HL7 FHIR Proposal for Variant Inheritance Basis

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Version 1

In JIRA ticket <u>FHIR-32101</u>, we propose to add a new component to the <u>R4 Variant</u> profile for expressing the method of determination for variant inheritance below.

- Component: variant-inheritance-basis
- Code: <u>LOINC 82309-6</u> (Basis for allelic phase)
- Value binding: LOINC Answer List LL4050-2
 - o Directly measured
 - o Family DNA
 - Family history
 - Inferred from population data

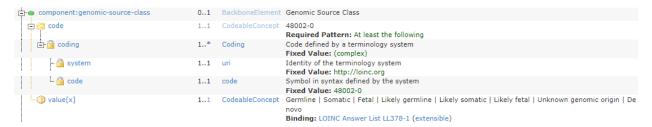
We are not able to think of other possible values, so we propose making the value set required rather than extensible, pending inputs from other Work Group members.

Epic currently uses an extension to send this information.

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



And for genomic-source-class:



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