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Data and text mining



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# **Interactive Integrated Electronic Health Records**

# Shane Crinion \*, Pilib Ó Broin

<sup>1</sup>School of Mathematics, Statistics & Applied Mathematics, National University of Ireland, Galway, H91 H3CY

\*To whom correspondence should be addressed.

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#### **Abstract**

Motivation: The a

Contact: name@bio.com

Supplementary information: Supplementary data are available at Bioinformatics online.

#### 1 Introduction

The widespread implementation of the electronic health record (EHR) has improved healthcare through reduced processing times and cost, improved quality of patient care and detailed clinical information through longitudinal storage Denny *et al.*, 2010. Longitudinal clinical data can be useful for applied clinical research like identifying drug adverse effects or respositioning of drugs. . . . . . . text follows.

$$\sum x + y = Z \tag{1}$$

#### 2 Approach

## 3 Methods

### 3.1 Population

The clinical data modelling diabetes patients was generated using modules from Synthea, a synthetic patient generator. The population consisted of 1000 individuals originating from Massachusetts, MA. The selection criteria were that they suffered from a metabolic disorder. The wellness encounters module provide was used to generate useful observations including BMI, insulin resistance and glycated haemoglobin . Diabetes is stored in 2 modules namely metabolic syndrome disease and metabolic syndrome care. These contain diabetes specific measurements including hemoglobin levels.

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2 Sample et al.

#### 3.2 Genotypic Data

Genotypic data was extracted from the Gene Expression Omnibus. Montesanto et. al indicate that CAT, FTO and UCP1 are all associated with type 2 diabetes. Search each of these genes with type 2 diabetes directs me to the same series: GSE25462. To model the effect of medication on gene expression, the commonly prescribed drug Simvastatin 20 MG Oral Tablet (n = 46) was used in the search. The search term

```
type[All Fields] AND 2[All Fields]
AND diabetes[All Fields]
AND Simvastatin[All Fields])
AND ("Homo sapiens"[Organism]
AND ("50"[n_samples]:"100"[n_samples])
```

was used to find gene expression variation associated with the disease and drug and the . The top result measured APOC3 levels in subgroups Type 2 diabetic and insulin-resistant but normoglycemic cohorts in skeletal muscle cells. The primary outcome of the search was to identify a suitable model for diabetes associated gene expression variation. Studies have suggested a linked between mitochondrial oxidative homeostasis and predisposition to diabetic vascular complications (Alberto Montesanto, 2018). Plasma APOC3 associates with almost half of HDL (Zvintzou E, 2017); HDL-associated apoA-I and lysosphingolipids protect against ATP reduction resulting from reverse cholesterol transport in mitochondria CR White, 2017). When sorting this by subset effect, TPM1 comes up as highest.

Genotyping was performed using [ HG-U133 Plus 2 ] Affymetrix Human Genome U133 Plus 2.0 Array which, according to their website, provides the greatest accuracy and reproducibility of any microarray platform.

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## 3.3 Development of integrated genomic and clinical data

The generated data contained numerous CSV files containing: allergies, careplans, conditions, encounters, imaging studies, observations, organizations, patients and procedures. The observations were merged with patients to put the name to each wellness and disease associated measurement. The medical coding or the associated observation description could then be used to compar

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# FPO

Fig. 1. Caption, caption

#### 3.4 Gene Expression Analysis

The Affymetrix GeneChip Human Genome U133 Plus 2.0 Array is a one-colour microarray that human array that identically represents the GeneChip Human Genome U133 set. The .CEL raw data files were imported and analysed using Bioconductor packages limma and oligo on R 3.5.3 "Great Truth".(possibly used Affy too?). Other software that can be used for the analysis is Galaxy, GenePattern, GeneSpring.

Quality control: R is used to make plots that can be used to identify poor quality probes or technical issues that could effect the results.

Normalisation prevents technical variation between assays from displaying as biological variation. The robust multi-array average (RMA) is used from the 'oligo' package to

Differential expression analysis is used to identify the genes that differ under biological conditions. Multiple testing correction is performed using the 'limma' method for log2 fold change ratio between test and control conditions and provides an 'adjusted' p-value for the differences due to multiple testing.

#### 4 Discussion

Talk about what the framework is and how easy to use the I2EHR is. Figure 2 Talk about a second use of I2EHR. Talk about how it was implemented through the treatment of type 2 diabetes patients. Bofelli *et al.*, 2000 might want to know about text text text The graphical data are a systemic way to analyse clinical observations and treatments and influence the focus for genomic research. This approach intends to give clinical researchers the insight required to improve diabetes treatment and help them to select the right genomic analysis that need to be performed to gain useful knowledge on the molecular background to the disease.

Synthea proved very useful for the acquisition of clinical data; Synthea improved efficiency by allowing instant usage and data manipulation to model the cohort effectively and eliminating waiting time. Efficiency is also improved by the elimination of issues associated with privacy including de-personalising the data and requesting new data privileges. Issue with Synthea is the discrepancies with demographic data that have been noted (ref). Synthea developers have addressed this and are continuously improving the likelyness to real data through their GitHub account (link).

Limitations of microarrays: limitations include cross-hybridisation induced high background limitations

# 5 Conclusion

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## Acknowledgements

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