Integrating genetics into diabetes care: a new role for DSNs

Maggie Shepherd, Amanda Stride, Sian Ellard, Andrew T Hattersley

Introduction

The importance of molecular genetic testing in diagnosing genetic subtypes of diabetes is well recognised as it can guide optimal treatment, help genetic counselling and explain the co-inheritance of apparently unrelated features (Stride, 2002). Many diabetes professionals do not recognise maturity onset diabetes of the young (MODY) and are unaware of the genetic tests available (Shepherd, 2001). MODY link nurses (MLNs) are trained in the genetics of diabetes and the genetic tests available. MLN s aim to increase awareness and recognition of MODY which should lead to an increase in accurate diagnosis and treatment. This article describes an exciting opportunity for DSNs to extend their role in the area of genetic testing in diabetes. This approach of training DSNs in genetics may provide a model for disseminating genetic information in other genetic disorders.

Diagnostic molecular genetic testing for people thought to have maturity onset diabetes of the young (MODY) has been available at the Royal Devon and Exeter NHS Healthcare Trust since December 1999. An educational initiative was funded by the Department of Health (DoH) to develop the integration of genetics services into diabetes care and has led to the appointment of 12 MODY link nurses (MLNs).

The identification of six genes in which mutations cause MODY has allowed the genetic cause of diabetes to be identified in over 80% of families who fit the clinical criteria of MODY (Owen, 2001). In these cases, a clinical diagnosis may be confirmed by molecular genetic testing for which Exeter is the UK referral centre. In the first 3 years, 361 samples have been received and a MODY gene mutation found in 63 out of 303 (20%) families tested. This lower detection rate suggests that the people who are sent for diagnostic testing frequently do not have MODY.

It is noticeable that the majority of the positive referrals have come from a limited number of well-informed clinicians. Molecular genetic diagnosis enables appropriate clinical management. People with MODY due to a mutation in the hepatocyte nuclear factor (HNF)-1α gene are sensitive to sulphonylureas (Pearson, 2003) whilst people with glucokinase (GCK) mutations require no treatment (Hattersley, 1998).

Healthcare professionals in diabetes are often unfamiliar with the key characteristics of monogenic forms of diabetes and misdiagnosis as type 1 or type 2 diabetes is common (Hathout, 1999, Lambert, 2003; Lehto, 1997; Moller, 1998). Professionals may be unfamiliar with the latest genetic findings and unsure of the most appropriate treatments. The best way to disseminate and integrate this information into clinical practice is uncertain. However, the DoH is encouraging initiatives to bring the benefits of genetics into mainstream clinical areas, particularly using the skills of specialist nurses (DoH, 2003).

Aims

The aims of the MLN project are to educate experienced diabetes nurses about genetic forms of diabetes and the genetic tests available, and for these nurses to disseminate the information to hospital diabetes teams within their area through presentations and case discussions.

MLN posts

The DoH funded six MLNs at the start of the project in July 2002; extra DoH funding has subsequently allowed the appointment of an additional six MLNs who started in post in May 2003. Funding is in place for these 12 MLNs until April 2004, and it is hoped that this will be extended. The 12

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1 MODY is characterised by autosomal dominant inheritance, type 2 diabetes and a young age of onset.

2 Patients are often misdiagnosed as having type 1 or type 2 diabetes.

3 Diagnostic genetic testing can confirm MODY and define the subtype which has implications for treatment.

4 MODY link nurses (MLNs) work in different locations throughout the UK.

5 The MLN role offers experienced DSNs the possibility of extending their role by developing and disseminating their knowledge of the genetics of MODY to hospital teams.

KEY WORDS

- **MODY link nurse**
- **Misdiagnosis**
- **Genetic testing**
- **Education**

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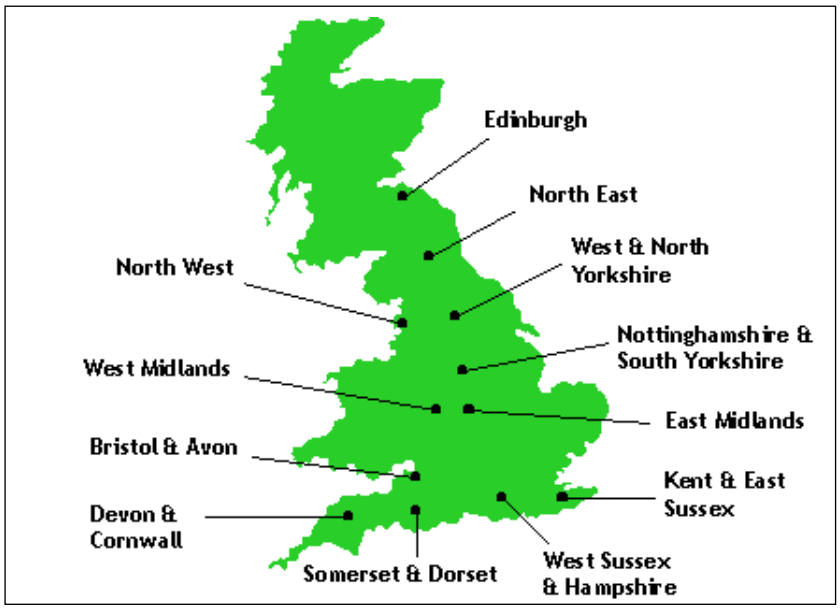

Figure 1. Location of MODY link nurses in post

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1 MLNs raise awareness about MODY and identify possible MODY families, following specific on-going training about genetics.

2 The MLNs assist in identifying which families are likely to have MODY, advise which genetic test would be the most appropriate and provide information regarding the process of genetic testing.

3 MLNs attend study days in Exeter at the start of the project, after 2 months and then every 4 months.

4 Close follow-up and support for the MLNs is provided by the Exeter team by phone and email.

MLNs are based in different regions of the UK (*Figure 1*). The nurses continue their current posts but are seconded to the MLN project for 3.5 hours per week. For practical reasons most of the nurses work on the project 1 day every fortnight.

The role of the MLN

The MLNs act as regional specialists and as a resource for their area. They help raise awareness about MODY and identify possible MODY families, following specific on-going training about genetics.

MLNs aim to visit 10–15 hospitals a year within their area. Letters were sent to the hospitals within each MLN's area asking if they would like the MLN to visit their department to give a presentation about MODY. The MLNs contact adult and paediatric diabetes teams, renal teams (where people with HNF-1β mutations are likely to be identified) and obstetric teams (where people with GCK mutations may be recognised). Presentations about MODY and information about the genetic tests available are given to departments or at regional meetings.

MLNs are involved in supporting treatment changes for patients, particularly those with HNF-1α MODY transferring from insulin to sulphonylureas (Shepherd, 2003) and visiting families thought to have MODY. They follow up family members of people in whom a MODY gene mutation has been found to ensure appropriate counselling and early diagnosis of those relatives at increased risk of developing diabetes. The nurses are able to assist in identifying which families are likely to have MODY, advise which genetic test would be the most appropriate and provide information about the process of genetic testing, including the type of sample required and completion of the MODY diagnostic genetic test request form.

Training in genetics

The MLNs attended study days in Exeter at the start of the project, after 2 months and then every 4 months. Training has included sessions on:

- The different types of MODY: GCK, HNF-1α, HNF-4α and HNF-1β.
- The implications of genetic testing for patients: genetic counselling; the impact of stopping insulin as a consequence of genetic testing; and patients' experiences.
- Molecular genetic techniques and interpretation of results.
- Identifying MODY families: case presentations, use of family trees and tracing diabetes within families.
- Other types of diabetes and their genetic links: young onset type 2 diabetes; the genetics of type 2 diabetes; genetic risk and tests in type 1 diabetes; and transient and permanent neonatal diabetes.

In addition, the MLNs have given presentations about their work during the previous 4 months, and discuss possible cases identified in their regions.

Close follow-up and support for the MLNs is provided by the Exeter team by phone and email. Possible cases of MODY and appropriateness of genetic testing is discussed with the nursing lead, the doctor lead for the group, the head of the molecular genetic laboratory, the clinical research fellows or the clinical scientists. Each of the MLNs in post from May 2003 has one of the original link nurses as a mentor for additional support.

Evaluation of the MODY link nurse project

The MODY link nurse project will be evaluated on several levels. The appropriateness of referrals for genetic testing according to clinical criteria will be assessed, with the aim of introducing guidelines for genetic testing in diabetes which would be of long term

benefit in reducing inappropriate referrals. The numbers of referrals from each region will be assessed before the MLNs start in post and on-going throughout the project in order to provide feedback to the link nurses and the hospital teams. The referrals will also be evaluated in terms of the numbers of mutations found.

In order to evaluate their development in the role, the MLNs completed assessment forms at the start of the project and will repeat this at the end of the project. Their understanding of genetic terms is assessed as well as their confidence performing activities related to the role, e.g. drawing family trees and giving talks on MODY. They are also given 10 case studies (at the start and end of the project) and asked to indicate the most likely type of diabetes, whether they would recommend genetic testing in that case and which genetic test would be the first priority.

The talks and presentations given by the MLNs to the hospitals allocated to them within their region are also evaluated (see below). The hospital teams are provided with evaluation sheets to complete which relate to the educational value of the session, the quality of the presentation and the overall usefulness of the session. The MLN is also asked to evaluate the session and completes a form with reflective comments about the perceived response to the presentation, the plan of action (such as future discussion of possible cases identified by the team) and any difficulties encountered.

The study days in Exeter are also evaluated and additional topics included to meet the needs of the MLNs. Evaluation of the project will be reported in the future. A longer term approach is needed due to the time taken to disseminate information about genetics, identify possible MODY families, perform genetic testing and analyse the results.

MLN perceptions of the role

Many of the nurses applying for the MLN role were happy in their existing post but welcomed the challenge and opportunity to learn more about monogenic diabetes and genetic testing:

'I thought I knew what MODY was but when I started doing this I realised how much I didn't know and the finding out has been excellent.'

Many of the nurses were unfamiliar with MODY before taking up the post:

'We had a few patients that had been labelled MODY and when we discussed it as a group nobody could define nor explain what MODY was! We were all in the dark.'

The nurses were amazed how much they learned about MODY and the genetics of diabetes at the ongoing study days:

'The study days were the best I have ever done. I learned so much and everyone keeps asking me questions about MODY.'

The MLNs and diabetes teams were delighted when they identified possible cases of MODY:

'It is brilliant when you discuss cases and you can see the dawn of realisation that their difficult patient actually fits into a recognised clinical pattern.'

Feedback about MLN presentations from the hospital teams

The nurses' presentations were well received by the diabetes teams they visited. In order to aid evaluation of the MLN project, the nurses giving the presentation and the audience were both encouraged to evaluate the session. Diabetes teams found the sessions interesting and informative:

'Very informative and useful, as personally, I did not know much about MODY, in particular diagnostic criteria.'

The talks raised awareness of MODY and also influenced practice which would aid identification of patients with MODY:

'Extremely interesting — will make me more specific when taking family history.'

Diabetes teams are more specific about taking family histories from people with diabetes as they are more aware of the autosomal dominant family history. Referrals for genetic testing are more specific as the key characteristics of MODY are discussed. Increased awareness regarding the management of people with MODY has resulted in the medication prescribed being changed if appropriate.

Prior misconceptions regarding MODY were also dispelled:

'I did not realise many of the distinctions between type 2 diabetes and MODY.'

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3 The study days in Exeter are also evaluated and additional topics included to meet the needs of the MLNs.

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Table 1. Details of MODY link nurses in post

North West Jane Houghton - 01772 523970	**Edinburgh** Jill Little - 0131 5371747
West Midlands Gill Salt - 07971 197364	**North East** Joanne Berry - 0191 3011533
Bristol and Avon Helen John - 01179 282892	**West and North Yorkshire** Sandra Dudding - 01274 364453
Devon and Cornwall Jackie Jones - 07929 023312	**Nottinghamshire and South Yorkshire** Heather McMahon - 01909 502640
Somerset and Dorset Lynne Balian - 07870 611317	**East Midlands** Ann Cartwright - 02476 540617
West Sussex and Surrey Jennie Brown - 01444 441881 x 4436	**Kent and East Sussex** Sue Petts - 01303 228827

The value of including paediatric diabetes teams was also recognised. The MLNs recognised that some diabetes teams were unfamiliar with the subtypes of MODY and that many mistakenly thought MODY was the same as young onset type 2 diabetes. Issues about the cost of genetic testing were raised in several cases:

'Interested, but some concerns about costs of genetic testing'.

As a result of the presentations many patients were identified as possibly having MODY:

'Some possible patients were identified in group discussion.'

In these cases, the MLN subsequently reviewed individual case notes to gain further details of the presentation of diabetes and family history to help the teams consider whether a diagnosis of MODY was likely, and whether genetic testing would be appropriate.

How to contact your local MLN

Any diabetes team that would like a presentation or update from the MODY link nurse in their region can contact their local nurse direct (*Table 1*). If you think you may have a patient with MODY who you would like to discuss, please contact your local nurse directly or the Exeter team. Further details of the MODY link nurse project, including the areas covered, the MODY link nurses email addresses and the Exeter team contact details may be obtained from this website: www.diabetesgenes.org. If you are interested in the possibility of taking up the role of MLN, please contact Maggie Shepherd.

Conclusion

The role of MLN offers DSNs the exciting opportunity to develop their role and increase their understanding of the genetics of diabetes and the genetic tests available. Becoming a MLN fulfils the possibility of continuing existing roles whilst learning new skills and knowledge. MLNs have been well received by the diabetes teams in their area and have helped to identify families previously misdiagnosed with type 1 or type 2 diabetes. Over 50 hospital diabetes teams have been visited since the start of the project and the MLN presentations have been well evaluated. The provision of this resource has been beneficial to both hospital teams and MODY families who have been supported in transferring from insulin to sulphonylureas. The approach of training experienced diabetes nurses in the genetics of diabetes may provide a useful model for the dissemination of genetic information in other conditions. ■

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