SPECIAL FEATURE

Gene genie

An important milestone has been reached with the advent of a genetic test to aid the diagnosis of amelogenesis imperfecta, says **Caroline Holland.**

Amelogenesis imperfecta (AI) is the inherited condition affecting the formation of tooth enamel. Dr Alan Mighell, a specialist in oral medicine with a research interest in the genetics of AI, announced the availability of the new NHS genetic test to aid the diagnosis of AI at the British Society of Paediatric Dentistry's (BSPD's) annual conference in Leeds in September 2016.

Starting his presentation with David Bowie's 'The Jean Genie', a song about taking a risk and doing things differently, Dr Mighell challenged his audience to get on board. AI is, according to Dr Mighell, more widespread than we might imagine. He tentatively suggests that up to around 1:700 people have the condition, which falls into one of two broad types – hypoplastic or hypomineralised – with many sub-types.

Patients with the condition and their families are familiar to dental hospital paediatric departments around the UK. Once seen by a paediatric specialist, the condition can usually be identified quickly. In addition to the evident visible traits, there are other indications. On a radiograph, the radiodensity of dentine and enamel of teeth affected by AI can appear the same and dental sensitivity is often an issue. AI can also be linked with other dental conditions, such as anterior open bite. Until now, however, it's been very difficult to recognise at an early stage whether the patients have other linked medical conditions.

Dr Mighell said: 'I get a lot of questions from parents and patients about whether the condition will be passed on to the next generation. They want to know why they have AI. And there is always the question: is it just AI or AI and something else?'

Many patients are very unhappy about their teeth and suffer from emotional distress. Very often, the condition goes in families and is multi-generational.

Moving forwards, said Dr Mighell, colleagues would be able to seek confirmation of a suspected diagnosis of AI which could identify the relevant genetic mutation. They would be able to find out whether they might be at risk of other health issues, such as renal calcification.

He described the role of nanotomography in visualising enamel affected by AI. 'Even the scrappiest bit of tooth can be phenomenally revealing of the underlying phenotype.'

Dr Mighell's talk was one of several in an entire session dedicated to AI, during which it emerged that this cohort of patients would be well served by greater clarity around their diagnosis.

The eminent paediatric dentist Kathy Harley, a speaker in the same session, in which she shared her restorative approach for young people with AI, later paid tribute to the 'fabulous work' that is taking place in Leeds. 'The whole genetic evolution is so exciting because ultimately we may be able to identify AI patients at birth and, over time, with differing interventions, these patients will need less and less treatment.'

However, the path to routine genetic testing for AI is not necessarily smooth. There are some considerations. Firstly, the fact that the test cannot deliver all the answers a family might want. Given that all the genes that cause AI are not yet known, the NHS test could simply be returned as negative and other avenues would need to be explored.

Then there is the cost to the NHS – £860 per test. As with many actuarial decisions, the fee which would potentially yield a diagnosis and an early treatment plan and inform improved care pales into insignificance when weighed up against the current cost of managing a young child with the condition where GA and long-term restorative costs are the norm.

Dr Mighell said there was an under recognition of the burden of AI on the NHS and on patients. Meanwhile, patient expectations are going up and there is a clear need for the translation of research into patient care. 'The genetic test fits well with the development of Managed Clinical Networks to improve standards of care.'

What's not clear is how many specialist dentists will want to request genetic testing. Suzanne Scott, senior lecturer in health psychology at King's College London Dental Institute, explained that gaining informed consent for a genetic test is essential, yet complex. There is research showing that patients who are not fully informed about the test or the implications of genetic testing can be more distressed by the result.

She said there is a considerable amount of guidance on the issue. It was important that the professionals have been appropriately trained in offering a genetic test while patients and their families need to be sufficiently supported when they receive the result.

'You need to be careful that if there is going to be a referral, it should happen early on – it should not be a last resort. And there should be clear criteria for referral. When you are dealing with genetics there are issues around family dynamics and there may be feelings of guilt or shame around passing on a condition.'

After the availability of a genetic test was announced at the BSPD conference, members of BSPD were invited to participate in focus groups to share their views. It's anticipated their feedback will help inform the transition to a routine use of the test. In the coming months, if you think a patient of yours has amelogenesis imperfecta (AI) then genetic testing can be requested via a specialist in paediatric dentistry.

Said Dr Mighell: 'We are in the middle of a genetics revolution because we can sequence people's DNA at a previously unimaginable rate and at relatively low cost and computers can handle and sort through the mass of data that this generates. Genetics is going to move to the forefront of healthcare in this and many other areas.'

Even bigger developments are on the horizon. Already, a new drug trial has found it's possible to switch off some AI genes in mice. Dr Mighell added: 'This may be possible in humans sooner than you might think.' ■