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Bioinformatics - Information technology applications

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PEDIATRIC REFERENCE INTERVAL APPROACH: LIVER FUNCTION TESTS

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INTRODUCTION
Appropriate reference intervals are essential in laboratory medicine, and the availability of gender- and age-specific reference values is also recommended. The CALIPER study aimed at establishing reference intervals for pediatric population (<19 years) by analyzing blood samples in healthy children.
Using the Laboratory Information System database, we aimed to retrospectively assess whether the intervals and reference values suggested for liver function tests (albumin (ALB), total bilirubin (BIL) and prothrombin time (PT)) are applicable in our region.

METHODS
Serum samples from pediatric patients (1-18 years) analyzed on the Architect-c16000 (Abbott) and ACLTop (Werfen) platforms between 2012-2016 were included. Samples with abnormal results for liver damage tests (ALT, GGT), total protein, and hemoglobin were excluded as well as samples from intensive care units. Outliers were removed based on Grubb's test. From this "presumably healthy population", 10th and 90th percentiles (lower and upper limits, respectively) were calculated, along with age/gender partitions, as described on CLSI C28-A3c. Results were compared with CALIPER (Clin Chem 2012) and statistical significance was set at 0.05.

RESULTS
A total of 9,749 non-hemolyzed patient samples were included. Gender differences were negligible. The reference values were: ALB(g/L): 40.1-46.0 (1-7 years), 40.2-46.3 (8-14 years), 39.9-47.4 (15-18 years); BIL(mg/dL): 0.22-0.69 (1-8 years), 0.28-0.84 (9-11 years), 0.30-1.02 (12-14 years), 0.32-1.13 (15-18 years); and PT(%): 73-103 (1-18 years). Higher values were obtained for BIL and narrower intervals for ALB.

CONCLUSION
Based on a retrospective reference interval approach, the liver function test values matched those previously described by CALIPER, using the same technology and manufacturer, although total bilirubin concentrations seem to be higher in our population.

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A NATIONAL SURVEY IN SWEDEN SHOWS THE CLINICAL APPLICATION OF THE NPU-TERMINOLOGY FOR LABORATORY MEDICINE

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The NPU-terminology (Nomenclature for Properties and Units) is an international clinical laboratory terminology which encompasses unique identifiers and definitions of examined properties, to enable safe transmission of patient laboratory data. The Swedish version of the terminology is curated at the national release center, and further developed together with the clinical laboratories in Sweden. It is not mandatory for the laboratories to use the terminology; however a signed agreement with each county in 2015 strongly encourages the commitment to use it in a pertinent manner throughout the health care system. With the major aim to enhance the national applicability of the NPU-terminology, the current study assessed the use of the terminology among the Swedish laboratories, focusing on the detailed clinical utilization and how the current support is perceived. An electronic survey was conducted among the clinical laboratories representing all counties in Sweden. The survey included 13 short questions with both multiple-choice and text comments to specifically address e.g. the primary usage domains, number of NPU-codes in the laboratory information systems, update frequency, local code usage and support to apply the codes. The questionnaire was completed by 31 persons (40%) representing all the different counties in Sweden. A broad usage of the terminology was confirmed (94%). The three major domains of application were the local laboratory information management systems (LIMS) (59%), electronic answers (55%) and description of methods (55%). The number of codes in the respective LIMS ranged from 44 to 900 NPU-codes, and the update frequency varied among the different laboratories. Support and education was considered to be sufficient for half of the survey participants, while the other half perceived a need for extended assistance. To this end, these results provide a unique starting point to improve the application, management and support of the NPU-terminology together with the clinical laboratories in Sweden.

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PHORESIS 2, THE NEW ALL-IN-ONE SEBIA SOFTWARE FOR THE MANAGEMENT OF SAMPLES, PATIENTS AND ANALYSIS PRODUCED BY SEBIA INSTRUMENTS.

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Background: Nowadays, due to gathering and growing demand, clinical laboratories increase their productivity, by using high-throughput and high-volume instruments (e.g. CE instrument CAPILLARYS 3 TERA, SEBIA). This leads to the production of large amount of results that need to be verified and commented in a short time without neglecting patients' traceability and history, quality and regulatory recommendations. Here we present PHORESIS 2, the new all-in-one software from SEBIA for the management of samples, patients and analysis produced by all SEBIA instruments.

Description: PHORESIS 2 has been developed based on Microsoft SQL Server. This technology enables, by different operators, samples management and results validation at the instrument site or from remote locations as well as multi-sites and multi-instruments control. The Host functions of PHORESIS 2 support bi-directional connection to LIS and SEBIA instruments, allowing reception of prescriptions and centralization of analysis demands for several SEBIA instruments. Data collected are organized centered on the patients, without storage limitation. PHORESIS 2 has different dashboards with user-friendly icon-driven functions. The Supervision dashboard is dedicated to the samples management. Pending samples, prescriptions and analysis to do are displayed, improving workload of the laboratories. Results review is done with the Validation dashboard that displays at a glance profiles (mosaic and full screen view), advanced functions for interpretations and automatic recall of the patient's history. Check the profiles is fast and easy thanks to a pre-filtered display of the data (working list, samples pending, pathological samples, results per technique, etc) with easy access to patient's history. Modifications on profiles are recorded, fulfilling quality and regulatory needs. The Search dashboard contains extended search and sort functions can be used to highlight any entries required. The Quality dashboard allows visualization of the controls and calibrators results. Levey-Jennings curves module ensures easy quality survey of the SEBIA instruments.

Conclusions: PHORESIS 2 is the new SEBIA software, enabling easy and fast result review, recall, and interpretation of electrophoresis results thanks to its user-friendly conception. Thinking to be centering on the patient, it displays features that help the operators to manage easily workload and review of the electrophoretic profiles.

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"AL-QUDS BLOOD BANK" BLOOD DONORS RECRUITMENT AND EDUCATION MOBILE APPLICATION

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The primary goal of blood banks is to provide blood products safely. Blood collection centers recruit, attract and encourage voluntary blood donors. Many obstacles are faced by blood banks and blood donors during the donation registration and acceptance process. Al-Quds Blood Bank (AQBB) Mobile application was developed to overcome these obstacles.

AQBB educate the blood donors by answering the FAQs about donation. Moreover, blood donors' eligibility assessed using the international standard full questionnaire available on the application. Then, blood donors can find the nearest blood donation center or blood donation campaigns. Furthermore, they can follow their tests results and referred to specialist in the viral infection cases.

On the other hand, AQBB will take advantage of the health information system which was recently applied in the governmental and private health care facilities. Our future plan is to activate AQBB Location-based Mobile Alert in case of emergencies or blood product shortage, also to test and evaluate AQBB availability and reliability.

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DEVELOPMENT OF A MOBILE HEALTH APPLICATION FOR THE SELF-MANAGEMENT AND THE EMPOWERMENT OF PATIENTS WITH DIABETES CONDITION

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Background
Diabetes Mellitus (DM) is a global public health crisis that threatens the economies of all nations and most importantly the quality of life of an exponentially increasing number of patients. The monitoring of glycaemia is critical for the self-management of DM. We aimed to develop a mobile health application that can facilitate both self-management and the empowerment of patients with DM condition.
Methods
We explored the potential benefits that lie in combining agile methods and User Experience (UX) engineering. Agile is a widely adopted approach to software development in which requirements and solutions evolve through collaboration between self-organizing, cross-functional teams. UX engineering is a field that is concerned with Human-Computer Interaction and specifically with producing user friendly interactive systems. A user-friendly interface is one that allows users to effectively and efficiently accomplish the tasks for which it was designed, and one that users rate positively on opinion or emotional scales.
Results
The resulting system (https://beta.egle.be/) includes a customizable dashboard and widgets for the monitoring of glycaemia, insulin intake, weight, meals, social and physical activities, and moving habits. In early design stages, iterative frequent lab-tests with a small number of participants (four patients diagnosed with Type 1 DM) were conducted in order to detect and fix usability issues. Participants were required to perform a set of tasks and to answer questions about their UX with the system. The participants were likely to be thinking aloud as they performed the tasks, while the experimenter recorded their behavior and answers. In later design stages, remote user testing with a larger number of participants (ten patients diagnosed with Type 1 DM) was conducted in order to collect user data about their behavior with the system. We observed that the reduction in use of the system led to more hypo- and hyper-glycaemia.
Conclusion
The reflection on these preliminary results allows us to propose the system as beneficial to self-management and the empowerment of patients with DM condition, and worthy of further elaboration. In particular, a widget for the monitoring of Hb1Ac levels is currently being integrated in the system.

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IDENTIFICATION OF BIOCHEMICAL SUB-PROFILES WITH PATTERN RECOGNITION TECHNIQUES: CAN IT BE USED FOR ESTABLISHING REFERENCE INTERVALS FROM BLOOD BANK REGISTRY?

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The indirect study of data already collected and stored in the laboratory information system (LIS) seems as an appealing approach to overcome the difficulties of a direct estimation of reference interval (RI). The present study explores the suitability of blood donors test values for indirect RI calculation. Pattern recognition techniques were further used in order to improve the accuracy of indirect RI calculation.
A sample of 157 individuals (77M/80F) from a healthy reference population was used for direct calculation of RI for 17 common biochemical analytes according to the CLSI C28-A3 guidelines. A total of 3580 records from blood donors were used for indirect RI calculation. The reference change value (RCV) was selected for determining the statistical significance of observed differences between the indirect and direct RI. Cluster analysis with hierarchical algorithm and Ward's criterion was applied in order to identify biochemical sub-profiles amongst 1830 blood donors.
There was no significant difference between indirect and direct RI for 15 out of 17 analytes. Indirect RI for uric acid (2.5th percentile for men) and γ-GT (97.5th percentile for both men and women) were significantly different from the corresponding direct RI. Cluster analysis revealed two sub-populations within the blood donors (1 and 2). Indirect RI derived from sub-population 1 (n=1396) did not differ significantly from the direct RI for all analytes. Indirect RI derived from sub-population 2 (n=434) had significant differences for the 2.5th percentile (uric acid and cholesterol) and for both the 2.5th and 97.5th percentile for AST, ALT and γ-GT compared to the direct RI.
In conclusion, blood bank registry can be used for establishing indirect RI values for most analytes, especially when pattern recognition techniques are applied. Hierarchical cluster analysis with Ward's criterion revealed a sub-population within a large sample of blood donors that had significantly different RI in common biochemical analytes compared to direct RI. Despite the strict criteria applied to blood donor selection, some of them may actually have subclinical pathology. Pattern recognition techniques may help in identifying biochemical sub-profiles and improve the indirect study of RI.

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SERUM PROTEIN ELECTROPHORESIS AND PRESCRICTIVE APPROPRIATENESS

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INTRODUCTION – The diagnostic objective of the request of serum protein electrophoresis (EF) is the detection of monoclonal components in serum and their monitoring. However, is common EF prescription for a medical general checkup in patients without particular clinical indications. Except for patients in active therapy for plasma cell dyscrasias, the scientific evidence highlights the futility of recurrent EF before a 60 day period, but sometimes the requests of this exam are more frequent. We adopted strategies to limit the request of EF before 60 days after the last run. The purpose of this study was to evaluate the impact of this approach.

METHODS – The Corelab NOCSAE Modena, has adopted informatic systems allowing addressing of adequate EF requests. A computer alert appears every time that the EF request from a hospital department is made if the exam was already carried out during the previous 60 days. The EF is scheduled but the analysis is not performed, and the report automatically shows the results of the last EF executed with a note: “Not performed analysis: the result is related to the dosage of the day dd/mm/yy. Scientific evidence considers it inappropriate for the EF application before 60 days since the last time". Oncology, haematology and nephrology departments are excluded from this rule.
For outpatients whose the medical history is not always known, EF is performed, but accompanied by a note: "Previous analysis performed on dd/mm/yy. Scientific evidence considers it inappropriate for the EF application before 60 days since the last time".

RESULTS – Our Laboratory in 2015 received 154,719 EF requests both from the hospital departments (33,697 = 21.8%) and blood collection centers (121,022 = 78.2%). The requests subject to the rule were 8,468 (5.4%) in total, specifically 4,939 (14.7%) coming from the inpatients and then not carried out, while 3,529 (2.9%) coming from the outpatients.

CONCLUSIONS – The information technology rule introduced allowed to limit the number of EF performed and to increase the appropriateness of the requests. It represents an important means of professional support and improvement of prescribing practices.

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QUANTITATIVE MPS ANALYSIS OF BRCA1/2 GENES. NOVEL CNEV ALGORITHM FOR FAST SCREENING OF LARGE GENOMIC REARRANGEMENTS

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Background
Massively Parallel Sequencing (MPS) pipelines are widely used in molecular diagnostics providing both qualitative and quantitative information from target sequences. Several type of genomic variations could be detected by means of MPS analysis including single nucleotide variants (SNVs), small insertions or deletions (indels), copy number variations (CNVs) and large genomic rearrangements (LGRs).
In this study a novel lightweight software, namely Copy Number eValuation (CNeV), was developed to predict both CNV and LGRs in BRCA1/2 genes from Hereditary Breast and Ovarian Cancer (HBOC) selected population. Furthermore, in-house High Resolution Melting Analysis (HRMA) protocol was performed to confirm in silico results.
Methods
Amplicon library were generated with Multiplicom BRCA MASTR Dx CE-IVD kit and assayed on Illumina/MiSeq platform following manufactures' instructions. Coverage data were produced and analyzed with CNeV software to predict quantitative status of germ-line BRCA1/2 exons. Custom HRMA strategy was then performed to confirm positive predictions of CNV status using Albumin amplicons as a reference on a Roche LightCycler® 480 Real-Time PCR System (Roche Diagnostics). MLPA analysis was used as a confirmatory test for all samples.
Results
A training set of 62 genotyped samples was used to validate the CNeV algorithm, all the 6 positive samples were detected (100% sensitivity), while 14/56 negative samples were false positive (75% specificity). All the positive results of CNeV algorithm were also confirmed by custom HRMA.
Conclusions
Large genomic rearrangements have recently been identified in HBOC families and account for a small but still significant proportion of cases. In fact, about 90 and 20 LGRs are reported as pathogenic variants in BRCA1 and BRCA2 carriers, respectively. Thus, structural variants must be included for a valid and complete molecular diagnostic workflow. In this scenario, we estimate that in silico analyses will provide an important reduction of costs and time instead of common "wet" quantitative ones.
Finally, CNeV software coupled with amplicon-targeted custom HRMA assay was able to drastically reduce the number of MLPA test in the diagnostic routine workflow. Our preliminary results reveal that this strategy could strongly reduces the costs and time for BRCA testing by about 74%.