SMART Genomics API

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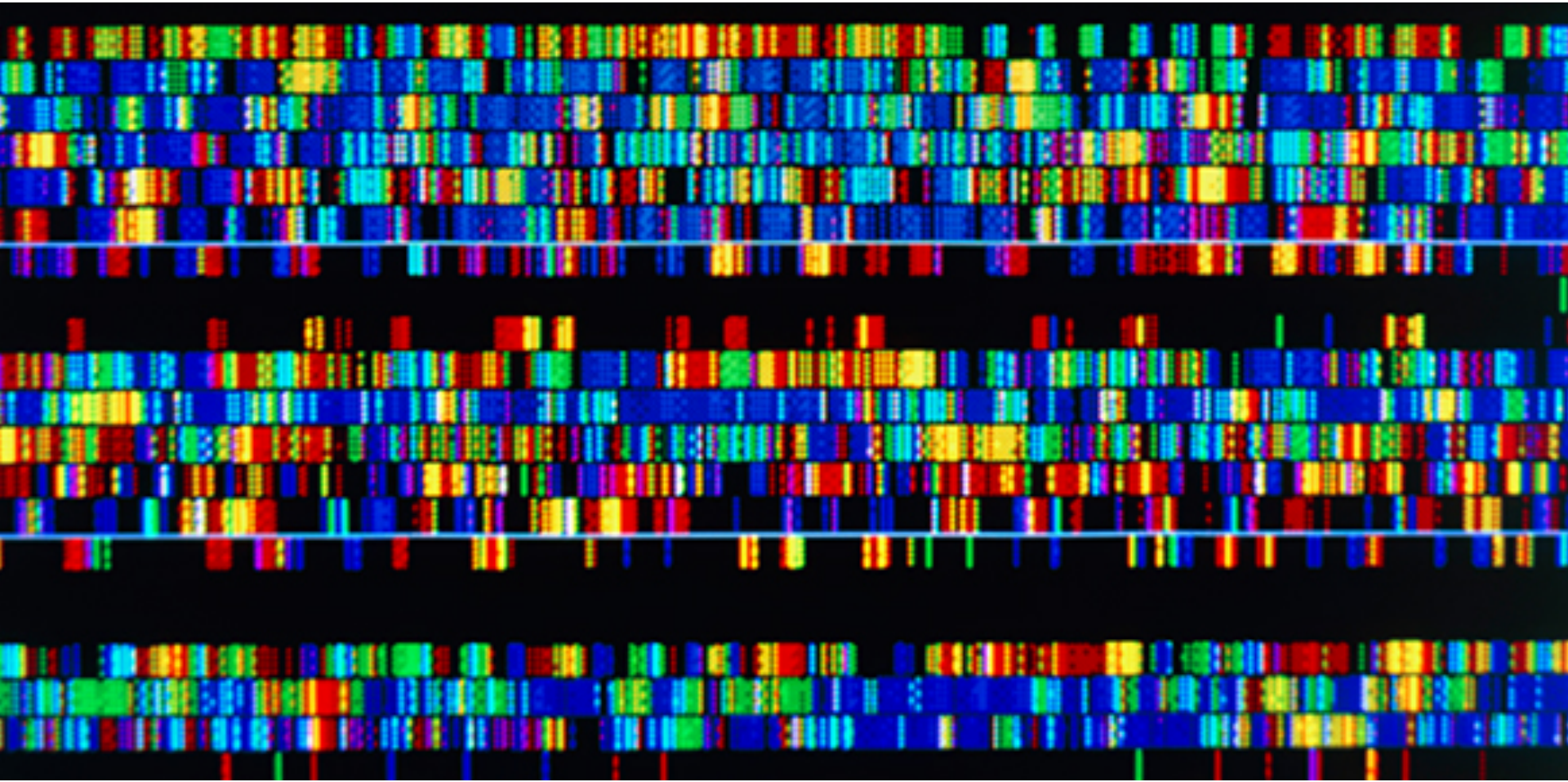
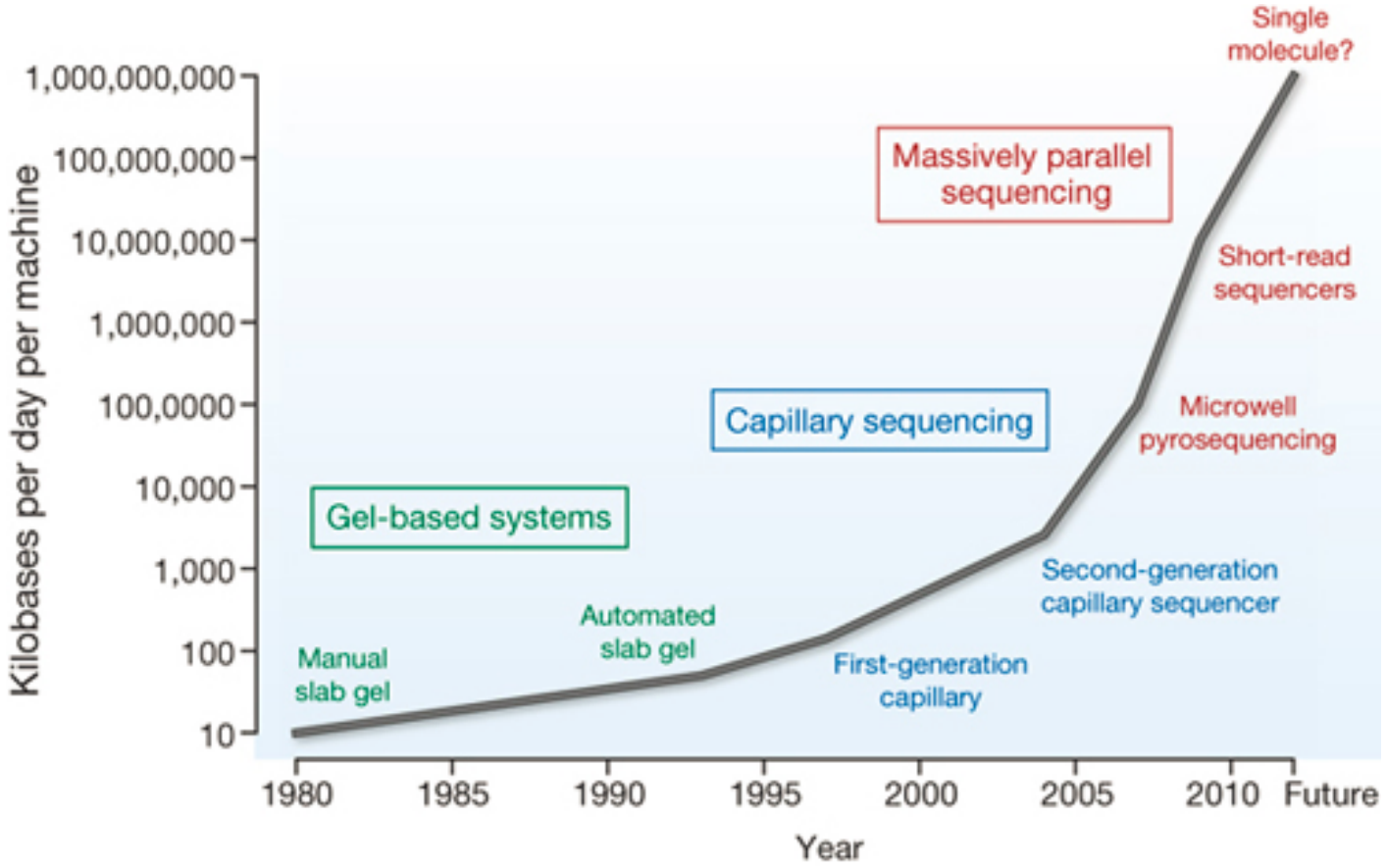
Mentor - Dr. Gil Alterovitz
PRIMES Conference
May 18, 2014



Status of Genomics

Status of Genomics

- Influx of data


Kilobases per day per machine
1,000,000,000
100,000,000
10,000,000
1,000,000
100,0000
10,000
1,000
100
10
1980
1985
1990
1995
2000
2005
2010
Future
Year
Gel-based systems
Capillary sequencing
Massively parallel sequencing
Manual slab gel
Automated slab gel
First-generation capillary
Second-generation capillary sequencer
Microwell pyrosequencing
Short-read sequencers
Single molecule?

Status of Genomics

- Influx of data

Status of Genomics

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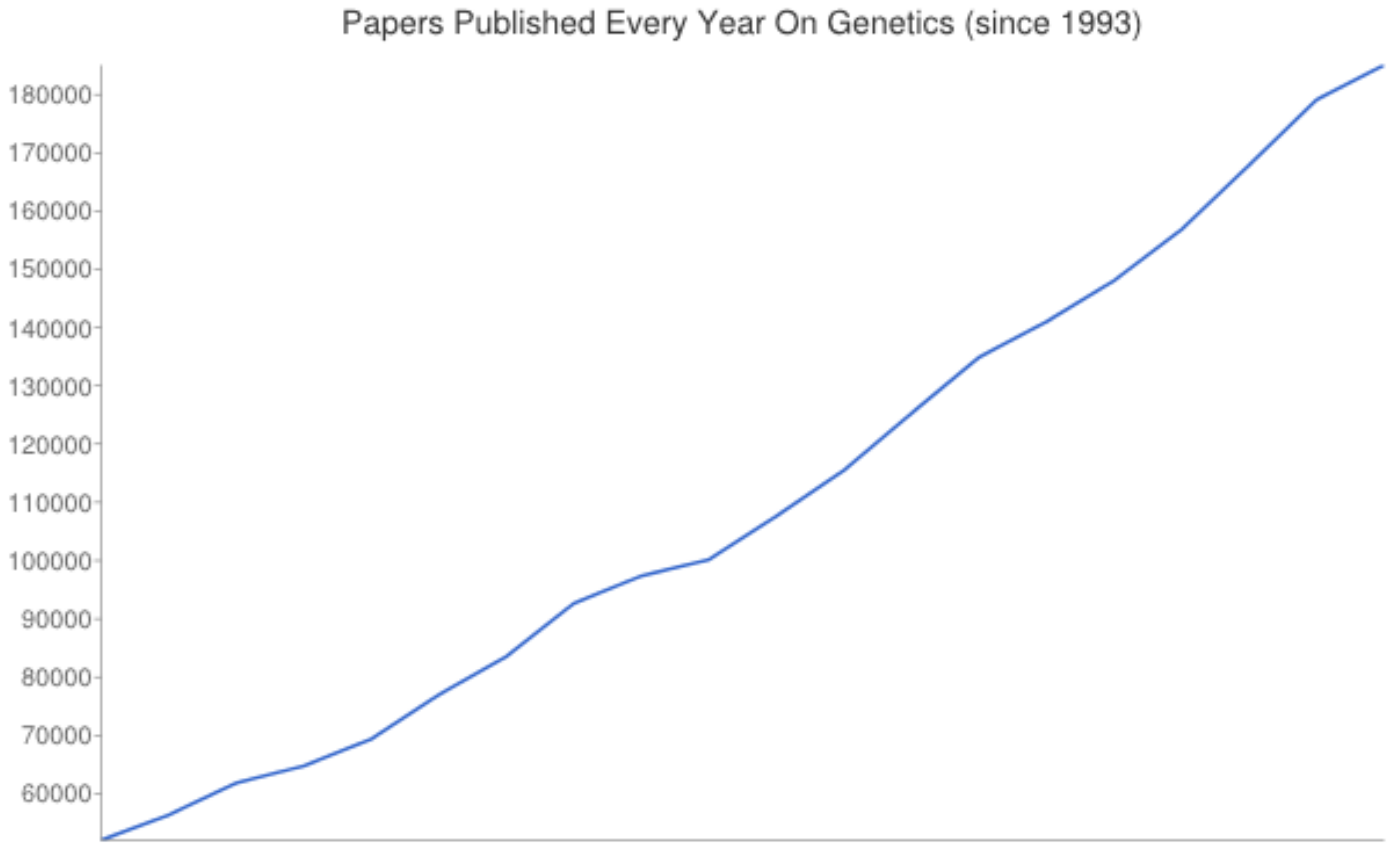
- A lot of research

Status of Genomics

- Influx of data
- A lot of research (?)


Papers Published Every Year On Genetics (since 1993)
180000
170000
160000
150000
140000
130000
120000
110000
100000
90000
80000
70000
60000

Status of Genomics

- Influx of data
- A lot of research!

Status of Genomics

- Influx of data
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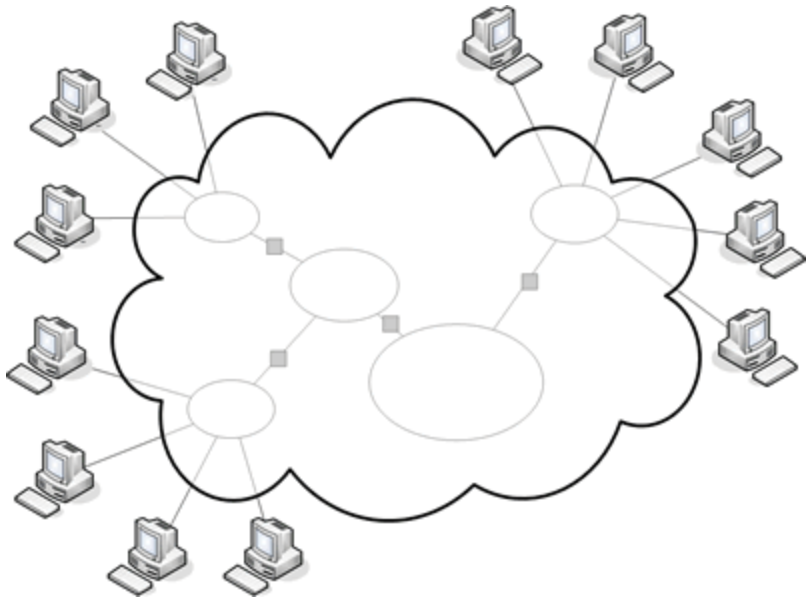
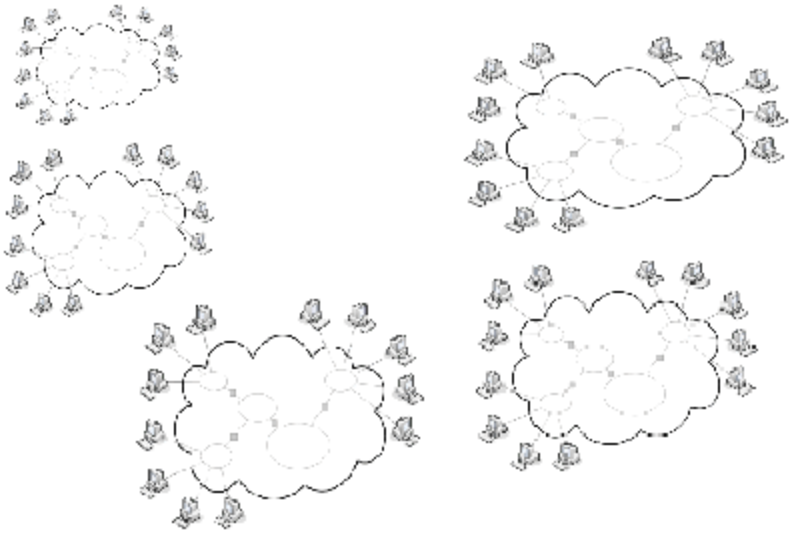
- Insufficient integration with clinics

Status of Genomics

- Influx of data
- A lot of research!
- Insufficient integration with clinics
- Insufficient collaboration/application





Solution?

A (NEMA 1-15 USA 2 pin)

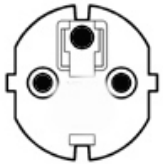
B (NEMA 5-15 USA 3 pin)

C (CEE 7/16)

D (BS546 5 A version of Type M)

E (French)

F (CEE 7/4 "Schuko")



G (BS1363 Fused 13 A,
5 A and 3A also in common use)

H (SI 32 Israel)

I (AS-3112 Argentina /
Australia / New Zealand)

J (SEV-1011 Switzerland)

K (SRAF 1962/DB Denmark)

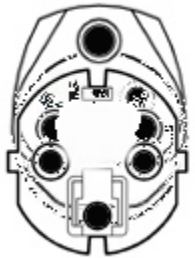
L (CEI 23-16 Chile / Italy)

M (15 A version of Type D BS546)

N Italy

O Denmark

P Israel



The ideal plug that makes everyone happy

SMART Project

- **S**ubstitutability **M**edical **A**pplication **R**eusable **T**echnology
- Funded by ONC
- Started as a Clinical project

SMART Genomics

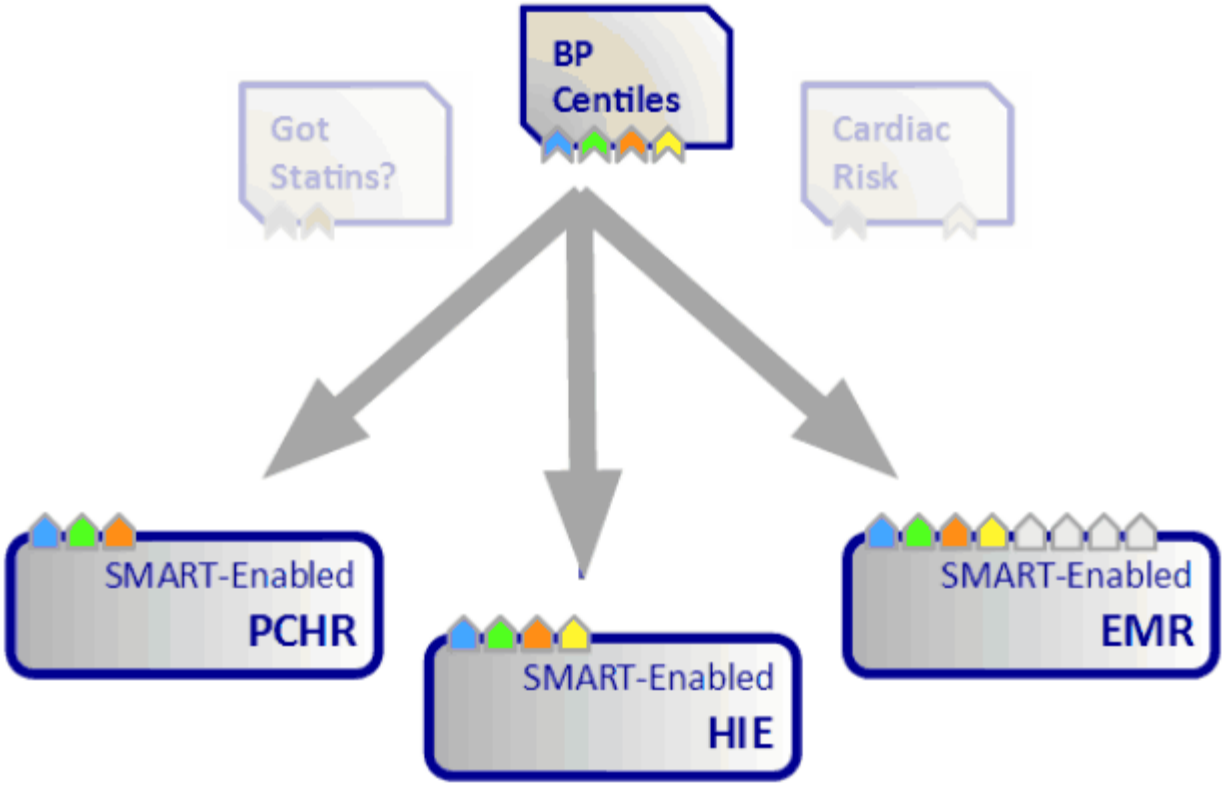
SMART Genomics

The object is simple.

SMART Genomics

The object is simple.

- Common API (**A**pplication **P**rogramming **I**nterface)


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

SMART Genomics

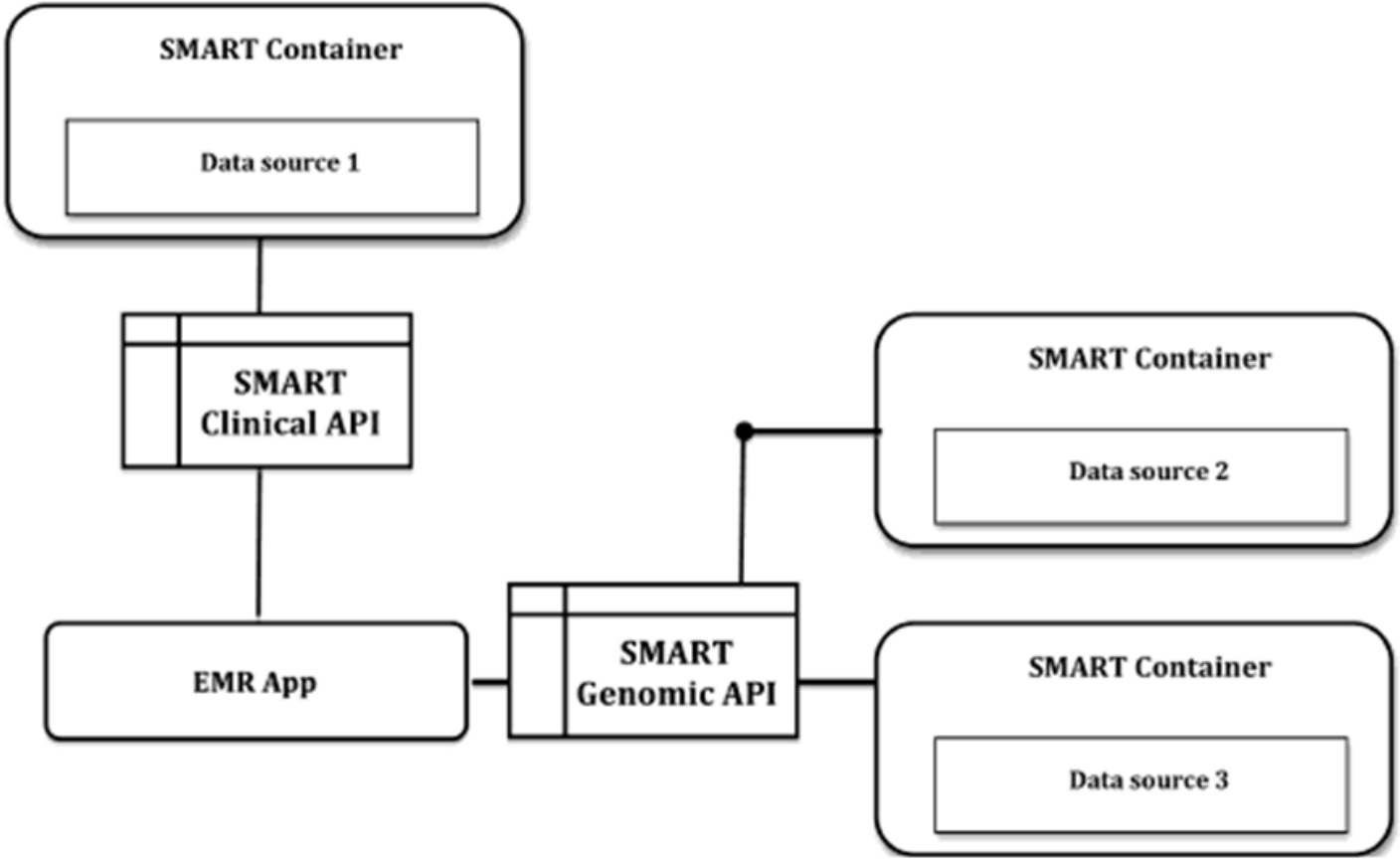
The object is simple.

- Common API (**A**pplication **P**rogramming **I**nterface)

SMART Genomics

The object is simple.

- Common API (**A**pplication **P**rogramming **I**nterface)
- Bridges labs and clinics


SMART Container
Data source 1
SMART
Clinical API
SMART Container
Data source 2
EMR App
SMART
Genomic API
SMART Container
Data source 3

Collaboration with the Industry

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- Clinic side - **FHIR** (**F**ast **H**ealth **I**nteroperability **R**esource)

Collaboration with the Industry

- Clinic side - **FHIR** (**F**ast **H**ealth **I**nteroperability **R**esource)
- Research side - **GA4GH** (**G**lobal **A**lliance for **G**enomics and **H**ealth)

Project Status

- Specification

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- Reference server

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- Demo applications

Project Status

- Specification
- Reference server
- Demo applications
  - Genomics Advisor (Web app and iOS app)
  - Variant Mapper

< Allen Vitalis >

Demo User · Logout

SMART

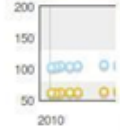
- Patient List
- Alerts
- Diabetes Monograph
- Genomics Advisor

SMART Diabetes Monograph

Vitalis, Al

DOB 2005-08-

BP goal < 130/



Genomics Ad

Patient ID:

Data format:

Last Known V

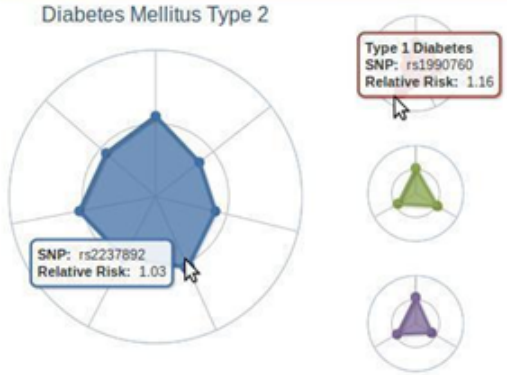
Ur tp	0-135
µalb/cre	< 30.0
SGOT	10-40
Chol	< 200
Tri	< 150
HDL	> 40
LDL	< 100
BUN	8%-25%
Cre	0.6-1.5
Glu	70-110
A1C	< 7%

SMART Diabetes Monograph Genomics Advisor

[Close]

SNP	Locus	CHRM	Code	Risk	Frequency
Type 1 Diabetes					
rs7202877	16Q23	16	TT	0.95	81.0%
rs5753037	22Q12	22	CC	0.93	37.2%
rs3087243	CTLA4	2	AG	0.98	49.4%
rs3825932	CTSH	15	CT	0.94	43.4%
rs1990760	IFIH1	2	TT	1.16	36.2%
rs6822844	IL2	4	GT	0.73	30.2%
rs3184504	SH2B3	12	CC	0.74	26.0%
				Total Relative Risk: 0.51	
Type 2 Diabetes					
rs2877716	ADCY5	3	CC	1.05	59.3%
rs2383208	CDKN2A	9	AG	0.88	28.2%
rs4402960	IGF2BP2	3	GG	0.92	46.2%
rs2237892	KCNQ1	11	CC	1.03	86.5%
rs2793831	NOTCH2	1	TT	0.97	79.9%
rs7578597	THADA	2	TT	1.03	81.4%
rs7961581	TSPAN8	12	TT	0.95	53.4%
				Total Relative Risk: 0.83	
Hypertension					
rs12413409	CYP17A1	10	GG	1.03	82.8%
rs17367504	MTHFR	1	AA	1.03	74.0%
rs3184504	SH2B3	12	CC	0.93	28.1%
				Total Relative Risk: 0.99	
Coronary Heart Disease					
rs1746048	CXCL12	10	CC	1.05	70.6%
rs3184504	SH2B3	12	CC	0.89	30.2%
rs6725887	WDR12	2	TT	0.95	74.0%
				Total Relative Risk: 0.89	

Radar Graphs



Genomics Advice

Disease Info

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

Drug Advice

SNP	Genotype	Advice

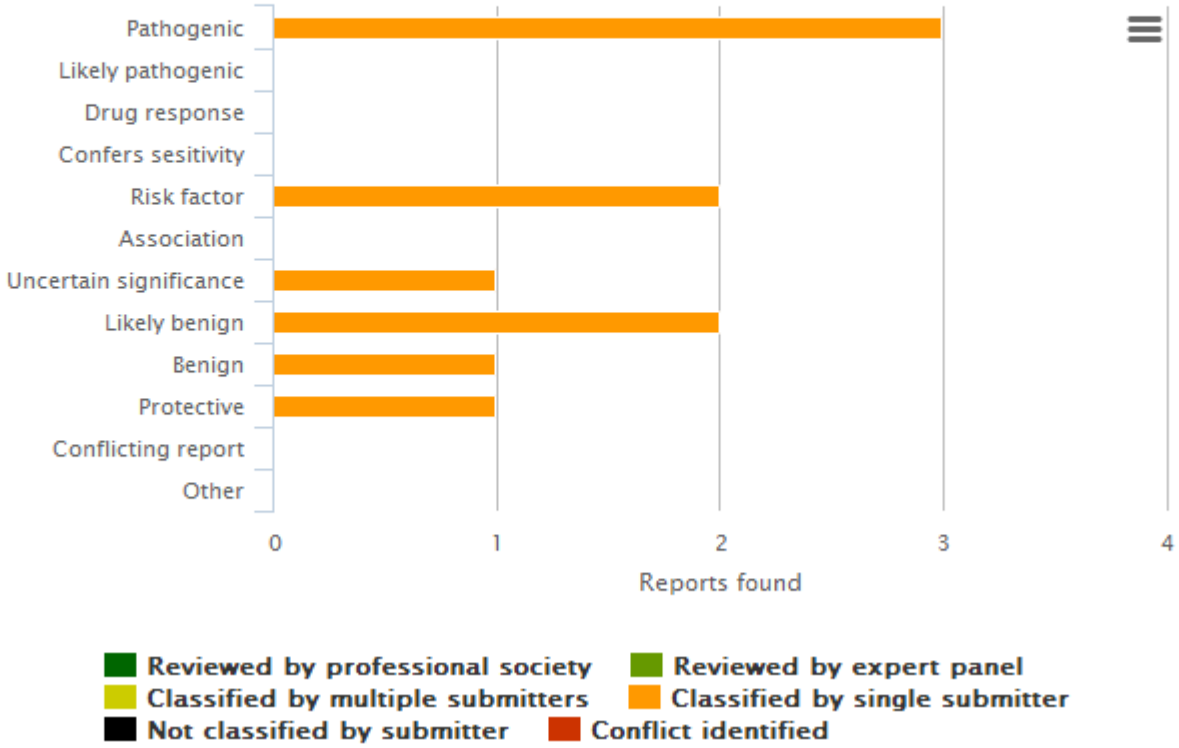
No Information Available

Menu | Disconnect | search for trait | Search | Lilly Demo

Trait/Phenotype

Familial Mediterranean Fever
Zellweger Syndrome Spectrum
Phenylketonuria
Crohn's Disease
Bipolar Disorder
Ulcerative Colitis
Celiac Disease
Hereditary Fructose Intolerance
Familial Hyperinsulinism
Age-related Macular Degenerati

Breast Cancer



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  - Partners Healthcare

Acknowledgements

- John Zhang
- Dr. Gil Alterovitz
- Community
- PRIMES