THE UNIVERSITY of EDINBURGH

News Release

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Family snaps help doctors pinpoint rare genetic disorders

Rare genetic diseases could be diagnosed with the aid of new computer software that scans ordinary photographs of patients' faces, scientists say.

The new computer programme uses facial recognition technology – similar to that used by Facebook – to build a description of the face's structure by identifying corners of eyes, the nose, the mouth and other features.

Many genetic conditions such as Down's syndrome and Angelman syndrome are associated with changes in facial appearance. These occur because of the large number of genes that are involved in the development of the face and skull as a baby grows in the womb.

Scanning photos of different individuals who have the same genetic condition can identify distinctive facial traits that are associated with that particular disorder. The approach works even in very rare disorders that affect only a few people in the world.

Doctors should one day be able to take a smartphone picture of a patient and run the computer analysis to quickly find out which genetic disorder the person might have, the scientists say. This will be particularly helpful in countries without easy access to genetic tests.

The programme was developed by software engineers at the University of Oxford. Experts from the Medical Research Council Institute of Genetics and Molecular Medicine at the University of Edinburgh provided guidance on the clinical images used to develop the software.

Professor David FitzPatrick, of the MRC Institute for Genetics and Molecular Medicine at the University of Edinburgh, said: "Thousands of babies are born each year in the UK with errors in their genetic makeup which mean that they do not develop normally. Getting to a firm diagnosis as quickly as possible is very important but the extremely large number of possible disorders makes this very challenging.

"This technology will help doctors to pick up extremely rare disorders and may be able to identify new conditions. It brings hope to those families living without a diagnosis and the prospect of information and tailored support to help them face the future."

The findings are reported in the journal eLife. The study was funded by the MRC, the Wellcome Trust, the National Institute for Health Research Oxford Biomedical Research Centre and the European Research Council.

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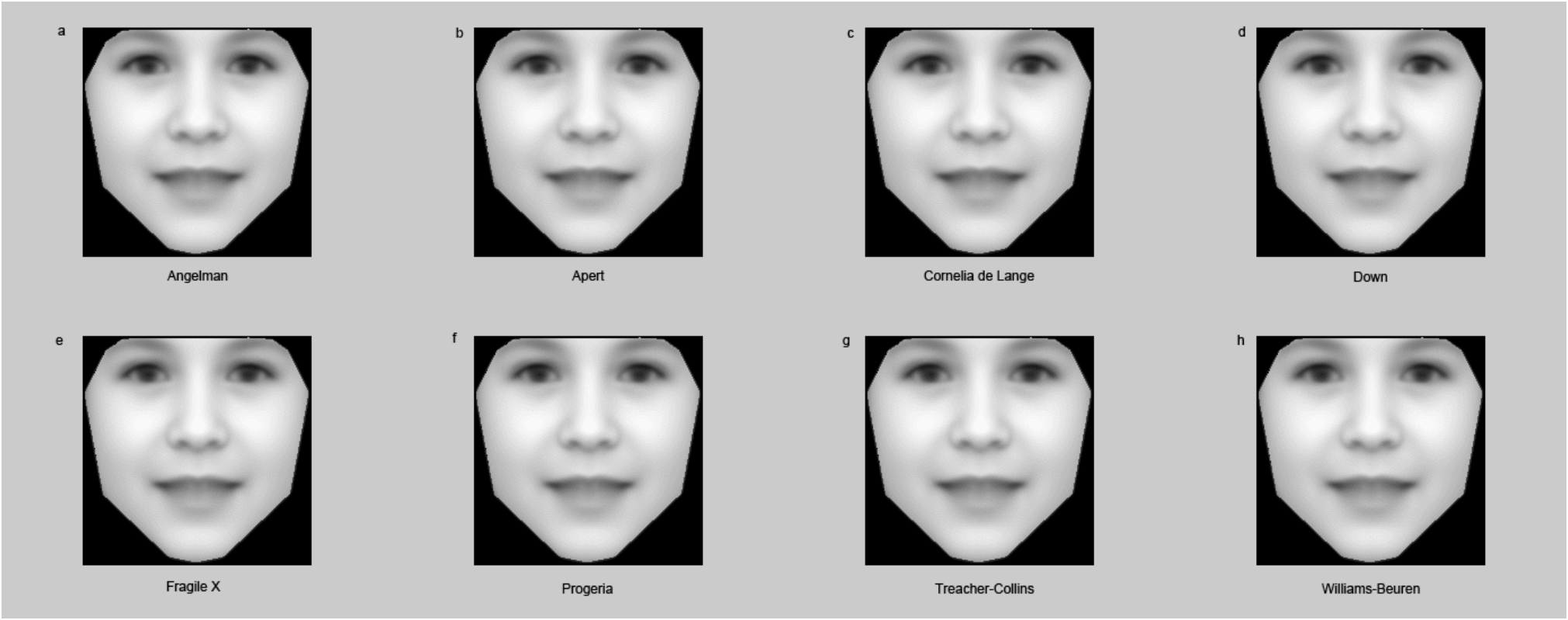
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