9-2020

Machine learning can identify abnormal chromosomes to facilitate initial screening in the cytogenomics laboratory

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Consolidating Molecular Pathology Data: A Low-Cost Composite Report Prototype

(Poster No. 136)

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Context: A bottleneck in ancillary/molecular test reporting was independently identified by 2 separate quality improvement projects (Lean Six Sigma in histology and Commission on Cancer Quality Study). The statuses and results of send-out molecular tests were not being uniformly entered into the electronic medical record with pathologists and clinicians not always being aware of all testing that had been ordered or completed.

Design: Our pathology library information system (Orchard Pathology, Carmel, Indiana) was used to create a novel report type, Ancillary Testing Aggregate Report Information (ATARI), which can be accessioned for each cancer diagnosis either at the time of appointment registration or at the time additional tests are ordered. Outside results are aggregated at the time of presentation and entered through customized library information system text menus to include the specimen identification number the test was performed on, where the test was performed, and the test results. Internally generated test requests also include the date the tissue was sent for testing, which allows for real-time status updates on expected turnaround time. The ATARI is electronically delivered to the pathology section of the hospital electronic medical record, and the report is amended as new data are generated, ensuring only one updated version is available.

Results: We demonstrated the feasibility of a low-cost, real-time data aggregation solution that is accessible in pathology, in the cancer center, and at our academic tertiary referral facility.

Conclusions: Our composite report is easily viewed by clinicians and pathologists. Future improvements include streamlining the data entry process and opportunities for interfaced discrete data from Orchard.

Machine Learning Can Identify Abnormal Chromosomes to Facilitate Initial Screening in the Cytogenomics Laboratory

(Poster No. 137)

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Context: Artificial intelligence/machine learning (AI-ML) has made great strides in recent years with applications in diagnostic medicine. We tested the hypothesis that AI-ML can permit initial screening for structural chromosome abnormalities at various band levels of resolution. Such technology could markedly reduce the downstream laboratory workload and increase turnaround time in cases positive for a clinically significant chromosome abnormality.

Design: We utilized partial karyotypes of normal chromosomes 4, 5, 7, 9, 13, 21, and 22 and abnormal derivative chromosomes 9 and 22 from the t(9;22)(q34;q11.2) associated with chronic myeloid leukemia to train our AI-ML system to identify normal from abnormal chromosomes. Two hundred chromosome images with equal class representation were used to train a 2-dimensional convolutional neural network, which is a deep learning algorithm commonly used in image classification. The parameters of the model were tuned using 5-fold cross-validation. Model performance was assessed by testing against a separate collection of 67 images.

Results: Normal and abnormal chromosomes could be differentiated by the computer with high accuracy. Our system correctly classified 96.5% of the 200 training images, whereas 63 of 67 test images were correctly classified, yielding overall test accuracy of 94.0% (Table).

Conclusions: This study demonstrates that AI-ML can potentially be of great value in the cytogenomics laboratory. Although not expected to replace the technologist, it can be used to provide an initial screen of each case, so that those cases with chromosome abnormalities can be quickly identified and evaluated, thus reducing turnaround time and facilitating earlier medical management.

Summary of Chromosome Classification Results by AI-ML

Chromosome	Correctly Identified, No. (%)	Misclassified, No. (%)	Total
Normal 4	5 (83.3)	1 (16.7)	6
Normal 5	5 (83.3)	1 (16.7)	6
Normal 7	6 (100.0)	0 (0.0)	6
Normal 13	6 (100.0)	0 (0.0)	6
Normal 21	7 (87.5)	1 (12.5)	8
Normal 22	11 (100.0)	0 (0.0)	11
Abnormal 9	12 (100.0)	0 (0.0)	12
Abnormal 22	11 (91.7)	1 (8.3)	12
Total	**63** (94.0)	**4** (6.0)	**67**

Performance Assessment of Pathologists Using Various Whole Slide Imaging Systems

(Poster No. 138)

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Context: Whole slide imaging (WSI) closely simulates conventional optical microscopy (OM) for routine diagnosis.

Design: We performed a comprehensive comparative evaluation using 240 cases, comprising 60 cases in each specimen category (biopsy, resection, frozen section, and cytology) as per College of American Pathologists recommendations. This study aimed at assessing the performance of pathologists on various WSI platforms and evaluating the diagnostic accuracy, andinterobserver and intraobserver concordances among the pathologists. The level of confidence and interpretation time for each case were recorded to assess the ease of using WSI for primary diagnosis. Each case was assessed by 7 pathologists (2 experts and 5 nonexperts) using 4 WSI systems. A training set of 10 cases was used for achieving self-familiarization, and each digital platform was evaluated after a wash-off period of 2 weeks.

Results: Overall diagnostic accuracy, when compared with reference standard for OM and WSI, was 95.44% and 93.32%, respectively. The major discordance rates were 2.08% (OM) and 2.4% (WSI) and the minor discordance rates were 2.48% (OM) and 4.28% (WSI). Both interobserver and intraobserver agreement between WSI and OM for primary diagnosis of biopsy, resection, frozen section and cytology specimens were substantial to perfect (Table), although diagnostic accuracy for cytology between WSI and OM (43.6%–87% versus 93.4%) with reference standard was >4%. Overall level of confidence for cytology evaluation was average irrespective of the scanner type. Diagnostic assessment time required for OM was less as opposed to WSI for all pathologists.

Conclusions: WSI can be safely adopted for biopsy, resection, and frozen section specimens. Further training and technical advancements are required for evaluation of cytology specimen by WSI.

Interobserver and Intraobserver κ Mean Values for Biopsy, Resection, Frozen, and Cytology

Specimen Type	Interobserver κ Mean	Intraobserver κ Mean
Biopsy	0.77 (0.76–0.80)	0.82 (0.97–0.72)
Resection	0.85 (0.84–0.87)	0.9 (0.98–0.82)
Frozen	0.91 (0.9–0.94)	0.94 (0.9–0.99)
Cytology	0.72 (0.7–0.73)	0.71 (0.55–0.82)

Development of Real-Time Blood Product Inventory Management Dashboards in a Multiple Hospital System

(Poster No. 139)

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