

STATISTICAL PROCESSING OF GENETIC INFORMATION IN THE MUDR RECORD

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Introduction

The important research task of the European Centre for Medical Informatics, Statistics and Epidemiology – Cardio (EuroMISE Centre – Cardio) is the applied research in the field of the electronic health record (EHR). In this frame, a pilot 3-tier EHR application named “MULTimedia Distributed Record” (MUDR) [1] was developed. Relations among collected attributes in this application are described by so-called knowledge base and the record itself is represented by a graph structure, where the knowledge base nodes are referenced from.

Applied Methods

The minimal data model of the cardiological patient [2] represented in MUDR by a cardiological knowledge base has been extended by adding *genetic semantic types* to enable representation of genetic information. On selected patients from the Obesity Sanatorium in Pobebrady and control groups from the Endocrinological Institute in Prague we monitored the APOE (*19q13.2*), LPL (*8p22*) and FABP2 (*4q28-q31*) genes. Knowledge nodes describing corresponding alleles have been added to the knowledge base.

Medical guideline libraries form an integrated part of the MUDR application layer. The main MUDR system uses XML defined by the MUDR XML Scheme [3] for the communication between clients and the application layer. We are using a MUDR application layer module *MUDR GeMo* in the form of a MUDR *guideline dynamic library* to access and evaluate genetic information in the record. This module processes genetic data of all stored patients and retrieves HTML formatted statistical results, which can be transferred via HTTP encoding in XML to the MUDR Client application. The MUDR GeMo module also uses various caching methods to accelerate the computation and data access. Computing the gene and allele frequency we verify the *Hardy-Weinberg's equilibrium* using the χ^2 Test.

Future Development

Simultaneously, we add a new functionality to the MUDR GeMo module. We intend to automatize comparing patients records with the information received from the *GenBank* and searches for dependences between patient's attributes and different ethnic groups.

Acknowledgment

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References

- [1] Hanzlicek P.: Development of Universal Electronic Health Record in Cardiology. Proceedings of MIE 2002. Amsterdam IOS Press, 2002. ISBN 1-58603-279-8, ISSN 0926-9630, pp. 356-360.
- [2] Tomeckova M., et al: Minimální datový model kardiologického pacienta - vyber dat. Cor et Vasa, Vol. 44, 2002, No. 4 Suppl., ISSN 0010-8650, pp. 123
- [3] Spidlen J.: MUDR XML Scheme: <http://www.euromise.cz/MUDRAPI.xsd>

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