

Computer program reads facial features to identify rare genetic diseases

Doctors faced with the tricky task of spotting rare genetic diseases in children may soon be asking parents to email their family photos. A computer program can now learn to identify rare conditions by analysing a face from an ordinary digital photograph. It should even be able to identify unknown genetic disorders if groups of photos in its database share specific facial features.

Rare genetic disorders are thought to affect 6 percent of people. Genetic tests exist for the more common conditions such as Down's syndrome, but many people with the rarer disorders never get a proper clinical diagnosis. Genetic tests aren't available for many conditions because the gene variants that cause them haven't been identified. This means doctors often have to rely on the pronounced facial features that occur in between 30 and 40 percent of rare disorders to make a diagnosis, but few people are trained to recognise them.

"Clinicians skilled in the use of facial features to support diagnosis are few and far between," says Alastair Kent, director of the charity Genetic Alliance UK. "As a result, families frequently experience long delays – years rather than months – before they receive a diagnosis for their child."

Read the full, original story: [Computer spots rare diseases in family photos](#)