Integration of the MODY link nurse project: 20-month evaluation

Maggie Shepherd, Andrew Hattersley and Sian Ellard

Introduction

The MODY link nurse project is an educational initiative funded by the Department of Health to develop the integration of new genetic knowledge into diabetes care. MODY (maturity-onset diabetes of the young) link nurses (MLNs) are experienced diabetes specialist nurses based throughout the UK who are seconded to the project for three-and-a-half hours per week. They receive ongoing training in Exeter to learn about the genetics of diabetes, genetic counselling and genetic testing. The MLNs disseminate this new genetic information to healthcare professionals in their allocated areas, assess patients with a possible diagnosis of MODY and provide support for known MODY families. This paper evaluates the introduction and impact of the first 12 MLNs in the UK after 20 months.

The Department of Health (DoH) is encouraging initiatives to bring the benefits of recent advances in genetic knowledge into mainstream clinical areas, particularly using the skills of specialist nurses (DoH, 2003). The MODY (maturity-onset diabetes of the young) link nurse (MLN) project commenced in 2002 (Shepherd, 2003a) with the appointment of six MLNs. Additional DoH funding has enabled the recruitment of a further 11 nurses in three phased intakes.

The MLNs aim to heighten awareness and aid recognition of MODY in their local areas, leading to an increase in the number of patients receiving an accurate diagnosis and the most appropriate treatment. This is important as the identification of six genes in which mutations cause MODY has allowed the genetic cause of diabetes to be identified in >80% of families who meet clinical criteria for MODY (Owen et al, 2002). In these cases a clinical diagnosis may be confirmed by molecular genetic testing.

A diagnostic service is provided by the Molecular Genetics Laboratory in Exeter for patients throughout the UK. A positive result may lead to a patient being able to stop insulin treatment and achieve good glycaemic control on sulphonylureas (Shepherd, 2003b). The diagnosis of a dominantly inherited form of diabetes has implications for other family members, who are at 50% risk of inheriting the affected gene and subsequently developing diabetes. Predictive genetic testing for these relatives will provide reassurance (for those who test negative) or may facilitate an early diagnosis (for those who have inherited the affected gene), enabling appropriate treatment and a reduced risk of diabetic complications. However, diabetes professionals are often unfamiliar with the key characteristics of monogenic forms of diabetes and misdiagnosis as type 1 or type 2 diabetes is common (Hathout et al, 1999; Lambert et al, 2003; Lehto et al, 1997; Moller et al, 1998).

Aim

The aim of this study was to evaluate the UK MODY link nurse project after an initial 20-month period.

Methods

The project has been evaluated in three key areas:

- the understanding and confidence of the MLNs in undertaking their new role in genetics
- MLN presentations to healthcare professionals

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1 This paper evaluates the introduction and impact of the first 12 UK MODY link nurses (MLNs) after 20 months.

2 The project is evaluated in three key areas: professional development, MLN presentations and genetic tests performed.

3 MLNs have increased knowledge of monogenic diabetes with 112 highly evaluated presentations and provided local support for families.

4 The MLNs have influenced referrals for genetic testing with an increase in the number of UK patients receiving a molecular genetic diagnosis of MODY.

5 This project offers a successful model for the integration of genetic information into practice.

KEY WORDS

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

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1 MLNs completed questionnaires at the start of their secondment and after one year to evaluate their increased understanding of genetics.

2 They were also given ten case studies at the start and after a year and asked to indicate the most likely type of diabetes.

3 All MLNs were allocated 10–15 hospitals in their area to contact and offer to give presentations about MODY and genetic testing and assist in identifying patients with possible MODY.

4 The number of UK patient referrals for genetic testing and positive tests were assessed before and during the project phases.

- molecular genetic testing.

The understanding and confidence of the MLNs in undertaking their new role in genetics

In order to evaluate their development and increasing understanding of genetics, the MLNs completed questionnaires at the start of their secondment and after one year in post. The aim was to assess their understanding of genetic terms and confidence in performing various activities relating to their role, e.g. drawing family trees and giving talks on MODY. They were also given ten case studies (at the start and after one year) and asked to indicate the most likely type of diabetes in each case, whether they would recommend genetic testing for those patients, and which genetic test would be most appropriate.

MLN presentations to healthcare professionals

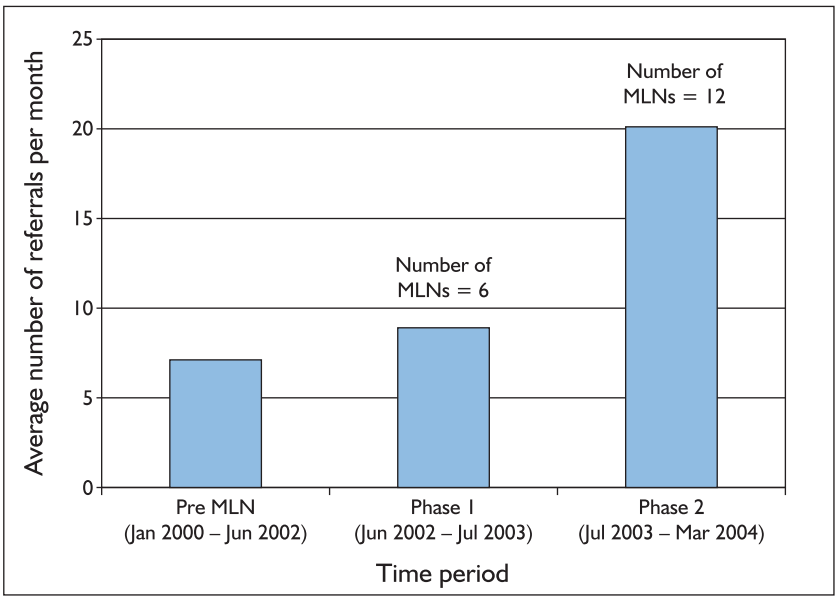
All MLNs were allocated 10–15 hospitals in their area to contact and offer:

- to give presentations about MODY and genetic testing and
- to assist in the identification of patients with a possible diagnosis of MODY.

The MLNs were given a template Microsoft PowerPoint presentation that included a description of the different genetic subtypes of MODY and how to differentiate MODY from other types of diabetes. They were also provided with copies of other presentations given at the Exeter study days that included examples of case studies and patients' perceptions of genetic information. The MLNs were then encouraged to tailor these presentations to suit the audience they were addressing.

Presentations given by the MLNs to healthcare professionals in their areas were evaluated by the attendees using standardised forms that asked them to indicate on a Likert scale (where one is poor and five is excellent) the educational value and content of the session, the presentation in terms of the delivery of information, the opportunity to ask questions and to rate overall how useful they found the session. Space was provided for additional comments. The MLNs also complete a feedback form following each presentation that highlights the numbers and job titles of staff seen, the form of the presentation, the perceived response, the future plan of action and any difficulties or problems encountered.

Figure 1. Average number of patients referred for genetic testing each month before and during the MODY link nurse project



Molecular genetic testing

The number of UK patient referrals from regions with/without MLNs was assessed:

- before the MLNs started in post (i.e. from the introduction of the diagnostic testing service in January 2000 until June 2002)
- during the first phase of the project (June 2002 to July 2003 when there were six MLNs), and
- during the second phase (July 2003 to March 2004 when there were 12 MLNs in post).

The number of positive tests from patients with a possible diagnosis of MODY and the number of samples received from families known to have MODY was also compared between MLN regions and non-MLN regions during these time periods. Test requests originating from Exeter were excluded from this analysis due to the local expertise and possibility of bias. The total number of UK patients who had a positive

test, and hence a genetic diagnosis of MODY, was also calculated.

Results

The understanding and confidence of the MLNs in undertaking their new role in genetics

Before attending their first training session in Exeter, <1% of the genetic terms described were understood by the MLNs and could be explained to others. After one year MLNs felt they understood and could explain 70% of the genetic terms to others.

At the start of their secondment MLNs only felt 'very confident' performing 8.5% of tasks relating to the role (e.g. drawing family trees) but this rose to 63% of tasks after one year. They initially felt 'confident or very confident' performing 48.5% of tasks relating to their role but after one year this increased to 91%.

For the hypothetical cases given to the MLNs at the start of the project, 40% of cases were correctly identified as having possible MODY (although the subtype of MODY was not identified). At one year, 97% of hypothetical cases were correctly identified as having MODY and the subtype of MODY was also accurately identified in 85% of cases.

MLN presentations to healthcare professionals

During the first 20 months of the project a total of 112 presentations were given at over 80 different hospitals throughout the UK. Many of these presentations were at multidisciplinary diabetes team meetings, although others were at much larger regional meetings. The audiences included consultant diabetologists, paediatricians, DSNs, dietitians, GPs and practice nurses. Ninety per cent of the presentations were rated by participants as 'very good' or 'excellent'.

Participants were encouraged to add individual comments to the feedback forms and diabetes teams indicated that they particularly valued information regarding:

- the difference between MODY and young-onset type 2 – 'I now have a good understanding of MODY compared to other types of diabetes'
- the importance of detailed family histories 'raised my awareness significantly in this area'
- the implications of genetic testing for clinical management, where it was 'interesting to understand the different treatments required', and 'informative – will alter practice'.

The presentations also prompted participants to identify patients who may have MODY, e.g. 'brought to mind a family who may fit MODY criteria'.

Others suggested the information about MODY was new to them, for example one person stated that it was the 'first time to have the opportunity to learn more about MODY – excellent'.

MLN feedback regarding the presentations showed that their local teams had a varied range of awareness of MODY and highlighted the perception that the costs of genetic tests may be a barrier to testing. Some centres requested that the MLN become a regular speaker at their meetings and in many cases the MLNs returned to discuss individual cases.

Molecular genetic testing

The number of UK patients referred for testing increased from an average of 7.1 per month before the project started in June 2002 to 20.1 per month during phase 2 of the project (*Figure 1*). There was only a small increase during phase 1 (to 8.8

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1 Understanding of genetic terms increased from <1% before training to 70% after one year in post.

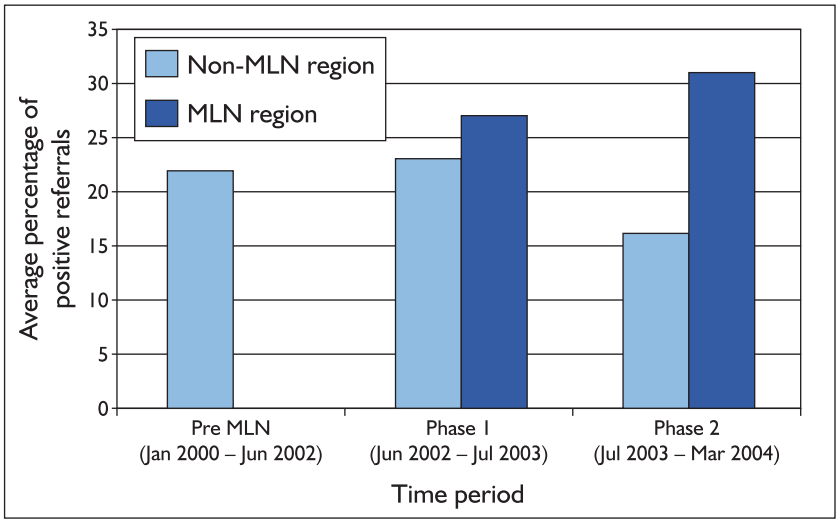
2 There was a increase in percentage of the role-related tasks MLNs felt 'confident' in carrying out from 48.5% before training to 91% after a year in post.

3 Forty percent of hypothetical cases were correctly identified as MODY at the start and increased to 97% after one year.

4 A total of 112 presentations were given in the first 20 months of the project to a wide range of healthcare professionals and were highly evaluated.

5 Numbers of patients referred for testing increased from an average of 7.1 per month before the project to 20.1 per month during phase 2.

Figure 2. Percentage of positive tests for patients with a posible diagnosis of MODY (index cases).



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1 The number of positive tests were higher in areas with MLNs compared to those without.

2 Following the introduction of MLNs, there has been a significant increase in the number of UK patients receiving a genetic diagnosis of MODY as a result of a positive molecular genetic test.

3 Training experienced diabetes specialist nurses to become regional MLNs has been successful.

4 Presentations given by MLNs to healthcare professionals in their areas have been well received and resulted in an increased number of patients referred for molecular genetic testing.

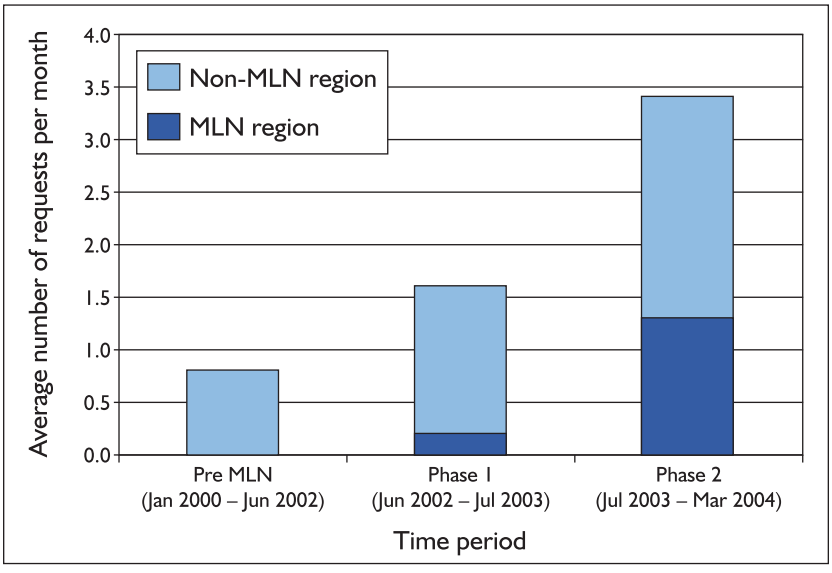
referrals per month) and this is likely to reflect a 'lead-in' period when the MLNs underwent training, made initial contact with their local teams, started giving presentations and assessed potential referrals.

The percentage of positive tests for patients with a possible diagnosis of MODY (index cases) prior to the project was 22% (43/194 tests). This increased to 25% during phase 1 (23/93 tests positive) and phase 2 (29/115 tests positive) of the project. The number of positive tests was higher in areas with MLNs (27% and 31% for phases 1 and 2) compared to those without (23% and 16%, as shown in *Figure 2*).

Between January 2000 and March 2004, diagnostic molecular genetic testing identified a mutation in one of the MODY genes in 95 index cases (out of a total of 402 patients tested). A further 68 requests for testing were received from 43 families in whom a mutation had been found (either through diagnostic testing or previous research studies).

Figure 3 shows the number of requests for testing relatives of patients known to have MODY. These include both affected family members seeking testing to confirm that they also have MODY and asymptomatic relatives at risk of having inherited a MODY mutation and hence developing diabetes in the future. Only 23 requests for testing were received during the 29-month period prior to the appointment of the MLNs (referral rate 0.8 per month) but this increased to 21 requests during phase 1 (1.6 per month) and 24 during phase 2 (3.4 per month). The increase in the number of tests for family members reflects both an increase in requests from non-MLN and MLN regions.

Figure 3. Average number of requests per month for testing relatives of patients known to have MODY.



Following the introduction of the MLN project there has been a significant increase in the number of UK patients receiving a genetic diagnosis of MODY as a result of a positive molecular genetic test. In the 29 months preceding the MLN project there were 70 positive tests. In the first 20 months of the project there have been 100 positive tests. This is an increase from 2.4 per month pre-introduction of MLNs to 3.5 per month in phase 1 and 7.7 per month in phase 2 (*Figure 4*).

Discussion

This interim assessment has established that the training of experienced diabetes specialist nurses to become regional MLNs has been successful. It therefore offers a model for the integration of new genetic knowledge into clinical care for other conditions and for the dissemination of new information within diabetes care. We have shown that the training has resulted in highly evaluated presentations, improved knowledge and confidence for the MLNs and an increased number of patients with a confirmed diagnosis of an inherited form of diabetes in the UK.

The presentations given by the MLNs to healthcare professionals in their areas have been well received and resulted in an increased number of patients referred for molecular genetic testing. This approach is needed as information on genetic subtypes is not routinely taught and recommended textbooks are inevitably out of date in a rapidly changing field like the genetics of diabetes. There are considerable advantages in small local meetings that involve multidisciplinary staff and allow considerable time for discussion compared to large lectures at national

and international meetings that are attended mostly by senior staff.

Feedback from these local meetings also highlighted concerns about genetic testing. The most common concern raised by a number of teams was the cost of testing and the availability of funding. Molecular genetic tests are more costly than biochemical or immunological tests, but a positive test may be considered cost-effective in terms of treatment options (reduced usage of insulin) and/or reduced likelihood of complications due to early diagnosis and appropriate treatment. Although it has been agreed by specialist commissioners that testing for MODY should be available for all patients in the UK who meet clinical criteria for testing (see www.diabetesgenes.org, accessed 17.02.05), funding streams are not clearly defined. While a centrally-funded laboratory service to test patients according to strict clinical criteria would be an attractive model for diabetes healthcare professionals, this is not an approach that fits current NHS financial flows.

The number of patients receiving a genetic diagnosis of MODY each month has tripled since the MLN project started. This increase in positive tests is mostly due to increased numbers of tests being performed, but the proportion of positive test results for patients with a possible diagnosis of MODY showed a small increase and was higher in areas with MLNs compared to those without. This suggests that the MLNs are contributing to an increase in the number of families with a definitive diagnosis of MODY, both through greater numbers of patients tested but also through a higher pick-up rate.

If a patient is found to have a mutation in one of the MODY genes, testing their relatives is straightforward and relatively inexpensive. One of the MLN roles is to offer genetic counselling for adult relatives who may be interested in a genetic test either to confirm that they also have MODY or to determine whether they have inherited the familial mutation and therefore have a high risk of developing diabetes in the future. Children whose parents seek predictive genetic testing on their behalf are referred to their local clinical genetics teams. The number of tests for family members has increased since the inception of the project and the network of MLNs throughout the UK facilitates testing for relatives who may reside in different parts of the country.

Patients with HNF-1α or HNF-4α MODY are sensitive to sulphonylureas and may achieve good glycaemic control on low doses. Stopping insulin, sometimes after many years, represents a major change in management (Shepherd, 2003c). The MLNs have been able to provide individual support for a total of seven patients during their transfer from insulin to sulphonylureas during the initial 20 months of the project.

Our previous studies had shown a lack of understanding of genetics by diabetes healthcare professionals (Shepherd, 2000) and this was confirmed by the initial evaluation of the MLNs' knowledge of genetic terms and inherited forms of diabetes. However, after one year in post, MLNs were confident about explaining the majority of genetics terms and correctly diagnosed 97% of hypothetical cases. The MLNs receive ongoing training every four months to further increase their genetics knowledge and they maintain close contact with the team in Exeter to seek

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1 MLNs are contributing to an increase in the number of families with a definitive diagnosis of MODY both through greater numbers of patients being tested but also through a higher pick-up rate.

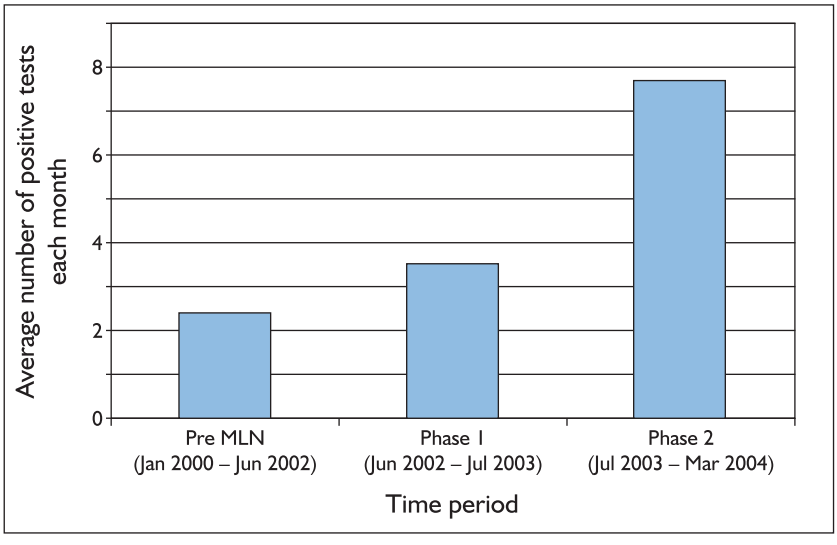
2 If a patient is found to have a mutation in one of the MODY genes, testing their relatives is straightforward and relatively inexpensive.

3 MLNs receive ongoing training every four months to further increase their genetics knowledge.

4 Diabetes teams have been keen to receive information about MODY from MLNs and to identify patients who would benefit from genetic testing.

5 MLNs have successfully increased awareness of MODY and provided local support.

Figure 4. Average number of positive tests (mutation screening of index cases and tests for family members) each month.



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1 The role of MLN has offered diabetes specialist nurses the exciting opportunity to develop their role and increase their understanding of the genetics of diabetes and the genetic tests available.

2 This approach may provide a useful model for the dissemination of genetic information in other conditions.

advice whenever required (Dudding et al, 2005).

Conclusion

During the initial 20 months of the MLN project, a group of 12 diabetes specialist nurses have already demonstrated a very clear increase in knowledge regarding genetic testing. Diabetes teams have been keen to receive information about MODY from the MLNs and to identify patients who would benefit from genetic testing. The MLNs have successfully increased awareness of MODY and provided local support for both professionals and MODY families. They have helped to identify families previously misdiagnosed as having type 1 or type 2 diabetes and have supported patients transferring from insulin to sulphonylureas.

The role of MLN has offered diabetes specialist nurses the exciting opportunity to develop their role and increase their understanding of the genetics of diabetes and the genetic tests available. Being seconded as a MLN has fulfiled the possibility of continuing their existing role while learning new skills and expanding knowledge (Dudding et al, 2005). 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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