U.S. Department of Health and Human Services

Office of the National Coordinator for Health Information Technology

Personalized Healthcare Detailed Use Case

March 21, 2008

Table of Contents

List of Figures

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) have been developed as the 2008 Use Cases which will be processed in the national HIT agenda activities in 2008.

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

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

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

This 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 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 by which healthcare providers can customize treatment and management plans for patients based on their unique genetic makeup. The 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 focuses on the exchange of genetic/genomic test information, personal and family health history, and the use of analytical tools in electronic health records (EHRs) to support clinical decision-making.

In specific terms:

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

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 information, personal and family health history, and analytical tools into EHRs to support clinical decision-making. Family health history relies on gathering data from disparate sources, increasing the need for interoperability. Ideally, family health history would be gathered at the point of care rather than retrospectively by interview during 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 other laboratory test information, may have 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.

While recognizing the issues and obstacles associated with these assumptions and to support these needs, the Personalized Healthcare 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 performs genetic or genomic testing after it receives genetic/genomic test orders and any accompanying information necessary for the testing in an interoperable form. 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 the management of their patients. Both clinicians and consumers have access to this information via the EHR or PHR.

While this topic is considered to be out-of-scope for this use case, it is recognized to be of high importance. The PHC Workgroup recognizes the uniqueness and complexity of newborn genetic screening (including in utero and other perinatal testing) - which shares characteristics with other genetic tests but has unique interoperability needs. 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. This important combination is discussed in more detail in Sections 7.0 and 8.0.

3.0 Use Case Stakeholders

Figure 3-1. Personalized Healthcare Use Case Stakeholders Table

Stakeholder	Contextual Description
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, physician assistants, nurses, psychologists, pharmacists, medical geneticists, genetic counselors, pathologists, and other licensed and credentialed personnel involved in treating patients.
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.
Electronic Health Record (EHR) System Suppliers	Organizations which provide specific EHR solutions to clinicians and patients such as software applications and software services. These suppliers may include developers, providers, resellers, operators, and others who may provide these or similar capabilities.
Genetic Specialists	Medical geneticists, genetic counselors, and clinicians (including pathologists) 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.
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.

Stakeholder	Contextual Description
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.
Knowledge Engineers	Knowledge engineers capture clinical knowledge in a structured form and incorporate it into tools supporting clinical practice. The knowledge can be represented in different ways such as rule sets, knowledge bases, guidelines, and other content to assist with a variety of different kinds of decision support.
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 interpretation of 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, in vitro diagnostic test manufacturers, medical device suppliers, etc.
Patients	Members of the public who receive healthcare services.
Personal Health Record (PHR) System Suppliers	Organizations which provide specific PHR solutions to clinicians and patients such as software applications and software services. These suppliers may include developers, providers, resellers, operators, and others who may provide these or similar capabilities.

Stakeholder	Contextual Description
Public Health Agencies/ Organizations (federal/state/local/ territorial/tribal)	Federal, state, local, territorial, and tribal 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. This also includes government agencies and professional associations which define, develop, and support registries. 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 meet regulatory standards for clinical laboratories and analyze specimens as ordered by providers to assess the health status of patients. Specifically, testing laboratories perform genetic/genomic and other 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. Some of these topics will appear in both the cross-cutting and use case-specific sections so that, in addition to the shared characteristics of the issue, considerations specific to a use case may be addressed.

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

- **Confidentiality, privacy, and security:**
    - In order for consumers to accept electronic health records, appropriate privacy and security protections may be needed to manage access to personal health information. Consumers may also want to decide who will view and communicate their personal health information. Privacy and security controls and the means of restricting data access are not standardized or regulated.
        - Without adequate 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 health information exchange activities 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, implementation, and maintenance 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.

In addition to the cross-cutting issues and obstacles described above, several other issues or obstacles exist that are specific to this use case. 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. While some issues will be addressed through HIT standardization and harmonization activities, policy development, the process of health information exchange, and other related initiatives, other issues will have to be specifically addressed within the context of personalized healthcare and genetic/genomic information and are delineated below.

- **Confidentiality, privacy, security and data access:**
    - Access to a personally controlled health record may help facilitate the acceptance of personalized healthcare. The consumer would have control of an actual medical record, rather then having only the passive ability to view their record through a portal. This would give the consumer access control, security and the choice of releasing appropriate information to other family members if recommended by their clinician.
        - Not using this approach may hinder the acceptance of personalized medicine due to concerns of privacy and access control on the part of the consumer.
    - The implementation of personalized healthcare may create additional risk of misuse of family history, disease risk, and predisposition information. In addition, whenever personal health information is stored, transmitted, archived or destroyed, it must be appropriately secured and audit trails should be made available.
        - Consumers may lose privacy protection or face unfair consequences (e.g., denial of health insurance or increased premiums) 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, dissimilar 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.
    - Information about who has accessed their records may be useful to patients. Specific to personalized healthcare is the situation in which family members may have access to information that would otherwise not be pertinent or available. State and federal laws vary in how they address consent and/or authorization. Therefore, these laws and associated policies need to be analyzed before rules for authorization and consent can properly protect patient privacy and confidentiality.

- Without the proper guidelines for information access, clinicians and consumers may not be willing to adopt some of the new technologies available for personalized healthcare.

- **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 harmonized 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 patients, authorized family members, and other authorized patient advocates.

  - 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 document entitled “Family Health History Multi-Stakeholder Dataset 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 and be in conflict with one another since this information 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.

  - Advances in the use of genetic and genomic information are being made at a rapid rate. This may cause situations in which, for example new information is discovered regarding a specific nucleotide sequence after a patient has been diagnosed and treated. Mechanisms that could incorporate this new knowledge and communicate it in a timely manner to both consumers and clinicians would promote personalized healthcare.

        - Without this type of surveillance and communications system in place, patients may not benefit from future advances in genetic/genomic knowledge.

    - Specific genetic information may be applicable to multiple disease processes, including pharmacogenetic information. The EHR could facilitate the cross utilization of genetic/genomic information in all relevant clinical settings.

        - Without a broad approach to genetic/genomic information, the full potential of personalized healthcare and the resulting benefits will not be realized.

    - 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 of 1996 (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.

- **Consumer and clinician literacy and education:**

    - Consumers may lack the experience, technical knowledge, and healthcare literacy regarding genetic testing and its implications to their own health. Moreover, this lack of understanding may drive consumer fear and lead consumers to resist sharing important information with clinicians.

        - Without the proper education for consumers, the pace of acceptance of genetic/genomic testing and personalized healthcare may be slow.

    - Likewise, the clinicians charged with delivering and handling this complex information may also lack training and knowledge in these new areas of clinical knowledge and technology.

        - Without the proper education and training for clinicians, the implementation, acceptance, and utilization of personalized healthcare may be hampered.

- **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.
- Keeping HIT tools in pace with genetic/genomic technology advances is a significant obstacle. Also, there is 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 harmonized standards for this information, it may be difficult to transmit information accurately between systems.
- Pharmacogenetics and pharmacogenomics are important and growing areas of personalized healthcare but the fields are still a developing medical practice. There is limited knowledge about which genes are involved in specific drug responses. Though significant knowledge is already available and in use, the workflows and data requirements are evolving.
    - Until the field matures and standard protocols and datasets are developed, it will be difficult to achieve interoperability.

5.0 Use Case Perspectives

The Personalized Healthcare Detailed Use Case describes personalized healthcare from 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, clinical specialists, advanced practice nurses, physician assistants, genetic counselors, medical geneticists, pathologists, psychologists, and other personnel that conduct clinical assessment and management activities and participate in evaluation, gathering of patient's personal and family heath history information, 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 laboratory director, laboratory supervisor, laboratory technicians, or other relevant staff. These personnel perform genetic/genomic and other laboratory tests ordered by clinicians to assess the genetic status of patients, generate test data, interpret the data in the context of other personal and family health information, perform a risk assessment in the context of family history information if needed, develop the patient report, and send the report to the ordering clinician.

- **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.

- **Information Exchange**

The information exchange perspective may include a variety of organizations including free-standing or geographic health information exchanges (e.g., Regional Health Information Organizations (RHIOs)), integrated care delivery networks, provider organizations, health record banks, public health networks, specialty networks, etc.

These entities may support specific functional capabilities which assist in facilitating health information exchange activities.

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 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 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 and 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. The testing laboratory performs the tests, analyzes the test data using genetic/genomic databases and repositories, and interprets the data. In addition, the testing lab considers other personal and family health information, performs a risk assessment in the context of family history information if needed, develops the patient report, and transmits the report back to the authorized providers. The other part of this scenario focuses on determining appropriate preventative action, treatment protocol, messaging, and clinical interpretation of test 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

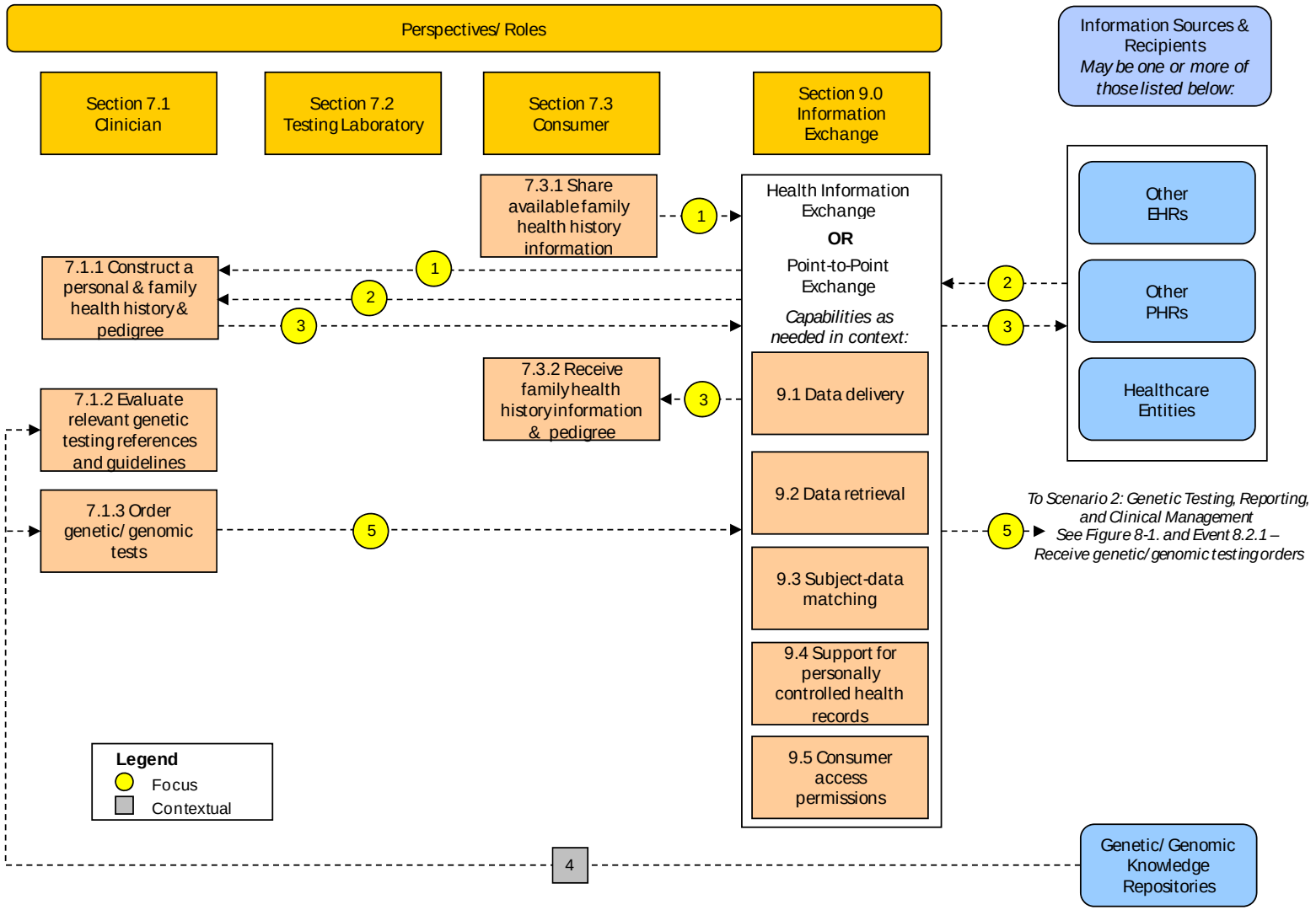

Figure 7-2. Clinical Assessment Scenario Flows

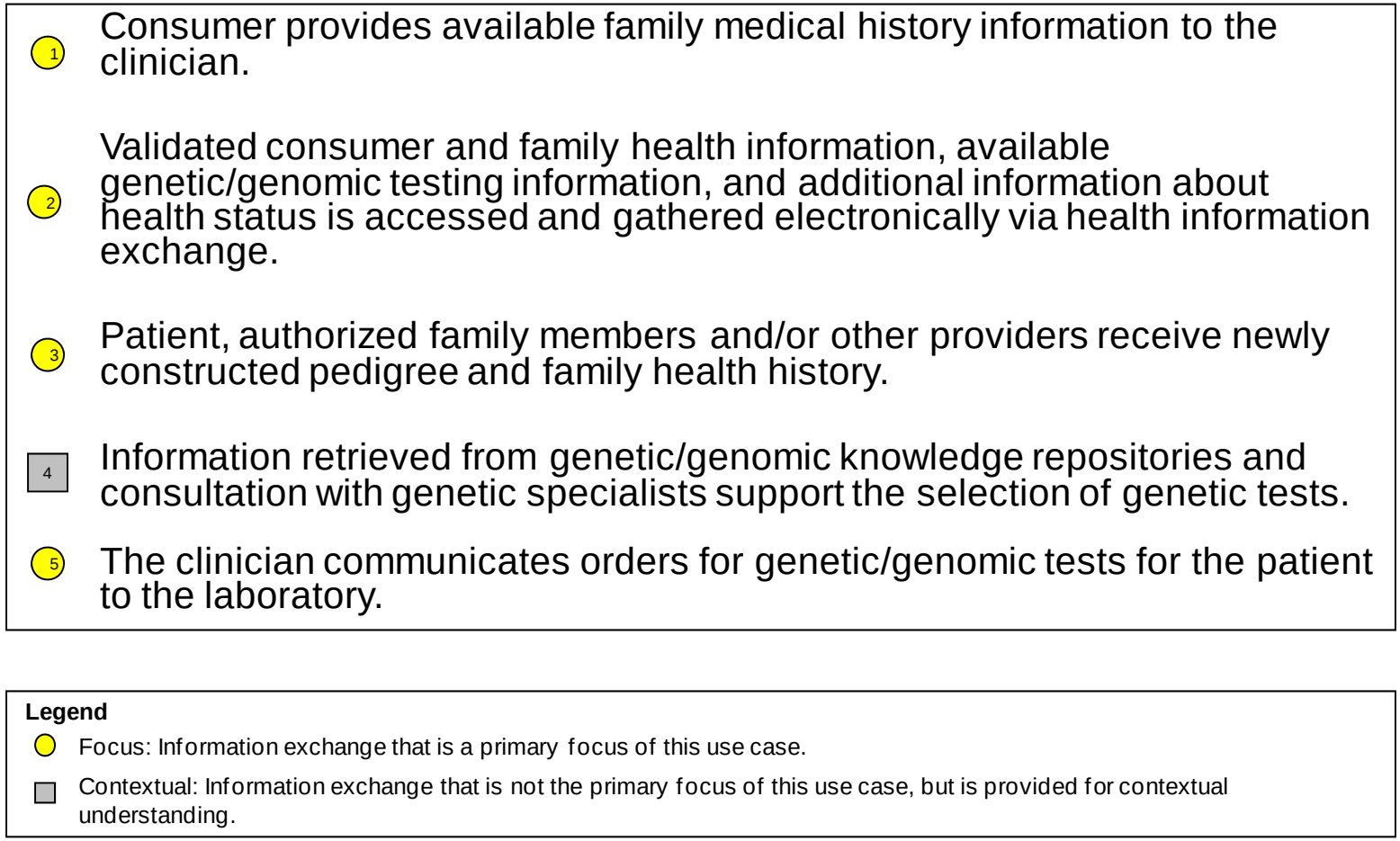

Figure 7-3. Clinical Assessment, Clinician Perspective

Code	Description	Comments
7.1.1	**Event:** Construct a personal & family health history & pedigree	**Figure 7-1, Flow 1, Flow 2, and Flow 3**
7.1.1.1	**Action:** 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 or other relevant diagnostic testing information from external sources to support the patient assessment and this corresponds to Figure 7-1, Flows 1 and 2. This may be in the context of testing for familial genetic disease, prenatal genetic testing, pharmacogenetic guidance for drug response, or any other personalized use of genetic or genomic testing. Consumer self-reported personal and family health history information may be available from the patient's PHR which corresponds to Figure 7-1, Flow 2. The current health status, along with the family and health history of the patient must form a transactionable and interoperable package. This information package includes standardized pedigree information joined with any information that is indicative of the current health status of the patient, predictive of various disease states, and helpful in elucidating drug interactions and responses. 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 and corresponds to Figure 7-1, Flows 1 and 2. In each case, the information source (e.g., authoritative clinical source, administrative source, or patient) should also be captured.
7.1.1.1a	**Alternative Action:** 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. In this alternative action, information may not be in an interoperable form, but would be electronic and corresponds to Figure 7-1, Flow 2. Enhanced functions as described in 7.1.1.1 would not be available.

Code	Description	Comments
7.1.1.1b	**Alternative Action:** 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. An additional consultative step may be necessary at this point between the clinician and patient. This action supports the rich family history currently utilized by clinicians as an intermediate step.
7.1.1.2	**Action:** 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, ideally with no duplications to gain the most comprehensive view of the patient's personal and family health history information. Clinicians would benefit from the ability to view personal and family health history information throughout the encounter. This action corresponds to Figure 7-1, Flow 2.
7.1.1.3	**Action:** 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 and this corresponds to Figure 7-1, Flow 3. Factors taken into account include duplication, currency, relevance to current clinical context, and data source.
7.1.1.4	**Action:** 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 and this corresponds to Figure 7-1, Flow 3. 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)), the consumer, and his/her authorized family members or proxies.
7.1.2	**Event:** Evaluate relevant genetic testing references and guidelines.	**Figure 7-1, Flow 4**

Code	Description	Comments
7.1.2.1	**Action:** Receive information from genetic/genomic knowledge repositories and/or decision support modules within EHRs.	Genetic/genomic knowledge repositories, registries or 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 EHR using clinical genetic databases such as Online Mendelian Inheritance in Man (OMIM), ALFRED, or other integrated or standalone decision support systems. These data interactions are typically contextual and ad hoc communications. However, the process of setting up decision support relies on standard vocabularies for family relationships, genetic test results, genetic test definitions, and genetic test interpretations as captured in the family and health history. This particular information exchange may present an opportunity for standardization in the future and corresponds to Figure 7-1, Flow 4.
7.1.2.2	**Action:** 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 and registries.
7.1.3	**Event:** Order genetic/genomic tests	**Figure 7-1, Flow 5**

Code	Description	Comments
7.1.3.1	**Action:** Write genetic/genomic test order.	The clinician may order genetic/genomic tests for the patient through a web application or via an available EHR and corresponds to Figure 7-1, Flow 5. The order specifies which laboratory test(s) need to be performed and the indication for performing these tests. These tests may be genetic/genomic in nature or they may be more generalized laboratory tests. In addition, the order includes patient demographic information, accompanying reference information such as patient and family health history (as described in section 7.1.1.1), general specimen information, billing and insurance information, and current physician and patient contact information. The clinician may utilize decision support tools and/or conduct consultations with a laboratory director, medical geneticist or pathologist to determine which tests to order. Test determinations are often based on probability percentages informed by the latest clinical knowledge.
7.1.3.2	**Action:** 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 has been selected to perform the testing and corresponds to Figures 7-1 and 8-1, Flow 5.

Figure 7-4. Clinical Assessment, Consumer Perspective

Code	Description	Comments
7.3.1	**Event:** Share available family health history information	**Figure 7-1, Flow 1**
7.3.1.1	**Action:** 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 and corresponds to Figure 7-1, Flow 1. The nature of this information has been described in section 7.1.1.1. In each case, the information source (e.g., authoritative clinical source, administrative source, or patient) is captured.

Code	Description	Comments
7.3.1.1a	**Alternative Action:** Patient uses an interoperable PHR to share his/her medical and family history with the clinician.	The consumer, in this case, uses an interoperable PHR which contains his/her personal health and family history. The information is accessed by appropriate clinicians to aid in diagnosis, analysis, and treatment and corresponds to Figure 7-1, Flow 1.
7.3.1.1b	**Alternative Action:** Patient self-reports personal medical and family history through an electronic portal.	The consumer self-reports personal medical and family history through an electronic portal which provides the appropriate data back to the clinician's EHR and corresponds to Figure 7-1, Flow 1. This information could potentially be transformed within the system to a standardized interoperable form.
7.3.1.1c	**Alternative Action:** Patient reports personal medical and family history by interview.	The consumer could be interviewed for all appropriate information during the course of a consultation with a clinician.
7.3.2	**Event:** Receive family health history information & pedigree	**Figure 7-1, Flow 3**
7.3.2.1	**Action:** 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 web access to the clinician's EHR. This action corresponds to Figure 7-1, Flow 3. 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.
7.3.2.1a	**Alternative Action:** Patient receives newly validated and updated personal and family health history information and pedigree, if appropriate, via an interoperable PHR.	The consumer, in this case, uses an interoperable PHR and incorporates his/her newly validated and updated personal and family health history information and pedigree from the clinician's EHR. This action corresponds to Figure 7-1, Flow 3. The information can be received from the appropriate clinician to aid in diagnosis, analysis, and treatment planning.

Code	Description	Comments
7.3.2.1b	**Alternative Action:** Clinician reports newly validated and updated personal and family health history information and pedigree, if appropriate, via patient consultation.	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.

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

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

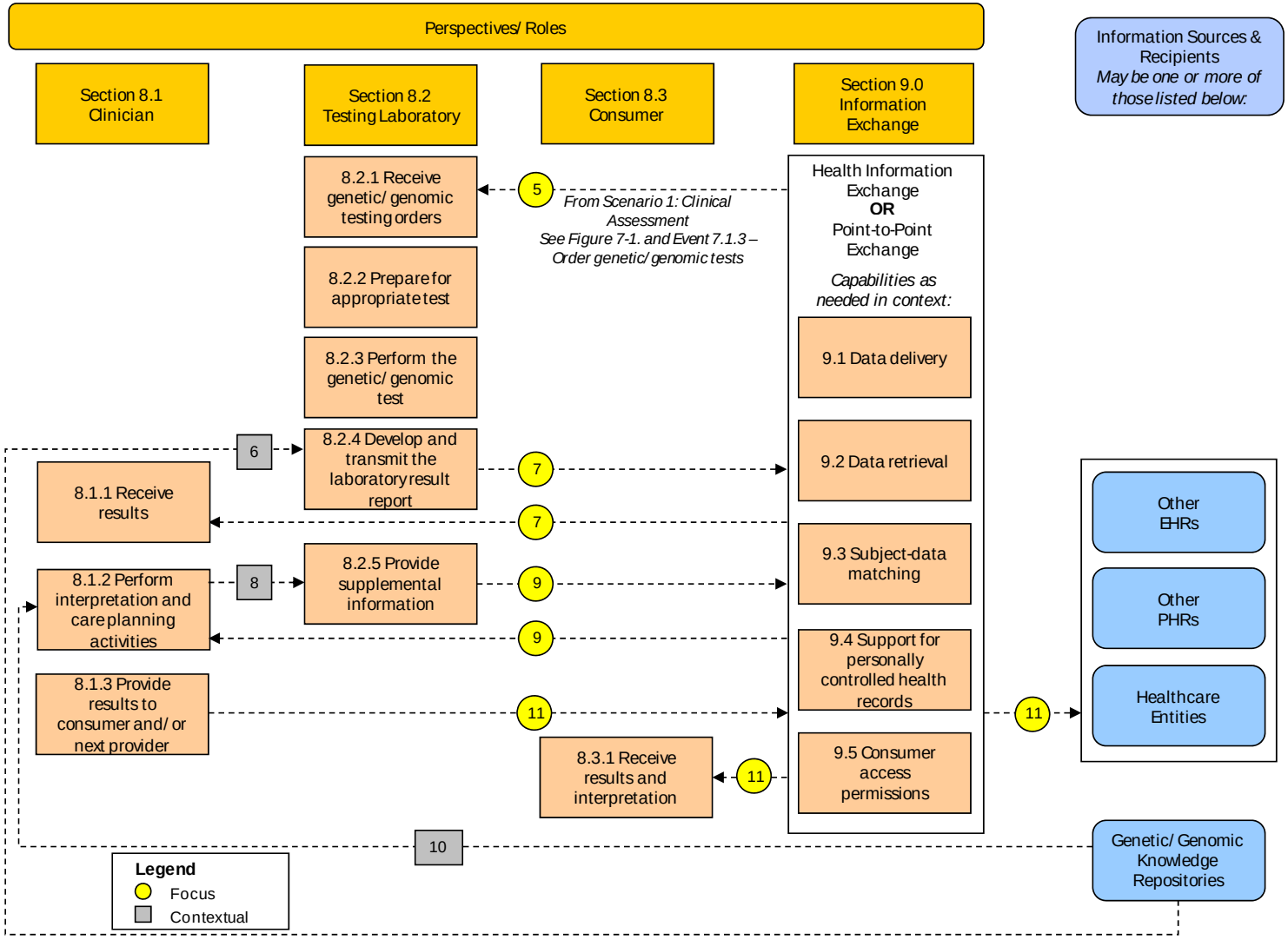

Figure 8-2. Genetic Testing, Reporting, and Clinical Management Scenario Flows

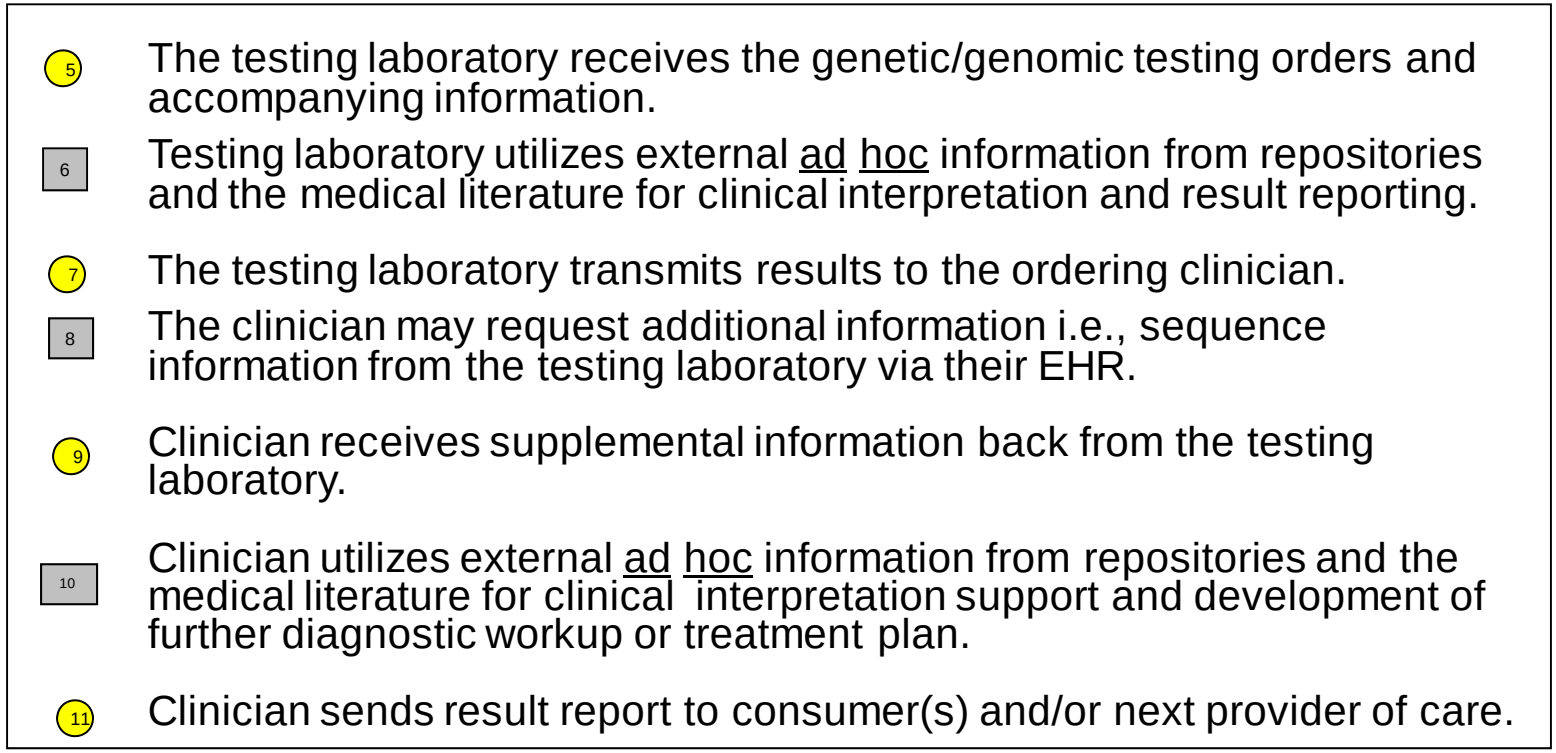

Figure 8-3. Genetic Testing, Reporting, and Clinical Management, Clinician Perspective

Code	Description	Comments
8.1.1	**Event:** Receive results	**Figure 8-1, Flow 7**
8.1.1.1	**Action:** 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. This action corresponds to Figure 8-1, Flow 7. 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. Interpretation of the complex primary laboratory data may be done by a qualified laboratory genetic specialist. The laboratory may provide key primary data such as nucleotide sequence or genetic variant information. There may also be a consultative interchange directly between the laboratory director/pathologist/medical geneticist and the ordering clinician.
8.1.2	**Event:** Perform interpretation and care planning activities	**Figure 8-1, Flow 8 and Flow 10**
8.1.2.1	**Action:** Perform interpretation and care planning activities.	The ordering clinician performs further clinical 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 Online Mendelian Inheritance In Man and corresponds to Figure 8-1, Flow 10. The ordering clinician may also consult with pathologists or other genetic testing experts 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.

Code	Description	Comments
8.1.2.2	**Action:** 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 may in the future be communicated to the clinician in a structured, standardized format. This corresponds to Figure 8-1, Flows 8 and 9. Standardization needs exist for clinicians and laboratories working with communication of genetic/genomic testing information. These future needs are focused on establishing more complete and robust data standards using 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.
8.1.3	**Event:** Provide results to the consumer and/or the next provider of care	**Figure 8-1, Flow 11**
8.1.3.1	**Action:** 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 clinicians' EHRs. This action corresponds to Figure 8-1, Flow 11. The information communicated may also include personal and family health information captured and organized by the ordering clinician such as a family pedigree as described in action 7.1.1.1. This result and interpretation would be most useful in a structured form.
8.1.3.2	**Action:** 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. This action corresponds to Figure 8-1, Flow 11. 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
8.2.1	**Event:** Receive genetic/genomic testing orders	**Figure 8-1, Flow 5**
8.2.1.1	**Action:** Receive and capture the genetic/genomic testing orders.	The testing laboratory receives the genetic/genomic testing orders, other important clinical information, and any previously reported genetic test results deemed necessary for the testing from a structured, standards-based electronic message. This action corresponds to Figure 8-1, Flow 5.
8.2.2	**Event:** Prepare for appropriate test	
8.2.2.1	**Action:** Prepare for and perform 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. Analysis may be necessary by the laboratorial staff to ensure that all information needed to perform the specialized tests has been received, validated, 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	**Alternative Action:** 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 ad hoc nature, but is an opportunity for focused communication in the future.
8.2.2.2	**Action:** 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	**Action:** 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.

Code	Description	Comments
8.2.3	**Event:** Perform the genetic/genomic test	
8.2.3.1	**Action:** Execute the steps required to perform the genetic/genomic test.	The testing laboratory performs the technical steps required to produce the genetic/genomic data.
8.2.4	**Event:** Develop and transmit the laboratory result report	**Figure 8-1, Flow 6 and Flow 7**
8.2.4.1	Action: Develop and transmit the laboratory result report.	The testing laboratory develops the laboratory report which may include specific test results and interpretation 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 directly from the LIS. This action corresponds to Figure 8-1, Flow 7. The patient report may also be transmitted to other authorized healthcare providers based on the consumer's preferences and/or needs. 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 and corresponds to Figure 8-1, Flow 6. The testing laboratory may also contract outside expertise for interpretation of these results if expertise is not available internally.
8.2.5	**Event:** Provide supplemental information	**Figure 8-1, Flow 8 and Flow 9**

Code	Description	Comments
8.2.5.1	**Action:** Provide supplemental information to the ordering clinician.	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 and standardized format. This action corresponds to Figure 8-1, Flows 8 and 9. 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 would help achieve the interoperable integration of genetic/genomic test information into EHRs.

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

Code	Description	Comments
8.3.1	**Event:** Receive results and interpretation	**Figure 8-1, Flow 11**
8.3.1.1	**Action**: Consumer receives laboratory results and clinical interpretation.	The consumer receives and incorporates available personal, family health, and/or genetic/genomic testing information via their PHR. This action corresponds to Figure 8-1, Flow 11. This information may have been self-reported earlier but may also be derived from clinicians' EHR systems, and other authoritative clinical and/or administrative data sources. Information in a standardized interoperable form may be sent to other providers of care using information exchange activities.
8.3.1.2	**Action:** Consumer provides consent and authorization to share information.	Consumers may additionally benefit from the ability to permit designated clinicians and other individuals (e.g., family members) to request and view information contained in the PHR (a.k.a., proxy access). This action corresponds to Figure 8-1, Flow 11. The consumer's authorized family members may also benefit from the ability to view, select, and/or incorporate information in the PHR, in accordance with the consumer's personal preferences.

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., Regional Health Information Organizations), 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, data receipt, and confirmation of delivery to EHRs, personally-controlled health records, other systems, and networks	Capability to securely deliver data to the intended recipient and 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, processes, and policies facilitating information exchange are utilized to confirm that the data available for retrieval match the person of interest to the clinician.

Code	Capability	Comments
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 consumer's 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.
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 not to make their health information available through network activities, those systems involved in exchanging health information may 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 purposes of this use case, health information exchange is the electronic movement of health-related data and information among organizations according to specific standards, protocols, and other agreed criteria. These functional capabilities may be provided fully or partially by a variety of organizations including free-standing or geographic health information exchanges (e.g., Regional Health Information Organizations (RHIOs)), integrated care delivery networks, provider organizations, health record banks, public health networks, specialty networks, and others supporting these capabilities. This term may also be used to describe the specific organizations that provide these capabilities such as RHIOs and Health Information Exchange Organizations.

Point-to-Point Exchange: For the purposes of this use case, point-to-point exchange includes direct interactions between two systems which do not involve intermediary information exchange functions to route and deliver the data. Representative architectures could include point-to-point messaging, service-oriented-architectures, or information exchange among participants using a common application platform.

10.0 Dataset Considerations

Currently, there are no standardized interoperable datasets and/or standards for the transfer of appropriate and necessary information to facilitate interoperable personalized healthcare delivery. As personalized healthcare is further defined, standardized datasets 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 and other clinical laboratory and pathology patient test reports. The first is critical in identifying and assessing patient risk and determining the genetic/genomic information that must be gathered or generated. The second is specific information identifying the genetic/genomic status of patients for the purposes of diagnosis, increased monitoring for early detection of disease, 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 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 and examples may be considered:

- Demographic information
    - Name
    - Unique identifier
    - Race/Ethnicity
    - Occupation
- Personal health information
    - History of specific disorders
    - Relevant non-genetic laboratory test and pathology data
    - Other clinical data such as radiology study results
    - Environmental exposure data
    - Any prior treatment for specific disorders
- Family history information

    - Disorders of family members
    - Ages of condition onset and/or death of various family members
    - Environmental exposure data
    - Relevant social data
    - Pedigree in structured form
- Personal genetic/genomic data
    - Prior genetic/genomic laboratory test results
    - Prior genetic status for specific disease
    - Full genome scan: deoxyribonucleic acid (DNA)
- Family genetic/genomic information
    - Genetic/genomic data of family members
    - Pedigree in structured form when appropriate
    - History of consanguinity
    - Consent/access allowance information

The document entitled “Family Health History Multi-Stakeholder Workgroup Dataset 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. There are parallel activities underway to develop specific terminology based on consensus throughout the industry.

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, physician assistants, nurses, psychologists, pharmacists, medical geneticists, genetic counselors, pathologists, and other licensed and 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: This refers to the fact that two persons have a common, recent ancestor. For example, unions/marriages contracted between second cousins or closer are categorized as consanguineous.

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: An activity 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 activity with specific components such as best practice guidelines, medication contraindication information, and access to recent research.

Department of Health and Human Services (HHS): 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): An electronic, cumulative record of information on an individual across more than one health care setting that is collected, managed, and consulted by professionals involved in the individual's health and care. This EHR description encompasses similar information maintained on patients within a single care setting (a.k.a., Electronic Medical Record (EMR)). This information may include patient demographics, progress notes, problems, medications, vital signs, past medical history, immunizations, laboratory and pathology information, and radiology reports.

Electronic Health Record (EHR) System Suppliers: Organizations which provide specific EHR solutions to clinicians and patients such as software applications and software services. These suppliers may include developers, providers, resellers, operators, and others who may provide these or similar capabilities.

Family Health History: Documentation of the health 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 (including pathologists) 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 an analysis of chromosomes, deoxyribonucleic acid (DNA), ribonucleic acid (RNA), or proteins.

Geographic Health Information Exchange/Regional Health Information Organizations: A multi-stakeholder entity, which may be a free-standing organization

(e.g., hospital, healthcare system, partnership organization) that supports health information exchange and enables the movement of health-related data within state, local, territorial, tribal, or jurisdictional participant groups. Activities supporting health information exchanges may also be provided by entities that are separate from geographic health information exchanges/Regional Health Information Organizations including integrated delivery networks, health record banks, and others.

Health Information Exchange (HIE): The electronic movement of health-related data and information among organizations according to specific standards, protocols, and other agreed criteria. These functional capabilities may be provided fully or partially by a variety of organizations including free-standing or geographic health information exchanges (e.g., Regional Health Information Organizations (RHIOs)), integrated care delivery networks, provider organizations, health record banks, public health networks, specialty networks, and others supporting these capabilities. This term may also be used to describe the specific organizations that provide these capabilities such as RHIOs and Health Information Exchange Organizations.

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.

Knowledge Engineer: Knowledge engineers capture clinical knowledge in a structured form and incorporate it into tools supporting clinical practice. The knowledge can be represented in different ways such as rule sets, knowledge bases, guidelines, and other content to assist with a variety of different kinds of decision support.

Laboratory Information System (LIS): A laboratory information system is a class of software which handles receiving, processing, transmitting, 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 interpretation of 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, in vitro diagnostic test manufacturers, 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 nation's health through the development of an interoperable harmonized health information infrastructure.

Pathologist: A physician, trained and board certified in Anatomic Pathology and/or Clinical Pathology, some of whom specialize in genetic/genomic testing and are subspecialty board certified in Molecular Genetic Pathology.

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): An electronic, cumulative record of health-related information on an individual, drawn from multiple sources, that is created, collected, and managed by the individual or an agent acting for the individual. The content of and rights of access to the PHR are controlled by the individual or agent. The PHR is also known as the electronic Personal Health Record (ePHR).

Personal Health Record (PHR) System Suppliers: Organizations which provide specific PHR solutions to clinicians and patients such as software applications and software services. These suppliers may include developers, providers, resellers, operators, and others who may provide these or similar capabilities.

Pharmacogenetics: Pharmacogenetics is a subset of pharmacogenomics and is defined as the study of variations in deoxyribonucleic acid (DNA) sequence as related to drug response.

Pharmacogenomics: The study of variations of DNA and ribonucleic acid (RNA) characteristics as related to drug response.

Point-to-Point Exchange: Point-to-point exchange includes direct interactions between two systems which do not involve intermediary information exchange functions to route and deliver the data. Representative architectures could include point-to-point messaging, service-oriented-architectures, or information exchange among participants using a common application platform.

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

Public Health Agencies/Organizations (federal/state/local/territorial/tribal): Federal, state, local, territorial, and tribal 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. This also includes government agencies and professional associations which define, develop, and support registries. 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 meet regulatory standards for clinical laboratories and 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 and other laboratory tests ordered by genetic specialists and clinicians to assess the genetic status of patients.