Integration and analysis of heterogeneous big data for precision medicine and suggested treatments for different types of patients.

http://project-iasis.eu

@Project_IASIS

iASiS: Big Data to Support Precision Medicine and Public Health Policy

Vassiliki Rentoumi
NCSR “Demokritos”, Greece
INNOVATHENS

SC1-PM-18-2016: Big Data supporting Public Health policies
iASiS Basic Facts

• Title: Integration and analysis of heterogeneous big data for precision medicine and suggested treatments for different types of patients

• Topic: H2020-SC1-PM-18-2016 - Big Data supporting Public Health policies

• Contract No.: 727658

• Budget: € 4.3M

• Project Officer: Gisele Roesems
Motivation

- Epidemiological data analysis is not sufficient for public health policies in the era of personalized/precision medicine.
- We also need explanations, e.g. why a treatment ought to work better for one type of patient than another.
- Therefore, we need to combine breadth (across a population) with depth (e.g. personal genome) in the analysis.
- Big data analysis can address both breadth and depth, under the appropriate framework. That’s iASiS!
Vision and Objectives

**iASiS Vision:**

Turn clinical, pharmacogenomics, and other Big Data into actionable knowledge for personalized medicine and health policy-making

**iASiS Objectives:**

- Integrate automated unstructured and structured data analysis, image analysis, and sequence analysis into a Big Data framework
- Use the iASiS framework to support personalized diagnosis and treatment
The iASiS Framework

- iASiS focuses on **two use cases**:
  - Lung cancer
  - Alzheimer’s disease

- General-purpose drugs are often ineffective

<table>
<thead>
<tr>
<th>Drug Type</th>
<th>Ineffective (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anti-depressants (SSRIs)</td>
<td>38%</td>
</tr>
<tr>
<td>Asthma Drugs</td>
<td>40%</td>
</tr>
<tr>
<td>Diabetes Drugs</td>
<td>43%</td>
</tr>
<tr>
<td>Arthritis Drugs</td>
<td>50%</td>
</tr>
<tr>
<td>Alzheimer’s Drugs</td>
<td>70%</td>
</tr>
<tr>
<td>Cancer Drugs</td>
<td>75%</td>
</tr>
</tbody>
</table>
The iASiS Framework

• iASiS analyzes:
  • EHRs (English & Spanish)
  • MRI & PET/CT images
  • Genomic data (e.g. liquid biopsy samples)
  • Related bibliography (e.g. PubMed)
  • Biomedical databases (e.g. DrugBank)
  • Biomedical ontologies (e.g. GO, UMLS)
The iASiS Framework

- Extracted knowledge is fused in the iASiS knowledge graph
  - Unified semantic schema
  - Linked data
  - Machine-processable knowledge

- iASiS end-users can:
  - Perform natural language questions
  - Receive answers along with justifications
  - Identify patterns in patient populations
  - Make informed decisions

- All steps of data management and analytics enforce privacy and access control
Alzheimer's Disease Pilot

Motivation:

• Approximately, **10% of people over 65** suffer from Alzheimer’s

• **Heterogeneity** of the symptoms impedes accurate diagnosis and treatments

iASiS will enable:

• **Discovery of patterns associated with** prognosis, outcomes and response to treatments

• **Association of medical and lifestyle advice to Alzheimer’s risk** and stages of severity
Alzheimer's Disease Pilot Data

- EHRs in English
- MRI Brain Images
- Genomic Data

- Pharmacological knowledge extracted from publicly available datasets
- Biomedical ontologies and taxonomies
  - terminology standardization
  - semantically describing the EHRs
iASiS Usage for Alzheimer's Disease (1)

- iASiS actors (e.g. Geneticist, Biological scientist, Clinician, Patient) ask:
  - Q1: How many Alzheimer’s patients have genetic maternal/paternal family history and how many don’t?
  - iASiS KG: Heterogeneous data get integrated and analyzed
    - Likely outcome: e.g. 20%
    - Personalized Medicine
      - this 20% may share a genetic variant, that might lead to early diagnosis
      - use or development of new drugs that target this genetic variant may lead to prevention
iASiS Usage for Alzheimer's Disease (2)

- iASiS actors (e.g. Geneticist, Biological scientist, Clinician, Patient) ask:
  - Q2: Are there comorbidities in the context of family history and genetic determinants?
  - iASis KG: correlation of Alzheimer’s with family history and comorbidities.

  - Likely outcome:
    - Yes, e.g. depression

  - Personalised medicine:
    - diagnosis of depression and identification of genetic determinants may lead to early diagnosis
    - may influence policy making on the importance of diagnosing depression
Lung Cancer Pilot

**Motivation:**
- Lung cancer among the most common and deadly diseases
- Lung cancer is a heterogeneous disease. Characteristics differ among patients and tumor regions

**iASiS will enable:**
- Discovery of correlations among tumor spread, prognosis, response to treatment
- Unraveling molecular mechanisms that predict response to different tumor types (signatures)
Beyond Data Analysis

• iASiS handles **sensitive patient data** from hospitals
  • EHRs, MRI and PET/CT images, blood and liquid biopsy samples
  • WP dedicated to Ethics Requirements

• Ethics Committee with external advisor to oversee the adherence to:
  • Guidelines for Good Clinical Practice – CPMP/ICH/135/95
  • Clinical Trials Directive – 2001/20/EC
  • WMA Declaration of Helsinki
  • International Cancer Genome Consortium Principles
  • Dementia Consortium Principles
  • Written informed consent from patients (option to opt out)
Beyond Data Analysis

• Data management plan
  • Procedures for data collection, storage, protection, retention and destruction
  • Data Protection Directive – 95/46/EC
  • Data Protection Act 1998
  • Human Rights Act 1998
  • Computer Misuse Act 1990
  • Copyright, Designs and Patents Act 1988

• Privacy and Security
  • Anonymized data
  • Data access regulated by European/National laws and hospital committees
  • Data stored in firewall protected servers accessible within trust’s premises only
iASiS Partners