Status of Genomics
Status of Genomics

- Influx of data
Status of Genomics

- Influx of data
Status of Genomics

- Influx of data
- A lot of research
Status of Genomics

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

- Influx of data
- A lot of research!
Status of Genomics

- Influx of data
- A lot of research!
- Insufficient integration with clinics
Status of Genomics

- Influx of data
- A lot of research!
- Insufficient integration with clinics
- Insufficient collaboration/application
Solution?
The ideal plug that makes everyone happy
SMART Project

- Substitutability Medical Application Reusable Technology
- Funded by ONC
- Started as a Clinical project
SMART Genomics
SMART Genomics

The object is simple.
SMART Genomics

The object is simple.

- Common API (*Application Programming Interface*)
SMART Genomics

The object is simple.

- Common API (Application Programming Interface)
The object is simple.

- Common API (Application Programming Interface)
- Bridges labs and clinics
Collaboration with the Industry
Collaboration with the Industry

- Clinic side - **FHIR (Fast Health Interoperability Resource)**
Collaboration with the Industry

- Clinic side - **FHIR** (Fast Health Interoperability Resource)
- Research side - **GA4GH** (Global Alliance for Genomics and Health)
Project Status

- Specification
Project Status

- Specification
- Reference server
Project Status

- Specification
- Reference server
- Demo applications
Project Status

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

**Type 1 Diabetes**

<table>
<thead>
<tr>
<th>SNP</th>
<th>Locus</th>
<th>CHRM</th>
<th>Code</th>
<th>Risk</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>rs7262877</td>
<td>16C23</td>
<td>16</td>
<td>TT</td>
<td>0.95</td>
<td>81.6%</td>
</tr>
<tr>
<td>rs5730307</td>
<td>22G12</td>
<td>22</td>
<td>CC</td>
<td>0.93</td>
<td>97.2%</td>
</tr>
<tr>
<td>rs3007243</td>
<td>2AG</td>
<td>2</td>
<td>AG</td>
<td>0.98</td>
<td>49.4%</td>
</tr>
<tr>
<td>rs3826921</td>
<td>15CTH</td>
<td>15</td>
<td>CT</td>
<td>0.94</td>
<td>43.4%</td>
</tr>
<tr>
<td>rs1990760</td>
<td>2TT</td>
<td>2</td>
<td>TT</td>
<td>1.16</td>
<td>36.2%</td>
</tr>
</tbody>
</table>

**Total Relative Risk: 0.51**

**Type 2 Diabetes**

<table>
<thead>
<tr>
<th>SNP</th>
<th>Locus</th>
<th>CHRM</th>
<th>Code</th>
<th>Risk</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>rs2877716</td>
<td>ADCYS</td>
<td>3</td>
<td>CG</td>
<td>1.05</td>
<td>59.3%</td>
</tr>
<tr>
<td>rs2332086</td>
<td>CDXN2A</td>
<td>9</td>
<td>AG</td>
<td>0.88</td>
<td>28.2%</td>
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<tr>
<td>rs4469260</td>
<td>IGF2B2</td>
<td>3</td>
<td>GG</td>
<td>0.92</td>
<td>46.2%</td>
</tr>
<tr>
<td>rs2237892</td>
<td>KCHN1</td>
<td>11</td>
<td>CC</td>
<td>1.03</td>
<td>86.5%</td>
</tr>
<tr>
<td>rs7585976</td>
<td>THADA</td>
<td>2</td>
<td>TT</td>
<td>1.03</td>
<td>81.4%</td>
</tr>
<tr>
<td>rs7941581</td>
<td>TSPAN8</td>
<td>12</td>
<td>TT</td>
<td>0.95</td>
<td>53.4%</td>
</tr>
</tbody>
</table>

**Total Relative Risk: 0.83**

### Disease Info

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

### Drug Advice

**SNP** | **Genotype** | **Advice**
<table>
<thead>
<tr>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>No Information Available</td>
</tr>
</tbody>
</table>

### Genomics Advisor

- **Type 1 Diabetes**
  - SNP: rs1990760
  - Relative Risk: 1.16

- **Type 2 Diabetes**
  - SNP: rs2337892
  - Relative Risk: 1.03
Project Status

- Specification
- Reference server
- Demo applications
- Usecase
Project Status

- Specification
- Reference server
- Demo applications
- Use case
  - Partners Healthcare
Acknowledgements

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