

81247-9 HL7 genetic variant reporting panel

PANEL HIERARCHY

LOINC#	LOINC Name	R/O/C	Cardinality	Ex. UCUM Units
81247-9	HL7 genetic variant reporting panel			
81294-1	Reference sequence coding system identifier panel			
81248-7	Transcript reference sequence coding system [Type]			
81249-5	Genomic reference sequence coding system			
81295-8	Local transcript reference sequence coding system OID	C		
81296-6	Local genomic reference sequence coding system OID	C		
48018-6	Gene [Identifier] in Blood or Tissue		0..*	
36908-2	Gene mutations tested for in Blood or Tissue by Molecular genetics method Nominal	C	0..*	
51959-5	Range(s) of DNA sequences examined	C	0..*	
81293-3	Range(s) of DNA sequences examined Narrative	C	0..1	
53577-3	Reason for study additional note [Text] in Blood or Tissue by Molecular genetics method Narrative	O	0..*	
51967-8	Genetic disease assessed [Identifier] in Blood or Tissue by Molecular genetics method	O	0..*	
51969-4	Genetic analysis summary report in Blood or Tissue Document by Molecular genetics method			
81303-0	HGVS version [Identifier]			
51968-6	Genetic disease analysis overall interpretation in Blood or Tissue by Molecular genetics method			
81250-3	Simple variant panel		0..n	
48003-8	DNA sequence variation identifier [Identifier] in Blood or Tissue by Molecular genetics method			
81252-9	Simple variant [Identifier]			
81292-5	Simple variant [Type]			
48018-6	Gene [Identifier] in Blood or Tissue			
51958-7	Transcript reference sequence [Identifier] in Blood or Tissue			
48004-6	DNA change			
48005-3	Amino acid change in Blood or Tissue by Molecular genetics method			
48019-4	DNA change type			
48006-1	Amino acid change type in Blood or Tissue by Molecular genetics method			
48013-7	Genomic reference sequence [Identifier] in Blood or Tissue			
69547-8	Genomic reference allele			
81254-5	Genomic allele location [Identifier]			
69551-0	Genomic alternate allele			
81255-2	dbSNP [Identifier]			
81256-0	COSMIC [Identifier]			
81257-8	CIGAR [Identifier]			
81258-6	Allelic frequency			%
48001-2	Chromosome region [Identifier] in Blood or Tissue by Molecular genetics method			
48002-0	Genomic source class [Type] in Blood or Tissue by Molecular genetics method			
53034-5	Allelic state in Blood or Tissue by Molecular genetics method			
53037-8	Clinical significance			
81259-4	Associated phenotype			
81251-1	Complex variant panel		0..n	
48008-7	Allele name [Identifier] in Blood or Tissue by Molecular genetics method			
81260-2	Complex variant [Identifier]			
81262-8	Complex variant HGVS name			
81263-6	Complex variant type			
81259-4	Associated phenotype			
53037-8	Clinical significance			
53034-5	Allelic state in Blood or Tissue by Molecular genetics method			
81250-3	Simple variant panel		0..n	
48003-8	DNA sequence variation identifier [Identifier] in Blood or Tissue by Molecular genetics method			
81252-9	Simple variant [Identifier]			
81292-5	Simple variant [Type]			
48018-6	Gene [Identifier] in Blood or Tissue			
51958-7	Transcript reference sequence [Identifier] in Blood or Tissue			
48004-6	DNA change			
48019-4	DNA change type			
48005-3	Amino acid change in Blood or Tissue by Molecular genetics method			
48006-1	Amino acid change type in Blood or Tissue by Molecular genetics method			
48013-7	Genomic reference sequence [Identifier] in Blood or Tissue			
69547-8	Genomic reference allele			
81254-5	Genomic allele location [Identifier]			
69551-0	Genomic alternate allele			
81255-2	dbSNP [Identifier]			
81256-0	COSMIC [Identifier]			
81257-8	CIGAR [Identifier]			
81258-6	Allelic frequency			%

48001-2	Chromosome region [Identifier] in Blood or Tissue by Molecular genetics method
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53034-5	Allelic state in Blood or Tissue by Molecular genetics method
53037-8	Genetic disease sequence variation interpretation in Blood or Tissue by Molecular genetics method
81259-4	Associated phenotype
81297-4	Structural variant panel
81286-7	Structural variant [Identifier]
81287-5	Structural variant reported start-end [NumRange]
81288-3	Precision of boundaries Nominal
81299-0	Structural variant reported arrCGH [Ratio]
81289-1	Structural variant [Type]
81300-6	Structural variant [Length]
81301-4	Structural variant outer start-end [NumRange]
81302-2	Structural variant inner start-end [NumRange]
81290-9	Structural variant HGVS name
81291-7	Structural variant ISCN name
81298-2	Structural variant cytogenetic location [Identifier]

PART DEFINITION/DESCRIPTION(S)**Part:** -

to be used for panels only

Source: Regenstrief LOINC

Selected information about each LOINC that is part of this panel**[81294-1](#) Reference sequence coding system identifier panel****PART DEFINITION/DESCRIPTION(S)****Part:** -

to be used for panels only

Source: Regenstrief LOINC

[81248-7](#) Transcript reference sequence coding system [Type]**TERM DEFINITION/DESCRIPTION(S)**

The coding system that is the source for the transcript reference sequence, such as NCBI, Ensembl, and LRG.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)**Part:** [Type](#)

Type

Source: Regenstrief LOINC

EXAMPLE ANSWER LIST ([LL3998-3](#))

SEQ#	Answer	Answer ID
1	NCBI <i>Description: National Center for Biotechnology Information/NIH</i>	LA26214-9
2	Ensembl	LA26215-6
3	LRG <i>Description: Locus Reference Genomic</i>	LA26216-4
4	Local	LA4583-6
5	Other	LA46-8

[81249-5](#) Genomic reference sequence coding system [Type]**TERM DEFINITION/DESCRIPTION(S)**

The coding system that is the source for the genomic reference sequence, such as NCBI, Ensembl, and LRG.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)**Part:** [Type](#)

Type

Source: Regenstrief LOINC

EXAMPLE ANSWER LIST ([LL3998-3](#))

SEQ#	Answer	Answer ID
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1	NCBI <i>Description: National Center for Biotechnology Information/NIH</i>	LA26214-9
2	Ensembl	LA26215-6
3	LRG <i>Description: Locus Reference Genomic</i>	LA26216-4
4	Local	LA4583-6
5	Other	LA46-8

81295-8 Local transcript reference sequence coding system OID

TERM DEFINITION/DESCRIPTION(S)

If use of a local coding system is reported for the transcript reference sequence coding system [\[LOINC: 81248-7\]](#), the OID of that coding system should be reported using this term.

Source: Regenstrief LOINC

CONDITION FOR INCLUSION

If the transcript reference sequence is from a local coding system, the OID should be provided.

81296-6 Local genomic reference sequence coding system OID

TERM DEFINITION/DESCRIPTION(S)

If use of a local coding system is reported for the genomic reference sequence coding system [\[LOINC: 81249-5\]](#), the OID of that coding system should be reported using this term.

Source: Regenstrief LOINC

CONDITION FOR INCLUSION

If the genomic reference sequence is from a local coding system, the OID should be provided.

48018-6 Gene [Identifier] in Blood or Tissue

TERM DEFINITION/DESCRIPTION(S)

HUGO Gene Nomenclature Committee (HGNC) identifier for a gene. List the gene(s) examined in full or in part by the study. If the study addresses multiple genes, these can be recorded in one OBX as a list separated by repeat delimiters or in multiple OBX's with one gene per OBX. The recommended coding system will use the HGNC gene symbol as the display text and HGNC gene ID as the code. For example, 21497^ACAD9^HGNC.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)

Identifier

Source: Regenstrief LOINC

ANSWER CARDINALITY

0..*

36908-2 Gene mutations tested for in Blood or Tissue by Molecular genetics method Nominal

TERM DEFINITION/DESCRIPTION(S)

For targeted mutation analysis, report the mutations that the study is designed to detect. These can be recorded in a list separated by repeat delimiters in a single OBX-5 or as separate OBX segments with a single mutation per OBX-5.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Prid](#)

Presence or Identity

Source: Regenstrief LOINC

CONDITION FOR INCLUSION

Either the mutations tested or the range(s) of DNA sequences examined (as structured or narrative data) should be included.

ANSWER CARDINALITY

0..*

51959-5 Range(s) of DNA sequences examined [NumRange]

TERM DEFINITION/DESCRIPTION(S)

This term is used to report the region(s) of interest for sequencing studies as one or more numeric ranges that identify the parts of the reference sequence that are sequenced. These can be recorded as one or more HL7 numeric ranges using repeat delimiters to separate multiple such ranges. They can also be recorded singly, one per OBX, using OBX-4 to distinguish these repeats with the same Observation ID. However, such detailed specification of the sequencing region of interest is rare, in part because this information is often proprietary, and the region of interest is reported as a text description instead, e.g., "Sequenced all of the coding, and appropriate flanking regions," using [\[LOINC: 81293-3\]](#).

Source: Regenstrief LOINC

CONDITION FOR INCLUSION

Either the mutations tested or the range(s) of DNA sequences examined (as structured or narrative data) should be included.

ANSWER CARDINALITY

0..*

81293-3 Range(s) of DNA sequences examined Narrative

TERM DEFINITION/DESCRIPTION(S)

This term is used to report a narrative description of the range(s) of DNA sequences examined in this sequencing study. Genetic test reports only rarely include explicit numeric ranges (which would be reported using [\[LOINC: 51959-5\]](#)) because they are often proprietary, and more often describe the regions examined in narrative. For example, "all coding regions and appropriate flanking regions." To report the region of interest (e.g., in terms of introns and exons) rather than the specific DNA sequences examined, [\[LOINC: 47999-8\]](#) may be used.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Txt](#)

Text

Source: Regenstrief LOINC

CONDITION FOR INCLUSION

Either the mutations tested or the range(s) of DNA sequences examined (as structured or narrative data) should be included.

ANSWER CARDINALITY

0..1

53577-3 Reason for study additional note [Text] in Blood or Tissue by Molecular genetics method Narrative

TERM DEFINITION/DESCRIPTION(S)

The freeform text that is entered by the ordering provider to further annotate the coded Reason for Study [\[LOINC: 51967-8\]](#) associated with an ordered test. In HL7 v2 messages, OBR-31 should be used to report the reason for study.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Txt](#)

Text

Source: Regenstrief LOINC

ANSWER CARDINALITY

0..*

51967-8 Genetic disease assessed [Identifier] in Blood or Tissue by Molecular genetics method

TERM DEFINITION/DESCRIPTION(S)

Coded identifier of the disorder being assessed but with exception to allow the recording of something not included in the controlled vocabulary that is being used. Various coding systems may be used, including ICD-9-CM, ICD-10-CM, SCT and NCBI MedGen.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)

Identifier

Source: Regenstrief LOINC

ANSWER CARDINALITY

0..*

51969-4 Genetic analysis summary report in Blood or Tissue Document by Molecular genetics method

TERM DEFINITION/DESCRIPTION(S)

Narrative report in disease diagnostic-based format.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Find](#)

Finding is an atomic clinical observation, not a summary statement as an impression. Physical, historical, review of systems and other such observations have a property of Finding. These may have a scale of NOM for coded findings or NAR for findings reported in narrative text.

Source: Regenstrief LOINC

81303-0 HGVS version [Identifier]**51968-6** Genetic disease analysis overall interpretation in Blood or Tissue by Molecular genetics method

TERM DEFINITION/DESCRIPTION(S)

Interpretation of all identified DNA Markers and/or Individual Alleles along with any known clinical information for the benefit of aiding clinicians in understanding the results overall. This is used for Symptomatic or Asymptomatic testing other than Carrier testing.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Imp](#)

(impression) is a diagnostic statement, always an interpretation or abstraction of some other observation (a series of test results, an image, or a total patient), and almost always generated by a professional.

Source: Regenstrief LOINC

PREFERRED ANSWER LIST ([LL541-4](#))

SEQ#	Answer	Answer ID
1	Positive	LA6576-8
2	Negative	LA6577-6
3	Inconclusive	LA9663-1
4	Failure	LA9664-9

[81250-3](#) Simple variant panel

TERM DEFINITION/DESCRIPTION(S)

This panel is used to report the information associated with a simple genetic variant, such as a single nucleotide change. It should not be used to report information related to structural variants.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: -

to be used for panels only

Source: Regenstrief LOINC

QUESTION CARDINALITY

0..n

[48003-8](#) DNA sequence variation identifier [Identifier] in Blood or Tissue by Molecular genetics method

TERM DEFINITION/DESCRIPTION(S)

A DNA Marker identifier conveys a universal or standard repository identifier for definitive characteristics of a DNA Marker. (recommend using NCBI dbSNP ids - rs#)

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)

Identifier

Source: Regenstrief LOINC

[81252-9](#) Simple variant [Identifier]

TERM DEFINITION/DESCRIPTION(S)

This term is used to report the unique identifier of the simple variant found in this study. The identifier may come from various sources, including NCBI's ClinVar and Ensembl. For example, the variant NM_014049.4(ACAD9):c.1249C>T (p.Arg417Cys) has the ClinVar ID 30880 and would be reported in OBX-5 as 30880^NM_014049.4(ACAD9):c.1249C>T (p.Arg417Cys)^ClinVar. [<http://www.ncbi.nlm.nih.gov/clinvar/variation/30880/>]

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)

Identifier

Source: Regenstrief LOINC

[81292-5](#) Simple variant [Type]

PART DEFINITION/DESCRIPTION(S)

Part: [Type](#)

Type

Source: Regenstrief LOINC

[48018-6](#) Gene [Identifier] in Blood or Tissue

TERM DEFINITION/DESCRIPTION(S)

HUGO Gene Nomenclature Committee (HGNC) identifier for a gene. List the gene(s) examined in full or in part by the study. If the study addresses multiple genes, these can be recorded in one OBX as a list separated by repeat delimiters or in multiple OBX's with one gene per OBX. The recommended coding system will use the HGNC gene symbol as the display text and HGNC gene ID as the code. For example, 21497^ACAD9^HGNC.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)
 Identifier
 Source: Regenstrief LOINC

[51958-7](#) Transcript reference sequence [Identifier] in Blood or Tissue**TERM DEFINITION/DESCRIPTION(S)**

This field carries the ID for the transcribed reference sequence, which is the part of the genomic reference sequence that is converted to messenger RNA (i.e., after the introns are removed). The transcript reference sequence ID may be reporting using various coding systems including NCBI's RefSeq ("NM_..."), Ensembl ("ENST..."), and LRG ("LRG..." plus "t1" to indicate transcript).
 Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)
 Identifier
 Source: Regenstrief LOINC

[48004-6](#) DNA sequence variation in Blood or Tissue by Molecular genetics method**TERM DEFINITION/DESCRIPTION(S)**

Human Genome Variation Society (HGVS) nomenclature for a single DNA marker. The use of the nomenclature must be extended to describe non-variations (aka. wild types) see samples for wild type examples.
 Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Find](#)
 Finding is an atomic clinical observation, not a summary statement as an impression. Physical, historical, review of systems and other such observations have a property of Finding. These may have a scale of NOM for coded findings or NAR for findings reported in narrative text.
 Source: Regenstrief LOINC

[48005-3](#) Amino acid change in Blood or Tissue by Molecular genetics method**TERM DEFINITION/DESCRIPTION(S)**

Human Genome Variation Society (HGVS) nomenclature for an amino acid sequence. This value is derivable from the DNA Marker value if available. It is provided for convenience. The use of the nomenclature must be extended to describe non-variations (aka. wild types) see samples for wild type examples.
 Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)**[48019-4](#) DNA sequence variation type in Blood or Tissue by Molecular genetics method****TERM DEFINITION/DESCRIPTION(S)**

Codified type for associated DNA Marker. DNA Marker's use the HGVS notation which implies the DNA Marker Type, but the concurrent use of this code will allow a standard and explicit type for technical and display convenience.
 Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Type](#)
 Type
 Source: Regenstrief LOINC

PREFERRED ANSWER LIST ([LL379-9](#))

SEQ#	Answer	Answer ID
1	Wild type	LA9658-1
2	Deletion	LA6692-3
3	Duplication	LA6686-5
4	Insertion	LA6687-3
5	Insertion/Deletion	LA6688-1
6	Inversion	LA6689-9
7	Substitution	LA6690-7

[48006-1](#) Amino acid change type in Blood or Tissue by Molecular genetics method**TERM DEFINITION/DESCRIPTION(S)**

Codified type for associated Amino Acid Marker. Amino Acid Marker's use the HGVS notation which implies the Amino Acid Marker Type, but the concurrent use of this code will allow a standard and explicit type for technical and display convenience.
 Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Type](#)
 Type
 Source: Regenstrief LOINC

PREFERRED ANSWER LIST ([LL380-7](#))

SEQ#	Answer	Answer ID
1	Wild type	LA9658-1
2	Deletion	LA6692-3
3	Duplication	LA6686-5
4	Frameshift	LA6694-9
5	Initiating Methionine	LA6695-6
6	Insertion	LA6687-3
7	Insertion and Deletion	LA9659-9
8	Missense	LA6698-0
9	Nonsense	LA6699-8
10	Silent	LA6700-4
11	Stop Codon Mutation	LA6701-2

[48013-7](#) Genomic reference sequence [Identifier] in Blood or Tissue**TERM DEFINITION/DESCRIPTION(S)**

This field carries the ID for the genomic reference sequence. The genomic reference sequence is a contiguous stretch of chromosome DNA that spans all of the exons of the gene and includes transcribed and non transcribed stretches. For this ID use either the NCBI genomic nucleotide RefSeq IDs with their version number (see: NCBI.NLM.NIH.Gov/RefSeq) or use the LRG identifiers, without transcript (t or p) extensions -- when they become available. (See- Report sponsored by GEN2PHEN at the European Bioinformatics Institute at Hinxton UK April 24-25, 2008).

The NCI RefSeq genomic IDs are distinguished by a prefix of "NG" for genes from the nuclear chromosomes and prefix of "NC" for genes from mitochondria. The LRG Identifiers have a prefix of "LRG_" Mitochondrial genes are not in the scope of LRG

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)

Identifier

Source: Regenstrief LOINC

[69547-8](#) Genomic reference allele [Identifier]**TERM DEFINITION/DESCRIPTION(S)**

Reference values ("normal") examined within the Reference Sequence. This is used in a genotyping test to define the reference and variable nucleotide strings. That is if the sequence variation is an insertion, then Reference Nucleotide will be blank and Variable Nucleotide will contain the inserted nucleotides. In contrast, if the sequence variation is a deletion, then the Reference Nucleotide will contain the deleted nucleotides, and the Variable Nucleotide will be blank.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Prid](#)

Presence or Identity

Source: Regenstrief LOINC

[81254-5](#) Genomic allele location [Identifier]**TERM DEFINITION/DESCRIPTION(S)**

The genomic allele location is the first genomic position in the reference allele that contains a change from the reference allele. For example, for the simple variant NM_014049.4(ACAD9):c.1249C>T (p.Arg417Cys), the genomic allele location is Chr3: 128906220 on Assembly GRCh38. [http://www.ncbi.nlm.nih.gov/clinvar/variation/30880/]

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)

Identifier

Source: Regenstrief LOINC

[69551-0](#) Genomic alternate allele [Identifier]**TERM DEFINITION/DESCRIPTION(S)**

The genomic alternate allele is the contiguous segment of DNA in the test sample that differs from the reference allele at the same location and thus defines a variant.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Prid](#)

Presence or Identity

Source: Regenstrief LOINC

[81255-2](#) dbSNP [Identifier]**TERM DEFINITION/DESCRIPTION(S)**

The unique identifier for the variant represented as a small nucleotide polymorphism (SNP). The dbSNP ID is used routinely as the base identifier in pharmacogenomics as well as arrCGH studies. For example, for the simple variant NM_014049.4(ACAD9):c.1249C>T (p.Arg417Cys), the dbSNP ID is 368949613. [http://www.ncbi.nlm.nih.gov/clinvar/variation/30880/]

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)

Identifier

Source: Regenstrief LOINC

[81256-0](#) COSMIC [Identifier]

TERM DEFINITION/DESCRIPTION(S)

The COSMIC ID is used to report the variant ID for the COSMIC database, which collects information about somatic cancer-related, non-structural mutations. Information in the COSMIC database includes an ID for the tissue source of the submission, a gene name, a reference sequence from Ensembl, pathogenicity, and HGVS names. For example, the COSMIC Mutation ID COSM34090 is for a missense mutation in the IDH2 gene with HGVS nomenclature c.516G>T and p.R172S. [<http://cancer.sanger.ac.uk/cosmic/mutation/overview?id=34090>]

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)

Identifier

Source: Regenstrief LOINC

[81257-8](#) CIGAR [Identifier]

TERM DEFINITION/DESCRIPTION(S)

This term is used to report the unique ID from CIGAR, a syntax for describing variation that is use most frequently during alignment in sequencing studies.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)

Identifier

Source: Regenstrief LOINC

[81258-6](#) Allelic frequency

TERM DEFINITION/DESCRIPTION(S)

The allelic frequency is the relative frequency of a particular allele in the population, expressed as a number from 0 to 1.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [NFr](#)

Number Fraction

Source: Regenstrief LOINC

[48001-2](#) Chromosome region [Identifier] in Blood or Tissue by Molecular genetics method

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)

Identifier

Source: Regenstrief LOINC

[48002-0](#) Genomic source class [Type] in Blood or Tissue by Molecular genetics method

TERM DEFINITION/DESCRIPTION(S)

The genomic class of the specimen being analyzed: Germline for inherited genome, somatic for cancer genome, and prenatal for fetal genome.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Type](#)

Type

Source: Regenstrief LOINC

PREFERRED ANSWER LIST ([LL378-1](#))

SEQ#	Answer	Answer ID
1	Germline	LA6683-2
2	Somatic	LA6684-0
3	Prenatal	LA10429-1
4	Likely germline	LA18194-3
5	Likely somatic	LA18195-0
6	Likely prenatal	LA18196-8
7	Unknown genomic origin	LA18197-6

[53034-5](#) Allelic state in Blood or Tissue by Molecular genetics method

TERM DEFINITION/DESCRIPTION(S)

The level of occurrence of a single DNA Marker within a set of chromosomes. Heterozygous indicates the DNA Marker is only present in one of the two genes contained in homologous chromosomes. Homozygous indicates the DNA Marker is present in both genes contained in homologous chromosomes. Hemizygous indicates the DNA Marker exists in the only single copy of a gene in a non-homologous chromosome (The male X and Y chromosome are non-homologous). Hemiplasmic indicates that the DNA Marker is present in some but not all of the copies of mitochondrial DNA. Homoplasmic indicates that the DNA Marker is present in all of the copies of mitochondrial DNA.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Find](#)

Finding is an atomic clinical observation, not a summary statement as an impression. Physical, historical, review of systems and other such observations have a property of Finding. These may have a scale of NOM for coded findings or NAR for findings reported in narrative text.

Source: Regenstrief LOINC

PREFERRED ANSWER LIST ([LL381-5](#))

SEQ#	Answer	Answer ID
1	Heteroplasmic	LA6703-8
2	Homoplasmic	LA6704-6
3	Homozygous	LA6705-3
4	Heterozygous	LA6706-1
5	Hemizygous	LA6707-9

[53037-8](#) Genetic disease sequence variation interpretation in Blood or Tissue by Molecular genetics method**TERM DEFINITION/DESCRIPTION(S)**

Single DNA Marker or Individual Allele interpretation in the context of the assessed genetic disease..

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Imp](#)

(impression) is a diagnostic statement, always an interpretation or abstraction of some other observation (a series of test results, an image, or a total patient), and almost always generated by a professional.

Source: Regenstrief LOINC

PREFERRED ANSWER LIST ([LL603-2](#))

SEQ#	Answer	Answer ID
1	Pathogenic	LA6668-3
2	Presumed Pathogenic	LA6669-1
3	Unknown Significance	LA6682-4
4	Benign	LA6675-8
5	Presumed Benign	LA6674-1

[81259-4](#) Associated phenotype**TERM DEFINITION/DESCRIPTION(S)**

The possible phenotype associated with the genetic variant found in this study.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Find](#)

Finding is an atomic clinical observation, not a summary statement as an impression. Physical, historical, review of systems and other such observations have a property of Finding. These may have a scale of NOM for coded findings or NAR for findings reported in narrative text.

Source: Regenstrief LOINC

[81251-1](#) Complex variant panel**TERM DEFINITION/DESCRIPTION(S)**

This panel is used to report information related to a complex genetic variant and includes a repeating subpanel for reporting specific information for each simple variation that the complex variant includes.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: -

to be used for panels only

Source: Regenstrief LOINC

QUESTION CARDINALITY

0..n

[48008-7](#) Allele name [Identifier] in Blood or Tissue by Molecular genetics method**TERM DEFINITION/DESCRIPTION(S)**

The published and commonly used name for a gene allele is recommended. For unpublished or non-established alleles, logical assumptions and conventions may be

used.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)

Identifier

Source: Regenstrief LOINC

[81260-2](#) Complex variant [Identifier]

TERM DEFINITION/DESCRIPTION(S)

This term is used to report the unique identifier of the complex variant found in this study. The identifier may come from various sources, including NCBI's ClinVar and Ensembl. For example, the variant NM_000106.5(CYP2D6):c.[886C>T;457G>C] – Haplotype has the ClinVar ID 16895. [<http://www.ncbi.nlm.nih.gov/clinvar/variation/16895/>]

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)

Identifier

Source: Regenstrief LOINC

[81262-8](#) Complex variant HGVS name

TERM DEFINITION/DESCRIPTION(S)

This term is used to report the name of the complex variant found in this study in HGVS format. For example, c.[886C>T;457G>C], which represents two separate base substitutions in one gene on one chromosome, or c.[886C>T];[457G>C], which represents two separate base substitutions in one gene on two different chromosomes.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Find](#)

Finding is an atomic clinical observation, not a summary statement as an impression. Physical, historical, review of systems and other such observations have a property of Finding. These may have a scale of NOM for coded findings or NAR for findings reported in narrative text.

Source: Regenstrief LOINC

[81263-6](#) Complex variant type

TERM DEFINITION/DESCRIPTION(S)

The type of complex variant, for example, compound heterozygous or haplotype.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Type](#)

Type

Source: Regenstrief LOINC

EXAMPLE ANSWER LIST ([LL3999-1](#))

SEQ#	Answer	Answer ID
1	Compound heterozygous	LA26217-2
2	Double heterozygous	LA26220-6
3	Haplotype	LA26218-0
4	Hemizygous	LA6707-9

[81259-4](#) Associated phenotype

TERM DEFINITION/DESCRIPTION(S)

The possible phenotype associated with the genetic variant found in this study.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Find](#)

Finding is an atomic clinical observation, not a summary statement as an impression. Physical, historical, review of systems and other such observations have a property of Finding. These may have a scale of NOM for coded findings or NAR for findings reported in narrative text.

Source: Regenstrief LOINC

[53037-8](#) Genetic disease sequence variation interpretation in Blood or Tissue by Molecular genetics method

TERM DEFINITION/DESCRIPTION(S)

Single DNA Marker or Individual Allele interpretation in the context of the assessed genetic disease..

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Imp](#)

(impression) is a diagnostic statement, always an interpretation or abstraction of some other observation (a series of test results, an image, or a total patient), and almost always generated by a professional.

Source: Regenstrief LOINC

PREFERRED ANSWER LIST ([LL603-2](#))

SEQ#	Answer	Answer ID
1	Pathogenic	LA6668-3
2	Presumed Pathogenic	LA6669-1
3	Unknown Significance	LA6682-4
4	Benign	LA6675-8
5	Presumed Benign	LA6674-1

53034-5 Allelic state in Blood or Tissue by Molecular genetics method**TERM DEFINITION/DESCRIPTION(S)**

The level of occurrence of a single DNA Marker within a set of chromosomes. Heterozygous indicates the DNA Marker is only present in one of the two genes contained in homologous chromosomes. Homozygous indicates the DNA Marker is present in both genes contained in homologous chromosomes. Hemizygous indicates the DNA Marker exists in the only single copy of a gene in a non-homologous chromosome (The male X and Y chromosome are non-homologous). Hemiplasmic indicates that the DNA Marker is present in some but not all of the copies of mitochondrial DNA. Homoplasmic indicates that the DNA Marker is present in all of the copies of mitochondrial DNA.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)**Part:** [Find](#)

Finding is an atomic clinical observation, not a summary statement as an impression. Physical, historical, review of systems and other such observations have a property of Finding. These may have a scale of NOM for coded findings or NAR for findings reported in narrative text.

Source: Regenstrief LOINC

PREFERRED ANSWER LIST ([LL381-5](#))

SEQ#	Answer	Answer ID
1	Heteroplasmic	LA6703-8
2	Homoplasmic	LA6704-6
3	Homozygous	LA6705-3
4	Heterozygous	LA6706-1
5	Hemizygous	LA6707-9

81250-3 Simple variant panel**TERM DEFINITION/DESCRIPTION(S)**

This panel is used to report the information associated with a simple genetic variant, such as a single nucleotide change. It should not be used to report information related to structural variants.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)**Part:** -

to be used for panels only

Source: Regenstrief LOINC

QUESTION CARDINALITY

0..n

48003-8 DNA sequence variation identifier [Identifier] in Blood or Tissue by Molecular genetics method**TERM DEFINITION/DESCRIPTION(S)**

A DNA Marker identifier conveys a universal or standard repository identifier for definitive characteristics of a DNA Marker. (recommend using NCBI dbSNP ids - rs#)

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)**Part:** [ID](#)

Identifier

Source: Regenstrief LOINC

81252-9 Simple variant [Identifier]**TERM DEFINITION/DESCRIPTION(S)**

This term is used to report the unique identifier of the simple variant found in this study. The identifier may come from various sources, including NCBI's ClinVar and Ensembl. For example, the variant NM_014049.4(ACAD9):c.1249C>T (p.Arg417Cys) has the ClinVar ID 30880 and would be reported in OBX-5 as 30880^NM_014049.4(ACAD9):c.1249C>T (p.Arg417Cys)^ClinVar. [<http://www.ncbi.nlm.nih.gov/clinvar/variation/30880/>]

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)
 Identifier
 Source: Regenstrief LOINC

[81292-5](#) Simple variant [Type]

PART DEFINITION/DESCRIPTION(S)

Part: [Type](#)
 Type
 Source: Regenstrief LOINC

[48018-6](#) Gene [Identifier] in Blood or Tissue

TERM DEFINITION/DESCRIPTION(S)

HUGO Gene Nomenclature Committee (HGNC) identifier for a gene. List the gene(s) examined in full or in part by the study. If the study addresses multiple genes, these can be recorded in one OBX as a list separated by repeat delimiters or in multiple OBX's with one gene per OBX. The recommended coding system will use the HGNC gene symbol as the display text and HGNC gene ID as the code. For example, 21497^ACAD9^HGNC.
 Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)
 Identifier
 Source: Regenstrief LOINC

[51958-7](#) Transcript reference sequence [Identifier] in Blood or Tissue

TERM DEFINITION/DESCRIPTION(S)

This field carries the ID for the transcribed reference sequence, which is the part of the genomic reference sequence that is converted to messenger RNA (i.e., after the introns are removed). The transcript reference sequence ID may be reporting using various coding systems including NCBI's RefSeq ("NM_..."), Ensembl ("ENST..."), and LRG ("LRG..." plus "t1" to indicate transcript).
 Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)
 Identifier
 Source: Regenstrief LOINC

[48004-6](#) DNA sequence variation in Blood or Tissue by Molecular genetics method

TERM DEFINITION/DESCRIPTION(S)

Human Genome Variation Society (HGVS) nomenclature for a single DNA marker. The use of the nomenclature must be extended to describe non-variations (aka. wild types) see samples for wild type examples.
 Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Find](#)
 Finding is an atomic clinical observation, not a summary statement as an impression. Physical, historical, review of systems and other such observations have a property of Finding. These may have a scale of NOM for coded findings or NAR for findings reported in narrative text.
 Source: Regenstrief LOINC

[48019-4](#) DNA sequence variation type in Blood or Tissue by Molecular genetics method

TERM DEFINITION/DESCRIPTION(S)

Codified type for associated DNA Marker. DNA Marker's use the HGVS notation which implies the DNA Marker Type, but the concurrent use of this code will allow a standard and explicit type for technical and display convenience.
 Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Type](#)
 Type
 Source: Regenstrief LOINC

PREFERRED ANSWER LIST ([LL379-9](#))

SEQ#	Answer	Answer ID
1	Wild type	LA9658-1
2	Deletion	LA6692-3
3	Duplication	LA6686-5
4	Insertion	LA6687-3
5	Insertion/Deletion	LA6688-1
6	Inversion	LA6689-9
7	Substitution	LA6690-7

48005-3 Amino acid change in Blood or Tissue by Molecular genetics method**TERM DEFINITION/DESCRIPTION(S)**

Human Genome Variation Society (HGVS) nomenclature for an amino acid sequence. This value is derivable from the DNA Marker value if available. It is provided for convenience. The use of the nomenclature must be extended to describe non-variations (aka. wild types) see samples for wild type examples.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)**48006-1 Amino acid change type in Blood or Tissue by Molecular genetics method****TERM DEFINITION/DESCRIPTION(S)**

Codified type for associated Amino Acid Marker. Amino Acid Marker's use the HGVS notation which implies the Amino Acid Marker Type, but the concurrent use of this code will allow a standard and explicit type for technical and display convenience.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Type](#)

Type

Source: Regenstrief LOINC

PREFERRED ANSWER LIST (LL380-7)

SEQ#	Answer	Answer ID
1	Wild type	LA9658-1
2	Deletion	LA6692-3
3	Duplication	LA6686-5
4	Frameshift	LA6694-9
5	Initiating Methionine	LA6695-6
6	Insertion	LA6687-3
7	Insertion and Deletion	LA9659-9
8	Missense	LA6698-0
9	Nonsense	LA6699-8
10	Silent	LA6700-4
11	Stop Codon Mutation	LA6701-2

48013-7 Genomic reference sequence [Identifier] in Blood or Tissue**TERM DEFINITION/DESCRIPTION(S)**

This field carries the ID for the genomic reference sequence. The genomic reference sequence is a contiguous stretch of chromosome DNA that spans all of the exons of the gene and includes transcribed and non transcribed stretches. For this ID use either the NCBI genomic nucleotide RefSeq IDs with their version number (see: NCBI.NLM.NIH.Gov/RefSeq) or use the LRG identifiers, without transcript (t or p) extensions -- when they become available. (See- Report sponsored by GEN2PHEN at the European Bioinformatics Institute at Hinxton UK April 24-25, 2008).

The NCI RefSeq genomic IDs are distinguished by a prefix of "NG" for genes from the nuclear chromosomes and prefix of "NC" for genes from mitochondria. The LRG Identifiers have a prefix of "LRG_" Mitochondrial genes are not in the scope of LRG

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)

Identifier

Source: Regenstrief LOINC

69547-8 Genomic reference allele [Identifier]**TERM DEFINITION/DESCRIPTION(S)**

Reference values ("normal") examined within the Reference Sequence. This is used in a genotyping test to define the reference and variable nucleotide strings. That is if the sequence variation is an insertion, then Reference Nucleotide will be blank and Variable Nucleotide will contain the inserted nucleotides. In contrast, if the sequence variation is a deletion, then the Reference Nucleotide will contain the deleted nucleotides, and the Variable Nucleotide will be blank.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Prid](#)

Presence or Identity

Source: Regenstrief LOINC

81254-5 Genomic allele location [Identifier]**TERM DEFINITION/DESCRIPTION(S)**

The genomic allele location is the first genomic position in the reference allele that contains a change from the reference allele. For example, for the simple variant NM_014049.4(ACAD9):c.1249C>T (p.Arg417Cys), the genomic allele location is Chr3: 128906220 on Assembly GRCh38.

[http://www.ncbi.nlm.nih.gov/clinvar/variation/30880/]

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)

Identifier

Source: Regenstrief LOINC

69551-0 Genomic alternate allele [Identifier]**TERM DEFINITION/DESCRIPTION(S)**

The genomic alternate allele is the contiguous segment of DNA in the test sample that differs from the reference allele at the same location and thus defines a variant.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)**Part:** [Prid](#)

Presence or Identity

Source: Regenstrief LOINC

81255-2 dbSNP [Identifier]**TERM DEFINITION/DESCRIPTION(S)**

The unique identifier for the variant represented as a small nucleotide polymorphism (SNP). The dbSNP ID is used routinely as the base identifier in pharmacogenomics as well as arrCGH studies. For example, for the simple variant NM_014049.4(ACAD9):c.1249C>T (p.Arg417Cys), the dbSNP ID is 368949613. [<http://www.ncbi.nlm.nih.gov/clinvar/variation/30880/>]

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)**Part:** [ID](#)

Identifier

Source: Regenstrief LOINC

81256-0 COSMIC [Identifier]**TERM DEFINITION/DESCRIPTION(S)**

The COSMIC ID is used to report the variant ID for the COSMIC database, which collects information about somatic cancer-related, non-structural mutations. Information in the COSMIC database includes an ID for the tissue source of the submission, a gene name, a reference sequence from Ensembl, pathogenicity, and HGVS names. For example, the COSMIC Mutation ID COSM34090 is for a missense mutation in the IDH2 gene with HGVS nomenclature c.516G>T and p.R172S. [<http://cancer.sanger.ac.uk/cosmic/mutation/overview?id=34090>]

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)**Part:** [ID](#)

Identifier

Source: Regenstrief LOINC

81257-8 CIGAR [Identifier]**TERM DEFINITION/DESCRIPTION(S)**

This term is used to report the unique ID from CIGAR, a syntax for describing variation that is use most frequently during alignment in sequencing studies.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)**Part:** [ID](#)

Identifier

Source: Regenstrief LOINC

81258-6 Allelic frequency**TERM DEFINITION/DESCRIPTION(S)**

The allelic frequency is the relative frequency of a particular allele in the population, expressed as a number from 0 to 1.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)**Part:** [NFr](#)

Number Fraction

Source: Regenstrief LOINC

48001-2 Chromosome region [Identifier] in Blood or Tissue by Molecular genetics method**PART DEFINITION/DESCRIPTION(S)****Part:** [ID](#)

Identifier

Source: Regenstrief LOINC

48002-0 Genomic source class [Type] in Blood or Tissue by Molecular genetics method

TERM DEFINITION/DESCRIPTION(S)

The genomic class of the specimen being analyzed: Germline for inherited genome, somatic for cancer genome, and prenatal for fetal genome.
Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Type](#)

Type
Source: Regenstrief LOINC

PREFERRED ANSWER LIST (LL378-1)

SEQ#	Answer	Answer ID
1	Germline	LA6683-2
2	Somatic	LA6684-0
3	Prenatal	LA10429-1
4	Likely germline	LA18194-3
5	Likely somatic	LA18195-0
6	Likely prenatal	LA18196-8
7	Unknown genomic origin	LA18197-6

53034-5 Allelic state in Blood or Tissue by Molecular genetics method**TERM DEFINITION/DESCRIPTION(S)**

The level of occurrence of a single DNA Marker within a set of chromosomes. Heterozygous indicates the DNA Marker is only present in one of the two genes contained in homologous chromosomes. Homozygous indicates the DNA Marker is present in both genes contained in homologous chromosomes. Hemizygous indicates the DNA Marker exists in the only single copy of a gene in a non-homologous chromosome (The male X and Y chromosome are non-homologous). Hemiplasmic indicates that the DNA Marker is present in some but not all of the copies of mitochondrial DNA. Homoplasmic indicates that the DNA Marker is present in all of the copies of mitochondrial DNA.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Find](#)

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Source: Regenstrief LOINC

PREFERRED ANSWER LIST (LL381-5)

SEQ#	Answer	Answer ID
1	Heteroplasmic	LA6703-8
2	Homoplasmic	LA6704-6
3	Homozygous	LA6705-3
4	Heterozygous	LA6706-1
5	Hemizygous	LA6707-9

53037-8 Genetic disease sequence variation interpretation in Blood or Tissue by Molecular genetics method**TERM DEFINITION/DESCRIPTION(S)**

Single DNA Marker or Individual Allele interpretation in the context of the assessed genetic disease..

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Imp](#)

(impression) is a diagnostic statement, always an interpretation or abstraction of some other observation (a series of test results, an image, or a total patient), and almost always generated by a professional.

Source: Regenstrief LOINC

PREFERRED ANSWER LIST (LL603-2)

SEQ#	Answer	Answer ID
1	Pathogenic	LA6668-3
2	Presumed Pathogenic	LA6669-1
3	Unknown Significance	LA6682-4
4	Benign	LA6675-8
5	Presumed Benign	LA6674-1

81259-4 Associated phenotype**TERM DEFINITION/DESCRIPTION(S)**

The possible phenotype associated with the genetic variant found in this study.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Find](#)

Finding is an atomic clinical observation, not a summary statement as an impression. Physical, historical, review of systems and other such observations have a property of Finding. These may have a scale of NOM for coded findings or NAR for findings reported in narrative text.

Source: Regenstrief LOINC

[81297-4](#) Structural variant panel

PART DEFINITION/DESCRIPTION(S)

Part: [-](#)
to be used for panels only
Source: Regenstrief LOINC

[81286-7](#) Structural variant [Identifier]

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)
Identifier
Source: Regenstrief LOINC

[81287-5](#) Structural variant reported start-end [NumRange]

TERM DEFINITION/DESCRIPTION(S)

Reported boundaries (start-end) of the structural variant in genomic coordinates relative to the reference sequence.
Source: Regenstrief LOINC

[81288-3](#) Precision of boundaries Nominal

TERM DEFINITION/DESCRIPTION(S)

The precision of the nucleotide boundaries that can be specified for a structural variant can vary widely, and in some cases the boundary can only be pinpointed down to a region that is on the order of megabases. This term should be used to report the narrative description of the boundary precision. The nucleotide ranges for the narrowest and widest possible boundaries of a structural variant can be reported using [\[LOINC: 81302-2\]](#) and [\[LOINC: 81301-4\]](#), respectively.
Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Find](#)
Finding is an atomic clinical observation, not a summary statement as an impression. Physical, historical, review of systems and other such observations have a property of Finding. These may have a scale of NOM for coded findings or NAR for findings reported in narrative text.
Source: Regenstrief LOINC

[81299-0](#) Structural variant reported arrCGH [Ratio]

PART DEFINITION/DESCRIPTION(S)

Part: [Ratio](#)
One measure divided by another that comes from the same system.
Source: Regenstrief LOINC

[81289-1](#) Structural variant [Type]

PART DEFINITION/DESCRIPTION(S)

Part: [Type](#)
Type
Source: Regenstrief LOINC

[81300-6](#) Structural variant [Length]

TERM DEFINITION/DESCRIPTION(S)

Length of the structural variant, which information may be ascertained in some but not all types of structural variants.
Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Len](#)
Length
Source: Regenstrief LOINC

[81301-4](#) Structural variant outer start-end [NumRange]

TERM DEFINITION/DESCRIPTION(S)

The genomic coordinates of the widest genomic range in which the structural variant might reside.
Source: Regenstrief LOINC

[81302-2](#) Structural variant inner start-end [NumRange]

TERM DEFINITION/DESCRIPTION(S)

The genomic coordinates of the narrowest genomic range in which the structural variant might reside.

Source: Regenstrief LOINC

[81290-9](#) Structural variant HGVS name

TERM DEFINITION/DESCRIPTION(S)

The name of a structural variant reported using HGVS nomenclature.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Find](#)

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Source: Regenstrief LOINC

[81291-7](#) Structural variant ISCN name

TERM DEFINITION/DESCRIPTION(S)

ISCN is a syntax for describing cytogenetic findings, from classical karyotypes to details that can be observed with copy number methodologies. Using ISCN nomenclature is highly recommended for reporting structural variants.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [Find](#)

Finding is an atomic clinical observation, not a summary statement as an impression. Physical, historical, review of systems and other such observations have a property of Finding. These may have a scale of NOM for coded findings or NAR for findings reported in narrative text.

Source: Regenstrief LOINC

[81298-2](#) Structural variant cytogenetic location [Identifier]

TERM DEFINITION/DESCRIPTION(S)

The chromosome region where the structural variant was found, for example, 17p12. This term may be used when more specific information, such as the structural variant reported start-end [\[LOINC: 81287-5\]](#), precision of boundaries [\[LOINC: 81288-3\]](#) and inner and outer start-end [\[LOINC: 81302-2\]](#) and [\[LOINC: 81301-4\]](#), respectively, cannot be or are not reported.

Source: Regenstrief LOINC

PART DEFINITION/DESCRIPTION(S)

Part: [ID](#)

Identifier

Source: Regenstrief LOINC

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