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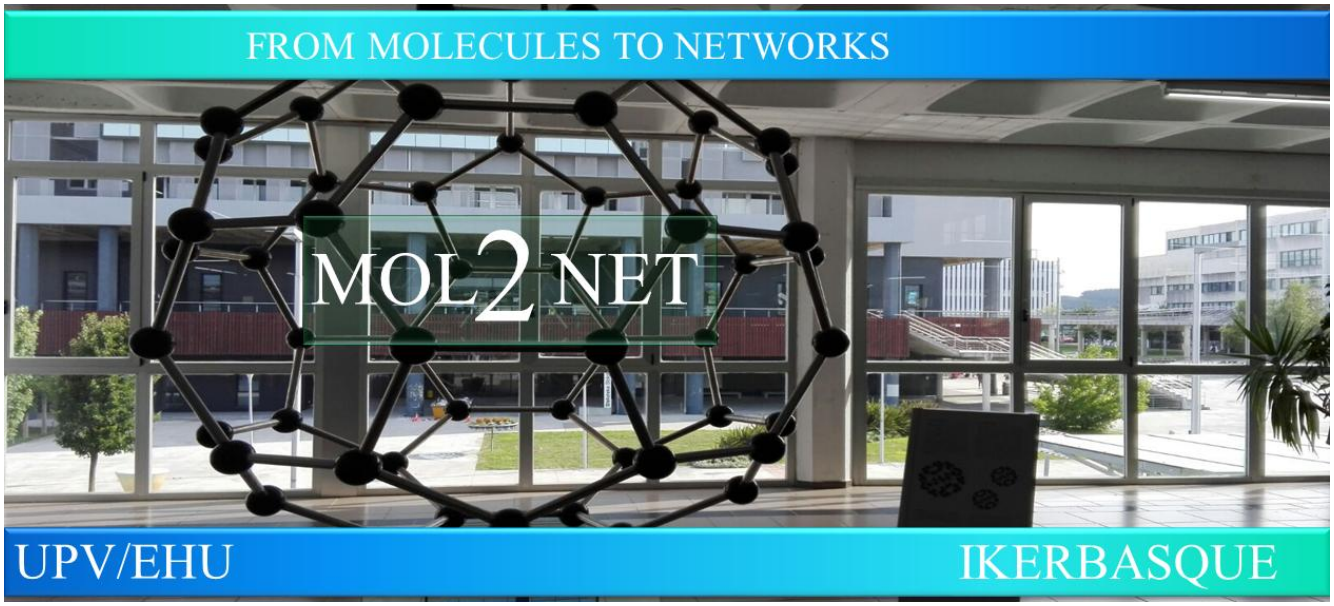

Genomics data and Artificial Intelligence

The first paper was published by Iman Hajirasouliha and Olivier Elemento in the American Society for Reproductive Medicine. The authors summed up the concepts of precision medicine and artificial intelligence, as well as the several limitations that these have in medicine when genomics data are used.

Precision medicine, known as, personalize medicine, incorporates more than genomics data, applying to epigenetics, proteomics, metabolomics, and imaging. Is important to underline, that the use of large-scale genomic sequencing lead to detect different mutations. As the amount of data is enormous, these studies require the use of Artificial Intelligence (AI) and Machine Learning (ML). ML is a form of AI in which a machine can learn and adapt to situations and data training. There are lots of ML techniques that can be used to create predictive models, as well as it can be used to predict responses to therapy based on genomic[1,2.] Some of the most frequently used techniques include logistic regression, random forests (RFs), naive Bayes classifiers, support vector machines (SVMs), Artificial Neural Networks (ANN), and Deep Neural Networks (DNN). This one has recently developed and has started applying in medicine.

Although the implementation of AI in medicine has a lot of benefits, also numerous challenges have been described[3]. For example, precision medicine and AI suffer from a relative lack of standardization. Also, apparently, the presence of biases in the data is used to learn new medical knowledge or train predictive models[4]. Finally, considering that AI software needs to integrate into existing and clinically validated workflows, implementation is also a challenge.

The second paper was published by Raquel Dias and Ali Torkamani in Genome Medicine. The authors summed up the recent successes and potential future applications of Artificial Intelligence (AI) in clinical diagnostics, highlighting the problems that can appear, and how can be resolved using AI techniques.

Examples of some of these problems are summarized:

On account of a large amount of individual genetic variants among the millions populating each genome, is needed extreme accuracy. Lots of systematic errors are generated by way of using standard tools[5]. Because of that is necessary the combination of statistics tools, which provides high precision, although it continued to appear biased errors[6]. AI algorithms have the capacity of learning this biased of one genome. This is the case of DeepVariant, a Convolutional Neural Networks (CNN) based variant caller that can do it without any specialized knowledge about genomics or sequencing platforms[7]. After variant calling, the molecular diagnostic of the disease needs to do it, which is the interpretation of the human genome data. This interpretation requires both the identification of candidate pathogenic variants and a determination of the correspondence between the diseased individual's phenotype and those expected to result from each candidate pathogenic variant. AI algorithms can significantly improve the mapping of phenotype to genotype by the extraction of higher-level diagnostic concepts that are integrated into medical images and Electronic Health Records (EHRs).

I. *Image to genetic diagnosis.* DeepGestalt, a CNN-based facial image analysis algorithm, is sufficiently precise to distinguish between molecular diagnostics that are mapped to the same clinical diagnosis[6]. When there are combined with genomic data, PEDIA, a genome interpretation system, can use phenotypic features extracted from facial photographs to accurately prioritize candidate pathogenic variants for 105 different monogenic disorders across 679 individuals[8].

II. *EHR to genetic diagnosis.* Owing to the high complexity and diversity of phenotypes and the way that this data is recapitulated (medical imaging, biochemical tests, etc) these are documented in an EHR. In this way, the diagnostic and the clinical decision-making are more precise avoiding possible biased.

Taking into account previous information published by Iman Hajirasouliha and Olivier Elemento in the American Society for Reproductive Medicine and by Raquel Dias and Ali Torkamani in Genome Medicine, the main message of both papers is that applying AI to medicine has lots of benefits, despite having some limitations. Comparing the second paper and the first paper, the second one (published by Raquel Dias and Ali Torkamani in Genome Medicine) is more focused on the capacity that has AI to solve the problems that can appear when genomics data is used. Although this paper explains the solutions to the major problems when AI is combined with genomics data, both of us share the same opinion about the need of applies AI due to the variety and complexity of this type of data. Finally, both papers have in common that more studies are needed.

Personally, I think that the two previous papers are complementary each to other. This is because the first one published by Iman Hajirasouliha and Olivier Elemento in the American Society for Reproductive Medicine explains the problems that may appear when genomics data and AI are used, whereas the second one published by Raquel Dias and Ali Torkamani in Genome Medicine suggest solutions using AI, so the authors of the first paper might take advantage of the second one when more studies related to this subject are done.

To sum up, in my opinion applying AI in medicine is a potential tool that in a near future should be implemented in almost public and private institutes. I agree that at the moment there exist some limitations, so in my view, more studies must be done.

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