FACEMATCH

USING FACE-MATCHING TECHNOLOGY TO HELP FIND A DIAGNOSIS FOR INDIVIDUALS WITH INTELLECTUAL DISABILITY

Dr Tracy Dudding-Byth

Despite the diagnostic power of advanced genomic sequencing, many individuals with moderate to severe intellectual disability (ID) are unable to be diagnosed due to the sheer number of genes with unknown function. With the vision of improving rate of diagnosis and health outcomes for this group of individuals, Dr Tracy Dudding-Byth and Professor Brian Lovell have recently launched the international FaceMatch Project, which uses computer face-matching technology to match the faces of children around the world with moderate to severe ID who remain undiagnosed following genetic testing.

Intellectual disability [ID] is one of the largest unmet challenges in health care. In developed countries, the most common cause of ID is genetic, and up to 50% of individuals with moderate to severe ID have associated craniofacial anomalies. Presently, only 30-40% of these children have a causative DNA variant in one of the known developmental disorder genes, and there are ~ 2000 ID genes still to be characterised. Furthermore, an estimated 70% of the genes in humans are of unknown function (Wright, Fitzpatrick & Firth, 2018). When DNA variants are detected in these genes, it is challenging to determine if they are normal variations or the cause of the ID. Comparing the DNA variants of two individuals with similar features can lead to novel ID gene discovery and diagnosis. When a diagnosis is not made by genetic testing, doctors manually presented facial images of their patients at medical conferences or record written information into databases hoping to locate another individual with similar features. To solve this clinical dilemma, FaceMatch has repurposed advanced face-recognition technology and developed a first of its kind innovative, international project to provide a means for effectively and efficiently comparing the facial images of the 70% of patients with ID who remain undiagnosed following genetic testing.

Computer face-matching technology

The technology within the FaceMatch Project was initially developed to match facial images of individuals for the primary purpose of recognizing blurry faces in CCTV for policing and counterterrorism. The software is being used to detect persons of interest in large crowd gatherings through various police and other security agencies. Since the software was developed for CCTV, high quality professional photographs are not required and even poor quality historical photographs can readily be used. This technology is unique as it uses low-resolution structural and frequency domain features rather than high resolution features. It is based on spatial textures and statistical models and is simultaneously insensitive to pose, illumination, expression, obscuration, blurring, decoding artefacts, and low-resolution images. Our health research FaceMatch team published an automated approach to matching the faces of non-identical individuals within the same syndrome subgroup within a database of 3,145 images and reported that, using two-dimensional photographs, the technology accurately matches the facial gestalt of unrelated individuals with the same syndrome form of intellectual disability ($p<0.000001$). (Dudding-Byth et al., 2017). Utilising this technology, FaceMatch has forged a solution to locating undiagnosed patients around the world who have similar facial features.

The FaceMatch Platform

The unique dual parent/doctor participation model is future focused and recognises that parents are partners in health care. Every new image is matched against all images within the database generating a ranking list and matching score which is more efficient and accurate compared to the human eye. This state-of-the-art internationally accessible model can be initiated by the parent or the doctor. Parents are asked to nominate a doctor to allow the FaceMatch team to work together with the parent and their own doctor. Images, medical information, and genetic data are securely stored. Notification emails are generated and maintained within the secure FaceMatch system. Parents, doctors, and the research team each have an individual dashboard which summarises status and outcome.

Benefits of an early diagnosis

Patients and families often endure long and isolating journeys toward a diagnosis, and the benefits of an early diagnosis for ID are substantial. A diagnosis can inform the child's prognosis and management; reduce parental isolation; provide information about possible recurrence risk in a subsequent child; reduce the number of invasive investigations and triage eligibility for novel therapeutic trials. Although each of these conditions are individually rare, collectively they inform our knowledge of the complex biological pathways involved in normal cognition and provide the first steps towards targeted treatments for ID.

How to participate

Participation is open to parent or guardians of:

1. Children (or adults) with moderate to severe intellectual disability who remain undiagnosed following review by a clinical geneticist or,
2. Children (or adults) who have a known genetic cause for their intellectual disability.

More information about the project including privacy, confidentiality and security can be found at facematch.org.au

Dr Tracy Dudding-Byth is an Australian Clinical Geneticist within the New South Wales Genetics of Learning Disability service and an Early Career Researcher within The University of Newcastle GrowUpWell Priority Research Center. Professor Brian Lovell is the Professor of ITEE at the University of Queensland.

References

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