SMART Genomics API

-- Standardizing genomics API to facilitate utilization of genetic data in clinics and laboratories

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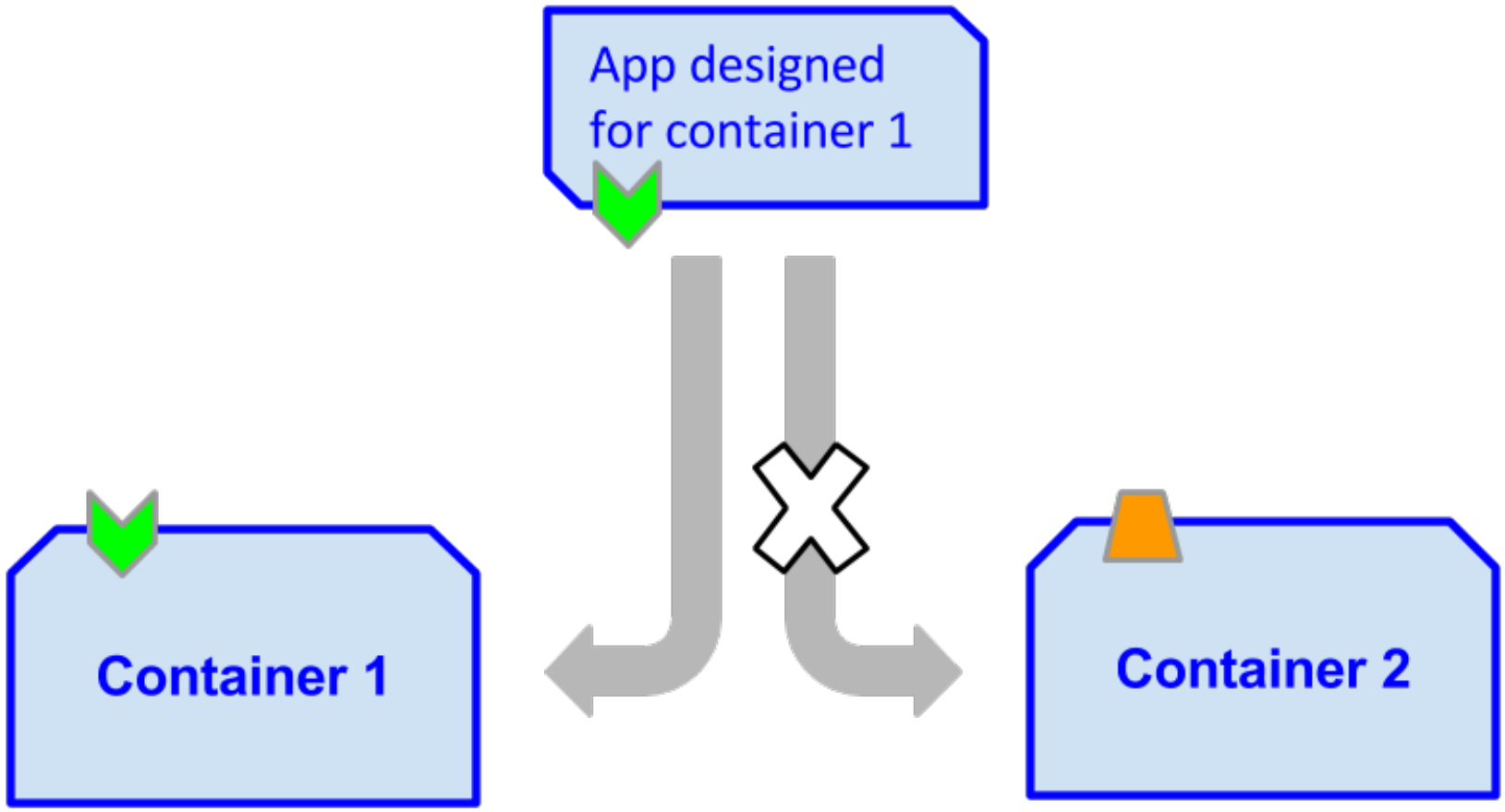
Problems of Current Genomics Communication

- No standard **API** (**A**pplication **P**rogramming **I**nterface)
- Lack means of communication
- Not integrated with clinical network


App designed
for container 1
Container 1
Container 2

"Plug" doesn't work

Representation of Genetic Data

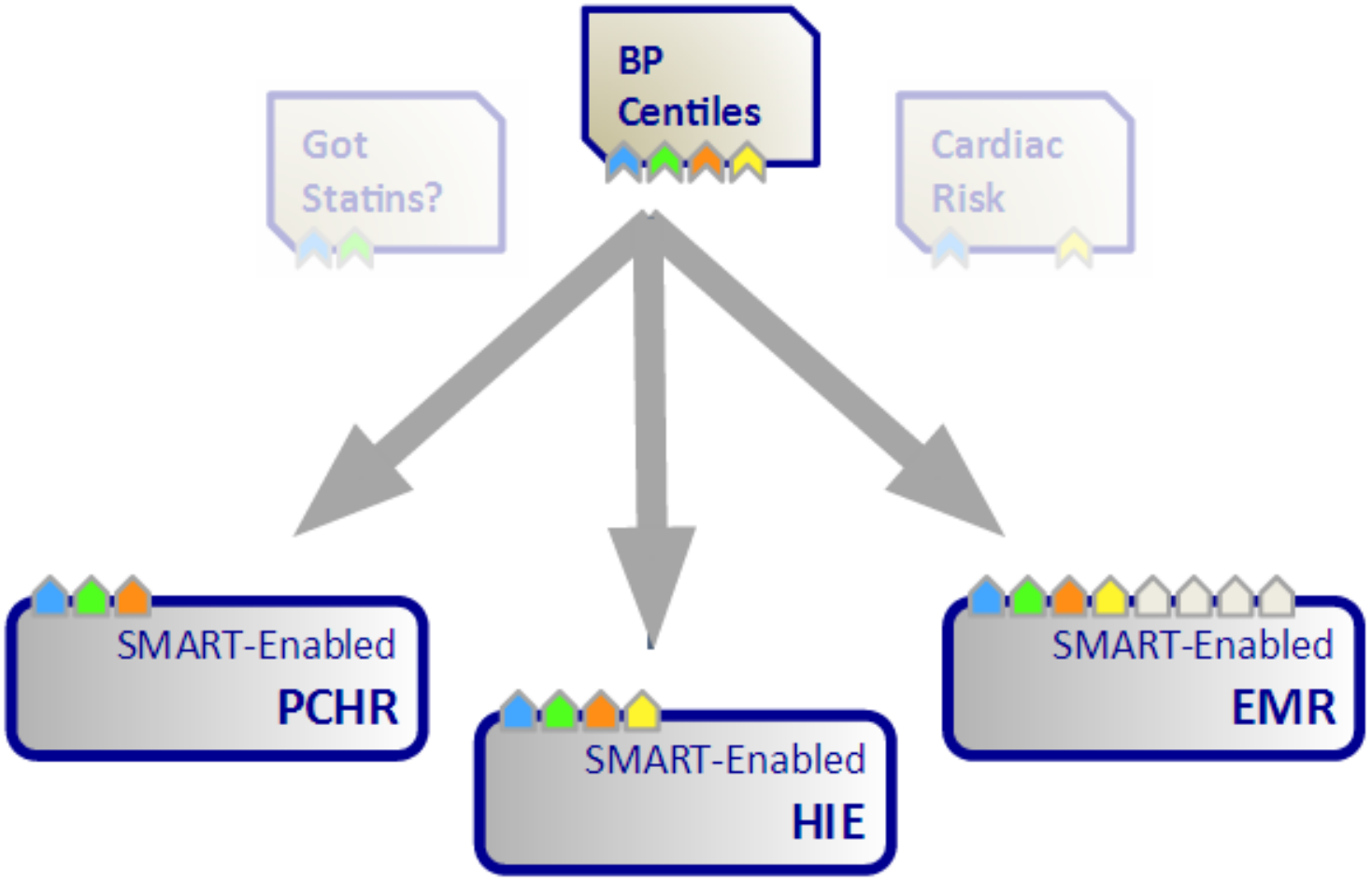
- Contents are essentially the same
  - Sequences (ATGC..) and genetic files
- Data are structured differently
- Different approaches of authentication
  - Most follow OAuth but implement differently

SMART Provides a Solution

- **S**ubstitutability **M**edical **A**pplications **R**euseable **T**echnology

- Generic design of API
  - Developers focus on the app

- Integration with clinical data

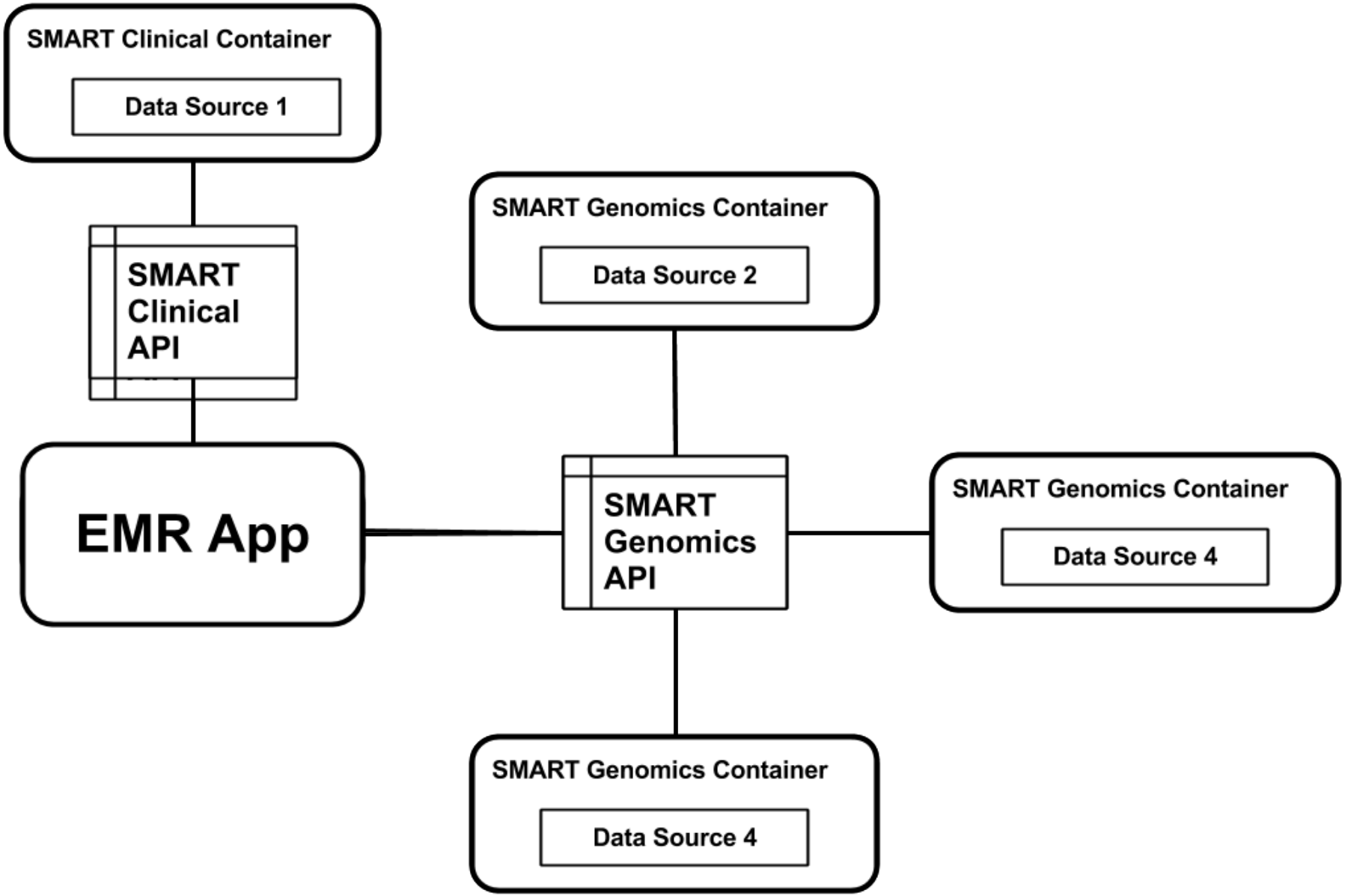

BP
Centiles
Got
Statins?
Cardiac
Risk
SMART-Enabled
PCHR
SMART-Enabled
HIE
SMART-Enabled
EMR

SMART Genomics API

- **Approaches substitutability differently because of differences between clinical and genomic data**

- **Integration of genetic data among heterogeneous sources**

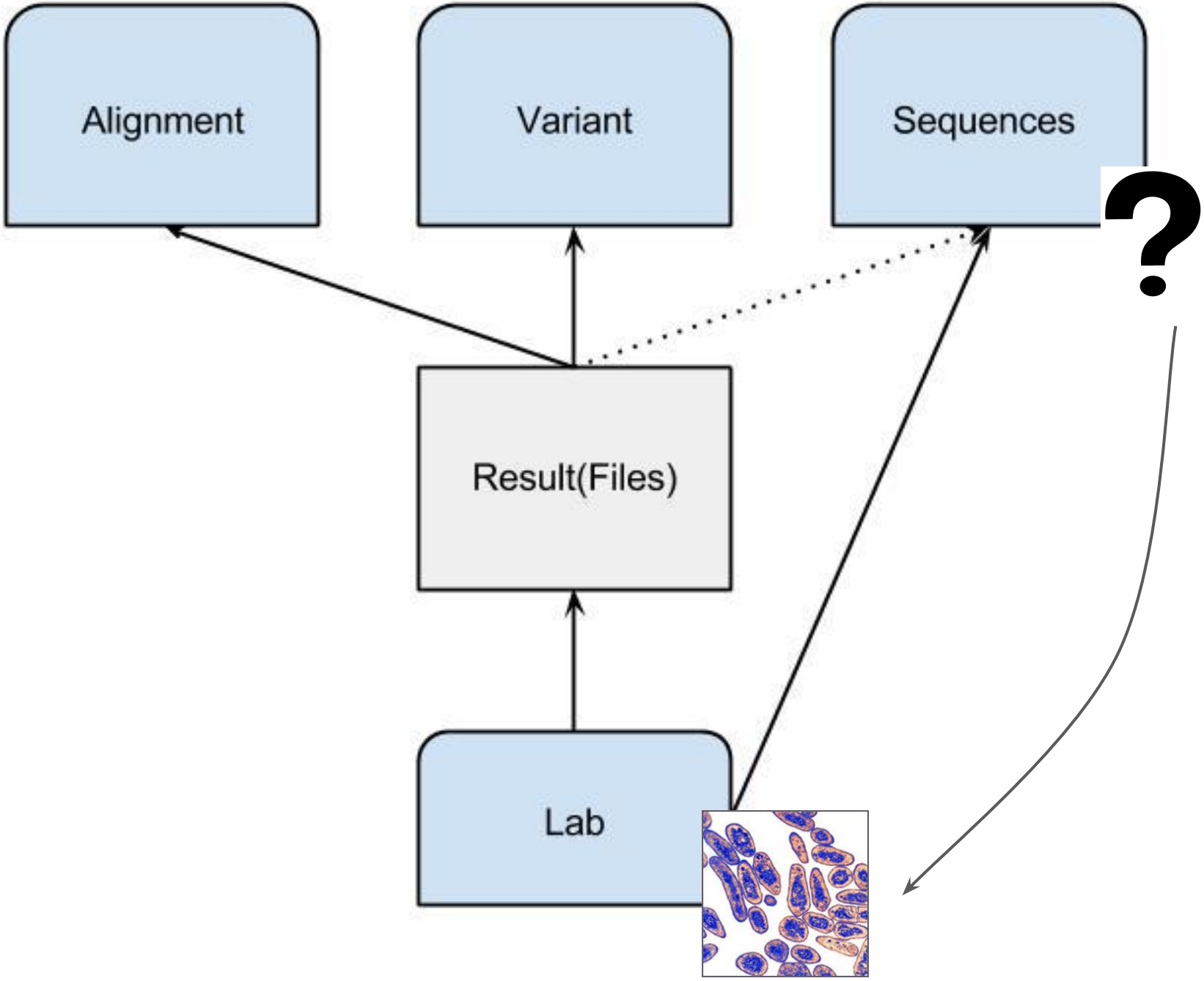
- **Bridging clinics by providing CCDA Genomics**
  - **CCDA**(**C**onsolidated **C**linical **D**ocument **A**rchitecture) -- an adopted format for transferring medical documents
  - Summary of a patient's genetic report

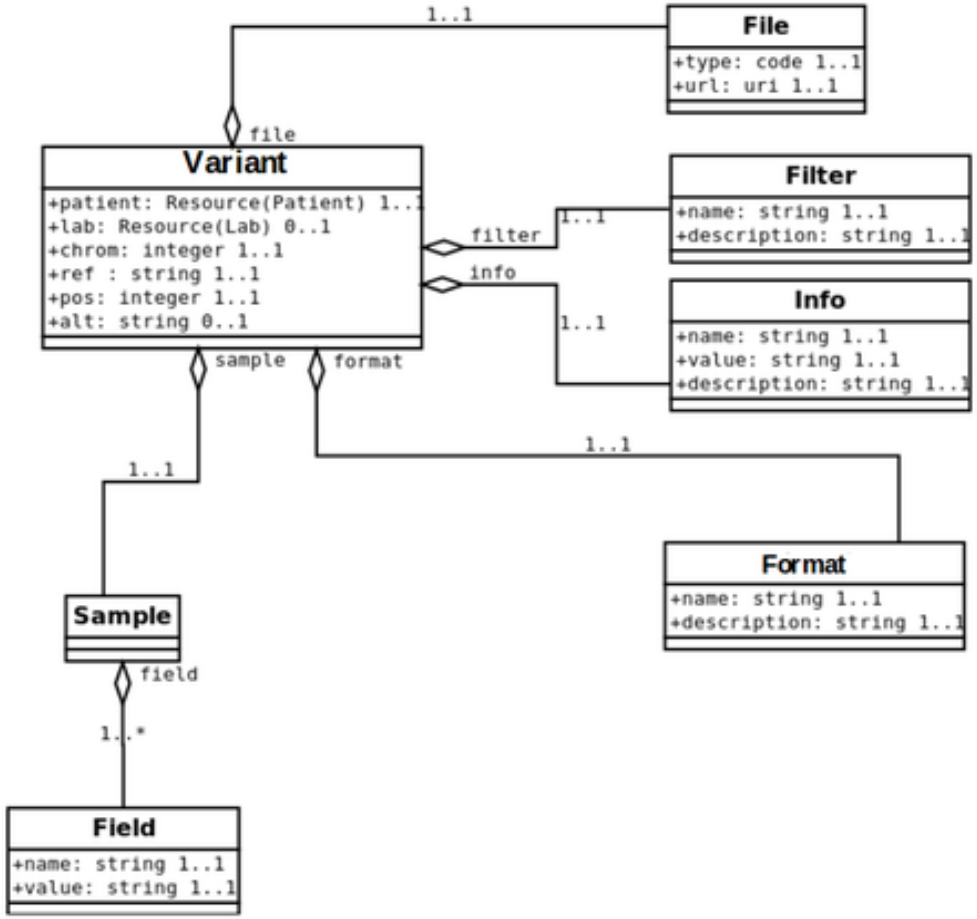

SMART Clinical Container
Data Source 1
SMART Clinical API
SMART Genomics Container
Data Source 2
EMR App
SMART Genomics API
SMART Genomics Container
Data Source 4
SMART Genomics Container
Data Source 4

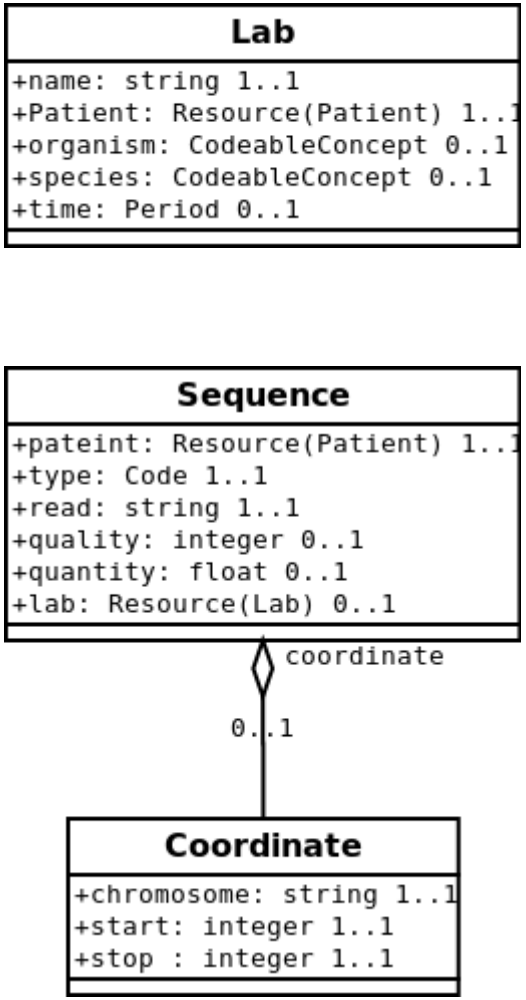
SMART Genomics API

- **Authentication**
  - OAuth2 - open standard for authentication
  - Two versions
    - Container - Container
    - App - Container

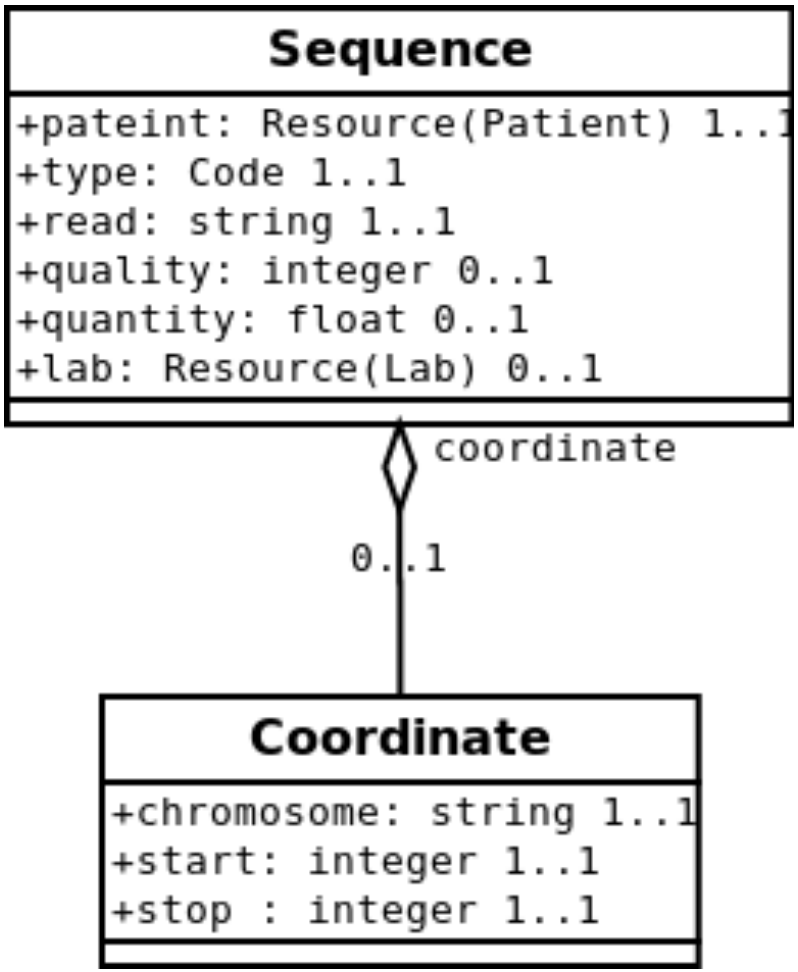
- **Data representation**
  - Structured in FHIR-style (Fast Health Interoperability Resource)
  - Interlinked data models
  - Grouped by **records**


Alignment
Variant
Sequences
?
Result(Files)
Lab


1..1
File
+type: code 1..1
+url: uri 1..1
file
Variant
+patient: Resource(Patient) 1..1
+lab: Resource(Lab) 0..1
+chrom: integer 1..1
+ref : string 1..1
+pos: integer 1..1
+alt: string 0..1
filter
1..1
Filter
+name: string 1..1
+description: string 1..1
info
1..1
Info
+name: string 1..1
+value: string 1..1
+description: string 1..1
sample
format
1..1
1..1
Format
+name: string 1..1
+description: string 1..1
Sample
field
1..*
Field
+name: string 1..1
+value: string 1..1


Lab
+name: string 1..1
+Patient: Resource(Patient) 1..1
+organism: CodeableConcept 0..1
+species: CodeableConcept 0..1
+time: Period 0..1
Sequence
+pateint: Resource(Patient) 1..1
+type: Code 1..1
+read: string 1..1
+quality: integer 0..1
+quantity: float 0..1
+lab: Resource(Lab) 0..1
coordinate
0..1
Coordinate
+chromosome: string 1..1
+start: integer 1..1
+stop : integer 1..1

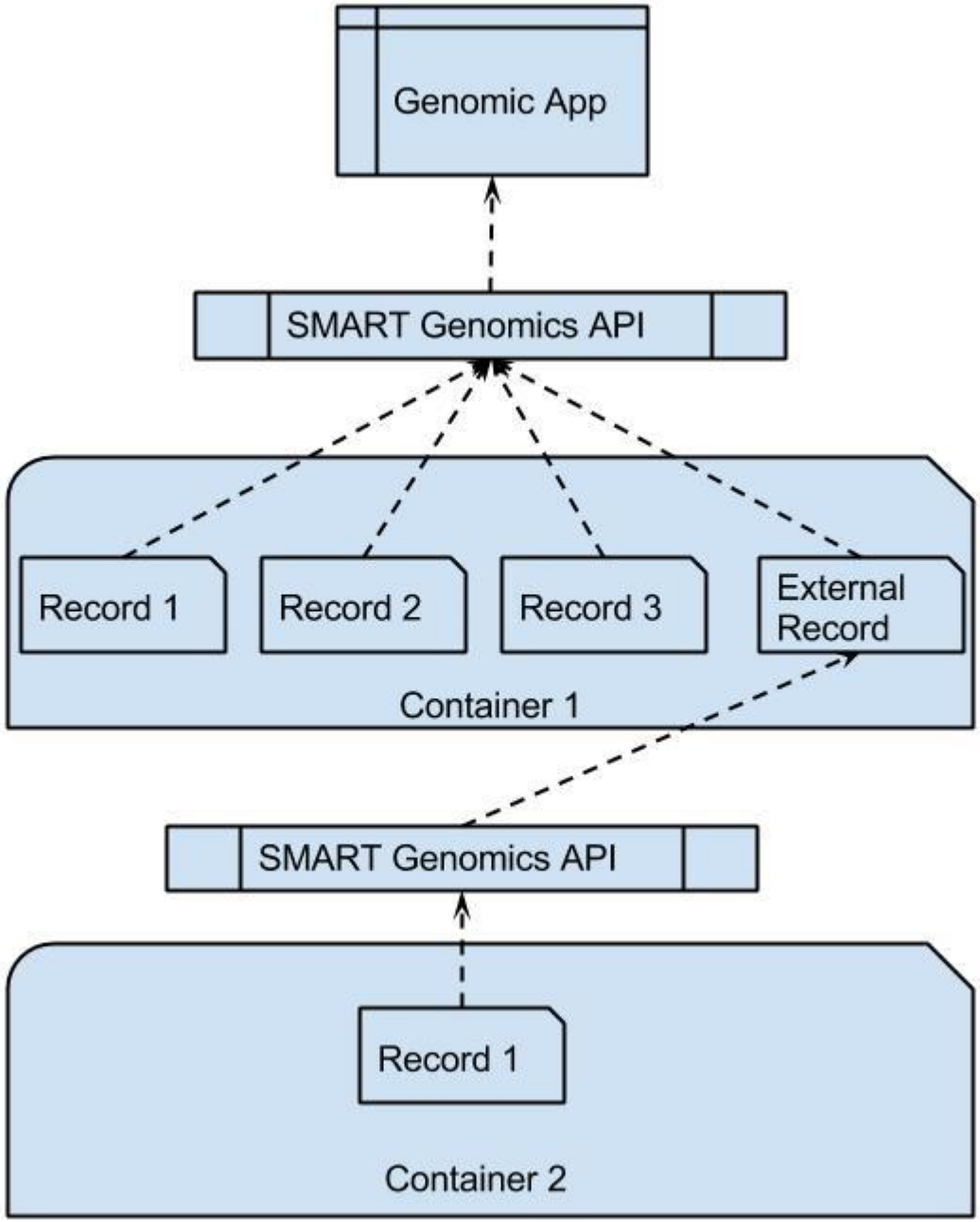
Resource Example



SMART Genomics API

Resources are organized by '**record**':

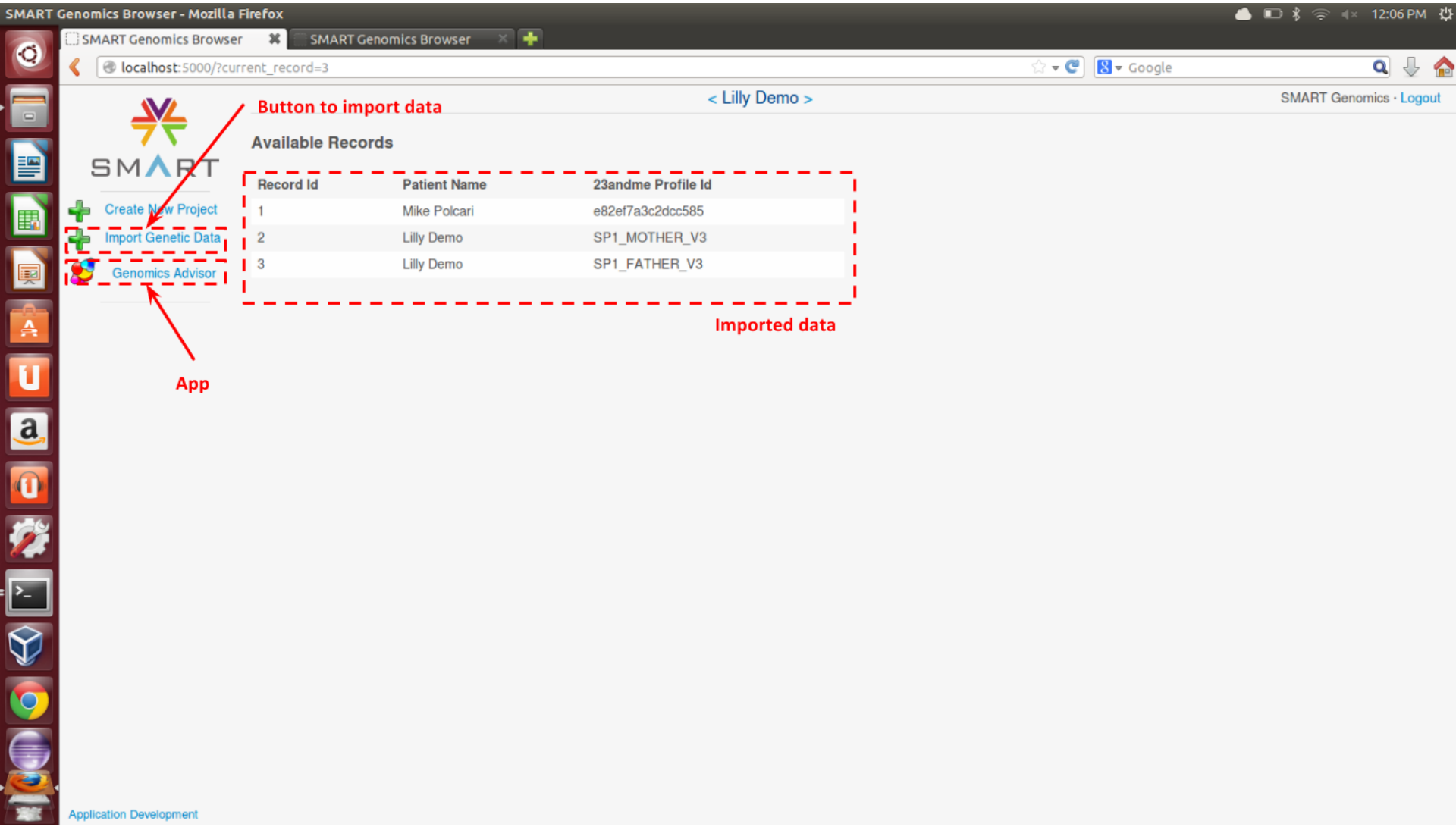
- A **record** groups all resources owned by a patient
- An account contains one or more **records**
- A **record** can either be internal or external
  - Both are perceived as the same by external source


Genomic App
SMART Genomics API
Record 1
Record 2
Record 3
External Record
Container 1
SMART Genomics API
Record 1
Container 2

Current Implementation

- **Imports data from cloud services and translates to SMART Genomics' format**

- **Demo app**
  - Genomics Advisor (collaboration with Peijin Zhang)


SMART Genomics Browser - Mozilla Firefox
12:06 PM
SMART Genomics Browser
SMART Genomics Browser
localhost:5000/?current_record=3
Google
< Lilly Demo >
SMART Genomics · Logout
Button to import data
SMART
Create New Project
Import Genetic Data
Genomics Advisor
Available Records
Record Id
Patient Name
23andme Profile Id
1
Mike Polcari
e82ef7a3c2dcc585
2
Lilly Demo
SP1_MOTHER_V3
3
Lilly Demo
SP1_FATHER_V3
Imported data
App
Application Development

SMART Genomics Browser - Mozilla Firefox

SMART Genomics Browser | SMART Genomics Browser

localhost:5000/?app=1

< Lilly Demo >

SMART Genomics · Logout

SMART

Create New Project

Import Genetic Data

Genomics Advisor

Application Development

Demo, Lilly

DOB 1999-12-08 AGE 13 SEX FEMALE

SNP	Locus	CHRM	Code	Risk	Frequency
Type 1 Diabetes					
rs5753037	22Q12	22	CC	0.93	37.2%
rs3129934	HLA-DRB1	6	CC	1.34	70.6%
rs2542151	PTPN2	18	TT	0.91	70.1%
rs3788013	UBASH3A	21	CC	0.88	30.2%
				Total Relative Risk: 1.0	
Type 2 Diabetes					
rs2877716	ADCY5	3	CC	1.05	59.3%
rs4402960	IGF2BP2	3	GG	0.92	46.2%
rs5215	KCNJ11	11	TT	0.90	42.2%
rs2237892	KCNQ1	11	CC	1.03	86.5%
rs2793831	NOTCH2	1	TT	0.97	79.9%
rs7903146	TCF7L2	10	CC	0.82	51.8%
rs7578597	THADA	2	TT	1.03	81.4%
rs7961581	TSPAN8	12	TT	0.95	53.4%
				Total Relative Risk: 0.7	
Hypertension					
rs12413409	CYP17A1	10	GG	1.03	82.8%
rs17367504	MTHFR	1	AA	1.03	74.0%
				Total Relative Risk: 1.06	
Coronary Heart Disease					
rs1746048	CXCL12	10	CC	1.05	70.6%
rs2291834	MIA3	1	CC	1.07	51.8%
rs9818870	MRAS	3	CC	0.96	71.6%
rs7739181	PHACTR1	6	GG	1.08	42.2%
rs6725887	WDR12	2	TT	0.95	74.0%
				Total Relative Risk: 1.11	

Disease Information

Patient is not at increased genomic risk for any Diabetes related comorbidities

Genomics Data

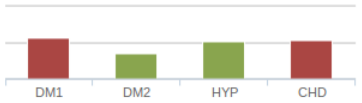
Patient ID: 3

Data format: SMART Genomics

Current Medications

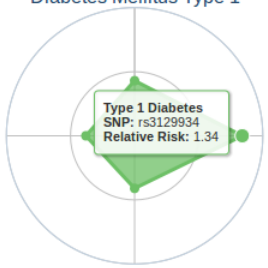
Amoxicillin, Amoxicillin

Risk Graph

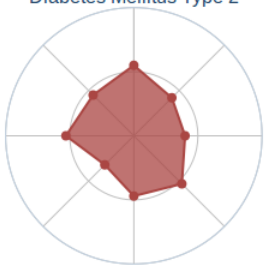


Radar Graphs

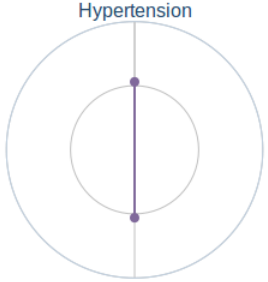
Diabetes Mellitus Type 1



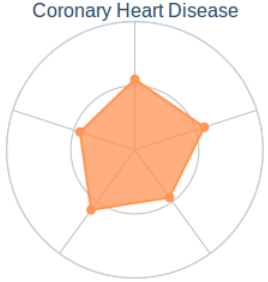
Diabetes Mellitus Type 2



Hypertension



Coronary Heart Disease

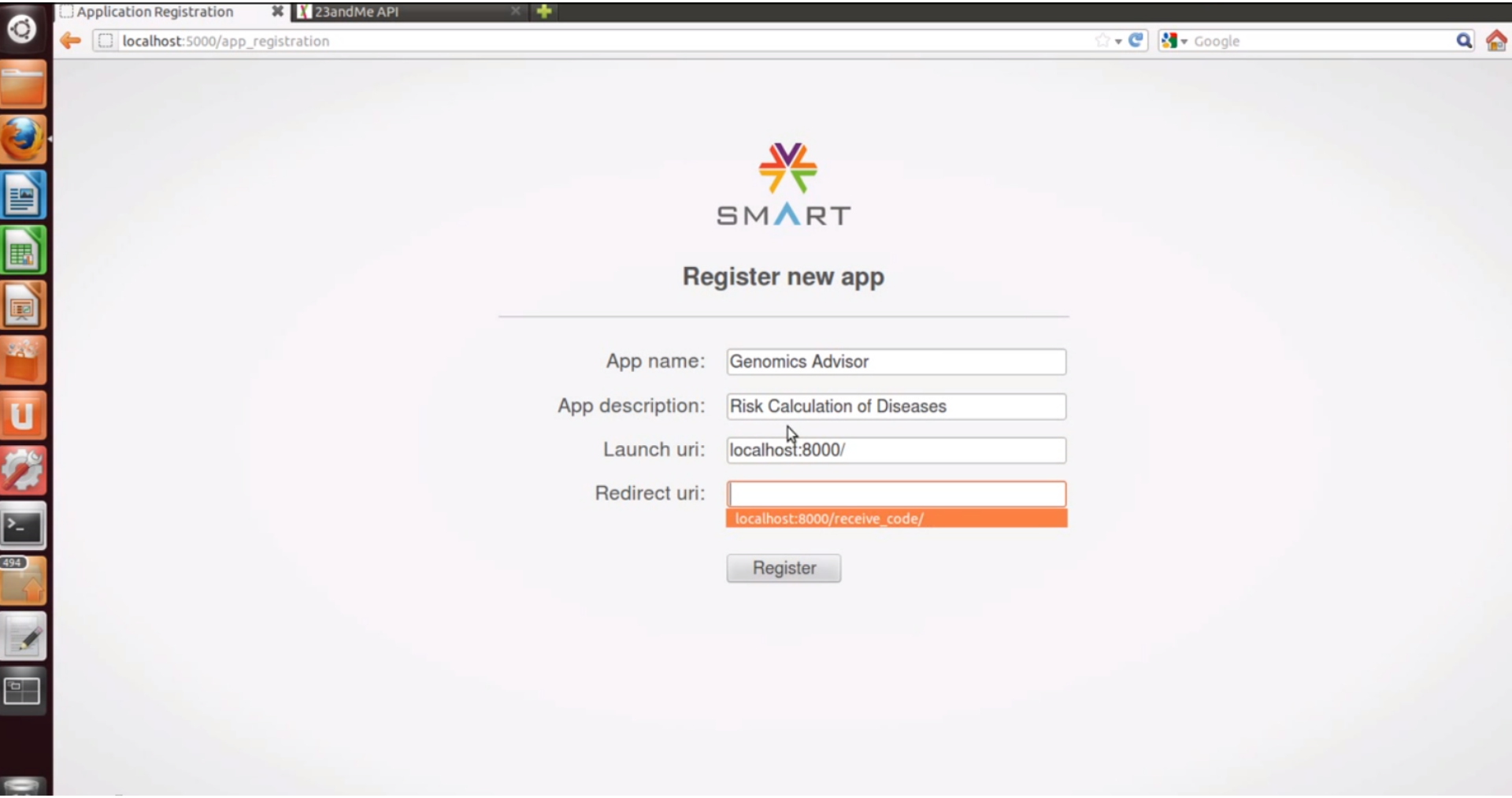


Drug Advice

No information available

Future plans

- **Design more data models**
  - Planning to add microarray (lab for gene expression)
- **Write more apps to demonstrate the API's functionalities**
  - A user friendly API playground for developer to understand basic aspects of the API
- **Distribute the code for the community to adopt**
- **Deploy as a reference container**
  - Developers can register their app and try it out


Application Registration
23andMe API
localhost:5000/app_registration
Google
SMART
Register new app
App name:
Genomics Advisor
App description:
Risk Calculation of Diseases
Launch uri:
localhost:8000/
Redirect uri:
localhost:8000/receive_code/
Register

Acknowledgements

- Dr. Gil Alterovitz for initiating this project and giving me advices
- Jason Evans for advices on designing the API calls
- Dr. Khovanova and other organizers of PRIMES for providing me this valuable opportunity
- Peijin Zhang for writing the demo app, Genomics Advisor
- My parents for supporting me

END

SMART Genomics API

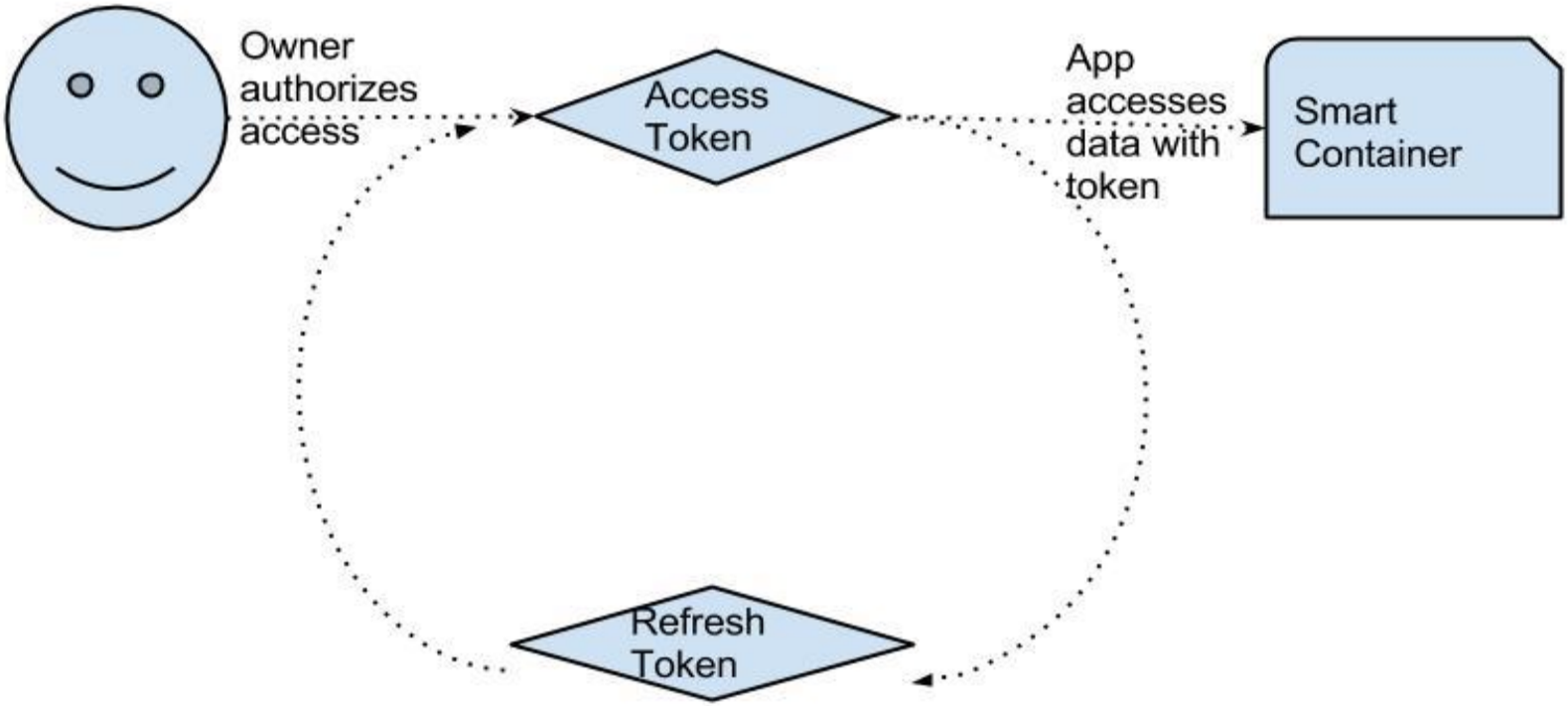
Two aspect of the API

- **Authentication**
  - OAuth2 - open standard for authentication

- **Data representation**
  - use gene coordinate as identifier -- e.g. chr13_123_124
  - Implemented FHIR
  - Resources grouped by **records**

Authentication

- Follows OAuth2 protocol
  - access token
  - refresh token
- Two versions
  - app-container -- throwaway access without refresh-token
  - container-container -- permanent access
- Enables data sharing


Owner
authorizes
access
Access
Token
App
accesses
data with
token
Smart
Container
Refresh
Token