



EVIDENCE >> INSIGHT >> ACTION

Evidence Brief:
Coordinating the Use of Genetic Tests and Related Services in British Columbia

19 June 2012

McMaster Health Forum

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The evidence brief was reviewed by a small number of policymakers, stakeholders and/or researchers in order to ensure its scientific rigour and system relevance.

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KEY MESSAGES

What's the problem?

- The ongoing planning, funding, delivery and evaluation of genetic tests and related services in British Columbia face a number of challenges.
 - Demand for genetic tests and related services appears to be growing exponentially, which can be explained by their increased availability (and affordability) coupled with their use for a broader scope of both rare and common diseases and conditions. This is expected to put increased pressure on current programs and services.
 - Current programs and services lack coordination, which limits their ability to efficiently respond to emerging pressures, especially for common diseases and conditions.
 - A variety of gaps exist in the health system arrangements within which genetic tests and related services are provided. These gaps exist in delivery arrangements (e.g., lack of overarching regulations or guidelines for the delivery of and quality assurance in all laboratory and non-laboratory services, and lack of coordination and communication across providers and settings), financial arrangements (e.g., lack of coordinated funding and reimbursement models to support a re-organization of existing resources), and governance arrangements (e.g., lack of robust medical and administrative frameworks and processes to inform coverage policies and decisions).

What do we know (from systematic reviews) about three viable elements of a comprehensive approach to address the problem?

- Element 1 – Develop a comprehensive policy framework for the ongoing planning, funding, delivery and evaluation of genetic tests and related services
 - A limited number of systematic reviews were identified that addressed this element. An older medium-quality review found benefits of a coordinated and intensified strategy to inform, consult with and engage stakeholders. One high-quality and one medium-quality review found insufficient evidence to support establishing a central ‘gatekeeper’ or ‘hub’ to coordinate decision-making. No reviews were found about establishing monitoring and evaluation mechanisms to support government’s role in the financing and delivery of genetic services, in strengthening capacity in health technology assessment, or in developing increased capacity in health human resources planning.
- Element 2 – Develop a quality framework for genetic tests and related services
 - Several high-quality reviews found benefits related to many sub-elements: strategies to evaluate the type of specialist resources needed to support credentialing processes; strategies to support the implementation of regulation and guidelines for quality assurance/control; and funding and remuneration mechanisms that support coordinated and appropriate use of services. Several medium-quality reviews found benefits in increased opportunities for the education and training of health professionals and implementing quality monitoring and improvement systems.
- Element 3 – Develop a framework to support consumer/patient/family decision-making about genetic tests and related services
 - A limited number of high-quality reviews found benefits of strategies to involve consumers/patients/families in health-related decisions as well as in developing decision aids or decision-support systems.

What implementation considerations need to be kept in mind?

- Potential barriers to the implementation of a comprehensive approach to coordinating the use of genetic tests and related services in B.C. can be identified at the level of patients/individuals (e.g., the asymmetry of knowledge and expertise between themselves and providers, frustration with past efforts), providers (e.g., perceived infringement on autonomy, lack of awareness of shared decision-making approaches, financial disincentives, challenges of standardizing professional practices), organizations (e.g., making decisions in a context of ‘media hype’ and legacy of platform technologies), and system level (e.g., challenges of cross-provincial coordination and standardization).

REPORT

Rapid technological advances in genetics and genomics hold promise for the diagnosis, treatment and even prevention of common and rare diseases. Nevertheless, they also raise concerns regarding their potential impacts on health systems, which has led many countries to reflect on the optimal models to coordinate genetic testing and related services,(1-5) as well as the optimal frameworks to guide policy and coverage decisions that could help embrace opportunities offered by new and emerging technologies while minimizing the use of harmful and ineffective ones.(6-10)

In Canada, existing structures and processes have been characterized by a lack of coordination to support ongoing planning, funding, delivery and evaluation of genetic tests and related services.(11) Given this, coordinating the present and future use of genetic tests and related services has been identified as an important policy issue in many provincial jurisdictions.(12-14)

Addressing this policy issue remains challenging since it will need to take place within a context of scarce resources, relatively fixed infrastructure, a constantly evolving landscape with new and emerging genetic and genomic technologies, and increased demands for genetic testing. Without a coordinated approach and medical and administrative leadership in planning, funding, delivering and evaluating genetic tests and related services, policymakers will continue to be faced with complex and contentious policy decisions.(9)

This evidence brief and the stakeholder dialogue it was prepared to inform were designed to guide the actions of those involved in the planning, funding, delivery and evaluation of genetic tests and related services in British Columbia (B.C.). The evidence brief reviews the research evidence about: 1) key features of the problem; 2) three elements of a comprehensive approach to address the problem; and 3) key implementation considerations for moving forward.

This evidence brief uses four key terms that need to be clarified at the outset: genetics, genomics, genetic tests and related services. Genetics can be defined as the study of single genes and their effects, whereas genomics is the study of the functions and interactions of all the genes in the genome, including interactions among them and environmental factors.(3;15;16)

Box 1: Background to the evidence brief

This evidence brief mobilizes both global and local research evidence about a problem, three elements of a comprehensive approach for addressing the problem, and key implementation considerations. Whenever possible, the evidence brief summarizes research evidence drawn from systematic reviews of the research literature and occasionally from single research studies. A systematic review is a summary of studies addressing a clearly formulated question that uses systematic and explicit methods to identify, select and appraise research studies and to synthesize data from the included studies. The evidence brief does not contain recommendations.

The preparation of the evidence brief involved five steps:

- 1) convening a Steering Committee comprised of representatives from key stakeholder groups and the McMaster Health Forum;
- 2) developing and refining the terms of reference for an evidence brief, particularly the framing of the problem and three viable elements of a comprehensive approach for addressing it, in consultation with the Steering Committee and a number of key informants and with the aid of several conceptual frameworks that organize thinking about ways to approach the issue;
- 3) identifying, selecting, appraising and synthesizing relevant research evidence about the problem, options and implementation considerations;
- 4) drafting the evidence brief in such a way as to present concisely and in accessible language the global and local research evidence; and
- 5) finalizing the evidence brief based on the input of several merit reviewers.

The three elements of a comprehensive approach for addressing the problem were not designed to be mutually exclusive. They could be pursued simultaneously or in a sequenced way, and each element could be given greater or lesser attention relative to the others.

The evidence brief was prepared to inform a stakeholder dialogue at which research evidence is one of many considerations. Participants' views and experiences and the tacit knowledge they bring to the issues at hand are also important inputs to the dialogue. One goal of the stakeholder dialogue is to spark insights – insights that can only arise when all of those who will be involved in or affected by future decisions about the issue can work through it together. A second goal of the stakeholder dialogue is to generate action by those who participate in the dialogue and by those who review the dialogue summary and the video interviews with dialogue participants.

Defining what constitutes a genetic test remains the subject of numerous debates. A recent review of ‘genetic testing’ definitions in international recommendations, guidelines and reports revealed that there is no widely agreed-upon definition, with it varying from narrow definitions focusing on methodologies or the material analyzed, to more contextually-based definitions.(17)

In this evidence brief, we draw from the work of Zimmern and Kroese who define a genetic test as an assay “to detect: 1) a particular genetic variant (or set of variants); 2) for a particular disease; 3) in a particular population; and 4) for a particular purpose.”(18)

This definition of genetic test reflects that most genetic tests are ‘closed’ or in other words, that the spectrum of mutations or abnormalities that the assay is designed to test is specified in advance. Yet, recent developments in genomic technologies allow ‘open ended’ searching for any mutation or abnormality within a gene or the entire genome.(6)

This definition also highlights the need to specify the purpose(s) for which a test is undertaken. Burke and Zimmern identified three ultimate purposes of genetic tests: 1) reducing morbidity or mortality; 2) providing information salient to the care of the patient or family members; and/or 3) assisting patients or family members with reproductive decision-making.(6)

Genetic tests may have numerous medical applications, such as:

- diagnostic testing - used to confirm or exclude the presence of a genetic mutation in an affected individual who has signs or symptoms;(19;20)
- predictive or susceptibility testing - used to identify which people have a higher chance of getting a disease before symptoms appear;(19)
- pre-symptomatic testing - a type of predictive testing that identifies individuals at risk for a certain condition suspected to be present in their family;(19)
- preconception/carrier testing - examines whether individuals have (or carry) a genetic mutation;(19)
- prenatal testing - used to confirm or exclude a genetic defect or congenital malformation in an ongoing pregnancy;(19;20)
- companion testing - examination of a person's genes to understand the differential response to a pharmacological treatment and side effects;(19;20)
- population screening - the application of a genetic test to a population or large population subgroup such as ethnic groups and neonates;(20)
- pre-implantation screening - the in-vitro selection of embryos for medically assisted procreation after a diagnosis of a genetic condition in the family;(20) and

Box 2: Equity considerations

A problem may disproportionately affect some groups in society. The benefits, harms, and costs of elements of a comprehensive approach to address the problem may vary across groups. Implementation considerations may also vary across groups.

One way to identify groups warranting particular attention is to use “PROGRESS,” which is an acronym formed by the first letters of the following eight ways that can be used to describe groups†:

- place of residence (e.g., rural and remote populations);
- race/ethnicity/culture (e.g., First Nations and Inuit populations, immigrant populations and linguistic minority populations);
- occupation or labour-market experiences more generally (e.g., those in “precarious work” arrangements);
- gender;
- religion;
- educational level (e.g., health literacy);
- socio-economic status (e.g., economically disadvantaged populations); and
- social capital/social exclusion.

The evidence brief strives to address all British Columbians, but (where possible) it also gives particular attention to two groups:

- patients with cancer; and
- pregnant women.

Many other groups warrant serious consideration as well, and a similar approach could be adopted for any of them.

† The PROGRESS framework was developed by Tim Evans and Hilary Brown (Evans T, Brown H. Road traffic crashes: operationalizing equity in the context of health sector reform. *Injury Control and Safety Promotion* 2003;10(1-2): 11–12). It is being tested by the Cochrane Collaboration Health Equity Field as a means of evaluating the impact of interventions on health equity.

- testing for somatic mutations in pathological tissues from cancer patients - used to better define prognosis and to suggest and monitor treatment options that are most likely to succeed.(20)

Genetic tests can be performed using a broad spectrum of techniques/technologies from different fields of laboratory genetics such as molecular genetics, biochemical genetics, cytogenetics, in-situ chromosomal hybridization with single or multiple gene or sub-telomeric gene probes, nucleic acid amplification, microarray analysis, and whole or partial genome sequencing. Certain definitions of genetic tests even go beyond DNA-based testing and include any test that can provide genetic information, such as a physical examination and a family history.(17) Such broad conceptions of genetic tests may reflect recent trends toward the integration of genetic medicine in primary and specialized care.(1;21)

We use the term “related services” in this evidence brief to embrace a broad scope of genetic services, ranging from laboratory services to non-laboratory services (e.g., genetic evaluations, counselling, treatment management, educational activities, family support and follow-up care).(3)

The scope of this evidence brief incorporates the above definitions and is focused on elements that contribute to coordinating the use of genetic tests *and* related services in B.C.. Although clinical genetic testing and public health genetics may be considered as two distinct arenas,(20) they were included in this evidence brief and the stakeholder dialogue it was prepared to inform in order to have a comprehensive lens to explore genetic tests and related services in the province. In addition, including clinical genetic testing and public health genetics is important as they both raise quality issues for laboratory services,(20) and are critical components for efforts towards improving the coordination of genetic tests and related services.(1)

Certain issues such as genetic research and patent reform will not be the focus of this evidence brief. In addition, while the privacy/confidentiality/circulation of genetic information among health professionals and health systems constitute important issues that need to be further explored,(1;22) they were too broad to address within the scope of this evidence brief.

The following key features of the health policy and system context in B.C. were also taken into account in preparation of this evidence brief:

- responsibility for coordinating the use of genetic tests and related services in Canada is divided across the federal government (e.g., for assessments of safety and effectiveness that are required for pre-market approval of medical technologies), provincial governments (e.g., for decisions about what to fund and for whom), regional health authorities (e.g., for coordinating delivery), and the clinical and public health settings in which they are provided;(11;23)
- there is currently no provincial strategy regarding genetic tests and related services in B.C., and the Provincial Medical Genetics Advisory Committee that previously advised the B.C. Ministry of Health from 1988 to 1992 is no longer in operation;
- the Provincial Health Services Authority has the mandate to ensure that B.C. residents have access to a coordinated network of high-quality specialized healthcare services, which includes several programs related to genetic testing and related services;
- the B.C. Ministry of Health has a number of agreements with the B.C. Medical Association regarding the provision of medical and laboratory services;
- the B.C. Ministry of Health recently directed four Health Authorities to consolidate clinical support services (including laboratories) in the lower mainland;
- accreditation of laboratories falls under the auspices of the Diagnostic Accreditation Program of the College of Physicians and Surgeons of B.C.; and
- the B.C. Ministry of Health established a Provincial Technological Assessment Committee in 2012.

THE PROBLEM

The challenges in the ongoing planning, funding, delivery and evaluation of genetic tests and related services in British Columbia can be understood by considering three sets of inter-related issues: 1) the increased availability of and demand for new genetic tests and related services; 2) the lack of coordinated programs and services; and 3) current health system arrangements that limit the ability of the system to efficiently manage these pressures.

Increased availability of and demand for new genetic tests and related services

Demand for new genetic tests and related services appears to be growing exponentially, which can be explained by their increased availability (and affordability) coupled with their use for a broader scope of both rare and common diseases and conditions.(24;25) In 2012, the Genetic Testing Registry established by the National Institutes of Health reported that genetic tests currently exist for more than 2,500 diseases, and that new discoveries are being made at an astonishing rate.(26)

Recent governmental initiatives have also been launched to support the uptake of new and emerging genomic technologies. For example, a joint initiative by Genome BC and the BC Cancer Foundation launched in 2011 will support the translation of genomic research into tailored and improved medical interventions for patients, as well as into disease prevention and early diagnosis.(27) In 2012, the Canadian Institutes of Health Research, through its Personalized Medicine Signature Initiative, and Genome Canada announced a \$65M joint initiative to support research projects in the fields of genomics and personalized medicine that could foster the development and validation of new genetic tests.(28)

At the provider and patient level, the enthusiasm towards genetics and genomics is also apparent. A survey of 341 Canadian family physicians, cardiologists and oncologists conducted in 2010 revealed that there is great optimism among physicians about the potential impact of genetic tests and personalized medicine on their practices, and 37% of respondents indicated that their patients had enquired about genetic testing and personalized medicine.(29) The rapid growth in the number of demands for genetic tests was also documented by an analysis conducted by the Molecular Oncology Task Force in Ontario, which revealed that referrals for cancer-related genetic testing rose by 61% in 22 clinics from 2002 to 2008.(30)

In B.C., it was estimated that 6,000 molecular genetic tests and 11,000 cytogenetic tests were conducted during the 2010-2011 period.(31) The BC Children's & Women's Hospital electronic laboratory reference manual lists approximately 60 genetics tests that are publicly funded for B.C. residents who are at risk for certain inherited diseases.(32) Other genetic tests are made publicly available through the BC Cancer Agency and are most often used for diagnosis, assessing prognosis, monitoring response to treatment and risk assessments for relapse for leukemias, lymphomas, breast, ovarian and colorectal cancer, as well as in cancer prevention (e.g., assessing susceptibility). Prenatal genetic tests are also routinely conducted through the BC

Box 3: Mobilizing research evidence about the problem

The available research evidence about the problem was sought from a range of published and “grey” research literature sources. Published literature that provided a comparative dimension to an understanding of the problem was sought using three health services research “hedgies” in MedLine, namely those for appropriateness, processes and outcomes of care (which increase the chances of us identifying administrative database studies and community surveys). Published literature that provided insights into alternative ways of framing the problem was sought using a fourth hedge in MedLine, namely the one for qualitative research. Grey literature was sought by reviewing the websites of a number of Canadian and international organizations, such as the Canadian Institute for Health Information, European Observatory on Health Systems and Policies, Genome BC, Genome Canada, Health Council of Canada, Health Evidence Network, Health Policy Monitor, and Organisation for Economic Co-operation and Development.

Priority was given to research evidence that was published more recently, that was locally applicable (in the sense of having been conducted in British Columbia or Canada), and that took equity considerations into account.

Prenatal Genetic Screening Program for congenital disorders such as Down syndrome and trisomy 18. Requests for genetic tests and related services have also increased significantly over the past years for a variety of diseases and associated conditions such as cardiovascular diseases, developmental delay, dysmorphic features, neurological disorders (e.g., neonatal seizures) and eye diseases (e.g., retinoblastoma).(33) In addition, gene-based testing for viral and bacterial infections (e.g., viral hepatitis) are increasingly identified as an important source of linked costs to genetic testing because the former are often processed using commercial testing kits rather than by leveraging the existing genetic lab testing infrastructure.

While the rapid growth in the number and types of genetic tests and related services that are becoming available may hold promise, there are concerns about the ‘media hype’ regarding genetic medicine and the resulting expectations that may contribute to increased demand.(25;34-38) Some have raised concerns that many genetic tests (e.g., mutation analysis, home testing and direct-to-consumer tests) are becoming available to clinicians and the public before their implications for patients and the health system have been fully examined.(24) For instance, while some genetic tests may hold promise in terms of health system cost savings, others may generate significant cost increases given the cascade of other tests, treatments and services that may be induced by them.(39) Thus, as Morgan et al. pointed out, “even good tests, when applied too broadly or without adequate information and support, can generate large, unwarranted cost impacts.”(39)

Furthermore, the ordering of genetic tests has shifted from being exclusively done by medical genetic specialists a decade ago to now include ordering by oncologists, neurologists, cardiologists, haematologists, ophthalmologists, microbiologists, pathologists and general practitioners. As a result, without greater coordination and processes for determining which tests and related services should be provided in what capacity, the increased availability of and demand for genetic tests and related services will put increased pressure on current programs and services in B.C., especially for common diseases and conditions.

Lack of coordinated programs and services

The Provincial Health Services Authority (PHSA) in British Columbia has the mandate to ensure that B.C. residents have access to a coordinated network of high-quality specialized healthcare services. With respect to genetic tests and related services, PHSA oversees:

- the BC Prenatal Genetic Screening Program, which focuses on three conditions (i.e., Down syndrome, trisomy 18 and open neural tube defects) and the detection of common chromosomal aneuploidies;
- the BC Cancer Agency’s Hereditary Cancer Program, which provides genetic counselling, testing, and related services for breast, ovarian and colorectal cancer, and several other hereditary cancer syndromes;
- the BC Cancer Agency’s genetic pathology laboratories, which focus on cancer cytogenetics and molecular oncology testing for leukemias, lymphomas, and certain solid tumors;
- the Provincial Medical Genetics Program hosted by the BC Children's Hospital and BC Women's Hospital & Health Centre, which provides counselling and testing services, as well as educational interventions (for the public, students, and health professionals) related to congenital anomalies and genetic diseases in fetuses, children and adults; and
- the Public Health and Microbiology Reference Laboratory at the BC Centre for Disease Control, which provides genomic testing and related services for viral and bacterial infections.

In addition to these services, PHSA enters into performance contracts with regional health authorities (e.g., Vancouver Coastal Health and Fraser Health) whose hospitals may deliver genetic testing and related services. Three regional health authorities (Vancouver Island Health Authority, Interior Health and Northern Health) have also established laboratory services that are independent of PHSA. When medically necessary genetic tests are not available locally, requests for out-of-province testing must be made to the Medical Service Plan, which provides a review and appeal process administered by Ministry of Health staff.

British Columbia is unique amongst Canadian provinces in how genetic testing is also provided by commercial laboratories on a fee-for-service basis when requested by a physician. Test menus are expanding

and include testing for hemoglobinopathies, coagulation disorders and (on a user-pay basis) paternity testing. Thus, there is little or no restriction on who can order a genetic test.

Despite the resources provided through these programs and services, access to genetic tests and related services may vary within the province (e.g., between regional health authorities) due to a lack of coordination. The discreteness of these programs and services also limits their ability to efficiently respond to requests for diseases not included specifically within their remit. This issue is not limited to B.C. as there appears to be a large disparity in waiting times across Canada for patients to obtain consultation and genetic test results, with wait times varying from a few months up to two years depending on where patients live.⁽¹¹⁾ Given relative uncertainty about whether available resources (e.g., medical geneticists, genetic counsellors and laboratory technologists) will be able to match future demand, the ability to stabilize (and eventually reduce) wait times for programs and services remains uncertain.⁽⁴⁰⁾

Current health system arrangements limit the ability to efficiently manage these pressures

A variety of features about the delivery, financial and governance arrangements within B.C.'s health system may also limit the ability to efficiently manage demand for current and future tests and related services.

Delivery arrangements

It can be argued that the problem may be less about the actual use of and/or demand for genetic tests and related services and more about the lack of coordinated delivery. Whereas genetic tests and related services were once limited in scope and easily coordinated through a medical genetics programmatic 'gatekeeper,' they can now be ordered by many types of providers (e.g., general practitioners and various specialists). Further, due to reductions in equipment costs and availability of direct-to-consumer kits, genetic tests can now be processed/analyzed in many types of settings, such as commercial labs on a fee-for-service or user-pay basis. This has resulted in a fragmented approach across the province for coordinating the delivery of genetic tests and related services and has fostered the entrenchment of disincentives to communication across providers and settings. This resonates with a 2007 study exploring the views of senior lab directors and clinicians at publicly funded Canadian predictive genetic testing facilities.⁽³⁸⁾ When asked about the nature of their relationship with federal and provincial government representatives, the majority were unable to identify who had a role in the implementation of predictive genetic tests and resource allocation at their facility.

Another key gap is the lack of strategy to evaluate access to the laboratory and non-laboratory genetic human and capital resources necessary to meet anticipated needs and to determine where they should be deployed across the B.C. health system. Limited access to genetic professionals (e.g., medical geneticists, genetic counsellors and laboratory staff) has been identified as a serious problem in many jurisdictions.^(1;14) As evidence of this, 834 genetic tests were sent for processing out of the province in 2011-2012 because they were not currently available in B.C. due to insufficient human and capital resources or the lack of expertise given the rarity of certain conditions, or because of the appropriateness of regional (supranational)/global centralization.⁽³¹⁾ This can also generate significant inequities in access to genetic services, which is an acute problem observed in rural and remote areas.⁽⁴¹⁾

Another important challenge in genetic service delivery is the lack of high quality, comprehensive and clear packages of care for specific populations (e.g., the genetic tests that should be included in prenatal or newborn screening, tests that should be included in a comprehensive assessment of developmental delay, and ones that result in outcomes that require interpretation by a geneticist), as well as for particular diagnoses and diseases (e.g., including pre- and post-test counselling, psychological support for patients and their families). The nature of new and emerging genetic tests is such that they cannot be considered as isolated investigations, but rather as part of an integrated package of care that could lead to improved outcomes.^(13;14;42)

Lastly, in a context where the public is becoming more assertive and knowledgeable on health matters and increasingly demanding access to the benefits of personalized medicine, the lack of coordinated and proactive

strategies to increase genetic and genomic literacy among the public may appear problematic.(3;14;29) Increased genetic and genomic literacy could also support the public/patients/families in making informed decisions given the complex implications of genetic testing (i.e., shared decision-making),(43) and also encourage citizen involvement at the system level to ensure that the organization and delivery of genetic tests and related services are equitable and aligned to citizens' values, needs, preferences and experiential knowledge.

Financial arrangements

Financial arrangements in B.C.'s health system contribute to a lack of a coordinated funding and remuneration models for genetic testing and related services. Resource allocation decisions appear to be largely regionalized and localized, with two predominant funding models: funding through the global budget of hospitals and regional health authorities, and through a fee-for-service system for some genetic laboratory services.(38) Both funding models have downsides. On the one hand, global budgets have been perceived as lacking transparency and making it difficult for the B.C. Ministry of Health to monitor certain indicators (e.g., which tests have been performed and at what volumes). On the other hand, the fee-for-service system has also been identified as problematic because it may create incentives to choose laboratory techniques/technologies based on the existence of a fee code for billing the Medical Service Plan for insured services, as opposed to choosing other techniques/technologies that may be more cost-effective for the system but that are not on the fee code list. In addition, the adoption of replacement technologies are potentially hampered by the impact that new, less costly tests may have on the revenues of service providers paid on a fee-for-service basis.

Another gap in financial arrangements is that, with the exception of the list of 60 genetic tests that are publicly funded and routinely delivered through actual programs (32) and the small but growing number of genetic tests eligible for fee-for-service reimbursement under the Medical Service Plan, there is currently a lack of clear lists of covered/reimbursed genetic tests and related services (e.g., for rare and orphan diseases).

Governance arrangements

Current governance arrangements also contribute to the lack of coordination in the planning, delivery, funding and evaluation of genetic tests and related services. For instance, Canada is characterized by a fragmented policy landscape that can be attributed to the division of responsibilities between the federal and provincial/territorial governments regarding healthcare. This fragmentation creates impediments to interprovincial cooperation in the delivery of genetic tests and related services, and has resulted in great diversity across the country in who makes policy and coverage decisions regarding genetic testing and related services, and in how such decisions are made, using what types of frameworks, and on what terms.(11;23) Furthermore, the fragmentation is also apparent in the wide variations across provinces, with some offering a broader scope of tests and services than others.(11)

Another important gap in the governance arrangements is the lack of robust criteria, frameworks and processes to inform coverage decisions. Requests to add new tests to the Medical Service Plan fee schedule are considered in isolation from the system of globally funded genetic tests and services. Similarly, there are no clear parameters helping to define exactly what testing is encompassed under programmatically funded genetic testing services. Furthermore, the current process for making funding decisions regarding genetic tests that are not on the 'local menu,' or for diseases outside of the remit of current programs and services, seems to generate frustration among both policymakers and healthcare providers in B.C.. On the one hand, policymakers sometimes perceive that requests to add new genetic tests and services to the menu neglect the wider impacts of these demands on the healthcare system. In addition, there are concerns that a piecemeal approach to coverage decisions may generate inequitable access to genetic testing services. On the other hand, healthcare providers approaching the Medical Service Plan to formally request genetic tests sometimes perceive the process to be cumbersome, lacking transparency and accountability, and ultimately not contributing to the quality of patient care.

As policy and coverage decisions regarding genetic tests and related services come under more intense scrutiny, there is a need for robust criteria, frameworks and processes (e.g., health technology assessment, overarching oversight) to produce more informed, transparent, accountable and legitimate decisions.(37;38) Establishing robust health technology assessment frameworks and processes to address the challenges of genetics and genomics has been identified as a priority area for more than a decade in Canada.(13;14) Building health technology assessment capacities appears to be especially important since coverage decisions must be made in the context of uncertainties, and inherently involve complex (and potentially contentious) value judgments. Giacomini and colleagues identified four ‘grey zones’ related to coverage decisions regarding new and emerging genetic tests:

- 1) many genetic tests have new purposes and effects;
- 2) there are often unclear standards regarding “how good is good enough” in terms of effectiveness, efficiency and other evaluative criteria;
- 3) evaluative information is often missing, ambiguous or incomplete; and
- 4) the technological and delivery contexts are rapidly evolving, and this may alter the purposes, effects, and costs of genetic tests after coverage decisions have been made.(9)

In response to these challenges, several researchers and advisory bodies have attempted to develop frameworks and processes to assess new and emerging genetic tests (e.g., clinical, economic, social, ethical, organizational and legal implications),(6-10) and to support ongoing stakeholder deliberation about how genetic services should be shaped and delivered.(3;44;45)

Another important issue related to governance arrangements is the lack of clarity in the scope of practice regarding who should make decisions about whether a genetic test should be done (e.g., primary care physicians, specialists, genetic counsellors, laboratory personnel, ministry of health administrators, public health practitioners, midwives and/or patients), on what basis, and whether all tests can be ordered by any health professional.(24;38) There is also no governance model that can leverage opportunities for more cost-effective genetic testing, including ones afforded by the effective convergence of previously separate laboratory disciplines (e.g., cytogenetics and molecular genetics). These issues are particularly important given the tendency towards the integration of genetics and genomics in various medical specialties and in primary care, which is likely to necessitate new core competencies for the medical workforce as well as a re-definition of professional roles and responsibilities.(1;14) While Canadian nurses and physicians may see themselves as capable of sharing important clinical tasks with professionals specialized in genetics, the integration of genetics is likely to necessitate educational interventions targeting health professionals to build capacity and professional skills (e.g., guidelines for what genetic test is clinically indicated, for counselling, and for interpreting results).(46;47) This appears consistent with the findings of a systematic review conducted by Scheuner and colleagues who concluded that “the primary care workforce, which will be required to be on the front lines of the integration of genomics into the regular practice of medicine, feels woefully underprepared to do so.”(48) These findings may suggest a lack of the genetic content in the healthcare workforce curriculum as well as in current continuing education interventions.

The lack of overarching regulations or guidelines for the delivery of and quality assurance in all laboratory and non-laboratory services constitutes another challenge. Indeed, rapid developments in the field of genetics may also overwhelm the capacity of regulatory agencies to ensure the quality and accuracy of such tests.(30) Although B.C. laboratories are required to meet accreditation standards of the Diagnostic Accreditation Program,(49) the delivery system for genetic tests and related services has been characterized in the country as a whole by a lack of overarching regulations or guidelines for quality assurance and delivery for all laboratory and non-laboratory services.(30) In recent years, there have been several examples of erroneous pathology testing across the country (B.C., Manitoba, Ontario, Quebec, New Brunswick, and Newfoundland and Labrador), which revealed variations in standards from province to province (and sometimes from one laboratory to another) and highlighted the need to review existing practices and identify optimal delivery arrangements for laboratory services.(50) In response to these incidents, the Canadian Pathology and Laboratory Medicine Leadership Council was given a mandate in 2010 to “develop and coordinate strategies and initiatives to strengthen the teaching of pathology and laboratory medicine, improve quality management in laboratories across the country, and support and enhance health human resource planning.”(51) While this initiative may support the development of national standards, the Council’s recommendations are not binding

for the provinces and the Council lacks regulatory authority.(50) Another contextual factor to consider is the internationalization of genetic testing and related services (e.g., out-of-country genetic laboratory tests and direct-to-consumer genetic tests available on the internet) which raises new challenges for current regulatory frameworks as healthcare providers and consumers may turn to unlicensed laboratories to perform tests.(30;52) This example illustrates the need for overarching regulations or guidelines with clear lines of accountability and effective performance-management systems to address these pressing challenges.

Lastly, current regulatory gaps regarding the controversial practice of direct-to-consumer genetic tests raise concerns about their impact on publicly funded health systems.(30;53;54) It is expected that a lack of policy regarding direct-to-consumer testing may increase the workload of health professionals, and consequently increase health system expenditures.(55;56) As evidence of this, a recent survey conducted in the United States revealed that 78% of those who consider using direct-to-consumer genetic testing services would ask their physician for help in interpreting test results.(57)

Additional equity-related observations about the problem

An important element of the problem that requires further discussion is how access to genetic testing may disproportionately affect certain groups or populations. Although virtually anyone can be affected by a genetic disease or condition, we will focus our attention in this evidence brief on patients with cancer and pregnant women for illustrative purposes. These two populations are most directly affected by the problem given the significant amount of cancer-related and prenatal testing that is performed within the province (and across Canada).

The number of Canadians diagnosed with cancer continues to increase annually. It is estimated that an average of 20 people will be diagnosed every hour of every day, with some type of cancer, and eight people will die from cancer (with 22,100 new cases and 9,300 deaths in B.C. alone).(58) Whereas 70-75% of cancers are sporadic, it is estimated that 20-25% of cancers are multifactorial (i.e., caused by the interplay of multiple genes and environmental factors which increase the risk of developing cancer) and 5-10% are hereditary (i.e., associated with a single susceptibility gene (e.g., BRCA1 and BRCA2) usually passed through a family in an autosomal dominant fashion).(59) In the United States at least, the burden of cancer, specifically the risk of developing and dying from it, is greater in ethnic minorities and medically underserved populations.(60) The unequal burden of cancer raises questions regarding equity of access to genetic tests and related services for different ethnocultural populations, and for vulnerable and geographically isolated populations, including what it means for the design of packages of care, who delivers them and where.

Women in decision-making positions with respect to pregnancy and childbirth are greatly affected by genetic tests and related services. Today, prenatal testing for congenital anomalies are routinely offered through the BC Prenatal Genetic Program, for Down Syndrome, which is the most common chromosome problem with an incidence estimated at one per 700 births, but also for trisomy 18 (one per 7,000 births) and open neural tube defects (one per 1,000 births). A systematic review revealed that some of the most often cited sources of difficulty for women in making the decision to undergo prenatal testing for Down syndrome were the undue pressures from others, the anxiety, worry and fear generated by the procedures or the potential implications, as well the lack of understanding of the risks and benefits of prenatal testing.(61) These findings resonate with the perception of “unequally shared decision-making” in prenatal genetic services.(62) Such concerns appear particularly important with the advent of a new non-invasive prenatal test for fetal chromosomal abnormalities and the anticipated surge in numbers of pregnant women in need of counselling to face difficult decisions.(63)

THREE ELEMENTS FOR ADDRESSING THE PROBLEM

Many elements could be selected as a starting point for deliberations about better coordinating the use of genetic tests and related services in British Columbia. To promote discussion about the pros and cons of potentially viable solutions, we have selected three elements (among many) of a comprehensive and coordinated approach to address the problem mentioned above. The three elements were identified and validated during consultation with the Steering Committee and key informants. They were selected because they tackle the problem at three levels: the policy level, the organizational level and the patient/provider level. The three elements were not designed to be mutually exclusive. They could be pursued simultaneously or sequentially, or components could be drawn from each element to create a new (fourth) element. They are presented separately to foster deliberations about their respective components, the relative importance or priority of each, their interconnectedness and potential of or need for sequencing, and their feasibility.

Element 1 – Develop a comprehensive policy framework for the ongoing planning, funding, delivery and evaluation of genetic tests and related services

This element includes: 1) the development of a comprehensive framework that takes into consideration the medical, social, legal, ethical, financial and governance issues raised by genetic tests and related services; 2) the development of a list of covered/reimbursed genetic tests and related services (e.g., for rare and orphan diseases) and packages of care for specific populations (e.g., the genetic tests that should be included in prenatal or newborn screening); 3) greater coordination and integration (within the province and across provinces) of existing programs and services to increase the efficiency with which existing genetic services are used; and 4) a multi-year framework for assessing the extent of deficiencies and addressing them.

This element might involve:

- appointing a government task force (which could be a policy forum, research-driven process or combination of both) to develop a comprehensive framework to support policy and coverage decisions regarding genetic tests and related services;
- establishing a central administrative ‘gatekeeper’ or

Box 4: Mobilizing research evidence about elements of a comprehensive approach for addressing the problem

The available research evidence about elements of a comprehensive approach for addressing the problem was sought primarily from Health Systems Evidence (www.healthsystemsevidence.org), which is a continuously updated database containing more than 1,800 systematic reviews of delivery, financial and governance arrangements within health systems. The reviews were identified by first searching the database for reviews containing “gen*” in the title and/or abstract. Additional reviews were identified by searching the database for reviews addressing features of the elements that were not identified using “gen*” as keywords. In order to identify evidence about costs and/or cost-effectiveness, the NHS Economic Evaluation Database (available through the Cochrane Library) was also searched using a similar approach.

The authors’ conclusions were extracted from the reviews whenever possible. Some reviews contained no studies despite an exhaustive search (i.e., they were “empty” reviews), while others concluded that there was substantial uncertainty about the element based on the identified studies. Where relevant, caveats were introduced about these authors’ conclusions based on assessments of the reviews’ quality, the local applicability of the reviews’ findings, equity considerations, and relevance to the issue. (See the appendices for a complete description of these assessments.)

Being aware of what is not known can be as important as being aware of what is known. When faced with an empty review, substantial uncertainty, or concerns about quality and local applicability or lack of attention to equity considerations, primary research could be commissioned, or an element could be pursued and a monitoring and evaluation plan designed as part of its implementation. When faced with a review that was published many years ago, an updating of the review could be commissioned if time allows.

No additional research evidence was sought beyond what was included in the systematic review. Those interested in pursuing a particular element may want to search for a more detailed description of the element or for additional research evidence about the element.

'hub' to coordinate decision-making and deployment of resources for providing genetic tests and related services;

- establishing monitoring and evaluation mechanisms to support the government's role in the financing and delivery of genetic services;
- strengthening capacity in health technology assessment for genetic tests and related services;(14)
- developing increased capacity in health human resource planning for genetics and putting in place a shared multi-year plan for genetic expertise in the health system;(14) and
- coordinating and intensifying a strategy to inform, consult and engage consumers and relevant stakeholders in the governance of genetic tests and related services.(14;44)

We found five systematic reviews that address this element, of which only one review that was older (i.e., the search of studies was conducted more than five years ago) and of low-quality (64) included studies focused on genetics. Of the other four reviews, two were recent, of which one was high-quality (65) and one was medium quality.(66) None of the reviews addressed the sub-elements related to establishing monitoring and evaluation mechanisms, strengthening capacity in health technology assessment or developing increased capacity in health human resource planning.

The five systematic reviews addressed the sub-elements focused on appointing a government task force, establishing a central 'gatekeeper' or 'hub' and coordinating and intensifying a strategy to inform, consult and engage consumers and stakeholders. An older, low-quality review focused on issues faced by the Food and Drug Administration (FDA) in the United States related to the application of pharmacogenomics.(64) There was consensus across the included studies that there is a need for guidance and regulation of pharmacogenomics and that the FDA was the appropriate 'central' agency to lead efforts to address these challenges. Studies highlighted initiatives that could be used by the FDA to address these challenges, which included advisory groups, interagency collaboration, and clear and transparent regulatory policy that includes provisions for making decisions on a case-by-case basis.

One recent and high-quality review addressed the sub-elements related to appointing a government task force and establishing a central 'gatekeeper' or 'hub.'(65) This review found insufficient evidence to support the use of interagency collaboration to improve health outcomes. It also noted that collaboration across multiple agencies is difficult and potentially expensive, and that successful collaborations require common objectives as well as a monitoring and evaluation plan.(65) A recent medium-quality review similarly found insufficient evidence for the effects of public health partnerships on health outcomes, although the qualitative studies included in the review suggested that some partnerships increased the profile of health inequalities on local policy agendas (thereby raising the prospect that the profile of a coordinated approach to genetic testing and related services could also be raised).(66)

For the sub-element related to coordinating and intensifying a strategy to inform, consult and engage consumers and relevant stakeholders, a high-quality but older review found a lack of research to reliably assess the impact of consumer involvement on healthcare policy development.(67) An older medium-quality review found that training of patients and healthcare professionals is an important component for successfully involving cancer patients in research, policy and planning and practice.(68)

A summary of the key findings from the synthesized research evidence is provided in Table 1. For those who want to know more about the systematic reviews contained in Table 1 (or obtain citations for the reviews), a fuller description of the systematic reviews is provided in Appendix 1.

Table 1: Summary of key findings from systematic reviews relevant to Element 1 – Develop a comprehensive policy framework for the ongoing planning, funding and delivery of genetic tests and related services

Category of finding	Summary of key findings
Benefits	<ul style="list-style-type: none"> • Coordinating and intensifying a strategy to inform, consult and engage consumers and relevant stakeholders in the governance of genetic tests and related services <ul style="list-style-type: none"> ○ An older high-quality review found that the benefits of consumer involvement (as compared to no involvement) were greatest in activities related to the creation of patient information materials.(67)
Potential harms	<ul style="list-style-type: none"> • None identified
Costs and/or cost-effectiveness in relation to the status quo	<ul style="list-style-type: none"> • Establishing a central ‘gatekeeper’ or ‘hub’ to coordinate decision-making and deployment of resources for providing genetic tests and related services <ul style="list-style-type: none"> ○ A recent high-quality review noted that the implementation of collaborations amongst multiple agencies can be difficult and more expensive than standard models.(65)
Uncertainty regarding benefits and potential harms (so monitoring and evaluation could be warranted if the element were pursued)	<ul style="list-style-type: none"> • Uncertainty because no systematic reviews were identified <ul style="list-style-type: none"> ○ Establishing monitoring and evaluation mechanisms to support the government’s role in the financing and delivery of genetic services: ○ Strengthening capacity in health technology assessment for genetic tests and related services ○ Developing increased capacity in health human resource planning for genetics and putting in place a shared multi-year plan for genetic expertise in the health system • Uncertainty because no studies were identified despite an exhaustive search as part of a systematic review <ul style="list-style-type: none"> ○ n/a • No clear message from studies included in a systematic review <ul style="list-style-type: none"> ○ Coordinating and intensifying a strategy to inform, consult and engage consumers and relevant stakeholders in the governance of genetic tests and related services <ul style="list-style-type: none"> ○ A high-quality but older review found a lack of research to reliably assess the impact of consumer involvement on healthcare policy development.(67)
Key components of the element if it was tried elsewhere	<ul style="list-style-type: none"> • Establishing a central ‘gatekeeper’ or ‘hub’ to coordinate decision-making and deployment of resources for providing genetic tests and related services <ul style="list-style-type: none"> ○ A recent high-quality review suggests that successful collaborations between multiple agencies requires objectives that are clearly defined and relevant to all engaged in the partnership as well as a monitoring and evaluation plan to assess the effectiveness of the collaboration.(65) • Coordinating and intensifying a strategy to inform, consult and engage consumers and relevant stakeholders in the governance of genetic tests and related services <ul style="list-style-type: none"> ○ An older medium-quality review found that training of patients and healthcare professionals is an important component for successfully involving cancer patients in research, policy and planning and practice.(68)
Stakeholders’ views and experience	<ul style="list-style-type: none"> • Appointing a government task force to develop a comprehensive framework to support policy and coverage decisions regarding genetic tests and related services <ul style="list-style-type: none"> ○ An older low-quality review reported on several recommendations for the FDA from authors of included articles in relation to the application of pharmacogenomics, which included: incorporating regulations to protect research participants and patient data, more involvement of other centres, clear and transparent regulatory policy (including provisions for making decisions on a case-by-case basis), and greater firmness in enforcing regulations for genetic tests.(64) • Establishing a central ‘gatekeeper’ or ‘hub’ to coordinate decision-making and deployment of resources for providing genetic tests and related services <ul style="list-style-type: none"> ○ The qualitative studies included in a recent medium-quality review suggested that some public health partnerships increased the profile of health inequalities on local policy agendas.(66)

Element 2 – Develop a quality framework for genetic tests and related services

This element includes the development of overarching regulations and guidelines for the delivery of and quality assurance in genetic tests and related services.(11)

This element might involve:

- establishing mechanisms to support the coordination and integration of services in the province (including integrating with existing regulations and accreditation processes for laboratory services);
- increasing opportunities for the education and training of health professionals in genetics and new genetic medicine (i.e., family physicians, pediatricians, medical geneticists, genetic counsellors and laboratory personnel);(14)
- strategies to evaluate how many genetic specialist resources are needed/demanded (and where) and support credentialing processes for providers of genetic services (e.g., counsellors);
- strategies to support the implementation of regulations and guidelines for quality assurance/control
- implementing quality monitoring and improvement systems; and
- funding and remuneration mechanisms that support coordinated laboratory services within the province and appropriate use of genetic tests and related services.

We found 20 systematic reviews addressing this element, however, none focused directly on genetics (and six reviews did not provide sufficient information to determine whether the focus of any of the included studies was on genetics). Specifically, we found several high-quality reviews that outlined benefits related to several of the sub-elements. First, a recent high-quality review assessed the effect of supports during pre-licensure education of health professionals as a means of managing health-worker supply, and found that that social, academic (e.g., career guidance and mentorship) and financial supports contributed to sustaining student enrolment to graduation.(69) In addition, several high- and medium-quality reviews outlined benefits for interventions to support behaviour change related to clinical practice (e.g., supporting the implementation of guidelines), which include: audit and feedback,(70) education (including distribution of materials, meetings and/or outreach visits),(71;72) and the use of reminders or prompts.(71-74) Lastly, a recent high-quality review found small effects in six of seven studies for financial incentives to improve the quality of care.(75)

A summary of the key findings from the synthesized research evidence is provided in Table 2. For those who want to know more about the systematic reviews contained in Table 2 (or obtain citations for the reviews), a fuller description of the systematic reviews is provided in Appendix 2.

Table 2: Summary of key findings from systematic reviews relevant to Element 2 – Develop a quality framework for genetic tests and related services

Category of finding	Summary of key findings
Benefits	<ul style="list-style-type: none"> • Establishing mechanisms to support the coordination and integration of services in the province (including integrating with existing regulations and accreditation processes for laboratory services) <ul style="list-style-type: none"> ○ A recent medium-quality review found that integrated care pathways are most effective in situations where deficiencies in service have been identified and where patient care pathways are predictable.(76) • Increased opportunities for the education and training of health professionals in genetics and new genetic medicine (i.e., family physicians, pediatricians, medical geneticists, genetic counsellors and laboratory personnel) <ul style="list-style-type: none"> ○ A recent medium-quality review found large positive effects for internet-based learning as compared to those receiving no intervention, but when compared to traditional methods, effects were comparable.(77) ○ Another medium-quality but older review found evidence to suggest that continuing medical education is effective in supporting knowledge acquisition and behaviour change (including attitudes, skills, practice behaviour and clinical practice outcomes).(78) • Strategies to evaluate how many genetic specialist resources are needed/demanded (and

	<p>where) and support credentialing processes for providers of genetic services (e.g., counsellors)</p> <ul style="list-style-type: none"> ○ A recent high-quality review assessed the effect of pre-licensure education of health professionals on health-worker supply and found that that student social, academic (e.g., career guidance and mentorship) and financial support contributed to sustaining student enrolment to graduation.(69) ● Strategies to support the implementation of regulation and guidelines for quality assurance/control <ul style="list-style-type: none"> ○ Several high- and medium-quality reviews found benefits for interventions to support behaviour change related to clinical practice, which include: audit and feedback,(70) education (including distribution of materials, meetings and/or outreach visits),(71;72) and the use of reminders or prompts.(71-74) ● Implementing quality monitoring and improvement systems <ul style="list-style-type: none"> ○ A medium-quality but older review found a positive effect for collaborative quality improvement interventions across nine randomized controlled trials on processes of care and patient care.(79) ○ Another review that was conducted recently but was of low quality found clinician/patient driven quality-improvement interventions were effective, and that manager/policymaker driven approaches were less effective.(80) ● Funding and remuneration mechanisms that support coordinated laboratory services within the province and appropriate use of genetic tests and related services <ul style="list-style-type: none"> ○ A recent high-quality review found small effects in six of seven studies for financial incentives to improve the quality of care.(75) ○ An older low-quality review found that alternative forms of remuneration or financial incentives such as fund-holding, capitation and caps on provider income can be used to reduce the use of healthcare resources, support adherence clinical guidelines or achieve health targets.(81)
Potential harms	<ul style="list-style-type: none"> ● Funding and remuneration mechanisms that support coordinated laboratory services within the province and appropriate use of genetic tests and related services <ul style="list-style-type: none"> ○ An older low-quality review noted that the risk of financial incentives is that they result in limited access to certain types of care, contribute to fragmentation of care across providers and settings, and produce conflicts of interest between providers and those receiving care.(81)
Costs and/or cost-effectiveness in relation to the status quo	<ul style="list-style-type: none"> ● Establishing mechanisms to support coordination and integration of services in the province (including integrating with existing regulations and accreditation processes for laboratory services) <ul style="list-style-type: none"> ○ An older high-quality review assessed the effectiveness of near patient testing and delivery systems between laboratory and general practice, but found insufficient data to be able to draw conclusions about cost-effectiveness.(82)
Uncertainty regarding benefits and potential harms (so monitoring and evaluation could be warranted if the element were pursued)	<ul style="list-style-type: none"> ● Uncertainty because no systematic reviews were identified <ul style="list-style-type: none"> ○ n/a ● Uncertainty because no studies were identified despite an exhaustive search as part of a systematic review <ul style="list-style-type: none"> ○ Establishing mechanisms to support coordination and integration of services in the province (including integrating with existing regulations and accreditation processes for laboratory services) <ul style="list-style-type: none"> ○ An older high-quality review assessed the effectiveness of near patient testing and delivery systems between laboratory and general practice, but found no evaluations.(82) ● No clear message from studies included in a systematic review <ul style="list-style-type: none"> ○ Establishing mechanisms to support coordination and integration of services in the province (including integrating with existing regulations and accreditation processes for laboratory services) <ul style="list-style-type: none"> ○ A recent but medium-quality review noted that the value of integrated care pathways is less certain in contexts where inter-professional working is well established.(76) ○ Increased opportunities for the education and training of health professionals in genetics and new genetic medicine (i.e., family physicians, pediatricians, medical geneticists, genetic counsellors, and laboratory personnel) <ul style="list-style-type: none"> ○ An older and low-quality review found mixed findings with respect to the effectiveness of internet-based continuing medical education as compared to traditional instruction methods.(83) ○ Funding and remuneration mechanisms that support coordinated laboratory services within the province and appropriate use of genetic tests and related services <ul style="list-style-type: none"> ○ An older, medium-quality review assessed the effectiveness of quality-based purchasing strategies, which included incentives such as providing additional fee-for-service

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	<p>payments, quality bonuses and public release of performance data, however, findings from the nine included trials for the 11 performance indicators were mixed.(84)</p>
<p>Key components of the element if it was tried elsewhere</p>	<ul style="list-style-type: none"> • Establishing mechanisms to support coordination and integration of services in the province (including integrating with existing regulations and accreditation processes for laboratory services) <ul style="list-style-type: none"> ○ A recent but low-quality review of models for genetics in healthcare found that multidisciplinary specialist clinics and coordinating services between professionals in first-, second- and third-line medical care are key components for providing genetics-related services in healthcare.(1) • Increased opportunities for the education and training of health professionals in genetics and new genetic medicine (i.e., family physicians, pediatricians, medical geneticists, genetic counsellors, and laboratory personnel) <ul style="list-style-type: none"> ○ Findings from a recent but low-quality review suggest that that online learning is most effective when it provides flexibility for students, provides support and rapid assessment for students and facilitates efficient communication.(85) • Implementing quality monitoring and improvement systems <ul style="list-style-type: none"> ○ A low-quality review that was published in 2007 (but in which the date of last search was not reported) examined practices for using public reporting of performance as a way of improving healthcare quality, and suggests that for public reporting to be effective it should focus on information directly related to a program’s objectives, audience, content, product, distribution and impacts.(86)
<p>Stakeholders’ views and experience</p>	<ul style="list-style-type: none"> • Increased opportunities for the education and training of health professionals in genetics and new genetic medicine (i.e., family physicians, pediatricians, medical geneticists, genetic counsellors, and laboratory personnel) <ul style="list-style-type: none"> ○ A recent but low-quality review assessing e-learning reported that health professionals found the flexibility provided to be an advantage as it allowed them to choose when and where they can participate in a course as well as the mediums used for the courseware.(85)

Element 3 – Develop a framework to support consumer/patient/family decision-making about genetic tests and related services

This element includes helping consumers, patients and their families to make informed decisions regarding genetic tests and their implications. It is designed to support a coordinated and standardized approach to engage patients and their families in decisions regarding genetic tests and related services, but also to support healthcare providers in offering consistent guidance.

This element might involve:

- strategies to involve consumers/patients/families in decisions about genetic tests and related services to encourage them to express their beliefs, values and preferences about treatments and care, or to optimize communication between them and their healthcare providers; and
- developing decision aids or decision support systems for consumers/patients about different screening or treatment options (pre and post-test).

We found 10 systematic reviews addressing this element, of which four addressed strategies to involve consumers/patients and their families in health-related decisions, and of which one high-quality (87) and two medium-quality reviews (88;89) reported including studies focused on genetics in the context of decision aids or decision support systems for consumers/patients (and three reviews did not provide sufficient information to determine whether the focus of any of the included studies was on genetics). One high-quality but older review found that consumer involvement helped produce information materials for patients that were more relevant, understandable and easy to read.(67) Another review that was older and of medium-quality found several benefits related to involving patients in healthcare such as improved self-esteem, updated and improved patient information resources, and development of an organizational culture that is supportive to patient involvement.(90) Two recent systematic reviews evaluating decision aids (one high-quality and one medium quality) found benefits in terms of knowledge of screening and treatment options, patient involvement and realistic perception of outcomes.(91;92) Another recent, medium-quality review found benefits for genetic counselling for improving the accuracy of risk perception.(88) Lastly, three systematic reviews (two high-quality and one medium-quality) found benefits for several strategies designed to communicate risk to patients.(87;89;93)

A summary of the key findings from the synthesized research evidence is provided in Table 3. For those who want to know more about the systematic reviews contained in Table 3 (or obtain citations for the reviews), a fuller description of the systematic reviews is provided in Appendix 3.

Table 3: Summary of key findings from systematic reviews relevant to Element 3 – Develop a framework to support consumer/patient/family decision-making about genetic tests and related services

Category of finding	Summary of key findings
Benefits	<ul style="list-style-type: none"> • Strategies to involve consumers/patients/families in decisions about genetic tests and related services to encourage them to express their beliefs, values and preferences about treatments and care, or to optimize communication between them and their healthcare providers (i.e., shared decision-making): <ul style="list-style-type: none"> ○ A high-quality but older review found research evidence indicating that the involvement of consumers during the creation of information materials for patients results in products that are more relevant, understandable and easy to read.(67) ○ An older review of medium-quality found several benefits related to the involvement of patients in the planning and development of healthcare, which include: improved self-esteem for patients; rewarding experience for healthcare staff; production of updated/improved patient information resources; simplified appointment procedures; more efficient transportation between treatment sites; improved access for people with disabilities; and organizational attitudes that are supportive to patient involvement.(90) • Developing decision aids or decision support systems for consumers/patients about different screening or treatment options (pre- and post-test): <ul style="list-style-type: none"> ○ Two recent reviews evaluated the use of decision-aids. A high-quality review found increases

	<p>in knowledge, patient involvement and realistic perception of outcomes among patients who received decision aids.(92) The review also found that decision aids reduced decision-related conflict and increased patient-practitioner communication. The second review, which was of medium-quality, found that cancer-related decision aids increased knowledge of screening and preventive/treatment options without increasing anxiety.(91)</p> <ul style="list-style-type: none"> ○ One recent medium-quality review assessed the impact of genetic counselling on the accuracy of risk perception. The review found an increase in the proportion of accurate risk assessment in participants after receiving counselling, and the changes were sustained at one-year follow-up.(88) ○ Three systematic reviews assessed strategies for communicating risk. An older high-quality review found three studies reporting that personalized risk communication interventions supported accurate risk perception, and three other studies found that it increased knowledge.(87) A high-quality recent review found that framing messages positively resulted in more positive perceptions of effectiveness, but did not change the persuasiveness of the message. One medium-quality review found that providing information about screening and framing the information with messages of loss led to a more positive perception of the effectiveness of what could be gained.(93) A medium-quality review published in 2009 (for which the year of last search was not provided) found that tailoring information about cancer risk and screening options increased both patient knowledge and accurate perception of risk as compared to those receiving generic information.(89)
Potential harms	<ul style="list-style-type: none"> ● None identified
Costs and/or cost-effectiveness in relation to the status quo	<ul style="list-style-type: none"> ● None identified
Uncertainty regarding benefits and potential harms (so monitoring and evaluation could be warranted if the element were pursued)	<ul style="list-style-type: none"> ● Uncertainty because no systematic reviews were identified <ul style="list-style-type: none"> ○ n/a ● Uncertainty because no studies were identified despite an exhaustive search as part of a systematic review <ul style="list-style-type: none"> ○ n/a ● No clear message from studies included in a systematic review <ul style="list-style-type: none"> ○ Strategies to involve consumers/patients/families in decisions about genetic tests and related services to encourage them to express their beliefs, values and preferences about treatments and care, or to optimize communication between them and their healthcare providers (i.e., shared decision-making) <ul style="list-style-type: none"> ○ A low-quality older review found limited evidence related to methods for communicating evidence to patients to help support their understanding and involvement in decisions. ○ Developing decision aids or decision support systems for consumer/patients about different screening or treatment options (pre- and post-test) <ul style="list-style-type: none"> ○ An older, high-quality review found limited evidence to determine whether personalizing risk communication (either in writing, verbally or visual presentations) increased the uptake of screening tests or supported informed decision-making.(87) ○ A medium-quality review found limited evidence from which to be able to assess the effectiveness of using a website tailored to cancer risk factors to increase patient's knowledge and perceptions of risk.(89)
Key components of the element if it was tried elsewhere	<ul style="list-style-type: none"> ● Strategies to involve consumers/patients/families in decisions about genetic tests and related services to encourage them to express their beliefs, values and preferences about treatments and care, or to optimize communication between them and their healthcare providers (i.e., shared decision-making) <ul style="list-style-type: none"> ○ A low-quality, older review provided recommendations based on limited evidence as well as expert opinion about key components that should be used by physicians to communicate clinical evidence to patients, which include: understanding the patient's experience and expectations; building partnership; providing evidence (including a discussion of limitations/uncertainties); presenting recommendations informed by clinical judgement and patient preferences; and checking for understanding and agreement.(94)
Stakeholders' views and experience	<ul style="list-style-type: none"> ● Strategies to involve consumers/patients/families in decisions about genetic tests and related services to encourage them to express their beliefs, values and preferences about treatments and care, or to optimize communication between them and their healthcare providers (i.e., shared decision-making) <ul style="list-style-type: none"> ○ A medium-quality older review of patient preferences for involvement in their cancer treatment found that most prefer a collaborative role in decisions about treatment options, with far fewer indicating they would prefer to take a passive or active role. However, findings from the same review indicate that the role they actually played in decision-making about treatment options was not reflective of their preferences.(95)

Additional equity-related observations about the three elements

Very few of the identified systematic reviews addressed the two populations prioritized in this evidence brief (patients with cancer and pregnant women) and those that did only provided information related to patients with cancer. One recent and high-quality systematic review analyzing the effect of changes in the method and level of payment on standards of care (which is a component of the second element) included studies related to cancer screening and found small effects of financial incentives on improving the quality of care.⁽⁷⁵⁾ In addition, three systematic reviews identified for the third element included studies with a focus on patients with cancer. One older, medium-quality review evaluated patient preferences for their involvement in cancer treatment and found that most prefer a collaborative role in treatment decision-making.⁽⁹⁵⁾ A medium-quality review found that tailored information related to cancer risk increased knowledge about screening and accurate perceptions of risk.⁽⁸⁹⁾ Lastly, another medium-quality review found cancer-related decision aids increased knowledge of preventative and treatment options.⁽⁹¹⁾

IMPLEMENTATION CONSIDERATIONS

Potential barriers to the implementation of a comprehensive approach to coordinating the use of genetic tests and related services in B.C. can be identified at the level of patients/individuals (e.g., the asymmetry of knowledge and expertise between patients and providers, frustration with past efforts), providers (e.g., perceived infringement on autonomy, lack of awareness of shared decision-making approaches, lack of financial incentives), organizations (e.g., making decision in a context of genetic ‘hype’), and system level (e.g., challenges of cross-provincial coordination and standardization). A detailed list of potential barriers to implementing the three elements is provided in Table 4. We found few empirical studies that helped to identify or establish the importance of these barriers, so we have listed those that were identified in a range of sources (not just empirical studies) and we have not rank ordered them in any way.

Table 4: Potential barriers to implementing the elements

Levels	Element 1 – Develop a comprehensive policy framework for the ongoing planning, funding, delivery and evaluation of genetic tests and related services	Element 2 – Develop a quality framework for genetic tests and related services	Element 3 – Develop a framework to support consumer/patient/family decision-making about genetic tests and related services
Patient/individual	<ul style="list-style-type: none"> • Asymmetry of knowledge and expertise between patients and providers (96) • Difficulty of engaging individuals who can articulate diverse viewpoints, which limits the influence of some communities on health system policymaking and leads to concerns about “tokenism” • Frustration among the public if their past efforts to inform health system policymaking were not influential 	<ul style="list-style-type: none"> • Asymmetry of knowledge and expertise between patients and providers (96) • Difficulty of engaging individuals who can articulate diverse viewpoints, which limits the influence of some communities on health system policymaking and leads to concerns about “tokenism” • Frustration among the public if their past efforts to inform health system policymaking were not influential 	<ul style="list-style-type: none"> • Lack of access to actual tests and related services (97;98) • Lack of applicability due to patient characteristics or clinical situation (97;98) • Perceived patient preferences for a model of decision-making that did not fit a shared decision-making approach (SDM) (97;98)
Service provider	<ul style="list-style-type: none"> • Service providers may be reluctant to undertake a redistribution or reconfiguration of roles and responsibilities (1) • Service providers may perceive the potential for infringement on their decision-making autonomy • Service providers are having to make decisions in the context of ‘hype’/heightened consumer demand for specific genetic tests and related services (37;38) • Service providers may be resistant to consumer involvement (67) • Service providers may be resistant to external/centralized oversight • Service providers may face a loss of revenue under service consolidation and replacement 	<ul style="list-style-type: none"> • Service providers may be reluctant to undertake a redistribution or reconfiguration of roles and responsibilities (1) • Lack of preparedness and/or training of non-genetics professionals (1) • Challenge of standardizing professional practices 	<ul style="list-style-type: none"> • Challenge of standardizing professional practices • Service providers may be reluctant to undertake a redistribution or reconfiguration of roles and responsibilities (1) • Service providers may be unwilling or hesitant to fully support a SDM approach (97;98) • Service providers may not have sufficient time and resources to implement SDM (97;98) • Service providers may not be aware of or in agreement with specific components of SDM approach (97;98) • Service providers may perceive a threat to their professional autonomy and authority (97;98) • Service providers may have concerns about malpractice liability (97;98)

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	<p>testing</p> <ul style="list-style-type: none"> • Unavailability of arms-length subject matter experts can expose the system to self-serving advisors 		<ul style="list-style-type: none"> • Financial incentives in the fee-for-service model do not support SDM (97;98) • Lack of preparedness and/or training of non-genetics professionals (1;48)
Organization	<ul style="list-style-type: none"> • Organizational leaders are having to make decisions in the context of 'hype'/heightened consumer demand for specific genetics tests and related services (37;38) • Organizational leaders may be resistant to consumer involvement (67) 	<ul style="list-style-type: none"> • Organizational leaders may have insufficient support / funding from government 	<ul style="list-style-type: none"> • Organizational leaders may have insufficient support / funding from government (97;98)
System	<ul style="list-style-type: none"> • Policymakers have to make decisions in the context of 'hype'/heightened consumer demand for specific genetics tests and related services (37;38) • Policymakers have to make decisions based on the legacy of platform technologies, which runs the risk of being overly conservative and failing to appropriately embrace opportunities • Challenges of cross-provincial coordination and standardization (38) • Multi-agency/collaborative initiatives are subject to changes outside the control of the partnership (65) • Policymakers may be resistant to consumer involvement (67) 	<ul style="list-style-type: none"> • Challenges of cross-provincial coordination and standardization (38) • Policymakers may face insufficient support and funding across the system 	<ul style="list-style-type: none"> • Policymakers may face insufficient support and funding across the system

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APPENDICES

The following tables provide detailed information about the systematic reviews identified for each element. Each row in a table corresponds to a particular systematic review and the reviews are organized by element (first column). The focus of the review is described in the second column. Key findings from the review that relate to the element are listed in the third column, while the fourth column records the last year the literature was searched as part of the review.

The fifth column presents a rating of the overall quality of the review. The quality of each review has been assessed using AMSTAR (A MeaSurement Tool to Assess Reviews), which rates overall quality on a scale of 0 to 11, where 11/11 represents a review of the highest quality. It is important to note that the AMSTAR tool was developed to assess reviews focused on clinical interventions, so not all criteria apply to systematic reviews pertaining to delivery, financial, or governance arrangements within health systems. Where the denominator is not 11, an aspect of the tool was considered not relevant by the raters. In comparing ratings, it is therefore important to keep both parts of the score (i.e., the numerator and denominator) in mind. For example, a review that scores 8/8 is generally of comparable quality to a review scoring 11/11; both ratings are considered “high scores.” A high score signals that readers of the review can have a high level of confidence in its findings. A low score, on the other hand, does not mean that the review should be discarded, merely that less confidence can be placed in its findings and that the review needs to be examined closely to identify its limitations. (Lewin S, Oxman AD, Lavis JN, Fretheim A. SUPPORT Tools for evidence-informed health Policymaking (STP): 8. Deciding how much confidence to place in a systematic review. *Health Research Policy and Systems* 2009; 7 (Suppl1):S8.

The last three columns convey information about the utility of the review in terms of local applicability, applicability concerning prioritized groups, and issue applicability. The third-from-last column notes the proportion of studies that were conducted in Canada, while the second-from-last column comments on the proportion of studies included in the review that deal explicitly with one of the prioritized groups. The last column indicates the review’s issue applicability in terms of the proportion of studies focused on genetics.

All of the information provided in the appendix tables was taken into account by the evidence brief’s authors in compiling Tables 1-3 in the main text of the brief.

Appendix 1: Systematic reviews relevant to Element 1 - Develop a comprehensive policy framework for the planning, funding and delivery of genetic tests and related services

Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
A government-appointed task force (policy forum or research-driven process) to develop a comprehensive framework to support policy and coverage decisions regarding genetic tests and related services	Evaluates the impact of collaboration between government agencies and local health agencies on health outcomes.(65)	<p>Interagency collaboration between local health agencies and government is often accepted as best practice. However, there is insufficient evidence to support the use of interagency collaboration to improve health outcomes.</p> <p>The review notes collaboration amongst multiple agencies is often difficult to implement, more expensive than standard service delivery, and may undergo changes due to external factors.</p> <p>Successful collaborations require clearly outlined objectives that are relevant to all agencies involved in the partnership, monitoring of all outcomes and include an evaluation process to assess effectiveness.</p>	2009	9/9 (AMSTAR rating from Program in Policy Decision-making)	0/11	0/11	0/11
	To identify issues faced by the Food and Drug Administration (FDA) relevant to the application of pharmacogenomics.(64)	There was consensus across the included studies that there is a need for guidance and/or regulation of pharmacogenomics for the protection of public safety, enhancement of drug effectiveness, and to create incentives to develop these new technologies. There is also consensus that the FDA is the appropriate agency to take a leadership role in meeting these challenges. Key initiatives from the FDA that were highlighted include the development of advisory groups and interagency collaborative efforts, sponsorship of conferences and symposia, development of staff	2006	2/9 (AMSTAR rating from Program in Policy Decision-making)	Not reported in detail- Description states: U.S.A.	0/210	210/210

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Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
		<p>expertise, and development of guidance for genomic data submission, new testing approaches, and co-development of tests and drugs.</p> <p>Several recommendations from articles by authors outside the FDA include regulating the appropriate protections for research participants and patient data, more involvement of multiple FDA centres including a new centre, regulatory policy that is transparent and clearly defined, regulation considered on a case-by-case basis for pharmacogenomics, and generally greater firmness by the FDA for regulating genetic tests</p>					
<p>Establishing a central 'gatekeeper' or 'hub' to coordinate decision-making and deployment of resources for providing genetic tests and related services</p>	<p>Evaluates the impact of collaboration between government agencies and local health agencies on health outcomes.(65)</p>	<p>Interagency collaboration between local health agencies and government is often accepted as best practice. However, there is insufficient evidence to support the use of interagency collaboration to improve health outcomes.</p> <p>The review notes collaboration amongst multiple agencies is often difficult to implement, more expensive than standard service delivery, and may undergo changes due to external factors.</p> <p>Successful collaborations require clearly outlined objectives that are relevant to all agencies involved in the partnership, monitoring of all outcomes and include an evaluation process to assess effectiveness.</p>	<p>2009</p>	<p>9/9 (AMSTAR rating from Program in Policy Decision-making)</p>	<p>0/11</p>	<p>0/11</p>	<p>0/11</p>

Coordinating the Use of Genetic Tests and Related Services in British Columbia

Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
	Impact of organizational partnerships on public health outcomes (health improvement and/or a reduction in health inequalities) in England between 1997 and 2008.(66)	<ul style="list-style-type: none"> Findings suggest that there is not yet any clear evidence of the effects of public health partnerships on health outcomes. However, qualitative studies suggested that some partnerships increased the profile of health inequalities on local policy agendas. Both the design of partnership interventions and of the studies evaluating them meant it was difficult to assess the extent to which identifiable successes and failures were attributable to partnership working. 	2008	7/10 (AMSTAR rating from the McMaster Health Forum)	0/15	0/15	0/15
Establishing monitoring and evaluation mechanisms to support the government's role in financing and delivery of genetic services	No reviews identified	n/a	n/a	n/a	n/a	n/a	n/a
Strengthened capacity in health technology assessment for genetic tests and related services	No reviews identified	n/a	n/a	n/a	n/a	n/a	n/a
Developing increased capacity in health human resource planning for genetics and putting in place	No reviews identified	n/a	n/a	n/a	n/a	n/a	n/a

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Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
a shared multi-year plan for genetic expertise in the health system							
Coordinated and intensified strategy to inform, consult and engage consumers and relevant stakeholders in the governance of genetic tests and related services	Evaluates the effects of consumer involvement and compares various methods of involvement in healthcare policy development and related research, clinical practice guidelines and information materials developed for patients.(67)	<p>The impact of consumer involvement on healthcare policy creation and related research and the development of clinical practice guidelines remains uncertain due to lack of research.</p> <p>The review notes the benefits of consumer involvement, in comparison to no consumer involvement, appear to be greatest in the creation of patient information materials</p> <p>Research evidence indicates involving consumers in the creation of information materials for patients produces material with improved readability and relevance, and that is more understandable to patients without elevating their anxiety.</p>	2005	9/11 (AMSTAR rating from www.rxforchange.ca)	0/6	Not reported	0/6
	Effectiveness of the agenda of involvement of people affected by cancer in research, policy and planning, and practice.(68)	<p>Training of patients and healthcare professionals is necessary for successful involvement of cancer patients in research, policy and planning, and practice.</p> <p>Patient involvement requires personnel and financial support.</p> <p>The opposing ideologies of individualism and collectivism are the most common rationales as to why people affected by cancer should be involved in research, policy and planning, and practice.</p>	2004	4/9 (AMSTAR rating from Program in Policy Decision-making)	Not reported in detail - Description states: Canada, U.K., U.S.A.	Not reported	0/131

Appendix 2: Systematic reviews relevant to Element 2 – Develop a quality framework for genetic tests and related services

Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
Establishing mechanisms to support coordination and integration of services in the province (including integrating with existing regulations and accreditation processes for laboratory services)	Analysis of current and emerging service delivery models for genetic services in Europe, North America and Australia, and associated integration challenges.(1)	<p>Coordination of the activities of first-, second-, and third-line medical care professionals is necessary for successful genetic adult care models.</p> <p>Political and cultural obstacles in the healthcare field must be overcome, including non-existent or weak relationships between professional organizations and poor levels of genetic proficiency by non-genetic professionals and the healthcare system.</p> <p>Relevant public health policies need to be implemented for the widespread implementation of delivery models.</p>	Not available.	2/9 (AMSTAR rating from Program in Policy Decision-making)	Not reported in detail-description states: Europe, North America and Australia	Not reported in detail (review provides findings related to cancer but unclear how many studies focus on cancer)	Not reported
	Effectiveness of integrated care pathways in children and adults, over a full range of health settings, which were successful.(76)	<p>Integrated care pathways are most effective in contexts where patient care trajectories are predictable.</p> <p>Their value in settings in which recovery pathways are more variable is less clear.</p> <p>Integrated care pathways are most effective in bringing about behavioural changes where there are identified deficiencies in services</p> <p>Their value in contexts where inter-professional working is well established is less certain.</p> <p>None of the studies reviewed included an economic evaluation and thus it is not</p>	2008	7/10 (AMSTAR rating from Program in Policy Decision-making)	U.K. (4); Australia (2); U.S.A. (2); Italy (1)	Not reported	Not reported

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Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
		known whether their benefits justify the costs of their implementation.					
	Effectiveness of near patient testing and the use of other delivery systems between laboratory and general practice.(82)	There is little evidence to support the general introduction of near patient testing in general practice. Near patient testing may provide additional value to patients in the specific clinical areas of early diagnosis, screening, and monitoring of chronic disease.	1995	9/10 (AMSTAR rating from Program in Policy Decision-making)	Not reported	Not reported	0
Increased opportunities for the education and training of health professionals in genetics and new genetic medicine (i.e., family physicians, pediatricians, medical geneticists, genetic counsellors, and laboratory personnel)	Analysis of the experiences of health professionals' with e-learning.(85)	The effectiveness of on-line learning is mediated by the learning experience. To enhance health professionals' experience of e-learning, courses need to address presentation and course design; they must be flexible, offer mechanisms for both support and rapid assessment, and develop effective and efficient means of communication, especially among the students themselves Health professionals reported the flexibility of e-learning as an advantage, both in terms of when and where they can choose to participate in the course, and also in terms of the mediums of courseware, such as CD-ROMs and printable materials.	2007	2/9 (AMSTAR rating from Program in Policy Decision-making)	0/19	0/19	0/19
	Effects of Internet-based learning for health professionals compared with no intervention and with non-internet interventions.(77)	Internet-based learning had a consistently positive effect compared to no internet intervention.	2007	6/11 (AMSTAR rating from www.rxfchange.ca)	Not reported	Not reported	0/201
	Effectiveness of specific continuing medical education tools and techniques for disseminating and retaining medical	Overall, the literature suggested that continuing medical education was effective in achieving and maintaining the	2006	7/11 (AMSTAR rating from www.rxfchange.ca)	Not reported	Not reported	0/145

Coordinating the Use of Genetic Tests and Related Services in British Columbia

Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
	knowledge.(78)	objectives studied: knowledge (22 of 28 studies); attitudes (22 of 26); skills (12 of 15); practice behaviour (61 of 105); and clinical practice outcomes (14 of 33). Live media, multimedia and multiple exposures were more effective than print, single media and single exposure, respectively.					
	Effectiveness of internet-based continuing medical education interventions for health care outcomes and physician performance.(83)	Six studies showed a positive change in participant knowledge with the use of internet-based continuing medical education over traditional avenues. Three studies showed a positive change in practice with the use of continuing medical education. The remaining seven studies showed no difference in knowledge between internet-based and traditional forms of continuing medical education.	2004	2/9 (AMSTAR rating from Program in Policy Decision-making)	Not reported	Not reported	0/16
Strategies to evaluate how many genetic specialist resources are needed/demanded (and where) and support credentialing processes for providers of genetic services (e.g., counsellors)	Assessment of the effect on health-worker supply of changes in the pre-licensure education of health professionals.(69)	Student support activities comprised successful interventions to maintain student enrolment in pre-licensure education. Social, academic and career guidance and mentorship, as well as financial assistance, all contributed to sustaining enrolment to graduation.	2007	10/10 (AMSTAR rating from Program in Policy Decision-making)	0/2	0/2	0/2
Strategies to support the implementation of regulation and	Assessment of the impact of audit and feedback on both patient outcomes and the practice of healthcare professionals.(70)	Audit and feedback alone compared to no other interventions (n=38), audit and feedback with educational meetings compared to no intervention (n=9), audit	2006	8/11 (AMSTAR rating from www.rxforchange.ca)	9/118	Not reported	0/118

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Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
guidelines for quality assurance/control		<p>and feedback as part of a multifaceted intervention compared to no intervention (n=41) and audit and feedback combined with complementary interventions compared to audit and feedback alone (n=24), and audit and feedback compared to other interventions (n=8), audit and feedback was found to be generally effective.</p> <p>Effectiveness of audit and feedback is relative and is influenced by baseline adherence to recommended practice, intensity of feedback, and if used in conjunction with educational meetings.</p> <p>The review notes the effectiveness of audit and feedback may improve if health professionals are actively involved and given responsibilities for bringing about change.</p>					
	Assessment of the relative effectiveness of various quality improvement strategies for enhancing healthcare.(80)	<p>Research evidence suggests clinician/patient driven quality improvement strategies are more effective compared to manager/policymaker driven approaches.</p> <p>The most effective quality improvement strategies included clinician-directed audit and feedback, decision support systems and the use of small-group discussions in continuing professional education.</p>	2008	2/11 (AMSTAR rating from Program in Policy Decision-making)	Not reported	Not reported	Not reported
	Effectiveness and efficiency of interventions to adjust outpatient referral rates or to enhance outpatient referral appropriateness.(72)	Multifaceted interventions (n=6) consisted mainly of educational meetings, reminders, and distribution of educational materials, which were	2002	7/11 (AMSTAR rating from www.rxforchange.ca)	0/17	Not reported	0/17

Coordinating the Use of Genetic Tests and Related Services in British Columbia

Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
		<p>generally effective for improving appropriate care outcomes.</p> <p>Local consensus processes (n=2) and educational outreach visits (n=2) reported mixed effects, while distribution of educational materials (n=2) was found to be generally ineffective.</p> <p>There was insufficient evidence regarding the effectiveness of audit and feedback, reminders and educational meetings for appropriate care.</p>					
	Effectiveness of providing general practitioner with costing information to change their clinical behaviour to reduce medical costs.(99)	A decrease in inappropriate prescribing of target drugs occurred with implementation of academic detailing and by providing physicians with costing information. An increase in generic prescribing occurred when there was computerized feedback on drug costs.	1996	3/10 (AMSTAR rating from Program in Policy Decision-making)	0/6	Not reported	0/6
	Effects of educational outreach visits on health professional practice or patient outcomes.(71)	<p>Educational outreach visits have small but consistent effects on prescribing. Eight trials compared educational outreach visit interventions with audit and feedback, and educational outreach visits were slightly superior.</p> <p>The effect of educational outreach visits on other types of professional performance varies greatly, and this review is unable to explain the variation.</p>	2007	8/11 (AMSTAR rating from www.rxfchange.ca)	3/69	Not reported	0/69
	Effectiveness of computer reminders delivered to clinicians during their routine activities.(74)	Computer reminders improved physician adherence to processes of care by a median of 4.2%. The median improvement was found to be 5.6%.	2008	6/11 (AMSTAR rating from Program in Policy Decision-making)	2/28	Not reported	0/28

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Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
		Larger improvements were seen for reminders that required physicians to enter a response.					
	Effectiveness of on-screen computer reminders delivered to clinicians at the point of care.(73)	Computer reminders lead to a 4.2% median improvement in process adherence for all outcomes, 3.3% for medication ordering, 3.8% for vaccinations and 3.8% for test ordering. Generally, point-of-care computer reminders achieve small improvements in physician behaviour.	2008	9/11 (AMSTAR rating from Program in Policy Decision-making)	Not reported	Not reported	0/28
Implementing quality monitoring and improvement systems	Assessment of the relative effectiveness of various quality improvement strategies for enhancing healthcare.(80)	Research evidence suggests clinician/patient driven quality improvement strategies are more effective compared to manager/policymaker driven approaches. The most effective quality improvement strategies included clinician-directed audit and feedback, decision support systems and the use of small-group discussions in continuing professional education.	2008	2/11 (AMSTAR rating from Program in Policy Decision-making)	Not reported	Not reported	Not reported
	Assesses the effectiveness of quality improvement collaboratives in enhancing the quality of care.(79)	Systematic review of nine controlled trials found a positive effect of quality improvement collaboratives on processes of care and patient outcomes. Review additionally examined the findings of 60 uncontrolled reports of which 53 trials indicated specific improvements in patient care and organizational performance due to participation in a quality improvement collaborative.	2006	4/11 (AMSTAR rating from www.rxforchange.ca)	Not reported	Not reported	Not reported
	Examines evidence on promising	Review suggests for public reporting to	Not reported	2/9 (AMSTAR rating from Program	1/13	Not reported	Not reported

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Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
	practices for effective public reporting on healthcare quality.(86)	<p>be effective attention must be focused on the reporting program’s objectives, audience, content, product, distribution and impacts.</p> <p>Review also indicates public reporting should be part of broader efforts to develop and nurture a relationship with the report’s intended audience in order to increase accountability and quality within the healthcare system.</p>		in Policy Decision-making)			
Funding and remuneration mechanisms that support coordinated laboratory services within the province and appropriate use of genetic tests and related services	To identify all financial incentives that had been proposed, described, or used regardless of their initial objective and, when possible, to assess the results of these incentives on costs, process or outcomes of care.(81)	Financial incentives can be used to reduce the use of healthcare resources, improve compliance with practice guidelines or achieve a general health target. It may be effective to use incentives in combination depending on the target set for a given healthcare program.	1999	1/11 (AMSTAR rating from www.rxchange.ca)	Not reported in detail-Description states: Canada; U.K.; U.S.A.	Not reported	0/89
	Effectiveness of quality-based purchasing strategies, and analysis of existing strategies.(84)	<p>Incentives used in the nine included randomized controlled trials were additional fee-for-service, quality bonuses, and public release of performance data.</p> <p>The results were mixed: among the 11 performance indicators evaluated, seven showed a statistically significant response to QBP strategies while four did not.</p> <p>Determining incentives for quality-based purchasing through measurements of outcomes is a practical action without causing unnecessary risk to the reputation or financial status of good hospitals.</p>	2003	4/10 (AMSTAR rating from Program in Policy Decision-making)	Not reported	Not reported	0/5045

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Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
	Analysis of the effect of changes in the method and level of payment on the standard of care provided by primary care physicians.(75)	<p>Of the seven studies, six showed positive but small effects of financial incentive on quality of care, but one study found no effect on quality of care.</p> <p>The use of financial incentives to improve quality of care for primary physicians is not sufficiently supported or not supported by evidence.</p>	2009	10/10 (AMSTAR rating from Program in Policy Decision-making)	0/7	2/7	0/7

Appendix 3: Systematic reviews relevant to Element 3 – Develop a framework to support consumer/patient/family decision-making about genetic tests and related services

Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
Strategies to involve consumers/patients/families in decisions about genetic tests and related services to encourage them to express their beliefs, values and preferences about treatments and care, or to optimize communication between them and their healthcare providers (i.e., shared decision-making)	Evaluates the effects of consumer involvement and compares various methods of involvement in healthcare policy development and related research, clinical practice guidelines and information materials developed for patients.(67)	<p>The review notes the benefits of consumer involvement, in comparison to no consumer involvement, appear to be greatest in the creation of patient information materials</p> <p>Research evidence indicates involving consumers in the creation of information materials for patients produces material with improved readability and relevance, and that is more understandable to patients without elevating their anxiety.</p> <p>The impact of consumer involvement on healthcare policy creation and related research and the development of clinical practice guidelines remains uncertain due to lack of trials.</p>	2005	9/11 (AMSTAR rating from www.rxforchange.ca)	0/6	Not reported	0/6
	To determine the preferences of patients in their involvement in cancer treatment.(95)	<p>Preferences of patients' involvement in cancer treatment vary, with the majority of patients preferring a collaborative role in treatment decision-making and a significant minority preferring a passive or active role.</p> <p>Many patients experience a dissonance between their preferred role and the role they perceive they actually played.</p> <p>It is inconclusive whether factors such as age, gender, marital status, socioeconomic status and health status</p>	2004	5/10 (AMSTAR rating from Program in Policy Decision-making)	Not reported	31/31	0/31

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Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
		affect preferences.					
	Identification of ways to communicate evidence to improve patient understanding, involvement in decisions and outcomes.(94)	<p>There is limited evidence available to guide how physicians can most effectively share clinical evidence with patients facing decisions.</p> <p>Based on the limited evidence available as well as expert opinion, the review recommends five components for efforts to frame and communicate clinical evidence: understanding the patient's (and family members') experience and expectations; building partnership; providing evidence, including a balanced discussion of uncertainties; presenting recommendations informed by clinical judgment and patient preferences; and checking for understanding and agreement.</p>	2003	0/10 (AMSTAR rating from Program in Policy Decision-making)	Not reported	Not reported	0/8
	Effects of patient involvement in the planning and development of health care.(90)	<p>Most patients who participated in involvement initiatives had improved self-esteem. Health care staff also found patient involvement rewarding, though some reported difficult relationships between patients and staff.</p> <p>Changes to service associated with increased patient involvement included: production of new or improved information sources for patients; simplifying appointment procedures; improving transport to treatment units; and, improving access</p>	2002	5/9 (AMSTAR rating from Program in Policy Decision-making)	2/40	Not reported	Not reported

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Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
		<p>for people with disabilities.</p> <p>Organizational attitudes shifted to become more open to patient involvement after patient involvement initiatives were implemented.</p>					
Developing decision aids or decision support systems for consumer/patients about different screening or treatment options (pre and post-test)	Effectiveness of decision aids for patients' treatment or screening decisions.(92)	<p>Decision aids increase patient involvement, improve knowledge and realistic perception of outcomes.</p> <p>Patients exposed to decision aids with explicit values clarification versus those without explicit values clarification were better informed and achieved decisions more consistent with their values.</p> <p>Decision aids, compared to typical care interventions, resulted in lower decisional conflict related to feeling uncertain about personal values and feeling uninformed, and reduced the number of passive patients in decision-making and those left feeling undecided post-intervention.</p> <p>In the four studies that measured this outcome, decision aids positively affect patient-practitioner communication.</p>	2009	9/11 (AMSTAR rating from www.rxforchange.ca)	Not reported in detail-description states: Australia; Canada; China; Finland; Netherlands; UK; USA	Not reported	?/86 (focus of studies not reported)
	Effectiveness of cancer-related decision aids to improve communication between health professionals and patients and to involve patients in their health care decisions.(91)	<p>Knowledge of screening options was significantly improved with the use of decision aids compared to regular practice, and similarly knowledge of preventive/treatment options was also increased.</p> <p>In a screening context, based on 34</p>	2007	4/11 (AMSTAR rating from Program in Policy Decision-making)	Not reported	34/34	0/34

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Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
		<p>randomized controlled trials, decision aids enhanced patient knowledge about screening options without augmenting anxiety.</p> <p>There was little difference between the different decision aids.</p> <p>Review concluded that cancer-related decision aids are effective in increasing patient knowledge compared with usual practice without increasing anxiety, particularly in the area of cancer screening.</p>					
	<p>Overview of the impact on risk perception accuracy of genetic counselling.(88)</p>	<p>Overall, studies found that an increased proportion of individuals correctly perceived their risk after counselling rather than before, and those who did not had smaller deviations from their objective risk than before counselling.</p> <p>The positive effects were sustained at follow-up one year later.</p> <p>Some studies observed no impact at all, or only observed an impact for low-risk participants</p>	2007	5/9 (AMSTAR rating from Program in Policy Decision-making)	Not reported	Not reported	19/19
	<p>To evaluate the effects of attribute (positive versus negative) framing and of goal (gain versus loss) framing of the same health information, on understanding, perception of effectiveness, persuasiveness, and behavior of health professionals, policymakers, and consumers.(93)</p>	<p>Attribute framing in a positive manner caused more positive perceptions of effectiveness than negatively-framed messages, but did not cause a change in persuasiveness of the message.</p> <p>For screening messages, loss messages led to a more positive perception of</p>	2007	10/11 (AMSTAR rating from Program in Policy Decision-making)	Not reported in detail- Description states: U.S.A. (4); Italy (3); Netherlands	Not reported	Not reported

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Element	Focus of systematic review	Key findings	Year of last search	AMSTAR (quality) rating	Proportion of studies that were conducted in Canada	Proportion of studies that deal explicitly with one of the prioritized groups	Proportion of studies that focused on genetics
		effectiveness than gain messages.			(3); Canada (1); Norway (1)		
	To assess the effects of different types of personalized risk communication for consumers making decisions about taking screening tests.(87)	<p>There was little evidence to suggest that personalized risk communication (written, spoken or visually presented) increases uptake of screening tests, or promotes informed decision-making by consumers.</p> <p>In three studies, personalized risk communication interventions lead to a more accurate risk perception, and three other trials reported that interventions lead to increased knowledge.</p> <p>More detailed personalized risk communication (i.e., those which present numerical calculations of risk) may be associated with a smaller increase in uptake of tests.</p>	2006	10/11 (AMSTAR rating from Program in Policy Decision-making)	2/22	Not reported	4/22
	Effectiveness of interventions that provide patients with cancer risk and cancer screening information tailored to their personal attributes.(89)	<p>Tailored information regarding cancer risk and screening led to increased cancer risk perception and knowledge of breast cancer compared to generic information.</p> <p>There is limited evidence to suggest that a website tailored on risk factors would be effective.</p>	Not reported	7/11 (AMSTAR rating from Program in Policy Decision-making)	0/40	40/40	40/40