

ASSIST: Association Studies assisted by Inference and Semantic Technologies



Project Leader

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Project Details

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Project Web Site

<http://assist.itl.gr/>

Keywords

Healthcare, medical archives, cervical cancer, association studies, data integration.

Overview

To facilitate association studies on genotypic and phenotypic factors related to cervical cancer, ASSIST resorts to medical inference applied on real patient data. Following the semantic approach, ASSIST is relying on available standards and recent research achievements in the area of semantics and soft computing in order to build its Medical Knowledge Base. The targeted virtual unification of the participating archives and interpretation of their content relies upon the semantic indexing of their records.

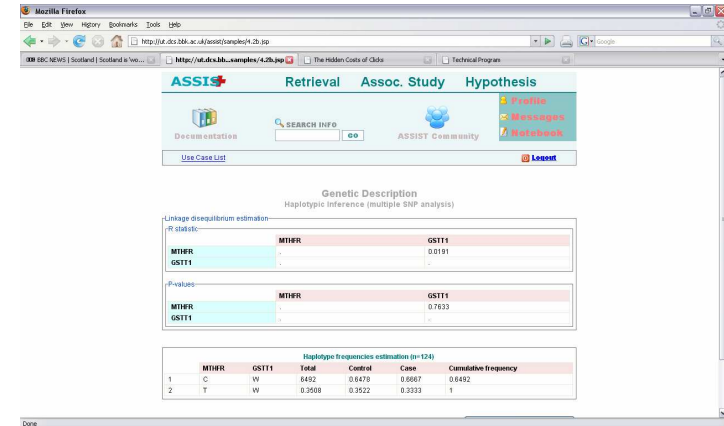
Objectives

Unlike the conventional way of treating stored medical information as alphanumeric data structures whose interpretation is carried out by the human user, ASSIST's inference engine:

- supports the virtual unification of the participating archives by translating medical concepts into syntactic values that the legacy systems of the participating archives may perceive; and
- undertakes the whole process of statistically evaluating medical hypotheses and producing medically important associations based on the collected data.

In addition to the inference engine, ASSIST incorporates two important interfacing modules:

- The first is the interface to its users, mainly medical researchers and geneticists. This graphical interface is medical knowledge aware in the sense that it allows expression of domain specific queries and particular hypotheses by referring to medical ontologies contained in the Medical Knowledge Base.
- The second type includes the interfaces to the participating medical archives, supporting exchange of data between them and ASSIST's core engine and in a way transparent to the end user.



The screenshot shows the ASSIST web application interface. It features a navigation menu with 'Retrieval', 'Assoc. Study', and 'Hypothesis' tabs. Below the navigation is a search bar and a 'Use Case List' link. The main content area displays 'Genetic Description' results for a 'Haplotype inference (multiple SNP analysis)'. It includes three tables:

| Linkage disequilibrium estimation | | |
|-----------------------------------|-------|--------|
| r (diseq) | MTHFR | GSTT1 |
| MTHFR | - | 0.0191 |
| GSTT1 | - | - |

| P-values | | |
|----------|-------|--------|
| | MTHFR | GSTT1 |
| MTHFR | - | 0.7633 |
| GSTT1 | - | - |

| Haplotype frequencies estimation (n=124) | | | | | | |
|--|-------|-------|--------|---------|--------|----------------------|
| | MTHFR | GSTT1 | Total | Control | Case | Cumulative frequency |
| 1 | C | W | 8492 | 0.6479 | 0.6887 | 0.6492 |
| 2 | T | W | 0.3508 | 0.3522 | 0.3113 | 1 |

ASSIST user interface: results of an association study.

Ethics

ASSIST respects and promotes the ethical principles that guide current medical research activities and is being designed in full compliance with the legal and ethical national and EU requirements and code of practice. Special care is being taken so as to avoid violation patient privacy during system operation. To this end, only anonymised patient information is handled by the ASSIST system, produced by state-of-the art anonymisation techniques and standards.

Impact

ASSIST aims to enable the cervical cancer medical researcher to use various HPV data, environmental, lifestyle and medical history items from diverse medical records, with minimal effort and cost. The investigation of associations among all these factors and genetic data will identify risk factors that can then be used at the point of care by gynecologists to identify women who are at high risk of developing cervical cancer.