

## **GENEYX ANALYSIS**

## **Application Programming Interface**

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## **Overview**

Geneyx Analysis is an NGS Analysis, Interpretation and Reporting platform that offers a complete VCF to Reporting solution for clinics, hospitals, diagnostic companies, and healthcare providers. Geneyx Analysis offers integrated variant prioritization, enhanced and rapid variant review and analysis, and automated reporting capabilities that allow for easy scaling of NGS operations while providing complete flexibility in customization and implementation. This document provides information about how to upload VCF samples into Geneyx Analysis using web API.

# **Create Sample**

This API call allows a user/integrator to upload a VCF file and create a corresponding Sample entry in Geneyx Analysis. Other than the VCF file, additional metadata may be included in the Sample. Optionally, the Sample entry may be associated with an existing Subject entry (by ID).

Request URL: <a href="mailto:kmailto:domain">domain</a> /api/createSample

Europe domain: <a href="https://analysis.geneyx.com">https://analysis.geneyx.com</a></a> China domain: <a href="https://fa.shanyint.com">https://fa.shanyint.com</a>

**Post Parameters** 

Category	Parameter	Description	Required
Auth	ApiUserId	The API user Id	Yes
	ApiUserKey	The API user key	Yes
	CustomerAccountKey	Geneyx Analysis Customer account key (for integrator this should be a parameter requested from the user). If the customer account key is missing the sample will be created at the current user's account.	No
Sample	SampleSerialNumber	Sample ID, may be the same as the ID of the sample in the LIMS	Yes
	SampleSequenceDate	Date when sample was sequenced	No
	SampleTakenDate	Date when sample was taken	No
	SampleReceivedDate	Date when sample was received	No
	SampleSequenceMachineId	ID of Sequencing Machine entry as defined in customer account	No
	SampleEnrichmentKitld	ID of Enrichment Kit entry as defined in customer account. If empty the default account enrichment kit will be used.	No
	SampleType	May be one of the following:  • RnaSeq • DnaSeq	No
	SampleTarget	May be one of the following:  WholeGenome  Exome  GenePanel  TargetRegion	No
	SampleSource	May be one of the following:      GermLine     Tumor     Blood     Buccal     Fetal	No

		Saliva	
		Other	
	SampleGenomeBuild	HG19_GCRH37 - default	No
		• GCRH38	
	SampleRelation	The sample relation, can be	No
		• Self	
		<ul> <li>Mother</li> </ul>	
		• Father	
		Sibling	
		• Twin	
		<ul> <li>MotherRelative</li> </ul>	
		<ul> <li>FatherRelative</li> </ul>	
		Other	
	SampleNotes	Free text up to 1024 characters	No
	SampleQcData	JSON structure for QC data	NO
	SnvFile	The name of the SNV file (that is attached	No
		below)	
	StructFile	The name of the structural variation file (that	No
		is attached below)	
	FastQUrl	The URL to the fastQ file	No
	BamUrl	The URL to the BAM file	No
	SnvFile	The name of the uploaded SNV file	No
	StructFile	The name of the structural variation file	No
Subject	SubjectId	Unique identified for Subject. May be any	Yes
		string. If an existing subject exists, the sample	
		will be associated with it.	
	SubjectName	Name of subject	No
	SubjectGender	May be "M" or "F"	No
	SubjectDateOfBirth	Date of birth of subject	No
	SubjectConsanguinity	The subject consanguinity	No
	SubjectPopulationType	Population type	No
	SubjectPaternalAncestry	Paterntal ancestry	No
	SubjectMaternalAncestry	Maternal ancestry	No
	SubjectFamilyHistory	The family history	No
Assignment	AssignedToUserId	The user id the sample is assigned to	No
	AssignedToFullName	The full name of the user the sample is assigned to	No
	Group Assignment	List of groups the sample is assigned to, each list item has:  • Code • Name	No
File	Files	Parameter of type File, must be .vcf or .vcf.gz	No

#### Notes

- It is possible to upload more than one file as part of one API call. The example for that would be to when uploading SNV file and a Structural variation file for the same sample.
- When uploading a Structural variation file or both SNV / Structural it is required to specify which file is which at the SnvFile/StructFile fields. The value should be the file name only without folder information.

## Response

A JSON object with the following fields

Code	success or error
Data	An optional returned data
Info	Textual message (or error message) related to the returned value

## Request Example:

```
Post Parameters:
"ApiUserId "
                    : "<Your API User Id>",
"ApiUserKey" : "<Your API User Key >",
"CustomerAccountKey": "<The customer account key>"
"SampleSerialNumber": "SAM-00123",
"SampleSequenceDate": '2016-05-01 08:00',
"SampleTakenDate" :'2016-01-01 08:00',
                :"RndSeq",
: "Exome",
"SampleType"
"SampleTarget"
"SampleSource"
                    : "Germline",
"SubjectId"
                     : "Subject1",
"SnvFile"
                     : "test.vcf"
File Parameter: c:\vcf\test.vcf
```

### Successful Response Example:

```
{ "Code": "success"}
```

```
{ "Code": "error", "Info": "SampleID SAM-00123 already exists" }
```

## **Create Case**

The create case API is used for creating a new case in Geneyx Analysis.

**URL**: <domain>/api/CreateCase

**Action**: POST

Category	Parameter	Description	Required
Auth	APIUserId	The API user Id	Yes
	APIUserKey	The API user key	Yes
	CustomerAccountKey	Geneyx Analysis Customer account key (for integrator this should be a parameter requested from the user). If the customer account key is missing the sample will be created at the current user's account.	No
Case	SerialNumber	The case serial number. In case the SN is not provided it shall be automatically generated by the server	No
	Name	The case Name. In case empty it shall be assigned as the protocol name	No
	Description	The clinical description	Yes
	PhenAlleleFreq	Phenotype allele frequency. In case empty the value is taken from the protocol	No
	Phenotypes	The phenotypes to use	No
	Owner	The case doctor owner	No
	OwnerDepartment	The case doctor department	No
	GeneList	A gene list to associate with the case	No
	Protocolld	The protocol ID to create the case from	Yes
Subject	SubjectId	An <u>existing</u> subject ID	Yes
Sample	ProbandSampleId	An <b>existing</b> main sample ID for the case	Yes
	AssociatedSamples	Array of the associated samples for the case. An example is the mother and father samples in a trio analysis. Each element in the array shall have the following fields	No
		Relation – the sample relation	Yes
		Sampleld - the sample id	Yes
		Affected – affect indication	No
Assignment	AssignedToUserId	The user id that case is assigned to	No
Assignment	Assigned To Full Name	The full name of the user the case is assigned to	No

### Sample Relation valid values

- Self
- Mother
- Father
- Sibling
- Twin
- MotherRelative
- FatherRelative
- Other

## Sample Affect valid values

- Unknown
- Affected
- Unaffected

## Request Example:

```
JSON Parameters:
{
        "ApiUserId"
                                : "<Your API User Id>",
        "ApiUserKey"
                                 : "<Your API User Key >",
        "CustomerAccountKey" : "<The customer account key>"
        "Protocolld"
                                : "RG_TRIO",
        "Description"
                                 : "Clinical Description 1",
        "PatientId"
                                 : 'Patient1',
        "MainSampleId"
                                 : 'Sample1',
        "AssociatedSamples"
                                 :[
                         {"SampleId": "MotherSample1", "Relation": "Mother", "Affected": "Unaffected"},
                        {"SampleId": "FatherSample1", "Relation": "Father", "Affected": "Unaffected"}
                                   ]
```

### Successful Response Example:

```
{ "Code": "success", Data: { the case object} }
```

```
{ "Code": "error", "Info": "MainSampleID SAM-00123 was not found" }
```

# **Case Assignment**

The case assignment API is used for updating the user assignment for a case

**URL**: <domain>/api/CaseAssignment

**Action**: POST

Payload: JSON structure

Category	Parameter	Description	Required
Auth	APIUserId	The API user Id	Yes
	APIUserKey	The API user key	Yes
Case	SerialNumber	The case Serial number	Yes
Assignment	AssignedToUserId	The user id that case is assigned to	No
	Assigned To Full Name	The full name of the user the case is assigned	No
		to	

#### Notes:

In case the user assignment is null, the user assignment shall be removed.

### Request Example:

```
JSON Parameters:

{

"ApiUserId" : "<Your API User Id>",

"ApiUserKey" : "<Your API User Key >",

"SerialNumber" : "CaseID",

"AssignedToUserId" : "<The user id GUID>",

"AssignedToFullName" : "John Dow",

]

}
```

## Successful Response Example:

```
{ "Code": "success", Data: { the case assignment object} }
```

```
{ "Code": "error", "Info": "Case not found" }
```

# **Sample Permission**

The sample assignment API is used for updating the user/group assignment for a sample

**URL**: <domain>/api/SampleAssignment

**Action**: POST

Payload: JSON structure

Category	Parameter	Description	Required
Auth	APIUserId	The API user Id	Yes
	APIUserKey	The API user key	Yes
Sample	SerialNumber	The sample Serial number	Yes
Assignment	AssignedToUserId	The user id that case is assigned to	No
	AssignedToFullName	The full name of the user the case is assigned	No
		to	
	Group Assignment	List of groups the sample is assigned to, each	No
		list item has:	
		• Code	
		Name	

#### Notes:

In case the user assignment is null, the user assignment shall be removed.

In case the group assignment is null or empty, the group assignment shall be removed

## Request Example:

```
JSON Parameters:

{

"ApiUserId" : "<Your API User Id>",

"ApiUserKey" : "<Your API User Key >",

"SerialNumber" : "SampleID",

"AssignedToUserId" : "<The user id GUID>",

"AssignedToFullName" : "John Dow",

]

}
```

## Successful Response Example:

```
{ "Code": "success", Data: { the sample assignment object} }
```

```
{ "Code": "error", "Info": "Sample not found" }
```

# **Report List**

The report list api is used to get new reports created on the account from a given date

URL: <domain>/api/ReportList?startTime=xxx

Action: POST

Payload: JSON structure

Category	Parameter	Description	Required
Auth	APIUserId	The API user Id	Yes
	APIUserKey	The API user key	Yes
URL	StartTime	The start time of reports retrieval	Yes

# **Report Url**

The report URL api is used to download a report by the report filename returned by the report list API

URL: <domain>/api/ReportUrl?fileName=xxx

**Action**: POST

Payload: JSON structure

Category	Parameter	Description	Required
Auth	APIUserId	The API user Id	Yes
	APIUserKey	The API user key	Yes
URL	FileName	The report file name to download	Yes

# **Sequence machines**

The sequence machine API is used to retrieve the list of sequence machines in the account

URL: <domain>/api/SequenceMachines

**Action**: POST

Category	Parameter	Description	Required
Auth	APIUserId	The API user Id	Yes
	APIUserKey	The API user key	Yes

## **Enrichment kits**

The sequence machine API is used to retrieve the list of enrichment kits in the account

URL: <domain>/api/enrichmentKits

Action: POST

Payload: JSON structure

Category	Parameter	Description	Required
Auth	APIUserId	The API user Id	Yes
	APIUserKey	The API user key	Yes

## **Protocols**

The sequence machine API is used to retrieve the list of protocols in the account

URL: <domain>/api/protocols

**Action**: POST

Payload: JSON structure

Category	Parameter	Description	Required
Auth	APIUserId	The API user Id	Yes
	APIUserKey	The API user key	Yes

# **Samples**

The sequence machine API is used to retrieve the list of samples in the account

**URL**: <domain>/api/samples

Action: POST

Category	Parameter	Description	Required
Auth	APIUserId	The API user Id	Yes
	APIUserKey	The API user key	Yes

## **Patients**

The sequence machine API is used to retrieve the list of patients in the account

**URL**: <domain>/api/patients

Action: POST

Payload: JSON structure

Category	Parameter	Description	Required
Auth	APIUserId	The API user Id	Yes
	APIUserKey	The API user key	Yes

## Cases

The sequence machine API is used to retrieve the list of cases in the account

URL: <domain>/api/cases

**Action**: POST

Category	Parameter	Description	Required
Auth	APIUserId	The API user Id	Yes
	APIUserKey	The API user key	Yes