

HL7 FHIR Proposal for Variant Inheritance Basis

Willie Chang

Version 1

In JIRA ticket [FHIR-32101](#), we propose to add a new component to the [R4 Variant](#) profile for expressing the method of determination for variant inheritance below.

- Component: variant-inheritance-basis
- Code: [LOINC 82309-6](#) (Basis for allelic phase)
- Value binding: [LOINC Answer List LL4050-2](#)
 - Directly measured
 - Family DNA
 - Family history
 - Inferred from population data

We are not able to think of other possible values, so we propose making the value set required rather than extensible, pending inputs from other Work Group members.

Epic currently uses an extension to send this information.

For reference, the variant-inheritance component has the following specification:

component:variant-inheritance	0..1	BackboneElement	Variant Inheritance
code	1..1	CodeableConcept	variant-inheritance Required Pattern: At least the following Code defined by a terminology system Fixed Value: (complex)
coding	1..*	Coding	
system	1..1	uri	Identity of the terminology system Fixed Value: http://hl7.org/fhir/uv/genomics-reporting/CodeSystem/tbd-codes-cs
code	1..1	code	Symbol in syntax defined by the system Fixed Value: variant-inheritance
value[x]	1..1	CodeableConcept	Maternal Paternal Unknown Binding: Variant Inheritances (extensible)

And for genomic-source-class:

component:genomic-source-class	0..1	BackboneElement	Genomic Source Class
code	1..1	CodeableConcept	48002-0 Required Pattern: At least the following Code defined by a terminology system Fixed Value: (complex)
coding	1..*	Coding	
system	1..1	uri	Identity of the terminology system Fixed Value: http://loinc.org
code	1..1	code	Symbol in syntax defined by the system Fixed Value: 48002-0
value[x]	1..1	CodeableConcept	Germline Somatic Fetal Likely germline Likely somatic Likely fetal Unknown genomic origin De novo Binding: LOINC Answer List LL378-1 (extensible)

The value sets for variant-inheritance and genomic-source-class do not overlap, which should address Robert Freimuth's concerns in the JIRA ticket.