Standards for Pathology Informatics in Australia (SPIA)

Reporting Terminology and Codes Genetic Pathology

(v3.0)

Superseding and incorporating the
Australian Pathology Units and Terminology Standards
and Guidelines (APUTS)

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Standards for Pathology Informatics in Australia (SPIA)

Previously known as the Australian Pathology Units and Terminology Standards and Guidelines (APUTS)

Generic Genetic Pathology Report Information Model Terminology

TRIM Document #: 156347

Document History:

Version	Reason for Change	Author	Date
1.0	Initial Terminology for Microbiology: Organisms mapped to SNOMED CT, published by the RCPA Pathology Units and Terminology Standardisation Project.	Michael Legg / Dr Christiaan Swanepoel	12-Feb-13
2.0	Added Document Revision History worksheet. Changed file name from PUTS-Genetics-Report-information-model to APUTS Genetics Report terminology reference set.	Donna Moore	06-Jun-14
2.1	After public feedback and approval by PITUS steering committee the following changes were made: - added comment for missing LOINC codes marked as 'XXXXX-X'. Comment added was 'LOINC code pending. Currently no code is available.'	Donna Moore	28-Oct-14
2.2	Add licence information.	Donna Moore	2-May-16
3.0	Added 'Version' and 'History' columns.	Donna Moore	14-Nov-16

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Generic Genetic Pathology Report Information Model Terminology

Generic Genetic Pathology Report Information Model Terminology												
Preferred term	**Example answers / response / Comments**	**LOINC**	**Component**	**Property**	**Timing**	**System**	**Scale**	**Method**	**Class**	**LongName**	**Version**	**History**
Specimen		31208-2	Specimen source	Prid	Pt	XXX	Nom		SPEC	Specimen source [Identifier] of Unspecified specimen	1.0	Added by PUTS project
	Specimen Type must be stated: This could be any of blood, cells, fluids [state type], and could include tissue, frozen sample, formalin fixed paraffin Embedded sample. etc *Note the complication that a sample from a fetus may contain DNA from both the fetus AND the mother*											
Request												
Genetic disease/condition assessed		51967-8	Genetic disease assessed	ID	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	Genetic disease assessed [Identifier] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
	A coded disease (recommend SNOMED) which is known to be caused by or identified by genomic DNA Markers. *ex: SCTID: 190905008 | Cystic fibrosis |* *alternative coding: HGNC:1881 | cystic fibrosis modifier 1*											
Clinical question		53577-3	Reason for study additional note	Txt	Pt	Bld/Tiss	Nar	Molgen	HL7.GENETICS	Reason for study additional note [Text] in Blood or Tissue by Molecular genetics method Narrative	1.0	Added by PUTS project
	The freeform text that entered by orderer to further annotate the coded Reason for Study associated with an ordered test. *Note: Although "Clinical Question" is shown as freeform text in this version, it is anticipated that a future "structured request framework" will be developed*											
Test												
Genetic Test												
Genomic Source class		48004-6	DNA sequence variation	Find	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	DNA sequence variation in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
	The genomic class of the specimen being analyzed:											
Genetic Level		XXXXX-X										
	Whole Genome *Chromosome* *Intergenic* *Gene*											
Gene Name or Locus												
Coding System		XXXXX-X										
HGNC Gene ID		48018-6	Gene identifier	ID	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	Gene [Identifier] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
	e.g. BRCA1 => HGNC:1100											
NCBI DNA Sequence Variation Number (dbSNP ids - rs#)		48003-8	DNA sequence variation identifier	ID	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	DNA sequence variation identifier [Identifier] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
	A DNA Marker identifier conveys a universal or standard repository identifier for definitive characterstics of a DNA Marker. (recommend using NCBI dbSNP ids - rs#)											
NCBI Genomic reference sequence		48013-7	Genomic reference sequence identifier	ID	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	Genomic reference sequence [Identifier] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
	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 *DMD (ref. sequence NM_000109)* *CFTR (ref. sequence NM_000492)* *HTT (ref. sequence NM_002111)*												
Code (actual code for the genetic test)		XXXXX-X										
	HGNC:1100 for BRCA1 *NM_000109 for DMD (ref. sequence)*											
Additional information		XXXXX-X										
Genetic Test Method		XXXXX-X										
Category		XXXXX-X										
Mutational analysis		XXXXX-X										
	The intention is to expand each of the following by one sublevel analogous to Chromosomal conditions so as to hold more detailed information relevant to each Type of Mutational Analysis (Next level of detail for these Mutational Analysis tests has not vet been shown)											
Tests for multiple mutations ...		XXXXX-X										
Test for selected mutations only ...		XXXXX-X										
Assay for size of triplet repeat only ...		XXXXX-X										
Chromosome microarray (CMA)												
Microarray platform		62375-1	Microarray platform	ID	Pt	Bld/Tiss	Nar	Molgen	HL7.CYTOGEN	Microarray platform [Identifier] in Blood or Tissue by Molecular genetics method Narrative	1.0	Added by PUTS project
Microarray platform version number		62376-9	Microarray platform version number	ID	Pt	Bld/Tiss	Nar	Molgen	HL7.CYTOGEN	Microarray platform version number in Blood or Tissue by Molecular genetics method Narrative	1.0	Added by PUTS project
Base pair start coordinate		62381-9	Base pair start coordinate	Num	Pt	Bld/Tiss	Qn	Molgen	HL7.CYTOGEN	Base pair start coordinate [#] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Base pair end coordinate		62381-9	Base pair end coordinate	Num	Pt	Bld/Tiss	Qn	Molgen	HL7.CYTOGEN	Base pair end coordinate [#] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Flanking normal region before start		62382-7	Flanking normal region before start	Find	Pt	Bld/Tiss	Nom	Molgen	HL7.CYTOGEN	Flanking normal region before start in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Analysis by Chromosomal Banding												
ISCN band level [#]		62358-7	ISCN band level	Num	Pt	Bld/Tiss	Ord	Molgen	HL7.CYTOGEN	ISCN band level [#] in Blood or Tissue Qualitative by Molecular genetics method	1.0	Added by PUTS project

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Generic Genetic Pathology Report Information Model Terminology

Generic Genetic Pathology Report Information Model Terminology												
Preferred term	**Example answers / response / Comments**	**LOINC**	**Component**	**Property**	**Timing**	**System**	**Scale**	**Method**	**Class**	**LongName**	**Version**	**History**
Chromosome band involved start		62379-3	Chromosome band involved start	Find	Pt	Bld/Tiss	Nom	Molgen	HL7.CYTOGEN	Chromosome band involved start in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Chromosome band involved end		62380-1	Chromosome band involved end	Find	Pt	Bld/Tiss	Nom	Molgen	HL7.CYTOGEN	Chromosome band involved end in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Chromosome banding method		62359-5	Chromosome banding method	Type	Pt	Bld/Tiss	Nom	Molgen	HL7.CYTOGEN	Chromosome banding method [Type] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Cells analyzed [#]		62360-3	Cells analyzed	Num	Pt	Bld/Tiss	Qn	Molgen	HL7.CYTOGEN	Cells analyzed [#] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Cells counted [#]		62361-1	Cells counted	Num	Pt	Bld/Tiss	Qn	Molgen	HL7.CYTOGEN	Cells counted [#] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Cells karyotyped.total [#]		55199-4	Cells karyotyped.total	Num	Pt	Bld/Tiss	Qn		MOLPATH	Cells karyotyped.total [#] in Blood or Tissue	1.0	Added by PUTS project
Colonies counted [#]		62362-9	Colonies counted	Num	Pt	Bld/Tiss	Qn	Molgen	HL7.CYTOGEN	Colonies counted [#] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Mosaicism detected		62363-7	Mosaicism detected	Find	Pt	Bld/Tiss	Nom	Molgen	HL7.CYTOGEN	Mosaicism detected in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Analysis by in situ hybridisation												
Cell phase		62368-6	Cell phase	Type	Pt	Bld/Tiss	Nom	Molgen	HL7.CYTOGEN	Cell phase [Type] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Probe gene name		62370-2	FISH probe gene name	ID	Pt	Bld/Tiss	Nom	Molgen	HL7.CYTOGEN	FISH probe gene name [Identifier] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Probe locus		62371-0	FISH probe locus	ID	Pt	Bld/Tiss	Nom	Molgen	HL7.CYTOGEN	FISH probe locus [Identifier] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Probe vendor		62372-8	FISH probe vendor	ID	Pt	Bld/Tiss	Nom	Molgen	HL7.CYTOGEN	FISH probe vendor [Identifier] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Cells analysed		62360-3	Cells analyzed	Num	Pt	Bld/Tiss	Qn	Molgen	HL7.CYTOGEN	Cells analyzed [#] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Other												
	Details of test method not already specified (e.g. MLPA or direct DNA sequencing or list mutations searched for)											
Linkage studies		XXXXX-X										
Genetic Test Result												
Cytogenetics												
Karyotype analysis result in ISCN expression		62356-1	Chromosome analysis result in ISCN expressio	Find	Pt	Bld/Tiss	Nom	Molgen	HL7.CYTOGEN	Chromosome analysis result in ISCN expression in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
	47,XY,+2 *46,XY,-18,+der(13q18q),t(13;18)(13q18q;13p18p)pat*											
Microarray analysis result in ISCN expression		XXXXX-X										
	arr 16p13.11(14,817,706-16,649,713)x3 dn *arr 1p36.22p31.3(9,240,258-67,037,576)x2 hmz*											
Molecular Genetics												
DNA sequence variation type		48019-4	DNA sequence variation type	Type	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	DNA sequence variation type in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
	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. *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*											
HGVS DNA Sequence Variation		48004-6	DNA sequence variation	Find	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	DNA sequence variation in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
	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. "Massively Parallel Sequencing [NGS/WGS] is anticipated to go here, but we have postponed specific inclusion pending the outcome of the MPS Working Party recommendations *e.g. c.3869A>C heterozygote*											
DNA region name		47999-8	DNA region name	ID	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	DNA region name [Identifier] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
	A human readable name for the region of interest. Typically Exon #, Intron # or other. NOTE: This is not standardized and is mainly for convenience and display purposes.											
RNA												
	This section is anticipated to hold the results of RNA-specific changes, such as RNA Editing, Transcriptomic results, etc											
Amino acid change		48005-3	Amino acid change	Find	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	Amino acid change in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
	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.											
Genomic RefSeq String		48013-7	Genomic reference sequence identifier	ID	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	Genomic reference sequence [Identifier] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
	Definition: Carries the literal string that represents the DNA sequence that is the basis for deciding what is a different (a variation) in the study subject The same information can also be reported more compactly by reference (see LOINC code 48013-7) if the reference sequence has been catalogued by a registration authority such as RefSeq at NCBI a. Reference sequences can be quite long *Example answer: 241 GGAGTAATCA GCAACTCAGG GGGACCTGTA CGAGTCTATA GCCTACCTGG TCGAGAAAAC 301 TATTCCTCAG TAGATGCCAA TGGCATCCAG TGTCAAATCC TTTCTACATC GTGTCCTTCT 361 TTCACACTAA GTA*											
Amino acid change type		48006-1	Amino acid change type	Type	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	Amino acid change type in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Allelic State		53034-5	Allelic state	Find	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	Allelic state in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
	"Includes Copy Number Variation [CNV], Loss of Heterozygosity [LOH], Quantitative gene dosage"											
Result modifier		62364-5	Test performance information	Find	Pt	XXX	Nar		HL7.CYTOGEN	Test performance information in Unspecified specimen Narrative	1.0	Added by PUTS project
RNA/Protein consequences												
Reports												
Co-occurrence with deleterious mutations												

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Preferred term	**Example answers / response / Comments**	**LOINC**	**Component**	**Property**	**Timing**	**System**	**Scale**	**Method**	**Class**	**LongName**	**Version**	**History**
Species conservation												
Biochemical												
	This modifier can be used to correlate genetic findings with (e.g.) pharmacogenomic or biochemical results											
Biophysical change												
In-silico analyses												
RNA Studies												
Functional studies												
Control data												
Segregation data												
Loss of normal allele in cancer												
Genetic Test Interpretation												
Limitations and Disclaimers (compulsory)		XXXXX-X										
	This section would describe the status of the result. Has it been validated? Is it research-use only? Are there known weaknesses? Is this study partial, in progress, complete, etc											
Result interpretation (compulsory)												
	This section summarises the laboratory findings. What were the results of testing? What are the biological and genetic implications of this result?											
Genetic Screen findings (optional)		19102-3	Genetic screen	Find	Pt	XXX	Nar	Molgen	MOLPATH	Genetic screen in Unspecified specimen by Molecular genetics method Narrative	1.0	Added by PUTS project
Carrier status summary (optional)		53039-4	Genetic disease analysis overall carrier interpr	Imp	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	Genetic disease analysis overall carrier interpretation [interpretation] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
	Carrier Identification 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.											
Sequence variation summary (optional)		53037-8	Genetic disease sequence variation interpreta	Imp	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	Genetic disease sequence variation interpretation [interpretation] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
Clinical Interpretation (compulsory)		51968-6	Genetic disease analysis overall interpretation	Imp	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	Genetic disease analysis overall interpretation [interpretation] in Blood or Tissue by Molecular genetics method	1.0	Added by PUTS project
	This section summarises the clinical significance (if any) of the laboratory findings. It can take into account Family History or Clinical History.											
Genotype clinical interpretation (optional)												
Phenotype clinical interpretation (optional)												
Conclusion (optional)		62365-2	Diagnostic impression	Imp	Pt	XXX	Nar	Molgen	HL7.CYTOGEN	Diagnostic impression [interpretation] in Unspecified specimen by Molecular genetics method Narrative	1.0	Added by PUTS project
	This is the final "tie it all together" field, if not already done so above.											
Interpretation modifier		XXXXX-X										
	As for Result modifier, see above											


Genetic Pathology Report Format
Specimen (compulsory)
Specimen Type must be stated: This could be any of blood, cells, fluids [state type], and could include tissue, frozen sample, formalin fixed paraffin Embedded sample. etc
Note the complication that a sample from a fetus may contain DNA from both the fetus AND the mother
Request
Genetic disease / condition assessed
"A coded disease (recommend SNOMED) which is known to be caused by or identified by genomic DNA Markers. eg SCT 190905008 | Cystic fibrosis |
Clinical question
Freeform text entered by requester to further annotate the coded Reason for Study associated with an ordered test.
Note : Although "Clinical Question" is shown as freeform text in this version, it is anticipated that a future "structured request framework" will be developed
Test
Genetic Test
Genomic Source class
Genetic Level
Whole Genome
Chromosome
Intergenic
Gene
Gene Name or Locus
Coding System
Code (actual code for the genetic test)
Additional information
Genetic Test Method
Category
Mutational analysis
The intention is to expand each of the following by one sublevel analogous to Chromosomal conditions so as to hold more detailed information relevant to each Type of Mutational Analysis. (Next level of detail for these Mutational Analysis tests has not yet been shown)
Tests for multiple mutations ...
Test for selected mutations only ...
Assay for size of triplet repeat only ...
Chromosome microarray (CMA)
Microarray platform
Microarray platform version number
Base pair start coordinate
Base pair end coordinate
Flanking normal region before start
Analysis by Chromosome Banding
ISCN band level [#]
Chromosome band involved start
Chromosome band involved end
Chromosome banding method
Cells analysed [#]
Cells counted [#]
Cells karyotyped.total [#]
Colonies counted [#]
Mosaicism detected
Analysis by in situ hybridisation
Cell phase
Probe gene name
Probe locus
Probe vendor
Cells analysed
Functional analysis
DNA methylation
DNA transcription
Other
Linkage studies
Details of test method not already specified (e.g. MLPA or direct DNA sequencing or list mutations searched for)
Genetic Test Result
Cytogenetics
Karyotype analysis result in ISCN expression
47,XY,+2
46,XY,-18,+der(13q18q),t(13;18)(13q18q;13p18p)pat
Microarray analysis result in ISCN expression
arr 16p13.11(14,817,706-16,649,713)x3 dn
arr 1p36.22p31.3(9,240,258-67,037,578)x2 hmz
Molecular Genetics
DNA sequence variation type
"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.
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
HGVS DNA Sequence Variation
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.
e.g. c.3869A>C heterozygote
"Massively Parallel Sequencing [NGS/WGS] is anticipated to go here, but we have postponed specific inclusion pending the outcome of the MPS Working Party recommendations
DNA region name
"A human readable name for the region of interest. Typically Exon #, Intron # or other. NOTE: This is not standardized and is mainly for convenience and display purposes.
RNA
This section is anticipated to hold the results of RNA-specific changes, such as RNA Editing, Transcriptomic results, etc
Amino acid change
"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.
Genomic RefSeq String
"Definition: Carries the literal string that represents the DNA sequence that is the basis for deciding what is a different (a variation) in the study subject The same information can also be reported more compactly by reference (see LOINC code 48013-7) if the reference sequence has been catalogued by a registration authority such as RefSeq at NCBI a. Reference sequences can be quite long
Example answer: 241 GGAGTAATCA GCAACTCAGG GGGACCTGTA CGAGTCTATA GCCTACCTGG TCGAGAAAAC 301 TATTCCTCAG TAGATGCCAA TGGCATCCAG TCTCAAATGC TTTCTAGATG GTCTGCTTCT 361 TTCACAGTAA CTA".
Amino acid change type
Allelic State
"Includes Copy Number Variation [CNV], Loss of Heterozygosity [LOH], Quantitative gene dosage"
Result modifier
RNA/Protein consequences
Reports
Co-occurrence with deleterious mutations
Species conservation
Biochemical
This modifier can be used to correlate genetic findings with (e.g.) pharmacogenomic or biochemical results
Biophysical change
In-silico analyses
RNA Studies
Functional studies
Control data
Segregation data
Loss of normal allele in cancer
Genetic Test Interpretation
Limitations and Disclaimers (compulsory)
This section would describe the status of the result. Has it been validated? Is it research-use only? Are there known weaknesses? Is this study partial, in progress, complete, etc
Result Interpretation (compulsory)
This section summarises the laboratory findings. What were the results of testing? What are the biological and genetic implications of this result?
Genetic Screen findings (optional)
Carrier status summary (optional)
"Carrier identification 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. "
Sequence variation summary (optional)
Clinical Interpretation (Compulsory):
This section summarises the clinical significance (if any) of the laboratory findings. It can take into account Family History or Clinical History.
Genotype clinical interpretation (optional)
Phenotype clinical interpretation (optional)
Conclusion (optional)
This is the final "tie it all together" field, if not already done so above.
Interpretation modifier
As for Result modifier, see above