

The following table contains a list of the coded concept fields within the eMERGE Report XML schema and their value sets. The Value Sets are recommendations not mandatory. If local, extensions or different coded concept sources are used it would require communication between the sender and receiver to agree upon those differences.

Coded Concept Fields	Coding System Constraints
Report.Genomic-Source	Codes from LOINC answer list LL378-1 Genetic Variation Source https://r.details.loinc.org/AnswerList/LL378-1.html
Report-Variant.Genomic-Source	
Report.Indication-Code	Local codes defined by eMERGE project.
Report.Overall-Interpretation	Local codes defined for eMERGE project, which conceptually map to the Codes from LOINC answer list LL541-1 Genetic disease analysis overall interpretation https://r.details.loinc.org/AnswerList/LL541-4.html
Report-Patient.Race-or-ethnicity	Codes from CDC-NESS coding system for HL7 Table 005 Race value set maintained by the CDC at https://phinvads.cdc.gov/vads/ViewValueSet.action?oid=2.16.840.1.114222.4.11.6065 Local codes added to extend base line set.
Report-Patient.Sex	Codes defined by the HL7 Table 001 for Administrative Sex. https://www.hl7.org/fhir/v2/0001/index.html
Report-Specimen.Type	Local codes defined by eMERGE project. SNOMED codes may be used if configured.
Report-Variant.Allele-State	Codes from LOINC answer list LL381-5 Genetic variant allelic state https://r.details.loinc.org/AnswerList/LL381-5.html
Report-Variant.Category	Codes from LOINC answer list LL4034-6 Clinical significance of genetic variation https://r.details.loinc.org/AnswerList/LL4034-6.html Local codes may supplement or overlap and map to LOINC codes.
Ref-Variant.Interpretation.Category	
Report.Patient-Disease.Code	Codes from OMIM, Disease Ontology, Orphanet, and other disease codes like those defined in the Monarch Initiative merged disease ontology. Supplemental local codes defined for diseases not available or mappable to available disease coding authorities.
Report.Interpreted-Disease.Code	
Reference-Variant.Interpretation-Disease.Code	
Reference-Variant.Gene-Identifier	HGNC, Entrez-Gene, OMIM, HPRD, Ensembl source identifiers can be used for genes.
Reference-Gene.Identifier	