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