

Connectathon 27 - FHIR Genomics Operations

Resources

- [Clinical Genomics Connectathon 27 Confluence page](#)
- [FHIR Genomics Operations undergoing ballot](#)
- [GACS Postman collection](#). Examples are included for
 - LiftOver
 - UncallableRegions
 - Targeted NGS, with regions not studied
- [Patients stored in the GACS server](#)
 - All data is from [1000 Genomes Project](#), publicly available
 - Most patient data is derived from WGS Build 37 VCFs
 - Patient NB6TK328 has data derived from WGS Build 38 VCF
 - Patient HG00403A has data derived from a targeted gene panel (list of studied regions is [here](#))
- Possible scenarios
 - Query for variants in a region, using the find-subject-variants operation.
 - Include a parameter to also get back information on uncallable regions.
 - For various SNPs of interest, where no variants are identified, determine if the region was studied and not deemed uncallable.
 - Explore liftOver capabilities (e.g. query a patient that has build37 data using a query using build38 parameters).
 - Compare Bundle vs. DiagnosticReport as operation response
- [\(DRAFT\) Potential new FHIR Genomics operations](#)
 - [Example of revised \\$find-subject-variants that supports multiple regions](#)
(\$find-subject-variants/?patientId=HG00404®ion=NC_000005.9:112043201-12181936,NC_000017.10:41196311-41277500,NC_000019.9:38924339-39078204)

Issues / proposals

- [\[Jira 31565\]](#) Add 'uncallableRegions' parameter to find-subject-variants operation
 - CHANGE: OUT:regionStudied cardinality from [0..*] to [0..1].
 - Where a server has information about the studied region, a regionStudied observation should be returned. If uncallableRegions parameter is set, but server doesn't have information on uncallable regions, the server shall return a single component:uncallable-regions with dataAbsentReason='unknown'
 - Long answer:
 - change OUT:regionStudied cardinality from [0..*] to [0..1].

- without the uncallableRegions flag
 - if studied region is unknown, do not include a regionStudied observation
 - if studied region is known, include a single regionStudied observation with
 - [1..1] component:genomic-ref-seq indicates the chromosome studied
 - [1..*] component:ranges-examined indicate the studied ranges that overlap with the queried region.
 - [1..1] component:coordinate-system, valued with '0-based interval counting'
 - [0..0] component:uncallable-regions is not included
- with the uncallableRegions flag
 - if studied region is unknown, do not include a regionStudied observation
 - if studied region is known, include a single regionStudied observation with
 - [1..1] component:genomic-ref-seq indicates the chromosome studied
 - [1..*] component:ranges-examined indicate the studied ranges that overlap with queried region.
 - [1..1] component:coordinate-system, valued with '0-based interval counting'
 - uncallable regions
 - if uncallable regions are not known
 - [1..1] component:uncallable-regions with dataAbsentRegion='unknown'
 - if uncallable regions are known
 - [1..*] component:uncallable-regions each indicating a contiguous sub-region within a reported ranges-examined that was deemed uncallable.
- allelic-state - should only be required if genomic-source-class = 'germline'
- Error codes:
 - Background: to move from single-region to multi-region queries, we need to reexamine the use of ERROR code 404 where the region hasn't been studied. With multi-region queries, some regions will and others may not have been studied. Also, we don't have a specific way to indicate a failed liftOver.
 - 200: Successfully executed request
 - if query range not studied, include regionStudied observation with a single component:ranges-examined with dataAbsentReason='not-performed'
 - 400 ERROR: Invalid query parameters
 - 404 ERROR: Patient not found

- 422 ERROR: Failed LiftOver
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- [] DiagnosticReport vs. Bundle for returning operation results
 - Options
 - Option 1: Bundle with full observations
 - Option 2: DiagnosticReport with referenced observations
 - Option 3: DiagnosticReport with contained observations
 - Considerations
 - Ability to query individual observations
 - Ability to retain bundle or report (to have a retained timestamp for when report was assembled)
 - Dynamic generation of FHIR instances and ability of server to keep track of assigned observation identifiers.
 - CDS pipeline vs. Lab-to-EHR pipeline, and the need for persistent identifiers in these scenarios. (Is the intent of the communication to have the contained observations stored discreetly in the EHR?)
- ... (any ballot comments)...