AUSTRALIAN PATHOLOGY UNITS AND TERMINOLOGY

(APUTS)

Reporting Terminology and Codes Genetic Pathology

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Terminology for Reporting Pathology: Generic Genetic Pathology Report

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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

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Example answers / response / Comments	LOINC	Component	Property	Timing	System	Scale	Method	Class	LongName
Specimen	31208-2	Specimen source	Prid	Pt	XXX	Nom		SPEC	Specimen source [Identifier] of Unspecified specimen
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
A coded disease (recommend SNOMED) which is known to be caused by or identified by genomic DNA Markers. ex: SCTID: 190905008 | Cystic fibrosis |									
alterntive 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
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
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
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
A DNA Marker identifier conveys a universal or standard repository identifier for definitive characteristics 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
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 yet 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
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
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
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
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
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
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
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
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
Cells analyzed [#]	62360-3	Cells analyzed	Num	Pt	Bld/Tiss	Qn	Molgen	HL7.CYTOGEN	Cells analyzed [#] in Blood or Tissue by Molecular genetics method
Cells counted [#]	62361-1	Cells counted	Num	Pt	Bld/Tiss	Qn	Molgen	HL7.CYTOGEN	Cells counted [#] in Blood or Tissue by Molecular genetics method
Cells karyotyped.total [#]	55199-4	Cells karyotyped.total	Num	Pt	Bld/Tiss	Qn		MOLPATH	Cells karyotyped.total [#] in Blood or Tissue
Colonies counted [#]	62362-9	Colonies counted	Num	Pt	Bld/Tiss	Qn	Molgen	HL7.CYTOGEN	Colonies counted [#] in Blood or Tissue by Molecular genetics method
Mosaicism detected	62363-7	Mosaicism detected	Find	Pt	Bld/Tiss	Nom	Molgen	HL7.CYTOGEN	Mosaicism detected in Blood or Tissue by Molecular genetics method
Analysis by in situ hybridisation									

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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
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
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
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
Cells analysed	62360-3	Cells analyzed	Num	Pt	Bld/Tiss	Qn	Molgen	HL7.CYTOGEN	Cells analyzed [#] in Blood or Tissue by Molecular genetics method
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 expressior	Find	Pt	Bld/Tiss	Nom	Molgen	HL7.CYTOGEN	Chromosome analysis result in ISCN expression in Blood or Tissue by Molecular genetics method
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
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
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
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
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
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	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
Allelic State	53034-5	Allelic state	Find	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	Allelic state in Blood or Tissue by Molecular genetics method
"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
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)	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?									

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Genetic Screen findings (optional)	19102-3	Genetic screen	Find	Pt	XXX	Nar	Molgen	MOLPATH	Genetic screen in Unspecified specimen by Molecular genetics method Narrative
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
Carrier Identification interpretation of all identified DNA Markers and/or Individual Alleles along with any known clinical information for the benefit of aidina clinicians in understandina the results overall.									
Sequence variation summary (optional)	53037-8	Genetic disease sequence variation interpretat	Imp	Pt	Bld/Tiss	Nom	Molgen	HL7.GENETICS	Genetic disease sequence variation interpretation [interpretation] in Blood or Tissue by Molecular genetics method
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
This section summarises the clinical significance (if any) of the laboratory findings. It can take into account Family Historv or Clinical Historv.									
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
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									


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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(9,240,258-67,037,576)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