ATTACHMENT 1

Genetic Testing Registry Data Fields

Introduction

This document delineates the proposed fields that will be used in the Genetic Testing Registry (GTR) to aggregate relevant data on genetic tests. This registry will provide for:

- Collection of data important in the use and evaluation of available genetic tests, including information on analytical validity, clinical validity, and clinical utility
- Dissemination of useful information for health care providers, consumers, payers, researchers
- Development of a user-friendly genetic testing database for reporting, exchanging, and updating test information

The GTR is designed to collect adequate information on genetic tests while taking into consideration the reporting burden on the laboratories, existing resources, standards and practices, and the practicalities of measuring some test attributes. The information that will be collected is divided into three top-level fields: laboratory, personnel, and test. There are approximately 31 minimal data fields; these fields represent the minimum amount of information that must be submitted in order to register a test in GTR. There are approximately 85 fields that are either recommended or optional; these fields collect relevant information that may not be available for some tests. For the sake of simplicity, elsewhere in the PRA the "recommended" and "optional" fields are grouped together and identified as "optional" fields, as neither set needs to be provided in order to participate in GTR. The difference between recommended and optional fields is in display: recommended fields that are not filled in by the submitter will display on the GTR website with the words "Not Provided," while optional fields that are not filled in by the submitter will not appear on the website. There are also approximately 24 fields that GTR can automatically complete on behalf of the submitter.

Each data element includes a definition of the element, the planned implementation, and references to outside resources that have suggested the data element or currently request it from laboratories. The following reference short names are listed with the corresponding complete citation:

AMP – Association for Molecular Pathology Survey and Response submitted to the Genetic Testing Registry Request for Information (RFI). Accessible at: http://oba.od.nih.gov/oba/gtr/comments/Association_for_Molecular_Pathology.pdf

McKesson – McKesson Advanced Diagnostics Management: Response submitted to the Genetic Testing Registry RFI and meetings with GTR staff members. Accessible at: http://oba.od.nih.gov/oba/comments/McKesson_Advanced_Diagnostic_Management.pdf

Javitt et al - Javitt G, Katsanis S, Scott J, Hudson K. Developing the blueprint for a genetic testing registry. Public Health Genomics. 2010; 13(2):95-105. PubMed ID: 19556748

CAP – College of American Pathologists Molecular Checklist. Accessible at http://www.cap.org

GA – Genetic Alliance: Zonno K, Terry S. A call for action from Genetic Alliance: Registry of genetic tests – A critical stepping stone to improving the genetic testing system. Genetic Testing and Molecular Biomarkers, 13:153-154, 2009.

MMWR – Centers for Disease Control and Prevention (CDC): Morbidity and Mortality Weekly Report (MMWR): Good Practices for Molecular Genetic Testing for Heritable Diseases and Conditions. (2009). Accessible at: www.cdc.gov/mmwr/preview/mmwrhtml/rr5806a1.html

eDOS – HL7 Version 2 Implementation Guide: Laboratory Test Compendium Framework, Release 1 (eDOS). 2010 Health Level Seven, International. Accessible at http://www.hl7.org/implement/standards/index.cfm

HL7 LOINC – HL7 Version 2 Implementation Guide: Clinical Genomics; Fully LOINC-Qualified Genetic Variation Model Release 1. 2009 Health Level Seven, International. Accessible at http://www.hl7.org/implement/standards/index.cfm

Visual Hierarchy Used in this Document

Data elements in this document are indicated by the following hierarchy:

Header (in most cases this is NOT a data field)

DATA FIELD OR SUB-GROUP OF DATA FIELDS

DATA FIELD

TYPES OF INFORMATION THAT SHOULD BE USED TO DEFINE THE UPPER LEVEL DATA FIELD

Data Fields

Laboratory Information:

It is expected that Laboratory Information will be provided and updated with a different time cycle than Test Information. Laboratory Information may be updated independently of Test Information and vice versa. For a CLIA laboratory, it is expected that some fields will correspond to elements in the CLIA database, as documented below.

References: 77% of AMP respondents are able to provide laboratory information.

NAME

In this section, the submitter can provide all information that identifies the submitting entity (in most cases laboratory is used in this document) by populating the fields below.

NAME OF LABORATORY: TEXT FIELD – MANUAL ENTRY – MINIMAL

Complete name of the laboratory performing the test.

References: CLIA [fieldname]
McKesson
Javitt et al

NAME OF LABORATORY ACRONYMS: TEXT FIELD – MANUAL ENTRY – OPTIONAL

Acronym(s) or short name(s) that identifies the laboratory. Field allows entering multiple acronyms. An auto-complete list of existing institutional acronyms will be included.

NAME OF INSTITUTION: TEXT FIELD – MANUAL ENTRY – OPTIONAL

Complete name of the institution to which the laboratory belong (e.g., hospital, university). It may be the same as "Name of Laboratory" for independent laboratories. This field may be auto-completed for existing institution names in GTR.

References: Javitt et al

INSTITUTION ACRONYMS: TEXT FIELD – MANUAL ENTRY – OPTIONAL

Acronym(s) or short name(s) that identify the institution to which the laboratory belong. Field allows entering multiple acronyms. An auto-complete list of existing institutional acronyms will be included.

NAME OF DEPARTMENT: TEXT FIELD – MANUAL ENTRY – OPTIONAL

This is the name of the department used in the Facility address.

References: Javitt et al

FACILITY ADDRESS

The information collected is shown in the fields below.

FACILITY ADDRESS CAN BE MADE PUBLIC – YES/NO CHECKBOX – MINIMAL DEFAULT: YES.FACILITY ADDRESS LINE 1: TEXT FIELD – MANUAL ENTRY - OPTIONAL

Street address of the facility (e.g., building number and street name).

References: Javitt et al
McKesson

FACILITY ADDRESS LINE 2: TEXT FIELD – MANUAL ENTRY - OPTIONAL

This is the subdivision number of the facility address if applicable (e.g., room or suite#).

References: Javitt et al
McKesson

FACILITY CITY: TEXT FIELD – MANUAL ENTRY – MINIMAL - VALIDATION

City will be validated along with state against the postal code entered.

FACILITY STATE/PROVINCE: PULL-DOWN MENU – MANUAL ENTRY – MINIMAL - VALIDATION

State will be validated along with city against postal code entered.

FACILITY POSTAL CODE: TEXT FIELD – MANUAL ENTRY – MINIMAL – VALIDATION

Postal code will be validated against state/city combination entered.

FACILITY COUNTRY: PULL-DOWN MENU – MINIMAL

Country used in the Facility address. United States will be set as default.

FACILITY ADDRESS OTHER: TEXT FIELD – MANUAL ENTRY – OPTIONAL

Submitter can specify any other Facility address as applicable.

References: Javitt et al
McKesson

FACILITY PHONE NUMBER: TEXT FIELD – MANUAL ENTRY - MINIMAL

General phone number for the laboratory. It could be specified as: Country name – Country-specific area code (set parameters) – phone number; field will be validated automatically if possible.

References: Javitt et al
McKesson

FACILITY FAX NUMBER: TEXT FIELD – MANUAL ENTRY - OPTIONAL

Existing general fax number for the laboratory to be made available on GTR website.

References: Javitt et al
McKesson

FACILITY EMAIL ADDRESS: TEXT FIELD – MANUAL ENTRY - MINIMAL

Existing general email address for the laboratory to be available to on GTR website.

References: Javitt et al
McKesson

LABORATORY MAIN WEBSITE URL: TEXT FIELD – MANUAL ENTRY - OPTIONAL

URL for the laboratory's website.

References: Javitt et al

LABORATORY TYPES OF SERVICE: PICK FROM LIST, PLUS ABILITY TO SUGGEST NEW - OPTIONAL

List of all services offered by the laboratory with the ability to multi-select. Services selected in this field represent all services offered by the laboratory and are not test-specific. The laboratory can add any new service not named in the list provided. An initial list is below.

- Genetic counseling
- Result interpretation
- DNA Banking
- RNA Banking
- Cord Blood Banking
- Tissue Banking
- Data Storage and Backup
- Confirmation of research findings
- Preimplantation Genetic Diagnosis (PGD)
- Custom Prenatal Testing

- Custom Deletion/Duplication Testing
- Custom Sequence Analysis
- Custom Balanced Chromosome Rearrangement Studies
- Marker Chromosome Identification
- Uniparental Disomy (UPD) Testing
- Identity Testing
- X-Chromosome Inactivation Studies
- Specimen Source Identification
- Other, specify_____

References: CAP MOL.05075
Javitt et al

LABORATORY SERVICES ORDER CODE: TEXT FIELD – MANUAL ENTRY – OPTIONAL

Order code that the laboratory uses for the particular service(s) selected in the field above. One order code field per laboratory service selected.

LABORATORY AFFILIATIONS – TEXT FIELD+URL – OPTIONAL

In this field a laboratory can identify if it is linked to a larger health care system and/or clinical unit that cares for individuals with a given disorder. Submitters can identify parent companies of fully owned subsidiaries in this field.

GENETESTS LABORATORY ID – TEXT FIELD – OPTIONAL

This field allows the submitter to provide its current GeneTests institution ID for identification purposes. NCBI may be able to auto-provide this in some cases. This data field would be shown to submitter only, not publically displayed.

LABORATORY PARTICIPATION IN EXTERNAL PROGRAMS – SELECT FROM LIST – OPTIONAL

Submitter can specify if the laboratory participates in external programs such as in the fields below. Submitter can select multiple programs. An initial list is below.

- ISCA Consortium (International Standards for Cytogenomic Arrays)
- Locus-specific Databases

STANDARDIZATION PROGRAMS

Does the laboratory participate in standardization programs?

DATA EXCHANGE PROGRAMS

Does the laboratory participate in data exchange programs?

Personnel Information:

In this section, the laboratory will provide information about its staff member(s) and will be able to make choices regarding if and how each person entered will be displayed in the GTR website or only stored in the database. The laboratory can enter multiple Persons.

At least one Person must be identified as the Laboratory Director for each laboratory.

The personnel information is provided only once for the laboratory and can be disseminated to all the tests the laboratory registers in GTR. Furthermore, specific contacts for specific tests can be linked from the test, rather than re-entered.

References: CAP MOL.40000 “Director Qualifications”; CAP MOL.40100 “Personnel – Technical Operations”; CAP MOL.40150 “Technologist Qualifications”
Javitt et al
McKesson

PERSON NAME: TEXT FIELD – MANUAL ENTRY – MINIMAL (FOR AT LEAST ONE PERSON)

The complete name of a person. This field will be used multiple times – once for each person entered.

PRIMARY LABORATORY CONTACT – YES/NO CHECKBOX – MINIMAL IF APPLICABLE

Is this person the primary laboratory contact for GTR staff? If checked, this person would receive communication from GTR staff if and when appropriate (e.g., annual update messages, test questions).

LABORATORY DIRECTOR – YES/NO CHECKBOX – MINIMAL IF APPLICABLE

Is this person a laboratory director?

DISPLAY PERSON ON GTR WEBSITE – YES/NO CHECKBOX – MINIMAL

Should this person be displayed in the GTR website? A person can be displayed in the GTR website but choose that their contact information be kept private.

PERSON ID – TEXT FIELD – HIDDEN (REQUIRED FOR BULK LOADS)

Unique key for the Person. It is only for laboratories providing data electronically to ensure that they can link a test uniquely to a person in bulk upload of data.

PERSON TITLE – PICK FROM LIST, PLUS ABILITY TO SUGGEST NEW – OPTIONAL

Person’s professional title in the laboratory as provided by the person. There will be a list of the most common titles to facilitate quick entry and an “Other” field to allow manual entry.
Suggested list:

- Lab Director
- Lab Associate Director
- Medical Director
- Genetic Counselor
- Nurse
- Research Nurse
- Administrator
- Staff

PERSON ACADEMIC DEGREE(S) – SELECT FROM LIST – OPTIONAL

The academic degree(s) the person holds. Submitter can select multiple academic degrees and whether each degree should be displayed after name on website. An initial list is below.

- M.D.
- D.O.
- D.P.M.
- Ph.D.
- M.S.
- M.A.
- B.S.

PERSON GENETIC CERTIFICATIONS – SELECT FROM LIST – OPTIONAL

The certification(s) the person holds from genetic colleges, boards, associations, and any other relevant organization or institution. An initial list is below.

- FACMG
- CGC

References: 39% of AMP respondents are able to provide this information.

PERSON OTHER CERTIFICATIONS – SELECT FROM LIST – OPTIONAL

The certification(s) the person holds from all colleges, boards, associations, or any other relevant organization or institution not named in the field above. An initial list is below.

- ABP
- FAAP
- ABIM
- FACP
- FACOG

PUBLICALLY DISPLAYED CREDENTIALS – AUTO-POPULATE BASED ON CERTIFICATION AND DEGREE

List of all credentials named on the prior 3 fields that will be publically displayed. Submitter may select if each credential should be displayed after name on website.

PERSON DATABASE PERMISSIONS – SELECT FROM LIST – MINIMAL

Role of the person in relation to GTR privileges/permissions. Permissions to view submitted information, add, edit or delete information, or all of the above. The suggested list is below.

- View Only
- Edit Only
- Add Only
- Delete Only
- All

PERSON PUBLIC PHONE NUMBER – TEXT FIELD; MANUAL ENTRY – OPTIONAL (MINIMAL IF TEST-SPECIFIC CONTACT)

Phone number to be made available for the public to contact the person. If the person is being entered as a test-specific contact, then a public phone number is required; for all other laboratory personnel, a public phone number is optional.

PERSON PRIVATE PHONE NUMBER – TEXT FIELD; MANUAL ENTRY – OPTIONAL (MINIMAL FOR AT LEAST ONE PERSON)

Person's direct phone number that will not be made public but may be used by GTR staff as needed. GTR requires the contact information of at least one laboratory staff member for communication about submission and maintenance of records. This number can be the same as the public phone number.

PERSON PUBLIC FAX NUMBER – TEXT FIELD; MANUAL ENTRY - OPTIONAL

Fax number to be made available for the public to contact the person.

PERSON PRIVATE FAX NUMBER – TEXT FIELD; MANUAL ENTRY - OPTIONAL

Person's fax number that will not be made public but may be used by GTR staff to contact as needed.

PERSON PUBLIC EMAIL ADDRESS – TEXT FIELD; MANUAL ENTRY – OPTIONAL (MINIMAL IF TEST-SPECIFIC CONTACT)

Email address to be made available for the public to contact the person. If the person is being entered as a test-specific contact, then a public email address is required; for all other laboratory personnel, a public phone number is optional.

PERSON PRIVATE EMAIL ADDRESS – TEXT FIELD; MANUAL ENTRY – OPTIONAL (MINIMAL FOR AT LEAST ONE PERSON)

The email account attached to NCBI login system by default. This field is optional for personnel being listed for the laboratory that are not involved in the submission or maintenance of information in the GTR.

PERSON CONTACT PUBLIC COMMENT – TEXT FIELD; MANUAL ENTRY – OPTIONAL

Comment to be displayed with public contact information for this person.

Licensure and Accreditation – Laboratory:

In this section, the submitter can provide information related to the different regulations that govern the laboratory such as Clinical Laboratory Improvement Amendments (CLIA) certifications and state licenses as shown in the fields below.

References: GA
MMWR

CLIA CERTIFICATION NUMBER – TEXT FIELD; MANUAL ENTRY – OPTIONAL (MINIMAL FOR U.S. LABORATORIES PROVIDING CLINICAL TESTS)

Certification number assigned by the Clinical Laboratory Improvement Amendments (CLIA) program to the laboratory. This field is mandatory for laboratories listing "clinical" tests. International laboratories and those registering research tests are not expected to have CLIA certification.

References: Javitt et al
McKesson
eDOS MSH-3 "Sending Application (CLIA ID sending Lab)"
MMWR

CLIA EXPIRATION DATE (MM/DD/YYYY) – TEXT FIELD; MANUAL ENTRY – OPTIONAL (MINIMAL FOR U.S. LABORATORIES PROVIDING CLINICAL TESTS)

Expiration date of the current CLIA certification for the laboratory. Some data will be valid for month and year only. This field is required for U.S. laboratories providing clinical tests that have entered a CLIA certification number.

References: Javitt et al
McKesson

STATE LICENSE – PULL-DOWN MENU – OPTIONAL

Name of the state under which the laboratory is licensed to practice. Submitter may select multiple.

STATE LICENSE # – TEXT FIELD; MANUAL ENTRY – OPTIONAL

License number issued by the state to the laboratory. It is mandatory for those with license numbers available.

STATE LICENSE EXPIRATION DATE (MM/DD/YYYY) – TEXT FIELD; MANUAL ENTRY – OPTIONAL

Expiration date on the state license. Mandatory for those with expiration dates.

OTHER CERTIFICATIONS/LICENSES – PULL-DOWN MENU – OPTIONAL

Name of all other certifications or licenses that the laboratories holds not named in the fields above. This field can include federal and international certifications/licenses such as ISO. Submitter may select multiple.

References: 69% of AMP respondents are able to provide this information
McKesson
Javitt et al
MMWR

OTHER CERTIFICATION/LICENSE # – TEXT FIELD; MANUAL ENTRY – OPTIONAL

This field is required for those laboratories that have license numbers available.

OTHER CERTIFICATION EXPIRATION DATE (MM/DD/YYYY) – TEXT FIELD; MANUAL ENTRY – OPTIONAL

This field is required for those laboratories that have certification/licenses with expiration dates.

Default Laboratory Values for Test Information:

Information stored in the default section can be copied to other fields and to each test offered by the laboratory. The submitter has the option to update all default values at once and override them as appropriate.

The default section is optional and has been designed for the purpose of saving submitters from entering the same information multiple times. The fields below can be provided in the default section if the submitter wants that answer to appear for all of their tests. The same fields appear in their appropriate sections both in this document and in the electronic submitter forms.

- Default How to Order: Text + URL – Manual Entry – Optional
- Default Specimen Source – Pull Down List – Optional
- Default Test Contact Policy – checkbox – Optional
- Default Test Orderable By – Pull-Down List – Optional
- Default Sample Negative Report – Optional
- Default Sample Positive Report – Optional
- Default Variants of Unknown Significance (VUS) policy and interpretation
    - What is the Protocol for Interpreting a Variation as a VUS? – Text Field – Manual Entry – Recommended
    - What Software is Used to Interpret Novel Variations? – Text Field – Manual Entry – Optional
    - What Is the Laboratory's Policy on Reporting Novel Variations? – Text Field – Manual Entry – Recommended
    - Are Family Members Who Have Defined Clinical Status Recruited to Assess Significance of VUS Without Charge? – Yes/No Checkbox with comments – Recommended
- Default VUS Report – Optional
- Default Will the Laboratory Re-contact the Ordering Physician if Variant Interpretation Changes? – Yes/No checkbox with comments – Optional

Test Information:

Each Test is a specific, orderable test from a particular laboratory, and receives a unique GTR accession number. The same or similar test performed by different laboratories gets a different accession number. Thus, a laboratory is free to define an orderable test exactly as they represent it in their catalog.

GTR ACCESSION ID (AUTO ASSIGNED BY NCBI WITH VERSIONING) – AUTOMATICALLY PROVIDED

A GTR accession ID has the format GTR00000001.1, a leading prefix "GTR" followed by 8 digits, a period, then 1 or more digits representing the version. When a laboratory updates a test, the accession stays the same, but the version increments. GTR accessions and versions are issued and controlled by NCBI. Any changes to the test information will result in a version change. Changes to laboratory and personnel information will not result in a version change. Access to archived versions of tests will be provided to submitters and users.

References: eDOS OM1-7 "Other Service/Test/Observation/IDs for the Observation"
McKesson
HL7

DATE LAST TOUCHED (AUTO PROVIDED BY NCBI) – AUTOMATICALLY PROVIDED

When a laboratory updates test-specific data fields, the date (format = MM-DD-YYYY) is recorded. This field is associated with the GTR Accession ID, where the accession stays the same but the version increments. The date last touched will update with the test versions. Any changes to the test information will result in a version change and subsequently update the date last touched. Changes to laboratory and personnel information will not result in a version change and will not update the date last touched for the test entry.

NAME OF TEST:

Name of test should include one or more of the following subfields.

References: 87% of AMP respondents are able to provide this information.

LABORATORY TEST NAME – TEXT FIELD; MANUAL ENTRY – MINIMAL

Test name that will appear as the default title on the GTR test detail page. It should be the test name the laboratory wishes to be commonly associated with the test. By default this name will be displayed as the test name.

References: eDOS OM1- 8 and 51 “Other Names (recognized by the producer for the observation)”
McKesson
MMWR

LABORATORY TEST SHORT NAME – TEXT FIELD; MANUAL ENTRY – OPTIONAL

Submitter’s short name or mnemonic for the test. This is a name that may be used in space limited reports such as lists and tables.

References: e-DOS OM1-10 “Preferred Short name or Mnemonic for the Observation”
MMWR

MANUFACTURER TEST NAME – TEXT FIELD; MANUAL ENTRY – OPTIONAL

Common commercial test name (e.g.,. OvaSure, FDA kit name). Manufacturer test could be an FDA approved test, a kit, or some other manufacturer test. A test may point (link) to another test with the ability to override certain values.

OTHER NAMES – TEXT FIELD; MANUAL ENTRY + PULL-DOWN LIST – OPTIONAL

The submitter can enter other synonyms and aliases by which the test can be searched. They can classify the type of name: archived, synonym, keyword, and so on.

References: eDOS OM1-11 “Preferred Long Name for the Observation”

TEST DEVELOPMENT – PULL-DOWN LIST – RECOMMENDED

Submitter can specify how the test was developed: whether the test is laboratory developed, FDA approved or cleared, an externally manufactured kit, a modified FDA-cleared/approved test, or combination as exemplified in the proposed list below. Please note that reflex testing is not included in this field.

- Laboratory Developed Test (no manufacturer test name)
- FDA-cleared/approved test (has FDA test name)

- Manufactured (research use only; not FDA-reviewed)
- Modified FDA (FDA-cleared/approved test, but with laboratory modifications/field changes)
- Combination (could include reflex & panels doing multiple tests) (list of tests – LDTs, FDA)

LABORATORY UNIQUE CODE – TEXT FIELD; MANUAL ENTRY – OPTIONAL (USED ONLY FOR BULK UPLOADS)

For tests provided in bulk electronically, the Laboratory Unique Code must be provided. This code is unique for the test from that laboratory. NCBI will use this code to determine if the laboratory is providing a new test or an update to an existing test, so it is critical that the same code be submitted for the same test.

References: e-DOS OM1-2 “Producer’s Service/Test/Observation ID”.

TEST-SPECIFIC LABORATORY SERVICES – TEXT FIELD; MANUAL ENTRY – OPTIONAL

Placeholder for other fields under “Test Information” that may become applicable to Services entries.

HOW TO ORDER – TEXT + URL; MANUAL ENTRY – RECOMMENDED

Description of the test ordering procedure and the laboratory website URL for more details.

References: CAP MOL.32300 and MOL.32350 “Requisition Information”
eDOS OM1-12 “Orderability”

SPECIMEN SOURCE – PULL-DOWN LIST + URL LINK; MANUAL ENTRY – RECOMMENDED

Pull-down list includes only the general specimen type required (e.g., whole blood, frozen tissue, fresh tissue, sputum). Detailed specimen requirements for the ordering physician should be found at the supplied URL. See ‘Default Specimen Source” for a sample list. Multiple entries allowed.

References: 73% of AMP respondents are able to provide this information.
CAP MOL.33050 “Specimen Collection/Handling Requirements”
eDOS OM4-6 “Specimen”
McKesson
Javitt et al
HL7

Submitter can enter one or more default specimen source or type from pre-determined list that will be displayed with all tests. Submitter can select all that apply. An initial list is below.

- Peripheral (whole) blood
- Buccal swab
- Saliva
- Amniocytes
- Amniotic fluid
- Bone marrow
- Cell culture
- Chorionic villi
- Cord blood
- Cystic hygroma fluid
- Dried blood spot (DBS) card

- Fetal blood
- Fresh tissue
- Fibroblasts
- Frozen tissue
- Paraffin block
- Product of conception (POC)
- Serum
- Skin
- Sputum
- Urine
- White blood cell prep
- Other, specify _____

TEST-SPECIFIC CONTACT PERSON – PULL-DOWN LIST – RECOMMENDED

In the interactive forms based update, the submitter may select from one of the personnel supplied earlier. For direct electronic submission of data, this field must be supplied as either a unique name matching the personnel list or a personnel ID previously supplied.

If no test-specific contact is given, the laboratory's general contact information will display by default.

References: eDOS OM1-17 "Telephone Number of Section"
MMWR

TEST-SPECIFIC CONTACT POLICY – CHECK BOX – RECOMMENDED

The overall policy of the laboratory regarding who (patients vs. health care providers) and when (pre-test/post-test/anytime) can contact the laboratory. The suggested options are:

- Pre-test email/phone consultation regarding genetic test results and interpretation is provided to patients/families.
- Post-test email/phone consultation regarding genetic test results and interpretation is provided to patients/families.
- Laboratory can only accept contact from health care providers. Patients/families are encouraged to discuss genetic testing options with their health care provider.

This check box can be auto selected by the default laboratory contact policy. Submitters can change the contact policy here and override the default settings.

INFORMED CONSENT REQUIRED – YES/NO CHECK BOX – RECOMMENDED (DEFAULT YES)

Submitters can choose to let users know if a test requires informed consent prior to testing. As default, all tests for all laboratories will likely need a disclaimer that informed consent is determined by the ordering physician's state laws.

Informed consent required:	Yes	No	Informed consent requirements are determined based on applicable state law

References: MMWR

GENETIC COUNSELING REQUIRED (PRE-TEST AND/OR POST-TEST) – YES/NO CHECKBOX – RECOMMENDED (DEFAULT YES)

Submitters can choose to let users know if a test requires genetic counseling and if so, whether prior to testing or prior to the release of test results.

References: MMWR

TESTING STRATEGY – TEXT + CITATIONS; MANUAL ENTRY – RECOMMENDED

Submitters can describe the suggested sequence of ordering tests, discuss reflex testing, and related issues. This field is for recommendations on how to order the different tests in sequence of relevance to the patient being tested. This field should not include discussion of methodology or test procedural protocols. Laboratories can describe whether a test has a required reflex test or a reflex mechanism. Each test component should be described. If a test is ordered, additional tests may be performed as necessary under certain circumstances based on initial results and that should be described in this field.

References: eDOS OM1-34 “Reflex Tests/Observations”

LABORATORY TEST ORDER CODE – TEXT FIELD; MANUAL ENTRY – RECOMMENDED

Laboratory’s order or catalog code for the test (i,e., the order code to put in the requisition to order the test from the laboratory).

References: eDOS OM1-2“Producer’s Service/Test/Observation ID”
MMWR

TEST CODES – URL – RECOMMENDED

Submitters can provide a URL for information on codes including LOINC, ICD-9, and ICD-10.

References: eDOS OM1-7 “Other Service/Test/Observation IDs for the Observation”
McKesson
HL7

URL FOR THE TEST – TEXT FIELD; MANUAL ENTRY – RECOMMENDED

Submitter-provided link to their website for test-specific information.

References: 26% of AMP respondents are able to provide this information.
Javitt et al

Availability:

This section identifies the location where different aspects of the test are performed. The intent is to increase transparency of where the test is performed. External means any laboratory/facility that does not belong to the reporting laboratory. For example, a test performed at an outside facility owned by the same company would be considered to be performed “externally.” Submitters can enter details and provide appropriate information for clarification of their entry as needed.

References: McKesson

TEST PERFORMED IN-HOUSE – CHECK BOX + TEXT – MINIMAL

Identification of where all parts of the test are performed. Suggested option list is below. Submitter can check multiple boxes and provide information in a single text field.

- Entire test performed in-house
- Entire test performed externally
- Specimen preparation performed in-house
- Specimen preparation performed externally
- Wet lab work performed in-house
- Wet lab work performed externally
- Interpretation performed in-house
- Interpretation performed externally
- Report generated in-house
- Report generated externally

References: eDOS OM1-27 "Outside Site(s) where Observation may be Performed" - optional
Javitt et al

IF TEST OR PART(S) OF TEST PERFORMED EXTERNALLY – CHECKBOX – MINIMAL

The following two questions will be required only if the test or any part of the test is performed externally. If submitter chooses that the entire or a portion of the test is performed externally, they may show whether they are authorized to enter the external collaborator's details of the test and whether the external collaborator had the ability to review the information for accuracy. Submitters will not be required to report the name of external collaborator/lab.

I AM AUTHORIZED TO ENTER DETAILS OF TEST – CHECKBOX YES/NO – (MINIMAL IF TEST OR PART(S) OF TEST PERFORMED EXTERNALLY)

Required if any portion of the test is performed externally.

THIS ENTRY HAS BEEN REVIEWED BY THE EXTERNAL COLLABORATOR(S) FOR ACCURACY – CHECKBOX YES/NO – (MINIMAL IF TEST OR PART(S) OF TEST PERFORMED EXTERNALLY)

Required if any portion of the test is performed externally. In the case that more than three facilities are involved in the testing process, if one facility has not reviewed the test and the other has, "No" should be selected. For example, if laboratory A is the reporting laboratory, wet lab work is performed in laboratory B (reviewed entry), interpretation in company C (did not review entry), the answer to this question is "No."

Accessibility:

This section identifies how the test can be ordered as described in the field below.

References: GA
MMWR

TEST ORDERABLE BY – PULL-DOWN LIST – OPTIONAL

Identification of who can order the test from this laboratory (e.g., specify Health Care Providers: Licensed physician, PA, RN, NP, GC). Submitter can select multiple choices. An initial list is below.

- Health Care Provider

- Public Health Mandate
- Out-of-State Patients
- In-State Patients
- Licensed Physician
- Physician Assistant
- Registered Nurse
- Genetic Counselor

Reporting of Test Results:

This section contains information on how the laboratory reports test results and clinical interpretation back to the ordering individual and lists the responsibilities the laboratory assumes for reporting results and providing clinical interpretations.

References: CAP MOL.35942 “Result Reporting”
eDOS OM1-32 “Interpretation of Observations”
McKesson
MMWR
HL7-LOINC 51969-4 “Genetic Analysis Summary Report”

SAMPLE NEGATIVE REPORT – UPLOAD DOCUMENT; MANUAL ENTRY – RECOMMENDED

Submitter can upload a sample negative report for the corresponding test (auto-populated if information is entered in the default section).

SAMPLE POSITIVE REPORT – UPLOAD DOCUMENT; MANUAL ENTRY – RECOMMENDED

Submitter can upload a sample positive report for the corresponding test (auto-populated if information entered in the default section).

VARIANTS OF UNKNOWN SIGNIFICANCE (VUS) POLICY AND INTERPRETATION

Submitter can enter information on how Variants of Unknown Significance (VUS) are handled in the laboratory by supplying the information in the subfields below.

WHAT IS THE PROTOCOL FOR INTERPRETING A VARIATION AS A VUS? – TEXT FIELD; MANUAL ENTRY – RECOMMENDED

Description of how the laboratory handles Variants of Unknown Significance.

WHAT SOFTWARE IS USED TO INTERPRET NOVEL VARIATIONS? – TEXT FIELD; MANUAL ENTRY – OPTIONAL

Examples of software applications for medical molecular genetics interpretation include: Melina II, MEME Suite, VISTACartagenia Bench, Alamut, SIFT, PolyPhen, Align-GVGD, GeneSplicer, laboratory proprietary internal software.

WHAT IS THE LABORATORY’S POLICY ON REPORTING NOVEL VARIATIONS? – TEXT FIELD; MANUAL ENTRY – RECOMMENDED

Description of how the laboratory reports novel variations, which could include who gets contacted and how (e.g., person ordering the test will be contacted via telephone as soon as VUS is identified).

ARE FAMILY MEMBERS WHO HAVE DEFINED CLINICAL STATUS RECRUITED TO ASSESS SIGNIFICANCE OF VUS WITHOUT CHARGE? – YES/NO CHECKBOX WITH COMMENTS – RECOMMENDED

Will the laboratory offer the test to family members free of charge?

SAMPLE VUS REPORT – UPLOAD DOCUMENT; MANUAL ENTRY – OPTIONAL

Submitter can upload a sample VUS report for this test (auto-populated if information entered in the default section).

WILL THE LABORATORY RE-CONTACT THE ORDERING PHYSICIAN IF VARIANT INTERPRETATION CHANGES? – YES/NO CHECKBOX WITH COMMENTS – RECOMMENDED

Description of how the laboratory deals with ongoing interpretation of genetic tests results after the initial report.

RESEARCH

The submitter can enter information on the procedures after-processing the sample by supplying the information in the subfields below.

IS RESEARCH PERFORMED AFTER CLINICAL TESTING IS COMPLETE? – TEXT FIELD; MANUAL ENTRY – RECOMMENDED

After clinical testing is complete, does the laboratory perform any research testing using the submitted specimen?

Indications for Use:

This section describes the reasoning for performing the test. Information is provided in the fields below by the submitter and parts are automatically filled with information from NCBI's ClinVar and/or OMIM databases.

References: CAP MOL.30670 "Clinical Indication/Clinical Utility"
MMWR

PURPOSE OF THE TEST – PULL-DOWN MENU; MULTI-SELECT – MINIMAL

Purpose(s) or indication(s) for use of the test. An initial list is are below and submitter can select multiple purposes.

- Diagnosis
- Screening
- Drug Response
- Risk Assessment
- Pre-symptomatic
- Mutation Confirmation (family specific or research results, etc)
- Pre-implantation genetic diagnosis

References: 61% of AMP respondents are able to provide this information.
McKesson

Javitt et al
MMWR

CLINICAL OR RESEARCH TEST OR BOTH – CHECK BOX – MINIMAL

Submitters can indicate whether the test is for clinical purposes or is a research test. Definitions/Rules for what qualifies as a clinical test must be determined. Clinical tests can only be provided by CLIA certified laboratories.

CONDITION(S) FOR WHICH TEST IS OFFERED – PICK FROM LIST PLUS ABILITY TO SUGGEST NEW – MINIMAL

Name of the disease/syndrome/drug response/etc. for which the test can be ordered.

References: Javitt et al
HL7 LOINC 51963-7 “Medication Assessed”
HL7-LOINC 51967-8 “Genetic Disease Assessed”
HL7-LOINC 53577-3 “Reason for Study, Additional Note”

LAB-DISPLAYED DISEASE NAME – MANUAL ENTRY – OPTIONAL

The submitter can provide the disease name they want associated with the test if the disease name is different from the one automatically provided by GTR.

DISEASE IDENTIFIER(S) – AUTOMATICALLY PROVIDED

NCBI will provide the SNOMED CT name and identifier as available.

References: McKesson

DISEASE SYNONYM(S) – AUTOMATICALLY PROVIDED WITH ABILITY TO SUGGEST NEW

The submitter can provide the disease synonym they want associated with the test if the disease synonym is different from the one automatically provided by GTR.

LAB-DISPLAYED DISEASE SYNONYM; MANUAL ENTRY – OPTIONAL

DISEASE PREFERRED ACRONYM – AUTOMATICALLY PROVIDED WITH ABILITY TO SUGGEST NEW
DISEASE ACRONYM(S) – AUTOMATICALLY PROVIDED WITH ABILITY TO SUGGEST NEW
LAB-DISPLAYED DISEASE ACRONYM – MANUAL ENTRY - OPTIONAL

The submitter can provide the disease acronym they want associated with the test if the disease acronym is different from the one automatically provided by GTR.

DISEASE TYPE – AUTOMATICALLY PROVIDED

This field names the type or category of the disease. An initial example list is below.

- Dysmorphology Syndrome

- Cancer Syndrome
- Neurology

DISEASE CLINICAL SUMMARY– AUTOMATICALLY PROVIDED
DISEASE CLINICAL FEATURE(S) – AUTOMATICALLY PROVIDED
DRUG INFORMATION – AUTOMATICALLY PROVIDED

For pharmacogenetic tests, additional information can be provided by NCBI in substitution for disease clinical summary and clinical features provided for other conditions.

MODE OF INHERITANCE – AUTOMATICALLY PROVIDED WITH ABILITY TO SUGGEST NEW

DISEASE MECHANISM – AUTOMATICALLY PROVIDED WITH ABILITY TO SUGGEST NEW

Identification of the disease mechanism (e.g., haploinsufficiency, imprinting, etc).

SIMILAR DISORDERS – AUTOMATICALLY PROVIDED WITH ABILITY TO SUGGEST NEW

This field contains the differential or a list of disorders similar to the one for which the test is used.

PREVALENCE – AUTOMATICALLY PROVIDED WITH ABILITY TO COMMENT OR ADD NEW CITATION – TEXT + CITATION + URL – OPTIONAL

The most current estimated number of cases of the disease in the population.

References: 34% of AMP respondents are able to provide this information.

DESCRIPTION OF THE TARGET POPULATION – TEXT + CITATIONS – RECOMMENDED

Explanation of which segment(s) of the population should be tested for this disease and why.

References: MMWR (recommended patient population)

Test Methodology:

This section contains technical information about the test as submitted by the laboratory.

METHOD CATEGORY– PULL-DOWN LIST – MINIMAL

Name of the general category the test belongs to. A list of examples is provided below.
- Method to detect nucleotide changes ≤5bp
- Method to detect deletions/duplications >5 and <250bp
- Method to detect deletions/duplications ≥250bp
- Method to detect enzyme/metabolite levels

References: 73% of AMP respondents are able to provide this information.

eDOS OM1-14 “Coded Representation of Method”
MMWR

PRIMARY TEST METHODOLOGY – PULL-DOWN LIST WITH ABILITY TO SUGGEST NEW – MINIMAL

Name of the test method used in the assay. A list of examples is provided below.
Examples:

- PCR-RFLP with Southern hybridization
- RT-PCR with gel analysis
- Trinucleotide repeat by PCR or Southern Blot
- Protein truncation
- Enzymatic levels
- Metabolite levels
- Gene expression profiling
- Comparative genomic hybridization
- GeneID
- Chromatin Immunoprecipitation on ChIP
- DamID
- SNP Detection
- Alternative splicing detection
- Fusion genes microarrays
- Tiling Arrays
- Other, specify_____

References: CAP MOL.30680 “Manufacturer Instructions”
CAP MOL.31705 “LDT Reporting”
CAP MOL.31935 “Modified FDA-Approved Assay”
HL7-LOINC 55233-1 “Genetic Analysis Master Panel”
HL7-LOINC 55232-3 “Genetic Analysis Summary Panel”
McKesson

PLATFORMS – LABORATORY-SPECIFIC PULL-DOWN LIST – RECOMMENDED

Information provided by the submitter, such as chips and arrays used in the test. Submitter will be able to select multiple items. Laboratories will be able to bulk upload platform information (e.g.,. their own array) and point to it.
Examples:

- Affymetrix GeneChip
- Agilent microarrays
- CodeLink Bioarray
- NimbleGen microarray
- Febit microarray
- Xeotron microarray
- Expression Array (Applied Biosystems)
- Spotted cDNA Array

References: CAP MOL.29290 “Reagent Data”
Javitt et al
HL7

INSTRUMENT(S) USED DURING TESTING – PULL-DOWN LIST + SUGGEST NEW – RECOMMENDED

Submitters can name the instruments used and point to them electronically. Submitter will be able to select multiple items.
Examples:

- Qiagen AutoPure LS
- Qiagen QIAcube
- Tecan Genesis Robotic Workstation 150
- PerkinElmer Victor3 1420 Multilabel Plate Reader
- Agilent 2100 Bioanalyzer
- Applied Biosystems 7900HT Sequence Detection System
- Applied Biosystems SOLiD v4 System Sequencer
- Applied Biosystems 9700 Thermal Cycler
- Covaris S2 Sonicator
- Roche LightCycler 480
- BioRad CFX96

References: eDOS OM1-13 “Identity of Instrument used to Perform this Study”
Javitt et al

DESCRIPTION OF TEST PROCEDURE/PROTOCOL – TEXT + CITATIONS – OPTIONAL

Summary of the methodology, which may include the description of the specific steps for each method used in the assay.

References: CAP MOL.34921 “Sequencing Assay Optimization”
eDOS OM1-41 “Description of Test Methods”

CONFIRMATION OF TEST RESULTS – TEXT FIELD; MANUAL ENTRY – RECOMMENDED

Submitters can provide further information about whether they confirm results, and how. Example: “Positive results are confirmed on a new DNA preparation using repeat sequence analysis”.

Analytes:

WHAT THE TEST MEASURES – PULL-DOWN LIST – MINIMAL

Category of the analyte being tested. A list of examples is provided below.

- Nucleotide Mutations
- Haplotypes
- Chromosome Rearrangements
- Full genome
- Enzymes
- Metabolites

References: HL7-LOINC 48006-1 “Amino Acid Change Type”
HL7-LOINC 48019-4 “DNA Sequence Variation Type”
Javitt et al
MMWR

GENE(S) BEING TESTED – TEXT FIELD; MANUAL ENTRY OR POSSIBLY PICK LIST – MINIMAL

Name of the gene that the test targets. When the gene name is identified, the gene symbol, synonym, location, family, and OMIM # will be provided automatically if available.

References: 87% of AMP respondents are able to provide this information.
CAP MOL.34914 “Gene Information”
Javitt et al
HL7-LOINC 48018-6 “HGNC Gene Identifier”
MMWR

GENE SYMBOL – AUTOMATICALLY PROVIDED

References: CAP MOL.36842 “Standard Nomenclature”
HL7-LOINC 48018-6 “HGNC Gene Identifier”

GENE SYNONYM(S) – AUTOMATICALLY PROVIDED WITH ABILITY TO SUGGEST NEW

References: HL7-LOINC 48008-7 “Allele Name”

GENE LOCATION – AUTOMATICALLY PROVIDED

References: HL7-LOINC 47999-8 “DNA Region Name”

GENE FAMILY– AUTOMATICALLY PROVIDED

Currently, this information is likely not available on most genes.

OMIM GENE # – AUTOMATICALLY PROVIDED
CHROMOSOMAL LOCATION BEING TESTED – TEXT FIELD; MANUAL ENTRY OR POSSIBLY AUTOMATICALLY PROVIDED – OPTIONAL (MINIMAL IF GENE NAME OR ANALYTE NOT PROVIDED)

Submitters must provide a gene name, analyte or chromosomal location. If a gene name is provided, the chromosomal location will be automatically provided if available. Submitters can provide the precise location being or just the chromosome band number.

References: HL7-LOINC 47999-8 “DNA Region Name”

EXON(S) BEING TESTED – TEXT FIELD; MANUAL ENTRY OR POSSIBLY PICK LIST – OPTIONAL

Submitter can specify which exons are being tested and their location.

MUTATION(S)/ANALYTE(S) TESTED – TEXT FIELD; MANUAL ENTRY OR POSSIBLY PICK LIST – MINIMAL

Submitter can specify which mutations are tested, their location or provide an identifier such as a dbSNP rs number. For biochemical tests, the submitter can specify which analyte is being tested.

References: 79% of AMP respondents are able to provide this information.
CAP MOL.34907 “Restriction Endonuclease Digestion Confirmation”
CAP MOL.34914 “Gene Information”
CAP MOL.34931 “Sense/Antisense Sequence”
HL7-LOINC 48003-8 “DNA Sequence Variation Identifier (dbSNP rs#)”
HL7-LOINC 48004-6 “DNA Sequence Variation (HGVS)”
HL7-LOINC 48005-3 “Amino Acid Change”

VARIANT IDENTIFIER (RS#/NSV#) – AUTO PROVIDED WITH ABILITY TO SUGGEST NEW

dbSNP (rs#) and dbVar (nsv#) associated with provided variants, if applicable.

References: HL7-LOINC 48003-8 “DNA Sequence Variation Identifier (dbSNP rs#)”

SEQUENCE LOCATION – AUTO PROVIDED WITH ABILITY TO SUGGEST NEW

Location of the chromosome, gene, or variant (tested regions) on the various genome assemblies. Needs to include the location and the assembly. Example: NG# on GRCh37.

SEQUENCE(S) BEING TESTED – TEXT FIELD; MANUAL ENTRY OR POSSIBLY BULK UPLOAD – RECOMMENDED

Submitters can provide as specific a location being tested as possible. If specific and detailed information is provided, it would be possible to map it to RefSeqGene and validate against NCBI resources. It would also make it possible to auto-populate the information on most gene and protein fields.

References: HL7-LOINC 48013-7 “Genomic RefSeq Identifier”
HL7-LOINC 51958-7 “Transcript RefSeq Identifier”
HL7-LOINC 47998-0 “DNA Sequence Variation Display Name”

PROBE(S) BEING TESTED – TEXT FIELD; MANUAL ENTRY OR POSSIBLY BULK UPLOAD – OPTIONAL

Submitters can identify the probes used in the test.

References: CAP MOL.34188 “Probe Characteristics”

PROTEIN(S) NAME – PICK FROM LIST OR AUTOMATICALLY PROVIDED

PROTEIN SYNONYM(S) – AUTOMATICALLY PROVIDED WITH ABILITY TO SUGGEST NEW

PROTEIN ACRONYM(S) – AUTOMATICALLY PROVIDED WITH ABILITY TO SUGGEST NEW

PROTEIN TYPE(S) – AUTOMATICALLY PROVIDED

This field will name the protein type or function. An initial list is below.

- Receptor
- Enzyme
- Structural

CLINICAL SIGNIFICANCE – PICK LIST + REFERENCES – RECOMMENDED

Proposed pick list:

- Pathogenic
- Presumed pathogenic
- Benign
- Presumed benign
- Unknown significance
- Drug response
- Other
- Not assessed
-

TEST COMMENT – TEXT FIELD; MANUALLY PROVIDED – OPTIONAL

This field will allow submitters to provide additional details on the targets for the test. For example, they may add details such as, “Bi-directional sequencing of exons 1-5 with concurrent analysis of Glu234Gly”.

Performance Characteristics:

This section contains information such as analytical validity and assay limitations, as provided by the laboratory. The suggested fields below will capture the test’s performance characteristics.

References: GA

ANALYTICAL VALIDITY – TEXT + CITATION – MINIMAL

Laboratories can provide an explanation of test accuracy and reliability by describing the available information on the fields below that represent the information available from a typical validation study (e.g., as required for CAP).

References: CAP MOL.30785 “Validation Studies – LDT’s”
CAP MOL.31475 “Validation Study”
MMWR

ANALYTICAL SENSITIVITY:

In this field, the submitter can describe the analytical detection rate.

References: 55% of AMP respondents are able to provide this information.
CAP MOL.31475 “Validation Study”
McKesson
Javitt et al

NUMBER OF SPECIMENS USED TO CALCULATE:

References: CAP MOL.30900 “Validation Studies – Specimen Selection”

ANALYTICAL SPECIFICITY:

In this field, the submitter can describe the analytical false positive rate.

References: 55% of AMP respondents are able to provide this information.
CAP MOL.31475 “Validation Study”
McKesson
Javitt et al

PRECISION:

In this field, the submitter can describe how close the results match those from independent sources (known to be the true results).

References: 37% of AMP respondents are able to provide this information.
CAP MOL.31475 “Validation Study”
eDOS OM2-3 “Range of Decimal Precision”
McKesson

ACCURACY:

In this field, the submitter can describe how close repeated results match each other.

References: 42% of AMP respondents are able to provide this information.
CAP MOL.31475 “Validation Study”
McKesson

REFERENCES TO SUPPORT ANALYTICAL VALIDITY

ASSAY LIMITATIONS: TEXT + CITATION – RECOMMENDED

In this field, the submitter can describe any factors that affect the value of the test for its intended use by providing information on test limitations and restrictions.

References: 58% of AMP respondents are able to provide this information.
CAP MOL.31245 “Reference/Reportable Range”
MMWR

LIMIT OF DETECTION:

References: 45% of AMP respondents are able to provide this information.

TEST RESTRICTIONS: (VALIDATED ONLY FOR CERTAIN SUBPOPULATIONS OR PARTICULAR USES)

Quality Control and Quality Assurance:

This section contains information on the test quality control (QC) and quality assurance (QA) methods, such as proficiency testing and validation procedures, as provided by the laboratory.

References: 37% of AMP respondents are able to provide this information.
CAP MOL.20000 “Documented QM/QC Plan”

PROFICIENCY TESTING PERFORMED ON THIS TEST – YES/NO CHECKBOX – RECOMMENDED

References: MMWR

METHOD USED FOR PROFICIENCY TESTING – CHECK BOX – RECOMMENDED

Submitter can specify which proficiency testing is performed for the laboratory; for example, whether the laboratory participates in a formal proficiency testing (PT) program or alternative assessment such as intra-laboratory sample exchanges. This field is mandatory if PT is performed on this test. An initial list is below.

- Formal PT program
- Alternative Assessment (e.g., Intra-Laboratory)

References: CAP MOL.10150 “PT Participation”
CAP MOL.10160 “Alternative Performance Assessment”

PT PROVIDER – PULL-DOWN LIST WITH ABILITY TO SUGGEST NEW – RECOMMENDED

Submitter can identify the institution or agency that provides PT for the test.

CAP TEST LIST – PULL-DOWN LIST – RECOMMENDED

This field only appears if CAP is chosen as PT provider for the test. Submitter can select multiple items.

DESCRIPTION OF PROFICIENCY TESTING METHOD – TEXT + CITATIONS – RECOMMENDED

Submitter can explain how PT is performed for the test and include information on PT results, reportable range, testing interval and number of specimens tested.

References: CAP MOL.10170 “PT Integration Routine Workload”

PROFICIENCY TESTING SCORE/RESULTS:

References: CAP MOL.10200 “PT Evaluation”

PROFICIENCY TESTING REPORTABLE RANGE:

References: CAP MOL.31245 “Reference/Reportable Range”

PROFICIENCY TESTING INTERVAL:

NUMBER SPECIMENS PER PT INTERVAL:

INTERNAL TEST VALIDATION METHOD DESCRIPTION – TEXT + CITATION – RECOMMENDED

Submitter can explain how the laboratory validates the test (initially or when test is changed).

References: CAP MOL.30785 “Validation Studies – LDTs”
CAP MOL.30900 “Validation Studies – Specimen Selection”
CAP MOL.30957 “Verification Studies- FDA cleared”
CAP MOL.31015 “Validation studies – Specimen Types”
CAP MOL.31130 “Validation Study Comparison”
CAP MOL.31475 “Validation Study”
Javitt et al

VALIDATION METHOD REPORTABLE RANGE:

References: CAP MOL.31245 “Reference/Reportable Range”
CAP MOL.31360 “Reference/Reportable Range Quantitative”

CLINICAL VALIDITY – TEXT + CITATION – RECOMMENDED

In this section, the submitter can provide available information on clinical specificity and sensitivity, describe the population and identify the number of specimens used in the validation procedure, and list calculated predictive positive and negative values as exemplified in the fields below.

References: 39% of AMP respondents are able to provide this information.
CAP MOL.31590 “Clinical Performance Characteristics”
McKesson
Javitt et al
GA
MMWR

CLINICAL SPECIFICITY:

Submitter can provide the proportion of negative test results obtained from patients who do not have the defined clinical presentation.

References: 29% of AMP respondents are able to provide this information.
CAP MOL.31590 “Clinical Performance Characteristics”
Javitt et al

CLINICAL SENSITIVITY:

Submitter can provide the proportion of positive test results obtained from patients with the defined clinical presentation.

References: 27% of AMP respondents are able to provide this information.
CAP MOL.31590 “Clinical Performance Characteristics”
Javitt et al

POPULATION(S) USED TO CALCULATE:

Submitter can describe the population used for clinical validity studies.

References: CAP MOL.31590 “Clinical Performance Characteristics”

NUMBER OF SPECIMENS USED TO CALCULATE:

POSITIVE PREDICTIVE VALUE:

The submitter can provide the lifetime risk to develop the disease if the test is positive.

References: CAP MOL.31590 “Clinical Performance Characteristics”

NEGATIVE PREDICTIVE VALUE:

The submitter can provide the probability not to develop the disease if the test is negative.

References: CAP MOL.31590 “Clinical Performance Characteristics”

Clinical Utility: Text + Citation – Recommended

In this section, the submitter can provide available information related to clinical utility such as: describe whether diagnosis can be made without the test; what the burdens are for the patient; cost effectiveness; impact of the test result to the patient (i.e., disease management, lifestyle, prevention); describe impact of test result to family members. Suggested fields to capture this information are shown below.

References: 40% of AMP respondents are able to provide this information.
CAP MOL.30670 “Clinical Indication/Clinical Utility”
McKesson
Javitt et al
GA
MMWR

DESCRIPTION OF HOW TEST IS USEFUL FOR THE PUBLIC:

Submitter can explain the impact the test may have to the general population.

References: Javitt et al

UTILITY OF OUTCOMES:

Submitter can describe the utility of the different outcomes of the test, for instance, in relation to clinical progression and treatment options, lifestyle, prevention, decision making, and whether there is evidence that the genetic test is useful for the patient or his/her relatives in the absence of immediate medical consequences.

References: Javitt et al

BENEFITS:

Submitter can describe the medical, clinical, personal and familial benefits of performing the test.

HARMS:

Submitter can discuss the potential harms of performing the test.

ADDED VALUE (WHEN COMPARED TO CURRENT MANAGEMENT WITHOUT GENETIC TESTING):

Submitter can describe how performing the test influences the patient and their health when compared to not having the test done (e.g., enabling predictive test in the family or prenatal diagnosis).

ASSOCIATED TREATMENTS:

Submitter can discuss the clinical treatment(s) available to patients that test positive for the variation.

Regulations – Test:

This section delineates information for the test-specific FDA regulations, applicable certifications and licenses as described in the fields below.

References: Javitt et al
GA
HL-7
MMWR

FDA REGULATORY CLEARANCES OF THE TEST:

Submitter can provide information related to the test's FDA approval or clearance by providing the applicable information in the fields below.

FDA CATEGORY DESIGNATION – PULL-DOWN LIST – RECOMMENDED

Default is Laboratory Developed Test (LDT) and submitter can choose from the proposed list below.

- IVD – In Vitro Device
- RUO – Research Use Only
- IUO – Informational/Investigational Use Only
- LDT – Laboratory Developed Test
- Not Applicable

References: CAP MOL.29290 "Reagent Data"

FDA REVIEW OF – PULL-DOWN LIST – OPTIONAL

Submitter can name item(s) for which they are providing FDA approval/clearance information. An initial list is below.

- Test kit(s)
- Assay(s)
- Reagent(s)
- Instrument(s)
- Not Applicable

FDA REGULATORY STATUS – CHECK BOX – OPTIONAL

Submitter can specify the status of the application for FDA approval/clearance of choice from above (e.g., test kit, assay, reagents, instruments). An initial list of options is below.

- Approved
- 510(k) Cleared
- PMA Approved
- HDE approved
- Pending
- Not Submitted

References: 50% of AMP respondents are able to provide this information.
Javitt et al

GA
HL7
MMWR

FDA APPLICATION # – TEXT FIELD; MANUAL ENTRY – OPTIONAL

Mandatory if FDA reviewed is chosen.

FDA APPROVAL DOCUMENTS – URL OR UPLOAD; MANUAL ENTRY – OPTIONAL

Submitter can provide links to the FDA website with information on approval or clearance of the genetic test or upload the documents.

TEST-SPECIFIC CERTIFICATIONS/LICENSES – PULL-DOWN MENU – RECOMMENDED

Submitters can select state-specific licenses and other test-specific certifications from the pull-down list. An example is NYSCLEP. “None” will be an option.

TEST-SPECIFIC CERTIFICATIONS/LICENSES APPROVAL # – TEXT FIELD; MANUAL ENTRY – OPTIONAL (MINIMAL IF TEST-SPECIFIC CERTIFICATIONS/LICENSES SELECTED ABOVE)

Certification/License number is mandatory if certification/license selected above.

TEST-SPECIFIC CERTIFICATIONS/LICENSES EXPIRATION DATE (MM/DD/YYYY) – TEXT FIELD; MANUAL ENTRY – OPTIONAL (MINIMAL IF TEST-SPECIFIC CERTIFICATIONS/LICENSES SELECTED ABOVE)

Expiration date is mandatory if certification/license selected above.

Research Test Data Fields:

Data fields that are specifically associated with research tests, not clinical tests.

RESEARCH LABORATORY’S POLICY ON RETURNING RESULTS – TEXT FIELD; MANUAL ENTRY – OPTIONAL

Description of how the laboratory releases the test results: person to whom results are delivered, who delivers the results, delivery method, etc.

IF A NOVEL VARIATION IS FOUND, IS THE SAMPLE SENT TO A CLIA-CERTIFIED LABORATORY FOR CONFIRMATION? – YES/NO CHECKBOX – OPTIONAL