



Amelogenesis imperfecta (AI) and Dentinogenesis imperfecta (DI) frequently asked questions

### **What do AI and DI mean?**

The words are really little more than descriptions; they come from the Latin roots:

*Amelo* (enamel) or *Dentino* (dentine - or dentin in the USA!)

*Genesis* (making, growth, development)

*Imperfecta* (poor, less than brilliant)

So, it should be perfectly OK to call these conditions "poorly-made enamel", or "poorly-made dentine" but that doesn't sound as important!

### **Are all blemishes of the enamel or dentine AI or DI?**

No, such blemishes may be congenital (literally "with birth") or acquired. AI and DI are congenital, in fact, they originate in the DNA. Acquired conditions result from such "outside" influences as severe systemic upset in infancy, which will have a generalised effect, or a blow to a primary tooth resulting in a mark on a permanent successor - a localised effect.

### **So, how are the conditions defined?**

AI has for a long time been defined as "A developmental condition, affecting predominantly the enamel and occurring in kindreds such that all, or nearly all of the teeth are affected in the same manner and to the same degree, in the absence of other findings."

This is still essentially true but work by Backman in Scandinavia has shown that the clinical appearance (phenotype) can in fact vary between family members (but see below - x-linked AI) and there are reports of AI affected people having other whole body findings which may or may not be related.

### **How do I know whether I (or my child) have this?**

AI and DI are genetically determined. They are inherited in the same way that our hair or our skin colour or our nose shape are.

DI and AI affect different parts of the teeth - the inner dentine and the outer enamel respectively. That's a matter of observation and a dentist should be able to help you out if you have any doubt about that. What you're really trying to do therefore is decide whether this is an inherited or an acquired condition.

So, there are three main questions to ask when trying to decide whether this is AI.

The first is "Does anyone else in the family have this or something like it"?

If the answer's "no" then this is either **not** AI, **or** it's a newly arisen change (technically but a bit spookily referred to as a new mutation) **or** you may be considering a recessively inherited condition - one where both biological parents are "carriers" of the genes for AI but in neither of whom is it expressed.

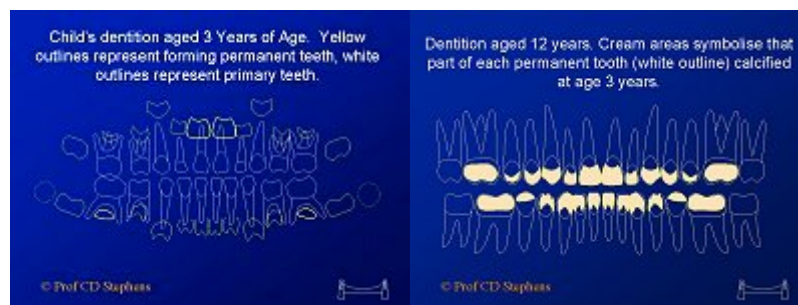
*Oh, a note for parents and practitioners! These new changes in genetic make-up (new mutations) do of course arise but it's much more likely that this is a family already affected by the condition. I do know of families where the adults have not yet "confessed" to the children that they are themselves affected but I always at some point have a look at both parent's teeth. A phrase such as "I'm going to embarrass Mum (or Dad)" works wonders!*

The second is "Are all the teeth affected equally"?

This should be read carefully. ALL and EQUALLY are important inclusions here. One of the side-agendas to all my work in this area was to try to find ways to diagnose between AI and Fluorosis. I nowadays am unlikely to allocate a diagnosis of AI for a youngster until the premolars or second molars (which form much later than the incisors) have erupted. An acquired insult which has EQUALLY affected those teeth as well as incisors must have had a pretty long duration!

The third question is "Is there a chronological distribution"?

This is unfortunately a bit difficult for a lay-person to assess (as it is for many of my undergraduates!). What it means is - is there a "tide-mark" amongst the teeth which suggests that the damage occurred at a particular point in time? If there is, then it speaks of a discrete insult, rather than the continuing developmental change brought about by AI (or DI).



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### **Are teeth affected by AI or DI weaker than other teeth?**

Often yes. There are a number of different types of appearance described in AI; some of them are predominantly hypoplastic (under-grown) some of them are hypomineralised (under-mineralised) some of them mix both findings.

The undergrown varieties have less enamel but it is usually of a reasonable quality. They will wear sooner but are not much more susceptible to decay than unaffected teeth. The under-mineralised group can be very weak and may crumble as they erupt (come through). The severe case on our [New Smiles web-page](#) is of a hypomineralised form. You can see the loss of enamel.

In DI, the teeth are small, bulbous and with shorter roots. The enamel is pretty well normal but the dentine scleroses (it increases in density and becomes less of a "shock absorber" with time). Along with this, the wear on the enamel can be rapid and it may even flake off.

### **Is gene therapy an option here?**

No. We know too little about this possible line of treatment for even life-threatening conditions as yet. As the man says - "Don't even go there"!

As a matter of interest, once the teeth are formed there is presently nothing (a probably theoretically nothing!) that can be done to alter their structure. So any attempt would have to work in the earliest years. Science fiction at present I'm afraid.

### **OK; so what can we do?**

Lots. I am really very concerned about the social aspects of these conditions. I have heard far too many harrowing reports from affected adults of their childhood experiences. This is an area of work that I want to follow up since I believe that Health Care Professionals haven't taken sufficient interest in it so far. If you're affected by either of these conditions and would be willing to take part in a questionnaire sometime in the future, please would you email me at [ai-di@bristol.ac.uk](mailto:ai-di@bristol.ac.uk)?

The dental care for people with AI and DI comes in three broad phases.

INFANCY (birth to 5 years-ish). Someone is going to have to discuss with the parents of the affected child the balance between intervention and leaving alone. This will depend upon factors such as the rate of destruction of teeth tissue either by wear or by decay; the cooperation of the youngster; the particular skills of the operator. There are now specialist Paediatric Dentists in many countries who have had particular training in these areas and it would be sensible to seek the advice of such specialist instead of - or as well as - a generalist.

Treatment at this stage - if recommended - is most likely going to involve the use of metal crowns to protect the back primary teeth from wear and sensitivity, together with either tooth-coloured crowns (strip crowns) or composite (tooth coloured) veneers to the upper front teeth. It is rare to treat the lower front primary teeth; they don't show a lot and they are fearsomely small to deal with - but it is possible!

The metal crowns may not look great but they are effective. They have the longest life-expectancy of any restoration for the primary teeth and long life -expectancy means fewer - or no - replacements!

This level of treatment - which is intended to be preventive and aesthetic (socially preventive?) is hard work for a small child. Careful thought will have to be put to how this is done, whether with the help of some sedation in those countries where that is used, or with a general anaesthetic. Your views and experiences would be welcomed by e-mailing [ai-di@bristol.ac.uk](mailto:ai-di@bristol.ac.uk).

CHILDHOOD and ADOLESCENCE (6 to mid / late teens)

A time of transition. The First Permanent Molars (Six-Year Molars; "Sixes") erupt from about 6 years of age (Surprise!). This is when we begin to be clearer about a diagnosis. It is possible that we were wrong all along, that what we saw in the primary teeth wasn't AI or DI at all (but if you worked through the three questions carefully then this is less than likely) OR, very rarely indeed, that the permanent teeth are going to be unaffected or at least less affected than the primary teeth were.

This is a very important time in a young person's life when they need all of our help and support. Restoratively, it's a really difficult time as teeth come through and reveal areas of disfiguration. It is possible to place composite filling on these teeth to mask or protect them but they will continue to appear to lengthen for a long time and there will be inevitable "tide-marks" between composite and gum as this happens. OK, this can be dealt with at regular reviews but it's frustrating for everyone concerned - and time-consuming and may be costly!

I am VERY reluctant to use restorations which require tooth-removal at this time in the young person's life for this reason and one other.

Any restoration, however well done, has a life span. You would not expect a new car - even a Rolls Royce say - to last for 10, 20 or 50 years without regular servicing and the occasional replacement of parts. Generally speaking, car parts can be replaced by unbolting one bit and adding on another. Sometimes, particularly with body panels, this means cutting away bits of surrounding damaged steel. In the end, you could still replace the whole wing, for example, and you'd be back where you started.

Teeth aren't like that. Yes, when you replace a restoration (filling, crown, veneer or whatever) you may have to cut away surrounding tooth damaged by decay or revealed by further tooth-growth but no, you can't ultimately replace the tooth other than with an artificial one.

So - Materials added to teeth without tooth preparation are the order of the day! This is ESPECIALLY true for DI where the dentine part of the tooth is brittle and tooth preparation may weaken it catastrophically. Pre-formed metal crowns for premolars are available - just like the ones used for primary molars - from [3M](#).

Care at this time may be provided by the Paediatric Dentist, should be supported by a generalist (a General dental Practitioner) and should be planned in conjunction with the advice of the Restorative Dentist who will be carrying out treatment in the last stage which follows

#### EARLY ADULTHOOD (17 years plus)

The British Specialty which deals with Crown and Bridge and implants and such is Restorative Dentistry. If child, family and Dentist have worked together effectively up to now, we should see a young person confident of their appearance, open to dental care under local anaesthetic and with the correct oral hygiene and dietary practices in place to move on to longer term restorations. Ten years ago I would have written "permanent" restorations; two or three years ago I would have said "definitive" restorations; now I'd say "longer term".

I've already explained about the life-span of any kind of restoration. With the improvements in adhesive dental materials, more Restorative Dentists are using composite materials to adjust the appearance of front teeth in the mid- to long-term. At the back of the mouth, the same adhesive technology can be used in adolescence to secure gold "onlays" (biting surfaces) again with minimal tooth-preparation. Tooth preparation for "crowns" may not be needed.

### **OK - that's the teeth, but what about bullying?**

This organisation [Changing Faces](#) is brilliant! Their motto is "Changing the way you face disfigurement". Most of their work has been with outwardly affected individuals although they agree that dental disfigurement can be just as disabling. They have previously agreed to provide support for some of the youngsters I work with. This is fairly recent so I haven't yet had any feed-back as a result. They can arrange all sorts of support, right down to visits to schools if necessary. Try them!

And please tell your dentist that this is an issue for you. Please.

If you need any more information, or to help us improve this page, please contact me at [ai-di@bristol.ac.uk](mailto:ai-di@bristol.ac.uk).

*Peter Crawford*

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