

# Opportunities for Knowledge Management Tools in Clinical Genetic Services

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## Abstract

This study aims to characterize the challenges in utilizing genetic information in health care and to identify opportunities for genetics knowledge management. Taking a grounded theory approach, we conducted semi-structured interviews with 48 New Zealand genetic services stakeholders. Several themes emerged from the data, including clinicians' lack of knowledge on genetic testing and a lack of support for test result dissemination. Knowledge management tools may have the potential to support key tasks in genetic services delivery, improve knowledge processes, and enhance knowledge networks. Stakeholder comments indicate opportunities for solutions such as decision support systems, electronic referral systems, electronic health record or personal health record systems, data submission and other knowledge processing tools, ontologies, and knowledge networking tools. Implementation of these technologies in clinical genetic services may help unleash the power of genetic information to improve healthcare outcomes and knowledge management performance.

*Keywords:* knowledge management; human genetics; clinical decision support systems.

## 1 Introduction

Since the completion of human genome sequencing in 2003, our understanding of the genetic component of human diseases has vastly increased (Engstrom et al., 2005). Human genetic variation knowledge identifies the associations between genetic variations and human conditions. Applying this knowledge, clinical genetic services provide genetic testing and results interpretation (IT Perspectives Inc., 2005). These services may improve patient care because genetic testing can produce valuable information for diagnosis, prognosis, treatment, and avoidance of adverse drug reaction. It is surprising that healthcare has not been transformed by the revolution of (molecular) genetic testing, as predicted by many, including (Bell, 1998, Bhandari et al., 2000, Glasner, 2002, McGuire et al., 2008, Zimmern, 1999, Personalized Medicine Coalition, 2006).

The management of medical genetic test results is far from systematic or efficient in terms of data storage, access, sharing, and usage. These results are not treated as other medical data in health systems. This is because

genetic information has familial and generational implications; it is predictive and the emphasis is not on treatment; its implications are sometimes severe, private and complex, introducing special ethical dilemmas (Kääriäinen et al., 2006). Given these challenges, this study aims to characterize the use of genetic information in health care and to inform how we might improve human genetic variation knowledge management.

Knowledge is a "fluid mix of framed experience, values, contextual information and expert insight that provide(s) a framework for evaluation and incorporating new experiences and information" (Davenport and Prusak, 1997). Accordingly, managing genetics knowledge requires a structured representation of genetic data and a framework in which to evaluate, incorporate, and share new experiences and information from routine genetic testing. It is valuable to support the processing of both explicit and tacit human genetic variation knowledge in order to advance genetics science as well as to apply it in health care. Knowledge management (KM) literature has identified three essential approaches: knowledge stock, knowledge process, and intellectual capital (Nonaka, 1994, Carlsson et al., 1996, Alavi and Leidner, 2001, Schultze and Leidner, 2002, Sambamurthy et al., 2003, Argote et al., 2003, Stankosky, 2005). Based on these theories, this research project aims to explore the principles and goals for human genetic variation knowledge management.

To realize the full potential of personalized medicine, policies must be implemented to appropriately protect the confidentiality, privacy, and security of genetic testing data with regard to access and use (McGuire et al., 2008). In the US, data models are being created to promote incorporation of genetic information in health record storage (Feero et al., 2008) and transfer (Shabo, 2007). New Zealand studies are needed to answer the critical questions around clinical genetic information management principles and frameworks. In this study, we analyse the range of practices in use of information relating to genetic services, and uncover the perceived gaps and opportunities within the existing information technology (IT) environment.

## 2 Methods

This study takes a grounded theory approach to provide groundwork for refining KM processes in genetic services. Grounded theory is an inductive methodology to generate theories through the emergence of conceptual categories (Glaser and Strauss, 1967, Rhine, 2008). The research data are collected by (1) reviewing relevant literature, policies, and organizational documents; (2) observing and communicating with participants about their roles, activities, and technology satisfaction; (3)

recording stakeholder comments on current or potential issues and challenges, mostly via face-to-face or telephone interviews; and (4) writing notes during observation, data collection, and data analyses. This multi-dimensional approach validates the accuracy of our findings as it triangulates different data sources by examining evidence from the sources and using it to build a coherent justification for themes (Creswell, 2003).

We collected the main data set of the study through semi-structured interviews with 48 participants. The interview questions encourage participants to talk about issues they feel are critical to genetic information management and KM. We used convenience sampling, snowballing, and theoretical sampling to identify participants that represent key stakeholder groups of the clinical genetic services in New Zealand. Their roles include those of patients and family members (numbering 8), healthcare providers (8), genetic counselling professionals (11), genetic testing laboratory scientists (3), governance (3), health IT professionals (4), researchers (5), directors of health institutions providing genetic services (7), and clinical advisors at indirect health services such as health insurance providers (5). Some participants have more than one role in the system, for instance as both clinician and director.

We conducted the data analysis as interviews proceed until data saturation. We use the classic grounded theory approach for inductive framework production from theoretical sampling, open coding, interpretive analysis, and constant comparison (Glaser and Strauss, 1967). We also apply a systematic multiple coding process that developed upon classic grounded theory, including three steps: (1) generating categories of information (open coding), (2) selecting one of the categories and positioning it within a theoretical model (axial coding), and then (3) explicating a story from the interconnection of these categories (selective coding) (Strauss and Corbin, 1998), see also (Creswell, 2003). Through multiple coding over interview data and constant comparison among all data sources, several data categories and themes, as well as one final theory, emerge based on our understanding of the New Zealand genetic service system processes and of stakeholder perspectives. One of these results indicates promising technologies for improving the performance of human genetic variation KM in clinical settings.

### **3 KM Challenges**

A number of topics surface from the interviews concerning genetic information management and knowledge management. In another paper, we reported the identified challenges and concluded that in order to realise the full potential of human genetic variation knowledge for better healthcare outcomes we need to overcome: the challenges of multiple service models, the inhibitors for genetic data sharing, and the barriers to wider use of genetic testing (Gu et al., 2011). In the current paper, we discuss the two themes that present major barriers for managing human genetic variation knowledge in the clinical setting, i.e., the clinician knowledge of genetic testing and the dissemination of genetic test results.

#### **3.1 Clinicians' Lack of Knowledge Regarding Genetic Testing**

In an earlier paper (Gu et al., 2010), we reported the gaps in clinician knowledge are a barrier for involving them in genetic services delivery. There is little information available to clinicians regarding genetic test's clinical utility and the service's referring processes. We collect clinicians' need for support in test referral decision-making, referral processing, and patient management. Critical information during these tasks includes disease risk probability, the test's clinical utility, prevalence, penetrance, and cost/benefit, as well as interpretation and limitation of test result. KM literature emphasizes support for knowledge processing, including knowledge storage, application, and reuse (Alavi and Leidner, 2001, Schultze and Leidner, 2002, Sambamurthy et al., 2003, Argote et al., 2003). Accordingly, genetics knowledge processing requires efforts to establish human genetic variation knowledge stocks, such as genetic service protocols and evidence-based clinical guidelines, as well as to facilitate knowledge flows that deliver critical information to clinicians.

#### **3.2 The Lack of Support for Circulating Genetic Information**

Our study also highlights a lack of support for distributing genetic test results. A regional genetic service director states that test results are disseminated only when a patient wishes them to be and the patient will benefit from the data sharing. However, there is no health information dissemination system to facilitate this data flow, among healthcare professionals or family members.

In New Zealand, most medical lab results are electronically transferred directly into practice management systems (PMS) of the GPs and specialists who order the tests. The participant comments in our study indicate that genetic test results are not always included in this system. Because of this, the test results are not delivered to healthcare providers in a timely manner, which delay the data distribution for quality healthcare delivery. A medical testing lab director points out the inability or unwillingness of genetic testing labs to issue reports electronically; and believes that the report should be distributed in the same way as all other lab reports, by electronic (or perhaps paper) transmission to the requesting clinician. On the other hand, it is claimed by genetic testing labs that all results are issued electronically through Delphic reporting system (Sysmex New Zealand Ltd., 2009); yet regional genetic services receive all results on paper. The inconsistency of the data management approach throughout the genetic services delivery system seems to be causing difficulties in data sharing.

In terms of patient's access to test results, the idea of an electronic patient access portal is proposed by both patients and healthcare providers in our interviews. The active involvement of patients in their own health management is believed to have potential for improving health outcomes and cost-effectiveness of medicine. A related topic is the support patients need when dealing with genetic information, including information support and emotional support. Patient support groups are highly

regarded by a patient who has her breasts preventively removed based on BRCA1 positive result and after hyperplasia was found,

There needs to be support readily available for other women/men who have experienced the same journey... It really lacked that for me. All I wanted was to meet someone my age who lost her breasts and could tell me it would be ok. ... I don't think there is enough focus on the options available to women. I was handed brochures which were 10 years old, not given any support group options, ... Most women I talk to say they feel alone until they meet other women who are in the same boat. It is easy for someone to explain your options from a piece of paper but there needs to be detailed information available for these women.

As this patient articulates, information support is needed in order to understand genetic testing and test result, to apply genetics knowledge in their health management so as to improve their health outcome. Moreover, patients have great needs for emotional support, particularly from other patients who are dealing with similar positive genetic test results and are facing similar decision-making.

Regarding family communication about genetic test results, we find that the data distribution firstly depends on the patient's decision whether to spread the word or not. Alternatively, in some Māori families, this decision is made by the elders in the Whānau (extended family). There is no systematic support in place to facilitate this family communication task. One genetic counsellor interviewed reported a case wherein a terminal cancer potentially could have been prevented if the patient had been informed about the risk by their family members. They often did not know that the condition was in the family because they had lost contact with another branch of their family. Our second finding on genetic data dissemination in families is that the task of contacting extended families and telling younger generations is impeded by the inadequate support for information storage and sharing. For instance, patients feel that they are not provided with sufficient educational materials regarding the implications of a test result, including the implications for families and particularly with recommendations for surveillance, management, intervention or cascade testing. This has made the distribution of genetic information a difficult job, especially in extended families, as expressed by a parent of a child patient with cystic fibrosis (CF):

Most people have a difficult time explaining to extended family the importance of them getting genetically tested for CF. Family are either ignorant/naive (i.e. "it's your problem not ours") or feel that you are just passing blame. ... I keep meaning to ask [managers] at the CF Assn [Cystic Fibrosis Association of New Zealand] if there is some information available that we can give to extended family in order for them to understand the genetic side of CF and how easily they can be tested to avoid another baby in the family being born with cystic fibrosis.

This demand for family communication support regarding genetic test results is confirmed by a study in

Australia where letters and booklets were found useful as supporting information (Gaff et al., 2005). In addition to these challenges for individuals' distribution of genetic test result, our interviews also identify barriers for aggregated knowledge sharing.

### **3.3 Difficulties in Knowledge Sharing with Global Knowledge-bases**

Genetic information and the associated clinical data collected in clinical genetic services are of high quality and of high relevance to the genetics research community (Gokhale et al., 2004). This presents an opportunity as well as challenge for data dissemination support from routine medical genetic testing up into literature and databases. Some of our participants in labs, genetic service offices, and clinics occasionally conduct research projects and contribute to journal publications; but this is difficult because they are very busy. Our participants report that there is no way of rewarding or encouraging genetic data sharing (e.g. encouraging online database submissions). From a testing lab's point of view, "If it were easy to do it, and particularly if there was some benefit to us, then we might do it. But at the moment it's neither easy nor much benefit to us." To create a system to reward data sharing may require international consensus in the research community. And IT support is needed to facilitate data submission. The participating lab scientists agreed that an ideal genetic information management system should respect patient privacy, should support lab report distribution to healthcare providers, and should assist automatic data dissemination into public databases.

We found that lab storage of genetic data often uses legacy packages from traditional medical test reporting systems and generic office information systems. For example, our participating lab scientists often keep a list of mutations and their interpretation in Microsoft Word, Access, or Excel. One problem with these methods is the lack of data structure, which leads to the flat files in individual labs and subsequently in public databases. Software applications that are specialised in genetic data analysis, such as Vector NTI (Invitrogen Corporation, 2008), Alamut (Interactive Biosoftware, 2009), and Geneious (Biomatters Ltd., 2005, Drummond, 2007) may also hold some fragments of genetic test data in the labs. But these tools don't offer efficient submission support to international databases either.

## **4 Opportunity for KM Tools**

Building upon the above themes that emerged from interviews, this section triangulates our findings with broader health informatics literature. We focus on identifying potential IT solutions that may improve genetics knowledge management, such as decision support systems, electronic referral systems, electronic health record or personal health record systems, data submission and other knowledge processing tools, ontologies, and knowledge networking tools.

### **4.1 Decision Support Systems (DSS)**

This study highlights the need to develop clinician competencies as genetic service providers. Our findings

suggest an opportunity for DSS developers to answer GPs' requests for risk and prognosis information support, as well as for process support in identifying and managing the patients at risk. A key task in delivering genetic services is the test referral decision-making. DSS may assist the task by providing clinicians with information on clinical utility of the test and probability of disease risk, as well as on prevalence, penetrance, and cost/benefit of the test. Another potential for DSS is to collect the right information for genetic counsellors and genetic testing labs. For instance, the family history validation process often occurs after the first meeting between a patient and a genetic counsellor. This process may start at the point of doctor referral by appropriate prompts from a DSS. In addition, DSS may provide information on the availability of genetic tests in the format of a user manual or "Lab Yellow Pages" (Pagon, 2006).

Applying up-to-date knowledge that links genetic tests to health management, DSS should aim not only to prompt referrals for testing, but also to translate test results into intervention recommendations such as surveillance, treatment, and drug dosage. Therefore, future DSS should incorporate evidence-based clinical guidelines and provide intervention recommendations. To provide recommendations rather than just assessments at the time and location of decision-making is a critical success factor for DSS (Kawamoto et al., 2005). The contribution of DSS algorithms to administrative functions such as prompting clinicians about surveillance schedules should be used in genetic testing in much the same way that they are used in primary care, such as recalling patients for cervical screening every three years. This may profoundly and beneficially change clinical practice by realising the value of medical genetics and pharmacogenetics knowledge. For instance, DSS may facilitate complying with established guidelines, such as warfarin initial dosing (US Food and Drug Administration, 2007b, US Food and Drug Administration, 2007a). The implementation of DSS in supporting this task may elicit a wider uptake of genetic testing technology in health care and deliver better health outcomes. Future DSS should also automatically check knowledge-base updates, inform healthcare providers about new scientific findings, identify patients with the relevant mutations that were previously interpreted as uninformative variants, and suggest further actions such as preventive procedures.

## **4.2 Electronic Referral Systems (E-Referral)**

In addition to including DSS functions as part of an e-Referral, the process of referral itself also requires a systematic approach to better support rapidly growing demand for genetic testing. E-Referrals may support streamlining the test referral, and referral triage, processes, may engender consistent business processes in genetic services, as well as may facilitate the tracking and sharing of test status. The New Zealand Health Information Strategy Advisory Committee (HISAC) envisaged that e-Referrals will include features such as referrals tracking, electronic status reports, acknowledging referral receipts, and generating alerts if service level timeframes are not met (New Zealand

Health Information Strategy Advisory Committee, 2009). Better information sharing and communication support would also help bridge the conventional boundaries of primary, secondary and tertiary care, improve provider-user trust and relationships, and ultimately would enhance the knowledge network in the genetic services delivery system.

## **4.3 Electronic Health Record (EHR) or Personal Health Record (PHR) Systems**

The Human Genetics Society of Australasia (HGSA) guidelines suggest genetic data should be kept separately in the genetic services and away from everyday clinical practice on the basis of the nature of the data and the consequences of misuse (Human Genetics Society of Australasia, 2007). Even though this is not a bad idea, it introduces the need for interoperability and for clearly defined rules regarding data sharing. Taking into account also the need to support family communication, we propose adopting shared EHR such as the United Kingdom (UK) HealthSpace SCR (UK National Health Service, 2009) and the United States (US) My HealtheVet (US Department of Veterans Affairs, 2009), or PHR such as Google Health (Google, 2009) and Microsoft HealthVault (Microsoft Corporation, 2010, Quest Diagnostics Inc., 2009) for electronically storing and sharing genetic test results. These systems feature secure storage and transfer of sensitive medical data, particularly with access control to lab results. Especially, PHR has the potential to reduce medical errors, improve disease management, and reduce overall health care costs by empowering patients as active participants in their own health care and facilitating communication between clinicians and patients (Reti et al., 2009, Markle Foundation, 2003). They are aligned with patient-driven health care models (Swan, 2009), which might be applicable for managing genetic test results, given the nature of genetic information as sensitive (personal and medical) and shared (with implications for family members).

## **4.4 Future KM Targets**

Based on participant comments, medical genetics KM requires efforts to facilitate knowledge processes (e.g., to establish knowledge stocks and to support knowledge flows) and to enhance knowledge networks (e.g., to develop core competencies).

### **4.4.1 Knowledge Processing**

To accelerate the knowledge processes of creation, integration, and reuse, one beginning point is to support data dissemination for research purposes. Clinical genetic services have the potential to create, validate and reuse genetics knowledge through collecting genetic and clinical data for genetics research community. The research use of these data may enhance our understanding of the underlying biological mechanisms of human diseases, thus, may lead to better management of the conditions. A key to realizing information sharing, as pointed out by lab scientists, is to make it easy and beneficial to data submitters. For instance, opportunities present themselves to develop systematic support for

database submission to central databases, locus specific databases, or to research projects including individual studies and coordinated projects such as the Human Variome Project (Human Variome Project, 2007). The data dissemination needs have to be appropriately balanced against individual confidentiality concerns. Therefore, de-identification of genetic data is a key to complying with privacy principles.

KM technologies might also support several other processes in the human genetics knowledge cycle, such as where to find knowledge, how to classify, store, maintain, and use it, how to ensure its quality, and how to motivate people to contribute. The codification of this knowledge may depend on standard data presentation in the domain. One key advance towards standardized documentation is the application of gene ontologies.

#### 4.4.2 Gene Ontologies

Because of the complexity of genetic data, standards are essential for human interpretation of genetic test results and for enabling interoperability between information systems. International standards are applied in testing labs, such as mutation nomenclature by Human Genome Organization (Antonarakis and Nomenclature Working Group, 1998, Dunnen and Antonarakis, 2000). However, interpretation of test results is not systematically coded in lab reports. A potentially useful tool for this problem is the well defined gene ontologies from the bio-informatics domain. By applying ontology-controlled terms and concepts, genetic sequence (variant) data would be represented within structure and semantics. Bio-ontologies will serve as: controlled vocabularies for annotation, the describing schema and the content of schema, domain maps, query mechanisms, resolution of semantic heterogeneity, and text analysis (Stevens et al., 2003). The success of the gene ontology (GO) project (Smith et al., 2007) in the bioinformatics research context is yet to be transferred into healthcare settings, especially for annotating the clinical significance of genetic variants. Meanwhile, health level seven (HL7) has organised a Clinical Genomics Special Interest Group, and is developing genotype models and a Family History Model in the interests of interoperability (Shabo, 2007). Further development of clinical genetics terminology using the systematized nomenclature of medicine (SNOMED) (International Health Terminology Standards Development Organisation, 2008) and the logical observation identifiers names and codes (LOINC) (Cimino et al., 2009, Bodenreider, 2008) may also help establish structured genetic test results as transferable health data.

#### 4.4.3 Knowledge Networking

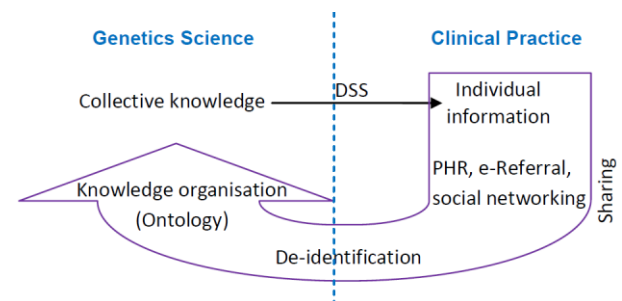
The issues that emerged in our study provide indications of how to enhance the knowledge network among all stakeholders in the genetic services system. First of all, the New Zealand genetic services, as a system, demonstrates ineffective communication and a lack of cooperation among health professionals, which is consistent with a UK report (Eeles et al., 2007). E-Referrals have the potential to facilitate cooperation across sectors, e.g., by sharing test status information, and

therefore to enhance the network of relationships among genetic services stakeholders. A second target in knowledge networking is to prepare healthcare professionals, especially primary and secondary providers, for genetic services delivery. As reported in the UK, a continued lack of awareness of cancer genetics is found amongst primary care practitioners (Iredale et al., 2007). Service protocols and guidelines as well as tools such as DSS may assist care givers in disease risk assessment, genetic testing referral decision-making, and patient management.

From the patient perspective, systematic assistance is needed for enhancing their communication with care givers and family members, e.g., through PHR. Patient contact with other patients in similar circumstances may be improved through online patient support groups or other social networking tools. Consumer health informatics technologies may be useful, including patient support groups such as PinkHope (Barter, 2010), and social networking websites such as PatientsLikeMe (PatientsLikeMe, 2010). With enhanced knowledge networks that involve patients and families, better health outcomes can be expected with more informed intervention choices such as preventive and personalised medicine.

#### 4.4.4 Genetic Services KM Framework

As a summary for the KM targets and promising IT tools based on the issues addressed in this study, we propose a KM framework for the clinical genetic services delivery system, as shown in Figure 1.



**Figure 1: KM framework for the clinical genetic services system**

Knowledge flow from genetics science to clinical practice will be supported by tools such as DSS for clinicians during key tasks in genetic service delivery. Feeding back to the body of the human genetic variation knowledge, data sharing will be facilitated by PHR among family members, by e-Referrals across health sectors for test status updates, by social networking tools such as online patient support groups, and by gene ontologies and submission support towards knowledge-bases with appropriate de-identification approach. Better health outcomes are expected from improved capacity of the genetic services system through technology implementation.

## 5 Discussion

The goal of this study was to examine the use of information relating to genetic services, identifying gaps, and exploring differences between “usual” health

information management and medical genetic information management so as to provide groundwork for refining KM processes to meet the needs of people using and working in genetic services delivery. Our findings have highlighted challenges surrounding medical genetic data sharing and usage, allowing us to suggest IT opportunities for supporting human genetic variation knowledge management. Promising tools include DSS, e-Referrals, and EHR or PHR. Implementing these health informatics technologies, as well as KM tools (e.g., data submission and other knowledge processing tools, ontology, and knowledge networking tools), would address the knowledge processes in the domain and potentially enhance knowledge networks.

A closely related investigation was the 2003 GP survey in New Zealand regarding their knowledge and practice on genetic testing (White and McLeod, 2003, Morgan et al., 2004). This national survey found that GPs felt that they lacked experience and knowledge of genetic testing, especially on the appropriate terminology and procedures. Our interviews have found little change regarding GP involvement in the genetic services delivery system. It appeared that clinicians, including GPs, need help to identify the utility of genetic tests. A 2009 study in the US highlighted the promotion of the inclusion of genetic information in the EHR (Sethi and Theodos, 2009). Our study concludes that IT might have great potential in the domain, such as DSS, e-Referrals, EHR/PHR, and KM tools. In particular, national implementation of DSS and e-Referrals is part of the current New Zealand national health IT plan (IT Health Board, 2010).

The main limitation of this study is that it is based upon personal perspectives from individual experience which might not represent accurately the entire New Zealand genetic services system. Strategic planning and framework development are needed to enable more widespread participation in the active use of genetic information among healthcare professionals, and to achieve a degree of uniformity and interoperability among nations. Establishment of such strategies and frameworks may require more research into the technical, organisational, and social issues that will open up public debate on the topics.

Future actions on the part of particular stakeholder groups include:

- *Clinicians* should require information and systematic support for the processes of genetic services, including electronic decision support and effective electronic referral systems, as well as clarification of ethical and legal framework.
- *Health IT innovators* should develop and implement quality systematic solutions to support key tasks in genetic service delivery.
- *Policy makers* should lead wide debates on the legal and ethical issues surrounding genetic information in order to develop appropriate frameworks that will facilitate better use of genetic data to improve health care.

Furthermore, patients and their families, of course, have the most at stake. They – as citizens generally, and through specific groups, such as cancer societies – should

insist on action on the three fronts above, as well as with respect to information, information management technology and support groups for their own direct use.

## 6 Conclusion

Taking a grounded theory approach, we explored the domain of human genetic variation knowledge management in the New Zealand clinical genetic services system. Clinicians' lack of knowledge of genetic testing and the lack of support for test result dissemination present barriers for unleashing the potential of genetics knowledge in health care. To overcome these challenges, we recommended a few KM technologies, including DSS, e-Referrals, EHR/PHR systems, data submission and other knowledge processing tools, ontologies, and knowledge networking tools. These will facilitate genetics knowledge processing by supporting the flow of genetic data such that it is appropriately structured and interpretable to all stakeholders. Moreover, knowledge networks among clinicians, genetic counsellors and lab scientists, as well as with patients and family members, will be enhanced through better communication support and more available information.

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