GeNeDit

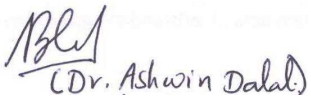
'Face to Face' with our Genes

Editorial

Human face is the mirror to our brains. Face is an important part of the body. We identify ourselves and others based on the facial characteristics. Human face gives identity to an individual. The embryonic development of human face involves complex interplay of large number of developmental events. Hence it is but obvious that face development would be affected in dysmorphic syndromes resulting from mutations in developmental genes. Most of the human dysmorphic syndromes have typical facial characteristics as an important component. The classic text on dysmorphic syndromes by Gorlin has been suitably titled as "Syndromes of Head and Neck". Most descriptions of dysmorphic syndromes include subjective information provided by individual investigators and this leads to lot of confusion for use in clinical practice. There have been many efforts to standardize the terms used in dysmorphology including the special issue of American Journal of Medical Genetics on "Elements of morphology". However the 'subjectiveness' in the assessment is always a confounding factor. Hence there have been efforts towards introducing 'objectiveness' in dysmorphic feature reporting. The initial attempts involved use of two dimensional photographs of individuals. Landmarks were plotted on the photographs at identifiable sites like 'corner of mouth', 'tip of nose' etc. and then a complex statistical analysis would be used to differentiate between 'normal' and 'dysmorphic' facies. Later the investigators used three dimensional images and landmark acquisition which further refined this technique. Recently a mobile application called Face2Gene has been extensively used for facial recognition of various dysmorphic syndromes. Facial gestalt recognition is an art perfected over years of practice but beginners can be significantly benefitted by the use of such apps/software which can aid in diagnosis as well as plan for genetic investigations.

The ability to include 'objectiveness' in face recognition has opened a Pandora's box of possibilities. Face recognition has been used in mobile phones as passwords as well as by law enforcement agencies to track and find criminals. The idea that study of human genetic makeup could help in prediction of 'human face' appears to be science fiction but the same has been recently demonstrated in an article published in Proceedings of National Academy of Sciences (PNAS), USA. The authors have used genetic information from whole genome sequencing data to predict the facial phenotype as well as other demographic details of an individual like eye colour, skin colour, ethnicity, etc with considerable accuracy. This has resulted in renewed discussions about privacy of human genomic data and possible misuse of such techniques for unlawful activities as well as for planning of 'designer babies' with beautiful facial characteristics. As is true with any new breakthrough in science, it remains to be seen how the technology shapes itself in the future.

As clinicians and medical geneticists, it is our responsibility to shape such discussions in future so that the new developments are used for better care of patients and families. 'Genetic clinics' is an effort towards achieving this goal through dissemination of useful and accurate information from the complex world of human genetics to the clinicians in practice.



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