

Preferred/example list in LOINC but required in IG:

Default proposal: change required lists to extensible (can send other codes if needed)

Patrick: for SO, we would “prefer” SO, etc. but for other cases extensible will be more suitable

AnswerList Link	List name	List contents	Notes	Final notes
https://r.detai ls.loinc.org/LOINC/69548-6.html?sections=Comprehensive	Variant Assess / Genetic variant assessment	Present Absent No call Indeterminate	Extensible, could still be required.	<i>Required Is ok</i>
https://r.detai ls.loinc.org/LOINC/48002-0.html	MG_2_Genetic variant source / Genetic variant source	Germline Somatic Fetal Likely germline Likely somatic Likely fetal Unknown genomic origin De novo	Alex had concerns with this list for tumor/vs/normal extensible	
https://r.detai ls.loinc.org/LOINC/48006-1.html	MG_4_Amino acid change type / Amino acid change type	Wild type Deletion Duplication Frameshift Initiating Methionine Insertion Insertion and Deletion Missense Silent Stop Codon Mutation	Required. AL shifted to a SO List: #22814	
https://r.detai ls.loinc.org/LOINC/53034-5.html	MG_5_Genetic variant allelic state / Genetic variant allelic state	Heteroplasmic Homoplasmic Homozygous Heterozygous Hemizygous	extensible	
https://r.detai ls.loinc.org/LOINC/81263-6.html	Complex variant types / Answers: ; Scale: Nom; Code: -; Score: -	Compound heterozygous Double heterozygous Haplotype Hemizygous	extensible	
https://r.detai ls.loinc.org/LOINC/53037-8.html	ACMG_Clinical significance of genetic variation	Pathogenic Likely pathogenic Uncertain significance Likely benign Benign	Mullai requested other items here Extensible Tracker:	Could extend the original list to clinvar, but may bleed into purpose of other

			Inherited Disease Pathogenicity - ValueCodeableConcept non LOINC value	
https://r detai ls.loinc.org/LOINC/53040-2.html	[CPIC metab act] Ultrarapid Rapid NI nterm Poor	Ultrarapid metabolizer Rapid metabolizer Normal metabolizer Intermediate metabolizer Poor metabolizer	preferred	
https://r detai ls.loinc.org/LOINC/51968-6.html	MG_8 / Genetic disease analysis overall predictive risk interpretation	Positive Negative Inconclusive Failure	preferred	
https://r detai ls.loinc.org/LOINC/83009-1.html	Genetic variation's effect on high-risk allele	Low risk High risk	Concern mentioned here at overall modelling approach extensible	
https://r detai ls.loinc.org/AnswerList/LL4049-4.html	Medication usage suggestion	Consider alternative medication Decrease dose Increase dose Use caution Normal response expected	extensible	
https://r detai ls.loinc.org/LOINC/62366-0.html	Genetic recommendation	Genetic counseling recommended Confirmatory testing recommended Additional testing recommended	extensible	

Unbound / TBD

Need to officially request codes to LOINC for all of these **(and make sure they can validate)**

Name / description / context / example answers

Can also request placeholders and use the LOINC template - but temporary codes may not validate on Grahame's server/IG publisher

Patrick can help prepare placeholders for URIs and can include temporary.

TBD / code	name	List contents (if provided)	Notes
TBD-mode-of-inheritance	MODE OF INHERITANCE (unbound, example)	Autosomal dominant Autosomal recessive X-linked dominant X-linked recessive Y-linked Codominant Mitochondrial	
TBD	Genetic variation's effect on transporter function (unbound, example)	Increased function Normal function Decreased function Poor function	53037-8??
TBD-grouper			
TBD-RegionCoverage		(quantity)	
TBD-somatic-diagnostic		E.g. Pathognomonic, Supportive, Argues Against, Rules Out	53037-8??
TBD-somatic-prognostic		E.g. Better outcome, poorer outcome	53037-8??
TBD-medication	(unbound, example)		
TBD-treatment	(unbound, example)		
TBD-somatic-predictive		E.g. Resistant, Responsive, Not-Responsive, Sensitive, Reduced-Sensitivity, Adverse Response	53037-8??
TBD-cancer	(unbound, example)		

No Binding listed (seems error)

https://s.details.loinc.org/LOINC/51961-1.html	(no binding mentioned) Preferred answer list : MG_6 / Drug efficacy	Resistant Responsive Presumed resistant Presumed responsive Unknown significance	
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	marker interpretation	Non-Responsive Presumed non-responsive	
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