Genetic Testing Registry

GTR Field Definitions - Version 2.1

12/22/2015

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INTRODUCTION

This document delineates the field definitions that will be used in the Genetic Testing Registry (GTR) to aggregate relevant data on genetic tests. This template will allow the following:

- 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 physicians, consumers, payers, researchers
- Development of a user-friendly genetic testing database for reporting, exchanging, and updating of information

The GTR is being designed to collect adequate information on clinical and research genetic tests while taking into consideration the reporting burden on the laboratories, existing resources, standards, and practices, and the practicalities of measuring some of these attributes in the best of conditions. The information that will be collected is divided into three top level fields, namely, laboratory, personnel and test. There are approximately 31

minimally required fields that represent the minimum amount of information that must be submitted in order to register a test in GTR. There are approximately 24 fields that GTR would be able to auto-complete in an attempt to decrease the burden on the submitter and there are approximately 85 fields that are either recommended (35) or optional (50); these fields collect relevant information that may not be available for some tests. Recommended filed will display the words "Not Provided" on the website when left blank by the submitter, optional fields will not appear in the website when left blank by the submitter.

Each data element includes a definition of the data 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

AHRQ – Update on Emerging Genetic Tests Currently Available for Clinical Use in Common Cancers. Available at: https://sp.ncbi.nlm.nih.gov/IEB/RCS/gtr/Shared%20Documents/Database/GeneTestsTracker/GeneTestTracker.pdf

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

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

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.

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

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

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

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

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

HEADER (NOT A DATA FIELD)

HEADER OF FAMILY OF DATA FIELDS

DATA FIELD OR FAMILY OF DATA FIELDS

DATA FIELD

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

References: In the survey administered to AMP members, 72% of responders said they were willing to participate in the GTR.

LABORATORY INFORMATION

It is expected that Laboratory Information is provided and updated with a different time cycle than Test Information. That is, 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.

None of the Laboratory fields map to the e-DOS, since the MSH-4 data element, "Sending Facility" is entirely site defined.

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

NAME OF LABORATORY/FACILITY

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 – MINIMAL

This is the complete name of the test provider.

Required field for both clinical and research tests.

References: CLIA [fieldname]
McKesson
Javitt et al

NAME OF LABORATORY ACRONYMS: TEXT FIELD – OPTIONAL

This is the acronym(s) or short name(s) that identify the test provider. Allow entering multiple acronyms.

Optional field for both clinical and research tests.

GENETESTS AT NCBI LAB ID, IF KNOWN: TEXT FIELD – OPTIONAL

This field only applies to laboratories who participated in the GeneTests Laboratory Directory when it was housed at NCBI-NLM-NIH. Laboratories that participated in the GeneTests Laboratory Directory but are registering a lab as new in GTR must provide their current GeneTests institution ID to help identify themselves and prevent multiple

lab records from displaying to the public. If the laboratory record is migrating from the GeneTests database, this field is automatically provided. This data is not publically displayed.

Optional field for both clinical and research tests.

NAME OF INSTITUTION: TEXT FIELD – OPTIONAL

This is the complete name of the institution that the lab is part of (ex. hospital, university, etc). It may be the same as “Name of Laboratory” for independent labs.

Optional field for both clinical and research tests.

References: Javitt et al

NAME OF INSTITUTION ACRONYMS: TEXT FIELD – OPTIONAL

This is the acronym(s) or short name(s) that identify the institution that the lab is part of. Allow entering multiple acronyms.

Optional field for both clinical and research tests.

NAME OF DEPARTMENT: TEXT FIELD – OPTIONAL

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

Optional field for both clinical and research tests.

References: Javitt et al

FACILITY ADDRESS

Address information of the test provider. In our forms we may use Lab but recognize that not all entities will be laboratories. There can be different mailing vs. shipping addresses that should be considered. The information collected is shown in the fields below and the submitter can choose whether to have the street address public or not.

Note: MGWG recommends that Mailing Address not be on the grid when searching for tests.

FACILITY STREET & NUMBER: TEXT FIELD - OPTIONAL

This is the street Facility address (ex. building number and street name).

Optional field for both clinical and research tests.

References: Javitt et al
McKesson

FACILITY ADDRESS OTHER: TEXT FIELD – OPTIONAL

Submitter can specify any other Facility address details as applicable.

Optional field for both clinical and research tests.

References: Javitt et al
McKesson

FACILITY CITY: TEXT FIELD – MINIMAL - VALIDATION

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

Required field for both clinical and research tests.

FACILITY STATE/PROVINCE: PULL-DOWN LIST – MINIMAL - VALIDATION

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

Required field for both clinical and research tests.

FACILITY POSTAL CODE: TEXT FIELD – MINIMAL – VALIDATION

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

Required field for both clinical and research tests.

FACILITY COUNTRY: PULL-DOWN LIST – MINIMAL

This is the country used in the Facility address. United States will be set as default.

Required field for both clinical and research tests.

FACILITY ADDRESS CAN BE MADE PUBLIC – YES/NO CHECKBOX – MINIMAL

Submitter can choose whether they want GTR users to have their address available in the oublic site.

Default: yes.

Required field for both clinical and research tests.

FACILITY CONTACT INFORMATION

PHONE NUMBER: TEXT FIELD - MINIMAL

This is the general phone number for the lab. It could be specified as: Country-specific area code (set parameters) – phone number.

Required field for both clinical and research tests.

References: Javitt et al
McKesson

FACILITY FAX NUMBER: TEXT FIELD - OPTIONAL

This is the existing general fax number for the lab which can be used by GTR users. It could be specified as: Country-specific area code (set parameters) – fax number.

Optional field for both clinical and research tests.

References: Javitt et al
McKesson

FACILITY EMAIL ADDRESS AND/OR URL TO WEB CONTACT FORM: TEXT FIELD - MINIMAL

This is the existing general email address for the lab or the URL for the lab's web contact form. This information will be available to all GTR users.

Required field for both clinical and research tests.

References: Javitt et al
McKesson

FACILITY MAIN WEBSITE URL: TEXT FIELD – OPTIONAL

This is the URL for the lab's website.

Optional field for both clinical and research tests.

References: Javitt et al

LABORATORY TYPES OF SERVICE

LABORATORY TYPES OF SERVICE NAME: PICK FROM LIST + 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.

Current list:

- Clinical Testing/Confirmation of Mutations Identified Previously
- Cord Blood Banking
- Custom Deletion/Duplication Testing
- Custom Sequence Analysis
- Custom mutation-specific/Carrier testing
- Data Storage and Backup
- Confirmation of research findings
- Custom Balanced Chromosome Rearrangement Studies
- Custom Prenatal Testing
- Custom microarray analysis
- DNA Banking
- Genetic counseling

- Identity Testing
- Insurance billing
- Marker Chromosome Identification
- Preimplantation Genetic Diagnosis (PGD)
- Result interpretation
- Tissue Banking
- Whole Exome Sequencing
- X-Chromosome Inactivation Studies
- Insurance appeals support
- Insurance preauthorization
- Mutation Confirmation
- RNA Banking
- Specimen Source Identification
- Uniparental Disomy (UPD) Testing
- Whole Genome Sequencing
- Other, specify_____

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Laboratory_services.txt

Optional field for both clinical and research tests.

Note: GTR uses the term services to classify tests that are not specific to a specific condition, set of conditions or genes, or chromosomal structures. Services are registered in the GTR with the registration of the laboratory, and not as a test. Included is the order or catalog code for the service used in the order requisition, if applicable.

References: CAP MOL.05075
Javitt et al

LABORATORY SERVICES ORDER CODE: TEXT FIELD - OPTIONAL

This is the order code that the laboratory uses for the particular service(s) selected in the field above.

Optional field for both clinical and research tests.

LABORATORY SERVICES COMMENT: TEXT FIELD – OPTIONAL

This field allows the submitter to provide additional information regarding the laboratory service selected from Laboratory Types of Service(s).

Optional field for both clinical and research tests.

LABORATORY AFFILIATIONS

Is the laboratory linked to a larger health care system, clinical unit that cares for individuals with a given disorder, academic institution? Submitters can identify parent companies of fully owned subsidiaries in this field.

Optional field for both clinical and research tests.

NAME OF AFFILIATE: TEXT FIELD – OPTIONAL

The name of institutions/programs the lab is affiliated or partnered with. Examples of affiliations are a larger health care system, clinical unit that cares for individuals with a given disorder, clinical research divisions and parent companies of fully owned subsidiaries. If the lab has multiple affiliations, all should be entered separately.

WEBSITE URL: TEXT FIELD – OPTIONAL

The URL for the website of the lab's affiliates

LABORATORY PARTICIPATION IN EXTERNAL PROGRAMS

Submitter can specify if lab participates in programs such as standardization or data exchange programs. Submitter can select multiple programs. ClinVar participation will be determined via NCBI's organization database for GTR and ClinVar and automatically reported on GTR records.

Optional field for both clinical and research tests.

PARTICIPATION IN STANDARDIZATION PROGRAMS: CHECKBOXES – OPTIONAL

Does the laboratory participate in standardization programs?

Current list:

- CETT Program (Collaboration Education and Test Translation)
- ISCA Consortium (International Standards for Cytogenomic Arrays)
- Locus-specific Databases
- Mutation-specific Databases
- Other

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Standardization_programs.txt

PARTICIPATION IN DATA EXCHANGE PROGRAMS: CHECKBOXES – OPTIONAL

Does the lab participate in data exchange programs?

Current list:

- CETT Program (Collaboration Education and Test Translation)
- ICCG (International Collaboration for Clinical Genetics) - Previously ISCA
- Locus-specific Databases
- Mutation-specific Database
- Other

Curent list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/External_programs.txt

PERSONNEL INFORMATION

In this section, the laboratory will provide the information for 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 just stored in the database. The laboratory can enter multiple people and identify them as staff or research personnel. Research collaborators may or may not work at the lab, their information is available to be displayed in research tests only.

At least one Person per laboratory is required to be identified as the Laboratory Director.

The personnel information is provided only once for the laboratory and can be disseminated to all the tests the laboratory offers. Furthermore, specific contacts for specific tests can be linked from the lab, 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

These fields collect the complete name of the staff member for which information will be provided in this section.

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

Required field for both clinical and research tests.

MIDDLE INITIAL: TEXT FIELD – OPTIONAL

Optional field for both clinical and research tests.

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

Required field for both clinical and research tests.

DISPLAY PERSON ON GTR WEBSITE: YES/NO RADIO BUTTON – MINIMAL

Should this person’s information be displayed in the GTR website for users to have available? A person can be displayed in the GTR website but choose that their contact information be kept private.

Required field for both clinical and research tests.

PRIMARY LABORATORY CONTACT: YES/NO RADIO BUTTON – MINIMAL

Is this person the primary lab contact for GTR staff? If checked, this person will receive communication from GTR staff if and when appropriate (e.g. annual update messages, submission questions). Each lab needs at least one person for whom ‘Yes’ is selected for this field.

Required (if applicable) field for both clinical and research tests.

LABORATORY DIRECTOR: YES/NO RADIO BUTTON – MINIMAL

Is this person a laboratory director? Each lab needs at least one person for whom ‘Yes’ is selected for this field.

Required (if applicable) field for both clinical and research tests.

PERSON ID: AUTOMATICALLY PROVIDED – HIDDEN (REQUIRED FOR XML BULK LOADS)

This is a unique key for the Person applicable only for test submission via xml. This is only for Laboratories providing data electronically to ensure that they can link a test uniquely to a person in XML bulk upload of data.

Hidden field for both clinical and research tests.

PERSON JOB TITLE: PICK FROM LIST + SUGGEST NEW - OPTIONAL

This is the person's professional title in the laboratory. Submitters can select multiple titles per person. There will be a list of the most common titles (below) to facilitate quick entry and an "Other" field to allow manual entry.

Optional field for both clinical and research tests.

Current list:

- Administrator
- CLIA License Holder
- Genetic Counselor
- Lab Associate Director
- Lab Director
- Medical Director
- Nurse
- Research Nurse
- Scientific Director
- Staff
- Other

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Personnel_titles.txt

PERSON ACADEMIC DEGREE(S): SELECT FROM LIST - OPTIONAL

The academic degree(s) the person holds. Submitter can select multiple academic degrees which will be displayed after the name on website. This information is just for display purposes so only need to select degrees they want displayed.

Current list:

- AA
- BASc
- BEng/BE
- BPharm
- BSc
- CN.A
- DDS/DMD
- DO
- DSc
- MD
- EngD
- JD
- LLB
- BA
- BAdm
- BMedSc/BMedSci
- BS
- BTech
- CRNA
- DNP
- DSW
- Deng
- PhD
- FNP
- JSD/SJD/LLD
- LLM

- LPN
- MBA
- MPharm
- MSc
- ND
- PharmD
- RN
- MA
- MPH
- MSW
- MS
- Other
- PsyD
- ScD

Optional field for both clinical and research tests.

PERSON PROFESSIONAL CERTIFICATIONS

The certification(s) the person holds from medical colleges, boards, associations, and any other relevant organization or institution. Examples of items on the list are below each of the fields below

PROFESSIONAL BOARD: SELECT FROM LIST – OPTIONAL

Current list:

- American Board of Allergy and Immunology
- American Board of Anesthesiology
- American Board of Colon and Rectal Surgery
- American Board of Dermatology
- American Board of Emergency Medicine
- American Board of Family Medicine
- American Board of Genetic Counseling
- American Board of Histocompatibility and Immunogenetics
- American Board of Internal Medicine
- American Board of Medical Genetics
- American Board of Neurological Surgery
- American Board of Nuclear Medicine
- American Board of Obstetrics and Gynecology
- American Board of Ophthalmology
- American Board of Orthopaedic Surgery
- American Board of Otolaryngology
- American Board of Pathology
- American Board of Pediatrics
- American Board of Physical Medicine and Rehabilitation
- American Board of Plastic Surgery
- American Board of Preventive Medicine
- American Board of Psychiatry and Neurology
- American Board of Radiology
- American Board of Surgery
- American Board of Thoracic Surgery
- American Board of Urology
- Genetic Nursing Credentialing Commission
- National Academy of Clinical Biochemistry

PROFESSIONAL BOARD SPECIALTY: SELECT FROM LIST – OPTIONAL

Current list:

- Advanced practice nurse in genetics
- Aerospace Medicine
- Allergy and immunology
- Anatomic Pathology and Clinical Pathology
- Anesthesiology
- Certified Histocompatibility Specialist
- Certified Histocompatibility Technologist
- Certified Histocompatiblity Associate
- Clinical Biochemical Genetics
- Clinical Cytogenetics
- Clinical Genetics
- Clinical Molecular Genetics
- Colon and rectal surgery
- Dermatology

- Diplomate of the ABHI
- Family medicine
- Genetics clinical nurse
- Medical genetics
- Neurology
- Nuclear medicine
- Occupational Medicine
- Orthopaedic surgery
- Pathology - Anatomic
- Pediatrics
- Plastic surgery
- Public Health and General Preventive Medicine
- Radiologic Physics
- Surgery
- Urology
- Emergency medicine
- Genetic counselor
- Internal medicine
- Neurological surgery
- Neurology with Special Qualification in Child Neurology
- Obstetrics and Gynecology
- Ophthalmology
- Otolaryngology
- Pathology - Clinical
- Physical Medicine and Rehabilitation
- Psychiatry
- Radiation Oncology
- Radiology - Diagnostic
- Thoracic surgery
- Vascular Surgery

PROFESSIONAL BOARD SUBSPECIALTY: SELECT FROM LIST – OPTIONAL

Current list

- Addiction Psychiatry
- Advanced Heart Failure and Transplant Cardiology
- Cardiovascular Disease
- Child and Adolescent Psychiatry
- Clinical Neurophysiology
- Congenital Cardiac Surgery
- Cytopathology
- Developmental-Behavioral Pediatrics
- Endocrinology, Diabetes and Metabolism
- Forensic Psychiatry
- Geriatric Medicine
- Gynecologic Oncology
- Hospice and Palliative Medicine
- Interventional Cardiology
- Medical Biochemical Genetics
- Medical Toxicology
- Neonatal-Perinatal Medicine
- Neurodevelopmental Disabilities
- Neuropathology
- Nuclear Radiology
- Pain Medicine
- Pathology - Forensic
- Pathology - Medical Microbiology
- Pathology - Pediatric
- Pediatric Critical Care Medicine
- Pediatric Emergency Medicine
- Pediatric Gastroenterology
- Pediatric Infectious Diseases
- Adolescent Medicine
- Blood Banking/Transfusion Medicine
- Child Abuse Pediatrics
- Clinical Cardiac Electrophysiology
- Clinical and Laboratory Dermatological Immunology
- Critical Care Medicine
- Dermatopathology
- Emergency Medical Services
- Epilepsy
- Gastroenterology
- Geriatric Psychiatry
- Hematology
- Infectious Disease
- Maternal and Fetal Medicine
- Medical Oncology
- Molecular Genetic Pathology
- Nephrology
- Neuromuscular Medicine
- Neurotology
- Orthopaedic Sports Medicine
- Pathology - Chemical
- Pathology - Hematology
- Pathology - Molecular Genetic
- Pediatric Cardiology
- Pediatric Dermatology
- Pediatric Endocrinology
- Pediatric Hematology-Oncology
- Pediatric Nephrology

- Pediatric Otolaryngology
- Pediatric Radiology
- Pediatric Rheumatology
- Pediatric Transplant Hepatology
- Plastic Surgery Within the Head and Neck
- Pulmonary Disease
- Rheumatology
- Spinal Cord Injury Medicine
- Surgery of the Hand
- Transplant Hepatology
- Vascular Neurology
- Pediatric Pulmonology
- Pediatric Rehabilitation Medicine
- Pediatric Surgery
- Pediatric Urology
- Psychosomatic Medicine
- Reproductive Endocrinology/Infertility
- Sleep Medicine
- Sports Medicine
- Surgical Critical Care
- Undersea and Hyperbaric Medicine
- Vascular and Interventional Radiology

PROFESSIONAL CREDENTIALS: SELECT FROM LIST – OPTIONAL

Current list

- ABA
- ABCRS
- ABEM
- ABIM
- ABNM
- ABO
- ABOS
- ABP
- ABPMR
- ABPS
- ABR
- ABTS
- APNG
- C(ASCP)
- CGC
- CHS(ABHI)
- CT(ASCP)
- DLM
- FAAD
- FAAN
- FAAOS
- FAAP
- FACAI
- FACMG
- FACOG
- FACP
- FACR
- FACS
- FACMG
- GCN
- HT(ASCP)
- M(ASCP)
- MLS(ASCP)
- Other
- SBB(ASCP)
- SCT(ASCP)
- ABAI
- ABD
- ABFM
- ABMG
- ABNS
- ABOG
- ABOto
- ABPM
- ABPN
- ABPath
- ABS
- ABU
- BB(ASCP)
- CG(ASCP)
- CHA(ABHI)
- CHT(ABHI)
- D(ABHI)
- DABMG
- FAAFP
- FAAO
- FAAOS
- FACAI
- FACEP
- FACNM
- FACP
- FACPM
- FACRM
- FCAP
- FACB
- H(ASCP)
- HTL(ASCP)
- MB(ASCP)
- MLT(ASCP)
- PA(ASCP)
- SC(ASCP)
- SH(ASCP)

- SM(ASCP)

Optional field for both clinical and research tests.

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

PERSON PUBLICLY DISPLAYED CREDENTIALS: AUTOPOPULATE BASED ON CERTIFICATION AND DEGREE

This field may display all credentials named on the prior 3 fields the way they will be displayed in the public site. Credentials to be displayed are automatically provided based on the information entered.

Autopopulate field for both clinical and research tests.

PERSON CONTACT INFORMATION

Contact information for personnel can be entered and display publicly or be kept private. A person's public telephone number and email address can be entered if a person will be selected as a test-specific contact, in other words, displayed on clinical test records so users can contact the person about the test. Information about how to contact a research person can be entered if a person will be selected as the study contact; that is, displayed on research test records so users can contact the person about the research test or study.

PERSON PUBLIC PHONE NUMBER: TEXT FIELD – OPTIONAL

This is the 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.

Format – US: area code-phone number and extension. Example: 202-555-5555 ext 123

International: +country code-area code-phone number (as applicable). Example: +351-234-555555

Optional field for both clinical and research tests. Required if test-specific contact for both clinical and research tests.

PERSON PRIVATE PHONE NUMBER: TEXT FIELD – OPTIONAL (MINIMAL FOR AT LEAST ONE PERSON PER LAB)

GTR requires the contact information, either a direct work phone number or email address, of at least one laboratory staff member for communication about submission and maintenance of records. This can be the same as the public phone number.

Format – US: area code-phone number and extension. Example: 202-555-5555

International: +country code-area code-phone number (as applicable). Example: +351-234-555555

Optional field for both clinical and research tests. Required for at least one person for both clinical and research tests.

PERSON PUBLIC EMAIL ADDRESS: TEXT FIELD – OPTIONAL

This is the email address or the URL to the lab web contact form 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 email address is optional.

Format: email@host.com Example: smith@lab.com

Optional field for both clinical and research tests. Minimal field for both clinical and research tests if test-specific contact.

PERSON PRIVATE EMAIL ADDRESS: TEXT FIELD – OPTIONAL (MINIMAL FOR AT LEAST ONE PERSON)

The email account attached to NCBI login system by default. This cannot be a web contact form because GTR staff needs to be able to contact this person directly about GTR submissions. This field is optional for personnel being listed for the lab that are not involved in the submission or maintenance of information in the GTR. This email will not be displayed to the public; however, it can be the same as the person's public email.

Format: email@host.com Example: smith@lab.com

Optional field for both clinical and research tests. Minimal field for at least one person for both clinical and research tests.

PERSON PUBLIC FAX NUMBER: TEXT FIELD - OPTIONAL

This is the fax number to be made available for the public to contact the person.

Format – US: area code-fax number and extension. Example: 202-555-5555

International: +country code-area code-fax number (as applicable). Example: +351-234-555555

Optional field for both clinical and research tests.

PERSON PRIVATE FAX NUMBER: TEXT FIELD - OPTIONAL

This is the person's work fax number that will not be made public but may be used by GTR staff to contact as needed.

Format – US: area code-fax number and extension. Example: 202-555-5555

International: +country code-area code-fax number (as applicable). Example: +351-234-555555

Optional field for both clinical and research tests.

PERSON SUPPLEMENTARY PUBLIC CONTACT COMMENT: TEXT FIELD – OPTIONAL

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

Examples: “Person is only available Monday-Thursday”, “Contact for information about mitochondrial disorders”.

Optional field for both clinical and research tests.

LABORATORY LICENSURE AND ACCREDITATIONS

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

References: GA
MMWR

CLIA CERTIFICATION

CLIA CERTIFICATION NUMBER: TEXT FIELD – OPTIONAL (MINIMAL FOR USA LABS PROVIDING CLINICAL TESTS)

This is the certification number assigned by the Clinical Laboratory Improvement Amendments (CLIA) program to the laboratory. This field is required for US labs listing clinical tests. International labs and labs registering research tests are not expected (or required) to have CLIA certification.

Minimal field for USA labs with clinical tests. Optional field for USA or international labs with research tests.

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

CLIA EXPIRATION DATE: TEXT FIELD – OPTIONAL (MINIMAL FOR USA LABS PROVIDING CLINICAL TESTS)

This is the expiration date of the current CLIA certification for the laboratory. This field is required for US labs providing clinical tests that have entered a CLIA certification number. Entered as (mm/dd/yyyy) or select date from the pop up calendar.

There is a calendar in this field, the submitter may choose to select a date on the calendar.

Minimal for USA labs with clinical tests. Optional for USA or international labs with research tests.

References: Javitt et al
McKesson

STATE LICENSE(S)

STATE LICENSE NAME: PULL-DOWN LIST – OPTIONAL

This is the name of the state under which the lab is licensed to practice. Submitter may select multiple.

Current list:

State Clinical Laboratory State Licensing Agency - Agency Accronym
AL - Alabama Department of Public Health ADPH
AK - Alaska Department of Health and Social Services DHSS
AZ - Arizona Department of Health Services ADHS
AR - Arkansas Department of Health ADH
CA - California Department of Public Health CDPH
CO - Colorado Department of Public Health and Environment CDPHE
CT - Connecticut Department of Public Health DPH
DE - Delaware Health and Social Services DHHS
DC - District of Columbia Department of Health DCDOH
FL - Florida Agency for Health Care Administration AHCA
GA - Georgia Department of Community Health DCH
HI - State of Hawai'i Department of Health DOH
ID - Idaho Department of Health and Welfare IDHW
IL - Illinois Department of Public Health IDPH
IN - Indiana State Department of Health ISDH
IA - Iowa Department of Public Health IDPH
KS - Kansas Department of Health and Environment KDHE
KY - Kentucky Cabinet for Health and Family Services CHFS
LA - State of Louisiana Department of Health and Hospitals DHH
ME - Maine Department of Health and Human Services DHHS
MD - Maryland Department of Health and Mental Hygiene DHMH
MA - Executive Office of Health and Human Services EOHHS
MI - Michigan Department of Community Health MDCH
MN - Minnesota Department of Health MDH
MS - Mississipi State Department of Health MSDH
MO - State of Missouri Department of Health and Senior Services DHSS
MT - Montana Department of Public Health and Human Services DPHHS
NE - Nebraska Department of Health and Human Services DHHS
NV - Nevada Department of Health and Human Services DHHS
NH - New Hampshire Department of Health and Human Services DHHS
NJ - State of New jersey Department of Health and Senior Services DHSS
NM - New Mexico Department of Health NMDOH
NY - New York State Department of Health NYSDOH
NY - New York Clinical Laboratory Evaluation Program NYCLEP
NC – North Carolina Department of Health and Human Services DHHS
ND - North Dakota Department of Health NDDoH
OH - Ohio Department of Health ODH
OK - Oklahoma State Department of Health OSDH
OR - Department of human Services DHS

PA - Pennsylvania Department of Health PADOH
RI - State of Rhode Island Department of Health RIDOH
SC - South Carolina Department of Health and Environmental Control DHEC
SD - South Dakota Department of Health SDDOH
TN - Tennessee Department of Health TDOH
TX - Texas Department of State Health Services DSHS
UT - Utah Department of Health UDOH
VA - Virginia Department of Health VDH
VT - Vermont Department of Health VDH
WA - Washington State Department of Health DOH
WV - West Virginia Department of Health and Human Resources DHHR
WI - Wisconsin Department of Health Services DHS
WY - Wyoming Department of Health WDH

Optional field for both clinical and research tests.

STATE LICENSE #: TEXT FIELD – OPTIONAL

This is the license number issued by the US state to the laboratory. It is required for labs that select a state license.

Optional field for both clinical and research tests, unless a state license is available.

STATE LICENSE EXPIRATION DATE: TEXT FIELD – OPTIONAL

This is the expiration date on the US state license. It is required for labs that select a state license.

There is a calendar in this field; the submitter may choose to select a date from the calendar. Entered as (mm/dd/yyyy).

Optional field for both clinical and research tests.

OTHER CERTIFICATION(S)/LICENSE(S)

OTHER CERTIFICATIONS/LICENSES NAME: PULL-DOWN LIST – OPTIONAL

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

Current list:

- American Association of Blood Banks, AABB
- College of American Pathologists, CAP
- Clinical Pathology Accreditation (UK) Ltd, CPA
- European Federation for Immunogenetics, EFI
- European Molecular Genetics Quality Network, EMQN
- NTSAD Tay-Sachs Carrier Testing QC Program, NTSAD

- New York State Clinical Laboratory Evaluation Program, NYS CLEP
- National Association for DNA Collection and Management, NADCM
- American Osteopathic Association, AOA
- American Society of Histocompatibility and Immunogenetics, ASHI
- Joint Commission on Accreditation of Heathcare Organizations, JCAHO
- International Organization for Standardization 15189, ISO15189
- Reference Institute for Bioanalytics, RfB
- Other, please specify _____

Optional field for both clinical and research tests.

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

OTHER CERTIFICATION/LICENSE #: TEXT FIELD – OPTIONAL

This is required for those labs which have license/accreditation/certification numbers available.

Optional field for both clinical and research tests.

OTHER CERTIFICATION/LICENSE EXPIRATION DATE: TEXT FIELD – OPTIONAL

This field is required for those labs which have certification/accreditation/licenses with expiration dates.

There is a calendar in this field; the submitter may choose to select a date from the calendar. Entered as (mm/dd/yyyy).

Optional field for both clinical and research tests.

DEFAULT LABORATORY VALUES FOR TEST INFORMATION

Information stored in the default section can be copied to each test offered by the lab while maintaining the submitter's ability to change them as the test is submitted.

The default section is optional and has been designed only to save submitters from entering the same information multiple times (per test) when entering clinical tests manually. It only applies to tests created after default values are submitted (it will not populate tests that were submitted prior to the default data was entered). The following fields can be provided in the default section if the submitter wants that answer to appear in all their tests. The same fields appear in their appropriate sections both in this document and in the electronic submitter forms.

Default Test Contact Policy: checkbox – Optional

Default Test Orderable By: – Pull-Down List - Optional

Default How to Order: Text + URL – Optional

Default URL to lab website with information about how to order this test Lab

Default Test-Specific Laboratory Services: Check box – Optional

Default Test-Specific Services Order Code: Manual Entry – Optional

Default Test-Specific Services Comment: Manual Entry – Optional

Default Test-Specific Laboratory Additional Services: Check box – Optional

- Custom mutation-specific/Carrier testingCustom Prenatal Testing

Default Test-Specific Additional Services Order Code: Manual Entry – Optional

Corresponds to custom mutation-specific/Carrier testing or Custom prenatal testing default fields

Default Test-Specific Additional Services Comment: Manual Entry – Optional

Corresponds to custom mutation-specific/Carrier testing or Custom prenatal testing default fields

Default Specimen Source – Pull Down List – Optional

Optional field for both clinical and research tests.

Default Variants of unknown significance (VUS) policy and interpretation

What is the Protocol for Interpreting a Variation as a VUS? – Text Field – Recommended

What Software is Used to Interpret Novel Variations? – Text Field – Optional

What Is the Laboratory's Policy on Reporting Novel Variations? – Text Field – Recommended

Are Family Members Who Have Defined Clinical Status Recruited to Assess Significance of VUS Without Charge? – Yes/NoDecline to answer Checkbox with comments - Recommended

Will the Laboratory Re-contact the Ordering Physician if Variant Interpretation Changes? – Yes/No/Decline to answer checkbox with comments – Optional

Comments about the laboratory procedure about the laboratory procedure to re-contact the ordering physician

Default Sample Negative Report – Optional

Default Sample Positive Report – Optional

Default VUS Report – Optional

CLINICAL TEST INFORMATION

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

GTR DATABASE TEST ATTRIBUTES

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.

Necessary for both clinical and research tests.

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

DATE LAST TOUCHED: AUTOMATICALLY PROVIDED

When a laboratory submits updates to 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.

Necessary for both clinical and research tests.

TEST TRACKING ID: TEXT FIELD – OPTIONAL (ONLY FOR BULK UPLOADS)

For tests provided electronically via one of the excel files or XML, one critical element MUST be provided here: a test tracking ID. This is a code which 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 always.

This field only applies to clinical tests.

References: e-DOS OM1-2 "Producer's Service/Test/Observation ID".

NAME OF CLINICAL 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 – MINIMAL

This is the test name which will appear as the title on the GTR public website (e.g. test lists, test detail page) and the title of the test record in the submission interface. It should be the clinical test name in the lab's catalog.

Minimal field for both clinical and research tests.

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

LABORATORY TEST SHORT NAME: TEXT FIELD – OPTIONAL

This is the submitter's short name or mnemonic for the test.

This field applies to clinical and research tests.

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

MANUFACTURER TEST NAME: TEXT FIELD – OPTIONAL

This is the common commercial test name (ex. OvaSure, FDA kit name, etc). Manufacturer test could be an FDA approved test, a kit, or some other manufacturer test.

This field only applies to clinical tests.

SEARCH TERMS: TEXT FIELD - OPTIONAL

The submitter can enter other synonyms and aliases they want the test to be searchable by. These can be names that have been archived, keyword(s) and so on.

This field only applies to clinical tests.

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

TEST DEVELOPMENT – PULL-DOWN LIST – RECOMMENDED

The submitter can categorize how the test was developed by specifying whether the test is laboratory developed (LDT), FDA reviewed, an externally manufactured kit, a modified FDA-reviewed test or combination as exemplified in the proposed list below. This field is to help delineate the differences in certification requirements between test types. Please note that reflex testing is not distinctly included in this field.

Current list:

- Test developed by laboratory (no manufacturer test name)
- FDA-reviewed (has FDA test name)
- Manufactured (research use only; not FDA-reviewed)
- Modified FDA (has FDA-reviewed entry, but with lab modifications/field changes)

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Test_development.txt

This field only applies to clinical tests.

TEST-SPECIFIC SERVICES

Laboratory services that are offered in conjunction with the specific test. This field is different from 'Laboratory services' which allows submitters to select general services the laboratory provides.

TEST-SPECIFIC SERVICES: PULL-DOWN LIST – OPTIONAL

Submitters can select multiple services related to the test.

Current list:

- Clinical Testing/Confirmation of Mutations Identified Previously
- Confirmation of research findings
- Custom Balanced Chromosome Rearrangement Studies
- Custom Deletion/Duplication Testing
- Custom Sequence Analysis
- Data Storage and Backup
- Genetic counseling
- Identity Testing
- Marker Chromosome Identification
- Maternal cell contamination study (MCC)
- Preimplantation Genetic Diagnosis (PGD)
- Result interpretation
- Specimen Source Identification
- Uniparental Disomy (UPD) Testing
- X-Chromosome Inactivation Studies
- Other, specify_____

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Test_services.txt

Optional field for both clinical and research tests.

TEST-SPECIFIC SERVICES ORDER CODE: TEXT FIELD – OPTIONAL

Lab's order code for test specific laboratory service.

Optional field for both clinical and research tests.

TEST-SPECIFIC SERVICES COMMENT: TEXT FIELD – OPTIONAL

This field allows the submitter to provide additional information regarding the test-specific service.

Optional field for both clinical and research tests.

TEST-SPECIFIC ADDITIONAL SERVICES

Additional test services are custom prenatal and/or mutation-specific testing. These are processed separately so they can display in different pages.

TEST-SPECIFIC ADDITIONAL SERVICES: PULL-DOWN LIST – OPTIONAL

These two test services were singled out for processing purposes; specifically, so they can show up in the lab comparison (grid) page. On the public display site, there is only one header/list for Test Services.

Laboratories can specify if they offer custom prenatal and/or mutation-specific testing. These additional services will be represented in GTR's test list page and in the lab comparison (grid) page.

Current list:

- Custom mutation-specific/Carrier testing
- Custom Prenatal Testing

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Test_additional_services.txt

Optional field for both clinical and research tests.

TEST-SPECIFIC ADDITIONAL SERVICES ORDER CODE: TEXT FIELD – OPTIONAL

The order code(s) corresponding to the test-specific custom prenatal and custom mutation-specific/carrier testing additional test services.

Display next to the specific Additional Test Services as "Order Code:" (on the testitem page).

Optional field for both clinical and research tests.

TEST-SPECIFIC ADDITIONAL SERVICES COMMENT: TEXT FIELD – OPTIONAL

This field allows the submitter to provide additional information regarding the test-specific additional services.

Optional field for both clinical and research tests.

HOW TO ORDER: TEXT + URL - RECOMMENDED

This is the description of the test ordering procedure and an URL to the lab website for more details about how the test can be ordered from the lab.

This field only applies to clinical tests.

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

SPECIMEN SOURCE(S): CHECKBOX + URL – RECOMMENDED

List only includes general specimen type required (whole blood, frozen tissue, fresh tissue, sputum, etc). Detailed specimen requirements for the ordering physician should be found at the supplied URL or in the 'How to order' text field. Multiple entries can be selected. Single URL may be supplied.

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

Example list:

- Amniocytes
- Amniotic fluid
- Bone marrow
- Buccal swab
- Peripheral (whole) blood
- Other, specify _____

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Specimen_sources.txt

This field only applies to clinical tests.

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

In the interactive forms, the submitter may select from one of the personnel supplied in the laboratory record. The name of the person must be selected in order to be displayed publicly. Multiple people may be selected and displayed as test specific contacts. For bulk submission via the full test spreadsheet, the person name should be entered exactly how it is in the database. For direct electronic submission of data (i.e. via XML), this field must be supplied as either a unique name matching the personnel list, or a personnel ID previously supplied.

Recommended field for both clinical and research tests.

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

TEST-SPECIFIC CONTACT POLICY: CHECKBOX – RECOMMENDED

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

Current list:

- 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.

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Contact_policy.txt

Recommended field for both clinical and research tests.

INFORMED CONSENT REQUIRED: YES/NO/BASED ON APPLICABLE STATE LAW/DECLINE TO ANSWER CHECKBOX – RECOMMENDED (DEFAULT DECLINE TO ANSWER)

Submitters can choose to let users know if a test requires informed consent prior to testing. It may be the case that all tests for all labs will likely need a disclaimer that informed consent is determined by the ordering physician's state laws.

Informed consent required choices:

- Yes
- No
- Decline to answer
- Informed consent requirements are determined based on applicable state law

This field only applies to clinical tests.

References: MMWR

PRE-TEST GENETIC COUNSELING REQUIRED: YES/NO/DECLINE TO ANSWER CHECKBOX – RECOMMENDED (DEFAULT DECLINE TO ANSWER)

Submitters can choose to let users know if a test requires genetic counseling prior to testing.

Pre-test genetic counseling choices:

- Yes
- No
- Decline to answer

This field only applies to clinical tests.

References: MMWR

POST-TEST GENETIC COUNSELING REQUIRED: YES/NO/DECLINE TO ANSWER CHECKBOX – RECOMMENDED (DEFAULT DECLINE TO ANSWER)

Submitters can choose to let users know if a test requires genetic counseling prior to the release of test results.

Informed consent required choices:

- Yes
- No

- Decline to answer

This field only applies to clinical tests.

References: MMWR

TESTING STRATEGY: TEXT + CITATIONS – 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.

This field only applies to clinical tests.

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

LABORATORY TEST ORDER CODE: TEXT FIELD - RECOMMENDED

This is the laboratory’s order or catalog code for the test (ie. The order code to put in the requisition to order the test from the lab).

This field only applies to clinical tests.

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

LOINC CODE(S): TEXT – OPTIONAL

Submitters can provide the LOINC code(s) that identify the test. Multiple codes can be entered as applicable to the test. There is a “search” link that takes the submitter to the LOINC website to search for the LOINC code for the test. Once identified, the code must be copied and pasted in this text box. GTR will automatically provide a url with the code entered to the respective LOINC record in the LOINC website for users to have access to it from the GTR test page.

This field only applies to clinical tests.

TEST CODES URL: TEXT – RECOMMENDED

Submitters can provide an URL for the lab’s website with information on codes including CPT, ICD-9, and ICD-10 that are applicable to this test.

This field only applies to clinical tests.

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

URL FOR THE TEST: TEXT FIELD – RECOMMENDED

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

Recommended field for both clinical and research tests.

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. To increase transparency of where the test is performed and to show whether the outside lab has the same qualifications as the reporting lab. External means any lab/facility that does not belong to the reporting lab. For example, a test performed at an outside facility owned by the same company would be considered to be performed "at an outside lab". Submitters can enter details and provide appropriate information for clarification of their entry as needed.

References: McKesson

PERFORMANCE SITE FOR TEST COMPONENTS: CHECKBOX + TEXT COMMENT – MINIMAL

Identification of where all parts of the test are performed. The complete list of options is below. Submitter can check multiple boxes. There is also a free text box for comments. The text box must be filled out if any part of the test is performed at an outside facility. It should contain information about the outside lab, the procedure or any other relevant information.

- Entire test performed in-house
- Specimen preparation performed in-house
- Specimen preparation performed at an outside lab
- Wet lab work performed in-house
- Wet lab work performed at an outside lab
- Interpretation performed in-house
- Interpretation performed at an outside lab
- Report generated in-house
- Report generated at an outside lab

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Test_location.txt

Minimal field for clinical tests.

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

ACCESSIBILITY

This section identifies who can order the test.

References: GA
MMWR

TEST ORDERABLE BY: CHECKBOX – OPTIONAL

Submitter should identify who can order the test from this lab (eg. specify Health Care Providers: Licensed physician, PA, RN, NP, GC, etc.). Submitter can select multiple choices.

Current list:

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

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Test_orderable_by.txt

This field only applies to clinical tests.

REPORTING OF TEST RESULTS

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

Suggestion: Standards for molecular reporting (AMP), ACMG sequence variants categories. Labs should be asked to define their criteria for interpretation (e.g. Pathogenic, benign, uncertain results).

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 – RECOMMENDED

Submitter can upload a sample negative report for the corresponding test. The report should not have any patient information. GTR staff will review reports to make sure there is no identifiable information before making the report public.

This field only applies to clinical tests.

SAMPLE POSITIVE REPORT: UPLOAD DOCUMENT – RECOMMENDED

Submitter can upload a sample positive report for the corresponding test. The report should not have any patient information. GTR staff will review reports to make sure there is no identifiable information before making the report public.

This field only applies to clinical tests.

SAMPLE VUS REPORT: UPLOAD DOCUMENT – OPTIONAL

Submitter can upload a sample VUS report for this test if applicable. The report should not have any patient information. GTR staff will review reports to make sure there is no identifiable information before making the report public.

This field only applies to clinical tests.

VARIANTS OF UNKNOWN SIGNIFICANCE (VUS) POLICY AND INTERPRETATION

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

WHAT IS THE PROTOCOL FOR INTERPRETING A VARIATION AS A VUS?: TEXT FIELD – RECOMMENDED

Description of how Variants of Unknown Significance are analysed and reported.

This field only applies to clinical tests.

WHAT SOFTWARE IS USED TO INTERPRET NOVEL VARIATIONS?: TEXT FIELD – 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.

This field only applies to clinical tests.

WHAT IS THE LABORATORY'S POLICY ON REPORTING NOVEL VARIATIONS?: TEXT FIELD – RECOMMENDED

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

This field only applies to clinical tests.

ARE FAMILY MEMBERS WHO HAVE DEFINED CLINICAL STATUS RECRUITED TO ASSESS SIGNIFICANCE OF VUS WITHOUT CHARGE?: YES/NO/DECLINE TO ANSWER/NOT PROVIDED - CHECKBOX + COMMENTS – RECOMMENDED

Will the lab offer the test to family members free of charge? The laboratory may choose to not disclose this information by selecting the “Decline to answer” option.

This field only applies to clinical tests.

WILL THE LABORATORY RE-CONTACT THE ORDERING PHYSICIAN IF VARIANT INTERPRETATION CHANGES?: YES/NO/DECLINE TO ANSWER/NOT PROVIDED – CHECKBOX + COMMENTS – RECOMMENDED

Description of how the lab deals with ongoing interpretation of genetic tests results after the initial report. The laboratory may choose not to disclose this information by selecting the “Decline to answer” option.

This field only applies to clinical tests.

IS RESEARCH ALLOWED ON THE SAMPLE AFTER CLINICAL TESTING IS COMPLETE?: TEXT FIELD – RECOMMENDED

After clinical testing is complete, does the laboratory perform any research testing using the submitted specimen? Are there any details to clarify this field? (e.g. are patients reconsented?)

This field only applies to clinical tests.

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 from sources such as ClinVar, OMIM, HPO and MeSH.

References: AHRQ
CAP MOL.30670 “Clinical Indication/Clinical Utility”
MMWR

PURPOSE OF THE TEST: PULL-DOWN LIST – MINIMAL

This is the purpose(s) or indication(s) for use of the test. Multiple purposes can be selected.

Current list:

- Diagnosis
- Drug Response
- Monitoring
- Mutation Confirmation
- Pre-implantation genetic diagnosis
- Predictive
- Pre-symptomatic
- Prognostic
- Recurrence

- Risk Assessment
- Screening
- Therapeutic management

Definition of the terms:

Diagnosis - Identification or confirmation of disease.

Drug response - Evaluation of genetic variability influencing an individual's reaction to specific medications, as in a pharmacogenetic test. Excludes immune-mediated adverse drug reactions (dose-independent drug allergies).

Monitoring - Periodic or continuous evaluation of a disease or condition over time, including a patient's response to medical treatment.

Mutation confirmation - Re-evaluation of a genetic test result to assess the validity of the initial result. For example, research test results or results from another laboratory.

Pre-implantation genetic diagnosis - Genetic testing performed on a small number of cells from a human embryo prior to uterine implantation as part of assisted reproduction procedures.

Predictive - Information from the test can be used to determine or predict the potential risk of eventually developing a disease or a disorder. Applicable to predictive biomarkers used in oncology, used to predict treatment response.

Pre-symptomatic - Genetic analysis of an asymptomatic or unaffected individual who is at risk of a specific genetic disorder.

Prognostic - Information from the test can be used to determine or predict the aggressiveness of the disease or overall outcome of the disease at the time of initial diagnosis and prior to initiation of treatment. Applicable to prognostic biomarkers used in oncology.

Recurrence - Used to detect disease recurrence in a patient who has already been diagnosed and treated for cancer.

Risk assessment -Evaluation of the likelihood of developing a specific condition based on genetic risk. Includes carrier testing in affected families.

Therapeutic management - Information can be used to determine therapeutic decision making.

Screening - Evaluation of a target population to identify a subgroup affected by a genetic condition or that have the potential to transmit the trait to their offspring. Includes newborn screening, ethnicity-based screening and pre-conceptual genetic testing.

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Purpose_of_the_test.txt

This field only applies to clinical tests.

References: 61% of AMP respondents are able to provide this information.
AHRQ
McKesson
Javitt et al

MMWR

CLINICAL OR RESEARCH TEST: CHECKBOX - MINIMAL

Submitters indicate whether the test is for clinical purposes or is a research test by clicking an initial button that opens a different set of forms since the information collected is different for these two types of tests.

MGWG supports inclusion of this field and recommends adding a field to indicate if data is used for research purposes (such as ISCA or CETT). MGWG also recommends a stronger connection at the lab display for when a lab does both clinical and research testing.

Minimal field for both clinical and research tests.

Condition for which test is offered: Pick from autocomplete dictionary + Suggest new – Minimal

Name of the disease, syndrome, drug response or phenotype for which the test can be ordered. Submitters can choose a condition name from the autocomplete dictionary. Start typing the condition name and the autocomplete dictionary will display choices. Try different spelings of the condition name if you do not see the name in the list. Search GTR for condition names.

Please note that condition names in this list may differ from the laboratory preferred name and the lab has the opportunity in the field below to type how it wants the condition to appear in their test page. The autocomplete dictionary list of diseases is comprised by names from sources like SNOMED CT, OMIM, GeneReviews, and other authoritative sources. This is an attempt to standardize disease nomenclature in GTR. When a condition is selected from the autocomplete dictionary, some fields will be automatically provided.

If the condition name is not in the autocomplete dictionary, the submitter has the opportunity to search the GTR database of condition names and then copy and paste it in this box.

If the test is being offered for a condition not found in the autocomplete dictionary (i.e. a newly published disease), the submitter can enter the new condition name in this box and provide a comment and all supportive evidence in the box at the bottom of the page entitiled “Private comment about the condition/phenotype to GTR staff”. GTR staff will review all documentation and add the condition name to the database.

A test may have multiple indications. This means that a submitter can enter multiple condition names for which the test is offered.

If the test has more than one condition (e.g. panels), one condition is selected as primary. By default, the first condition entered is identified as the primary condition. The primary condition in a test dictates the information displaying publicly in the Indication tab of the test and in the relevant sections of the navigation panel of the test (right hand corner of the test page).

Minimal field for both clinical and research tests.

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”

MGWG: discussion of pick list.

LAB-DISPLAYED CONDITION NAME: TEXT FIELD – OPTIONAL

The submitter can provide the condition name they want associated with the test if the condition name is different from the one selected in the autocomplete dictionary provided in the field above. This field is optional and only needs to be filled if the submitter wants to provide a different condition name or preferred synonym. This name will be displayed in the public test record.

Optional for both clinical and research tests.

CONDITION IDENTIFIER(S): AUTOMATICALLY PROVIDED

NCBI will provide the SNOMED CT name as the preferred condition name when available. The term is connected to the same concepts from sources such as OMIM, Orphanet and GeneReviews.

Field applies to both clinical and research tests.

References: McKesson

CONDITION SYNONYM(S): AUTOMATICALLY PROVIDED + SUGGEST NEW

Once the condition/phenotype is identified, the condition synonym(s) will automatically display for the submitter to review. The submitter can also provide the condition synonym(s) they want associated with the test if the condition synonym is different from the one(s) automatically provided by GTR by suggesting new synonym(s).

Field applies to both clinical and research tests.

CONDITION PREFERRED ACRONYM: AUTOMATICALLY PROVIDED

The condition preferred acronym will be automatically provided when a condition is selected from the autocomplete dictionary.

Field applies to both clinical and research tests.

CONDITION ACRONYM(S): AUTOMATICALLY PROVIDED + SUGGEST NEW

The condition alternate acronyms will be automatically provided when a condition is selected from the autocomplete dictionary. The submitter can type other condition acronyms that the lab uses and that do not appear on the list. These acronyms will only be displayed in the test item page for this lab.

Field applies to both clinical and research tests.

LAB-DISPLAYED CONDITION PREFERRED ACRONYM: TEXT FIELD – OPTIONAL

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

This field only applies to clinical tests.

INDICATION TYPE: PULL-DOWN LIST – AUTOMATICALLY PROVIDED + SUGGEST NEW

NCBI assigns broad categories to the name of a condition.

Current list:

- blood group
- disease
- finding
- named protein variant
- pharmacological response

This fields applies to both clinical and research tests.

DISEASE CLINICAL SUMMARY: AUTOMATICALLY PROVIDED

This information is based on the primary condition selected for the test. It will be displayed to the public and is not currently available in the submission interface.

Field automatically provided for both clinical and research tests.

DISEASE CLINICAL FEATURE(S): AUTOMATICALLY PROVIDED

This information is based on the primary condition selected for the test. It will be displayed to the public and is not currently available in the submission interface.

Field automatically provided for both clinical and research tests.

MODE OF INHERITANCE: AUTOMATICALLY PROVIDED + SUGGEST NEW

This information is based on the primary condition selected for the test. The mode of inheritance will be automatically provided when available and the submitter will have the opportunity to suggest a new mode of inheritance by selecting from a menu list. If the condition may be inherited according to multiple modes of inheritance, please select 'Other' and type all the applicable modes of inheritance.

Current list:

- Autosomal dominant inheritance
- Autosomal dominant inheritance with maternal imprinting
- Autosomal dominant inheritance with paternal imprinting
- Autosomal recessive inheritance
- Autosomal unknown
- Codominant
- Genetic anticipation
- Mitochondrial inheritance

- Multifactorial inheritance
- Oligogenic inheritance
- Sex-limited autosomal dominant
- Somatic mutation
- Sporadic
- Unknown mechanism
- X-linked dominant inheritance
- X-linked inheritance
- X-linked recessive inheritance
- Y-linked inheritance
- Other, please specify

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Mode_of_inheritance.txt

Field applies to both clinical and research tests.

DISEASE MECHANISM: PULL-DOWN LIST + SUGGEST NEW – OPTIONAL

This is the disease mechanism for the condition/phenotype in relation to the test target (measurement). This is the identification of the reasoning or biological process that causes the disease.

Current list:

- Gain of function
- Loss of function
- Other, please specify

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Disease_mechanism.txt

Field applies to both clinical and research tests.

PREVALENCE: AUTOMATICALLY PROVIDED + SUGGEST NEW + URL + CITATIONS – OPTIONAL

The prevalence, or the most current estimated number of cases of the disease in the population. Citation(s) can be provided to support the prevalence statement and a URL to a page with more information.

This field applies to both clinical and research tests.

AMA CPT CODE(S): SELECT FROM LIST – OPTIONAL

AMA CPT MoPath code(s) is the set of molecular pathology Current Procedural Terminology (CPT®) codes - 'MoPath' codes - from the American Medical Association (AMA) are available for selection into the test record. The MoPath CPT codes were developed to represent nucleic acid analyses to detect germline or somatic variation or for HLA Class I and Class II typing, all of which are in scope for GTR. Tier 1, Tier 2, and 'Unlisted' codes are available in GTR to submitters who accept the AMA license agreement. The so-called 'stacking codes' have been deleted from the AMA CPT codebook.

This field only applies to clinical tests.

TARGET POPULATION: TEXT + CITATIONS – RECOMMENDED

This is the explanation of which segment(s) of the population should be tested for this disease using this specific test, and why. Citation(s) can be provided to support the information about target population.

This field only applies to clinical tests.

References: MMWR (recommended patient population)

PRIVATE COMMENT ABOUT THE CONDITION/PHENOTYPE TO GTR STAFF: TEXT – OPTIONAL

This is a place for submitters to provide explanations or suggestions about a condition/phenotype name that differs from those in the GTR autocomplete dictionary. GTR staff will review and process the new condition/phenotype name as appropriate.

This field applies to both clinical and research tests.

TEST METHODOLOGY

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

MGWG: Methodology is one of the most valuable fields to clinicians and needs to be easily viewable on public retrieval.

MAJOR METHOD CATEGORY: PULL-DOWN LIST – MINIMAL

Category of test methods by major method category which subsets the menu choices for subsequent fields.

Current list:

- Biochemical Genetics
- Cytogenetics
- Molecular Genetics

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Major_method_category.txt

This field applies to both clinical and research tests.

References: eDOS OM1-18 “Nature of Service/Test/Observation”
McKesson
MMWR
If this is by test type (molecular, biochemical, etc.) we could map the data from GeneTests method categories.
HL7-LOINC 48002-0 “Genomic Source Class” (somatic/germ)

METHOD CATEGORY: PULL-DOWN LIST - MINIMAL

Name of the general category the test belongs to. Can select multiple categories per test. Each category will have a single higher level category which is assigned by NCBI based on category selected. Multiple sets of higher level method category, method category and test methodology can be entered per test.

Example list for cytogenetics:

FISH-metaphase	Cytogenetics
FISH-interphase	Cytogenetics
Chromosome breakage studies	Cytogenetics
Sister chromatid exchange	Cytogenetics
Multicolor FISH (M-FISH)/Spectral Karyotyping (SKY)	Cytogenetics
Fluorescence in situ hybridization (FISH)	Cytogenetics
Karyotyping	Cytogenetics

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Method_category.txt

This field is field for both clinical and research tests.

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 + SUGGEST NEW – MINIMAL

Name of the test method used in the assay. Allows multiple selection and ability to enter value not on the list. Each methodology is selected after selection of a category. Once a method category is selected, one or more test methodologies relevant to that category can be selected or a new method entered. If a user selects multiple method categories after each selection of a category they will be given a list of relevant test methodologies to choose from (or enter new).

Example list:

- Allele-specific primer extension (ASPE)
- Alternative splicing detection
- Bi-directional Sanger Sequence Analysis
- C-banding
- Chromatin Immunoprecipitation on ChIP
- Other, specify_____

This field is for clinical and research tests.

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: PULL-DOWN LIST - RECOMMENDED

Name of the platform used in the test, if any. This field is not currently displaying to the public.

Example list:

- Affymetrix CytoScan HD Array
- Agilent Human CpG Island Microarray Kit, 1x244K
- Illumina Infinium HD HumanCytoSNP-12
- Manufacturer-specific test
- None/not applicable
- Other, specify_____

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Platforms.txt

This field applies to both clinical and research tests.

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 instrument(s) used for a specific methodology. If the instrument used is not on the list there is the ability to provide new instrument names.

Example list:

- Applied Biosystems 3730 capillary sequencing instrument
- BioRad CFX96
- Covaris S2 Sonicator
- PerkinElmer Victor3 1420 Multilabel Plate Reader
- Roche LightCycler 480
- Other, specify

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Instruments.txt

This field only applies to both clinical and research tests.

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

TEST PROCEDURE/PROTOCOL: TEXT + CITATIONS – OPTIONAL

This is the summary of the methodology which may include the description of the specific steps for each method used in the assay.

This field applies to both clinical and research tests.

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

CONFIRMATION OF TEST RESULTS: TEXT FIELD – RECOMMENDED

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

This field applies to both clinical and research tests.

TEST TARGET(S)

This section describes the analytes, chromosomal or mitochondrial regions, genes and variants, or proteins that are measured in the test. Every target must be linked to at least one specific indication.

GERMLINE, SOMATIC OR BOTH: RADIO BUTTON – MINIMAL (GERMLINE SELECTED BY DEFAULT)

Identify whether the test target is germline or somatic (e.g. cancer) or both.

This field applies to both clinical and research tests.

WHAT THE TEST MEASURES: PULL-DOWN LIST – MINIMAL

This field names the category of the analyte being tested.

Current list:

- Analyte
- Chromosomal region/mitochondrion
- Gene
- Protein

This field applies to both clinical and research tests.

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 – SELECT FROM AUTOCOMPLETE DICTIONARY - MINIMAL

This is the symbol of the gene that the test interrogates. There is an autocomplete dictionary that provides choices when the gene name or symbol is typed. When the gene name is identified, the gene symbol, synonym, location, family, OMIM # will be provided automatically in the public site, if available.

This field applies to both clinical and research tests.

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 LOCATION: AUTOMATICALLY PROVIDED

This information will be displayed to the public and is not currently available in the submission interface.

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

This field applies to both clinical and research tests.

ASSOCIATED REFERENCE SEQUENCE: TEXT FIELD – OPTIONAL

Submitters must provide a gene name, analyte, protein or chromosomal location. If a gene name is provided, the chromosomal location will be automatically provided as available. Submitters can provide as specific a location being tested as possible or just the chromosome band number. There is an autocomplete dictionary that provides choices as you typin (eg. NM).

This field applies to both clinical and research tests.

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

RELEVANT EXON(S): TEXT FIELD – OPTIONAL

Submitter can specify which exons are being tested and their location. When specifying the exons tested, the reference sequence must be identified as well.

This field applies to both clinical and research tests.

VARIANT(S): TEXT FIELD OR AUTOCOMPLETE DICTIONARY + SUGGEST NEW – OPTIONAL

Submitter can specify which variant(s) are tested by HGVS name or expression, common name. There is an autocomplete dictionary that provides choices as the NM or NP or the variation are typed. Please select from the autocomplete dictionary.

This field applies to both clinical and research tests.

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”

CLINICAL SIGNIFICANCE: PULL-DOWN LIST + CITATIONS – RECOMMENDED

Select the clinical significance of the variant to condition selected by selecting from the menu list. Submitters also have the opportunity to provide citations.

Current list:

- Benign
- Benign/Likely benign
- Likely benign
- Likely pathogenic
- Pathogenic
- Pathogenic/Likely pathogenic
- Uncertain significance
- association
- confers sensitivity
- conflicting data from submitters
- drug response
- not provided
- protective
- risk factor
- Other, specify

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Clinical_significance.txt

This field applies to both clinical and research tests.

VARIANT IDENTIFIER (RS#/NSV#): AUTOMATICALLY PROVIDED

dbSNP (rs#) and dbVar (nsv#) associated with provided variants, if applicable. This information will be displayed to the public and is not currently available in the submission interface.

This field applies to both clinical and research tests.

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

ANALYTE(S): AUTOCOMPLETE DICTIONARY + SUGGEST NEW – MINIMAL (IF APPLICABLE)

For biochemical tests, the submitter can specify which analyte or enzyme is being tested. There is an autocomplete dictionary that provides choices as the analyte name is typed.

This field applies to both clinical and research tests.

CHROMOSOMAL REGION/MITOCONDRION: AUTOCOMPLETE DICTIONARY + SUGGEST NEW – MINIMAL (IF APPLICABLE)

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.

This field applies to both clinical and research tests.

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

PROTEIN NAME(S): PICK FROM AUTOCOMPLETE DICTIONARY + SUGGEST NEW

There is an autocomplete dictionary that provides choices as the protein or enzyme name is typed.

This field applies to both clinical and research tests.

PROTEIN SYNONYM(S): AUTOMATICALLY PROVIDED

This information will be displayed to the public and is not currently available in the submission interface.

This field applies to both clinical and research tests.

PROTEIN ACRONYM(S): AUTOMATICALLY PROVIDED

This information will be displayed to the public and is not currently available in the submission interface.

This field applies to both clinical and research tests.

TEST COMMENT: TEXT FIELD – OPTIONAL

This field will allow submitters to provide additional details on the test and its targets. For example, add details such as, “Bi-directional sequencing of exons 1-5 with concurrent analysis of Glu234Gly”. Please note that adding details about the test in this field does not preclude from providing details about the targets the test interrogates in the test target section.

This field applies to both clinical and research tests.

PERFORMANCE CHARACTERISTICS

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

References: GA

ANALYTICAL VALIDITY: TEXT- MANUAL ENTRY – MINIMAL

Defined as the accuracy and reliability of the test for measuring the component of interest. Labs can provide an explanation of how accurate and reliable the test is by describing the available information on analytical sensitivity and the number of specimens used to calculate it, analytical specificity, precision and accuracy that represent the information available from a typical validation study (ex. as required for CAP). These fields have been categorized in the fields below for clarity but please note that all this information will be entered in one text box (citations will

be entered separately) in an effort to reduce reporting burden and ease data entry by submitters. Labs are also encouraged to include internal data about analytical validity in this field, especially, if there are no citations or published papers available.

This field only applies to clinical tests.

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

ANALYTICAL VALIDITY CITATION(S): TEXT – RECOMMENDED

Laboratories are encouraged to provide citations to support their analytical validity statements. PubMed citations (PMID or PMCID) are encouraged when available as this will facilitate linking to the full citation from the GTR website. The submission user interface enables submitters to search the PubMed database by providing a link called “Search PubMed” next to the citations box. Once the desired paper is identified, submitters must copy the PMID or PMCID and past it in the citations box of the GTR submission user interface. When a PMID or PMCID are provided, citations are displayed as links to the PubMed record so they are easily available to users of the GTR public site.

This field only applies to clinical tests.

ASSAY LIMITATION(S): TEXT + CITATIONS – 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.

This field only applies to clinical tests.

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:

For example, describe whether the test was validated only for certain subpopulations or particular uses.

QUALITY CONTROL AND QUALITY ASSURANCE

This section contains information on the test QC and QA methods, such as PT testing and validation procedures, as provided by the lab.

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 RADIO BUTTON – RECOMMENDED

In this section, the submitter can provide information on proficiency testing if the lab performs it for this test. Proficiency testing can be defined as the use of blind testing to assess whether the laboratory can perform a test correctly. Usually the samples are provided by an organization independent of the laboratories performing the test and that information can be provided in the fields below.

This field only applies to clinical tests.

References: MMWR

METHOD USED FOR PROFICIENCY TESTING: – PULL-DOWN LIST + SUGGEST NEW – RECOMMENDED

Submitter can specify which type of proficiency testing is performed for the lab; for example, whether the lab participates in a formal PT program, alternative assessment or intra-laboratory sample exchanges. Submitter can select multiple choices. This field is minimal if PT is performed on this test.

Current list:

- Alternative Assessment
- Formal PT program
- Inter-Laboratory
- Intra-Laboratory
- Other, specify_____

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Proficiency_testing_method.txt

This field only applies to clinical tests.

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

PROFICIENCY TESTING PROVIDER: PULL-DOWN LIST + SUGGEST NEW – RECOMMENDED

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

This field only applies to clinical tests.

Current list:

- American College of Medical Genetics / College of American Pathologists, ACMG/CAP
- American College of Physicians - Medical Laboratory Evaluation, ACP MLE
- Association for Molecular Pathology, AMP
- Belgian Official EKE Schemes, GLP Monitoring Programme
- Canadian External Quality Assessment Laboratory, CEQAL
- Centers for Disease Control and Prevention Newborn Screening Quality Assurance Program, CDC DLS
- Clinical Pathology Accreditation (UK) Ltd., CPA
- European Concerted Action on Thrombosis External Quality Assessment Program, ECAT EQAP
- European Molecular Genetics Quality Network, EMQN
- European Research Network for the Evaluation and Improvement of Screening Diagnosis and Treatment of Inherited Metabolic Disorders - External Quality Assessment Schemes, ERNDIM EQAS
- External Quality Assessment for Molecular Genetic Testing for Phenylketonuria, EQA-PKU
- Human Genetic Society of Australasia, HGSA
- Institute for Standardization and Documentation in the Medical Laboratory, INSTAND
- Pacific Northwest Regional Genetics Group, PacNoRGG
- Society for Inherited Metabolic Disorders, SIMD

- Other, specify

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Proficiency_testing_provider.txt

CAP INFORMATION

If the test PT provider is CAP, submitter can enter the appropriate information. Multiple options can be selected. Choices for controlled lists were taken from 2012 CAP Survey catalog. Available at: http://www.cap.org/apps/docs/proficiency_testing/2012_surveys_catalog.pdf

This field only applies to clinical tests.

MAJOR CAP CATEGORY: PULL-DOWN LIST – RECOMMENDED

Example list:

- Alpha-1 Antitrypsin (SERPINA1) Genotyping
- Biochemical Genetics
- Cytogenomic Microarray Analysis
- Defective DNA Mismatch Repair/HNPCC
- Molecular Hematologic Oncology
- Pharmacogenetics

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/CAP_test_categories.txt

CAP CATEGORY: PULL-DOWN LIST – RECOMMENDED

Example list for biochemical genetics:

Acylcarnitines, qualitative and quantitative	Biochemical Genetics
Amino acids, qualitative and quantitative	Biochemical Genetics
Carnitine, qualitative and quantitative	Biochemical Genetics
Glycosaminoglycans (mucopolysaccharides), qualitative and quantitative	Biochemical Genetics
Organic acids, qualitative and quantitative	Biochemical Genetics
Educational challenge	Biochemical Genetics

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/CAP_test_list.txt

CAP TEST LIST: PULL-DOWN LIST – RECOMMENDED

Example list for pharmacogenetics:

PGx	Allele detection (genotype)
PGx	Interpretation
PGx	CYP2C19
PGx	CYP2C9

PGx1	IL28B
PGx2	HLA-B*5701

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/CAP_test_codes.txt

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.

This field only applies to clinical tests.

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 + CITATIONS - RECOMMENDED

Submitter can explain how the laboratory validates the test (initially or when test is changed) and describe the validation method reportable range.

This field only applies to clinical tests.

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 + CITATIONS – RECOMMENDED

Defined as how consistently and accurately the test detects or predicts the intermediate or final outcomes of interest. 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.

This field only applies to clinical tests.

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

CLINICAL SENSITIVITY:

Submitter can provide the proportion of positive test results among 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

MGWG: recommendation that this is valuable information for clinicians and should consider adding as a distinct field that is easily viewable

CLINICAL SPECIFICITY:

Submitter can provide the proportion of negative test results among patients without the defined clinical presentation.

References: 29% 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:

Submitter can provide the number of specimens used to calculate the clinical sensitivity and specificity of the test.

POSITIVE PREDICTIVE VALUE:

The submitter can provide the chance of having the phenotype among those that test positive.

References: CAP MOL.31590 “Clinical Performance Characteristics”

NEGATIVE PREDICTIVE VALUE:

The submitter can provide the the chance of not having the phenotype among those that test negative.

References: CAP MOL.31590 “Clinical Performance Characteristics”

CLINICAL UTILITY: PULL-DOWN LIST + URL + CITATIONS – RECOMMENDED

In this field, 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. A medical definition of clinical utility is defined narrowly (How likely the test is to significantly improve patient outcomes), but GTR uses a broader definition including possible impact on family, lifestyle planning, etc.

Submitters can select categories from a pull-down menu describing how a test might be used and provide references from the peer-reviewed literature that support the categories choices or a URL to a page with such information. Submitters can select multiple items from the list. Each item selected must be supported by citation(s) or references to recommendations or practice guidelines for the test that have been issued by authoritative groups (e.g., U.S. Preventive Services Task Force, professional societies such as ACMG, EuroGentest) or a URL to such information.

The categories for the pull-down list are:

- Avoidance of invasive testing
- Establish or confirm diagnosis
- Guidance for management
- Guidance for selecting a drug therapy and/or dose
- Lifestyle planning
- Predictive risk information for patient and/or family members
- Reproductive decision-making
- Sufficient research has not been conducted to establish utility of the test
- Other, specify_____

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Clinical_utility_categories.txt

Submitters can also provide a URL to their lab’s webpage with more information on the clinical utility of the test; for example, a description of how the test is useful for the public, utility of outcomes (clinical, personal, decision making, informational, etc), harms, benefits, added value (when compared to current management without genetic testing) and associated treatments.

References that are in PubMed or PubMedCentral will be automatically categorized as per PubMed/PubMedCentral’s tags. All other references must be manually categorized by submitters into one of the following three categories:

- Published studies

- Published guidelines
- Third-party reviews and/or recommendations

If no evidence of utility is available, submitters could choose to display a statement explaining that sufficient research has not been conducted to demonstrate the utility of test.

This field only applies to clinical tests.

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

REGULATIONS – TEST

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

This section only applies to clinical tests.

References: Javitt et al
GA
HL-7
MMWR

FDA REGULATORY CLEARANCES OF THE TEST

Submitter can provide information related to the test or part(s) of the test that has been submitted for review by the FDA. There is one set of the fields per item or part of the test that has FDA information which is selected in the field “FDA review of”. If there has been no FDA review, then only the "FDA Category Designation" field is applicable. If the test is a test developed by the laboratory (LDT) and the FDA exercises enforcement discretion, then the submitter only needs to fill out the field “FDA Category Designation”.

If multiple parts of the test have been reviewed by the FDA, the submitter can provide multiple sets of information for each part of the test reviewed by the FDA. For example, if both the reagents and the instrument used during the test have been reviewed by the FDA, the submitter can provide two sets of “FDA review of, FDA category designation, FDA regulatory status, FDA application #, and upload FDA approval documents”. If the FDA application # is provided, GTR will automatically provide a link to the FDA website with information on that specific application # so the submitter does not have to upload the documents manually. The submitter is encouraged to provide at least the “FDA Category Designation”.

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/FDA_Category_Pt_1.txt

This field only applies to clinical tests.

HAS THERE BEEN FDA REVIEW OF THE TEST?: YES/NO CHECKBOX – OPTIONAL

This field only applies to clinical tests.

FDA REVIEW OF: PULL-DOWN LIST + SUGGEST NEW – OPTIONAL

Submitter can name which item(s) they are providing FDA review/clearance info for. Submitter can select multiple items and each item needs its own set of "FDA review of, FDA category designation, FDA regulatory status, FDA application #, and upload FDA approval documents".

Current list:

- ASR – Analyte Specific Reagent(s)
- Assay(s)
- Instrument(s)
- IVDMIA - In Vitro Diagnostic Multivariate Index Assay(s)
- Test kit(s)
- Other, specify _____

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Items_FDA_reviewed.txt

This field only applies to clinical tests.

FDA CATEGORY DESIGNATION: PULL-DOWN LIST – RECOMMENDED

Default is "FDA exercises enforcement discretion" which applies to tests developed by the laboratory which have not been evaluated by the FDA. Submitter can choose from the list below.

Current list:

- IVD – In Vitro Device
- RUO – Research Use Only. Not for use in diagnostic procedures.
- IUO – Investigational Use Only. The performance characteristics of this product have not been established.

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/FDA_Category_Pt_1.txt

This field only applies to clinical tests.

References: CAP MOL.29290 "Reagent Data"

FDA REGULATORY STATUS: PULL-DOWN LIST - OPTIONAL

Submitter can specify the status of the application for FDA clearance of choice from above (test kit, assay, reagents, instruments, etc). Submitter can choose from the list below.

Current list:

- FDA Cleared/Approved
- FDA exempt
- Pending

- Not Submitted
- Other, specify _____

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/FDA_status.txt

This field only applies to clinical tests.

References: 50% of AMP respondents are able to provide this information.
Javitt et al
GA
HL7
MMWR

FDA APPLICATION #: TEXT FIELD – OPTIONAL

Required if "FDA Cleared/Approved" is chosen. NCBI will automatically provide a link to the FDA page with the approval documents using the application #. Submitter can review the link.

This field only applies to clinical tests.

FDA APPROVAL DOCUMENTS: UPLOAD – OPTIONAL

NCBI will provide a link to the FDA page with the approval documents using the application # entered above. However, the submitter can also upload these documents directly into GTR.

This field only applies to clinical tests.

NEW YORK STATE CLEP CERTIFICATION

Submitters can select whether the test has New York State Clinical Laboratory Evaluation Program certification.

This field only applies to clinical tests.

NYSCLEP CERTIFICATION STATUS: PULL-DOWN LIST - OPTIONAL (MINIMAL IF NYSCLEP SELECTED ABOVE)

Current list:

- Approved
- Grandfathered
- Exempt
- Pending

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/NYS_CLEP_status.txt

NEW YORK STATE CLEP CERTIFICATION #: TEXT FIELD – OPTIONAL (MINIMAL IF NYSCLEP SELECTED ABOVE)

Certification/License number is minimal if NYSCLEP selected above.

This field only applies to clinical tests.

NEW YORK STATE CLEP CERTIFICATION EXPIRATION DATE: TEXT FIELD – OPTIONAL (MINIMAL IF NYSCLEP SELECTED ABOVE)

Expiration date is minimal if certification/license selected above. Entered as (mm/dd/yyyy).

This field only applies to clinical tests.

RESEARCH TEST INFORMATION

For the purposes of the GTR, a research test is defined as a test that is performed for the purpose of contributing to generalizable knowledge or for a laboratory to generate data in order to make technical improvements to a test.

The following data fields that are specifically associated with research tests, not clinical tests. The fields that are collected for both test types are identified in throughout the document.

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: AUTOMATICALLY PROVIDED

When a laboratory submits updates to 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 RESEARCH 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 – MINIMAL

This is the test name which will appear as the title on the GTR public website for the research test (e.g. test lists, test detail page) and the title of the test record in the submission interface. It should be the research 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 – OPTIONAL

This is the submitter’s short name or mnemonic for the test.

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

WHAT IS THE PURPOSE OF THE RESEARCH TEST?: RADIO BUTTON – MINIMAL

For the purposes of the GTR, a research test is defined as a test that is performed for the purpose of contributing to generalizable knowledge or for a laboratory to generate data in order to make technical improvements to a test. 'Contribute to generalizable knowledge' is applicable if the intent is to publish the study results and/or the study protocol is aproved by a research ethics committee (e.g., Institutional Review Board).

Current list:

- Contribute to generalizable knowledge
- For the laboratory to generate data in order to make technical improvements to a test

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Purpose_of_research_test.txt

RESEARCH STUDY INFORMATION

RESEARCH STUDY NAME: TEXT – OPTIONAL

Complete study name from protocol approved by research ethics committee, if applicable. Otherwise, name of study used by the laboratory.

SHORT STUDY NAME: TEXT – OPTIONAL

Shortened study name one line in length to appear in lists and summaries.

CLINICALTRIALS.GOV IDENTIFIER: TEXT – OPTIONAL

If study is registered at www.ClinicalTrials.gov, enter Identifier in format NCT00000000.

URL FOR THE STUDY: TEXT – OPTIONAL

Enter URL to the study website, if one is available.

PROTOCOL NUMBER IF THE STUDY IS APPROVED BY A RESEARCH ETHICS COMMITTEE: TEXT – OPTIONAL (RECOMMENDED IF THE PURPOSE OF THE TEST IS TO CONTRIBUTE TO GENERALIZEABLE KNOWLEDGE)

If the study is approved by a research ethics committee (e.g., Institutional Review Board), the protocol number should be provided.

WHAT IS THE STUDY TYPE?: RADIO BUTTON – OPTIONAL

Current list:

- Interventional study (or clinical trial)
- Observational study
- Expanded access
- Not applicable

Definitions from ClinicalTrials.gov:

Interventional - Participants are assigned to receive diagnostic, therapeutic, or other types of interventions.

Observational - Participants are assessed for biomedical or health outcomes. Interventions may be received but are not assigned as part of the study.

Expanded Access - Investigational new drugs are provided to patients with serious conditions through an FDA-regulated process.

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Study_type.txt

DESCRIPTION OF RESEARCH: TEXT FIELD + CITATIONS – MINIMAL

Single open text field to describe the research study/test and citations to support the description. There can be multiple citations and citations are optional.

STUDY AIMS AND HYPOTHESES: TEXT – OPTIONAL

Specify study aims and hypotheses. Copy/paste from study protocol, if available.

UPLOAD STUDY PROTOCOL: UPLOAD – OPTIONAL

Upload study protocol as a pdf or word file. The file should not have any proprietary information.

RESEARCHERS AND CONTACT INFORMATION

Research personnel are entered in the Laboratory section of the submission site, then selected from within test submissions. The Laboratory personnel section in the lab has information about the required information for each of the research person type.

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Research_personnel.txt

PERSON RESPONSIBLE FOR THE STUDY: SELECT FROM LIST – MINIMAL

Name of principal investigator or lab director who is ultimately responsible for the conduct of the research.

STUDY CONTACT: SELECT FROM LIST – MINIMAL

Person designated as the contact to enroll participants in study.

CO-INVESTIGATOR: SELECT FROM LIST – OPTIONAL

Individual(s) involved with the Principal investigator in the scientific development or execution of the research project.

RESEARCH CONTACT POLICY: TEXT – OPTIONAL

Description of when and how Study contact should be contacted.

STUDY PARTICIPATION INFORMATION

UPLOAD PARTICIPANT CONSENT FORM: UPLOAD - OPTIONAL

Upload consent form (as a pdf or word file) that participants are required to review and sign. GTR users will be able to view and print the document.

IS THE STUDY CURRENTLY RECRUITING PARTICIPANTS?: RADIO BUTTON – RECOMMENDED

Indicate whether enrollment is currently active or not. This information should be kept up-to-date by editing the test whenever recruitment status changes.

Current list:

- Yes
- No
- Unknown

ELIGIBILITY CRITERIA: TEXT – RECOMMENDED

Listing of the inclusion and exclusion criteria for study participation e.g. age range, gender, diagnostic features, etc.

RECRUITMENT SITES: TEXT – OPTIONAL

Description of the site(s) or location(s) where participants would go to enroll and participate in the study.

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 from sources such as ClinVar, OMIM, HPO and MeSH.

References: AHRQ
CAP MOL.30670 “Clinical Indication/Clinical Utility”
MMWR

CONDITION FOR WHICH TEST IS OFFERED: PICK FROM AUTOCOMPLETE DICTIONARY + SUGGEST NEW – MINIMAL

Name of the disease, syndrome, drug response or phenotype for which the test can be ordered. Submitters can choose a condition name from the autocomplete dictionary. Start typing the condition name and the autocomplete dictionary will display choices. Try different spelings of the condition name if you do not see the name in the list. Search GTR for condition names.

Please note that condition names in this list may differ from the laboratory preferred name and the lab has the opportunity in the field below to type how it wants the condition to appear in their test page. The autocomplete dictionary list of diseases is comprised by names from sources like SNOMED CT, OMIM, GeneReviews, and other authoritative sources. This is an attempt to standardize disease nomenclature in GTR. When a condition is selected from the autocomplete dictionary, some fields will be automatically provided.

If the condition name is not in the autocomplete dictionary, the submitter has the opportunity to search the GTR database of condition names and then copy and paste it in this box.

If the test is being offered for a condition not found in the autocomplete dictionary (i.e. a newly published disease), the submitter can enter the new condition name in this box and provide a comment and all supportive evidence in the box at the bottom of the page entitiled “Private comment about the condition/phenotype to GTR staff”. GTR staff will review all documentation and add the condition name to the database.

A test may have multiple indications. This means that a submitter can enter multiple condition names for which the test is offered.

If the test has more than one condition (e.g. panels), one condition is selected as primary. By default, the first condition entered is identified as the primary condition. The primary condition in a test dictates the information displaying publicly in the Indication tab of the test and in the relevant sections of the navigation panel of the test (right hand corner of the test page).

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”
MGWG: discussion of pick list.

LAB-DISPLAYED CONDITION NAME: MANUAL ENTRY - OPTIONAL

The submitter can provide the condition name they want associated with the test if the condition name is different from the one selected in the autocomplete dictionary provided in the field above. This field is optional and only needs to be filled if the submitter wants to provide a different condition name or preferred synonym. This name will be displayed in the public test record.

CONDITION IDENTIFIER(S): AUTOMATICALLY PROVIDED

NCBI will provide the SNOMED CT name as the preferred condition name when available. The term is connected to the same concepts from sources such as OMIM, Orphanet and GeneReviews.

References: McKesson

CONDITION SYNONYM(S): AUTOMATICALLY PROVIDED + SUGGEST NEW

Once the condition/phenotype is identified, the condition synonym(s) will automatically display for the submitter to review. The submitter can also provide the condition synonym(s) they want associated with the test if the condition synonym is different from the one(s) automatically provided by GTR by suggesting new synonym(s).

CONDITION PREFERRED ACRONYM: AUTOMATICALLY PROVIDED

The condition preferred acronym will be automatically provided when a condition is selected from the autocomplete dictionary.

CONDITION ACRONYM(S): AUTOMATICALLY PROVIDED + SUGGEST NEW

The condition alternate acronyms will be automatically provided when a condition is selected from the autocomplete dictionary. The submitter can type other condition acronyms that the lab uses and that do not appear on the list. These acronyms will only be displayed in the test item page for this lab.

LAB-DISPLAYED CONDITION PREFERRED ACRONYM: MANUAL ENTRY - OPTIONAL

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

INDICATION TYPE: SELECT FROM LIST – AUTOMATICALLY PROVIDED + SUGGEST NEW

NCBI assigns broad categories to the name of a condition.

Current list:

- blood group
- disease
- finding
- named protein variant
- pharmacological response

DISEASE CLINICAL SUMMARY: AUTOMATICALLY PROVIDED

This information is based on the primary condition selected for the test. It will be displayed to the public and is not currently available in the submission interface.

DISEASE CLINICAL FEATURE(S): AUTOMATICALLY PROVIDED

This information is based on the primary condition selected for the test. It will be displayed to the public and is not currently available in the submission interface.

MODE OF INHERITANCE: AUTOMATICALLY PROVIDED + SUGGEST NEW

This information is based on the primary condition selected for the test. The mode of inheritance will be automatically provided when available and the submitter will have the opportunity to suggest a new mode of inheritance by selecting from a menu list. If the condition may be inherited according to multiple modes of inheritance, please select 'Other' and type all the applicable modes of inheritance.

Current list:

- Autosomal dominant inheritance
- Autosomal dominant inheritance with maternal imprinting
- Autosomal dominant inheritance with paternal imprinting
- Autosomal recessive inheritance
- Autosomal unknown
- Codominant
- Genetic anticipation
- Mitochondrial inheritance
- Multifactorial inheritance
- Oligogenic inheritance
- Sex-limited autosomal dominant
- Somatic mutation
- Sporadic
- Unknown mechanism
- X-linked dominant inheritance
- X-linked inheritance
- X-linked recessive inheritance
- Y-linked inheritance
- Other, please specify

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Mode_of_inheritance.txt

DISEASE MECHANISM: SELECT FROM LIST + SUGGEST NEW– OPTIONAL

This is the disease mechanism for the condition/phenotype in relation to the test target (measurement). This is the identification of the reasoning or biological process that causes the disease.

Current list:

- Gain of function
- Loss of function
- Other, please specify

PREVALENCE: AUTOMATICALLY PROVIDED + SUGGEST NEW + URL + CITATIONS – OPTIONAL

The prevalence, or the most current estimated number of cases of the disease in the population. Citation(s) can be provided to support the prevalence statement and a URL to a page with more information.

PRIVATE COMMENT ABOUT THE CONDITION/PHENOTYPE TO GTR STAFF: TEXT – OPTIONAL

This is a place for submitters to provide explanations or suggestions about a condition/phenotype name that differs from those in the GTR autocomplete dictionary. GTR staff will review and process the new condition/phenotype name as appropriate.

TEST METHODOLOGY

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

MGWG: Methodology is one of the most valuable fields to clinicians and needs to be easily viewable on public retrieval.

MAJOR METHOD CATEGORY: PULL-DOWN LIST – MINIMAL

Category of test methods by major method category which subsets the menu choices for subsequent fields.

Current list:

- Biochemical Genetics
- Cytogenetics
- Molecular Genetics

References: eDOS OM1-18 “Nature of Service/Test/Observation”
McKesson
MMWR
If this is by test type (molecular, biochemical, etc.) we could map the data from GeneTests method categories.
HL7-LOINC 48002-0 “Genomic Source Class” (somatic/germ)

METHOD CATEGORY: PULL-DOWN LIST - MINIMAL

Name of the general category the test belongs to. Can select multiple categories per test. Each category will have a single higher level category which is assigned by NCBI based on category selected. Multiple sets of higher level method category, method category and test methodology can be entered per test.

Example list for cytogenetics:

FISH-metaphase	Cytogenetics

FISH-interphase	Cytogenetics
Chromosome breakage studies	Cytogenetics
Sister chromatid exchange	Cytogenetics
Multicolor FISH (M-FISH)/Spectral Karyotyping©„¢ (SKY©)	Cytogenetics
Fluorescence in situ hybridization (FISH)	Cytogenetics
Karyotyping	Cytogenetics

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Method_category.txt

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 + SUGGEST NEW – MINIMAL

Name of the test method used in the assay. Allows multiple selection and ability to enter value not on the list. Each methodology is selected after selection of a category. Once a method category is selected, one or more test methodologies relevant to that category can be selected or a new method entered. If a user selects multiple method categories after each selection of a category they will be given a list of relevant test methodologies to choose from (or enter new).

Example list:

- Methylation-specific PCR
- Microarray
- Multiplex Ligation-dependent Probe Amplification (MLPA)
- Next-Generation (NGS)/Massively parallel sequencing (MPS)
- Oligonucleotide Ligation Assay (OLA)
- Other, specify_____

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Primary_test_methodology.txt

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: PULL-DOWN LIST - RECOMMENDED

Name of the platform used in the test, if any. This field is not currently displaying to the public.

Example list:

- Affymetrix GeneChip Human Genome U133 Plus 2.0 Array
- Illumina Infinium HD HumanCytoSNP-12

- Life Technologies TaqMan OpenArray MicroRNA Panels
- Manufacturer-specific test
- None/not applicable
- Other, specify_____

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Platforms.txt

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 instrument(s) used for a specific methodology. If the instrument used is not on the list there is the ability to provide new instrument names.

Example list:

- Affymetrix HotStart-IT Probe qPCR Master Mix with UDG (2X)
- Agilent 2100 Bioanalyzer
- Applied Biosystems 3730 capillary sequencing instrument
- Roche LightCycler 480
- Tecan Genesis Robotic Workstation 150
- Other, specify

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/Instruments.txt

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

TEST PROCEDURE/PROTOCOL: TEXT + CITATIONS – OPTIONAL

This is the 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 – RECOMMENDED

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

TEST TARGET(S)

This section describes the analytes, chromosomal or mitochondrial regions, genes and variants, or proteins that are measured in the test. Every target must be linked to at least one specific indication.

GERMLINE, SOMATIC OR BOTH: RADIO BUTTON – MINIMAL (GERMLINE SELECTED BY DEFAULT)

Identify whether the test target is germline or somatic (e.g. cancer) or both.

WHAT THE TEST MEASURES: PULL-DOWN LIST – MINIMAL

This names the category of the analyte being tested.

Current list:

- Analyte
- Chromosomal region/mitochondrion
- Gene
- Protein

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 – SELECT FROM AUTOCOMPLETE DICTIONARY - MINIMAL

This is the symbol of the gene that the test interrogates. There is an autocomplete dictionary that provides choices when the gene name or symbol is typed. When the gene name is identified, the gene symbol, synonym, location, family, OMIM # will be provided automatically in the public site, 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 LOCATION: AUTOMATICALLY PROVIDED

This information will be displayed to the public and is not currently available in the submission interface.

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

ASSOCIATED REFERENCE SEQUENCE: TEXT FIELD – OPTIONAL

Submitters must provide a gene name, analyte, protein or chromosomal location. If a gene name is provided, the chromosomal location will be automatically provided as available. Submitters can provide as specific a location being tested as possible or just the chromosome band number. There is an autocomplete dictionary that provides choices as you typin (eg. NM).

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

RELEVANT EXON(S): TEXT FIELD – OPTIONAL

Submitter can specify which exons are being tested and their location. When specifying the exons tested, the reference sequence must be identified as well.

VARIANT(S): TEXT FIELD OR AUTOCOMPLETE DICTIONARY + SUGGEST NEW – OPTIONAL

Submitter can specify which variant(s) are tested by HGVS name or expression, common name. There is an autocomplete dictionary that provides choices as the NM or NP or the variation are typed. Please select from the autocomplete dictionary.

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”

CLINICAL SIGNIFICANCE: PULL-DOWN LIST + CITATIONS – RECOMMENDED

Select the clinical significance of the variant to condition selected by selecting from the menu list. Submitters also have the opportunity to provide citations.

Example list:

- Affects
- Benign
- Likely benign
- Likely pathogenic
- Pathogenic
- Uncertain significance
- association
- confers sensitivity
- drug response
- not provided
- protective
- risk factor
- Other, specify

Current list: ftp://ftp.ncbi.nlm.nih.gov/pub/GTR/standard_terms/CAP_test_list.txt

VARIANT IDENTIFIER (RS#/NSV#): AUTOMATICALLY PROVIDED

dbSNP (rs#) and dbVar (nsv#) associated with provided variants, if applicable. This information will be displayed to the public and is not currently available in the submission interface.

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

ANALYTE(S): AUTOCOMPLETE DICTIONARY + SUGGEST NEW – MINIMAL (IF APPLICABLE)

For biochemical tests, the submitter can specify which analyte or enzyme is being tested. There is an autocomplete dictionary that provides choices as the analyte name is typed.

CHROMOSOMAL REGION/MITOCONDRION: AUTOCOMPLETE DICTIONARY + SUGGEST NEW – MINIMAL (IF APPLICABLE)

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"

PROTEIN NAME(S): PICK FROM AUTOCOMPLETE DICTIONARY + SUGGEST NEW

There is an autocomplete dictionary that provides choices as the protein or enzyme name is typed.

PROTEIN SYNONYM(S): AUTOMATICALLY PROVIDED

This information will be displayed to the public and is not currently available in the submission interface.

PROTEIN ACRONYM(S): AUTOMATICALLY PROVIDED

This information will be displayed to the public and is not currently available in the submission interface.

TEST COMMENT: TEXT FIELD – OPTIONAL

This field will allow submitters to provide additional details on the test and its targets. For example, add details such as, "Bi-directional sequencing of exons 1-5 with concurrent analysis of Glu234Gly". Please note that adding details about the test in this field does not preclude from providing details about the targets the test interrogates in the test target section.

REFERENCES:

These are references used in the writing of this document.
PubMed ID(s):
OMIM Disease
OMIM Gene
AHRQ
GeneReviews
Orphanet Reviews
Orphanet Case Articles
Genetics Home Reference Disease
Genetics Home Reference Gene

ACMG Guidelines
ACOG Guidelines
NSGC Guidelines
SIMD Guidelines
ASHG Guidelines
ACLA Guidelines
CAP Guidelines
EGAPP; ACCE Guidelines; GeT-RM Program
Other CDC Resources: Lab Practice Evaluation Team; Newborn Screening and Related Conditions Team
ACT Newborn Screening Guidelines
NCHPEG GeneFacts Decision-Support Pages
CLSI Guidelines (Clinical and Laboratory Standards Institute)
NCCLS MMIA Guidelines (National Committee for Clinical Laboratory Standards)
APHL Guidelines (American Public Health Laboratories)
NIST Guidelines
USPSTF Guidelines (US Preventive Services Task Force)
DOE Task Force Guidelines [Need to check and see if any of these are disease or method related]
NICE Guidelines
Patient Support/Advocacy Groups
Genetic Alliance

Create an information sheet that the labs can use that gathers all information together seamlessly.

COMMENTS:

PHASE II FIELDS FOR RESEARCH TESTS

RESEARCH LABORATORY'S POLICY ON RETURNING RESULTS – TEXT FIELD – OPTIONAL

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

Phase II field for research tests only.

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

Phase II field for research tests only.

DROPPED FIELDS:

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.

This field has been removed merged with Person Professional Credentials.

Optional field for both clinical and research tests.

TEST TURN-AROUND-TIME: TEXT FIELD - OPTIONAL

Ability to provide a range of TATs. Possibly asking for minimum, average and maximum TATs.

References: CAP MOL.20300 “Turn Around Time”
eDOS OM1-23 “Typical Turn Around Time”
McKesson
Javitt et al

MGWG expressed concern that this may vary widely depending on circumstances, and that an ordering group would have to discuss this with the testing lab anyway. MGWG did not think it would be used to decide which test lab to select. MGWG recommended dropping this field from GTR.

PRICE

‘Price’ was determined to be removed at this time due to significant controversy and difficulty in consistency in reporting.

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

The following two questions will only be required 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 permitted 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 who the external collaborator/lab is.

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 lab A is the reporting lab, wet lab work is performed in lab B (reviewed entry), interpretation in company C (did not review entry), the answer to this question is No.

This field only applies to clinical tests.

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 field only applies to clinical tests.

DRUG INFORMATION: AUTOMATICALLY PROVIDED

For pharmacogenetic tests, additional information about the drug can be obtained from the links to PharmGKB and DailyMed that will be displayed in the Dicovery panel of the test item page.

Field applies to both clinical and research tests.

OTHER NAMES TYPE: PULL-DOWN LIST – OPTIONAL

The submitter can classify the type of test name entered above. Examples include: archived, synonym, keyword, or they can specify other.

This field only applies to clinical tests.

PROBE(S) BEING TESTED: TEXT FIELD OR POSSIBLY BULK UPLOAD - OPTIONAL

Submitters can identify the probes used in the test.

This field only applies to clinical tests.

References: CAP MOL.34188 “Probe Characteristics”