

ACGS-GeL CIP-API Workshop

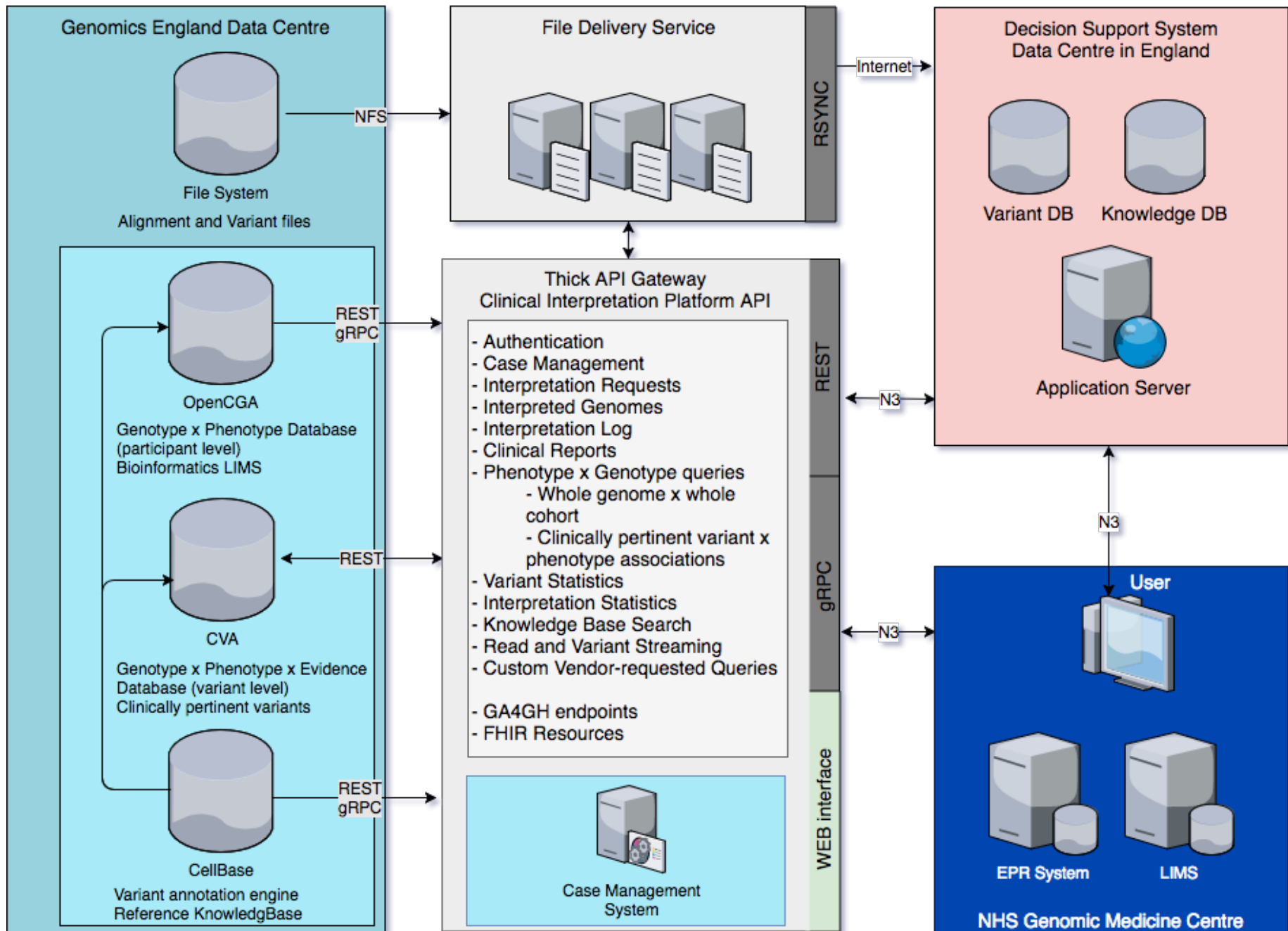
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- *Clearly defined methods of communication between software components.*
- *exposing objects or actions a developer needs, an API simplifies programming*
- Web APIs
 - HTTP request messages
 - GET, PUT, POST
 - JSON

Background

- CIP-API
 - Created by Genomics England to distribute results to Clinical Interpretation Providers
 - Congenica, Fabric (Omicia), Nextcode and Illumina
 - Also available over N3 to NHS-GMCs (subject to DSA)
- Interpretation Portal
 - GUI for visualizing cases in the CIP-API
 - Links to CIP systems and Clinical Data
 - Completion of Outcomes Questionnaires (closing cases)

Clinical Interpretation Platform Architecture



GeL Bioinformatics data models

- <http://gelreportmodels.genomicsengland.co.uk>

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Protocols

ClinicalReportsCancer org.gel...

Types

org.gel.models.report.avro

ActionType Enum

Actions

Ancestries

CancerDemographics

CancerParticipant

CancerSample

ChSquare1KGenomesPha...

ClinicalReportCancer

ConsentStatus

EthnicCategory Enum

FeatureTypes Enum

GelPhase Enum

GenomicFeatureCancer

InbreedingCoefficient

KGPopCategory Enum

KGSuperPopCategory En...

MatchedSamples

Method Enum

Penetrance Enum

PersonKaryotypicSex Enum

org.gel.models.report.avro

ClinicalReportCancer

Type	Field	Default Value	Description
string	interpretationRequestID		This is the interpretation Request Id, first number in XXX-123-1
string	interpretationRequestVersion		This is the version of the interpretation Request Id, second number in XXX-123-2
string	reportingDate		Date of this report
string	user		Author of this report
array<ReportedSomaticVariants>	candidateVariants		Candidate Variants - as defined in CommonInterpreted
array<ReportedSomaticStructuralVariants>	candidateStructuralVariants		Candidate Structural Variants - as defined in CommonInterpreted
string	genomicInterpretation		Summary of the interpretation, this should reflect the positive conclusions of this interpretation
null array<string>	references		References (pubmed Ids)
map<string>	referenceDatabasesVersions		This map should contain the version of the different DBs used in the process
map<string>	softwareVersions		This map should contain the version of the different DBs software in the process
map<array<map<string>>>	genePanelsCoverage		This map of key: panel_name, value: (arrays of (map of key: gene, value: gene coverage))
CancerParticipant	cancerParticipant		

CIP-API: Interpretation Requests

- *JSON data - results of GeL's Interpretation pipeline*
- A case can have multiple interpretation requests (versions)
- http://gelreportmodels.genomicsengland.co.uk/html_schemas/org.gel.models.report.avro/3.0.0/InterpretationRequestCancer.html#/
- Contains:
 - Case status (see CIP-API statuses slide later)
 - Same type (cancer or rare disease)
 - Participant information (identifiers, type, origin, file paths, priority)
 - Details of Tiered Variants

CIP-API: Interpreted Genome

- Results of third-party system Interpretation of GeL's results
 - OMICIA's interpretation (Phevor and VAAST scores)
 - Exomiser scores
 - Any other service that supports the model

CIP-API: Clinical Reports

- *JSON data in Clinical Report*
- An interpretation request can have multiple clinical reports
- http://gelreportmodels.genomicsengland.co.uk/html_schemas/org.gel.models.report.avro/3.0.0/ClinicalReportCancer.html#/schema/org.gel.models.report.avro.ClinicalReportCancer
- Contains:
 - Reporter, Date
 - Participant information
 - Details of primary findings
- Everything you see in the HTML report

CIP-API Statuses

API status	Interpretation Portal	Description
waiting_payload	To Be Dispatched	GeL interpretation Pipeline has completed
interpretation_generated	To Be Dispatched	Interpretation Request has been created, ready to send to CIP
files_copied	To Be Dispatched	BAM, VCF and tiering files have been sent to the CIP
dispatched	To Be Dispatched	Interpretation Request has been sent to CIP
transfer_ready	To Be Dispatched	CIP has completed their analysis of the case
transfer_complete	To Be Dispatched	CIP has transferred their interpreted genome to the API
gel_qc_failed	To Be Dispatched	The case has failed QC and will be sent back to the CIP
rejected_wrong_format	To Be Dispatched	Interpreted Genome has failed API validation
gel_qc_passed	To Be Dispatched	The case has passed GeL QC checks
sent_to_gmcs	To Be Reviewed	The case has been sent to the GMCs for them to generate the report in the CIP portal in Inuvika/web interface
report_generated	To Be Closed	A report has been generated for this case by the GMC. More than one report can be generated
report_sent	Archived Cases	The reporting outcomes questionnaire has been completed
blocked	NA	Further interpretation of the case has been blocked

CIP-API end-points

swagger

Hello, cboustred

Django Logout

Authorize

Genomics England Interpretation Platform API

ClinicalReport	Show/Hide	List Operations	Expand Operations
ExitQuestionnaire	Show/Hide	List Operations	Expand Operations
GelQCOutcome	Show/Hide	List Operations	Expand Operations
GelStatus	Show/Hide	List Operations	Expand Operations
GelTieringQCOutcome	Show/Hide	List Operations	Expand Operations
Download_file	Show/Hide	List Operations	Expand Operations
docs	Show/Hide	List Operations	Expand Operations
get-token	Show/Hide	List Operations	Expand Operations
interpretationRequests	Show/Hide	List Operations	Expand Operations
dateSummary	Show/Hide	List Operations	Expand Operations
stats	Show/Hide	List Operations	Expand Operations
case-priority	Show/Hide	List Operations	Expand Operations
dispatch	Show/Hide	List Operations	Expand Operations
request_status	Show/Hide	List Operations	Expand Operations
interpretationRequestsList	Show/Hide	List Operations	Expand Operations
test_cip	Show/Hide	List Operations	Expand Operations
release	Show/Hide	List Operations	Expand Operations
download_token	Show/Hide	List Operations	Expand Operations

Authentication

- Username and Password
 - Controlled by GeL service desk (LDAP)
- Users associated with a GMC and multiple Local Delivery Partners (LDPs)
- Interpretation Requests linked by LDP
 - Access to cases therefore controlled at LDP level
- Successful authentication
 - POST username and password to “get-token” endpoint
 - Returns a “token” which has to be used in subsequent API interactions

API Client Libraries

- Code Library that helps the user interact with an API
- E.g.
 - A Python library that helps a user with basic tasks such as authentication
- Currently no Client Library for CIP-API
 - Hope can use these workshops to start building one!

Interpretation Portal

- <https://cipapi.genomicsengland.nhs.uk/interpretationportal>
- GUI for visualizing cases in the CIP-API
- Following functionality:
 - Overview of Rare Disease and Cancer cases ready for NHS GMC review and track overall case status.
 - Hyperlink to submitted clinical data in Lab Key.
 - Download available files including VCFs & reports.
 - Review GeL generated cancer preliminary and supplementary analysis reports.
 - Complete a reporting outcomes questionnaire to :close” a case
- Lacks integration with your local systems / data

Documentation

<https://cipapi.genomicsengland.nhs.uk/api/documentation/index.html>

The screenshot shows the 'Usage' section of the Genomics England Interpretation API documentation. On the left is a sidebar with a search bar and a list of navigation links: 'Usage', 'Basic Usage', 'Authentication Endpoint', 'RequestList Endpoint', and 'Interpretation Request Endpoint'. The main content area is titled 'Usage' and includes a 'View page source' link. It contains a bulleted list of sub-topics: 'Basic Usage', 'Authentication Endpoint', 'RequestList Endpoint', and 'Interpretation Request Endpoint'. The 'Basic Usage' section explains the API's purpose and provides a link to a full list of endpoints. The 'Authentication Endpoint' section lists details for a POST request, including URL, method, content type, and content. A 'Usage Example' section shows a Python code snippet using the 'requests' library to obtain a token and make a protected API call.

Genomics England Interpretation API

Search docs

Usage

- Basic Usage
- Authentication Endpoint
- RequestList Endpoint
- Interpretation Request Endpoint

Docs » Usage [View page source](#)

- Usage
 - Basic Usage
 - Authentication Endpoint
 - RequestList Endpoint
 - Interpretation Request Endpoint

Usage

Basic Usage

Genomics England Interpretation API, the aim of this documentation is guide the user through the basic uses cases. Python and bash code will be used to illustrate call to the webservices, but this can be extrapolated to any other language. This application also provide a friendly web interface to use the webservices, that can be used to get familiar with its usage. Not all the endpoints will cover in this documentation but a full list can be found in: </api/docs/>

Authentication Endpoint

- URL: `/api/get-token/`
- Method: POST
- Content Type: `'application/json'`
- Content: `{ "username": "user", "password": "password" }`

Usage Example:

```
import requests

token = ''
response = requests.post('{host}/api/get-token'.format(host='https://.....'), {'username': 'user', 'password': 'password'})
if response.status_code == 200:
    token = response.json()['token']

response2 = requests.get('protected-url', headers={"Authorization": "JWT " + token})
```