The Diversity of Genetic Services Delivery Models in New Zealand

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Abstract

Medical genetic testing is starting to change clinical practice in many nations. Our New Zealand (NZ) genetic services stakeholder interviews identified four distinct service delivery models, namely Patient-Doctor-Counsellor Model, Patient-Doctor-Lab Model, Patient-Counsellor-Lab Model, and Patient-Lab (Commercial) Model. This paper applies unified modeling language (UML) activity diagrams to describe the workflows in each of the four models. Based on participant comments, we analyse the strengths and weaknesses of each model, finding problems such as insufficient knowledge and little communication among stakeholders. With the different referral processes and non-standardised documentation in the multiple models, the communication of genetic testing results and their implications is not managed in a systematic manner. In particular, inadequate information is available to stakeholders, incomplete documentation is recorded in some models, and there is little support for communication or result dissemination. Targeting these problems, information technology (IT) development has the potential to meet genetic services stakeholders' information and communication needs. Future IT implementation in the domain should be tailored to assist key tasks such as test referral, risk assessment, documentation recording, and result dissemination. Helpful health IT support might include clinical decision support systems (DSS), e-Referrals, shared electronic health records (EHR), and personal health records (PHR).

1. Introduction

Medical genetic testing produces valuable information for diagnosis, treatment, prognosis, and the avoidance of adverse drug reaction. A few examples include hemochromatosis gene (HFE) testing for haemochromatosis diagnosis, human epidermal growth factor receptor 2 gene (HER2) testing for choosing breast cancer therapy, huntingtin gene (HTT) testing for predicting risk of Huntington's disease, and thiopurine S-methyltransferase (TPMT) gene testing for prescribing azathioprine. Genetic services, which offer genetic testing and result interpretation, have an increasing impact on clinical practice, by providing evidence for preventive prognoses and personalised treatments [1]. One type of genetic services delivery model operating in New Zealand (NZ) is through a formal Genetic Service Program. It is a tertiary health service consisting of two regional offices: Northern Regional Genetic Service, and Central & Southern Regional Genetics Service. They provide individuals and families with education and information, family history documentation and assessment, clinical assessment and diagnosis, and patient management [2]. Questions remain, however concerning the lack of systematic storage and transfer of genetic testing results. Aiming to develop genetic information management principles, we conducted a semi-structured interview study with NZ genetic services stakeholders. Their experience and perceptions identified how the genetic services system does (or does not) work, and their information requirements, expectations, and concerns suggested how the system should work in the future.

2. Methods

This paper is based on comments made by forty-eight participants representing ten significant roles in NZ genetic services – patient and family member, general practitioner (GP), specialist, clinical geneticist, genetic counsellor (genetic associate), genetic testing laboratory scientist, director of a health institution that is directly involved in genetic services delivery (such as hospital, medical lab, or regional genetic service), clinical advisor at an indirect health service (for example, health insurance provider), manager at the Ministry of Health, advisor at the Privacy Commission, information technology (IT) advisor at the Ministry of Health or a District Health Board (DHB), health IT vendor, health IT researcher, and genetics researcher. Our observation and interview questions focused on participants'

operating procedures and their actual experience. Implementing adapted grounded theory [3, 4], data themes are identified from participant comments only. Taking a critical view [5], we then compare the differences between multiple perspectives. Unified modeling language (UML) activity diagrams with swimlanes are used to model the workflows of business processes in the system. Swimlane diagrams partition activity states into groups, such as patients, doctors, genetic counsellors, general medical labs, and genetic testing labs. Each group represents the business organisation responsible for those activities [6]. The participants' judgements of these models are summarised into their strengths and weaknesses.

3. Results

Based on participant comments, four NZ genetic services delivery models are identified: Patient-Doctor-Counsellor Model, Patient-Doctor-Lab Model, Patient-Counsellor-Lab Model, and Patient-Lab (Commercial) Model.

3.1. UML Swimlane Presentation of NZ Genetic Services Delivery Models

The Patient-Doctor-Counsellor Model is a basic NZ genetic services delivery pathway, involving patients, doctors, genetic counsellors, general medical labs, and genetic testing labs. In this model, each locus where activities occur is depicted as one swimlane, in Figure 1; for instance, all activities that occur at a doctor's office are grouped in the DOCTOR swimlane, the doctor being responsible for these activities. The Patient-Doctor-Counsellor Model starts when a patient presents with symptoms or concerns of a genetic disorder at a doctor's office (GP or specialist). Based on relevant knowledge and experience, the doctor might believe a genetic test to be useful for diagnosis or treatment. Therefore, they make a referral to a Regional Genetic Service office, where genetic counsellors evaluate and sort all referrals in a triage process. In a face-to-face meeting with the patient, a genetic counsellor or a clinical geneticist assesses the disease risk; if a genetic test is relevant and available, they suggest the testing to the patient and explain possible testing implications. For instance, having a genetic test might have implications for insurance. If the patient consents to the test, this is then arranged by genetic counsellors. Firstly, the patient needs to go to a general medical lab for sample collection (often a blood sample), which is sent to an accredited genetic testing lab according to a waiting list in the Regional Genetic Service office. Lab scientists perform the test and record any detected abnormality and its interpretation in a lab report. Based on this report, genetic counsellors write an explanatory letter suggesting surveillance recommendation and/or management intervention. After counselling the patient about implications of the test result, genetic counsellors store the doctor referral, family history, family tree, lab report, and explanatory letter into a family folder. This folder will be kept indefinitely in the Genetic Service office as a reference for family members and future generations. Upon patient consent, copies of the lab report and explanatory letter can be sent to health care providers.

The Patient-Doctor-Lab Model in Figure 2 demonstrates that clinicians can order some tests directly from a genetic testing lab without routing through genetic services, such as the HFE gene test for haemochromatosis. These doctors make direct contact with the genetic testing lab, which in some cases is overseas. They arrange sample transfer, receive the lab report, explain result implications to patients, and file the lab report in medical notes. This process could change to a Patient-Doctor-Counsellor Model if the testing lab suggests that genetic counsellors become involved, for example in prenatal testing or in pre-symptomatic testing of an incurable condition such as Huntington's disease.

On the other hand, the Patient-Counsellor-Lab Model bypasses the doctors. As Figure 3 indicates, if a family folder already exists in a Regional Genetic Service office with information about a pathogenic mutation in the family, members of the family might phone up for a disease risk assessment and sometimes a subsequent genetic test. Although genetic counsellors prefer a doctor referral, self-referrals are sometimes accepted. In such cases, there would be no doctor data stored, and doctors would not be informed unless patients requested it.

Figure 4 – the Patient-Lab (Commercial) Model is based on 'direct to consumer' genetic testing by private companies typically through Internet advertisements [7]. In this process, patients pay for the test, take their own sample at home, send it to the lab, and receive the report directly. Therefore, no documentation is stored in the health system.

3.2. Strengths and Weaknesses of NZ Genetic Services Delivery Models

According to stakeholder experience, all existing models have pros and cons. We summarise key aspects of each model in terms of strengths and weaknesses in Table 1. Some problems are consistent among several models, such as insufficient knowledge and little communication support among stakeholders. The parties of interest include patients and family members, health care providers, Regional Genetic Services, genetic testing labs, and health care funding

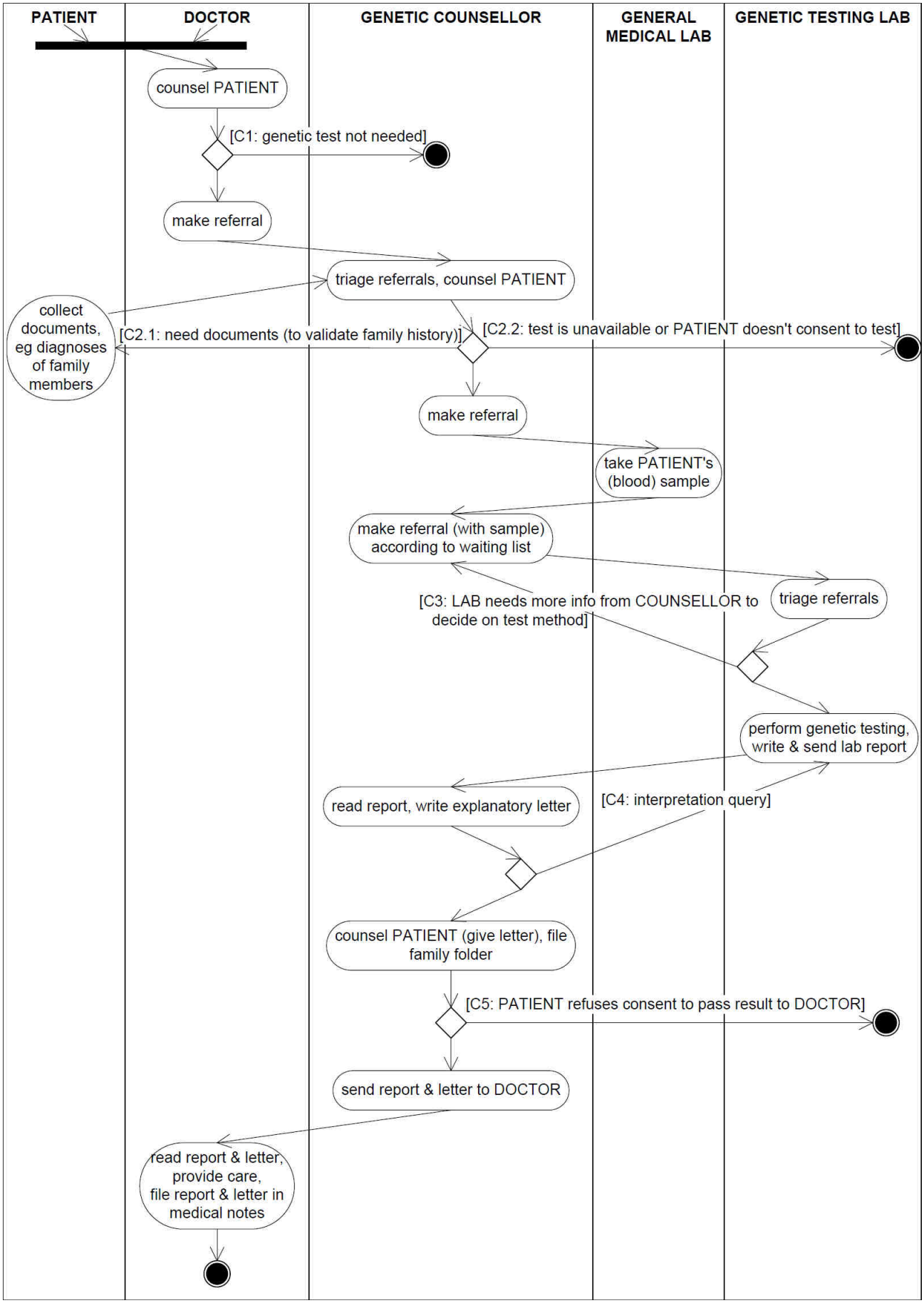

Figure 1 – UML Swimlanes of Patient-Doctor-Counsellor Model

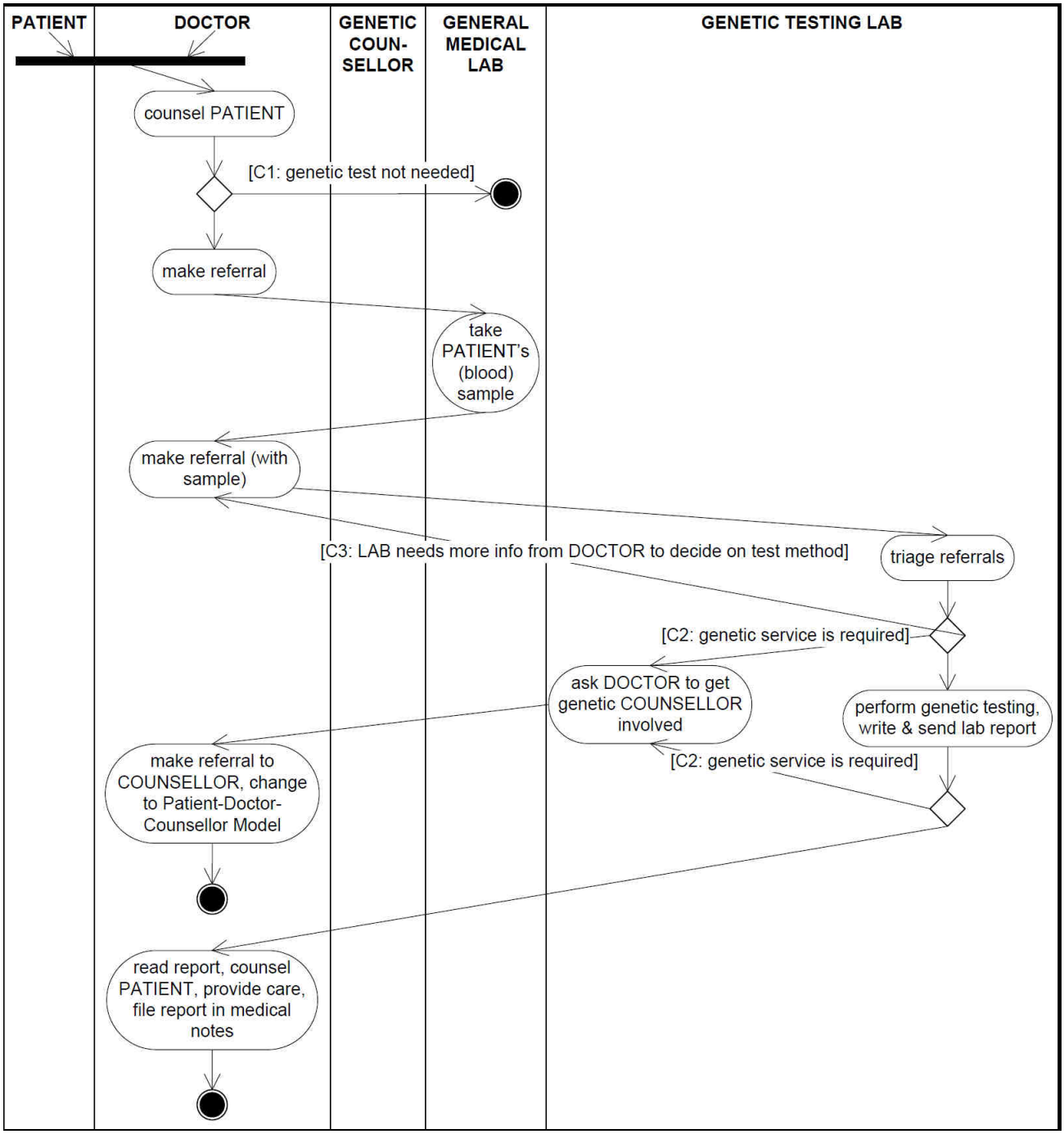

Figure 2 – UML Swimlanes of Patient-Doctor-Lab Model

agencies in both the public sector (such as DHBs) and the private sector (such as insurance providers). However, the knowledge level in many is less than satisfactory. A Privacy Commission advisor stated:

> That's a particular problem, because there's not really the knowledge out there. There's not really understanding. I think concerns and, I mean it's not ignorance, it's just the lack of knowledge of the issue tend to breed fare, quite understandably. ... I don't know if there's much knowledge of genetic issues at primary care level, who are directly involved in obtaining and providing genetic services. And a lot need to happen at the primary care level.

A GP related: "This is such a tiny aspect to GPs' work at the moment (I realise that it will increase in the future)."

Information is needed to involve health care providers in the genetic service delivery system, such as information about the clinical utility and availability of genetic tests, referral processes, and risk assessment protocols, as mentioned by a

GP: “Until we have much clearer information about risk and prognosis, primary care is not likely to be significantly involved, and will likely remain in the specialist sphere for the foreseeable future anyway.”

A good understanding of a genetic test result, including its limitations in predicting a health outcome, is required by health insurance providers. Explanations are sometimes necessary in order for health service providers to understand the value of a management intervention based on a genetic test. A genetic counsellor related: “Sometimes misinterpretation of genetic test results can occur, leading to misinformation.”

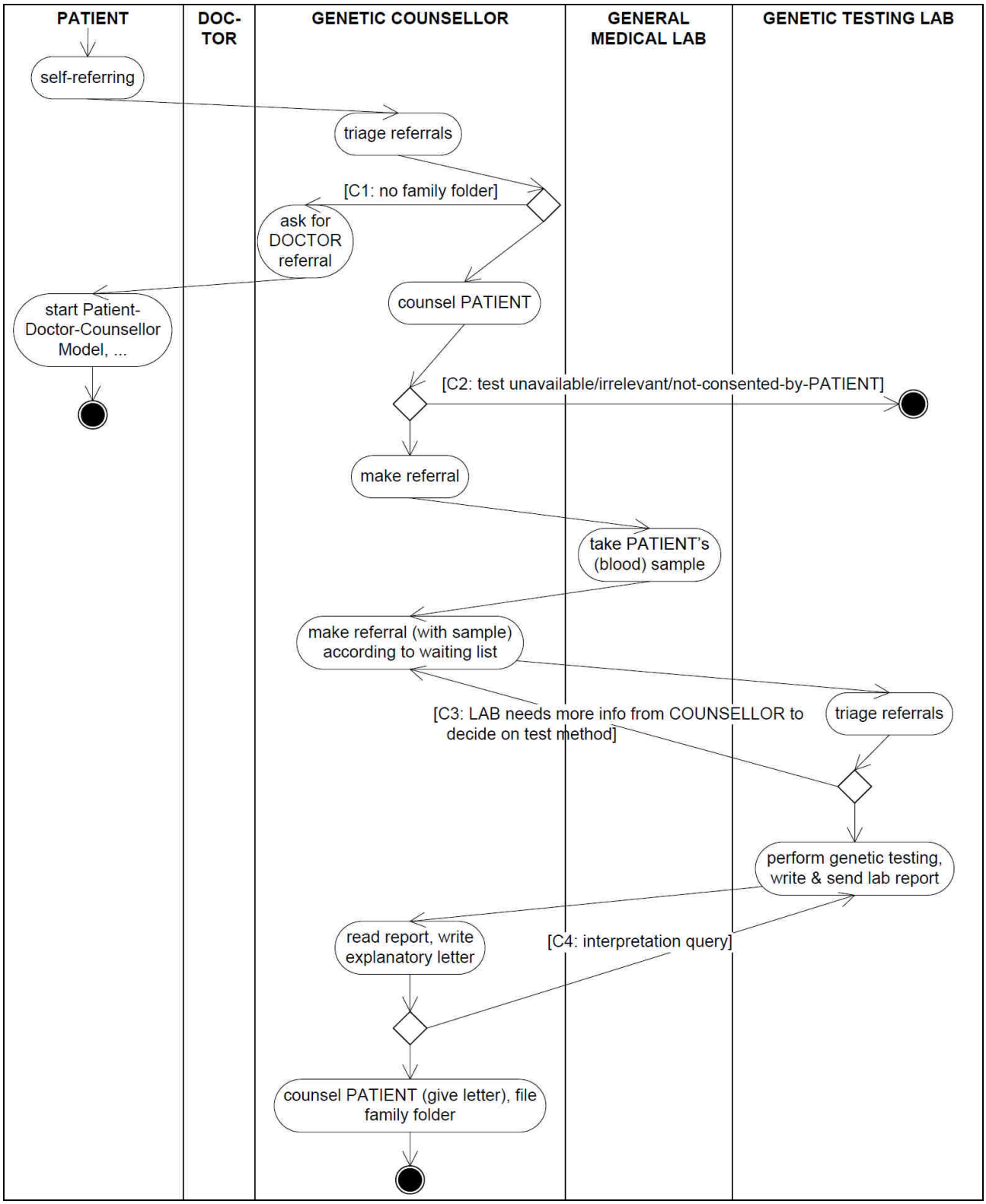

Figure 3 – UML Swimlanes of Patient-Counsellor-Lab Model

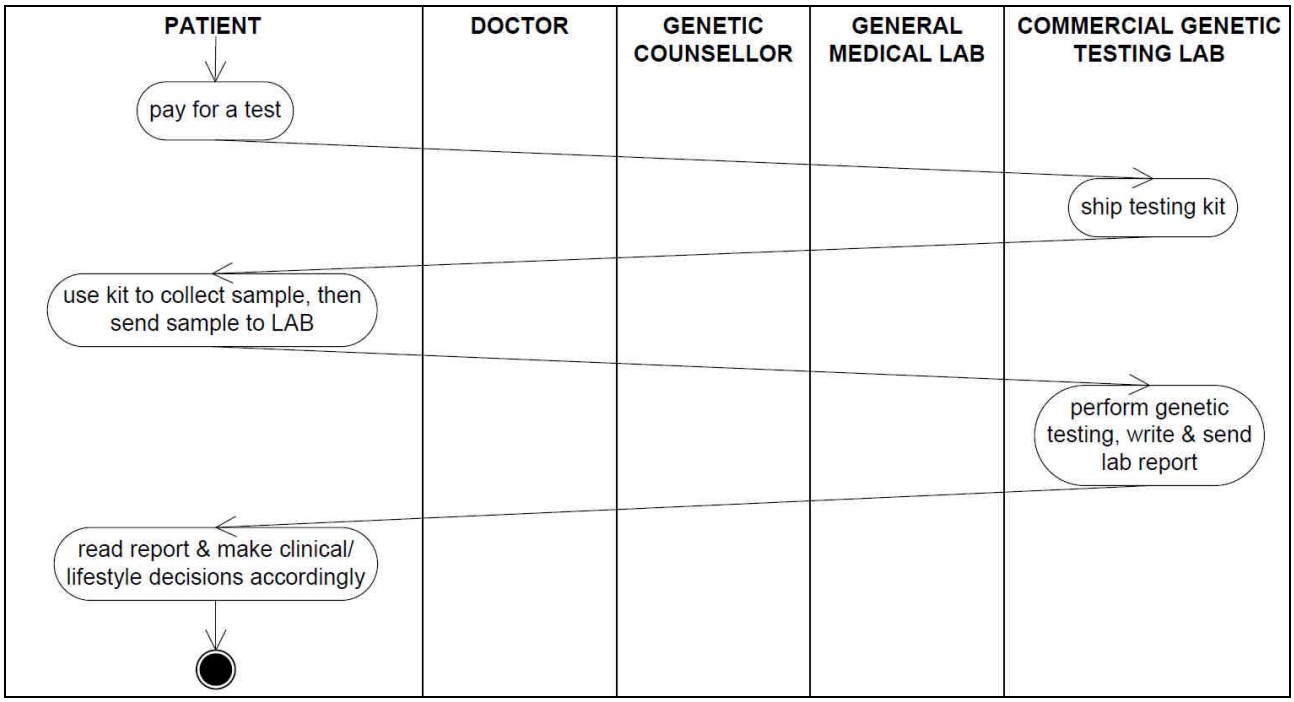

Figure 4 – UML Swimlanes of Patient-Lab (Commercial) Model

The availability of information is also essential to support the dissemination of genetic testing results in families for a better health outcome, given the fact that the result distribution in extended families is already a difficult task for patients. As expressed by a parent of a child 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.

In addition to the issue of lack of knowledge, genetic services delivery processes could be time consuming. Taking the Patient-Doctor-Counsellor Model as an example, transfer of doctor referrals and testing results often depends on postal courier services and communication during the process is seldom adequate. Several participants concur that in the months of waiting for an appointment with a genetic counsellor, then for the test results, patients and referring doctors hear little about the status of the appointments, of testing, or about the results. A medical specialist described the experience: "You just don't know what's happening, you send off a request and it goes into a long black hole."

Another difficulty in the Patient-Doctor-Counsellor Model is the validation process of family history, which requires patients to obtain relevant family members' clinical diagnoses. This involves records from multiple health institutes, sometimes overseas or dated decades before. One patient met numerous difficulties in collecting death certificates: "Their records didn't go back that far." "The records you are requesting have been destroyed."

In brief, there are four genetic services delivery models identified in the NZ system, each with its own strengths and weaknesses. The major differences among the four models – such as doctor referral processes and various documentation styles – present challenges for the systematic management of genetic testing related information. Many participants believe more effective information management would improve service quality and lead to better health outcomes. Observing these results, we will discuss a few information and communication technologies with their possible usefulness and applicability in genetic information management.

Table 1 - Strengths and Weaknesses of NZ Genetic Services Delivery Models

Service Model	Strengths	Weaknesses
Patient-Doctor-Counsellor Model	• Patient is counselled by genetic counsellor and managed by doctor • Genetic services maintain a family folder	• Requires doctor's knowledge of genetic testing • Difficult for patient to collect required documents, such as clinical diagnoses of family members • Waiting list at Regional Genetic Services • Little communication between doctor and Genetic Services
Patient-Doctor-Lab Model	• Quick turn-around time back to doctor • Patient is counselled and managed by doctor	• Requires doctor's knowledge of genetic testing, connection with testing labs, and ability to explain results • No family folder is created
Patient-Counsellor-Lab Model	• Quick, targeted testing in lab • Patient is counselled and managed by genetic counsellor • Family folder is maintained	• Family folder must be already established • Waiting list at Regional Genetic Services • Depends on family communication to inform disease risk • Health care provider is not involved unless patient requires it
Patient-Lab (Commercial) Model	• Third party, such as insurance, will not be able to access results • Patient-centred process	• Testing might not comply with authority recommendations • Health care provider is not involved • Proper counselling might not be offered • Patient must pay by private fund • No documentation is kept in NZ health system

4. Discussion

Medical genetic testing is a relatively new technology, only now starting to change clinical practice, especially at the primary care level. One example of genetic testing being potentially useful in routine clinical practice is that of drug response testing, including for azathioprine [8, 9] and warfarin [10, 11]. However, the knowledge required to deliver quality genetic services has not been effectively transferred to health care professionals. There is little information available with regard to the utility and availability of genetic tests, the test referral processes, and risk assessment protocols. Education and information services might be necessary to involve clinicians in the genetic service delivery system. Protocols and handbooks [12], in the format of a "Lab Yellow Pages" or a user manual [13], could help health care professionals with service delivery. One crucial decision is to assess whether it is indeed valuable to use genetic testing, taking into account the clinical utility of the test, the probability of disease risk, the prevalence of the test, and the cost/benefit of the test. Technologies such as clinical decision support systems (DSS) might empower service providers with knowledge and tools in this assessment. Support is also needed to facilitate referral processing and stakeholders' communications.

Technologies such as electronic referral systems (e-Referrals) might help streamline the test referral, and referral triage, processes. Ideally, e-Referrals would engender consistent business processes, facilitate the tracking of appointments and tests status, and would support the sharing of status information among service providers and users. Such tools have the potential to improve service effectiveness and efficiency, as well as to bridge the conventional boundaries of primary, secondary, and tertiary care. Ultimately, better communication among genetic service providers would enhance the knowledge network in the service delivery system.

The NZ system of multiple genetic services delivery models differs from the single Patient-Doctor-Counsellor Model of the United Kingdom (UK) [14]. This diversity presents challenges in the standardising of genetic services documentation, which could be a key to the systematic management of genetic information. Currently, genetic service delivery depends on a paper-based information storage and transfer mechanism. To achieve standardised documentation in all delivery models, the implementation of effective health IT tools is tritical. Relevant solutions might include shared electronic health records (EHR) and personal health records (PHR), such as the UK HealthSpace Summary Care Record (SCR) [15], the United States (US) MediCare PHR [16], Google Health [17], and Microsoft HealthVault [18]. These systems feature the secure storage and transfer of sensitive medical data, particularly with regard to access control of lab results. They are aligned with patient-driven health care models [19], which might be applicable for managing genetic testing results, given the nature of genetic information being sensitive (personal and medical) and being shared (with

implications for family members). Such applications for securely storing and appropriately sharing genetic information may eventually improve genetic services quality and lead to better health outcomes.

The main limitation of this study is that it is based upon personal perspectives from individual experience. It might not represent accurately the whole NZ genetic services system. The qualitative data collected are not statistically significant. The sample is not randomly selected from the population, but rather deliberately sought to enhance sample diversity.

5. Conclusion

Our New Zealand genetic service stakeholder interviews identified four genetic service delivery models: Patient-Doctor-Counsellor Model, Patient-Doctor-Lab Model, Patient-Counsellor-Lab Model, and Patient-Lab (Commercial) Model. They are presented in this paper, using specifications in UML swimlane diagrams. Patients, doctors, genetic counsellors, general medical labs, and genetic testing labs are responsible for the key activities in these models. Some problems are found, such as insufficient knowledge and little communication among stakeholders. The diversity of the four models is expressed in the various test referral processes and documentation styles. Partly due to this diversity, the communication of genetic testing results and their implications is not managed in a systematic manner. In particular, inadequate information is available to stakeholders, incomplete documentation is recorded in some models, and there is little support for communication or result dissemination. With these problems, a few health IT tools could be helpful, including DSS, e-Referrals, shared EHR, and PHR. Development utilising these tools has the potential to meet stakeholders' information and communication needs. Future IT implementation should be tailored to assist health professionals and patients with key tasks such as test referral, risk assessment, documentation recording, and result dissemination.

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