10:40am (PO20-0001)

LMM-22: An Enhanced Linear Mixed Model (LMM) Approach for Genome-Wide Association Studies (GWAS) for the Prediction of Diseases and Traits among Humans from Genomics Data
Siddharth Sharma (Stanford)

Increasingly, genomics is being used for the prediction of specific traits and diseases (phenotypes) among humans. Wider availability of genomics data through multiple research projects (such as International HapMap Project1 and 1000 Genomes2) has been a catalyst in that direction. With the recent advances in machine learning and big data analysis, data computation resources and data models needed for genomics data analysis are readily available. However, the prediction of traits and diseases has its own challenges in terms of computational requirements and computational analysis, statistical analysis (example: confounding variables), and limited quality of data collection. Linear Mixed Models (LMM, a type of linear regression) is a common approach for Genome-wide Association Studies (GWAS) for the prediction of common traits among humans using genomics. This paper researches the existing LMM-based approaches for Genome-wide Association Studies (GWAS), describes the experiment performed on FaST-LMM approach from Microsoft Research, and then proposes an enhanced approach (called LMM-22) on how to address computational and statistical issues. LMM-22 focuses on the parallelization of LMM computations and execution of LMM-22 on General Purpose Graphics Processing Units (GPU) as against CPUs to accelerate the LMM approach for GWAS studies.

10:50am (PO20-0037)

Development and validation of a melanoma genomic index (MGI) focused on CNVs and AOH from whole-genome SNP aiding in histological assessments of complex melanocytic lesions.
Varun Agarwal

Majority of melanomas can be accurately diagnosed on an adequate biopsy. However, specific subsets of melanocytic proliferations contain conflicting and ambiguous features that preclude a consensus diagnosis histologically. Artificial intelligence (AI) based methodologies are diversifying into genomics, especially in biomarker analysis and clinical diagnostics. Given major variability in diagnostic approaches for melanocytic lesion assessment, generating an algorithm for better stratification of genomic aberrations in melanoma identified by whole-genome SNP microarray - previously validated at 100% sensitivity and specificity - is key. A diagnostic melanoma genomic index (MGI) focused on CNVs and AOH of 6 markers (RREB1, CDKN2A, MYC, MYB, CCND1, and BRAFv600e) and holistic genome variance was developed and validated. One hundred cases of melanoma (n = 36) and histologic mimics of melanoma (n = 64) were investigated utilizing OncoScan data to create a machine-learning (ML) MGI algorithm that expedites data analysis and stratification of malignant and benign melanocytic lesions. The algorithm processes OSCHP genome probe data in HDF5 binary and uses boosted random forests and Bayesian networks to evaluate CNVs, Smooth Signal, BAF, and Log2Ratios to produce legitimate variant calls (discarding, merging, or verifying mutations). Variant regions intersect an hg19 reference genome to output a summative prognostic score based on numerically weighted classification of intergenic and intragenic variants per chromosome and cumulatively over the genome while providing an evaluation of BRAF mutation status and FISH marker variance. The development of an ML-driven data-anonymizing, synthetic data generator to model sample data trends has been used to expand algorithm training. Genomic data from 100 test cases (with variant call mosaicism), prognostic marker states, and clinical scores corresponding with manual review were generated (100% melanoma classification accuracy, 98% scoring accuracy). An average normal genome holistic variance score of 15% versus a melanoma positive MGI of 85% was established against the genomic markers, indicating high confidence in melanoma diagnosis. MGI serves as a valuable tool to stratify melanocytic lesions based on genomic aberrations and assists in histological assessments of these complex melanocytic lesions.
Simple Retooling of EAGLE CAD for Designing Complex Macro to Micro Fluidic Circuits
Pallavi Balivada (Boston University)

Designing routing for microfluidic chips requires tools that can create complex, modular, and multi-layered devices. Tools that are currently used to design such systems are limited in terms of usability, scaling, high level design abstraction, and integration with other platforms. The development of Electronic Design Automation (EDA) tools solved similar challenges in electronic systems, suggesting that an EDA tool such as EAGLE could prove to be an accessible, easy-to-use and customizable framework for designing fluidic systems. Furthermore, using a standard design tool could formalize fluidic design, which could in turn facilitate important collaborative efforts such as common design repositories and libraries. Here, we present a guide to adapting EAGLE to design fluidic routing using custom EAGLE libraries, scripts and design rules. Our framework streamlines the design process for various manufacturing methods and applications, from droplet microfluidics to 3D printed millifluidics. Additionally, EAGLE's separate schematic and board tools allow abstraction to the extent that designers without specialized knowledge of fluidics could still use the tool to create high-level descriptions of novel and feasible devices. We demonstrate one application of this framework to designing pneumatic digital logic architectures, highlighting the benefit of a modular approach. We show that the system can be used to design common logic elements such as a ring oscillator, a 4-bit adder and a shift register, and also present data validating a pneumatic inverter built at the millifluidic scale.

Demonstration of Doppler Ultrasound Based Innovative Diagnostic Tool for Cancer
Saksham Saksena (Houston High School / University of Memphis)

Cancer is one of the top killers in the United States. Though various health agencies have come up with elaborate screening strategies, they are expensive and cumbersome. Hence, they are not widely accepted. Recently, circulating tumor cell clusters (CTCC) have been detected during early stages of Cancer. CTCC can be measured with a variety of immunohistochemical and flow cytometry tools [1]. However, these restrict distinction of certain types of cancer, and yield from small aliquots of blood sample is low. CTCC (90” 1000 µm) detected during early stages of Cancer can be measured with a variety of immunohistochemical and flow cytometry tools [1].

For this study, a closed circulation system with an Ultrasound Phantom (UP) with wall-less vessels was created using the following steps. A vessel tube was placed in a cartridge. 5.5% agar solution heated to 90°C and cooled to 55°C, mimicking soft tissue, was poured into the cartridge, and refrigerated at 4°C for 12 hours to solidify. The vessel tube was carefully removed and intravenous (IV) tube connectors were fastened to the two ends of the wall-less cavity in the solidified agar. One end of the IV tubing was connected to the inflow connector and the output port of the 250ml 0.9% sodium chloride, normal saline (NS), IV bag to create an incoming flow into the UP. This section of IV tubing was threaded through Braun Vista infusion pump. The outflow connector was connected to the input port of the IV bag to create a closed circuit. To such IV bags, 25ml of respective sample solutions, as described below, were injected. This closed circulatory circuit (CCC) represented the systemic circulation of the body with the Infusion Pump, mimicking the heart, was set at infusion rate of 800ml/hr. Each sample added to the IV bag was passed through our CCC, changing the tubing between each sample, and flushing the UP with 25ml of NS. Doppler ultrasound was performed over UP for 15-minute duration using a 2-MHZ probe of GE LOGIQ e machine. Sample Descriptions: Instant yeast (Baker's Corner distributed by Aldi Inc. Batavia, IL) was used. 0.7g was added to 100ml H2O at 37°C. Four yeast samples were prepared. In the first sample, yeast was only mixed with water to represent normal blood cells (5 µm). The second yeast sample was incubated with sugar (9g per 100ml) and starch (9g per 100ml) for 1 minute to represent CTCC’s (20 µm). After 1 minute, for every 24ml sample solution, 1ml 4.8 mmol sodium fluoride (NaF) solution was added to block enolase, hence arresting further yeast division by blocking glucose metabolism. The third sample was incubated with starch and sugar (same as sample 2) for 15 minutes to represent CTCC (60-100 µm). After 15 minutes, NaF was added in similar proportions as above. A fourth yeast sample was made by mixing the other 3 samples in 8:1:1 proportion (mimicking circulating blood of early cancer). Fifth sample had NS only. Results / Conclusions

Results / Conclusions
Doppler signals from the third sample of yeast, representing CTCC surrogate (CTCCs), varied significantly from that of the first, second, and fifth samples in terms of intensity. This allowed us to distinguish the CTCCs from other particles in the fourth sample. Sample 2 could not be distinguished from sample 1. Adapting the backscatter ratio for this situation, we get V_c/V_n =V_c/V_n , where I_c is the intensity of the signal when a CTCCs is passing (in sample 4), I_n is the intensity of the “normal” signal (sample 1), V_c is the volume of the CTCCs, and V_n is the volume of the tubing scanned. Using this ratio, a more precise size of the yeast was determined, allowing for better detection of CTCCs. This provides a proof of principle demonstration, through the CCC system, suggesting that CTCC can be detected through real-time doppler ultrasound over carotid artery. Using such an approach for diagnosis, ultrasound detection of cancer could be made faster, more effective, and more universal.
Seizure Detection in Epilepsy Patients using Machine Learning Algorithm
Aribah Baig, Alessandro Barbiellini Amidei (Foxborough High School, Saint Sebastian's School)

There are over 50 million people who currently suffer from epilepsy. Seizures, the primary feature of this disease, are defined broadly as abnormal electrical activity in the brain during a certain period of time. Electroencephalography (EEG) is a test that measures the electrical activity in the brain and is used in the field of seizure onset detection. The primary focus of this project is to develop a patient-specific machine learning model that can efficiently classify whether or not an event is a seizure using EEG data. At the end of the project, a model was created which had a seizure accuracy rate of 69%, a non-seizure accuracy rate of 98%, and a latency period of 0.48 seconds when tested on an hour of EEG data.

Multiband Theory of Superconductivity
Joseph Cain (New York University)

The most current Bardeen-Cooper-Schrieffer (BCS) Theory of Superconductivity uses a vector formulation to accurately model the onset of superconductivity of materials that are of a single band. However, a multiband theory has been proven to model the behavior of many more materials, eliminating the single band requirement. We bridge these two ideas and propose a multidimensional, vector-formulated, multiband BCS equation. When the kernel is a positive function, we are able to establish the existence of a critical temperature, and therefore, the onset of superconductivity.

Silk-Derived Highly Dispersed Ni Supported by N-Doped Carbon Nanosheet for Electrochemical CO2 Reduction
Shicheng Hu (Massachusetts Institute of Technology)

Highly-dispersed metal catalysts supported by carbon materials have shown distinctive catalytic properties from their bulk counterparts, and are thus considered as efficient catalysts for electrochemical CO2 reduction reaction (CO2RR). Silk, on the other hand, has been demonstrating its applications in clothing, cosmetics and biocompatible materials since ancient times due to its unique chemical structure and properties. In this work, we investigated the usage of silk in CO2RR as a catalyst. We successfully synthesized highly dispersed Ni supported by N-doped carbon nanosheet (Ni/N-C) from silk and Ni(NO3)2. The silk's rich coordination sites and ability to maintain morphology upon pyrolysis were found to be essential. The ability to maintain morphology upon pyrolysis was related to the direct formation of aromatic conjugated system. The catalyst demonstrated high activity and selectivity for CO2RR, with the Faradaic Efficiency (FE) for toward CO production approaches 100% at 0.95V vs. reversible hydrogen electrode (RHE) with excellent stability.
E-cigarette use among teens has gotten so bad that the Surgeon General has called it an "epidemic". It is estimated that over 40% of the high school graduating class of 2019 has vaped. One of the most popular locations for high school vaping is the bathroom, as it allows students safety from surveillance. Privacy laws prohibit the use of cameras in bathrooms, making it difficult for school administrators to effectively crack down on high school vaping. A sensor that detects e-cigarette vapor would be legal for use in bathrooms and would assist school administrators in combating the teen vaping outbreak. In this study, an Indium Tin Oxide (ITO) nanofiber gas sensor was fabricated to detect e-cigarette vapor. ITO nanofibers were fabricated by electrospinning a solution of Indium (III) Isopropoxide, tin (IV) Isopropoxide, and Polyvinylpyrrolidone onto a substrate of silicon. Silver electrodes were applied with a 2mm gap, and wires were fastened to each using conductive copper tape. The sensor was then exposed to vapor from a JUUL e-cigarette in a controlled testing chamber. Nanofiber resistance was monitored. It was discovered that ITO has a good, albeit delayed reaction to e-cigarette vapor, demonstrating a 28% jump in resistance after 3-4 minutes of exposure.