The Standard Genetics Profile extends the DiagnosticReport resource to enable reporting of structured genetic test results. It denotes condition context for genetic testing, which may influence reported variants and interpretation for large genomic testing panels and provides references to the FamilyMemberHistory resource.

This profile supports reporting of variants at the genomic, cDNA, and protein change level. In addition, a condition context may be provided, as AssessedCondition. For large genomic tests, a condition may be used as an input into the analytic pipeline to aid in the identification of clinically relevant variants related to the test order.
Hi7 Clinical Genomics Work Group emphasizes the importance of transmitting structured genetic findings within the clinical, translational, and research environments fully integrated with other clinical data, in order to drive outcomes analysis, operational decision making, discovery research, and public health reporting.

Ownership[edit | edit source]

Owning committee name[edit | edit source]
CG

Contributing or Reviewing Work Groups[edit | edit source]

Expected implementations[edit | edit source]

gForge Users[edit | edit source]

FHIR Profile Development Project Insight ID[edit | edit source]

Plans[edit | edit source]

Timelines[edit | edit source]

• TargetDateForInternalReview

Balloting Requirements[edit | edit source]

Choose one:

• Ballot with next FHIR DSTU or Normative Edition
• or Ballot independently as DSTU
• or Realm specific ballot
• or No Ballot

Desired Ballot Date

• PutDesiredBallotDateHere