GeNeDit

Importance of Artificial Intelligence in Medical Genetics

Editorial

Artificial intelligence (AI) is making all areas of medicine swift, sharp, speedy and hence, effective. AI-based tools can do many things which human intelligence cannot. However, AI is not new for medical geneticists. AI-based next-generation sequencing (NGS) data analysis and variant interpretation has made life easy for medical and clinical geneticists. In this issue, the review article about the clinical implications of splice site variants has discussed the use of many AI-based tools such as SpliceAI. The functional effects of splice site variants can be tested in laboratories but may not be possible for every variant detected in clinical situations. Deep learning has been incorporated into bioinformatic algorithms for genome analysis, protein structure classification, prediction, regulatory genomics and cellular imaging. This is especially important in this era where sequencing three billion nucleotides is easy but the challenges of interpretation of variations of unknown significance are bigger than that of sequencing. Tools for changing uncertainties to certainties and help in decision making for patient care have been developed. The use of AI in various areas of genomics is facilitating wider applications of genomics in patient care and also in population screening.

With use of NGS-based testing for varied phenotypes presenting at all ages including antenatally detected anomalies to late-onset genetic disorders, for cancers and familial cancer predisposition syndromes, for population-based screening for monogenic disorders in neonates, for pharmacogenetics and for cancer screening, a large population is getting NGS-based testing for some or the other clinical reason. This not only needs high throughput and speedy results of NGS, but also needs clinicians empowered with clinical knowledge about when to order the test, deep and reverse phenotyping, and pretest and post-test counselling.

AI is going to help all clinicians in a great way as it is doing for NGS laboratories. The GenExpress in this issue highlights the various ways in which this can be done. Image analysis tools have greatly improved over the last decade and are being successfully used in getting diagnosis/ differential diagnosis for dysmorphology, diagnosis of retinal disorders, histopathological diagnosis etc. This helps 'non-experts' to get an expert opinion, spreading services to remote places. As medical genetics is encompassing all branches of medicine, the need for knowledge about this speciality in clinicians is increasing. There is not only the need of superspecialist medical geneticists but all clinicians to be empowered with the principles and tools of genetics in addition to the knowledge of genetic disorders.

Management of genetic disorders has genetic counselling as an important component. Genetic counselling is also essential before NGS-based testing and while communicating the positive, negative or uncertain reports. AI-based communication methods are being found to be suitable for these purposes. Studies have shown that the chatbot can be effectively used to provide pretest counselling. The GenExpress in this issue has covered two articles about successful and efficient use of chatbots for genetic counselling for breast cancer, etc. As the applications of genetic testing are increasing, the need for genetic counsellors is greatly increasing and even developed countries which have training programs in genetic counselling are not able to cope up with increasing demands. The use of AI-based counsellors appears to be a solution for providing personalized counselling. One article in GenExpress also talks about the efficient use of AI for selecting cases from the neonatology unit for whole genome sequencing.

On the one hand the application of genetics in medicine needs a high level of intelligence, while on the other hand interaction with patients needs a sensitive heart as well. AI cannot replace clinical geneticists when it comes to handling the emotional and ethical issues related to genetic diseases. The GeneFocus and HearToHearTalk articles in this issue talk about the emotional

strain clinicians carry, and the ethical dilemmas associated with prenatal diagnosis, which AI cannot address.

The human mind and intelligence and its power of imagination will find out many more applications of AI for genetics in medicine. We are passing through a revolution of molecular medicine and AI is becoming an important aid for clinicians and hospitals getting zapped with the data, novel diagnoses and new therapeutics. Hence the need for clinicians who are knowledgeable and comfortable with the new form of genetic medicine. The next generation of doctors in medical colleges need to get exposure and experience of this next-generation medicine. This will be possible if the teachers are trained in medical genetics as applied to their specialities. Medical colleges need departments of medical genetics and teachers should be empowered to provide genetics services to the patients. The whole world is thinking of ways to update genetic curricula and upscale the existing knowledge about genetics among medical and paramedical workers. In India, we have our training courses such as the GeneTOP (online) and ICMR Course in Medical Genetics & Genetic Counseling in addition to sessions on genetics in various conferences, and online teaching modules of the Indian Academy of Pediatrics and other societies. Many institutes like the Centre for DNA Fingerprinting and Diagnostics (CDFD), Hyderabad are conducting courses on NGS analysis. The eyes of the medical fraternity in India have opened to the importance of genetics in medicine. We have to put our mind to this and see how to incorporate medical genetics in the medical care system of India and use our imagination to make the best use of AI.

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