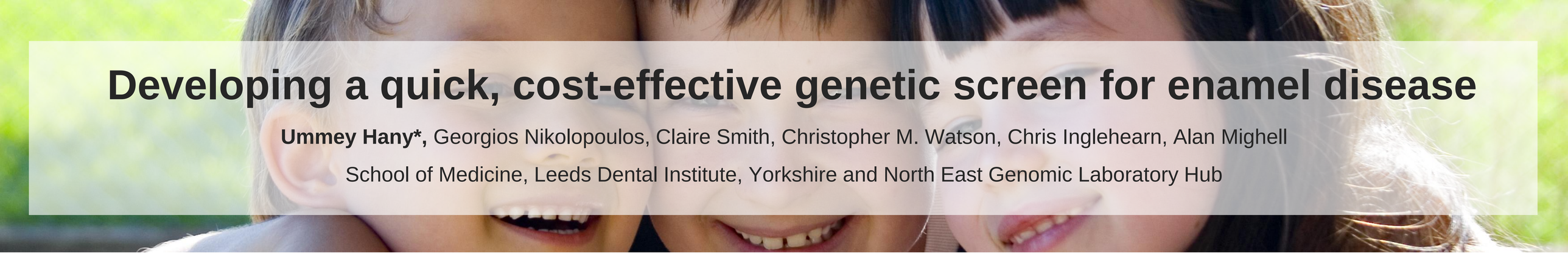


Developing a quick, cost-effective genetic screen for enamel disease

Ummey Hany*, Georgios Nikolopoulos, Claire Smith, Christopher M. Watson, Chris Inglehearn, Alan Mighell

School of Medicine, Leeds Dental Institute, Yorkshire and North East Genomic Laboratory Hub

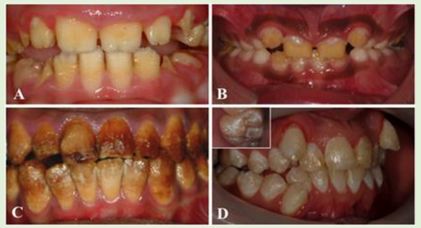
Amelogenesis Imperfecta (AI)

Our smile is central to the way we present ourselves to the world.

AI is the result of a group of rare inherited diseases of enamel formation.

It causes discoloured, misshapen, brittle or sometimes soft teeth. Patients experience pain, frequent infections and early tooth loss.

They also suffer from high levels of distress, social avoidance, and emotional problems.

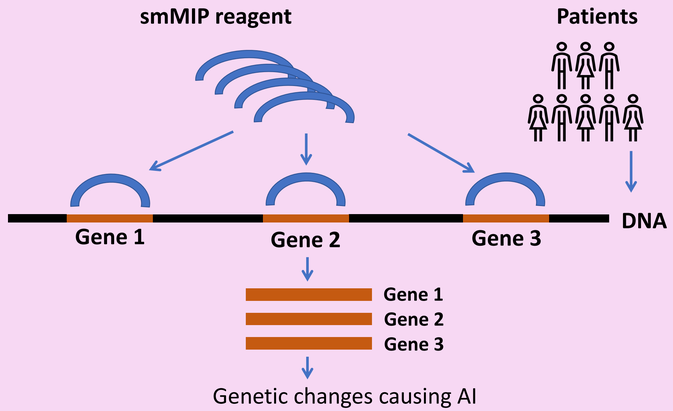


AI usually means lots of trips to dentist throughout childhood. Having AI can be tough.

Screen development

Defects in many different genes can cause AI, and the resulting enamel defects are highly variable.

We have developed a screening method that allows simultaneous screening of all AI genes, in hundreds of patients, cheaply in one test.

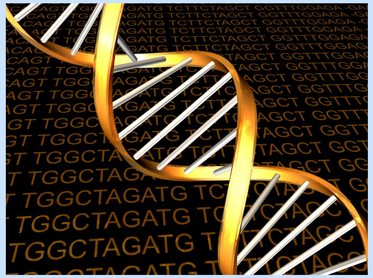


We have successfully screened 20 patients, and are ready to screen DNA from over 300 new AI patients already recruited from centres around the UK.

Research benefits of testing

Helps to uncover all the genes and mutations involved in AI.

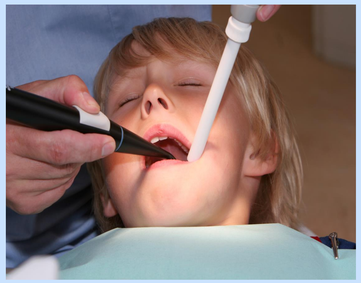
Gives us a better understanding of how teeth form, relevant to other common enamel problems.



Clinical benefits of testing

Gives patients and their families information about future risks and outcomes.

Means patients can be given a personalised treatment that is best for them.



Improved patients pathways lead to better care for all by raising standards and development of new treatments.

The Leeds Teaching Hospitals NHS Trust

References: 1. Children images and DNA image were downloaded from Dreamstime Copyright © 2000-2020
2. Smith et al (2017). *Front Physiol*. 2017;8:435. doi:10.3389/fphys.2017.00435
* Contact: u.hany@leeds.ac.uk

UNIVERSITY OF LEEDS