHealth solutions. European and international experts and stakeholders developed a series of overview use cases, standards, and profiles following the eHealth interoperability framework (Antilope, 2015).

'eHealth interoperability is one of the greatest challenges for the whole healthcare community – and not just for healthcare IT only' (Antilope, 2015). A recent project relating to interoperability is called ANTILOPE. Between 2013 and 2015, national and international organisations worked together to define and select eHealth standards and specifications. The Antilope project has provided regional, national and international projects with practical guidance on how to converge eHealth platforms and practices by using international profiles and standards. The Antilope project results demonstrate how EU Projects carry forward its approach. Recommendations from the project suggest how the process will be used and further disseminated to advance eHealth Interoperability (Antilope, 2015).

## 2.5 Standardisation

'Understanding information in EHR systems: Paving the Road for Semantic Interoperability through standards' has some interesting points. This reference discusses the challenges and difficulties of current applications to interpret information exchanged between sender and receiver. Some examples of health information formats developed over the last few years are Health Level Seven (HL7), Clinical document architecture (CDA) and HL7 Fast Interoperability Healthcare Resources (FHIR) (Orlova and Salyards, 2016).

Arellano and Weber mention the lack of standards in data collection for demographics and master patient index (MPI) cause poor record linkage (Arellano and Weber, 1998). Arellano

and Weber describe the concept of an enterprise person index (EPI) and MPI as the 'glue' which supports relevant patient care information across systems. They identified three main issues with combining EPI's, '1) There are no accepted standards for either collecting or storing substantive information, 2) There are no standards for default values, allowing registrars to assign any value for unknown data fields rather than the desired blank or empty field, 3) Data for identifying critical fields are subject to error because of changes that occur over time, registrar error, or patient misinformation(Arellano and Weber, 1998). The importance of standardisation is highlighted as a key weakness in collecting and recording data. This lack of standard contributes to the problems with linking data.

In a paper entitled, 'Contribution of standards and profiles to the interoperability in eHealth' the abstract explains the paper will provide an overview of the standards that are used in eHealth with sharing medical data. Bourquard describes the standards in eHealth as 'reaching maturity'. It discusses the use of a European set of profiles by eHealth projects promoting interoperability harmonisation among European countries. Bourquard says this will 'accelerate the delivery of quality and efficiency in the care processes' (Bourquard, 2011). Care of patients is described as 'no longer the role of a single practitioner' but as 'team across several specialities, locations'. Europe wide there is an emphasis on IT product solutions and their interoperability to increase the quality and efficiency of care. Some examples Bourquard uses are telemedicine and ambient assisted living (AAL) tools.

Bourquard writes about allowing the sharing of health data and two key elements that must be considered. One, the communication infrastructure and secondly the interoperability of the IT systems and the devices interconnected with the communication structure. Some requirements would be the identification of the professionals and patients who are involved in the care. A secured environment with data protection, which raises a question of, who is responsible for the medical data and who has the right to access the data. One other requirement as mentioned before is the interoperability between systems in which data is exchanged or shared. For interoperability to be successful the systems need to implement standards and protocols at the semantic, syntactical and technical levels. Terminologies such as LOINC, Snomed and ICD-10 are described as key elements to achieve interoperability (Bourquard, 2011).

A hospital is seen as a main user of standards. The hospital information system is composed of patient management systems, EHR systems, laboratory information systems (LIS), radiology information systems (RIS), picture archiving communication system (PACS), pharmacy information system to name a few. Data is exchanged among all these systems. Bourquard describes the problem of a transition phase when proprietary standards, international standards, and integration profiles are working together. There is the challenge of trying to make this transition phase as short as possible, with the balance of future vendors uptake on new standards versus current or other vendors not seeing the advantage of knowing the new standards (Bourquard, 2011).

Five reasons are mentioned as to why the standards outlined have not been widely adopted. They are quality safety issues, adoption of the international standards, semantic interoperability, communication and training and governance. Bourquard concludes that 'Interoperability eHealth is now a concept with increasing maturity and its strategic impact is much better understood'. Because of this, increased harmonisation, and quality of products, will, in turn, enhance patient safety (Bourquard, 2011).

Boussadi and Zapletal write about the differentiating the representation of data instances from the definition of clinical information models. They talk about the most recent initiative called Fast Healthcare Resources (FHIR). The aim of their study was to investigate the application of the FHIR standard to modelling and exposing EHR data. They were able to conclude and show the feasibility of implementing a FHIR layer over an i2b2 database model to expose data of the clinical data warehouse (CDW) as a set of FHIR resources using HAPI FHIR API (Boussadi and Zapletal, 2017).

Piho et al. wrote a paper entitled 'Archetypes based meta-modelling towards evolutionary, dependable, and interoperable healthcare information systems'. They discuss the evolutionary aspect of the ability of information systems to change and evolve similarly to how organisations and business processes change. Information systems should be able to communicate and understand each other's data, this is the interoperability aspect and lastly, they should be dependable, 'work correctly and securely as expected'. Archetypes and archetype patterns were used as meta-models. Piho et al., state these three elements of dependability, interoperability and evolutionary criteria as extremely important for laboratory software (Piho et al., 2014).

From the second international workshop on meta-modelling for healthcare systems, Piho et al. redesigned the previous archetypes and archetype patterns and compared them to HL7 version 3: Reference information model and openEHR Reference Model. In the comparison, they put an emphasis on 'semantical aggregation of health data across heterogeneous data sources, as well as the interoperability and evolution models and software'. They mention one of the issues with domain models developed by different independent parties is semantic heterogeneity. This is an obstacle when developing interoperable software systems. Piho et al., describe some data mining tools used to deal with semantic heterogeneity, for example, Microsoft BizTalk Server. They explain the concept of many actions in one task and in each action, there may be many outcomes. An example given was a laboratory measurement, the ordering doctor is the consumer and the laboratory is the provider. '" Sending an order", "sample collecting" and "receiving a report" can be the actions, and results reported to the laboratory can be the outcomes of the "receiving a report" action'. Piho et al. concluded that the analysis performed demonstrates the capability to semantically aggregate health data across heterogeneous data sources (Piho et al., 2015).

As described in chapter one, health ecosystems are an important mechanism for developing innovative solutions to issues with technical interoperability. But the success of an eHealth strategy is dependent on fundamental enablers like a standards based platform (Health, 2016). As part of developing a roadmap for eHealth standards a project called *e*Standards was proposed by HL7, CEN/TC 251, IHE and eHealth competence centres in Europe, with support. The *e*Standards project aims to advance eHealth interoperability and align global standards. Using European and Global stakeholder involvement, *e*Standards aims to build consensus on eHealth Standards, provide an arena for knowledge sharing and promote rapid adoption of standards.

This concludes this chapter of reviewing the literature relating to and influencing the research topic. The next chapter will discuss the overview of phenylketonuria and how patients are managed.

# 3 Overview and management of PKU

This chapter will discuss the overview and management of PKU. The chapter is sectioned into firstly presenting the domain analysis seen in figure 9, followed by information about dietary treatment, blood monitoring, clinic visits, benefits from defero texting system and potential disadvantages to the system. The chapter concludes with the 2017 published European guidelines for the management of patients with PKU.

Phenylketonuria (PKU) is a rare genetic disorder that is present from birth. In PKU, the body is unable to break down an amino acid called Phenylalanine which then builds up in the blood and the brain and can cause brain damage and other problems when untreated.

Ireland was the first country in the world to begin a national screening programme for PKU. At NCIMD National Centre for Inherited Metabolic Disorders, we have been treating patients with PKU from birth since it was added to the National Newborn Screening Programme in 1966. 1 in 4,600 babies born in the Republic of Ireland has PKU.

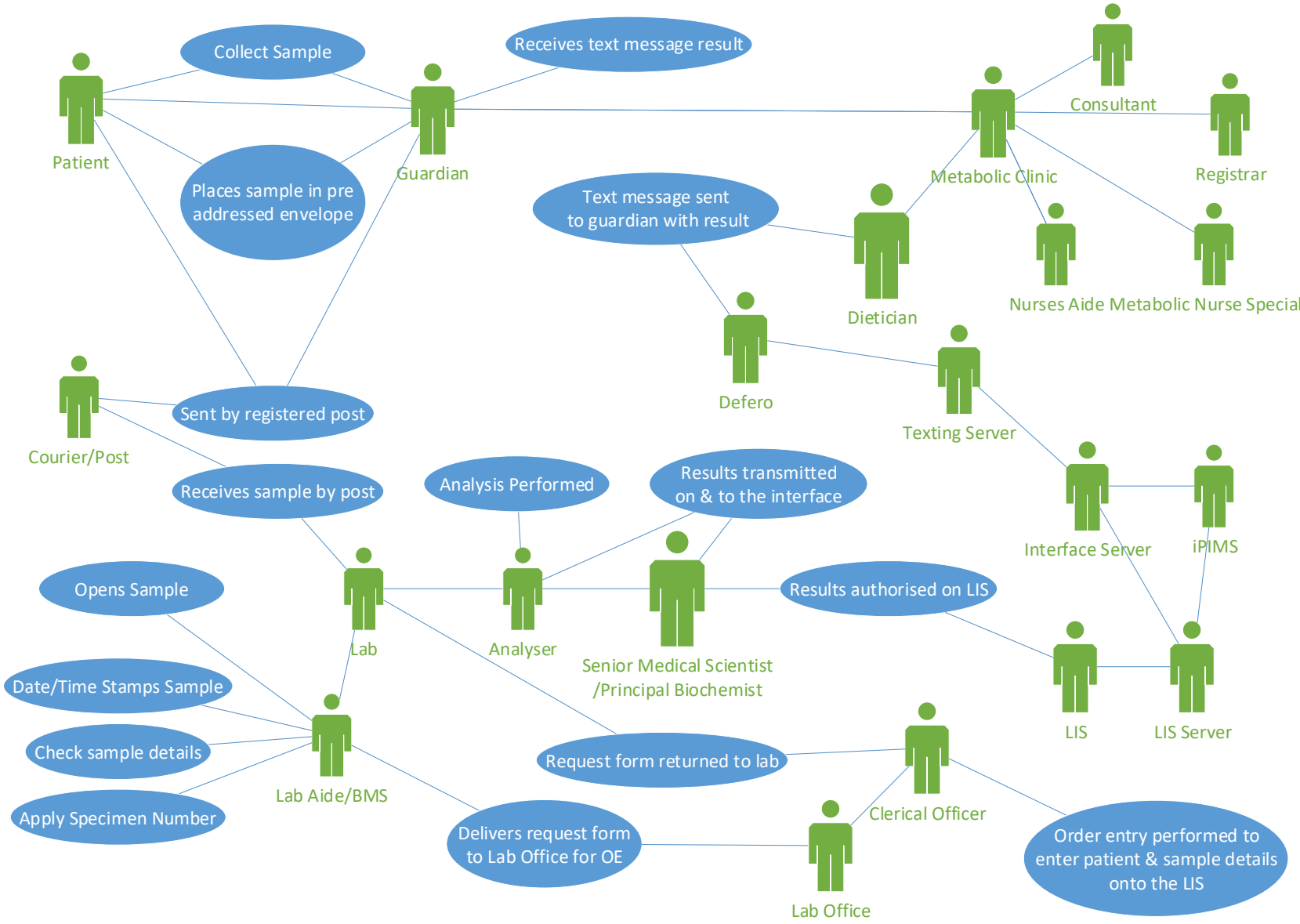



*Figure 9 Domain analysis of PKU Use Case*

## 3.1 Dietary Treatment

Treatment by Dietary protocol commences for all infants whose levels exceed 600µmol/L and infants whose levels remain between 400µmol/L and 600µmol/L for more than seven days. Diet for life is recommended. The monitoring frequency in the table below is only a guideline and depends on individual circumstances. The diet has three components, synthetic protein, protein exchanges and free foods. Synthetic protein or phenylalanine-free protein substitute is a drink that is essential for growth and development. To ensure levels are within the recommended range, it is advised that the patient intakes the drink evenly over the day. Most protein substitutes have vitamins and minerals included; others need these added to make them complete. Protein exchanges or natural protein contained in e.g. meat, fish, cheese, eggs, soya, and nuts are not allowed in the protein-restricted diet. An exchange is the amount of any food which contains 1g of protein. The number of exchanges allowed will vary between individuals and is changed from time to time in accordance with blood levels and growth. Protein-free foods as the name suggests, do not contain protein and may be consumed liberally to provide energy and calories and variety in the diet. Some examples of protein free food are most fruit and vegetables, butter, jam, honey, and special low protein products such as low protein milk.

## 3.2 Blood Monitoring

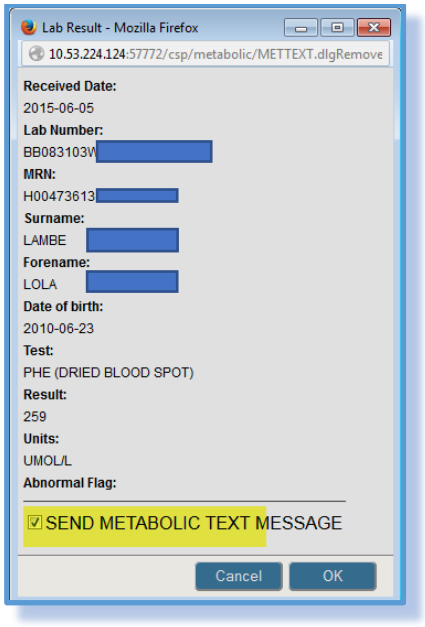
Regular blood tests are an essential part of the overall management of PKU. Monitoring is the way of assessing acceptable control. The frequency and method of blood tests differ depending on age, control, and other factors. A liquid blood sample is required up until one year of age and in maternal PKU. Otherwise, a sample of dried blood spots on a special card (Guthrie card) is required. Treating PKU involves a protein-restricted diet for life and taking regular dietary supplements which contain amino acids, vitamins, and minerals. The aim is to keep the blood Phenylalanine level within a specific target range. This is where self-monitoring is important. SMS text messaging is a useful tool for contacting patients to let them know to continue with set diet or adjust based on their levels.

## 3.3 Clinic Visits

In the metabolic (OPD) Out Patient Department in TSCUH, approximately 15 patients are seen every week. At each clinic visit, there is a dietary review, a general health review, and

biochemical screening from 10 years for the following, B12 Folate, Zinc, Selenium, Quantitative amino acids, bone and renal profile, ferritin, FBC, and a psychology review.

In Temple Street Children's University Hospital (TSCUH), there is a text message service for phenylketonuria (PKU) patients to receive their Phenylalanine (PHE) results. PKU patients require regular monitoring of their blood PHE levels. Patients collect their samples at home by finger pricking to release a spot of blood which is applied to a dried blood spot (DBS) card. Once dry, the card is posted to the Department of Paediatric Laboratory Medicine (DPLM) for testing. The metabolic texting software webpage, when run, reads lab data from the data warehouse but will only take outpatients that have been designated as part of the pilot. Other criteria include only taking results that have the tests DBPHE or PKPHE and patient date of birth falling within the range 18 months to 4 years. By default, all entries are displayed with the "TEXT" flag set on.




The user can scroll through the list and pick the entries that he/she wants to suppress a text message. This is done by double-clicking on an entry and un-ticking the "Send text" option and clicking OK to save or Cancel to quit.

Once an entry has been flagged as "no text message" it will appear as a red colored line entry and have its TEXT flag set to N.

*Figure 10 Web view of the lab result and selection box to send metabolic text message*

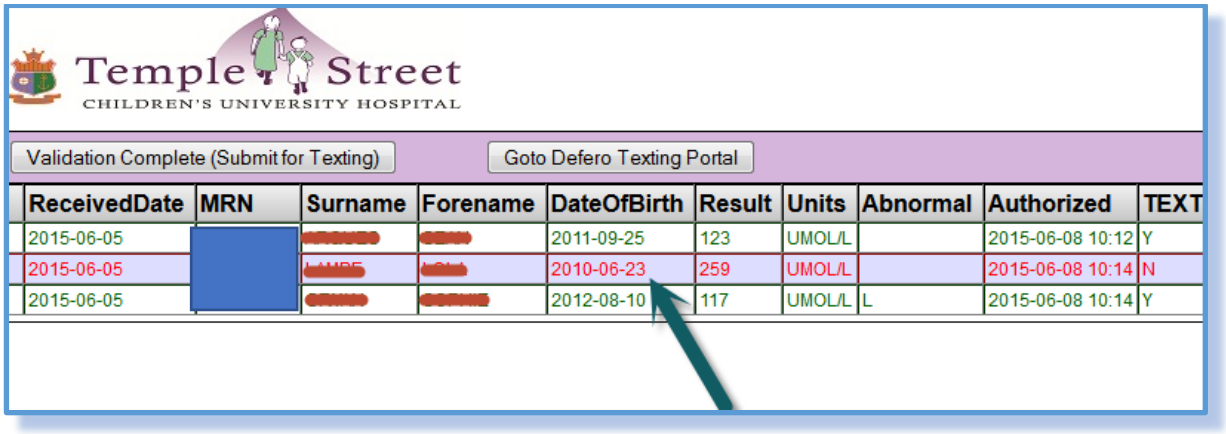



*Figure 11 Validation screen containing examples of the identities used to confirm patient demographics before sending a text message*

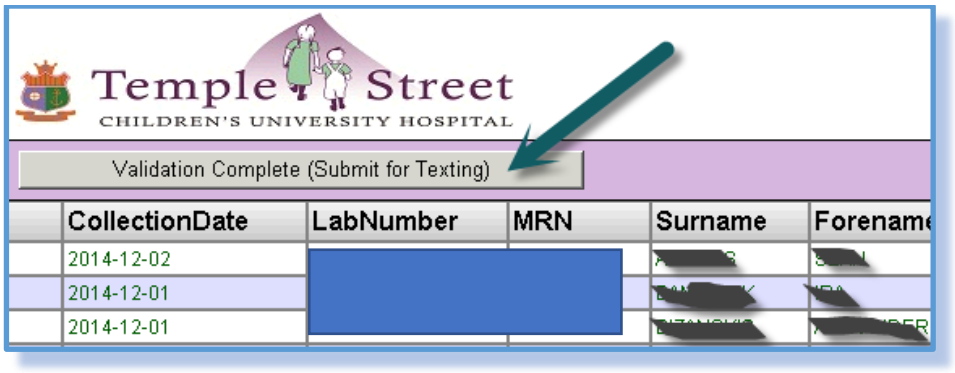



*Figure 12 Validation screen with confirmation button for submitting list for text message sending*

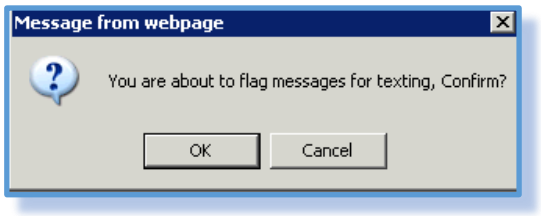



*Figure 13 Confirmation prompt box*

When the user is happy enough having validated the list, they will click on the "Submit for texting" button.

As a precaution, the user will be prompted to confirm the action. When the user selects OK, the list is cleared down and no longer available until the next run.

At this stage, a background process runs which picks up the validated results and passes an appropriate text message on to the text messaging software Defero. Users can log onto

Defero to check whether text messages were sent out via the portal link on the metabolic texting web page.

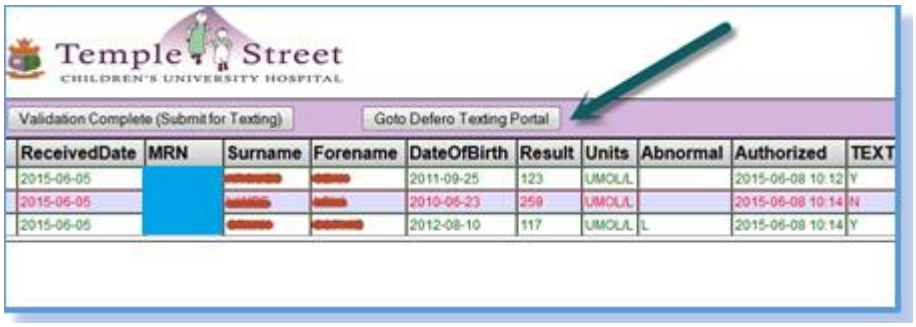



*Figure 14 Displaying the button link to the Defero Texting Portal*

To date, 280 patients are enrolled to receive results via text message. Currently, there are approximately 6,000 PHE tests performed per annum. Over 50% of results are within normal limits. This text message service has saved dieticians an estimated 580 hours per annum, with an estimated cost saving of €3,000 per annum. 97% of parents surveyed felt the system saved them time.

### 3.4 Benefits not measured

Apart from the aim of a better patient outcome, because of quicker results and potentially improved adherence to diet, there are benefits which cannot be measured. One such benefit is the time gained by the dieticians, with a significant reduction in the number of hours spent ringing each patient with phenylalanine levels and dietary adjustments. Their time can be spent with patients who may not have gotten the desired attention before the introduction of the Defero system. Focus can be switched to identifying patients with a greater need of closer monitoring of diet and guidance with the treatment plans determined by the dietician.

### 3.5 Potential Disadvantage

Up until the age of 16 patients attend TSCUH for treatment and consultations. After their 16$^{th}$ birthday, patients are transferred to the AMET Adult Metabolic Clinic in the Mater Hospital. A potential disadvantage of the SMS text message system is that this service will be lost when patients are transferred. This is an aspiration for the future to have the Mater hospital on the same Defero system, but currently, this is not available.

Upon receipt of the DBS, the laboratory ensures the sample meets criteria set for testing, e.g. data quality, patient demographics, sample quality. Each DBS is assigned a unique specimen

ID, which is entered in iLAB against the patient demographics. Patient demographics are maintained in the patient management system (PMS) iPIMS. The tandem analyser is interfaced with the laboratory information system (LIS) iLAB which is in turn interfaced with the (PMS) patient management system iPIMS.

Samples are processed using a Tandem MS/MS analyser. Results are entered in the laboratory LIS called iLAB. They are clinically authorised by a Scientist or Principal Biochemist. A message is generated and sent to the data warehouse, called cache. Cache is a temporary storage location for the files, so they can be accessed again at a later point in the process. A web page is viewed and displays the data from the warehouse. This is the point where the dietician interacts with the web page and validates the results generated by the lab and selects the results which are ready to be processed for receiving an SMS text message.

Ensemble is the integration engine used to perform ETL. ETL stands for Extraction, Transformation, and Loading. Ensemble extracts the data from the warehouse (cache), also collects mobile phone number and checks patient demographics data from the iPIMS system, then transforms the message and loads the data into the Defero system. A list of patients is sent to Defero and is ready for the last step of transmission. The SMS is then sent to the patient.

PHE results that are 'normal' will be sent a text message containing the patient MRN, date of sample, numeric PHE value and advised that no dietary changes are necessary. However, there is a different process for 'abnormal' results. Abnormal PHE results will not be given a numeric value and will be asked to phone the metabolic dietician for advice.

## 3.6 European Guidelines

In January 2017, a review entitled 'Key European guidelines for the diagnosis and management of patients with phenylketonuria' was published. The main objective of this review was to optimise phenylketonuria (PKU) care. It identified diet as an important role in treatment. PKU is a lifelong treatment and management is scheduled according to age, adherence to treatment and clinical status. 'Nutritional, clinical and biochemical follow up is necessary for all patients, regardless of therapy.' This extract from the report highlights the nutritional importance in the management of care.

Over a three-year period from October 2012 to December 2015, the guidelines were developed using a method called the Scottish Intercollegiate Guidelines Network (SIGN). From the literature conducted seventy recommendations were formulated. Of the seventy, ten were chosen to be of highest priority and referred to in the review. The guidelines were created by seventeen European PKU experts. They included paediatric and adult metabolic physicians, paediatric neurologist, psychologists, neuropsychologists, a biochemist, and metabolic dieticians. They were divided into five working groups, 1) nutritional treatment and biochemical or nutritional follow up, 2) neurocognitive outcomes, 3) Psychosocial outcome, 4) Adult and Maternal PKU and 5) Diagnosis of PKU including treatment initiation and drugs in PKU.

The previous paragraph highlights the need for interoperability. This chapter describing the overview and management of patients with PKU is concluded. The next chapter will describe the research methodology used to conduct the research process in figure five.

# 4 Research Methodology

This chapter describes the methodology used to answer the research question proposed in chapter 1. It will provide the rationale for selecting the method and tools used to gather data and validate processes. For the purposes of this research, I would describe myself as a T-shaped Researcher (Brown et al., 2015).

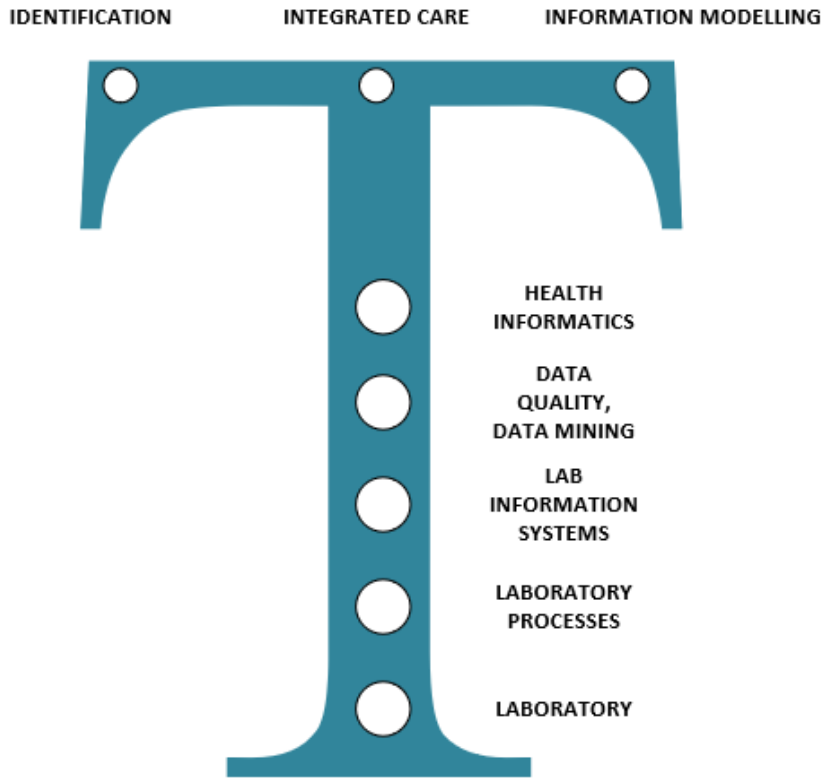



*Figure 15 T-shape Researcher Diagram*

## 4.1 Research Question

The research question What role does identification play to support Metabolic Dieticians and Scientists for care and research of patients with Phenylketonuria (PKU) – An Information modelling perspective will be answered by reviewing literature, creating a domain analysis of the PKU Case, conducting three focus groups, creating a UML activity diagram, creating a patient, scientist and dietician personas, creating use cases, collection of the identities and uses and the categorisation of the identities into tables.

## 4.2 Focus Groups

The literature outlining the guidelines for the diagnosis and management of patients with PKU identify the key stakeholders are Metabolic Dieticians and Metabolic Scientists to optimise

PKU care. The purpose of this research was to gather data and receive validation from key stakeholders identified in the literature and domain analysis relating to identification in the care and research of a patient with PKU. The metabolic scientists and dieticians were asked 'As Metabolic dieticians and Scientists you generate, collect and store data. From your experience, what are uses of data for care and research of patients with PKU?'. Once the uses for care and research were listed a secondary question was asked 'What are the identities you need to conduct those uses?'.

## 4.3 Complex Systems and 1-2-4-all Liberating Structure

The Oxford English dictionary defines the word complex as "Consisting of many different and connected parts", "A group or system of different things that are linked in a close or complicated way; a network"(Dictionary, 2018). In a briefing document created by the University of Victoria, the definition of complexity science is stated as "Complexity science is the study of a system. It is concerned with complex systems and problems that are dynamic, unpredictable, and multi-dimensional, consisting of a collection of interconnected relationships and parts" (Victoria, 2012). Healthcare can be best described as a complex system. From primary care to tertiary care it comprises many different sectors. It has many governing structures and practices to manage performance. The structural properties of the healthcare system in Ireland limit its adaptive capacity and alignment with complex system requirements for future integration. The complex system needs to become a complex adaptive system.

1-2-4-All is a technique developed by Henri Lipmanowicz and Keith Mc Candless. This type of structure allows for the participation of everyone in the group. Everyone is given an equal opportunity to contribute. The sequence of steps involves silent self-reflection for one minute, generate ideas in pairs for two minutes, building on ideas from self-reflection. Share and develop ideas from your pair in foursomes for four minutes, taking note of similarities and differences. The group then shares the ideas with everyone in the group for 5 minutes. This type of structure allows for every individual to engage, expands the diversity of input. The exercise is simple, results focused and inclusive.

The next section will discuss the results gathered using the research method discussed in this section.

# 5 Results

In chapter one, the overview of the research process outlines the steps taken to answer the research question. This chapter discusses the results gathered from conducting the focus groups, the validation of the UML activity diagram and the creation of a dietician, scientist, and patient personas. Finally, this section will address the data gathered and how it interacts with information modelling perspectives.

Three separate focus groups were held. The first focus group was with the Metabolic Scientists, a week later a session was held with Metabolic Dieticians and finally a group held with health informaticians. The liberating structure 1-2-4-all method was used to conduct the focus groups as discussed in the research methodology section.

As displayed in the overview of the research process, the UML activity diagram figure five created was validated by a Senior Metabolic Dietician and Principal Biochemist. After the focus groups and UML validation were completed, the literature review was repeated to link any findings to the literature, investigate the impact on standards and relate the data gathered back to answering motivations of the research. The results from the focus groups were illustrated and identities further categorised into table form. The dissemination of the results involved categorising the identities into three, patient, sample and other.

In Chapter six, synthesis of literature review and practical work, the results from chapter five were used to create a patient, scientist and dietician personas. Using the domain analysis of a PKU patient from figure nine and the UML activity diagram figure 5, use cases were created. Finally

## 5.1 Focus Group Results

Results from the focus groups have been illustrated in figures 16,17,18,19,20 and 21. These results were then categorised and tabulated in tables 3,4,5,6,7,8,9 and 10 to display the selected stakeholders in a combined view.

- MRN
- Surname
- Forename
- DOB
- Sex
- Gender ID

- MRN
- Sex
- Age
- Home Location
- Clinical Details

- Address
- Ward/Clinic
- Consultant
- External Hospital Address
- External Hospital Phone
- Requesting Doctor
- Bleep No
- Clinical Details
- Reason for request
- Medications/Antibiotics


- Reason for request
- Units
- Reference ranges
- Test Codes
- Quality Control
- Result

- Sample Collection Date
- Sample Collection Time
- Investigations Required
- Sample Type
- Fasting/Non-Fasting
- Priority -Routine/Urgent
- Lab Specimen Number
- Sample Received Date
- Sample Received Time
- Units
- Result
- Reference Ranges
- Test codes
- Test method

*Figure 16 Brainstorming diagram of Researcher Identities*

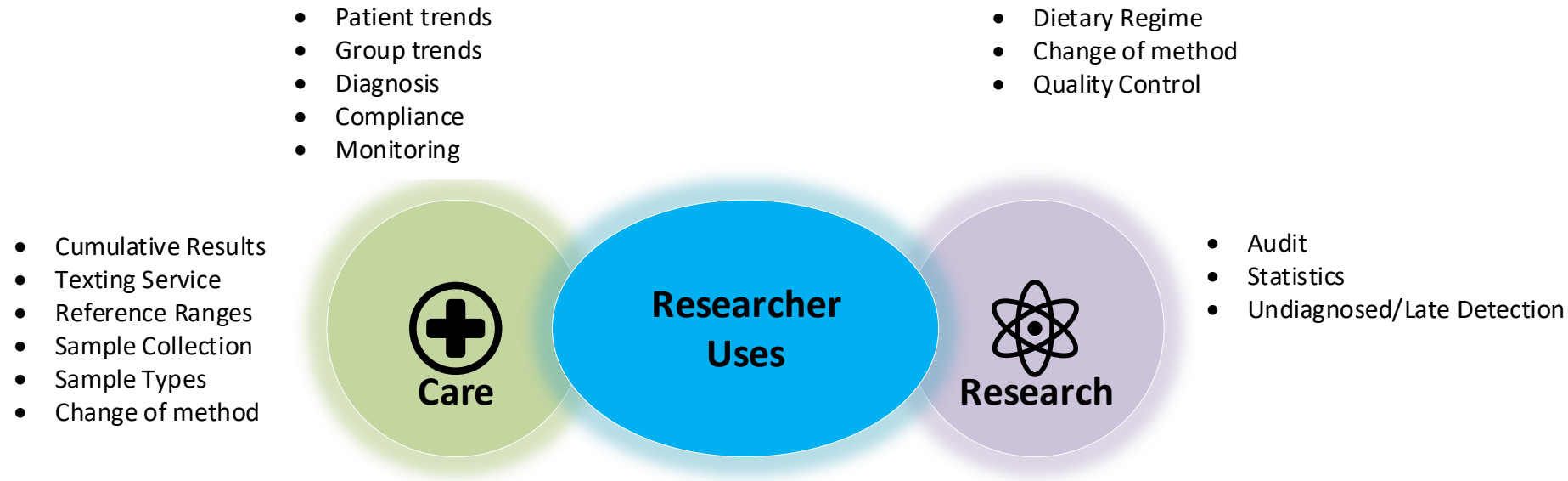



*Figure 17 Brainstorming diagram of Researcher Uses*

## 5.2 Brainstorming results from the researcher

Figure 16 and 17, illustrate the identities and uses respectively, identified by the researcher. These results were gathered after the creation of the domain analysis use case for the PKU patient seen in figure five. The results were categorised into care and research as addressed in the research question. Results were categorised again into patient, sample, and other identities, and cross-referenced with care and research. Focus groups were held with the scientists and the results can be seen in figure 18 and 19.

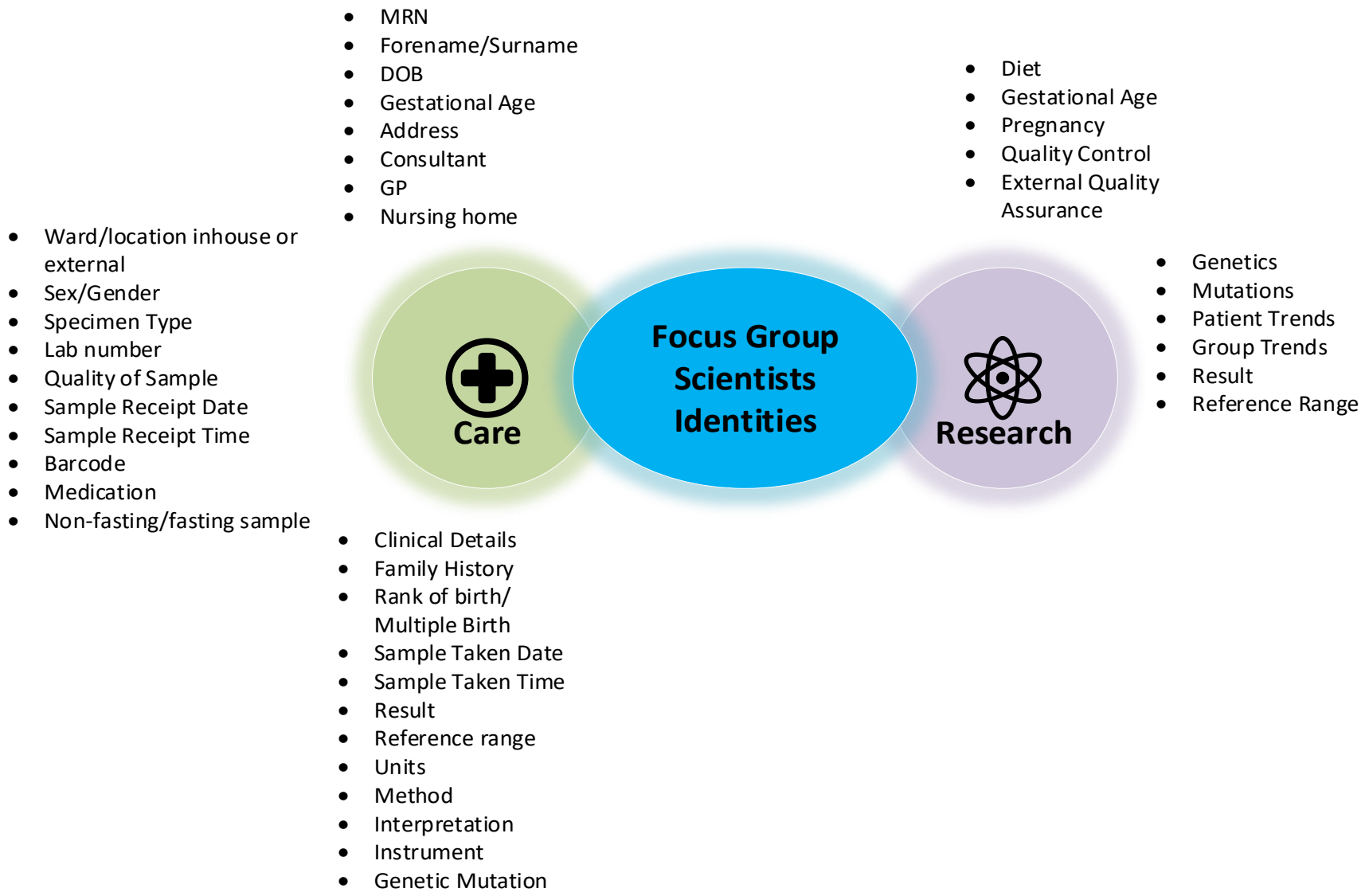



*Figure 18 Brainstorming diagram of Identities from Focus Group Results of Scientists*

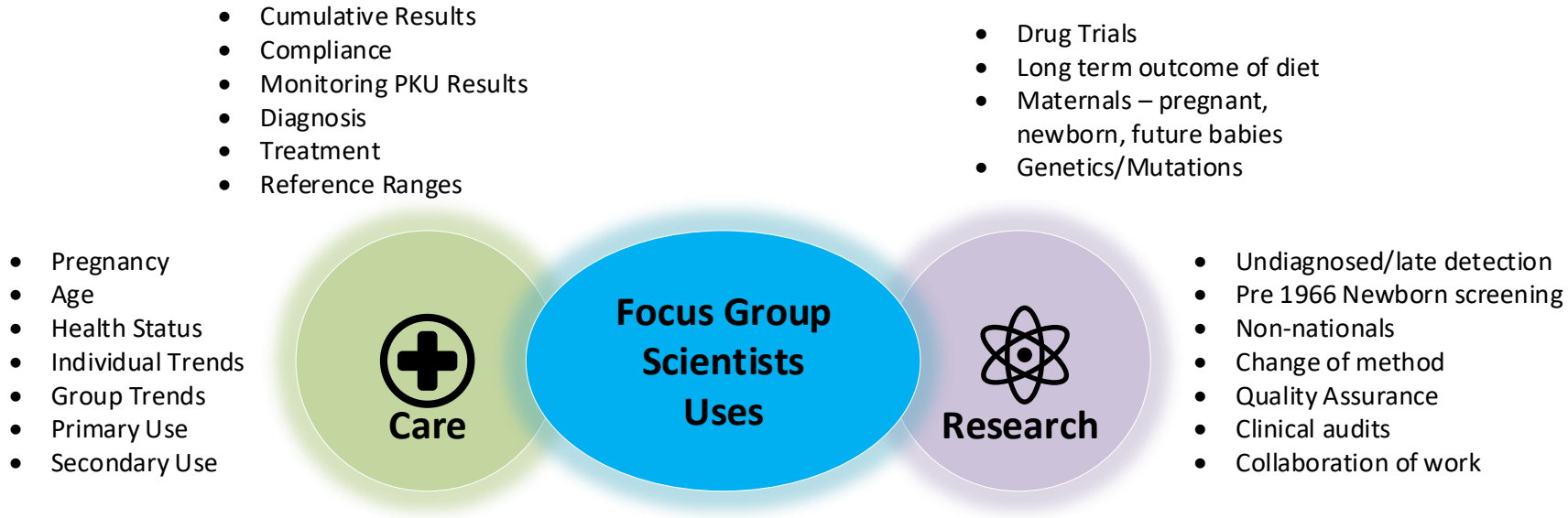



*Figure 19 Brainstorming diagram of uses from Focus Group Results of Scientists*

## 5.3 Brainstorming results from the scientists

Figure 18 and 19, illustrate the identities and uses respectively, identified by the scientists. These results were gathered after asking the following questions:

1) As Metabolic Scientists you generate, collect and store data. From your experience, what are uses of data for care and research of patients with PKU?
2) What are the identities you need to conduct those uses?

The results were categorised into care and research as addressed in the research question. Focus groups were then held with the dieticians and the results can be seen in figure 20 and 21.

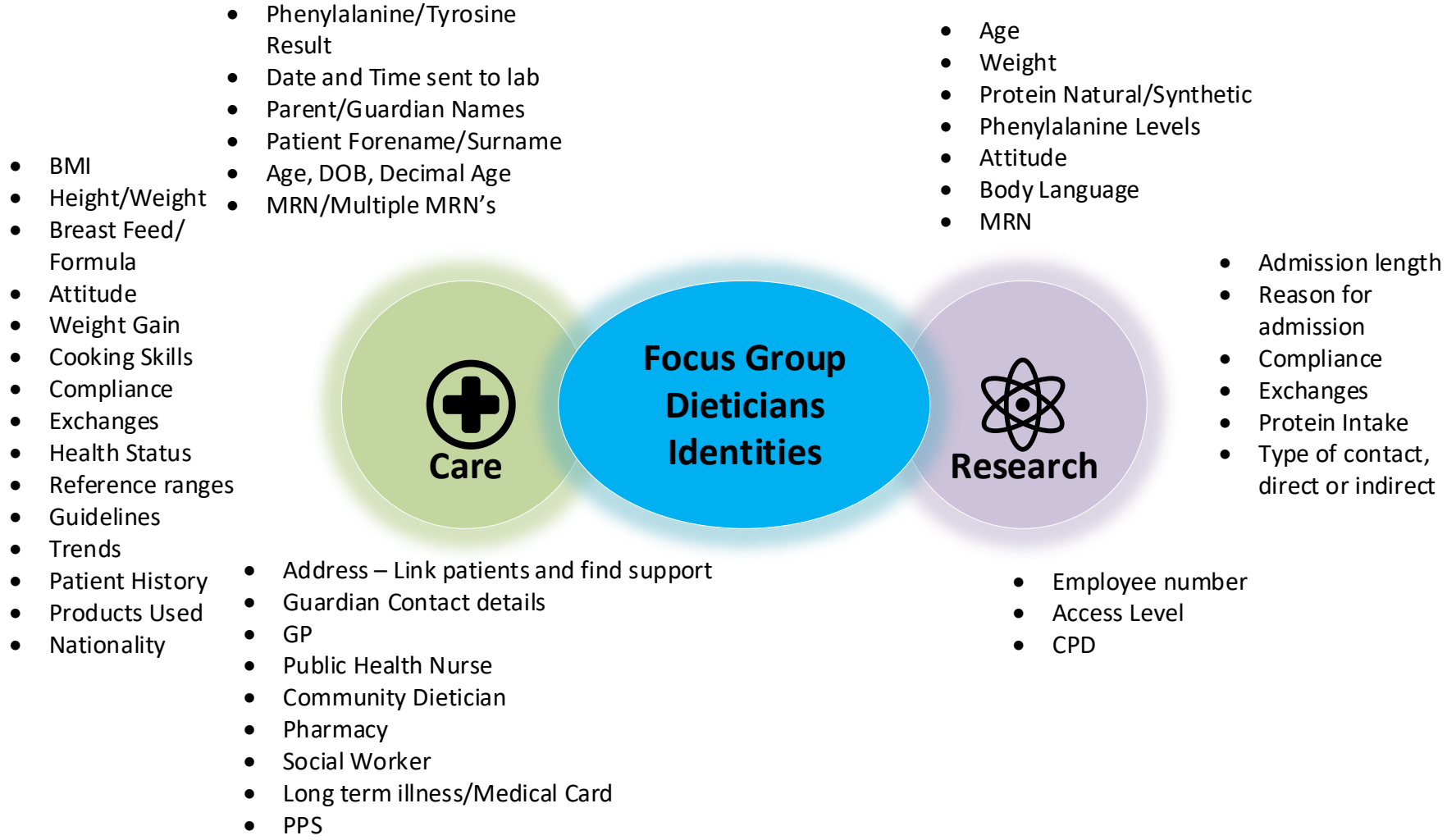



*Figure 20 Brainstorming diagram of Identities from Focus Group Results of Dieticians*

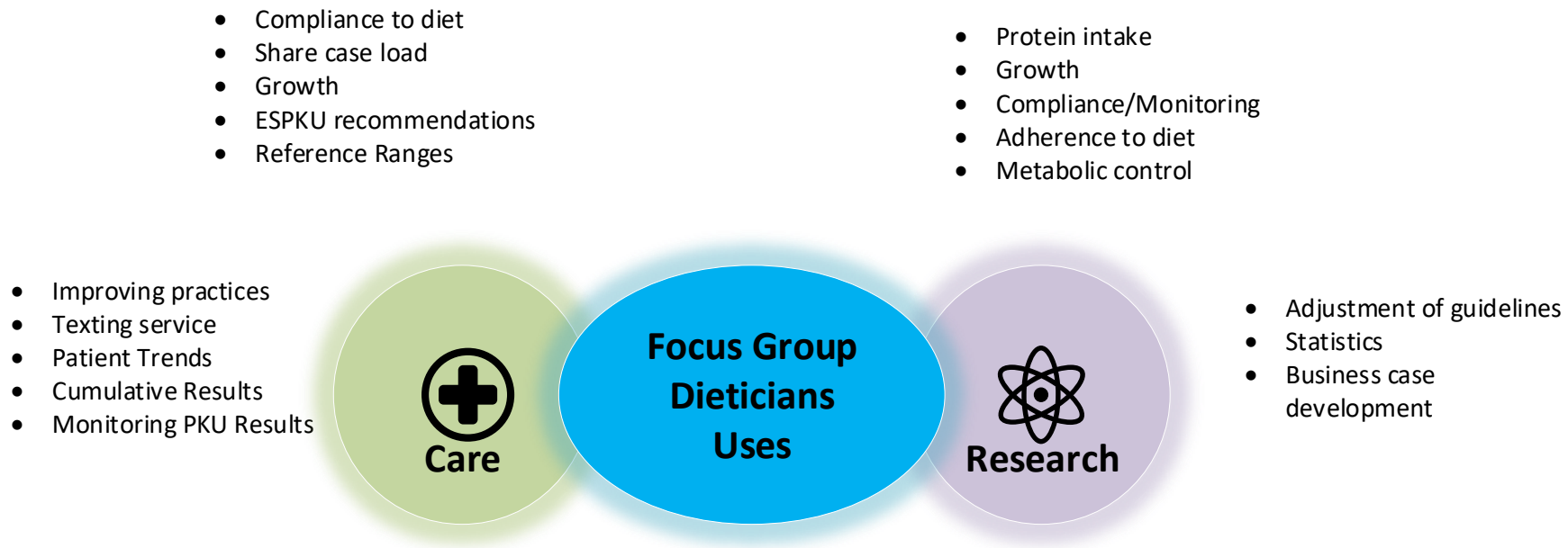



*Figure 21 Brainstorming diagram of uses from Focus Group Results of Dieticians*

## 5.4 Brainstorming results from the Dieticians

Figure 20 and 21, illustrate the identities and uses respectively, identified by the Dieticians. These results were gathered after asking the following questions:

1) As Metabolic Dieticians you generate, collect and store data. From your experience, what are uses of data for care and research of patients with PKU?
2) What are the identities you need to conduct those uses?

The results were categorised into care and research as addressed in the research question. These results from the researcher, scientists and dieticians were categorised and tabulated into a combined view.

*Table 3 Care uses cross referenced with patient identities from researcher, scientists, and dieticians*

| Patient Identities | MRN/ Hospital No. | Forename | Surname | Sex/ Gender ID | Date of Birth | Nationality | Decimal Age | Gestational Age | Rank of Birth | Address | Clinical Details | Height/ Weight | BMI | Attitude to disease management |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| **Care Uses** | | | | | | | | | | | | | | |
| Cumulative Results | R/S/D | R/S/D | R/S/D | R/S/D | R/S/D | S | D | S | S | R/S/D | R/S | D | D | D |
| Compliance/Monitoring | R/S/D | R/S/D | R/S/D | R/S/D | R/S/D | S | D | S | S | R/S/D | R/S | D | D | D |
| Diagnosis Information | R/S/D | R/S/D | R/S/D | R/S/D | R/S/D | S | D | S | S | R/S/D | R/S | D | D | D |
| Individual Patient Trends | R/S/D | R/S/D | R/S/D | R/S/D | R/S/D | S | D | S | S | R/S/D | R/S | D | D | D |
| Group Patient Trends | R/S/D | R/S/D | R/S/D | R/S/D | R/S/D | S | D | S | S | R/S/D | R/S | D | D | D |
| Treatment Regime | S/D | S/D | S/D | S/D | S/D | S | D | S | S | S/D | S | D | D | D |
| Current Health Status | S/D | S/D | S/D | S/D | S/D | S | D | S | S | S/D | S | D | D | D |
| Reference Ranges | R/D | R/D | R/D | R/S/D | R/S/D | - | D | - | - | R/D | R | D | D | D |
| Texting Results | R/D | R/D | R/D | R/D | R/D | - | D | - | - | R/D | R | D | D | D |
| Change of Method | R | R | R | R | R | - | - | - | - | R | R | - | - | - |
| Sample Collection Data | R | R | R | R | R | - | - | - | - | R | R | - | - | - |
| Sample Types | R | R | R | R | R | - | - | - | - | R | R | - | - | - |
| Pregnancy Status impact on care | S | S | S | S | S | S | - | S | S | S | S | - | - | - |
| Primary Care Use | S | S | S | S | S | S | - | S | S | S | S | - | - | - |
| Secondary Care Use | S | S | S | S | S | S | - | S | S | S | S | - | - | - |
| Age Profiling | S | S | S | S | S | S | - | S | S | S | S | - | - | - |
| Multi-discipline Team Case Load | D | D | D | D | D | - | D | - | - | D | - | - | - | - |
| Effect on Growth | D | D | D | D | D | - | D | - | - | D | - | D | D | D |
| Improving Practices | D | D | D | D | D | - | D | - | - | D | - | D | D | D |
| ESPKU Recommendations | D | D | D | D | D | - | D | - | - | D | - | D | D | D |

R = Researcher S = Scientist D = Dietician

## 5.5 Care Uses cross-referenced with patient identities

Table three illustrates the care use cases identified cross-referenced with the patient identities gathered from the focus groups. The letter R represents the researcher, S represents the Scientist and D represents the Dietician. These letters were entered in the tables to show which identities were required for each use care and by which stakeholder.

*Table 4 Care uses cross-referenced with sample identities from researcher, scientists, and dieticians*

| Sample Identities | Ward/Clinic | Consultant | External Hospital Address | External Hospital Phone | Non Hospital - Nursing Home | Priority – Routine/Urgent | Bleep No | Reason for Request | Medications/ Antibiotics | Sample Collection Date | Sample Collection Time | Investigations Required | Sample Type | Lab Specimen Number |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| **Care Uses** | | | | | | | | | | | | | | |
| Cumulative Results | R/S | R/S | R/S | R | S | R | R | R | R/S | R/S | R/S | R/S | R/S | S |
| Compliance/Monitoring | R/S | R/S | R/S | R | S | R | R | R | R/S | R/S | R/S | R/S | R/S | S |
| Diagnosis Information | R/S | R/S | R/S | R | S | R | R | R | R/S | R/S | R/S | R/S | R/S | S |
| Individual Patient Trends | R/S | R/S | R/S | R | S | R | R | R | R/S | R/S | R/S | R/S | R/S | S |
| Group Patient Trends | R/S | R/S | R/S | R | S | R | R | R | R/S | R/S | R/S | R/S | R/S | S |
| Treatment Regime | S | S | S | S | S | S | S | S | S | S | S | S | S | S |
| Current Health Status | S | S | S | - | S | - | - | - | S | S | S | S | S | S |
| Reference Ranges | R | R | R | R | S | R | R | R | R | R | R | R | R | - |
| Texting Results | R | R | R | R | S | R | R | R | R | R | R | R | R | - |
| Change of Method | R | R | R | R | S | R | R | R | R | R | R | R | R | - |
| Sample Collection Data | R | R | R | R | S | R | R | R | R | R | R | R | R | - |
| Sample Types | R | R | R | R | S | R | R | R | R | R | R | R | R | - |
| Pregnancy Status impact on care | S | S | S | - | S | - | - | - | S | S | S | S | S | S |
| Primary Care Use | S | S | S | - | S | - | - | - | S | S | S | S | S | S |
| Secondary Care Use | S | S | S | - | S | - | - | - | S | S | S | S | S | S |
| Age Profiling | - | - | - | - | - | - | - | - | S | S | S | S | S | S |
| Multi-discipline Team Case Load | - | - | - | - | - | - | - | - | - | - | - | - | - | - |
| Effect on Growth | - | - | - | - | - | - | - | - | - | - | - | - | - | - |
| Improving Practices | - | - | - | - | - | - | - | - | - | - | - | - | - | - |
| ESPKU Recommendations | - | - | - | - | - | - | - | - | - | - | - | - | - | - |

R = Researcher S = Scientist D = Dietician

*Table 5 Care uses cross-referenced with sample identities from researcher, scientists, and dieticians (Continued)*

| Sample Identities | Sample Received Date | Sample Received Time | Units | Result | Reference Range | Test Code | Test Method | Analyser/ Instrument |
|---|---|---|---|---|---|---|---|---|
| **Care Uses** | | | | | | | | |
| Cumulative Results | R/S | R/S | R/S | R/S/D | R/S/D | R | R/S | S |
| Compliance/Monitoring | R/S | R/S | R/S | R/S/D | R/S/D | R | R/S | S |
| Diagnosis Information | R/S | R/S | R/S | R/S/D | R/S/D | R | R/S | S |
| Individual Patient Trends | R/S | R/S | R/S | R/S/D | R/S/D | R | R/S | S |
| Group Patient Trends | R/S | R/S | R/S | R/S/D | R/S/D | R | R/S | S |
| Treatment Regime | S | S | S | S/D | S | - | - | S |
| Current Health Status | S | S | S | S/D | S | - | - | S |
| Reference Ranges | R | R | R/S | R/S/D | R/S/D | R/S | R/S | - |
| Texting Results | - | - | - | D | D | - | - | - |
| Change of Method | R | R | R | R | R | R | R/S | S |
| Sample Collection Data | R | R | R | R | R | R | R | - |
| Sample Types | | | R | R | R | R | R | - |
| Pregnancy Status impact on care | S | S | S | S | S | - | R/S | S |
| Primary Care Use | S | S | S | S | S | - | S | S |
| Secondary Care Use | S | S | S | S | S | - | S | S |
| Age Profiling | S | S | S | S | S | - | S | S |
| Multi-discipline Team Case Load | - | - | - | D | D | - | - | - |
| Effect on Growth | - | - | - | D | D | - | - | - |
| Improving Practices | - | - | - | D | D | - | - | - |
| ESPKU Recommendations | - | - | - | D | D | - | - | - |

R = Researcher S = Scientist D = Dietician

## 5.6 Care Uses cross-referenced with sample identities

Table four and five illustrate the care use cases identified cross-referenced with the sample identities gathered from the focus groups. The letter R represents the researcher, S represents the Scientist and D represents the Dietician. These letters were entered in the tables to show which identities were required for each use care and by which stakeholder.

*Table 6 Care uses cross-referenced with other identities from researcher, scientists, and dieticians*

| Other Identities | Feeding Method – Breast or Formula | Cooking Skills | Dietary Exchanges | Guardian Name | Guardian Contact Details | Medical Card/PPS No. |
|---|---|---|---|---|---|---|
| **Care Uses** | | | | | | |
| Cumulative Results | D | D | D | D | D | D |
| Compliance/Monitoring | - | - | - | - | - | - |
| Diagnosis Information | - | - | - | - | - | - |
| Individual Patient Trends | - | - | - | - | - | - |
| Group Patient Trends | - | - | - | - | - | - |
| Treatment Regime | - | - | - | - | - | - |
| Current Health Status | - | - | - | - | - | - |
| Reference Ranges | - | - | - | - | - | - |
| Texting Results | D | D | D | D | D | D |
| Change of Method | - | - | - | - | - | - |
| Sample Collection Data | - | - | - | - | - | - |
| Sample Types | - | - | - | - | - | - |
| Pregnancy Status impact on care | - | - | - | - | - | - |
| Primary Care Use | - | - | - | - | - | - |
| Secondary Care Use | - | - | - | - | - | - |
| Age Profiling | - | - | - | - | - | - |
| Multi-discipline Team Case Load | - | - | - | - | - | - |
| Effect on Growth | D | D | D | D | D | D |
| Improving Practices | D | D | D | D | D | D |
| ESPKU Recommendations | D | D | D | D | D | D |

R = Researcher S = Scientist D = Dietician

## 5.7 Care Uses cross-referenced with other identities

Table six illustrates the care use cases identified cross-referenced with the other identities gathered from the focus groups. The letter R represents the researcher, S represents the Scientist and D represents the Dietician. These letters were entered in the tables to show which identities were required for each use care and by which stakeholder.

*Table 7 Research uses cross-referenced with patient identities from researcher, scientists, and dieticians*

| Patient Identities | MRN/ Hospital No. | Forename | Surname | Sex/ Gender ID | Date of Birth | Nationality | Decimal Age | Gestational Age | Rank of Birth | Address | Clinical Details | Height/ Weight | BMI |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| **Research Uses** | | | | | | | | | | | | | |
| Growth | D | D | D | D | D | D | D | - | D | - | D | D | D |
| Reference Ranges | S | S | S | S | S | - | - | S | S | S | - | - | - |
| Business Case Development | - | - | - | - | - | - | - | - | - | - | - | - | - |
| Change of Method | - | - | - | R/S | R/S | - | - | S | - | R/S | - | - | - |
| Dietary Regime | R | R | R | R | R | - | - | - | R | R | - | - | - |
| Audit | R/S | R/S | R/S | R/S | R/S | - | R | R/S | R/S | R/S | - | - | - |
| Statistics | | | | R/D | R/D | - | R/D | - | R/D | R | - | - | - |
| Undiagnosed/Late Detection | R/S | R/S | R/S | R/S | R/S | - | R | R/S | R/S | R/S | - | - | - |
| Pre-1966 Newborn Screening | S | S | S | S | S | - | - | S | S | S | - | - | - |
| Non-Nationals | S | S | S | S | S | - | - | S | S | S | - | - | - |
| Drug Trials | S | S | S | S | S | - | - | S | S | S | - | - | - |
| Maternal – Pregnant/Newborn/Future | S | S | S | S | S | - | - | S | S | S | - | - | - |
| Long Term Outcome of Diet | S | S | S | S | S | - | - | S | S | S | - | - | - |
| Quality Assurance/Quality Control | - | - | - | R/S | R/S | - | R | R/S | | R | - | - | - |
| Genetics/Mutations | S | S | S | S | S | - | - | S | S | S | - | - | - |
| Collaboration of Work | S | S | S | S | S | - | - | S | S | S | - | - | - |
| Protein Intake | D | D | D | D | D | - | D | - | D | - | D | D | D |
| Compliance/Adherence to Diet | D | D | D | D | D | - | D | - | D | - | D | D | D |
| Metabolic Control | D | D | D | D | D | - | D | - | D | - | D | D | D |
| Adjustment of Guidelines | D | D | D | D | D | - | D | - | D | - | D | D | D |

R = Researcher S = Scientist D = Dietician

## 5.8 Research Uses cross-referenced with patient identities

Table seven illustrates the research use cases identified cross-referenced with the patient identities gathered from the focus groups. The letter R represents the researcher, S represents the Scientist and D represents the Dietician. These letters were entered in the table to show which identities were required for each use care and by which stakeholder.

*Table 8 Research uses cross-referenced with sample identities from researcher, scientists, and dieticians*

| Sample Identities | Ward/Clinic | Consultant | External Hospital Address | External Hospital Phone | Non Hospital - Nursing Home | Priority – Routine/Urgent | Bleep No | Reason for Request | Medications/ Antibiotics | Sample Collection Date | Sample Collection Time | Investigations Required | Sample Type |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| **Research Uses** | | | | | | | | | | | | | |
| Growth | - | - | - | - | - | - | - | - | - | - | - | - | - |
| Reference Ranges | - | - | - | - | - | - | - | - | - | - | - | R/S | R/S |
| Business Case Development | - | - | - | - | - | - | - | - | - | - | - | - | - |
| Change of Method | - | - | - | - | - | - | - | - | S | S | S | R | R/S |
| Dietary Regime | R | R | R | R | - | - | - | R | R | R | R | R | R |
| Audit | R/S | R/S | R/S | R | - | R | R | R | R/S | R/S | R/S | R | R/S |
| Statistics | R | R | R | R | - | R | R | R | R | R | R | R | R |
| Undiagnosed/Late Detection | R/S | R/S | R/S | R | - | R | R | R | R/S | R/S | R/S | R | R/S |
| Pre-1966 Newborn Screening | S | S | S | - | - | - | - | - | S | S | S | - | S |
| Non-Nationals | S | S | S | - | - | - | - | - | S | S | S | - | S |
| Drug Trials | S | S | S | - | S | - | - | - | S | S | S | - | S |
| Maternal – Pregnant/Newborn/Future | S | S | S | - | S | - | - | - | S | S | S | - | S |
| Long Term Outcome of Diet | S | S | S | - | S | - | - | - | S | S | S | - | S |
| Quality Assurance/Quality Control | S | S | S | - | S | - | - | - | S | S | S | R | R/S |
| Genetics/Mutations | S | S | S | - | S | - | - | - | S | S | S | - | S |
| Collaboration of Work | S | S | S | - | S | - | - | - | S | S | S | - | S |
| Protein Intake | - | - | - | - | - | - | - | - | - | - | - | - | - |
| Compliance/Adherence to Diet | - | - | - | - | - | - | - | - | - | - | - | - | - |
| Metabolic Control | - | - | - | - | - | - | - | - | - | - | - | - | - |
| Adjustment of Guidelines | - | - | - | - | - | - | - | - | - | - | - | - | - |

R = Researcher S = Scientist D = Dietician

*Table 9 Research uses cross-referenced with sample identities from researcher, scientists, and dieticians (Continued)*

| Sample Identities | Sample Received Date | Sample Received Time | Units | Result | Reference Range | Test Code | Test Method | Analyser/ Instrument |
|---|---|---|---|---|---|---|---|---|
| **Research Uses** | | | | | | | | |
| *Growth* | - | - | - | - | - | - | - | - |
| *Reference Ranges* | R | R | R | R | R | R | R | R |
| *Business Case Development* | - | - | - | - | - | - | - | - |
| *Change of Method* | - | - | R/S | R/S | R/S | R | R | R |
| *Dietary Regime* | R | R | R | R | R | R | R | R |
| *Audit* | R/S | R/S | R/S | R/S | R/S | R | R | R |
| *Statistics* | R | R | R | R/D | R | R | R | R |
| *Undiagnosed/Late Detection* | R/S | R/S | R/S | R/S | R/S | R | R | R |
| *Pre-1966 Newborn Screening* | S | S | S | S | S | - | - | - |
| *Non-Nationals* | S | S | S | S | S | - | - | - |
| *Drug Trials* | S | S | S | S | S | - | - | - |
| *Maternal – Pregnant/Newborn/Future* | S | S | S | S | S | - | - | - |
| *Long Term Outcome of Diet* | S | S | S | S | S | - | - | - |
| *Quality Assurance/Quality Control* | S | S | R/S | R/S | R/S | R | R | R |
| *Genetics/Mutations* | S | S | S | S | | - | - | - |
| *Collaboration of Work* | - | - | S | S | S | - | - | - |
| *Protein Intake* | - | - | - | D | - | - | - | - |
| *Compliance/Adherence to Diet* | - | - | - | D | - | - | - | - |
| *Metabolic Control* | - | - | - | D | - | - | - | - |
| *Adjustment of Guidelines* | - | - | - | D | - | - | - | - |

R = Researcher S = Scientist D = Dietician

## 5.9 Research Uses cross-referenced with sample identities

Table eight and nine illustrate the research use cases identified cross-referenced with the sample identities gathered from the focus groups. The letter R represents the researcher, S represents the Scientist and D represents the Dietician. These letters were entered in the tables to show which identities were required for each use care and by which stakeholder.

*Table 10 Research uses cross-referenced with other identities from researcher, scientists, and dieticians*

| Other Identities | Breast Feed/Formula | Cooking Skills | Exchanges | Guardian Name | Guardian Contact Details |
|---|---|---|---|---|---|
| **Research Uses** | | | | | |
| Growth | D | D | D | D | D |
| Reference Ranges | - | - | - | - | - |
| Business Case Development | D | D | D | D | D |
| Change of Method | - | - | - | - | - |
| Dietary Regime | - | - | - | - | - |
| Audit | - | - | - | - | - |
| Statistics | D | D | D | D | D |
| Undiagnosed/Late Detection | - | - | - | - | - |
| Pre-1966 Newborn Screening | - | - | - | - | - |
| Non-Nationals | - | - | - | - | - |
| Drug Trials | - | - | - | - | - |
| Maternal – Pregnant/Newborn/Future | - | - | - | - | - |
| Long Term Outcome of Diet | - | - | - | - | - |
| Quality Assurance/Quality Control | - | - | - | - | - |
| Genetics/Mutations | - | - | - | - | - |
| Collaboration of Work | - | - | - | - | - |
| Protein Intake | D | D | D | D | D |
| Compliance/Adherence to Diet | D | D | D | D | D |
| Metabolic Control | D | D | D | D | D |
| Adjustment of Guidelines | D | D | D | - | - |

R = Researcher S = Scientist D = Dietician

## 5.10 Research Uses cross-referenced with other identities

Table ten illustrates the research use cases identified cross-referenced with the other identities gathered from the focus groups. The letter R represents the researcher, S represents the Scientist and D represents the Dietician. These letters were entered in the tables to show which identities were required for each use care and by which stakeholder.

## 5.11 Validation of UML Activity Diagram

Senior Metabolic Dietician and Principal biochemist were asked separately to consider and comment on the UML activity diagram for the monitoring use case for a PKU patient. The Senior Metabolic Dietician and the Principal Biochemist stated that “The diagram contained all the participants in the use case”. They agreed with the use cases represented.

## 5.12 Results from focus group with Health Informaticians

A focus group was held with two health informaticians using the 1-2-4-all liberating structure to ask their perspective on the research gathered. They stated that the linking between systems with integrable data was a key point. A generalised model is a way to approach this research topic. A model could provide a structure for easily exchanging data syntax and improve the efficiency of accessing data. The model would help with flexibility and reusability. There is a danger that without a generalised model that the implementation could be vendor locked and this could lessen the innovation required for progression. The goal is to capture the end user benefits which will lead to patient benefits.

## 5.13 Outcomes from the research process

Table 11 displays the outcomes from each stage of the research process. Each outcome contributed to the practical work in chapter six.

*Table 11 Outcomes from the research process*

| ***Stage*** | **Description** | **Outcome** |
|---|---|---|
| *Stage 1* | Research question | Question defined focusing on identification from an information modelling perspective using a patient with PKU |
| *Stage 2* | Create Domain Analysis of PKU Use Case | Key stakeholders identified |
| *Stage 3* | Conduct Literature Review | Strategies and themes identified linked to the research question |
| *Stage 4* | Apply for Ethical Approval Temple Street Children's University Hospital | Apply for Trinity Ethics |
| *Stage 5* | Apply for Ethical Approval Trinity College Dublin | Perform research |
| *Stage 6* | Conduct Focus Groups | Identities and uses |
| *Stage 7* | Validation of UML Activity Diagram | Validation of the use cases |
| *Stage 8* | Literature Review Repeated | Reviewing the linkage to the research topic after the focus groups |
| *Stage 9* | Create Personas | Personas could be used for future work |
| *Stage 10* | Create Use Cases | Use cases could be used for future work |
| *Stage 11* | Review data, standards and information modelling perspective. | The data gathered could influence standards and show the importance and role of identities in information modelling |

# 6 Synthesis of Literature Review and Practical Work

This chapter reflects on the practical work performed from the results generated in the previous chapter. Each of the elements created from the research process is synthesised with the literature reviewed in chapter two. The synthesis resulted in the creation of PKU patient persona and scientist and dietician personas, UML activity diagram of monitoring use case and from the UML, use cases were created according to the Antilope project template. In figure 22 and 23, the use case of monitoring has been selected to demonstrate how other use cases can be broken down. This use case was selected, as it was common to both stakeholders selected.

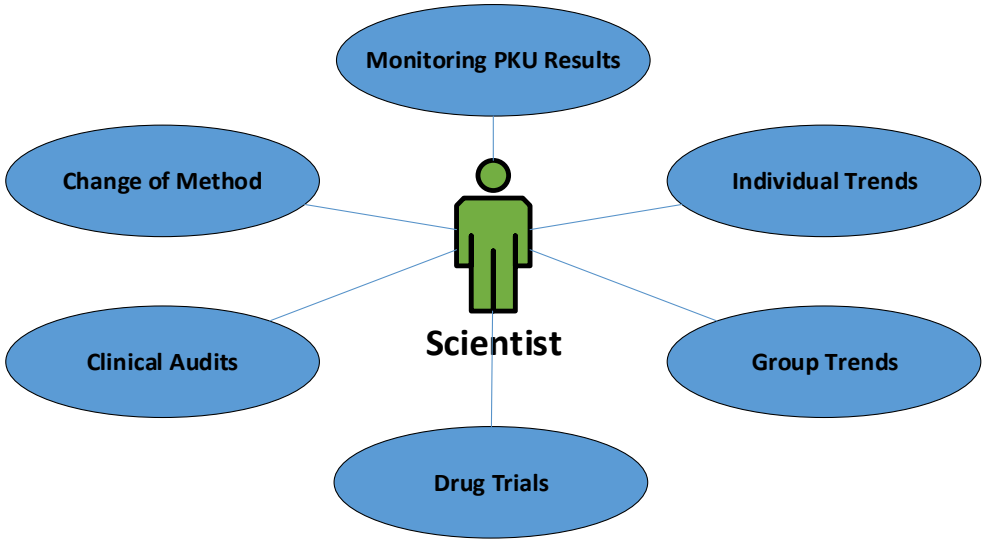



*Figure 22 Selection of Use Cases from Scientist Stakeholder*

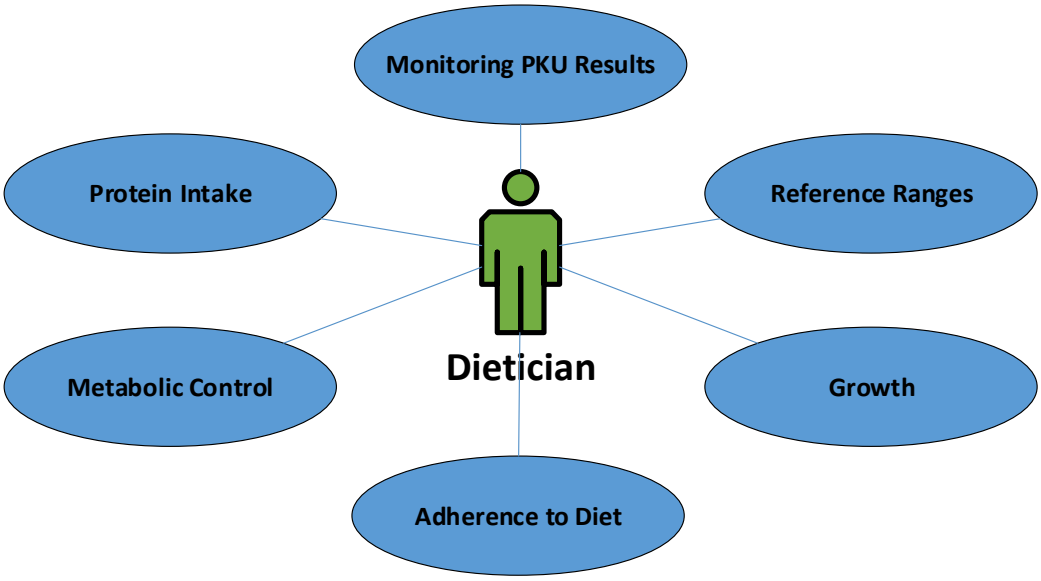
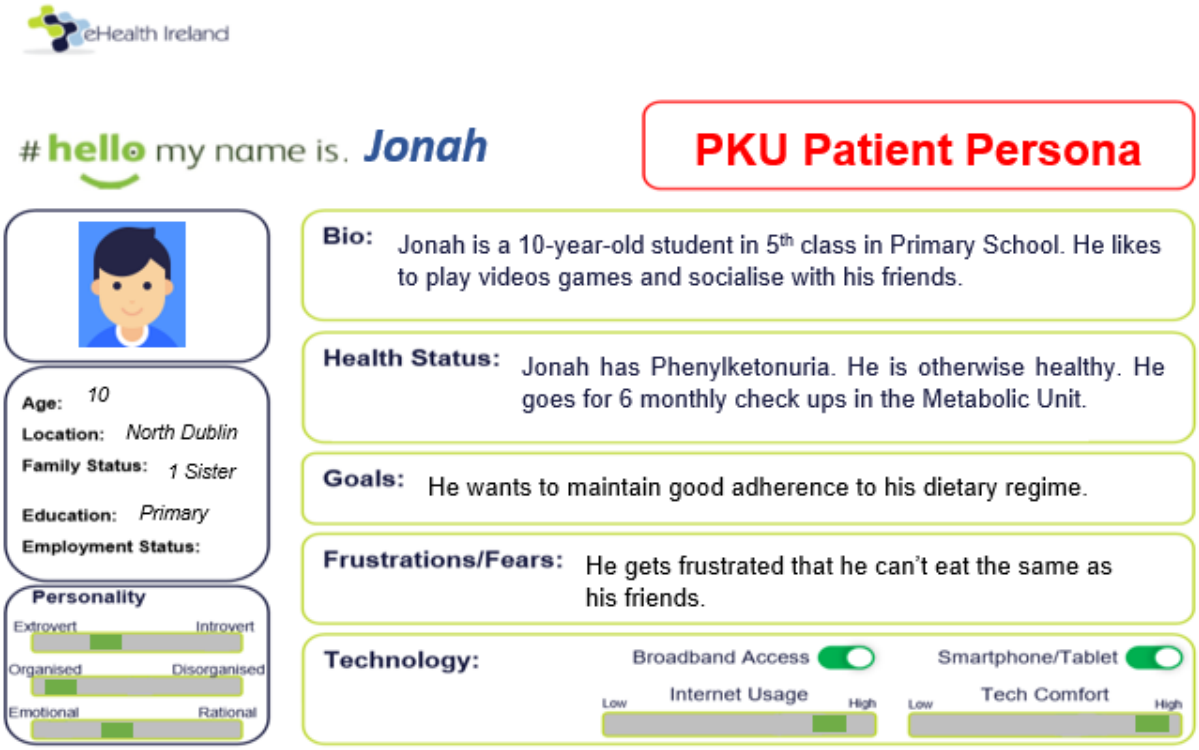



*Figure 23 Selection of Use Cases from Dietician Stakeholder*

## 6.1 Personas

As part of the electronic health record programme, personas and scenarios were created. These personas will illustrate want people want from future health systems. They will form a building block for the National EHR programme and clinical strategy programme. Together the personas and scenarios will create a vision to help enable a better healthcare in Ireland. Figure 24,25 and 26 were created using the sample template from eHealth Ireland. Each persona represents a key stakeholder in the monitoring use case of a patient with PKU.




**Scenario:** Patient diagnosed with Phenylketonuria (PKU)

**Actors:** Patient (Jonah Smith), Guardian (Mary Smith), Consultant (Dr X), Dietician (Anne)

Jonah Smith, 10, attends National Centre for Inherited Metabolic Disorders, Temple Street Children's University Hospital for a 6 Monthly review.

Each visit Jonah has a clinical nutritional assessment, which involves the recording of anthropometric parameters (weight, height, BMI) and is checked for clinical features of micronutrient and Phenylalanine deficiency.

Jonah and Mary send in samples fortnightly to check his Phenylalanine levels. He has Plasma Amino Acids checked annually. There is a biochemical nutritional assessment done annually. This involves measuring plasma homocysteine or methylmalonic acid, or both, haemoglobin, mean corpuscular volume and ferritin.

Jonah does not currently require neurocognitive or neurological examination. His memory, inhibitory and motor control are excellent.

There is a discussion between Jonah, Mary and the consultant about any behaviour issues, psychosocial functioning and wellbeing. There is no requirement for Psychiatric examination currently.

Jonah and Mary discuss the Phenylalanine levels with the dietician Sam and recommends that Jonah stay on his current dietary regime until further instructed. Jonah expresses he is finding it difficult to see friends eating foods that may affect his adherence. Mary will receive a text message with the results once approved by the dietician on the defero texting system.

*Figure 24 PKU Patient Persona*

eHealth Ireland

# hello my name is. Geraldine

## Metabolic Scientist Persona

**Age:** 40
**Location:** Dublin
**Family Status:** Married
**Education:** Masters
**Employment Status:**

**Personality**

Extrovert — Introvert
Organised — Disorganised
Emotional — Rational

**Bio:** Geraldine is 40 years old and has a family with two teenage kids. She works part-time. She has a strong work ethic and organisation skills.

**Role:** Works as Metabolic Scientist. Manages maintenance of analysers, processing and resulting samples.

**Goals:** To deliver the correct result for the correct patient, maintain a good work life balance

**Frustrations/Fears:** Incorrect result to wrong patient

**Technology:**
Broadband Access
Smartphone/Tablet
Internet Usage (Low — High)
Tech Comfort (Low — High)

**Scenario:** A Day in the life of a Metabolic Scientist

**Actors:** Metabolic Scientist (Geraldine), Patient (Jonah), Clerical Officer

On Monday morning Geraldine arrives into the laboratory. She checks her roster to see which analyser/bench she will be working on today. She is on the PKU Dried blood spot bench. Geraldine plans her day, firstly checking the maintenance schedule for the Mass Spectrometer Analyser. She performs the full weekly maintenance check list for the analyser in preparation for performing tests in the afternoon.

Samples arrive into the laboratory throughout the morning. Geraldine receives the samples including Jonah's, opens them, and date stamps each one with the collection date and collection time. She check the patient demographics are provided on each sample. Each specimen gets the application of laboratory specimen number before it is delivered to the laboratory office for Order Entry by Clerical Officers.

Once samples are returned to the laboratory, sample are prepared for analysis following a standard operating procedure (SOP). The SOP is a step by step guide for performing the preparation, analysis, resulting and authorisation of PKU results.

At 2pm the samples are analysed, and results are generated. Geraldine processes the results to be transmitted on the LIS interface. Numerical values are transferred into the LIS. Results are reviewed by a second checker, usually another scientist.

Results are then authorised and are viewable for healthcare providers to access the LIS. Geraldine can now clean the preparation area, ready for the next day.

*Figure 25 Scientist Persona*

eHealth Ireland

# hello my name is. Anne

## Metabolic Dietician Persona

**Age:** 40
**Location:** Dublin
**Family Status:**
**Education:** Masters
**Employment Status:**

**Personality**

Extrovert — Introvert
Organised — Disorganised
Emotional — Rational

**Bio:** Anne is a Senior Metabolic Dietician. She is very dedicated to her work. She has experience in metabolic disorders.

**Role:** Anne trains and supports other dieticians in the department. She is part of the MDT for metabolic patients.

**Goals:** Deliver the best care to metabolic patients. Be part of patient EHR

**Frustrations/Fears:** Manual processes, time taken away from patient care, IT systems don't fit to needs.

**Technology:**

Broadband Access
Internet Usage (Low — High)
Smartphone/Tablet
Tech Comfort (Low — High)

**Scenario:** A day in the life of Metabolic Dietician – reviewing PKU results

**Actors:** Senior Metabolic Dietician (Anne), Patient (Jonah), Guardian (Mary)

Anne arrives for work at 8am. She checks the patient summary sheet for attendances today. She manually makes notes and comments to discuss with each patient and guardian. A meeting is held with the multidisciplinary team (MDT) to plan for the OPD appointments scheduled today.

Patients arrive throughout the morning and afternoon and are seen by metabolic consultants, registrars, healthcare assistants, nurse specialists.

Anne sees patients Jonah and mother Mary in the Metabolic Unit. She discusses how good Jonah's adherence is to his diet which is reflected in this Phenylalanine levels. Jonah expresses he is finding it difficult when eating with his peers. Anne reassures him that maintaining his compliance and continued monitoring will be of great benefit to his overall health status.

Once clinic is completed, Anne opens the defero texting system. She reviews and transmits most results to the guardian mobiles by text message. There are 3 results that require contacting the guardian via phone call to discuss.

Anne finds the manual aspects of transcribing patient results to different systems difficult and time consuming and is fraught with possible human error. She would like the metabolic unit to have an electronic health record for each patient. The IT systems currently used are not fit for purpose.

*Figure 26 Dietician Persona*

## 6.2 UML Activity Diagram

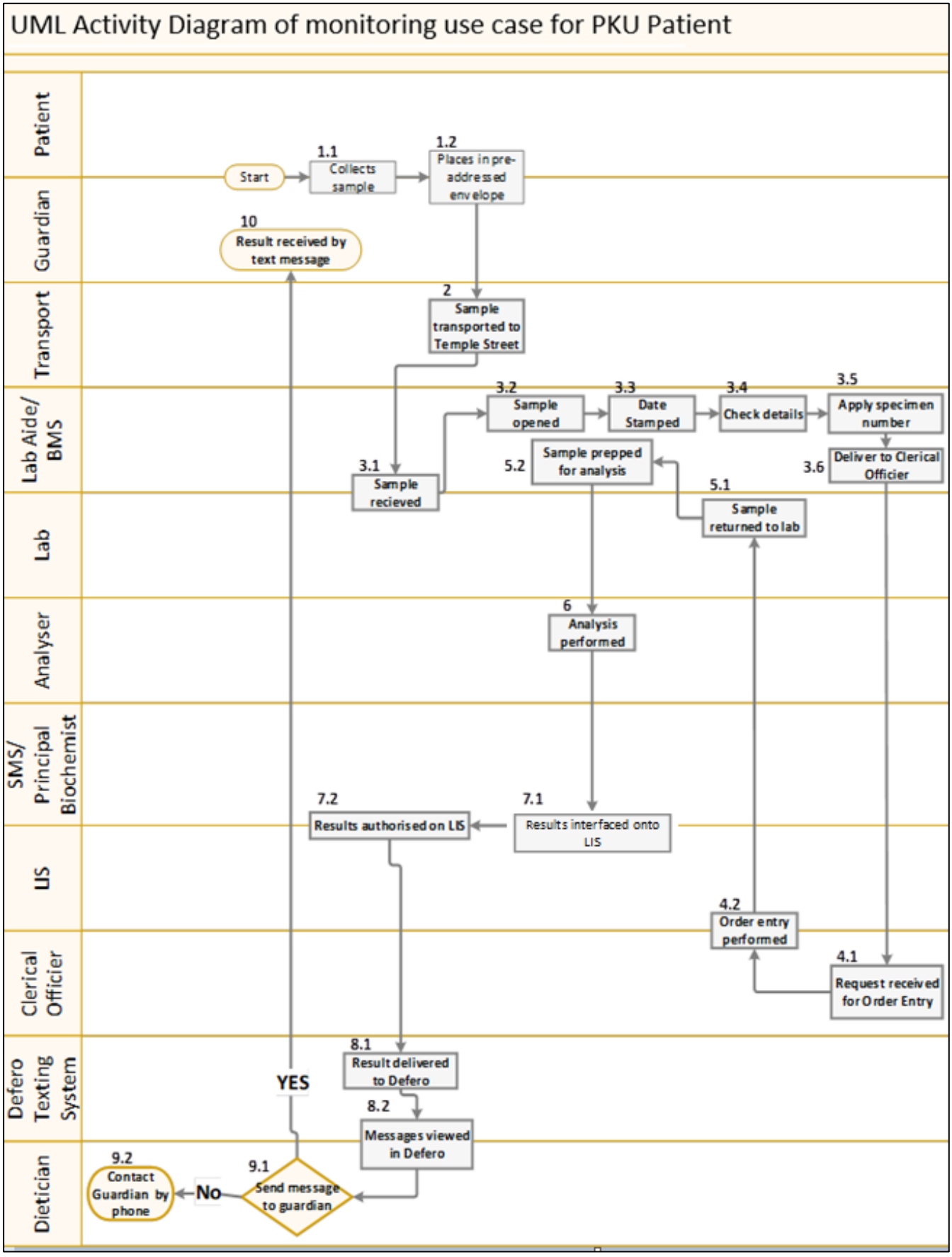



*Figure 27 UML activity diagram of monitoring use case for PKU Patient*

The UML activity diagram was created to provide a clear overview of the stakeholders and use cases involved the monitoring use case. Each of the use cases can be seen in a cross-function manner to display the involvement of each stakeholder. Each use case is numbered to link to the next section., where each use case is designed from the Antilope project template.

## 6.3 Monitoring Use for PKU patient

This section contains use cases created from the UML activity diagram figure 26, of the monitoring use of a patient with PKU. The use cases that follow were designed on the template from the Antilope Project Use Case Repository. The template has been modified to include identities. The use cases created in the monitoring use are important because they will provide a platform and structure for future use cases.

Use Case: 1.1/1.2 Sample Collection/Placed into Pre-addressed Envelope

**Relevance:** Clinical diagnosis and monitoring of Phenylketonuria (PKU) and other diseases are performed in a specialised laboratory service located in National Centre for Inherited Metabolic Disorders, Temple Street Children's University Hospital. Patients are directed by healthcare providers to collect samples at intervals (e.g. weekly) depending on their health status. Samples are collected by patients or from patients by guardians. Results determine treatment plans and are an indicator of adherence to treatment plans. This use case should ensure the correct steps are followed for the collection of the sample, application of patient demographics, data regarding sample collection are supplied to the laboratory and the sample is correctly packaged for transportation to the laboratory for analysis. The risk with incorrect collection, wrong patient demographics, incorrect sample collection data and improper packaging of the sample for transportation will prevent consistency of data. If the steps are correctly followed and supplied this will ensure a timely, complete and consistent patient information within the hospital organisation promoting good data quality.

**Domain:** Participatory healthcare

**Context:** Not applicable

**Scale:** Patient

**Systems:** Patient Administration System

**Information:** Sample Collection, Laboratory Request/Sample Form

**Participants:** Patient, Guardian, Metabolic Dietician

**Identities:** Patient Forename, Patient Surname, MRN, DOB, Address, Clinic, Consultant, Sample Collection Date, Investigations Required, Sample Type, Address for Metabolic Laboratory

**Process Flow:**

1. Metabolic dietician informs patient/guardian of the frequency of testing.
2. Patient/Guardian get Guthrie card to collect sample.
3. Ensure finger is warm.
4. Cleanse.
5. Dry.
6. Squeeze skin taut.
7. Puncture finger with lancet.
8. Apply to centre of circle on Guthrie card.
9. Repeat steps to fill next three circles.
10. Allow blood to dry at room temperature for at least three hours.
11. Place addressograph containing patient demographics on the card.
12. Fill in the date of collection on the card.
13. Place card in Tyvex envelope.
14. Affix pre-addressed label and stamp to envelope and post.

Use Case: 2. Sample transported to Temple Street

**Relevance:** Transportation of samples in the correct manner to be received by the National Centre for Inherited Metabolic Disorders, Temple street for analysis. Patients are directed to package and post samples following the guidelines. This use case should be used to ensure the samples are correctly transported. The risk with incorrect transportation could cause an effect on the analysis of the sample or if the sample is lost in transportation, it could affect the treatment plan for the patient. Correct transportation will result in the patient having complete consistent patient results within the hospital organisation.

**Domain:** Transportation

**Context:** Not applicable

**Scale:** Inter-organisational

**Systems:** Post

**Information:** Sample Packaging, Transportation by Post

**Participants:** Patient, Guardian, Transportation by Post

**Identities:** Address of Metabolic Laboratory

**Process Flow:**

1. Pre-addressed envelope collected by post.
2. Pre-addressed envelope processed by post.
3. Pre-addressed envelope transported by post.

Use Case: 3.1/3.2/3.3/3.4. Sample Received/Opened/Date Stamped/Details Checked

**Relevance:** Sample are received by post daily, Monday to Friday. Patient demographics and sample quality are checked pre-analysis. This use case should be used for checking patient demographics and sample quality. Following the SOP will ensure the necessary steps are taken to ensure correct patient samples are analysed by using patient information provided and results are generated in a timely manner within the hospital organisation.

**Domain:** Laboratory

**Context:** Not applicable

**Scale:** Intra-organisational

**Systems:** No systems involved

**Information:** Sample request received/opened, Sample date stamped with the time of receipt, Check patient demographics and sample details

**Participants:** Transportation by Post, Laboratory, Laboratory Aide/Basic Medical Scientist

**Identities:** Patient Forename, Patient Surname, MRN, DOB, Address, Clinic, Consultant, Sample Collection Date, Investigations Required, Sample Received Date, Sample Received Time, Sample Type

**Process Flow:**

1. Pre-addressed envelope received into Laboratory at Temple Street Hospital.
2. Pre-addressed envelope opened by Laboratory Aide/Basic Medical Scientist.
3. Sample date stamped by Laboratory Aide/Basic Medical Scientist.
4. Patient demographics checked by Laboratory Aide/Basic Medical Scientist.

Use Case: 3.5/3.6. Specimen number applied/Delivered to Clerical Officer

**Relevance:** The application of a specimen number by Laboratory Aides/Basic Medical Scientist adds additional data to the sample. The specimen number is required for entry in the LIS. The specimen number provides an additional unique identifier to the sample and links patient demographics to that sample. This use case should ensure the correct application of a unique specimen number to assist with providing the correct result for a patient for a specific collection date. The addition of a specimen number to the patient sample, linked to the patient demographics in the LIS, will provide complete data quality and traceability for the specimen when results are reviewed by clinical teams.

**Domain:** Laboratory

**Context:** Not applicable

**Scale:** Intra-organisational

**Systems:** No systems involved

**Information:** Specimen number applied to sample, Sample delivered to clerical officer

**Participants:** Laboratory Aide/Basic Medical Scientist, Clerical Officer

**Identities:** Patient Forename, Patient Surname, MRN, DOB, Address, Clinic, Consultant, Sample Collection Date, Investigations Required, Sample Received Date, Sample Received Time, Sample Type, Laboratory Specimen Number, Test Code

**Process Flow:**

1. Specimen number applied by Laboratory Aide/Basic Medical Scientist.
2. Test Code written on request.
3. Sample is delivered to the Clerical Officer.

Use Case: 4. Request received for Order Entry/Order Entry performed in LIS

**Relevance:** Sample requests are performed by clerical officers in the laboratory office. Entry of patient demographics is required. These details are provided on the addressograph added by the patient after the collection of the sample. Sample details are also recorded. Specimen number, date and time of collection are required data items for order entry in the LIS. The clerical officer will enter the tests to be performed on the sample. The entry of patient demographics, sample details and tests required on the LIS will ensure complete and consistent patient information within the hospital organisation promoting good data quality.

**Domain:** Laboratory Office

**Context:** Not applicable

**Scale:** Intra-organisational

**Systems:** Laboratory Information System, Patient Administration System

**Information:** Request received, Order Entry of patient demographics and sample details in LIS

**Participants:** Clerical Officer

**Identities:** Patient Forename, Patient Surname, MRN, DOB, Address, Clinic, Consultant, Sample Collection Date, Investigations Required, Sample Received Date, Sample Received Time, Sample Type, Laboratory Specimen Number, Test Code

**Process Flow:**

1. Request received in laboratory office.
2. Order Entry performed in LIS by clerical officer.
3. Patient demographics entered in LIS.
4. Sample details entered in LIS.
5. Tests to be performed entered in LIS.

Use Case: 5.1/5.2. Sample returned to lab/Sample prepped for analysis

**Relevance:** Following order entry from the clerical officer, the sample is returned to the laboratory for preparation of the sample before analysis. The sample requires preparation before analysis. The laboratory has standard operating procedures which provide steps to follow to prepare and conduct analysis until a result is generated.

**Domain:** Laboratory

**Context:** Not applicable

**Scale:** Intra-organisational

**Systems:** Standard Operating Procedures (SOP)

**Information:** Sample returned for analysis

**Participants:** Laboratory

Laboratory Aide/Basic Medical Scientist

**Identities:** Patient Forename, Patient Surname, MRN, DOB, Address, Clinic, Consultant, Sample Collection Date, Investigations Required, Sample Received Date, Sample Received Time, Sample Type, Laboratory Specimen Number, Test Code

**Process Flow:**

1. Sample returned to lab.
2. Sample prepared for analysis.

### Use Case: 6 Analysis Performed

**Relevance:** The Laboratory Aide (LA) or Basic Medical Scientist (BMS) will follow the standard operating procedure to prepare the sample for analysis. By interfacing, the LIS populates the analyser and prepares a visual worksheet to guide the LA or BSC to the correct placement of samples. Analysis is performed, and results interfaced into the LIS for checking before authorisation.

**Domain:** Laboratory

**Context:** Not applicable

**Scale:** Intra-organisational

**Systems:** Laboratory Information System (LIS), LIS interface, Analyser

**Information:** Sample Analysis

**Participants:** Laboratory, Laboratory Aide, Basic Medical Scientist, LIS, LIS interface

**Identities:** Patient Forename, Patient Surname, MRN, DOB, Address, Clinic, Consultant, Sample Collection Date, Investigations Required, Sample Received Date, Sample Received Time, Sample Type, Laboratory Specimen Number, Test Code

**Process Flow:**

1. Analysis is performed on the analyser
2. Results are interfaced onto LIS.

Use Case: 7.1/7.2. Results interfaced on LIS/Results Authorised on LIS

**Relevance:** Interfacing and authorisation is the final stage were demographics can be visually checked by a senior metabolic scientist. Before authorisation, the scientist will check the demographics and laboratory specimen number in the LIS. Once complete the results are authorised and transmitted to the defero texting system.

**Domain:** Laboratory

**Context:** Not applicable

**Scale:** Intra-organisational

**Systems:** Laboratory Information System (LIS)

**Information:** Result interfacing, authorisation of results on LIS

**Participants:** Laboratory, Senior Metabolic Scientist, LIS, LIS Interface, Defero texting system

**Identities:** Patient Forename, Patient Surname, MRN, DOB, Address, Clinic, Consultant, Sample Collection Date, Investigations Required, Sample Received Date, Sample Received Time, Sample Type, Laboratory Specimen Number, Test Code

**Process Flow:**

1. Results interfaced on LIS by Senior Metabolic Scientist.
2. Demographics and results visually checked on LIS.
3. Results authorised on the LIS.

Use Case: 8.1/8.2. Result delivered to Defero texting system/Message viewed in Defero

**Relevance:** Transmission of results from the LIS to the defero texting system is the second last electronic message transferred containing patient demographics. The dietician will review the message in a browser window. Once confirmed the results are approved for a text message to be sent to the guardian.

**Domain:** Texting system

**Context:** Not applicable

**Scale:** Inter-organisational

**Systems:** Laboratory Information System (LIS), Defero Texting System

**Information:** Results delivered to Defero, results viewed in Defero

**Participants:** Defero texting system, Metabolic Dietician

**Identities:** Patient Forename, Patient Surname, MRN, DOB, Address, Clinic, Consultant, Sample Collection Date, Investigations Required, Sample Received Date, Sample Received Time, Sample Type, Laboratory Specimen Number, Test Code

**Process Flow:**

1. Results are delivered to the Defero Texting System.
2. Results are viewed by the dietician in the Defero Texting System.
3. Results are approved by the dietician and selected for sending a text message to the guardian.

Use Case: 9.1/9.2 Send Message to Guardian or Contact Guardian by phone

**Relevance:** Texting of results is a service available for patients over the age of 2 who attend the Metabolic Unit at Temple Street Children's University Hospital. This service was introduced to help elevate the time spent by dieticians, phoning patient guardians with results. With this new process, normal results can be text after a review in the browser of the Defero Texting System. Patients who require an intervention will receive a text asking them to contact the dieticians at the metabolic unit.

**Domain:** Metabolic Unit

**Context:** Not applicable

**Scale:** Intra-organisational

**Systems:** Defero Texting System

**Information:** Message sent to guardian or guardian contacted by phone

**Participants:** Defero Texting System, Dietician

**Identities:** Patient Forename, Patient Surname, MRN, DOB, Address, Clinic, Consultant, Sample Collection Date, Investigations Required, Sample Received Date, Sample Received Time, Sample Type, Laboratory Specimen Number, Test Code

**Process Flow:**

1. Dietician sends message to guardian or Guardian contacted by phone depending on the result.

Use Case: 10 Result received by Guardian

**Relevance:** Results received indicate to the guardian whether the result is normal and to proceed with dietary regimes. Otherwise, the guardian is asked to contact the dietician to discuss modifying the regime.

**Domain:** Participatory healthcare

**Context:** Not applicable

**Scale:** Guardian

**Systems:** Mobile

**Information:** Results received by guardian

**Participants:** Guardian

**Identities:** Patient Forename, Patient Surname, MRN, DOB, Address, Clinic, Consultant, Sample Collection Date, Investigations Required, Sample Received Date, Sample Received Time, Sample Type, Laboratory Specimen Number, Test Code

**Process Flow:**

1. Result received by guardian.

# 7 Conclusion

This aim of this research was to answer the question of 'What role does identification play to support metabolic dieticians and scientists for care and research of patients with PKU – An information modelling perspective'. To answer this question, it has been broken down into three points to address:

1) What is the role of identifiers in integrated care and research using an EHR?
2) What are the identifier issues focusing on modelling and quality?
3) To identify uses carried out by dieticians and scientists using information for care and research of patients with PKU

The research process consisted of focus groups informed by the literature review. The literature review was repeated to link any findings to the literature, investigate the impact on standards and relate the data gathered back to answering the motivations of the research. Results from focus groups were illustrated and tabulated. Further categorisation of the results formed the tables into care and research uses with patient, sample, and other identities.

From the results gathered, a UML activity diagram was created, and the monitoring use was selected to demonstrate how this could be augmented to other uses. The use case template from the Antilope project was employed to create use cases representing those seen in the UML activity diagram figure 26. Identifiers were added to the created use cases, to illustrate the importance for potential future inclusion. Following the template from eHealth Ireland persona project, three personas were created to represent the patient, scientist, and dietician from the domain analysis of a PKU case. The key findings from this work are presented below.

Strategies were introduced which highlighted the current state and plans for health care in the future. For successful execution of an eHealth strategy, research showed it to be dependent on fundamental enablers being present. The key themes presented were integration, interoperability, and standardisation. The literature review identified a gap where the importance of identifiers should be included for future research and work.

The research found that not all stakeholders are included in the development process which leads to the lack of interoperability and poor data quality. The importance of stakeholder engagement in the standards development process has been highlighted at national,

European and International forums. This research, using the example of the condition PKU, showed how vital stakeholder engagement is in the creation of standards.

As part of the eHealth strategy in Ireland, the provision of an individual health identifier is acknowledged. The IHI will uniquely and safely identify a person. The absence of an IHI will contribute to an ineffective EHR system. The role identifiers play in integrated care using an EHR is vital. Current standards for reference modelling do not recognise identities other than patients and physicians. This research shows the requirement for inclusion of other identities, for example, the laboratory.

## 7.5 Limitations of the research

This research demonstrated the importance and role of identifiers play to support dieticians and scientists in the care and research of patients with PKU, from an information modelling perspective. There are some limitations recognised in this research.

The number of people to question for the research was restricted. This is because it was conducted in the National Metabolic Laboratory and National Centre for Inherited Metabolic Diseases. Key stakeholders were chosen to conduct the research and it is a good starting for future work.

There is also the time restriction given for the research to be conducted. Along with scheduling, it can be difficult to gather groups of similar expertise at the same time. With small numbers of specialised experts, the demands for their time can be restricted.

## 7.6 Influence on future work

The European interoperability framework study published in 2013 influenced the Antilope project. The use cases, profiles, and standards were created following framework guidelines. Most recently the European Union's Horizon 2020 research funded the EURO-CAS project. The aim of the EURO-CAS project is to promote the adoption of interoperability testing of eHealth solution against identified eHealth standards. In summary, EURO-CAS, European and international standard organisations like CEN and ISO along with national strategies like the EHR persona project from eHealth Ireland together create a standards accreditation process. The positive is that stakeholders from National, European and International forums influence the decision making. The benefit of having a wide decision-making base is that the process is rendered more robust and complete.

There needs to be some caution with having one vision for the successful implementation of eHealth strategies. The potential for unintended consequences may not be realised until a later stage. This process and its outcomes should make allowances for technological and future innovations with integration, interoperability, and standards.

In this research, the patient was not included. Patient-centric care is in the vision for future eHealth strategies. In future work, the patient input would contribute a valuable aspect.

In May 2018, the 'Stay Left, Shift Left' strategy was introduced by Martin Curley new CIO for the HSE. This strategy will support the implementation of Sláintecare. The concept of the 'Stay Left, Shift Left' strategy is to shift care from acute care to community and home settings. With this strategy combined with Sláintecare strategy, Ireland builds towards a proactive, predictive and preventative healthcare system.

This research about the role of identities from an information modelling perspective, along with the creation of personas, UML activity diagram and uses cases will hopefully influence future standards for EHR communication.

# 8 References


ANTILOPE. 2015. *Antilope Handover Mission - 4 Key Enablers* [Online]. Available: https://www.antilope-project.eu/wp-content/uploads/2015/01/Antilope4KeyEnablersFINAL.pdf

ARELLANO, M. G. & WEBER, G. I. 1998. Issues in identification and linkage of patient records across an integrated delivery system. *J Healthc Inf Manag,* 12**,** 43-52.

BOOK, I. S. 2014. Health Identifiers Act 2014. *In:* OIREACHTAS, H. O. (ed.).

BOURQUARD, K. 2011. Contribution of Standards and Profiles to the Interoperability in eHealth. *Communications and Strategies***,** 71-86.

BOUSSADI, A. & ZAPLETAL, E. 2017. A Fast Healthcare Interoperability Resources (FHIR) layer implemented over i2b2. *BMC Medical Informatics & Decision Making,* 17**,** 1.

BROWN, R. R., DELETIC, A. & WONG, T. H. F. 2015. Interdisciplinarity: How to catalyse collaboration. *Nature,* 525**,** 315-317.

CEUSTERS, W. & SMITH, B. 2006. Strategies for referent tracking in electronic health records. *Journal of Biomedical Informatics,* 39**,** 362-378.

CHEN, X., BERRY, D. & STEPHENS, G. Using a generalised identity reference model with archetypes to support interoperability of demographics information in electronic health record systems. 2015 37th Annual International Conference of the IEEE Engineering in Medicine and Biology Society (EMBC), 25-29 Aug. 2015 2015. 6834-6839.

COMMISSION, E. 2012. eHealth Action Plan 2012-2020.

COMMISSION, E. 2017. *eHealth Interoperability Conformity Assessment Scheme for Europe* [Online]. Available: https://www.euro-cas.eu/#!project

COMMISSON, E. 2016. Standardisation Policy.

CONSULTING, D. 2013. *eHealth Interoperability Framework Study* [Online]. Available: https://ec.europa.eu/digital-single-market/news/ehealth-interoperability-framework-study-0

CURLEY, M. 2018. *Innovating for Sláintecare: Care Closer to home* [Online]. Available: http://www.ehealthireland.ie/Stakeholder-Engagement/eHealth%20Ireland%20EcoSystem/Innovating-for-Slaintecare-Care-Closer-to-Home/Martin-Curley.pdf

DELOITTE 2013. eHealth Interoperability Framework Study. *In:* COMMISSION, E. (ed.).

DICTIONARY, O. E. 2018. Complex.

ECHA. 2018. *European Connected Health Alliance* [Online]. Available: https://echalliance.com/page/Aboutus

ETHEL. 2017. *ETHEL About Us* [Online]. Available: https://www.ehtel.eu/about-us

HEALTH, D. O. 2012. *eHealth Strategy for Ireland* [Online]. Available: https://health.gov.ie/wp-content/uploads/2014/03/Ireland_eHealth_Strategy.pdf

HEALTH, D. O. 2016. Statement of strategy.

HIMSS. 2013. *Definition of Interoperability* [Online]. Available: https://www.himss.org/sites/himssorg/files/FileDownloads/HIMSS%20Interoperability%20Definition%20FINAL.pdf

HIQA. 2010. *International review of unique health identifiers for individuals* [Online]. Available: https://www.hiqa.ie/reports-and-publications/health-information/international-review-unique-health-identifiers

HITCH. 2010. *Healthcare interoperability and conformance harmonisation* [Online]. Available: https://joinup.ec.europa.eu/document/healthcare-interoperability-testing-and-conformance-harmonisation-hitch

HSE. *Individual Health Identifier* [Online]. Available: http://www.ehealthireland.ie/Strategic-Programmes/IHI/

HSE. 2016. *Integrated Care* [Online]. Available: http://www.hse.ie/eng/about/Who/clinical/integratedcare/

HSE 2017a. Patient Narrative Project. *In:* HEALTH, D. O. (ed.).

HSE 2017b. YourVoiceMatters. *In:* HEALTH, D. O. (ed.).

IPPSOI. 2015. *Knowledge and Information Strategy - Delivering the Benefits of eHealth in Ireland* [Online]. Available: http://www.ipposi.ie/our-work/health-innovation/ehealth/

IRELAND, E. 2018a. *Electronic Health Record* [Online]. Available: http://www.ehealthireland.ie/Strategic-Programmes/Electronic-Health-Record-EHR-/

IRELAND, E. 2018b. *Innovating for Sláintecare: Care closer to home* [Online]. Available: http://www.ehealthireland.ie/Stakeholder-Engagement/eHealth%20Ireland%20EcoSystem/Innovating-for-Slaintecare-Care-Closer-to-Home/

IRELAND, I. A. E. 2016. *Patient Centred Care* [Online]. Available: http://www.ipposi.ie/wp-content/uploads/2016/09/eHealth-outcome-report-FINAL.pdf

MCCANDLESS, H. L. K. 2010. Liberating Structures: Innovation by Including and unleashing everyone. *E&Y Performance.*

OPENEHR. 2018. *What is openEHR* [Online]. Available: https://www.openehr.org/what_is_openehr#

ORLOVA, A. & SALYARDS, K. 2016. Understanding Information in EHR Systems: Paving the Road for Semantic Interoperability through Standards. *Journal of AHIMA,* 87**,** 44-47.

PIHO, G., TEPANDI, J., THOMPSON, D., TAMMER, T., PARMAN, M. & PUUSEP, V. 2014. Archetypes based Meta-modeling towards Evolutionary, Dependable and Interoperable Healthcare Information Systems. *Procedia Computer Science,* 37**,** 457-464.

PIHO, G., TEPANDI, J., THOMPSON, D., WOERNER, A. & PARMAN, M. 2015. Business Archetypes and Archetype Patterns from the HL7 RIM and openEHR RM Perspectives: Towards Interoperability and Evolution of Healthcare Models and Software Systems. *Procedia Computer Science,* 63**,** 553-560.

SOCIETY, H. I. A. M. S. 2018. *Electronic Healthcare Record* [Online]. Available: https://www.himss.org/library/ehr

VICTORIA, U. O. 2012. *Complexity Science* [Online]. Available: https://www.uvic.ca/research/groups/cphfri/projects/currentprojects/complexity/index.php