MedicalXpress

Gaza strip families give first clue to condition causing blindness and tooth decay

February 12 2009

Scientists studying an inherited condition resulting in blindness and crumbling teeth have found a single defective gene can affect both eye function and normal tooth development.

A previously undiscovered and unexpected link between the formation of teeth and eyes has been uncovered by researchers from the University of Leeds, through studies in two families living in a village in the war-torn Gaza strip.

Funded by the Wellcome Trust and Yorkshire Eye Research, the project team sought to identify the cause of a condition they named Jalili syndrome, in which related individuals suffered loss of eyesight, almost from birth, and poorly developed teeth.

Chris Inglehearn, Professor of Molecular Ophthalmology at Leeds Institute of Molecular Medicine, said: "What interested us was the idea that there might be a single process or protein essential in both teeth and eyes, not something you'd normally think of as having much in common.

"Working with colleagues in the Leeds Dental Institute, genetic changes were found that disrupt the function of a protein called CNNM4 and that are passed on from one generation to the next in these families. This protein is present in the cells that lay down tooth enamel and also in the various layers of the retina, the light sensitive 'film' at the back of the eye."

Prof Inglehearn added that little is yet known about the role of this vital protein but it is possible that it is involved in transporting magnesium or calcium to cells in the body. Both minerals are known to be important in visual function and in teeth and bone formation.

Although a rare condition, Jalili syndrome sufferers from France, Scotland, Iran, Bosnia and Guatemala were also studied. Further research could help scientists gain a better understanding of more common forms of blindness, weak teeth and bone diseases such as osteoporosis, and in time may lead to better treatments.

The possibility of using gene replacement therapy to reverse the effects of this condition is still a long way off and would need to be applied soon after birth to be effective.

Martin McKibbin, Consultant Ophthalmologist at St James University Hospital and Chair of Yorkshire Eye Research, said: "The findings of this research are surprising but will give doctors and scientists a greater understanding of the causes of inherited blindness and may identify novel treatments in the future.

"Yorkshire Eye Research is pleased to be able to support this and other research projects in Yorkshire. The charity relies on donations from individuals and organisations in Yorkshire to continue its work."

Jalili syndrome is named after team member Ismail Jalili, who travelled regularly to Gaza and the occupied West Bank trouble spots to recruit the Gaza strip families into the research, until the escalation of conflict made such trips impossible.

More information: The paper "Mutations in CNNM4 cause Jalili syndrome, consisting of autosomal recessive cone-rod dystrophy and amelogenesis imperfect" is published in *American Journal of Human*

Genetics.

Source: University of Leeds