

Deep Learning for Healthcare: Transforming Biomedical Data into Actionable Insights

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ABSTRACT

The evolution of technology has both positive and negative impacts on health care due to the tremendous increase of biomedical data. Medical data, being complex and heterogeneous, poses difficulties for traditional data mining and statistical learning techniques, which is why deep learning has gained popularity recently. Unlike its predecessors, deep learning offers end-to-end learning, thus allowing extraction of useful information from various data sources like genomic devices, wearable gadgets, electronic health records, and even medical imaging. This paper focuses on recently integrated deep learning algorithms that enhance prediction and diagnosis accuracy of various diseases to optimize treatment processes. Even in its advancement, the widespread use of deep learning algorithms for clinical purposes is hindered by data heterogeneity and low interpretability as well as the need for large volumes of high quality data. We will address those limitations by developing deep learning algorithms with more domain knowledge, better interpretability, and federated learning techniques. Overcoming these limitations could allow deep learning to transform medical research and patient care by enabling streamlined and customized health care solutions.

Keywords: Deep Learning, Healthcare, Artificial Intelligence, Machine Learning, EHRs, Medical Imaging, Biomedical Data

INTRODUCTION

Healthcare is transitioning into a new era where the massive volume of biomedical data will have greater relevance than ever before. For instance, in precision medicine, minutiae like variability in molecular features, imaging, geographic information systems (GIS), and even a patient's lifestyle are integrated into EHRs, so medicine can be provided to 'the right patient at the most appropriate time' [1].

The large availability of biomedical data brings tremendous opportunities and challenges to health care research. In particular, exploring the associations among all the different pieces of information in these data sets is a fundamental problem to develop reliable medical tools based on data-driven approaches and machine learning. To this aim, previous works tried to link multiple data sources to build joint knowledge bases that could be used for predictive analysis and discovery [2]. Although existing models demonstrate great promises (e.g. [3]), predictive tools based on machine learning techniques have not been widely applied in medicine [4]. In fact, there remain many challenges in making full use of the biomedical data, owing to their high-dimensionality, heterogeneity, temporal dependency, sparsity and irregularity [5].

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A common approach in biomedical research is to have a domain expert to specify the phenotypes to use in an ad hoc manner. However, supervised definition of the feature space scales poorly and misses the opportunities to discover novel patterns. Alternatively, representation learning methods allow to automatically discover the representations needed for prediction from the raw data [6]. Deep learning methods are representation-learning algorithms with multiple levels of representation, obtained by composing simple but nonlinear modules that each transform the representation at one level (starting with the raw input) into a representation at a higher, slightly more abstract level [7]. Deep learning models demonstrated great performance and potential in computer vision, speech recognition and natural language processing tasks [8].

Given its demonstrated performance in different domains and the rapid progresses of methodological improvements, deep learning paradigms introduce exciting new opportunities for biomedical informatics. Efforts to apply deep learning methods to health care are already planned or underway.

However, deep learning approaches have not been extensively evaluated for a broad range of medical problems that could benefit from its capabilities. There are many aspects of deep learning that could be helpful in health care, such as its superior performance, end-to-end learning scheme with integrated feature learning, capability of handling complex and multi-modality data and so on. To accelerate these efforts, the deep learning research field as a whole must address several challenges relating to the characteristics of health care data (i.e. sparse, noisy, heterogeneous, time-dependent) as need for improved methods and tools that enable deep learning to interface with health care information workflows and clinical decision support. In this article, we discuss recent and forthcoming applications of deep learning in medicine, highlighting the key aspects to significantly impact health care. We do not aim to provide a comprehensive background on technical details (see e.g. [6], [9], [10]) or general application of deep learning (see e.g. [11]). Instead, we focus on biomedical data only, in particular those originated from clinical imaging, EHRs, genomes and wearable devices. While additional sources of information, such as metabolome, antibodyome and other omics information are expected to be valuable for health monitoring, at this point deep learning has not been significantly used in these domains. Thus, in the following, we briefly introduce the general deep learning framework, we review some of its applications in the medical domain and we discuss the opportunities, challenges and applications related to these methods when used in the context of precision medicine and next-generation health care.

LITERATURE REVIEW

The use of deep learning for medicine is recent and not thoroughly explored. In the next sections, we will review some of the main recent literature (i.e. 32 papers) related to applications of deep models to clinical imaging, EHRs, genomics and wearable device data. Table 1 summarizes all the papers mentioned in this literature review, in particular highlighting the type of networks and the medical data considered. To the best of our knowledge, there are no studies using deep learning to combine neither all these data sources, nor a part of them (e.g. only EHRs and clinical images, only EHRs and genomics) in a joint representation for medical analysis and prediction. A few preliminary studies evaluated the combined use of EHRs and genomics (e.g. see [12], [13]), without applying deep learning though; for this reason, they were not considered relevant to this review. The deep architectures applied to the health care domain have been mostly based on convolutional neural networks (CNNs) [14], recurrent neural networks (RNNs) [15], Restricted Boltzmann Machines (RBMs) [16] and Autoencoders (AEs) [17]

Author	Model	Application	Reference
Liu et al. (2014)	Stacked Sparse AE	Early diagnosis of Alzheimer disease from brain MRIs	[18]
Brosch et al. (2013)	RBM	Manifold of brain MRIs to detect modes of variations in Alzheimer	[19]
Prasoon et al. (2013)	CNN	Automatic segmentation of knee cartilage MRIs to predict the risk of osteoarthritis	[20]
Yoo et al. (2014)	RBM	Segmentation of multiple sclerosis lesions in multi-channel 3D MRIs	[21]
Cheng et al. (2016)	Stacked	Diagnosis of breast nodules and lesions from ultrasound images	[22]
Gulshan et al. (2016)	CNN	Detection of diabetic retinopathy in retinal fundus photographs	[23]
Esteva et al. (2017)	CNN	Dermatologist-level classification of skin cancer	[24]
Liu et al. (2015)	CNN	Prediction of congestive heart failure and chronic obstructive pulmonary disease from longitudinal EHRs	[25]
Lipton et al. (2015)	LSTM RNN	Diagnosis classification from clinical measurements of patients in pediatric intensive unit care	[26]
Pham et al. (2016)	LSTM RNN	Deep Care: a dynamic memory model for predictive medicine based on patient history	[27]
Miotto et al. (2016)	Stacked Denoising AE	Deep Patient: an unsupervised representation of patients that can be used to predict future clinical events	[28]
Liang et al. (2014)	RBM	Automatically assign diagnosis to patients from their clinical status	[29]
Koh et al. (2017)	CNN	Prevalence estimates for different chromatin marks	[30]
Fakoor et al. (2013)	Stacked Sparse AE	Classification of cancer from gene expression [31] profiles	
Lyons et al. (2014)	Stacked Sparse AE	Prediction of protein backbones from protein sequences	[32]
Hammerla et al. (2016)	CNN/RNN	HAR to detect freezing of gait in PD patients	[33]
Zhu et al. (2015)	CNN	Estimation of EE using wearable sensors	[34]
Jindal et al. (2016)	RBM	Identification of Photoplethysmography signals for health monitoring	[35]
Nurse et al. (2016)	CNN	Analysis of electroencephalogram and local field potentials signals	[36]

Table 1. Summary of the articles described in the literature review with highlighted the deep learning architecture applied and the medical domain considered

Challenges and opportunities

Despite the promising results obtained using deep architectures, there remain several unsolved challenges facing the clinical application of deep learning to health care. In particular, we highlight the following key issues: Data volume, Data quality, Temporality, Domain complexity and Interpretability.

All these challenges introduce several opportunities and future research possibilities to improve the field. Therefore, with all of them in mind, we point out the following directions, which we believe would be promising for the future of deep learning in health care.

- Feature enrichment
- Federated inference
- Model privacy
- Incorporating expert knowledge
- Temporal modeling
- Interpretable modeling

CONCLUSION

Deep learning has emerged as a transformative tool in healthcare, enabling the extraction of actionable insights from complex and heterogeneous biomedical data. By leveraging advanced neural network architectures, deep learning facilitates disease prediction, diagnosis, and treatment optimization across various medical domains, including electronic health records, medical imaging, genomics, and wearable device data. However, significant challenges remain, particularly in terms of data heterogeneity, model interpretability, and the need for large, high-quality datasets. Addressing these challenges through improved feature enrichment, federated learning, privacy-preserving models, and the integration of domain knowledge will be critical for advancing deep learning in clinical practice. With continued research and technological advancements, deep learning has the potential to revolutionize medical research and patient care, paving the way for more personalized, efficient, and intelligent healthcare solutions.

REFERENCES

- [1] G. H. ; M. Lyman Harold L., "Biomarker Tests for Molecularly Targeted Therapies: Key to Unlocking Precision Medicine - Biomarker Tests for Molecularly Targeted Therapies — The Key to Unlocking Precision Medicine," *N Engl J Med*, vol. 375, no. 1, pp. 4–6, 2016, doi: 10.1056/nejmp1604033.
- [2] R. L. Xu Li; Wang QuanQiu, "dRiskKB: a large-scale disease-disease risk relationship knowledge base constructed from biomedical text.," *BMC Bioinformatics*, vol. 15, no. 1, p. 105, 2014, doi: 10.1186/1471-2105-15-105.
- [3] N. P. ; Y. Tatonetti Patrick Peiyong; Daneshjou Roxana; Altman Russ B., "Data- Driven Prediction of Drug Effects and Interactions," *Sci Transl Med*, vol. 4, no. 125, pp. 125ra31-NA, 2012, doi: 10.1126/scitranslmed.3003377.
- [4] R. Z. Bellazzi Blaz, "Predictive data mining in clinical medicine: Current issues and guidelines," *Int J Med Inform*, vol. 77, no. 2, pp. 81–97, 2006, doi: 10.1016/j.ijmedinf.2006.11.006.
- [5] G. A. Hripcsak David J., "Next-generation phenotyping of electronic health records," *J Am Med Inform Assoc*, vol. 20, no. 1, pp. 117–121, 2012, doi: 10.1136/amiajnl-2012-001145.
- [6] Y. Bengio, *Neural Networks: Tricks of the Trade (2nd ed.) - Practical recommendations for gradient-based training of deep architectures*, vol. NA, no. NA. 2012. doi: 10.1007/978-3-642-35289-8_26.
- [7] Y. Lecun, Y. Bengio, and G. Hinton, "Deep learning," *Nature*, vol. 521, no. 7553, pp. 436–444, May 2015, doi: 10.1038/NATURE14539.
- [8] O. M. Abdel-Hamid Abdelrahman; Jiang Hui; Deng Li; Penn Gerald; Yu Dong, "Convolutional neural networks for speech recognition," *IEEE/ACM Trans Audio Speech Lang Process*, vol. 22, no. 10, pp. 1533–1545, 2014, doi: 10.1109/taslp.2014.2339736.
- [9] Y. Bengio, *Learning Deep Architectures for AI*, vol. NA, no. NA. 2009. doi: NA.

- [10] Y. Bengio, *Neural Networks: Tricks of the Trade* (2nd ed.) - Practical recommendations for gradient-based training of deep architectures, vol. NA, no. NA. 2012. doi: 10.1007/978-3-642-35289-8_26.
- [11] Y. B. LeCun Yoshua; Hinton Geoffrey E., "Deep learning," *Nature*, vol. 521, no. 7553, pp. 436–444, 2015, doi: 10.1038/nature14539.
- [12] L. C. Li Wei-Yi; Glicksberg Benjamin S.; Gottesman Omri; Tamler Ronald; Chen Rong; Bottinger Erwin P.; Dudley Joel T., "Identification of type 2 diabetes subgroups through topological analysis of patient similarity," *Sci Transl Med*, vol. 7, no. 311, pp. 311ra174-NA, 2015, doi: 10.1126/scitranslmed.aaa9364.
- [13] M. P. Gerstung Elli; Martincorena Inigo; Bullinger Lars; Gaidzik Verena I.; Paschka Peter; Heuser Michael; Thol Felicitas; Bolli Niccolo; Ganly Peter; Ganzer Arnold; McDermott Ultan; Döhner Konstanze; Schlenk Richard F.; Döhner Hartmut; Campbell Peter J., "Precision oncology for acute myeloid leukemia using a knowledge bank approach," *Nat Genet*, vol. 49, no. 3, pp. 332–340, 2017, doi: 10.1038/ng.3756.
- [14] Y. B. LeCun Yoshua; Hinton Geoffrey E., "Deep learning," *Nature*, vol. 521, no. 7553, pp. 436–444, 2015, doi: 10.1038/nature14539.
- [15] E. B. Choi Mohammad Taha; Schuetz Andy; Stewart Walter F.; Sun Jimeng, "Doctor AI: Predicting Clinical Events via Recurrent Neural Networks.," *JMLR Workshop Conf Proc*, vol. 56, no. NA, pp. 301–318, 2016, doi: NA.
- [16] T. N. Tran Tu Dinh; Phung Dinh; Venkatesh Svetha, "Learning vector representation of medical objects via EMR-driven nonnegative restricted Boltzmann machines (eNRBM)," *J Biomed Inform*, vol. 54, no. NA, pp. 96–105, 2015, doi: 10.1016/j.jbi.2015.01.012.
- [17] P. L. Vincent Hugo; Lajoie Isabelle; Bengio Yoshua; Manzagol Pierre-Antoine, "Stacked Denoising Autoencoders: Learning Useful Representations in a Deep Network with a Local Denoising Criterion," *Journal of Machine Learning Research*, vol. 11, no. 110, pp. 3371–3408, 2010, doi: NA.
- [18] B. Liu and J. Liu, "Overview of Image Denoising Based on Deep Learning," *J Phys Conf Ser*, vol. 1176, no. 2, Mar. 2019, doi: 10.1088/1742-6596/1176/2/022010.
- [19] T. T. Brosch Roger, "MICCAI (2) - Manifold Learning of Brain MRIs by Deep Learning," *Med Image Comput Comput Assist Interv*, vol. 16, no. Pt 2, pp. 633–640, 2013, doi: 10.1007/978-3-642-40763-5_78.
- [20] A. P. Prasoon Kersten; Igel Christian; Lauze François; Dam Erik B.; Nielsen Mads, "MICCAI (2) - Deep Feature Learning for Knee Cartilage Segmentation Using a Triplanar Convolutional Neural Network," *Med Image Comput Comput Assist Interv*, vol. 16, no. Pt 2, pp. 246–253, 2013, doi: 10.1007/978-3-642-40763-5_31.
- [21] Y. B. Yoo Tom; Traboulsee Anthony; Li David K.B.; Tam Roger, *MLMI - Deep Learning of Image Features from Unlabeled Data for Multiple Sclerosis Lesion Segmentation*, vol. NA, no. NA. 2014. doi: 10.1007/978-3-319-10581-9_15.
- [22] J.-Z. N. Cheng Dong; Chou Yi-Hong; Qin Jing; Tiu Chui Mei; Chang Yeun- Chung; Huang Chiun-Sheng; Shen Dinggang; Chen Chung-Ming, "Computer-Aided Diagnosis with Deep Learning Architecture: Applications to Breast Lesions in US Images and Pulmonary Nodules in CT Scans," *Sci Rep*, vol. 6, no. 1, p. 24454, 2016, doi: 10.1038/srep24454.
- [23] V. P. Gulshan Lily; Coram Marc; Stumpe Martin C.; Wu Derek; Narayanaswamy Arunachalam; Venugopalan Subhashini; Widner Kasumi; Madams Tom; Cuadros Jorge; Kim Ramasamy; Raman Rajiv; Nelson Philip C.; Mega Jessica L.; Webster Dale R., "Development and Validation of a Deep Learning Algorithm for Detection of Diabetic Retinopathy in Retinal Fundus Photographs," *JAMA*, vol. 316, no. 22, pp. 2402–2410, 2016, doi: 10.1001/jama.2016.17216.
- [24] A. K. Esteva Brett; Novoa Roberto A.; Ko Justin M.; Swetter Susan M.; Blau Helen M.; Thrun Sebastian, "Dermatologist-level classification of skin cancer with deep neural networks," *Nature*, vol. 542, no. 7639, pp. 115–118, 2017, doi: 10.1038/nature21056.
- [25] Y. Liu, J. Yang, Y. Huang, L. Xu, S. Li, and M. Qi, "MapReduce Based Parallel Neural Networks in Enabling Large Scale Machine Learning," *Comput Intell Neurosci*, vol. 2015, 2015, doi: 10.1155/2015/297672.
- [26] Z. C. Lipton, D. C. Kale, C. Elkan, and R. Wetzel, "Learning to Diagnose with LSTM Recurrent Neural Networks," Nov. 2015, [Online]. Available: <http://arxiv.org/abs/1511.03677>
- [27] T. Pham, T. Tran, D. Phung, and S. Venkatesh, "DeepCare: A Deep Dynamic Memory Model for Predictive Medicine," Jan. 2016, [Online]. Available: <http://arxiv.org/abs/1602.00357>

- [28] R. L. Miotto Li; Dudley Joel T., "ECIR - Deep Learning to Predict Patient Future Diseases from the Electronic Health Records," in *Lecture Notes in Computer Science*, vol. NA, no. NA, 2016, pp. 768–774. doi: 10.1007/978-3-319-30671-1_66.
- [29] Z. Liang, G. Zhang, J. X. Huang, and Q. V. Hu, "Deep learning for healthcare decision making with EMRs," in *2014 IEEE International Conference on Bioinformatics and Biomedicine (BIBM)*, IEEE, Nov. 2014, pp. 556–559. doi: 10.1109/BIBM.2014.6999219.
- [30] P. W. P. Koh Emma; Kundaje Anshul, "Denoising genome-wide histone ChIP- seq with convolutional neural networks.," *Bioinformatics*, vol. 33, no. 14, pp. i225–i233, 2017, doi: 10.1093/bioinformatics/btx243.
- [31] R. Fakoor, A. Nazi, and M. Huber, "Using deep learning to enhance cancer diagnosis and classification," 2013. [Online]. Available: <https://www.researchgate.net/publication/281857285>
- [32] J. D. Lyons Abdollah; Heffernan Rhys; Sharma Alok; Paliwal Kuldip K.; Sattar Abdul; Zhou Yaoqi; Yang Yuedong, "Predicting backbone C α angles and dihedrals from protein sequences by stacked sparse auto-encoder deep neural network.," *J Comput Chem*, vol. 35, no. 28, pp. 2040–2046, 2014, doi: 10.1002/jcc.23718.
- [33] N. Y. Hammerla, S. Halloran, and T. Ploetz, "Deep, Convolutional, and Recurrent Models for Human Activity Recognition using Wearables," Apr. 2016, [Online]. Available: <http://arxiv.org/abs/1604.08880>
- [34] J. Zhu, A. Pande, P. Mohapatra, and J. J. Han, "Using Deep Learning for Energy Expenditure Estimation with wearable sensors," in *2015 17th International Conference on E-health Networking, Application & Services (HealthCom)*, IEEE, Oct. 2015, pp. 501–506. doi: 10.1109/HealthCom.2015.7454554.
- [35] V. B. Jindal Javad; Pouyan M. Baran; Nourani Mehrdad, "EMBC - An adaptive deep learning approach for PPG-based identification," *Annu Int Conf IEEE Eng Med Biol Soc*, vol. 2016, no. NA, pp. 6401–6404, 2016, doi: 10.1109/embc.2016.7592193.
- [36] E. Nurse, B. S. Mashford, A. J. Yepes, I. Kiral-Kornek, S. Harrer, and D. R. Freestone, "Decoding EEG and LFP signals using deep learning: Heading truenorth," in *2016 ACM International Conference on Computing Frontiers - Proceedings*, Association for Computing Machinery, Inc, May 2016, pp. 259–266. doi: 10.1145/2903150.2903159.