Researchers link facial structure to kidney disease

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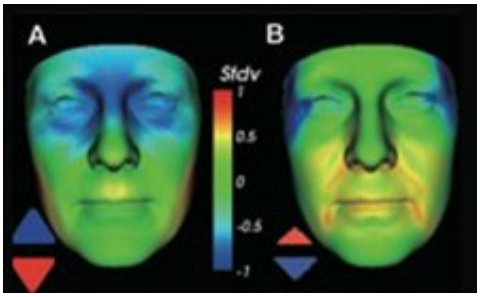

(Medical Xpress)—Researchers at King's College London's Dental Institute have shown that people with a certain kind of kidney disease have characteristic facial features that may reflect the genetic mutation they carry.

A team led by Professor Sharpe—an expert in craniofacial development and stem cell biology at the Institute—has published a paper detailing this phenomenon in the journal *Human Molecular Genetics*.

Autosomal Dominant Polycystic Kidney Disease (ADPKD) is the most common genetic kidney disorder. In the UK, it accounts for around 1 in 10 people on dialysis and 1 in 8 of those with a kidney transplant. Among ADPKD families, about 64-85 per cent of families have mutations in the PKD1 gene and about 15-36 per cent have mutations in

the related, PKD2 gene.

Mice with mutations in these genes show similar kidney disease to humans. In this recent collaborative study with groups at University College London and the University of Cambridge, Professor Sharpe and his team identified specific facial and dental abnormalities in PKD2 mutant mice. These features develop after birth and correlate with the function of PKD2 as a mechanoreceptor, and thus influence the structure of the face by influencing jaw strength and other features.

To the naked eye, patients with ADPKD are not known to have any characteristic facial or dental features. To test whether there was any relationship in humans with these mutations, 3D facial shape analysis—using techniques developed by Professor Peter Hammond's lab at UCL's Institute of Child Health—was carried out on a small group of patients (11 female, 8 male, mean age 48 years). None of the patients had yet reached the stage where dialysis or a kidney transplant was necessary. 'Surprisingly this analysis revealed specific characteristics of ADPKD patient faces, some of which correlated with those of the mutant mice,' said Professor Sharpe, who is also affiliated to the MRC Centre for Transplantation at King's. Patients with ADPKD had a slight vertical lengthening of their faces, slightly longer noses, and less symmetrical faces. 'Our results suggest that PKD2 mutations are thus not only responsible for kidney disease but also craniofacial anomalies in mice and characteristic human features,' he adds.

The complexity of the human face means that a very wide variety of molecular processes are involved in its development. Defects in such processes that are manifested as a specific disease such as ADPKD are also likely to have effects on facial development. Such effects may not always be obvious but the use of 3D shape analysis can reveal subtle characteristics that can correlate with disease phenotypes. 'To what extent analysis of facial features can be used to diagnose disease is an

intriguing question that requires further investigation,' concluded Professor Sharpe.

Provided by King's College London