

**U.S. Department of Health and Human Services**

**Office of the National Coordinator for Health Information Technology**



**Personalized Healthcare Draft Detailed Use Case**

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## 1.0 Preface

Use cases developed for the American Health Information Community (AHIC) are based on the priorities expressed by the AHIC, which include needs expressed by the AHIC Workgroups. These high-level use cases focus on the needs of many individuals, organizations, and systems rather than the development of a specific software system. The use cases describe involved stakeholders, information flows, issues, and system needs that apply to the multiple participants in these arenas.

The use cases strive to provide enough detail and context for standards harmonization, certification considerations, architecture specifications and detailed policy discussions to advance the national health information technology (HIT) agenda. These high-level use cases focus, to a significant degree, on the exchange of information between organizations and systems rather than the internal activities of a particular organization or system.

During the January 2007 AHIC meeting, nine priority areas (representing over 200 identified AHIC and AHIC workgroup detailed issues and needs) were discussed and considered. Three of these areas (Consumer Access to Clinical Information, Medication Management, and Quality) were selected for use case development and the final 2007 Detailed Use Cases were published in June, 2007.

The remaining six priority areas from the January 2007 AHIC meeting (Remote Monitoring, Patient-Provider Secure Messaging, Personalized Healthcare, Consultations & Transfers of Care, Public Health Case Reporting, and Immunizations & Response Management) are now being developed into the 2008 Use Cases which will be processed in the national HIT agenda activities in 2008.

The 2008 Use Cases are being developed by the Office of the National Coordinator for Health Information Technology (ONC) with opportunities for review and feedback by interested stakeholders within both the private and public sectors. To facilitate this process, the use cases are being developed in two stages:

- The **Prototype Use Cases** describe the candidate workflows for the use case at a high level, and facilitate initial discussion with stakeholders; and
- The **Detailed Use Cases** document all of the events and actions within the use case at a detailed level.

This document is the Draft Detailed Use Case. Feedback received on the Prototype Use Case has been considered and incorporated where applicable into this document.



This Draft Detailed Use Case is divided into the following sections:

- Section 2.0, Introduction and Scope, describes the priority needs identified by one or more AHIC workgroups and includes draft decisions made regarding the scope of the use case.
- Section 3.0, Use Case Stakeholders, describes individuals and organizations that participate in activities related to the use case and its components.
- Section 4.0, Issues and Obstacles, describes issues or obstacles which may need to be resolved in order to achieve the capabilities described in the use case.
- Section 5.0, Use Case Perspectives, describes how the use case combines similar roles (or actors) to describe their common needs and activities. The roles are intended to describe functional roles rather than organizations or physical entities.
- Section 6.0, Use Case Scenarios, describes how various perspectives interact and exchange information within the context of a workflow. Use case scenarios provide a context for understanding information needs and are not meant to be prescriptive.
- Sections 7.0 and 8.0 provide a greater level of detail for each scenario and include information flows. Specific events and actions for each perspective and scenario are presented and discussed. These are also not intended to be prescriptive.
- Section 9.0, Information Exchange, describes the role of information exchange in the use case at a high level.
- Section 10.0, Dataset Considerations, identifies specific information opportunities relevant to this use case that may support future standardization and harmonization activities.
- Appendix A, the Glossary, provides draft descriptions of key concepts and terms contained in the draft detailed use case.

Following receipt of feedback from interested stakeholders, ONC will develop a final detailed use case.



## 2.0 Introduction and Scope

In January 2007, the AHIC approved a recommendation to develop a use case addressing personalized healthcare. Personalized healthcare describes processes where healthcare providers can customize treatment and management plans for patients based on their unique genetic makeup. AHIC established seven workgroups, one of which is the Personalized Healthcare (PHC) Workgroup. The PHC Workgroup was given the broad charge of making recommendations to the AHIC on personalized healthcare and has been one of the key drivers for the development of this use case. While the PHC Workgroup works to meet its broad charge, the personalized healthcare use case will focus on the exchange of genetic/genomic test information, personal and family health history, and the use of analytical tools in the electronic health record (EHR) to support clinical decision-making.

In specific terms:

- Consumers and clinicians will benefit from the inclusion of family and personal health history, combined with genetic/genomic testing results in EHRs. This will provide useful predictive information that can lead to earlier disease detection.
- Clinicians will benefit from capabilities that link large, medically related genetic datasets to individual-level genetic/genomic data.
- Clinicians will be better able to manage individual patients if access to education and information providing guidance on genetic/genomic test selection, risk analysis tools, and family health history information are made available to them.
- Genetic/genomic information can be helpful in health maintenance, prevention, and disease management, which may lead to a reduction in overall health care costs by encouraging early detection.

One of the goals of the AHIC is to establish a pathway, based on common data standards, to facilitate the incorporation of clinically useful, genetic, personal and family health history information, and analytical tools into EHRs to support clinical decision-making. Family health history takes time to gather from a disparate number of sources, increasing the need for interoperability. Ideally, family health history would be gathered concurrently as new events occur at the point of care rather than retrospectively by interview at different encounters. Similarly, accurately recording the data from genetic/genomic tests, as well as having a complete record of all genetic/genomic tests performed for a consumer, regardless of the ordering clinician, is important. Genetic/genomic information, unlike much other laboratory test information, has lifelong significance.

This use case assumes the developing presence and implementation of EHRs, interfaces between personal health records (PHRs) and EHRs, and health information exchange including those engaged in activities related to standards, interoperability, architecture,



policy development, and certification. While recognizing the issues and obstacles associated with these assumptions and to support these needs, the Personalized Healthcare Draft Detailed Use Case focuses on the exchange of family and personal health history and genetic/genomic testing information between stakeholders in two scenarios:

- **Clinical Assessment.** A family health history is gathered from or by the consumer in an interoperable form to be used by consumers and clinicians. This information is accessed by clinicians and used in conjunction with personal medical history, current health status, and personal preferences to develop a diagnostic plan.
- **Genetic Testing, Reporting, and Clinical Management.** A medical testing laboratory which performs genetic or genomic testing receives and captures genetic/genomic test orders and any accompanying information necessary for the testing. The testing laboratory performs the tests, develops the patient report, and transmits this information back to authorized providers. Clinicians utilize this new diagnostic information for management of their patients. Both clinicians and consumers have access to this information via the EHR or PHR.

The PHC Workgroup recognizes the uniqueness and complexity of newborn genetic screening - which shares characteristics with other genetic tests but has unique data flow and interoperability issues. While this topic is not considered to be in-scope for this use case, it is recognized to be of high importance. The Newborn Screening subgroup of the PHC Workgroup is currently working to advance this topic for future recommendations.

Combining genetic/genomic testing information with family and personal health history information in the EHR can facilitate improvements in medical decisions and increase patient participation in healthcare management.



### 3.0 Use Case Stakeholders

**Figure 3-1. Personalized Healthcare Use Case Stakeholders Table**

Stakeholder	Contextual Description
<b>Clinicians</b>	Healthcare providers with patient care responsibilities, including physicians, advanced practice nurses, pharmacists, physician assistants, nurses, medical geneticists, genetic counselors, and other credentialed personnel involved in treating patients.
<b>Clinical Genetic Databases</b>	Organizations that maintain resources, such as online servers and databases which provide detailed contextual knowledge specific to genetic diseases and the impact of genetic status on medical treatments. These databases may also provide references to the relevant medical literature.
<b>Consumers</b>	Members of the public that include patients as well as caregivers, patient advocates, surrogates, family members, and other parties who may be acting for, or in support of, a patient receiving or potentially receiving healthcare services.
<b>Electronic Health Record (EHR) Service Providers</b>	Organizations which assist in providing EHR capabilities to consumers. These capabilities may include providing services using information which is gathered/stored from a separate organization. Organizations that provide these capabilities may include: vendors, healthcare providers, health data banks, healthcare payors, etc.
<b>Genetic Specialists</b>	Medical geneticists, genetic counselors, and clinicians who participate in evaluation, diagnostic planning, and genetic/genomic test ordering and result interpretation activities.
<b>Genetic/Genomic Knowledge Repositories</b>	Organizations that maintain resources which provide raw genetic/genomic information. The information may include human genetic sequence data, structured nomenclature regarding specific genetic disease, or other similar data types.
<b>Health Information Management (HIM) Personnel</b>	Personnel who manage healthcare data and information resources, encompassing services in planning, collecting, aggregating, analyzing, and disseminating individual patient and aggregate clinical data.
<b>Health Researchers</b>	Organizations or individuals who use health information to conduct research.





Stakeholder	Contextual Description
<b>Healthcare Entities</b>	Organizations that are engaged in or support the delivery of healthcare. These organizations could include hospitals, ambulatory clinics, long-term care facilities, community-based healthcare organizations, employers/occupational health programs, school health programs, dental clinics, psychology clinics, care delivery organizations, pharmacies, home health agencies, hospice care providers, and other healthcare facilities.
<b>Healthcare Payors</b>	Insurers, including health plans, self-insured employer plans, and third party administrators, providing healthcare benefits to enrolled members and reimbursing provider organizations. As part of this role, they provide information on eligibility and coverage for individual consumers, as well as claims-based information on consumer health history. Case management or disease management may also be supported.
<b>Laboratory Organizations</b>	Advocacy/professional organizations or societies such as the College of American Pathologists (CAP) or the National Committee for Clinical Laboratory Standards (NCCLS) which are concerned with the appropriate use of laboratory technology and laboratory information in clinical medicine.
<b>Manufacturers/Distributors</b>	Entities which may be involved in the following activities: research, development, testing, production, storage, distribution, surveillance, and communication regarding medical/healthcare products at the community, regional, and national level, such as pharmaceutical manufacturers, drug wholesalers, medical device suppliers, etc.
<b>Patients</b>	Members of the public who receive healthcare services.
<b>Personal Health Record (PHR) Service Providers</b>	Organizations which assist in providing PHR capabilities to consumers. Organizations that provide these capabilities may include: vendors, healthcare providers, health data banks, healthcare payors, etc.
<b>Public Health Agencies/ Organizations (local/state/territorial/federal)</b>	Local, state, territorial, and federal government organizations and clinical care personnel that exist to help protect and improve the health of their respective constituents.
<b>Registries</b>	Organized systems for the collection, storage, retrieval, analysis, and dissemination of information to support health needs. This also includes government agencies and professional associations which define, develop, and support registries. These may include registries of phenotypic and genotypic information.



Stakeholder	Contextual Description
<b>System Vendors</b>	Organizations that develop and provide health information technology solutions. These solutions may include applications, data repositories, web services, etc., that contain or support the organization of genetic/genomic information.
<b>Testing Laboratories</b>	Medical testing laboratories, either within a hospital, ambulatory, or clinician office environment and/or operating as a free-standing entity, which analyze specimens as ordered by providers to assess the health status of patients. Specifically, testing laboratories which perform genetic/genomic laboratory tests ordered by genetic specialists and clinicians to assess the genetic status of patients.



## 4.0 Issues and Obstacles

Realizing the full benefits of HIT is dependent on overcoming a number of issues and obstacles in today's environment. Inherent is the premise that some of these issues and obstacles will be cross-cutting and therefore shown in all use cases, while others are unique to this specific use case.

Issues and Obstacles which are applicable across use cases appear below in problem and consequence form:

- **Confidentiality, privacy, and security:**
  - Consumers may require privacy controls for personal health information that govern how patient data is accessed, viewed, and communicated. Privacy controls and the means of restricting data access, which are available for paper-based records, are not currently available for electronic records.
    - Without permissions and controls, consumer participation in the act of electronic health information exchange may be limited.
  - There are regulations concerning the storage, transmission, or destruction of electronic health information. These regulations are inconsistent across federal, state, and local jurisdictions.
    - Without consistent standards, the viewing, accessing, or transmitting of electronic health information may be inhibited.
- **Information integrity, interoperability, and exchange:**
  - Incomplete, inaccurate, or proprietarily-formatted information prevents efficient exchange or utilization of electronic health information.
    - Without data standards that promote compatibility and interoperability, longitudinal patient medical records may be incomplete or of questionable integrity.
- **EHR and HIT adoption:**
  - The processes identified in the use cases rely upon successful integration of EHRs into clinical activities. Because this integration may not align with current workflow and may require additional upfront costs, it may not be widely pursued or implemented.



- Low adoption of HIT, particularly within rural areas and long-term care settings, may create disparate service levels and may adversely affect healthcare for these populations.
- **Lack of business model and infrastructure:**
  - Financial incentives are not currently sufficient to promote the business practices necessary for sustainable HIT.
    - If sufficient reimbursement policies and other financial incentives are not established, HIT adoption may be difficult or unsustainable.
  - Activities involving health information exchange will require additional technical infrastructure, functionality, and robustness, beyond what is currently available.
    - Unless the requisite infrastructure for health information exchange capabilities is established, improved upon, and sustained, these capabilities may have limited success and provide few benefits.
- **Clinical Decision Support:**
  - The capabilities, requirements, and standards needed for consistent development and implementation of Clinical Decision Support have not been identified.
    - The utility and benefits of Clinical Decision Support cannot be fully realized without the development of workflows and standards demonstrating benefits for consumers, patients, and providers.

Genetic/genomic information can provide information on disease status, predisposition to various diseases, the risk of passing on a disease to offspring, and potentially adverse or positive responses to therapeutic actions using pharmacogenetic guidance for oncology and other disciplines. Realizing the full benefits of personalized healthcare will be dependent on overcoming a number of issues and obstacles in today's environment. While some issues will be addressed through HIT standardization and harmonization activities, policy development, the process of health information exchange, and other related initiatives, others will have to be specifically addressed within the context of personalized healthcare and genetic/genomic information and are delineated below. In addition to the cross-cutting issues and obstacles described above, several other issues or obstacles exist that are specific to this use case.



- **Confidentiality, privacy, security and data access**

- Because EHRs are typically administered by healthcare providers, consumers may only have passive access to their medical record.
  - Without access to a personally controlled health record, the consumer may not be able to sufficiently restrict or grant access to sensitive information related to personalized healthcare.
- The implementation of personalized healthcare may create additional risk of misuse of family history, disease risk, and predisposition.
  - Consumers may lose privacy rights or face unfair consequences (e.g., health insurance denied or premiums raised) through improper disclosure of family history, disease risk, and predisposition information unless proper safeguards are put in place.
- There may be secondary uses of personalized healthcare information (for research or public health) which are not directly addressed by privacy agreements.
  - Secondary use of data may violate patient privacy and confidentiality.
- In some cases, non-standardized regulations may act as an obstacle to the exchange of genetic/genomic information, particularly across state boundaries.
  - Patients may not have access to information, thereby preventing appropriate care.

- **Family health history information interoperability and privacy**

- Family health history is typically obtained by interviewing the patient and/or other related individuals in an ad hoc and non-standardized manner. No industry guidelines exist to standardize this information or the manner in which it is gathered.
  - Without standards and consistent nomenclature, interoperable systems may be difficult to develop. These guidelines may facilitate information exchange between clinicians, between clinicians and patients, and between and among patients and authorized family members or other authorized consumers.



- Data included in a family health history are not always precise. Also, current terminologies do not always incorporate metrics reflecting the level of certainty at which this information can be obtained.
  - Without the proper structure and form, interoperable systems may be difficult to develop. A new document entitled “Family Health History Multi-Stakeholder Data Requirements Summary” may be advanced through the PHC Workgroup. This document is available for review on the Personalized Healthcare Use Case website located at <http://www.hhs.gov/healthit/usecases>.
- Several separate instances of family health history information may exist but be in conflict with one another since it is often gathered at disparate places and times, and by multiple individuals. Therefore, a process for the arbitration/reconciliation of this data may be needed.
  - Without a validation and reconciliation of this information, care of a particular family member or consumer may be compromised.
- Because of the sensitive nature of family health history information, several specific and unique issues related to privacy of patient information are of concern; particularly those related to the sharing of this information with authorized family members or other patient advocates. Currently, genetic information is treated as any other protected health information (PHI) under the Health Insurance Portability and Accountability Act (HIPAA).
  - Without adherence to and proper interpretation of these guidelines as they relate to genetic/genomic information, this information could be used in an inappropriate manner.
- **Genetic/Genomic data interoperability**
  - The exchange of genetic/genomic information across systems, sites and settings of care is constrained by a fragmented nomenclature for ordering and reporting of tests and testing results.
    - The fragmented and unstructured nature of this information may lead to difficulties in the act of information exchange and put at risk the quality of care.
  - Because of the recent and ongoing development of genetic technologies (for example – Polymerase Chain Reaction and DNA Microarrays), keeping pace with technological advances is a significant obstacle. There is also a lack of standards for information transferred to and from laboratory instrumentation



involved in genetic/genomic testing. Some of these data are reported outside the laboratory and must be standardized to ensure interoperability with EHRs and PHRs

- Because of the lack of standardization, the integrity of information being exchanged with EHRs and PHRs may become compromised.
- The format of most genetic test results is a combination of specific diagnostic information (which might be chromosome- or gene- specific nomenclature or genetic sequence data) and interpretive text which may be thought of as metadata related to a genetic test result. There are no agreed upon industry standards for this information.
  - Without an agreed-upon standard for this information, it may be difficult to transmit information accurately between systems.



## 5.0 Use Case Perspectives

The Personalized Healthcare Draft Detailed Use Case will describe personalized healthcare from the viewpoint associated with three perspectives. The perspectives included in the use case are intended to indicate roles and functions, rather than organizations or physical locations. Each perspective is described below:

- **Clinician**

The clinician perspective includes family physicians, pediatricians, obstetricians, oncologists, internists, advanced practice nurses, physician assistants, genetic counselors, medical geneticists, and other personnel that conduct clinical assessment and management activities and participate in evaluation, diagnostic planning, genetic/genomic test ordering, and result interpretation activities. The clinician may also be working from within the testing laboratory.

- **Testing Laboratory**

The testing laboratory perspective includes medical laboratory personnel such as the lab director, lab supervisor, lab technicians, or other relevant staff who perform genetic laboratory tests ordered by clinicians to assess the genetic status of patients, generate data, and report results.

- **Consumer**

The consumer perspective includes members of the public who receive healthcare services, as well as caregivers, patient advocates or surrogates, family members, and other parties who may be acting for, or in support of, a patient. The consumer self-reports family health history information, requests and views available family health history and genetic/genomic testing information, and considers personalized prevention messages and/or treatment information.

These perspectives are the focus of the events detailed in the scenarios described in Section 6.0.





## 6.0 Use Case Scenarios

The Personalized Healthcare Draft Detailed Use Case focuses on the exchange of personal health, family health history, and genetic/genomic testing information between consumers and clinicians in two scenarios.

- **Clinical Assessment**

This scenario is focused on gathering past medical history, current medical status, and family health history information from or by the consumer in an interoperable form to be used by consumers and clinicians. This information is accessed by clinicians and used in conjunction with personal medical history, current health status, and personal preferences to develop a diagnostic plan.

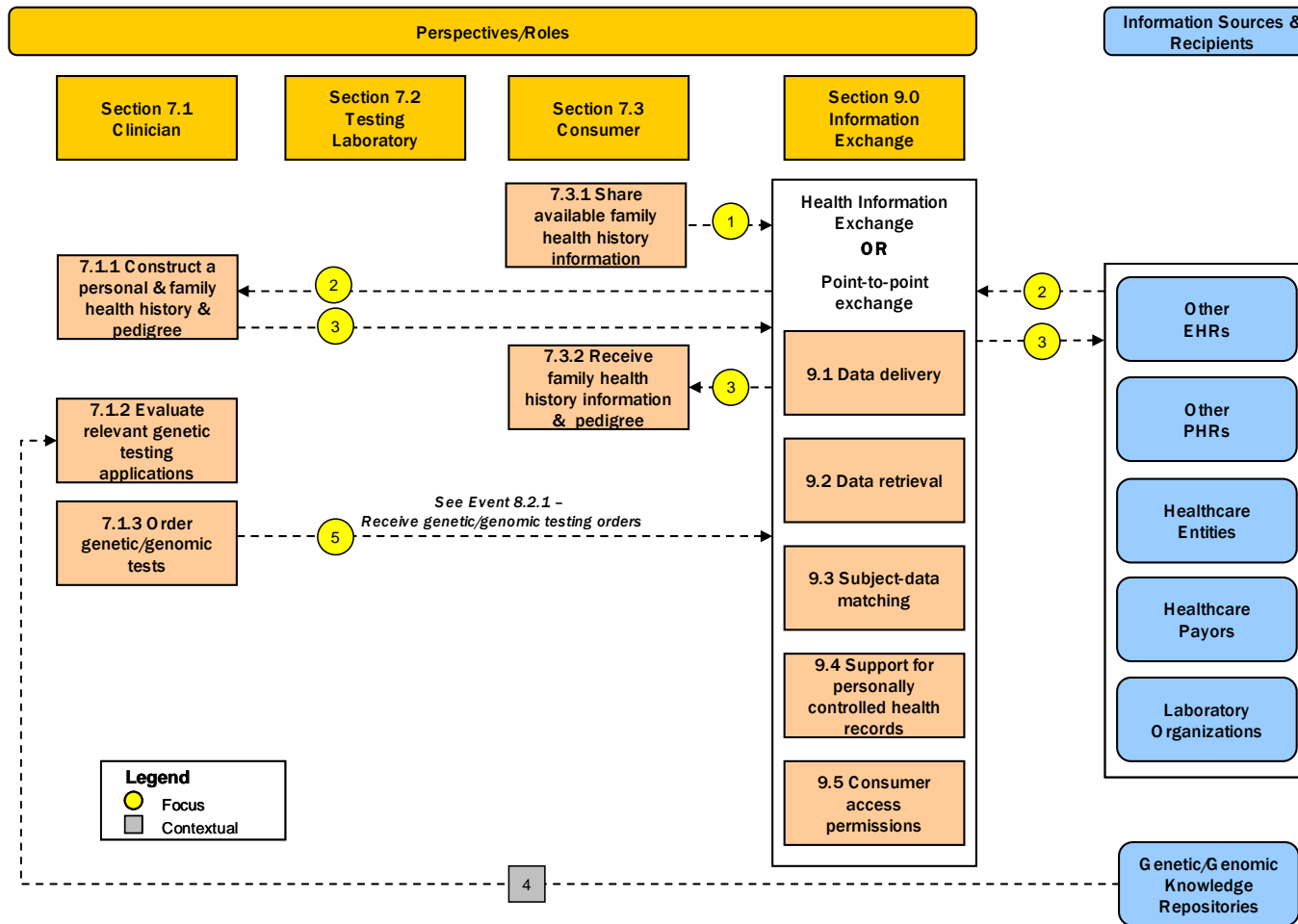
- **Genetic Testing, Reporting, and Clinical Management**

This scenario highlights both the genetic/genomic testing and reporting functions as well as the clinical management that follows the receipt of information from the testing. Part of this scenario is focused on a testing laboratory receiving and capturing genetic/genomic test orders and any accompanying information necessary for the testing, as well as the ability to exchange genetic/genomic laboratory test results among laboratories and ordering clinicians with appropriate privacy and security considerations. A medical testing laboratory which performs genetic/genomic testing receives and captures test orders and any accompanying information necessary for the testing. The testing laboratory performs the tests, develops the patient report, and transmits this information back to authorized providers. The other part of this scenario focuses on determining appropriate preventative action, treatment protocol, messaging, and interpretation of results and analysis utilizing decision support tools, and genetic/genomic knowledge repositories, as well as the consumer's ability to permit designated individuals to request and view information in their PHR.



## 7.0 Scenario 1: Clinical Assessment

Figure 7-1. Clinical Assessment





**Figure 7-2. Clinical Assessment Scenario Flows**

- ① Consumer shares available family medical history information.
- ② Validated consumer and family health information, any genetic/genomic testing information, and additional information from past consumer encounters or family members is accessed and gathered electronically via health information exchange.
- ③ Patient receives newly constructed pedigree and family health history.
- ④ Information from genetic/genomic knowledge repositories support the selection of genetic tests.
- ⑤ The Clinician orders genetic/genomic tests for the patient.



**Figure 7-3. Clinical Assessment, Clinician Perspective**

Code	Description	Comments
7.1.1	<b>Event:</b> Construct a personal & family health history & pedigree	
7.1.1.1	<b>Action:</b> Request and gather available personal and family health history information in interoperable electronic form.	The clinician gathers current patient personal health history, family health history and any past genetic/genomic testing information from several external sources to support the patient assessment. This may be in the context of testing for familial genetic disease, prenatal genetic testing, pharmacogenetic guidance for oncology treatment or any other personalized use of genetic or genomic testing. Consumer self-reported personal and family health history information may be available from the patients PHR. Additional available information could be gathered electronically via information exchange, from hospital EHRs, ambulatory EHRs (such as from a Primary Care Physician (PCP)), and/or other sources (such as healthcare payors, other providers, or family members) that hold information about the patient. Ideally, this information could be provided in an integrated view without duplications. In each case, the information source (e.g., authoritative clinical source, administrative source, or patient) could also be captured.
7.1.1.1a	<b>Alternative Action:</b> Request and gather available personal and family health history information in viewable electronic form.	The clinician views summary personal and family health history information from external sources.
7.1.1.1b	<b>Alternative Action:</b> Gather personal and family health history information via interview.	The clinician and support staff gather personal and family health history information by interviewing the patient, patient's family, significant others, and/or caregivers – and in some instances, by contacting the patient's PCP.



Code	Description	Comments
7.1.1.2	<b>Action:</b> View consolidated available personal and family health history information.	After information is gathered from multiple sources, the clinician views the information in a consolidated format to gain the most comprehensive view of the patient's personal and family health history information. Clinicians require the ability to view personal and family health history information throughout the encounter.
7.1.1.3	<b>Action:</b> Select personal and family health history information.	After viewing the patient's personal and family health history information, the clinician makes determinations regarding which information will be incorporated into the EHR. Factors taken into account include duplication, currency, relevance to current clinical context, and data source.
7.1.1.4	<b>Action:</b> Incorporate personal and family health history information.	The clinician executes the necessary steps to store the patient's personal and family health history information in the patient's EHR. This compiled list of verified, current information will be available for viewing throughout the encounter. In addition this information will be reviewed and communicated to other clinicians, such as the next provider of care (if applicable), and the consumer and his/her authorized family members or proxies at the end of the encounter.
7.1.2	<b>Event:</b> Evaluate relevant genetic testing applications	
7.1.2.1	<b>Action:</b> Receive information from genetic/genomic knowledge repositories and/or decision support modules within EHRs.	Genetic/genomic knowledge repositories and other sources provide data and reference information to support the selection of genetic tests, incidence figures and other decision support capabilities. These act in conjunction with, and may be integrated into, the clinician's local EHR using clinical genetic databases such as Online Medelian Inheritance in Man (OMIM), ALFRED, or other integrated or standalone decision support systems. These data interactions are typically contextual and <u>ad hoc</u> communications. However, the process of setting up decision support relies on standard vocabularies for family relationship nomenclature and other data points related to family health history.



Code	Description	Comments
7.1.2.2	<b>Action:</b> Perform interpretation, assembly, validation, and evaluation activities.	The clinician performs interpretation, assembly, validation, and evaluation activities with support from decision support tools. The clinician analyzes the relevant medical and health information for genetic and/or birth defect risks and assesses and interprets the risk for occurrence of genetic conditions. The clinician may access decision support tools that utilize data tables, risk assessment algorithms, and/or other information from the genetic/genomic knowledge repositories.
7.1.3	<b>Event:</b> Order genetic/genomic tests	
7.1.3.1	<b>Action:</b> Write genetic/genomic test order.	The clinician may order genetic/genomic tests for the patient through a web application or via an available EHR. The clinician may utilize decision support tools and/or conduct consultations with a molecular pathologist to determine which tests to order.
7.1.3.2	<b>Action:</b> Communicate genetic/genomic test order to the medical laboratory performing the genetic/genomic testing.	The genetic/genomic test order is communicated to the medical laboratory that is performing the genetic/genomic testing. The order may also include accompanying reference information such as patient and family information to be considered in analysis and interpretation of the results, general specimen information, billing information, and physician and patient contact information.



**Figure 7-4. Clinical Assessment, Consumer Perspective**

Code	Description	Comments
<b>7.3.1</b>	<b>Event:</b> Share available family health history information	
<b>7.3.1.1</b>	<b>Action:</b> Patient may self-report personal and family health history information.	The consumer could use a PHR or direct or web access to the clinician's EHR to record personal and family health history information. This information could be available to a clinician via retrieval from the consumer's PHR or made available to the clinician based on the consumer's preferences.
<b>7.3.1.1a</b>	<b>Alternative Action:</b> Patient uses an interoperable PHR to share his/her medical and family history with the clinician.	The consumer, in this case, is equipped with an interoperable PHR which includes his/her personal health and family history. The information can be quickly sent to the appropriate clinician to aid in diagnosis, analysis and treatment.
<b>7.3.1.1b</b>	<b>Alternative Action:</b> Patient self-reports personal medical and family history through an electronic portal.	The consumer would be presented with an electronic form which would provide the appropriate data back to the clinician's EHR. This information could potentially be transformed within the system to a standardized interoperable form.
<b>7.3.1.1c</b>	<b>Alternative Action:</b> Patient reports personal medical and family history by interview.	The consumer would be interviewed for all appropriate information during the course of a consultation with a clinician.
<b>7.3.2</b>	<b>Event:</b> Receive family health history information & pedigree	
<b>7.3.2.1</b>	<b>Action:</b> Patient receives newly validated and updated personal and family health history information and pedigree, if appropriate, from clinician.	The consumer could receive newly validated and updated personal and family health history information and pedigree using a PHR or direct or web access to the clinician's EHR. This information could be available to a consumer via retrieval from the clinician's EHR or provided automatically by the clinician based on consumer's preferences.



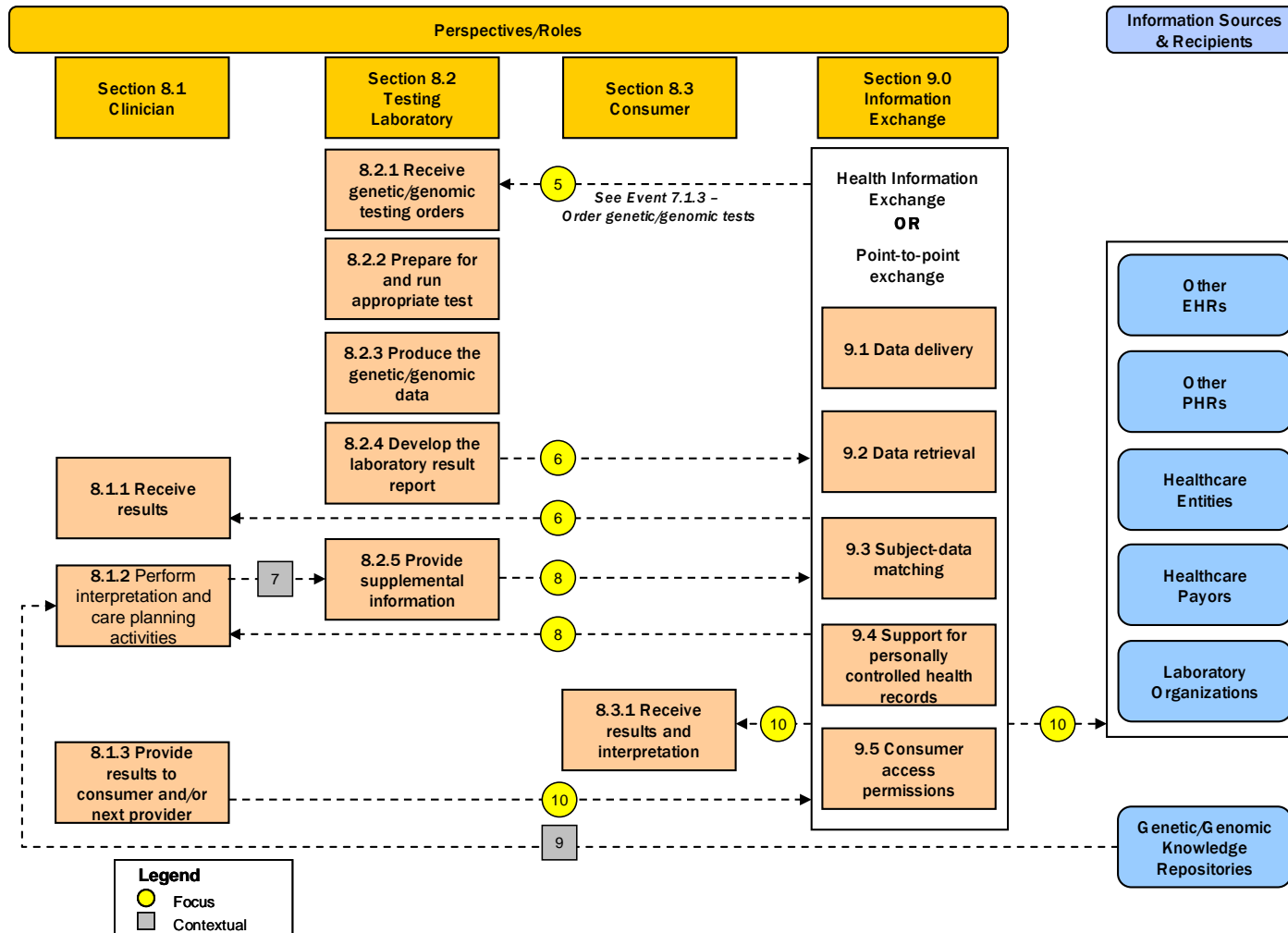
Code	Description	Comments
7.3.2.1a	<p><b>Alternative Action:</b> Patient receives newly validated and updated personal and family health history information and pedigree, if appropriate, via an interoperable PHR.</p>	<p>The consumer, in this case, is equipped with an interoperable PHR which carries his/her newly validated and updated personal and family health history information and pedigree from the clinician's EHR. The information can be received from the appropriate clinician to aid in diagnosis, analysis and treatment plan.</p>
7.3.2.1b	<p><b>Alternative Action:</b> Clinician reports newly validated and updated personal and family health history information and pedigree, if appropriate, via patient consultation.</p>	<p>The clinician would communicate all appropriate newly validated and updated personal and family health history information and pedigree information during the course of a consultation with the consumer.</p>





## 8.0 Scenario 2: Genetic Testing, Reporting, and Clinical Management

Figure 8-1. Genetic Testing, Reporting, and Clinical Management





**Figure 8-2. Genetic Testing, Reporting, and Clinical Management Scenario Flows**

- 5 The testing laboratory receives and captures the genetic/genomic testing orders and accompanying information.
- 6 The testing laboratory transmits results to the ordering clinician.
- 7 The clinician may request additional information i.e. – sequence information from the testing laboratory via their EHR. This may have been previously 'pushed' to the clinician.
- 8 Clinician receives additional information back from the testing laboratory.
- 9 Clinician utilizes external ad hoc data from repositories for interpretation support.
- 10 Clinician sends result report to consumer(s) and/or next provider of care.



**Figure 8-3. Genetic Testing, Reporting, and Clinical Management, Clinician Perspective**

Code	Description	Comments
<b>8.1.1</b>	<b>Event:</b> Receive results	
<b>8.1.1.1</b>	<b>Action:</b> The ordering clinician receives results from the testing laboratory.	The clinician receives the genetic/genomic test results via an EHR or other clinical data system. All identified genetic variants, a full description of what was tested (possibly to include sequence level information), and the interpretation could be included in the structured representation.
<b>8.1.2</b>	<b>Event:</b> Perform interpretation and care planning activities	
<b>8.1.2.1</b>	<b>Action:</b> Perform interpretation and care planning activities.	The ordering clinician performs interpretation, care planning, and care plan implementation activities utilizing several forms of decision support including, but not limited to, external data repositories and integrated decision support information and algorithms built into the clinician's own EHR. Information may also be retrieved from external data repositories such as OMIM. The ordering clinician may also consult with molecular pathologists during the process of analyzing and interpreting the genetic/genomic test results. This particular information exchange may present an opportunity for standardization in the future.
<b>8.1.2.2</b>	<b>Action:</b> Request and view additional information from the testing laboratory.	The clinician may also request additional information from the testing laboratory, such as genomic sequence information, which could be communicated to the clinician in a structured, standardized format. Standardization needs exist for clinicians and laboratories working with communication of genetic/genomic testing information. These needs are focused on establishing more complete and robust data standards using a healthcare terminology, along with a standardized format, both of which are needed to help achieve the interoperable integration of genetic/genomic test information into EHRs.



Code	Description	Comments
<b>8.1.3</b>	<b>Event:</b> Provide results to the consumer and/or the next provider of care	
<b>8.1.3.1</b>	<b>Action:</b> Communicate results and additional interpretation from the testing laboratory to the next provider of care.	The ordering clinician provides results and additional interpretation from the testing laboratory to other clinicians, such as the next provider of care. This information can be incorporated into clinician's EHRs. The information communicated may also include personal and family health information captured and organized by the ordering clinician such as a family pedigree.
<b>8.1.3.2</b>	<b>Action:</b> Communicate results and additional interpretation from the testing laboratory to the patient and other authorized family members.	The ordering clinician provides results and additional interpretation from the testing laboratory to the consumer(s). The information communicated may also include personal and family health information captured by the ordering clinician. The information could be hand-written or printed out from the clinician's EHR. It could also be communicated in a standardized interoperable form for inclusion into the patient's PHR. The patient may also be provided with a care plan.

**Figure 8-4. Genetic Testing, Reporting, and Clinical Management, Testing Laboratory Perspective**

Code	Description	Comments
<b>8.2.1</b>	<b>Event:</b> Receive genetic/genomic testing orders	
<b>8.2.1.1</b>	<b>Action:</b> Receive and capture the genetic/genomic testing orders.	The testing laboratory receives and captures the genetic/genomic testing orders and any accompanying information necessary for the testing from a structured, standards-based electronic message.
<b>8.2.2</b>	<b>Event:</b> Prepare for and run appropriate test	



Code	Description	Comments
8.2.2.1	<b>Action:</b> Prepare for and run the appropriate test based on the genetic/genomic testing orders received.	The laboratorial staff prepares for the genetic/genomic test based on the testing orders received. A level of analysis is necessary by the laboratorial staff to ensure that all the accurate and correct information to run the specialized tests has been received and properly set up within the Laboratory Information System (LIS). It is possible, based on the capabilities of the LIS, that some of this analysis may be completed within the LIS automatically.
8.2.2.1a	<b>Alternative Action:</b> Communicate with the ordering clinician to get clarification.	Because of the specialized nature of genetic testing and the evolving technologies, situations may arise in which the testing laboratory will need to communicate back to the ordering clinician to ensure that the correct testing has been ordered and all the necessary information has been gathered to enable testing to take place. The clinician responds to communication from the testing laboratory. This information exchange is likely to be of an <u>ad hoc</u> nature.
8.2.2.2	<b>Action:</b> Make revisions to orders, as necessary.	Based on the review, the testing laboratory may revise the order after consulting with and informing the clinician.
8.2.2.3	<b>Action:</b> Return information on order status or any order changes.	Information on order status and any order modifications could be incorporated into the EHR for access by clinicians involved in the patient's care.
8.2.3	<b>Event:</b> Produce the genetic/genomic data	
8.2.3.1	<b>Action:</b> Perform the steps required to produce the data.	The testing laboratory performs the technical steps required to produce the genetic/genomic data.
8.2.4	<b>Event:</b> Develop the laboratory result report	



Code	Description	Comments
8.2.4.1	Action: Develop the laboratory result report.	<p>The testing laboratory develops the laboratory report and transmits it in both narrative and structured form in a standards-based electronic message to the ordering clinician's EHR or other clinical data system. The patient report may also be transmitted to other authorized healthcare providers based on the consumer's preferences and/or needs.</p> <p>All identified genetic variants, a full description of what was tested (possibly to include sequence level information), and the interpretation could be included in the structured representation. Genetic/genomic knowledge repositories may be utilized by the testing laboratory to support this activity.</p>
8.2.5	<b>Event:</b> Provide supplemental information	
8.2.5.1	<b>Action:</b> Provide supplemental information to the ordering clinician.	<p>The testing laboratory may receive requests from the ordering clinician for additional information, such as sequence information or raw instrument data, which could be communicated to the clinician in a structured, standardized format. Standardization needs exist for clinicians and laboratories working with communication of genetic/genomic testing information. These needs are focused on establishing more complete and robust data standards using a healthcare terminology, along with a standardized format, both of which are needed to help achieve the interoperable integration of genetic/genomic test information into EHRs.</p>

**Figure 8-5. Genetic Testing, Reporting, and Clinical Management, Consumer Perspective**

Code	Description	Comments
8.3.1	<b>Event:</b> Receive results and interpretation	



Code	Description	Comments
<p><b>8.3.1.1</b></p>	<p><b>Action:</b> Consumer receives laboratory results and clinical interpretation.</p>	<p>The consumer receives available personal, family health, and/or genetic/genomic testing information via their PHR. This information may have been self-reported earlier, or may be derived from their clinicians' EHR systems, other authoritative clinical sources and/or administrative data sources. Information in a standardized interoperable form would move through an information exchange and could be sent to other providers of care. Consumers would additionally benefit from the ability to permit designated clinicians and other individuals (e.g., family members) to request and view information in their PHR (a.k.a., proxy access). The consumer's authorized family members would also benefit from the ability to view, select, and/or incorporate information into their own PHRs, in accordance with the consumer's personal preferences.</p>



## 9.0 Information Exchange

This section highlights selected information exchange capabilities which enable the scenarios described in this use case. These functional capabilities may be provided fully or partially by a variety of organizations including free-standing or geographic health information exchanges (e.g., RHIOs), integrated care delivery networks, provider organizations, health record banks, public health networks, specialty networks, and others supporting these capabilities.

**Figure 9-1. Personalized Healthcare Information Exchange Capabilities**

Code	Capability	Comments
9.1	Data delivery – including secure data delivery, confirmation of delivery to EHRs, personally controlled health records, other systems and networks	Capability to securely deliver data to the intended recipient, confirm delivery, including the ability to route data based on message content if required. For example, routing may be applicable to identify the destination testing laboratory which is to receive the genetic/genomic testing orders.
9.2	Data retrieval – including data lookup, retrieval and data location registries	Capability to locate and retrieve requested data subject to consumer access decisions and local policies. For example, retrieving the consumer's family health history information involves determining the availability of the requested information as well as delivery to the requestor
9.3	Subject-data matching	Capability to match available data to the appropriate person during retrieval or routing. For example, when a clinician makes a request for family health history information for a specific person, the systems involved in the information exchange confirm that the data available for retrieval match the person of interest to the clinician.
9.4	Support for personally controlled health records – including managing consumer-identified locations to store their personally controlled health information; support consumer requests for information as well as routing of information to the consumer's preferred personally controlled health record	Capability to maintain and implement information identifying the consumers preferred personally controlled health record (e.g., a PHR or health record bank) to support data routing and retrieval. For example, the consumer may identify a specific personally controlled health record which holds their available family health history information.





Code	Capability	Comments
9.5	Consumer-controlled access decisions – including managing consumer-controlled providers of care and access permissions information; including consumer choice to not participate in network activities	Capability to manage and implement consumer-controlled access decisions during information exchange activities. For example, if a consumer chooses to not make their health information available through network activities, those systems involved in exchanging health information would need to maintain awareness of this decision and honor it when requests for the consumer’s data are processed.

While not described in this section, other capabilities support information exchange including: data integrity and non-repudiation checking; subject and user identity arbitration with like identities during information exchanges; access logging and error handling for data access and exchange; consumer review of disclosure and access logs; and routing consumer requests to correct data.

**Health Information Exchange (HIE):** For the purpose of this use case, the functional capability to exchange health information between networks in order to exchange the health information of individuals or populations. These functional capabilities may be provided fully or partially by a variety of organizations including free-standing or geographic health information exchanges (e.g., RHIOs), integrated care delivery networks, provider organizations, health record banks, public health networks, specialty networks, and others supporting these capabilities.

**Point-to-Point Exchange:** A direct link or communication connection with defined endpoints.



## 10.0 Dataset Considerations

Currently, there are no data set and/or standards for the transfer of appropriate and necessary information to facilitate personalized healthcare delivery. As personalized healthcare is further defined, the dataset will become central to the adoption and success of personalized healthcare. At the request of AHIC, the PHC Workgroup has been working on the development of a dataset specific to personalized healthcare. The dataset that is most specific to personalized healthcare falls into two broad categories: 1) family health history information and 2) genetic/genomic test result information. The first is critical in assessing patient risk and determining which genetic/genomic information must be gathered or generated and the second is specific information identifying the genetic/genomic status of patients for the purposes of diagnosis or treatment of various disease states, including familial forms of various genetic diseases, as well as guiding treatment of cancer and other illnesses by using pharmacogenetic information.

The core dataset is still being developed under the direction of the PHC Workgroup. For the purposes of addressing the scenarios in this use case, the following non-exhaustive information categories with limited examples may be considered:

- Demographic information
  - Name
  - Numerical identifier
  - Race
- Personal health information
  - History of specific disorders
  - Relevant non-genetic laboratory test data
  - Any prior treatment for specific disorders
- Family history information
  - Disorders of family members
  - Ages of death of various family members
  - Relevant social data



- Personal genetic/genomic data
  - Prior genetic/genomic lab results
  - Prior genetic status for specific disease
  - Full genome scan: DNA
- Family genetic/genomic information
  - Genetic/genomic data of family members
  - Pedigree in structured form when appropriate
  - History of consanguinity

The document entitled “Family Health History Multi-Stakeholder Workgroup Data Requirements Summary” is available for review and public feedback at the Personalized Healthcare Use Case website located at <http://www.hhs.gov/healthit/usecases>.



## Appendix A: Glossary

These items are included to clarify the intent of this use case. They should not be interpreted as approved terms or definitions but considered as contextual descriptions.

**AHIC:** American Health Information Community; a federal advisory body chartered in 2005, serving to make recommendations to the Secretary of the U.S. Department of Health and Human Services regarding the development and adoption of health information technology.

**Care:** Relieving the suffering of individuals, families, communities, and populations by providing, protecting, promoting, and advocating the optimization of health and abilities.

**Clinical Genetic Databases:** Organizations that maintain resources, such as online servers and databases which provide detailed contextual knowledge specific to genetic diseases and the impact of genetic status on medical treatments. These databases may also provide references to the relevant medical literature.

**Clinicians:** Healthcare providers with patient care responsibilities, including physicians, advanced practice nurses, pharmacists, physician assistants, nurses, medical geneticists, genetic counselors, and other credentialed personnel involved in treating patients.

**CMS:** Centers for Medicare & Medicaid Services; a federal agency within the Department of Health and Human Services that administers Medicare, Medicaid and the State Children's Health Insurance Program.

**Consanguinity:** Relationship by blood or by a common ancestor that may influence disorders of patient, family members, and/or relatives.

**Consumers:** Members of the public that include patients as well as caregivers, patient advocates, surrogates, family members, and other parties who may be acting for, or in support of, a patient receiving or potentially receiving healthcare services.

**Decision Support:** A discipline that enables improved analysis and conclusions based on related information, recent research, algorithms, or other resources. In a clinical environment, decision support can help clinicians make more informed care decisions based on these resources. Clinical decision support is a related discipline with specific components such as best practice guidelines, medication contraindication information, and access to recent research.

**Department of Health and Human Services (HHS):** Department of Health and Human Services; the United States federal agency responsible for protecting the health of the nation and providing essential human services with the assistance of its operating divisions that include: Administration for Children and Families (ACF), Administration on Aging (AOA),



Agency for Healthcare Research and Quality (AHRQ), Agency for Toxic Substances and Disease Registry (ATSDR), Centers for Disease Control and Prevention (CDC), Centers for Medicare & Medicaid Services (CMS), Food and Drug Administration (FDA), Health Resources and Services Administration (HRSA), Indian Health Services (IHS), National Institutes of Health (NIH), Program Support Center (PSC), and Substance Abuse and Mental Health Services Administration (SAMHSA).

**Electronic Health Record (EHR):** The electronic health record is a longitudinal electronic record of patient health information generated in one or more encounters in any care delivery setting. This information may include patient demographics, progress notes, problems, medications, vital signs, past medical history, immunizations, laboratory information, and radiology reports.

**Electronic Health Record (EHR) Service Providers:** Organizations which assist in providing EHR capabilities to consumers. These capabilities may include providing services using information which is gathered/stored from a separate organization. Organizations that provide these capabilities may include: vendors, healthcare providers, health data banks, healthcare payors, etc.

**Family Health History:** Documentation of the healthcare problems suffered by blood relatives for the purposes of assessing an individual's risk for these disorders.

**Genetic Counselors:** Professionals engaged in the educational counseling process for individuals and families who have a genetic disease or who are at risk for such a disease.

**Genetic Specialists:** Medical geneticists, genetic counselors, and clinicians who participate in evaluation, diagnostic planning, and genetic/genomic test ordering and result interpretation activities.

**Genetic/Genomic Knowledge Repositories:** Organizations that maintain resources which provide raw genetic/genomic information. The information may include human genetic sequence data, structured nomenclature regarding specific genetic disease, or other similar data types.

**Genetic/Genomic Test:** A specific laboratory test intended to provide data regarding the genetic/genomic status of an individual. This test can be at the molecular, chromosomal or sequence level.

### **Geographic Health Information Exchange/Regional Health Information**

**Organizations:** A multi-stakeholder entity, which may be a free-standing organization i.e. – hospitals, healthcare systems, partnership organizations, etc. that supports health information exchange which enable the movement of health-related data within state, local, territorial, or jurisdictional participant groups. Activities supporting health information exchanges may also be provided by entities which are separate from geographic health



information exchanges/Regional Health Information Organizations and may include: integrated delivery networks, health data banks, etc.

**Health Information Exchange (HIE):** The functional capability to exchange health information between networks in order to exchange the health information of individuals or populations. These functional capabilities may be provided fully or partially by a variety of organizations including free-standing or geographic health information exchanges (e.g., RHIOs), integrated care delivery networks, provider organizations, health record banks, public health networks, specialty networks, and others supporting these capabilities.

**Health Information Management (HIM) Personnel:** Personnel who manage healthcare data and information resources, encompassing services in planning, collecting, aggregating, analyzing, and disseminating individual patient and aggregate clinical data.

**Health Researchers:** Organizations or individuals who use health information to conduct research.

**Healthcare Entities:** Organizations that are engaged in or support the delivery of healthcare. These organizations could include hospitals, ambulatory clinics, long-term care facilities, community-based healthcare organizations, employers/occupational health programs, school health programs, dental clinics, psychology clinics, care delivery organizations, pharmacies, home health agencies, hospice care providers, and other healthcare facilities.

**Healthcare Payors:** Insurers, including health plans, self-insured employer plans, and third party administrators, providing healthcare benefits to enrolled members and reimbursing provider organizations. As part of this role, they provide information on eligibility and coverage for individual consumers, as well as claims-based information on consumer health history. Case management or disease management may also be supported.

**HITSP:** The American National Standards Institute (ANSI) Healthcare Information Technology Standards Panel; a body created in 2005 in an effort to promote interoperability and harmonization of healthcare information technology through standards that would serve as a cooperative partnership between the public and private sectors.

**Laboratory Information System (LIS):** A laboratory information system is a class of **software** which handles receiving, processing and storing information generated by **medical laboratory** processes. These systems often must interface with **instruments** and other information systems such as **hospital information systems**. An LIS is a highly configurable application which is customized to facilitate a wide variety of laboratory workflow models.



**Laboratory Organizations:** Advocacy/professional organizations or societies such as the College of American Pathologists (CAP) or the National Committee for Clinical Laboratory Standards (NCCLS) which are concerned with the appropriate use of laboratory technology and laboratory information in clinical medicine.

**Manufacturers/Distributors:** Entities which may be involved in the following activities: research, development, testing, production, storage, distribution, surveillance, and communication regarding medical/healthcare products at the community, regional, and national level, such as pharmaceutical manufacturers, drug wholesalers, medical device suppliers, etc.

**Medical Geneticist:** A physician, trained and board certified in the subspecialty of Medical Genetics.

**ONC:** Office of the National Coordinator for Health Information Technology; serves as the Secretary's principal advisor on the development, application, and use of health information technology in an effort to improve the quality, safety and efficiency of the nations health through the development of an interoperable harmonized health information infrastructure.

**Patients:** Members of the public who receive healthcare services.

**Pedigree:** A pedigree is a graphic, visual presentation of a family's health history and genetic relationships for the purpose of health risk assessment. It provides, at a glance, the distribution of a medical condition in a group of close relatives. If the condition clusters among relatives or follows a clear pattern of inheritance, then the risk for the condition can be assessed for the unaffected family members.

**Personal Health History:** The medical history of an individual.

**Personal Health Record (PHR):** A health record that can be created, reviewed, annotated, and maintained by the patient or the caregiver for a patient. The personal health record may include any aspect(s) of the health condition, medications, medical problems, allergies, vaccination history, visit history, or communications with healthcare providers.

**Personal Health Record (PHR) Service Providers:** Organizations which assist in providing PHR capabilities to consumers. Organizations that provide these capabilities may include: vendors, healthcare providers, health data banks, healthcare payors, etc.

**Pharmacogenetics:** The merger of pharmacology and genetics into a field that pertains to the hereditary responses to drugs.

**Point-to-Point Exchange:** A direct link or communication connection with defined endpoints.



**Providers:** The healthcare clinicians within healthcare delivery organizations with direct patient interaction in the delivery of care, including physicians, nurses, and other clinicians. This can also refer to healthcare delivery organizations.

**Public Health Agencies/Organizations (local/state/territorial/federal):** Local, state, territorial, and federal government organizations and clinical care personnel that exist to help protect and improve the health of their respective constituents.

**Registries:** Organized systems for the collection, storage, retrieval, analysis, and dissemination of information to support health needs. These may include registries of phenotypic and genotypic information.

**System Vendors:** Organizations that develop and provide health information technology solutions. These solutions may include applications, data repositories, web services, etc., that contain or support the organization of genetic/genomic information.

**Testing Laboratories:** Medical testing laboratories, either within a hospital, ambulatory, or clinician office environment and/or operating as a free-standing entity, which analyze specimens as ordered by providers to assess the health status of patients. For this use case, these are testing laboratories which perform genetic/genomic laboratory tests ordered by genetic specialists and clinicians to assess the genetic status of patients.