

<https://bioethicsarchive.georgetown.edu/pcsbi/sites/default/files/Clinician%20Primer%20Incidental%20Findings%2010.30.16.pdf>

drExtension:

secondary-finding

Profile (propose only adding it to one of our profiles if we make it more specific to CG?):

Genetic Observation Common Properties

(<http://build.fhir.org/ig/HL7/genomics-reporting/obs-base.html>)

Type:

CodeableConcept

Control:

0..1

Terminology Binding (Preferred):

Code	Display	Definition
ACMG-SF-V1	ACMG Version 1	First release (2013): ACMG Recommendations for Reporting of Incidental Findings in Clinical Exome and Genome Sequencing https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3727274/
ACMG-SF-V2	ACMG Version 2	Second release (2016): Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update (ACMG SF v2.0): a policy statement of the American College of Medical Genetics and Genomics. https://www.ncbi.nlm.nih.gov/pubmed/27854360

Definition:

Secondary findings are genetic test results that provide information about variants in a gene unrelated to the primary purpose for the testing, most often discovered when Whole Exome Sequencing (WES) or Whole Genome Sequencing (WGS) is performed. This extension should be used to denote when a genetic finding is being shared as a secondary finding, and ideally refer to a corresponding guideline or policy statement.

For more detail, please see:

<https://ghr.nlm.nih.gov/primer/testing/secondaryfindings>