GeNeXpreSS

Applications of Artificial Intelligence (AI) in Medical Genetics: From Patient Counseling to Training of Students.

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Automated prioritization of sick newborns for whole genome sequencing using clinical natural language processing and machine learning

(Peterson et al., 2023)

Human Phenotype Ontology (HPO)-based phenotypic descriptions for whole genome sequencing (WGS) in the neonatal intensive care unit (NICU) is an established practice. It is done manually and at one or two isolated points of time. Constant vigilance for changes in laboratory results, diagnostic imaging, and clinical trajectories is crucial to identify infants most likely to benefit from WGS. It is labour-intensive and requires experts, making it a bottleneck. Automated clinical natural language processing (CNLP) technologies can help generate structured descriptions from unstructured clinical notes, aiding in triaging neonates for WGS.

The Mendelian Phenotype Search Engine (MPSE) combines the CNLP workflow with a machine learning-based prioritization tool, facilitating patient review. Diagnostic rates for CNLP datasets were consistently at or above the cohort diagnostic fraction of 43% at every MPSE score percentile. In contrast, MPSE scores from manually curated phenotypes showed weaker diagnostic performance. This difference can be from the average number of phenotype terms derived from each method. The CNLP method produced an average of 114.8 and 64.5 phenotype terms, compared to 4.1 and 9.5 from manual curation in two independent cohorts.

The CNLP/MPSE workflow prioritized patients for rapid WGS (rWGS) with high accuracy [area under the curve (AUC)=0.86], enriching diagnostic yields in the top-scoring quartile. MPSE automatically surveys all NICU admissions and updates their scores daily based on health record content. These MPSE score cutoffs can be used to prioritize patients for further physician review.

A randomized trial comparing the effectiveness of pretest genetic counseling using an artificial intelligence automated chatbot and traditional in-person genetic counseling in women newly diagnosed with breast cancer

(Al-Hilli et al, 2023)

Alternative service delivery models are critically needed to address the increasing demand for genetics services and the limited supply of genetics experts available to provide pre-test counseling. Al-Hilli et al. conducted a prospective randomized controlled trial of women with stage 0 to III breast cancer not meeting the National Comprehensive Cancer Network (NCCN) criteria for genetic testing. Patients were randomized to pretest counseling with a chatbot or a certified genetic counselor (GC). Nineteen were randomized to the chatbot and 18 to traditional genetic counseling. Out of the total number of participants, 38.2% had a family member with breast cancer but did not meet the NCCN criteria. Participants completed a questionnaire assessing their knowledge of breast cancer genetics and a survey assessing satisfaction with their decision regarding pretest counseling. After the pretest counseling, all patients opted to undergo genetic testing. There were no significant differences in the median knowledge score between the chatbot and traditional counseling (11 vs. 12, $p = 0.09$) or median patient satisfaction score (30 vs. 30, $p = 0.19$). No patients had a delay in time-to-treatment due to genetic testing turnaround time, nor did any patients undergo

additional risk-reducing surgery. The scores of satisfaction and comprehension in these patients using either an automated hatbot or an in-person Genetic counselor did not significantly differ. The study suggests that utilizing a chatbot for pretest counseling is as effective as traditional counseling by a certified genetic counselor regarding patient knowledge, satisfaction, and comprehension. This alternative approach can alleviate the strain on genetic counseling services by providing a viable, efficient option for pretest counseling, especially when genetic experts are in limited supply.

Evaluation of the Rosa Chatbot providing genetic information to patients at risk of hereditary breast and ovarian cancer: Qualitative interview study

(Siglen et al, 2023)

Genetic testing has become integral for patients with breast or ovarian cancer, necessitating reliable access to genetic information. To meet this demand, a chatbot named Rosa was developed for human-like conversations about testing of the BRCA genes. This study aimed to evaluate the perceived utility and trust in Rosa among healthy individuals at risk of hereditary cancer and its influence on their handling of sensitive information. A total of 175 at-risk individuals were invited to test Rosa before and after genetic counseling, recruited from all cancer genetic clinics in Norway to ensure diversity. Among them, 61 (34.9%) consented to individual interviews, with a selected subgroup of 16 (26%) participating in in-depth video interviews. These semi-structured interviews explored usability, perceived usefulness, trust in the information, the chatbot's influence, and future digital tool use in healthcare. The findings indicated that participants welcomed Rosa, valuing its 24/7 availability and role in preparing for and reviewing genetic counseling sessions. The information provided by Rosa, created by healthcare professionals, was considered medically accurate, making it more reliable than general internet searches. Key themes emerged: “Anytime, anywhere”; “In addition, not instead”; and “Trustworthy and true.” Notably, none of the participants reported increased worry after using Rosa.

In conclusion, Rosa offers easy access to consistent, quality-assured genetic information, reassuring patients at risk of hereditary breast and ovarian cancer. The participants did not support its use as a replacement for genetic counseling when hereditary cancer is confirmed. Thus, Rosa serves as a complementary tool, enhancing but not replacing traditional genetic counselling.

Recognition of genetic conditions after learning with images created using generative artificial intelligence

(Waikel et al., 2024)

The study aimed to compare the ability of pediatric residents to recognize Kabuki syndrome (KS) and Noonan syndrome (NS) after exposure to one of four educational interventions, including generative artificial intelligence (AI) methods. Participants categorized 20 images following exposure to one of four educational interventions: text-only descriptions, authentic images, and two types of AI-generated images. For KS, sensitivity with text descriptions was 48.5%, not significantly different from random guessing. Sensitivity improved with natural images (60.3%) and AI-generated images (57.0% and 59.6%). For NS, text descriptions had a sensitivity of 65.3%, compared to 74.3% with authentic images and 68.0% and 71.0% with AI-generated images. In terms of specificity, none of the interventions showed a significant difference from the text-only approach. For KS, the number of participants unsure about diagnostic features decreased from 52.8% to 7.6%, and for NS, it decreased from 24.5% to 4.7%. There was a significant correlation between confidence levels and sensitivity for real and AI-generated images.

In conclusion, the study found that real and AI-generated images enhanced the recognition of KS and NS among pediatric residents, with real photos proving most effective. While slightly less effective than real images, AI-generated images were not inferior and could serve as a valuable adjunctive tool, particularly for educating about rare conditions. It highlights the potential role of AI-generated images in medical education to improve the recognition of genetic syndromes.

References

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