EDITORIAL

Next Generation Clinical Practice – It's Man *Versus* Artificial Intelligence!

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Artificial intelligence (AI) is gradually matching up to the human skills in many medical specialties, including clinical genetics. Studies have shown that facial analysis technologies measured up to the capabilities of expert clinicians in recognizing various developmental and genetic disorders [1-4]. Clinical dysmorphologists have developed their skill of recognizing a genetic syndrome based on the gestalt, gradually over the years with experience. This was perhaps the skill that made a clinical dysmorphologist, an expert amongst the physicians. This is now being challenged by AI! With free availability of softwares and applications on the web and mobile phones, it is now easy to obtain a genetic diagnosis, almost instantly, with increasing accuracy. AI can achieve a clinical diagnosis now, without the help of any laboratory or imaging modality. Narayanan, *et al*. [5] have attempted just that through their study published in this issue of *Indian Pediatrics*. The study involved testing the software to make an accurate diagnosis in 51 previously confirmed cases of dysmorphic genetic syndromes.

Face2gene is a promising AI-driven software that exploits a facial image analysis framework, DeepGestalt [6]. This in turn is based on computer vision and deep-learning algorithms to quantify the similarities and differences between various syndromes. A two-dimensional image of the patient is to be uploaded by the physician. Given an image, the face is first detected using a cascaded deep convolutional neural network (DCNN) based methods. The face is further divided and cropped into multiple regions using certain facial landmarks, which are geometrically normalized. Scaling is done for each region compared to a fixed size and converted to grayscale. Specialized DCNNs process the facial regions. It then predicts the probability for each syndrome per region of the face that was initially landmarked. A Gestalt model for syndrome classification is then aggregated. Gestalt refers to the information contained in the facial morphology. Finally, a list of top thirty most likely syndromes is displayed based on a combination of facial gestalt as well as features provided by the user.

Though the computer-based recognition of a genetic syndrome may seem at par with clinical recognition, there are certain challenges in this algorithm, which include limited data, as these rely on comparison of the images with established diagnosis and subtle facial patterns. Also important to address are the ethnic differences that exist not only at the global level but also at regional level. For example, it is not possible to use the same measurements for children from southern and eastern parts of India. As more and more images are being added to the database, the deep learning algorithms are expected to become more robust and specific. Another challenge for face2gene is the ability to recognize a normal face. It is unable to do so because of lack of facial digital data from normal individuals for comparison.

The study by Narayanan, *et al*. [5] is the first of its kind from India, which paves a path for use of this handy software in the clinics. The diagnostic accuracy in this cohort is encouraging. A 72.5% diagnostic rate in patients with dysmorphism gives immense hope to physicians and geneticists to identify rare genetic syndromes while saving precious time and money. However, most of the syndromic diagnoses have been for the more commonly observed disorders, and its efficiency remains to be seen for the rarer syndromes. Further, the study does not have a representation of chromosomal disorders (other than aneuploidies), which are common causes of dysmorphism. Top ten hits seem to be a very liberal criteria to be considered for a positive diagnosis, as it may not reduce the genetic testing algorithms much. Nevertheless, this study proves a place for this application, when used carefully amalgamating the clinical acumen with technology.

In our experience, this tool has been proven to be valuable with diagnosis being established in approximately one-third of patients using this application. The syndromes that were correctly identified included William syndrome, Angelman syndrome, Rubinstein Taybi, Kauffman oculocerebrofacial syndrome, Nicolaides Barraister, Schwartz Jampel syndrome, Coffin Siris, Osteopathica striata with cranial sclerosis and Weidmann Steinner syndrome. One of the challenges observed is the

inability of the software to pick Noonan and Turner syndrome, as well as the low gestalt for ear and limb anomalies.

The tool is useful as it saves not only time for a particular patient and family, but also unnecessary costs incurred due to innumerable tests that are required to achieve the diagnosis. The diagnostic odyssey is averted, many a times. There are many more applications of the facial digital recognition technology, including that of pain assessment in young children, and a potential use in unidentified persons data repositories [7,8]. Perhaps, it is only the start of an era of digital technology assisted clinical practice.

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