

Personalized Healthcare
Draft Detailed Use Case Feedback Instructions

The Draft Detailed Personalized Healthcare (PHC) Use Case is available for public feedback. If you would like to provide feedback about this document please provide your organization's information and feedback below. Suggestions for providing your feedback are listed below.

- 1.) Whenever possible, identify the specific section of the document to which your feedback refers.

Example A:

6.1 Reporting and Data Sharing

6.2 Investigation

- 2.) Please save your document using a naming convention e.g.: (YRYR.MO.DY-Organization Name-
PHC Feedback.doc)

Example B:

2008.01.18- ABC Organization-PHC Draft Detailed Feedback.doc.

- 3.) Email this document to usecase3@hhs.gov by the close of business on Friday, February 15th, 2008.
- 4.) You will receive an acknowledgement email confirming that ONC has received your feedback. Disposition of all feedback received will be available upon request.

Thank you.

Organization Contact Information

Please provide the contact information for your organization: Organization Name, Contact Person Name, Contact Person Telephone, Contact Person Email Address, Date Feedback was Submitted.

College of American Pathologists

Susan Eads Role

202-354-7102

srole@cap.org

February 15, 2008

Draft Detailed Use Case

Please review the Draft Detailed Personalized Healthcare Use Case and provide feedback below.

The College of American Pathologists (CAP) is providing the following written response to the Office of the National Coordinator for Health Information Technology's (ONC) request for feedback on the Draft Detailed Personalized Healthcare Use Case. The CAP is a national medical specialty society representing more than 17,000 physicians who practice clinical and/or anatomical pathology and laboratory medicine. CAP members practice

their specialty in a variety of settings, including community hospitals, independent clinical laboratories, academic medical centers, and federal and state health facilities.

The concept of personalized healthcare relies heavily on genotypic (laboratory-derived) information as part of the electronic health record (EHR) to be correlated to a patient's family history, clinically relevant information, outcomes, exposures, etc. Laboratory testing already provides key health information that may influence as much as 60 to 70 percent of health care decision-making¹ and is likely to increase in importance. For that reason, it is crucial that pathologists, as directors of medical laboratories, be involved in almost every group that is engaged in standard setting for this initiative.

The CAP believes that a fundamental change is required in the organization of this use case in order to more accurately represent how genetic tests are used and interpreted. Throughout the current draft of the use case, the laboratory and testing functions are incorrectly separated from the interpretative role of a laboratory physician and incorrectly delegated to the clinician directly caring for the patient. It is true that clinicians do use genetic test interpretations in developing management plans for their patients, however, the clinician responsible for interpreting test data from the medical laboratory is *the pathologist working within the laboratory*. The pathologist interprets the data produced in the laboratory in the context of the personal and family health information and family history, includes this information in the patient test report, and transmits the information in both a narrative and structured form to another clinician (who often lacks training and experience in interpretation of genetic/genomic test data). The patient's test report may include specific sequence information, genetic mutations or variants, expression levels, a risk assessment if appropriate, and the clinical significance of the test results. Raw data currently is not transmitted, nor would it be useful. In addition, it is likely there will be consultative interchange between the primary patient care clinician and the pathologist to discuss unusual or unexpected results or for further clarification about the significance of the test result in the context of the specific patient and/or the patient's family members. We have identified in our detailed comments below specific instances where the role of the pathologist in the interpretation of personalized healthcare tests should be represented in the document.

A pathologist is a physician, trained and board certified in Anatomic Pathology and/or Clinical Pathology, who is qualified to direct a clinical laboratory. The pathologist will have prior exposure to genetic/genomic testing and may be subspecialty board certified in Molecular Genetic Pathology. Because genetic/genomic information from laboratory tests will increasingly be integrated into every aspect of the practice of medicine, the CAP believes it is preferable to use the more general term "pathologist" throughout the document rather than the more specific term "molecular pathologist."

¹ Forsman R.W., Why Is the Laboratory an Afterthought for Managed Care Organizations? Clin Chem 1996; 42:813-16.

2.0 Introduction and Scope

In the description of the Genetic Testing, Reporting, and Clinical Management Scenario, the term “testing” appears. As used, “testing” involves performing the test and interpreting the results. For clarity, the CAP recommends changing the word “testing” at the end of the first sentence of the scenario description to the phrase “*test performance and interpretation of test results.*”

Currently, genetic/genomic test data is interpreted within the laboratory by the pathologist and not sent to the clinician for interpretation. The clinician only interprets the reported test results for patient management, additional diagnostic testing, or treatment plan development. The CAP recommends the second sentence of the Genetic Testing, Reporting, and Clinical Management Scenario description be changed to: “The testing laboratory *under the direction of a pathologist* performs the tests, *interprets test results in the context of the clinical information and the medical literature*, develops the patient report, and transmits this information *with recommendations* back to authorized providers.” (Italics indicate new text.)

The CAP recommends clarification of the following statements in section 2.0:

- **“Genetic/genomic information, unlike much other laboratory test information, has lifelong significance.”**

This assertion is only true for tests looking at heritable variants, but is generally not true for cancer genomic testing, which can vary as the cancer progresses and is not always heritable. Cancer may have both genetic and somatic components because underlying risk of a cancer can be inherited, but not all cancers have a heritable risk component, at least not that has been identified. Somatic changes refer to genetic changes only in the cancer cells, which are not present in all cells of the patient and, therefore, cannot be passed on to offspring. Because this use case addresses the broader scope of genomic testing, and not just genetic heritable testing, this statement should be reconsidered.

- **“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.”**

This approach is not realistic given the Health Insurance Portability and Accountability Act regulations, but may be possible if a consenting process could be developed that permits transfer of medical information from one patient to another related patient’s medical record.

3.0 Use Case Stakeholders

The contextual description of “Clinicians” lists physicians and medical geneticists, who are physicians. It does not list pathologists, who also are physicians. The CAP recommends the contextual description be changed to: “Healthcare providers with patient care responsibilities, including physicians, *such as primary care physicians, family practice physicians, medical geneticists, pathologists, and other specialty physicians*, advanced practice nurses, pharmacists, physician assistants, nurses, genetic counselors, and other credentialed personnel involved in treating patients.” (Italics indicate new text.)

Similarly, the contextual description of “Genetic Specialists” lists clinicians and medical geneticists, who are clinicians. It does not list pathologists, who also are clinicians. The CAP recommends the contextual description be changed to: “*Physician specialists, including medical geneticists and pathologists, as well as genetic counselors and other clinicians who participate in evaluation, diagnostic planning, and genetic/genomic test ordering and result interpretation activities.*” (Italics indicate new text.)

The contextual description of Laboratory Organizations does not refer to the appropriate interpretation of laboratory information. The CAP recommends that the contextual description be changed to: “Advocacy/professional organizations or societies such as the College of American Pathologists (CAP) or the National Committee for Clinical Laboratory Standards (NCCL) which are concerned with the appropriate use of laboratory technology and *interpretation of laboratory information in clinical medicine.*” (Italics indicate new text.)

The contextual description of “Electronic Health Record (EHR) Service Providers” states such service providers assist in providing EHR capabilities to consumers. These service providers also assist in providing EHR capabilities to healthcare providers. The CAP recommends the first sentence of the contextual description be changed to: “Organizations which assist in providing EHR capabilities to *healthcare providers and consumers.*” (Italics indicate new text.)

The contextual description of “Manufacturers/Distributors” does not include manufacturers of in vitro diagnostic tests, which are central to genetic/genomic testing. The CAP recommends that “*in vitro diagnostic test manufacturers*” be added to the contextual description.

4.0 Issues and Obstacles

Under Clinical Decision Support, there is no discussion of the need to update clinical decision support tools. The CAP recommends adding a sub-bullet as follows: “*Because medical knowledge and practice standards advance, clinical decision support tools need to be updated frequently. The basis for such updates needs careful input from physicians and other*

healthcare providers to establish when new standards are ready for practice in the evolution of our knowledge.”

Also in the last paragraph under Clinical Decision Support, there is no discussion of specific genetic/genomic information applying to multiple disease processes. The CAP recommends adding the following sentences after the first sentence in that paragraph: *“Specific genetic information may be applicable to multiple disease processes, especially pharmacogenetic information. The EHR should facilitate the cross utilization of genetic/genomic information in all relevant clinical settings.”*

The CAP recommends clarification of the following statement under the heading 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.” Only non-genetic specialists gather family health history in an “ad hoc and non-standardized manner.” Medical geneticists and genetic counselors currently take a systematic history, and this systematic method could be built into EHR systems to facilitate the taking of a more systematic family health history by non-genetic specialists.

5.0 Use Case Perspectives

In the description of the clinician perspective, a number of health care professionals are listed who “...conduct clinical assessment and management activities and participate in evaluation, diagnostic planning, genetic/genomic test ordering, and result interpretation activities.” Pathologists are health care professionals who analyze and interpret results of genetic/genomic tests for other physicians. Pathologists also consult with those physicians regarding diagnostic planning. The description also includes the following sentence: “The clinician may also be working within the testing laboratory.” The CAP recommends replacing this sentence with the following sentence: *“Pathologists are physicians who work from within the testing laboratory.”*

In the description of the testing laboratory perspective, there needs to be greater emphasis on the head of the testing laboratory, the pathologist laboratory director. In addition, the services performed by the testing laboratory need to be expanded to include the interpretation of test data, performance of risk assessments, and development of the patient report. The CAP recommends that the description be replaced by the following sentence: *“The testing laboratory perspective includes the pathologist laboratory director and other medical laboratory personnel such as the supervisor, 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 transmit the report to the ordering clinician.”* (Italics indicate new text.)

6.0 Use Case Scenarios

In the Genetic Testing, Reporting, and Clinical Management Scenario description, the services performed by the testing laboratory need to be expanded to include the interpretation of test data and performance of risk assessments. The CAP recommends that the fourth and fifth sentences be replaced with the following sentences: “The testing laboratory performs the tests, *analyzes the test data using genetic/genomic databases and repositories, interprets the data in the context of the medical literature as well as 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 the 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.” (Italics indicate new text.)

7.0 Scenario 1: Clinical Assessment

The CAP recommends that the following changes and clarifications be made in the diagram and tables of the Clinical Assessment Scenario:

- **Figure 7-1. Clinical Assessment – Add step of consultative interaction with testing laboratory to the diagram. The CAP suggests adding a box containing this step between box 7.1.2 and 7.1.3.**
- **Figure 7-2. Clinical Assessment Scenario Flows, #4 – Add new text as indicated in the following sentence: “Information from genetic/genomic knowledge repositories *and consultation with genetic specialists* support the selection of genetic tests.” (Italics indicate new text.)**
- **Figure 7.3. Clinical Assessment, Clinician Perspective, 7.1.1.1 –**
 - **Add new text as indicated in the following sentence: “The clinician gathers current patient personal health history, family health history and any past genetic/genomic testing information, *other laboratory and pathology testing information as well as other diagnostic information, such as radiology study results,* from several external sources to support the patient assessment.” (Italics indicate new text.)**
 - **Delete the word “oncology” from the following sentence as indicated: “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.” Pharmacogenetic guidance may be indicated for treatments other than oncology.

- **Figure 7.3. Clinical Assessment, Clinician Perspective, 7.1.1.1** – Clarify the text containing references to obtaining information from primary care physicians. The current text suggests that a genetic specialist will collect the information when it is highly likely that primary care physicians, family practice physicians, and other non-genetic specialists will be doing this in the future.
- **Figure 7.3. Clinical Assessment, Clinician Perspective, 7.1.1.1b** – Delete “ by contacting the patient’s PCP” at the end of the sentence.
- **Figure 7.3. Clinical Assessment, Clinician Perspective, 7.1.3.1** – Replace “molecular pathologist” with “*genetic specialist*.” In other sections, the CAP recommended that the definition of “genetic specialist” be changed to include pathologists and that a definition of “*Pathologist*” be added.
- **Figure 7.3. Clinical Assessment, Clinician Perspective, 7.1.3.1 and 7.1.3.2** – Clarify text to acknowledge that pathologists routinely work with ordering clinicians to select testing and focus overly broad requests toward more cost-effective initial targeted requests.
- **Figure 7.3. Clinical Assessment, Clinician Perspective 7.1.3.1** – Add text to clarify that informed consent should be obtained from the patient prior to the ordering or performing of genetic/genomic testing.
- **Figure 7.3. Clinical Assessment, Clinician Perspective, 7.1.3.2** – Add new text as indicated in the following sentences: “The genetic/genomic *and other laboratory* test orders *are* 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, *including historical and physical findings to allow pathologists to confirm expected predictive value of tests performed*, general specimen information, billing information, and physician and patient contact information.” (Italics indicate new text.) Additionally, clarify the text to acknowledge that other diagnostic tests may be needed, such as radiology tests or biopsy procedures with histologic and phenotypic evaluation.
- **Figure 7-4. Clinical Assessment, Consumer Perspective, 7.3.1** – Clarify text to acknowledge: (1) the patient also may have paper reports and records from previous clinical encounters, *e.g.*, test results and physician summary notes, which will need to be incorporated into the EHR, possibly by scanning of the documents; and (2) the patient may give the clinician permission/consent to directly contact and

collect information from other physicians who have previously cared for the patient. This information also needs to be incorporated into the EHR either by scanning of paper documents provided or through electronic transmission from one EHR system to another, which should be made possible through addressing EHR compatibility up front.

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

The CAP recommends that the following changes and clarifications be made in the diagram and tables of the Genetic Testing, Reporting, and Clinical Management Scenario:

- **Figure 8-1. Genetic Testing, Reporting, and Clinical Management – Clarify text to acknowledge that the interpretation of primary laboratory data of this complexity (e.g., raw DNA sequence tracings) is done by the pathologist and is not transmitted to primary patient care clinicians for interpretation. The pathologist may need information from genetic/genomic data repositories and databases for the interpretation of the test data and the generation of the patient report. Almost invariably, such clinicians lack training and experience in genetic/genomic data interpretation. The pathologist will provide key primary data, such as nucleotide sequence variants, in the synthesized report with the interpretation of their significance. It is very likely there will be consultative interchange between the primary patient care clinician and the pathologist to discuss unusual or unexpected results or the significance of a result in the context of the specific patient and/or the patient’s family members. The clinician integrates the laboratory data, which has already been evaluated and interpreted by the pathologist, into the overall clinical picture of the patient to make an assessment and then a management plan. Add a box between 8.2.3 and 8.2.4 containing the following new text: “*pathologist performs interpretation.*” (Italics indicate new text.) Add new text as indicated to box 8.1.1 “*Receives results and interpretation.*” (Italics indicate new text.) Add new text as indicated to box 8.1.2 “*Performs additional interpretation and care planning activities.*” (Italics indicate new text.) See also comments below regarding Figure 8-3. Genetic Testing, Reporting and Clinical Management, Clinician Perspective, 8.1.2.1 and Figure 8-4. Genetic Testing, Reporting, and Clinical Management, Testing Laboratory Perspective, 8.2.4.1.**
- **Figure 8-2. Genetic Testing, Reporting, and Clinical Management Scenario Flows – Add another numbered step for the laboratory to access genetic/genomic databases and repositories and the medical literature for the interpretation of test results and the generation of the patient report. Note that this access may be from within the LIS rather than from the EHR.**
- **Figure 8-2. Genetic Testing, Reporting, and Clinical Management Scenario Flows, #9 – Add new text as indicated in the following sentence: “Clinician utilizes external**

ad hoc data from repositories *and the medical literature* for clinical interpretation support *and development of further diagnostic workup or treatment plan.*” (Italics indicate new text.)

- **Figure 8-2. Genetic Testing, Reporting, and Clinical Management Scenario Flows, #10** – Add new text as indicated in the following sentence: “Clinician sends result report to consumer(s) and/or next provider of care *or proceeds with next diagnostic cycle or treatment plan.*” (Italics indicate new text.)
- **Figure 8-3. Genetic Testing, Reporting, and Clinical Management, Clinician Perspective, 8.1.2.1** – At beginning, add new sentence as indicated: “*The pathologist performs interpretation and transmits this information to the ordering clinician.*” (Italics indicate new text.) Add new text as indicated in the following sentence: “The ordering clinician performs *additional* interpretation, care planning, and care plan implementation activities utilizing several forms of decision support including, but not limited to, *medical literature*, external data repositories and integrated decision support information and algorithms built into the clinician’s own EHR.” (Italics indicate new text.) In the following sentence, delete “molecular” and add new text as indicated: “The ordering clinician may also consult with ~~molecular~~ *pathologists* during the process of analyzing and interpreting the genetic/genomic *and other laboratory* test results.” (Italics indicate new text.)
- **Figure 8-3. Genetic Testing, Reporting, and Clinical Management, Clinician Perspective, 8.1.3** – Add new text as indicated in the following phrase: “Event: Provide results to the consumer and/or the next provider of care, *or develop further diagnostic workup or treatment plan and interface with pharmacy or medical specialists*” (Italics indicate new text.)
- **Figure 8-4. Genetic Testing, Reporting, and Clinical Management, Testing Laboratory Perspective, 8.2.2.1** – Add new text as indicated in the following sentences: “*The pathologist and laboratory* staff prepares for the genetic/genomic test based on the testing orders received. A level of analysis is necessary by the *pathologist and laboratory* 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).” (Italics indicate new text.)
- **Figure 8-4. Genetic Testing, Reporting, and Clinical Management, Testing Laboratory Perspective, 8.2.2.1a** – Add new text as indicated in the following sentence: “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 *and proper test interpretation* to take place.” (Italics indicate new text.) Additionally,

add new text as indicated in the following sentence: “This information exchange is likely to be of an **ad hoc** nature, *but may be standardized within the test ordering sections of the EHR in the future.*” (Italics indicate new text.)

- **Figure 8-4. Genetic Testing, Reporting, and Clinical Management, Testing Laboratory Perspective, 8.2.3.1** – Add new sentence as indicated: “The testing laboratory performs the technical steps required to produce the genetic/genomic data. *Instrument data may be directly transferred to the LIS through a direct interface.*” (Italics indicate new text.)
- **Figure 8-4. Genetic Testing, Reporting, and Clinical Management, Testing Laboratory Perspective, 8.2.4.1** – At beginning, add new sentence as indicated: “*The pathologist interprets the data in the context of the personal and family health information and family history as well as the medical literature and generates the patient test report, which may include specific sequence information, genetic mutations or variants, expression levels, a risk assessment if appropriate, and the clinical significance of the test results.*” (Italics indicate new text.) Add new text as indicated in the following sentence: “*The patient test report is transmitted in both narrative and structured form within the LIS along with transmission of a standards-based electronic message to the ordering clinician’s EHR or other clinical data system.*” (Italics indicate new text.)
- This scenario does not take into account the role of a “local” pathologist, who can serve as an intermediary in the selection of and consultation with a reference laboratory to which personalized healthcare test orders may be sent. This role would include follow-up consultations on site with the ordering clinician and/or discussions with the reference laboratory, if the ordering clinician has questions regarding the results or process.

10.0 Dataset Considerations

The CAP recommends the following clarifying additions:

- **First paragraph:** “...2) genetic/genomic *and other clinical laboratory and pathology patient test reports.*” (Italics indicate new text.)
- **First main bullet, third sub-bullet:** “*Race/Ethnicity*” (Italics indicate new text.)
- **Second main bullet, second sub-bullet:** “*Relevant non-genetic laboratory test and pathology data*” and add two additional sub-bullets as follows: “*Other clinical data such as radiology study results*” and “*Environmental exposure data*” (Italics indicate new text.)

- **Third main bullet, second sub-bullet:** “Ages of *disease onset and/or* death of various family members” and add additional sub-bullet “*Environmental exposure data*” (Italics indicate new text.)
- **Fourth main bullet, first sub-bullet:** “Prior genetic/genomic *laboratory test* results” (Italics indicate new text.)

Appendix A: Glossary

The CAP recommends the following clarifying additions:

- **“Clinicians:** Healthcare providers with patient care responsibilities, including physicians, *such as primary care physicians, family practice physicians,* medical geneticists, *pathologists, and other specialty physicians,* advanced practice nurses, pharmacists, physician assistants, nurses, genetic counselors, and other credentialed personnel involved in treating patients.” (Italics indicate new text.)
- **“Decision Support:** A discipline that enables improved analysis and conclusions based on related information, *published research studies,* algorithms, or other resources.” (Italics indicate new text.) “Published research studies” is different than the current “recent research” because it is necessary for the science to be peer-reviewed at a minimum, but likely the evidence for clinical decision support needs to be evidence-based with demonstration of clinical validity and utility.
- **“Electronic Health Record (EHR):** The electronic health record is a longitudinal electronic record of patient health information generated in one of more encounters in any care delivery setting. This information may include patient demographics, progress notes, problems, medications, vital signs, past medical history, immunizations, *laboratory and pathology* information, and radiology reports.” (Italics indicate new text.)
- **“Genetic Specialists:** “*Physician specialists, including medical geneticists and pathologists, as well as* genetic counselors and *other* clinicians who participate in evaluation, diagnostic planning, and genetic/genomic test ordering and result interpretation activities.” (Italics indicate new text.)
- **“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, DNA, RNA or proteins.*” (Italics indicate new text.) Defining a “genetic test” has been quite controversial for a long time. The first sentence of the definition is broad, but the second sentence in the draft does not contribute to the definition because it is not clear what “molecular” means. The new text in the second sentence is suggested to clarify the types of molecules that can be analyzed.

- **“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.” (Italics indicate new text.) The laboratory data is only useful if it gets out of the LIS to other information systems.
- **“Laboratory Organizations:** Advocacy/professional organizations or societies such as the College of American Pathologists (CAP) or the National Committee for Clinical Laboratory Standards (NCCL) which are concerned with the appropriate use of laboratory technology and *interpretation of laboratory* information in clinical medicine.” (Italics indicate new text.)
- **“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.” (Italics indicate new text.)
- **“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.*” (Italics indicate new text.) This new definition is parallel to the definition of “Medical Geneticist.”
- **“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 testing laboratories which perform *genetic/genomic and other* laboratory tests ordered by genetic specialists and clinicians to assess the genetic status of patients.” (Italics indicate new text.)