MODY link nurses: pushing the boundaries of diabetes nursing

Sandra Dudding, Heather McMahon and Maggie Shepherd

Introduction

The role of the MODY (maturity-onset diabetes of the young) link nurse (MLN) has enabled diabetes specialist nurses (DSNs) to step beyond the boundaries of their existing roles. The MLN role has added a new dimension that provides possibilities for personal and professional development, learning new skills, expanding knowledge regarding the genetics of diabetes, and networking with professionals both locally and nationally. Since July 2002 the Department of Health has funded the appointment of MLNs throughout England and Scotland, with 17 MLNs now in post. These MLNs are DSNs who have regular training and support to enable them to increase recognition and awareness of MODY and other monogenic forms of diabetes amongst hospital teams in their region, leading to an increase in the proportion of patients accurately diagnosed and treated. This article describes how these DSNs have extended their role in the area of genetic testing in diabetes.

The MODY link nurse (MLN) project is an educational initiative funded by the Department of Health (DoH) to develop the integration of genetics into diabetes care. The Royal Devon and Exeter NHS Foundation Trust provides diagnostic molecular genetic testing for maturity-onset diabetes of the young (MODY) for patients throughout the UK via the UK Genetics Testing Network. Prior to the appointment of MLNs, only 20% of the samples received in the laboratory for testing for mutations in MODY genes were positive, with the majority of these being from a minority of well-informed clinicians (Shepherd et al, 2003). It was also noted that many areas of the UK were not referring patients for genetic testing in MODY. We hoped that employing experienced diabetes specialist nurses (DSNs) throughout the UK to educate healthcare professionals in their regions would lead to an increase in the number of patients accurately diagnosed as having a monogenic form of diabetes. An evaluation of the project after the first 20 months is reported in a paper submitted for publication in a future issue of the *Journal of Diabetes Nursing*.

Six MLN posts were initially funded by the DoH from July 2002 for 18 months. Due to the success of the project, additional DoH funding has subsequently enabled a further three phases of nurses to be seconded to the project, with 15 nurses now in post throughout England and two in Scotland supported by funding from the Scottish Executive (*Figure 1*).

What is MODY?

MODY is a monogenic type of diabetes: this means that in a family the diabetes is caused by a change in a single gene. It is estimated to affect 1–2% of people with diabetes, which is approximately 20–40 000 people in the UK (Shepherd et al, 2001). Although this equates to a small proportion of the population with diabetes, a correct diagnosis confirmed by molecular genetic testing can lead to positive treatment changes for many individuals. The average UK diabetes clinic with 5000 patients would be expected to have approximately 50 patients with MODY. The identification of mutations in six genes that cause MODY means that both predictive and diagnostic testing is now possible in most (approximately 89%) of MODY families. There are further MODY genes still to be identified, as mutations are not found in 11% of MODY families (Shepherd, 2001).

MODY is characterised by three main features:

- Early onset diabetes, which is usually

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1 The MODY link nurse (MLN) project is a DoH-funded educational initiative to develop the integration of genetics into diabetes care.

2 Seventeen MLNs in the UK are seconded to work on the project.

3 The MLNs aim to increase awareness of monogenic diabetes within their area and support local families with MODY.

4 The MLN role has enabled DSNs to increase their skills and knowledge regarding the genetics of diabetes.

5 This successful approach could provide a model for disseminating genetic information in other genetic disorders.

KEY WORDS

- **Maturity-onset diabetes of the young**
- **MODY link nurse**
- **Genetics**
- **Education**

Sandra Dudding is a DSN at Bradford Royal Infirmary Foundation Teaching Hospital, Bradford, Heather McMahon is a DSN at Doncaster and Bassetlaw NHS Foundation Hospital, Worksop, and Maggie Shepherd is Honorary Clinical Senior Lecturer at the Peninsula Medical School, Royal Devon and Exeter Foundation Hospital, Exeter

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1 Maturity-onset diabetes of the young (MODY) is characterised by three main features: early onset diabetes, non-insulin dependent diabetes and autosomal-dominant inheritance.

2 Different sub-types of MODY have distinct clinical characteristics.

3 MODY link nurse (MLN) training every four months includes detailed information about the different types of MODY, sessions on genetic counselling and discussion of patient cases.

diagnosed under the age of 25 in at least one family member.

- Non-insulin dependent diabetes, defined by treatment without insulin for five years or a measurable C-peptide.
- Autosomal-dominant inheritance, with diabetes being passed from an affected individual in one generation to the next. It can usually be traced back through two to three generations (Hattersley, 1998).

Different types of MODY have distinct clinical characteristics. Patients with mutations in the glucokinase (GCK) gene have slightly raised hyperglycaemia from birth. It remains stable throughout life, and complications are rare in this group.

In contrast, patients with MODY due to a mutation in the HNF-1α gene are born with normal glucose tolerance but usually develop diabetes around adolescence. At onset they are often mistaken to have type 1 diabetes. This is due to the presence of hyperglycaemia in slim young adults combined with professionals' unfamiliarity with the importance of family history. Individuals with diabetes due to an HNF-1α mutation are prone to complications; particularly retinopathy (Stride and Hattersley, 2002). However they are usually sensitive to small doses of sulphonylureas (Pearson et al, 2000). If the diagnosis of HNF-1α MODY is confirmed by genetic testing, then providing the patient has not progressed through sulphonylureas to insulin treatment previously, they may be able to stop insulin injections and improve their glycaemic control with a low-dose sulphonylurea (Pearson et al, 2000).

The other types of MODY are rare. HNF-4α MODY has a similar presentation to HNF-1α MODY and HNF-4α patients are also sensitive to sulphonylureas (Pearson, personal communication). HNF-1α mutations (also described as renal cysts and diabetes or RCAD) again affects only a small proportion (approximately 3%) of MODY patients. These patients may well be identified in renal clinics as they usually present with variable renal histology and may progress to developing diabetes later (Bingham et al, 2001).

Figure 1. Locations of the MODY link nurses.

MLN training in Exeter

The MLNs attend an initial three-day course in Exeter. This includes a description of the MLN role, the different types of MODY, a visit to the molecular genetics lab and details of the process of genetic testing, how to take family histories, sessions on genetic counselling and discussion of patient cases. Study days are attended for one-and-a-half-days every four months. During each visit to Exeter the MLNs are expected to present an interesting case history to the rest of the MLNs, and further information and in-depth knowledge is gained about MODY and other genetic forms of diabetes. The Exeter team provide an information and support network to all of the MLNs throughout the year.

The role of a MLN

Each MLN is already an experienced DSN and, with the additional ongoing training from the team in Exeter, has been able to take on the increased role and responsibilities required. The MLNs are seconded from their Trust to work for 3.5 hours a week on the project. Initially, the majority of the time has been spent visiting other diabetes centres, as well as renal and obstetric units that have

been allocated within the MLN's region, giving presentations about MODY and other genetic forms of diabetes and providing details of the genetic tests available.

The MLN's role is also to provide a link for patients and professionals with the Exeter team. MLNs take referrals from local clinicians and may advise on the genetic tests that would be appropriate. They also support the patient and their families by providing genetic counselling before, during and after genetic testing. The results of the tests not only have implications for the patient but also for their family and may mean that other members of the family not known to have diabetes may request a predictive genetic test. The MLNs can arrange predictive tests for adults, but requests for predictive tests in young children are referred to local clinical genetics services due to the ethical issues involved.

Identification of a mutation within one of the MODY genes may result in a change of treatment for some patients. For example, someone with a GCK mutation will not need treatment for their hyperglycaemia. Patients with an HNF-1α mutation may be able to discontinue their current insulin regimen in certain circumstances and commence sulphonylurea treatment with appropriate support and guidance from their local MLN.

The benefits of being a MLN

The MLN role has provided the opportunity to learn new skills and increase knowledge. The MLNs are able to apply their vast experience of caring for patients with diabetes and their newly acquired knowledge of genetics in the post-genomic era not only in MODY but in other genetic forms of diabetes including maternally-inherited diabetes and deafness, permanent neonatal diabetes, transient neonatal diabetes, and familial partial lipodystrophy. The training has also raised awareness amongst the MLNs of many issues surrounding the diagnosis of diabetes and the appropriate use of biochemical and immunological tests (for example measurement of C-peptide and glutamic acid decarboxylase [GAD] autoantibodies) that have implications for their general clinical practice. Networking with the other MLNs between the training sessions in Exeter ensures mutual support for one another and has prevented isolation of the role. Following up families with MODY who live in other parts of the UK encourages communication between the MLNs. It has also allowed them the opportunity to exchange ideas and share other areas of their practice not necessarily associated with MODY.

The Exeter genetics of diabetes team includes clinicians, nurses and scientists from both a service and research background. They provide support by regular phone and e-mail contact, which has been invaluable as a way of discussing individual clinical cases with the MLNs. The team in Exeter will also endeavour to assist with other problems encountered in the role, such as funding for genetic tests.

Meeting with other diabetes teams while giving presentations has given MLNs the opportunity to forge new professional links, enabling other diabetes teams to use the MLNs as regional experts. MLNs have also developed skills and expertise in genetic counselling and frequently visit families in their region who are thought may have MODY to discuss the issues both on an individual level and with their wider family.

This new role has encouraged the MLNs to further their personal and professional development. Specific examples include:

- being involved in setting up and running a specialist MODY clinic locally
- a poster presentation at Diabetes UK evaluating the MLN project in South East England
- successful application to the Scottish Executive for funding of MLNs in Scotland
- writing for publication for the first time
- presentations about MODY at both regional and national conferences.

Teaching and presentation skills have also been developed as part of the role and all these facets have helped each MLN to develop both personally and professionally.

As MLNs have been appointed in successive phases, those already in post have acted as mentors for newly appointed MLNs. This has involved mentoring a MLN, usually in a locality near by, to give support by taking the new MLNs out on home visits to possible MODY families or when giving presentations to hospital teams about MODY. This has increased the confidence of

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1 MLNs initially spend most of their time visiting other diabetes centres, and renal and obstetric units giving presentations about MODY, other genetic forms of diabetes and genetic testing.

2 MLNs provide a link for patients and professionals with the Exeter diabetes and genetics team.

3 They support patients and their families by providing genetic counselling as appropriate.

4 The role provides the opportunity to learn new skills, increase knowledge, forge new professional links, and develop personally and professionally.

5 The Exeter genetics of diabetes team provide support and give advice regarding individual cases.

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1 Those already in post have acted as mentors for newly appointed MLNs.

2 Patient referrals to MLNs come from a variety of sources, usually following a MLN presentation.

3 The Exeter website (www.diabetesgenes.org) provides details about MODY, genetic testing and MLN location.

4 The DoH is encouraging initiatives such as the MLN project. It is hoped to secure funding for MLN posts in Wales and Ireland.

5 The success of this project could provide a model for DSNs to extend their role by learning new skills in other key areas.

not only those being mentored but also those mentoring, and provides an additional point of support for the new MLN.

MLN presentations within the authors' allocated regions

Heather McMahon has given 15 talks to the diabetes teams in her locality in a 14-month period, including one to a group of practice nurses with a special interest in diabetes. The number attending these presentations totalled 201 medical and nursing staff.

Sandra Dudding has given 18 talks to diabetes teams in the Yorkshire region in a 13-month period. Two of these were to a group of practice nurses who run mini clinics at their surgery. Sandra has given a presentation about MODY at the UK association of DSNs, and another with Professor Andrew Hattersley at a regional paediatric conference in Leeds.

Both MLNs have further talks planned at both local, regional and national level.

Patient referrals

Patient referrals are received from a variety of sources including consultants, medical staff and other DSNs, usually following a presentation from the MLN. Healthcare professionals may access the Exeter website (www.diabetesgenes.org), which provides details about MODY and genetic testing and provides information about the location of the MLNs.

Examples of referrals

Since commencing in post in May 2003, Heather has had 14 referrals from 10 different sources, one of which was a three-week-old baby with diabetes, which was later confirmed as transient neonatal diabetes. Four of the referrals were regarding patients that have already had a positive genetic test for MODY but further support and testing was requested for other members of the family.

Sandra has had 25 referrals from eight different sources. One of these patients was confirmed on genetic testing to have HNF-1α MODY, which led to two other members of their family also having HNF-1α MODY confirmed. Consequently, two members of this family have successfully been able to discontinue insulin, and this process has been closely supervised by the MLN. Sandra has also been involved in diagnosing a woman with GCK during pregnancy, enabling her to manage her pregnancy without insulin injections. Six of the referrals received by Sandra were found to be inappropriate for genetic testing as they were more likely to have type 2 diabetes.

Conclusion

The DoH is encouraging initiatives such as the MLN project that can bring the benefits of genetics into mainstream clinical areas. The successful approach of training specialist nurses in genetics could provide a model for disseminating genetic information in other genetic disorders. It could also provide a model for DSNs to extend their role by learning new skills in other key areas.

The MLN posts have provided experienced DSNs with the possibility of expanding their role across new boundaries into the field of genetics and encouraged the possibilities of personal and professional development and networking, both locally and nationally. It is hoped to secure funding to appoint MLNs for Wales and Ireland in the future.

If you would like to contact your local MLN to discuss individual patients or to request a presentation to your diabetes team, please contact your nearest MLN as indicated on the website www.diabetesgenes.org. ■

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ACKNOWLEDGMENTS

We are very grateful to the Department of Health for funding this project.