Monogenic diabetes: information seeking and genetic testing access via e-mail

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Abstract

Background: Confirmation of monogenic diabetes by molecular genetic testing has allowed many patients, often previously assumed to have type 1 diabetes, to transfer from insulin injections to sulphonylurea tablets, with improvements in glycaemic control and quality of life: www.diabetesgenes.org provides information about monogenic diabetes and genetic testing.
Aim: To investigate key issues raised by individuals who e-mailed the monogenic diabetes team about genetic testing and monogenic diabetes management.
Methods: Sixty e-mail enquiries, received over a six-month period from patients and professionals worldwide, were analysed using a qualitative thematic content approach.
Results: Five themes emerged: 1. Accessing genetic technology: patients and professionals both enquired about access to testing; 2. Presentation of evidence: medical facts presented by patients and professionals included characteristics specifically relevant to diagnosing monogenic diabetes; 3. Experiences of healthcare: patients often researched their condition online and some felt dissatisfied with routine consultations; 4. Seeking specialist advice regarding treatment: specific information was sought relating to management of neonatal diabetes or monogenic diabetes and pregnancy; 5. Searching for a cure through genetic technology: patients questioned whether genetic advances would lead to a cure for diabetes.
Conclusion: This project offers the first insights into use of e-mail as a means of gaining access to a specialist monogenic team and information about genetic testing. Although providing advice via e-mail can prove complicated, particularly when received from patients under the care of other clinicians, it is an efficient means of communicating specialist knowledge. Study findings will aid development of a 'frequently asked questions' section of www.diabetesgenes.org.

Key words

Monogenic diabetes; genetic testing; information seeking; e-mail enquiry

Introduction

The possibility of genetic testing has raised expectations among patients with a range of conditions who seek information about the cause of their disease. However, the introduction of genetic technology provides challenges for professionals and patients and may contradict previous understanding of disease and treatment.[1] This is the case for many individuals with monogenic diabetes, who are often assumed to have type 1 diabetes prior to genetic testing. The benefits of genetic technology in diabetes can be extremely positive:[2–5] the confirmation of monogenic diabetes by genetic testing allows many patients, previously assumed to have type 1 diabetes and treated with insulin, to transfer to sulphonylurea tablets, leading to improvements in glycaemic control and quality of life.[6–8] This includes patients diagnosed in the neonatal period[9–15] and those with *HNF1A, HNF4A* or *Glucokinase* maturity onset diabetes of the young (MODY).[16–19]

Using the internet as an educational resource allows patients and healthcare professionals (HCPs) to access the same information about monogenic diabetes and genetic testing. Patients' use of the internet for retrieving health information is escalating, but it can be difficult to identify how they use it.[20,21]

The Royal Devon and Exeter NHS Foundation Trust is the UK referral centre for genetic testing in diabetes and receives samples from all over the world. Its website, www.diabetesgenes.org, which was developed in 2001 provides information on monogenic diabetes and genetic testing, and has had over 72,000 'hits'. The Exeter monogenic diabetes team receives e-mail enquiries through this website from patients and professionals worldwide. This paper provides the first insights into the issues raised by patients and professionals through e-mail communication regarding genetic testing in diabetes.

Aims

To assess why individuals e-mail members of the monogenic diabetes team with queries relating to monogenic diabetes and genetic testing, and to identify the characteristics of those sending enquiries.

Methods

This project was a systematic analysis of e-mail enquiries from individuals across the world to the Exeter monogenic diabetes team regarding the possibility of genetic testing or the

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Received: 20 January 2010
Accepted in revised form: 23 March 2010

management of individuals already known to have monogenic diabetes. Sampling ensured variation and included enquiries from patients and professionals. All queries were anonymised, but codes were used to identify whether the query originated from a patient or professional, and their country of origin. E-mails sent to members of the monogenic diabetes team within a six-month period were identified.

A qualitative thematic content analysis was used to identify key themes and issues arising from the e-mails, including identification of what patients and professionals wanted to know about monogenic diabetes or genetic testing. To enable easier analysis of quantitative data, codes rather than free-text fields were used to classify data such as country of origin and whether the enquirer was a patient or a HCP.

This study was approved by the North and East Devon Research Ethics Committee. Individuals were e-mailed to ask for their consent to analyse the contents of their original e-mail and to publish the findings in anonymised form. All e-mails were responded to by the Exeter monogenic diabetes team.

Results

Sixty e-mail enquiries received within the specified six months were selected; they all related to cases of monogenic diabetes or genetic testing. Enquiries originated from at least 15 countries, with a substantial proportion from the UK and USA (Table 1). Similar numbers of enquiries were received from patients 28 (47%) and HCPs 32, (53%), with the vast majority of professionals being doctors (n=28, 47%); only two (3%) enquiries were from nurses. The average enquiry was approximately 200 words long.

Participants were asked how they found the diabetes genes website: 12 (20%) had visited the site previously; 9 (15%) found it through a search engine; 9 (15%) followed a link from the International Study of Paediatric and Adolescent Diabetes (ISPAD) website; 7 (11.6%) knew a member of the Exeter diabetes team and e-mailed them directly; 6 (10%) heard about the website during a conference presentation by a member of the Exeter diabetes team; 5 (8.3%) had been referred to the site by their doctor; 5 (8.3%) had read about the website in a magazine; 8 (13%) were unknown. Five key themes were identified:

Country	E-mails analysed, n (%)
UK	19 (31.6)
USA	16 (26.6)
Canada	2 (3.3)
India	2 (3.3)
New Zealand	2 (3.3)
Sweden	2 (3.3)
Australia	1 (1.6)
Croatia	1 (1.6)
France	1 (1.6)
Germany	1 (1.6)
Italy	1 (1.6)
Luxemburg	1 (1.6)
Mexico	1 (1.6)
Trinidad & Tobago	1 (1.6)
UAE	1 (1.6)
Unknown	8 (13.3)

Table 1. Countries of origin for e-mail enquiries to diabetesgenes.org, concerning genetic testing or monogenic diabetes (n=60, 6-month analysis)

1. Accessing genetic testing technology

Both patients and HCPs used e-mail as a means of enquiring directly about access to testing:

"I request your opinion regarding the possibility of doing a genetic analysis." (HCP)

"I think I have a high probability of being MODY, can we test for it ?"(Patient)

The majority of enquiries 28 (46%) were considered likely to have a monogenic cause of diabetes from the information provided, and directly asked about genetic testing. In these cases, further information was provided by the Exeter team including details of the blood sample required to enable genetic testing to be performed. In eight (13%) enquiries further details were required to judge if genetic testing was appropriate, and further e-mail dialogue ensued. This often included requests for additional clinical information and other non-genetic test results (*eg* glutamic acid decarboxylase antibodies, C-peptide) prior to genetic testing. In seven cases (12%), the information provided suggested that a monogenic cause of diabetes was unlikely; reasons for this were explained and genetic testing was not recommended. In 12 cases (20%), the enquiries related to individuals who were already confirmed to have monogenic diabetes, who requested specialist advice regarding their management.

2. Presentation of evidence

Of interest were the ways in which patients presented diabetes information. Medical terminology was used in a similar manner by patients and professionals. Details included characteristics specifically relevant to a monogenic diabetes diagnosis, such as age at diagnosis and family history:

"She is 19 years old and presented with diabetes at 14…paternal grandfather, paternal grandmother and an uncle all have diabetes…she has done extremely well on insulin 18 units morning, 8 units evening." (HCP)

"I am 29. In 1998 (aged 20) I was diagnosed with type 1 based on a fasting sugar of 7.8...I was put straight onto insulin… 4 units of Mixtard per day… My brother is 27 and was diagnosed this week…My father and paternal grandmother have type 2."(Patient)

Information from patients and professionals included key characteristics relating to monogenic diabetes, such as age at diagnosis, family history of diabetes and current or previous treatment. The presentation of

this evidence indicates that patients are 'credible knowers' and their queries were considered to be legitimate. These information seekers have gleaned what medical information is considered important from personal experience. However, this use of 'medical terminology' and the level of information provided may also reflect the selection bias of those most likely to access websites and e-mail HCPs directly. Responses to these enquiries contained similar information for both patients and professionals, including the attachment of published articles that provided additional details, where appropriate.

3. Experiences of healthcare and interactions between patients and HCPs

Patients with monogenic diabetes frequently try to make sense of their condition, which they often consider to be different. A proportion of patients (number unknown) use the internet to research their diabetes, to equip themselves with information about the possibility of genetic testing and treatment change. Six (10%) enquirers questioned professionals' comprehension of their condition and were clearly seeking answers about the nature of their diabetes, for example:

"I felt 'fobbed off' when asking my diabetic consultant why my diabetes is atypical. The two consultants I broached the issue of MODY with seemed reluctant to talk about it, and annoyed I had done my own research." (Patient)

Searching for additional knowledge, particularly regarding conditions deemed to be rare, may affect the power balance within healthcare consultations, for example:

"The consultant/diabetic nurses I see don't seem particularly interested in scientific advances but I would like to know the facts." (Patient)

Although a minority of enquirers felt unhappy with discussions about their type of diabetes with their clinical team, in other cases HCPs clearly communicated their thoughts:

"I have shared with her my concern that she might actually have type 2 diabetes and whether we should be allowing her a trial off insulin." (HCP)

A desire to clarify the diagnosis and most appropriate treatment was considered helpful by patients and professionals:

"I think to make the diagnosis would rule out uncertainty and the mother is clearly keen for this." (HCP)

Communication within the healthcare consultation can impact on the effectiveness of the consultation and may affect patients' perceptions of professionals' credibility, particularly when dealing with rare conditions such as monogenic diabetes.

4. Seeking specialist advice regarding treatment

Professionals frequently sought specific information about the clinical management of patients. Twelve enquiries (20%) related to individuals with confirmed monogenic diabetes (*ie* the patients had undergone molecular genetic testing and received a positive result), and the majority of these were seeking treatment advice. Twelve of 60 queries (20%) related to neonatal diabetes management, where the option of sulphonylurea treatment was relatively new:

"How much did the smallest infant weigh who was treated with sulphonylureas? This baby is <1kg so I am anxious about oral agents as first line treatment." (HCP)

"'Should I be concerned about starting my son on a new treatment at such a young age? Have you ever treated anyone as young as him before?" (Mother of patient with neonatal diabetes)

These enquiries frequently related to a change from insulin to the use of sulphonylureas as the result of a genetic diagnosis. This was particularly of concern to those caring for babies with neonatal diabetes (who had no prior experience of sulphonylurea use in young children) or individuals assumed to have type 1 diabetes, who had previously considered insulin necessary for survival. In both cases, sulphonlyurea use challenged their experiences and beliefs, and the reassurance of a specialist team was sought.

5. Searching for a cure for diabetes through genetic technology

The impact of diabetes on individuals and families led many to hope for improved treatments and ultimately a cure. 'Science hype' relating to genetics and the potential benefits it might offer (frequently reported in the media) were also reflected in patients' information-seeking behaviours:

"I was told this form of diabetes cannot be cured even by stem cell advances." (Patient)

Perceptions of the possible implications of research into genetics may have led to hopes that are presently unrealistic:

"All we can do is hope for a miracle for this heart-breaking disease." (patient)

Eight enquiries (13%) came from individuals considered unlikely to have monogenic diabetes, including patients with type 1 diabetes who had heard about the possibility of transferring from insulin to oral therapy and were hoping that this applied to them.

Discussion

E-mail provides patients and professionals with access to specialist teams and information about genetic testing. Such communication enhances chronic disease management,[22] is convenient and efficient,[23,24] and some patients may feel more able to ask questions via e-mail compared with face-to-face communication.[24] However, providing advice via e-mail can prove difficult, particularly when received directly from patients who are under the care of other clinicians.

In this study, advice was given to patients that could be discussed with their own clinical team, who would be required to refer them for genetic testing or prescribe treatment.

Previously, concerns have been raised about patients' ability to use e-mail appropriately,[25] but this was not considered problematic in our study. E-mail enables communication between patients and professionals at a time convenient to both groups.[26] Indeed, in this study, e-mails facilitated a rapid response that is rarely available within a traditional healthcare system: the majority of e-mails were answered within 48 hours of receipt. However, the issue of reimbursement for online medical communication has previously been highlighted in US studies.[22,27] The time spent by the Exeter monogenic team responding to e-mails is not currently quantified and as such is a service with little recognition.

There are some limitations to this study, such as sampling limitations and the wide variety of questions that were e-mailed. However, although our research analysed e-mails over a specific time period there is no indication that the issues raised varied from those received on a regular basis.

Conclusion

Our study provides the first insights into the use of e-mail as a means of gaining access to a specialist team and information on genetic testing in diabetes. E-mail communication allowed rapid access to a team specialising in a rare form of diabetes, and advice was sought regarding both general and specific issues. It is envisaged that such enquiries will continue to increase as use of the internet and awareness of genetic testing in diabetes escalate. Ways to address recognition of this service could be considered. Findings from this study will be used to improve the information provided on www.diabetesgenes.org, by developing a 'frequently asked questions' section.

Acknowledgements

MS is supported by an NIHR post doctoral fellowship and by the Peninsula NIHR Clinical Research Facility.

Conflicts of interest

None.

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