The research commercialisation office of the University of Oxford, previously called **Isis Innovation**, has been renamed **Oxford University Innovation**

All documents and other materials will be updated accordingly. In the meantime the remaining content of this Isis Innovation document is still valid.

URLs beginning www.isis-innovation.com/... are automatically redirected to our new domain, www.innovation.ox.ac.uk/...

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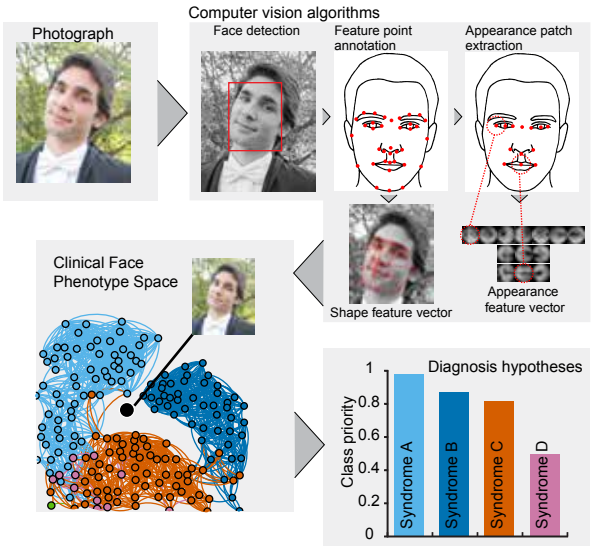
Diagnosis of rare diseases with computational analysis of photographs

Dr Christoffer Nellaker and his team at Oxford's Department of Physiology Anatomy and Genetics are developing computer vision algorithms to automatically analyse photographs of faces for disease-relevant phenotypes.

The approach will aid clinicians to narrow the search space to find the correct diagnosis for rare diseases.

Genetic disorders affect almost 8 per cent of people, about a third of whom will have symptoms that greatly reduce their quality of life. There are over 7,000 known inherited disorders and the path to diagnosis is often very difficult.



Clinical dysmorphology is a key discipline within clinical genetics which requires an enormous breadth of experience to correctly classify and diagnose ultra-rare diseases.

The team are applying computer vision research to enable analyses based on ordinary photographs to be performed for the purpose of detecting disease phenotypes.

They have developed Clinical Face Phenotype Space, an algorithm which will automatically detect faces in photographs, annotate locations of key anatomical parts and extract machine readable feature descriptions of the facial gestalt.

The approach uses machine learning to create a multidimensional space shaped to account for spurious variations such as lighting, pose, occlusions, and image quality. The Clinical Face Phenotype Space algorithm locates patients in the context of known syndromes, and can help to generate disease hypotheses.

Above: automatic average faces from six of the different syndromes used in training the Clinical Face Phenotype Space algorithms

With Clinical Face Phenotype Space, the team are aiming to create an impartial means of narrowing the search space to suspected rare diseases. This could augment the prioritisation of testing in clinical investigations and allow the clustering of patients by phenotype even when no known syndrome diagnosis exists, aiding disease identification.

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