

Be prepared to be surprised by MODY



Tim Butler

Does familiarity breed contempt? MODY is a cluster of conditions which are characterised by single gene defects leading to diabetes. At first it often appears to be typical diabetes, but with attention to detail, the unusual features of MODY can be identified. This presents a challenge to us all in how can we maintain this attention to detail as care moves into the community. The increasing burden of diabetes upon an already beleaguered primary care sector means that more complex care is carried out by practitioners, who are generalists and not specialists. Those of us who are specialists operating with the community have a responsibility to encourage our colleagues to watch out for the unusual: whatever it may be.

With the explosion in the incidence of diabetes, the increasing complexity of treatments and the system's drive for relentless efficiency, we are developing systems which cater for those who 'fit'. It's the patients that don't fit, the atypical, who present the challenge. The targets are for population management but increasingly, individuals are getting lost as priorities are directed towards the average person with diabetes, not the atypical one. Indeed it is the overall complexity of the condition and multifactorial presentation and phenotypes of diabetes that has bedevilled the streamlining and delivery of services. No 'one size fits all'. MODY, defined as a heterogeneous group of disorders, is yet another example of this diversity within the field of diabetes.

Many of our complex patients remain within secondary care while others receive care from their GP surgeries or local GPSI services. This also presents a challenge which we are well familiar with. For our non-complex patients, often described as those without complications, an even greater proportion are cared for in the community. With MODY accounting for between 1 and 2% of our population with diabetes, these patients are certainly not the average,

yet, they are not necessarily regarded as the 'complex patient'. As a result many will exist and survive within the primary care sector without ever seeing a specialist. With no quick easy way to test for MODY, the skill to identify those suitable for screening relies on the attention to detail, looking for the unusual and awareness of the condition. Indeed, the desire of the Department of Health to move an even greater proportion of diabetes care into the community adds to this challenge. (DoH, 2000, 2002).

With the above in mind, I strongly encourage readers to take the time to read the following article by Webster and Owen. Why? It may make all the difference; the difference between developing or not developing complications or between a lifetime on insulin therapy or not. The authors provide an appropriately pitched introduction to MODY and highlight the need to be aware of the diagnosis. In the next issue of the journal case studies will be presented which illustrate the potential impact of a diagnosis. I refer readers to the excellent website www.diabetesgenes.org to find further details. Our limited experience of this service has been extremely helpful. I would also suggest that primary care practitioners contact their local laboratories before they start to embark upon c-peptide and autoantibody studies as these can prove problematic in the community. Finally, if I had to recommend any take-home message, it would be to look at the inheritance pattern in their younger type 1 patients: If relatives are affected then look into it further.

So does familiarity breed contempt? Yes, but it is in looking for these surprises and spotting the atypical which makes a specialist team. With 1–2% of people with diabetes affected, there are certainly surprises out there. ■

DoH (2000) The NHS Plan: a plan for investment, a plan for reform. DoH, London

DoH (2002) The National Service Framework for Diabetes. DoH, London

Tim Butler is a GPSI in Diabetes for County Durham PCT, and an Honorary Clinical Lecturer in Diabetes at the University of Newcastle Upon Tyne.