Prioritization of future genetics education for general practitioners: a Delphi study

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INTRODUCTION

It has been argued that the greatest public health benefit of advances in understanding the human genome may be realized for common chronic diseases such as cardiovascular disease, diabetes mellitus, and cancer.\(^1\) International attempts to integrate such knowledge into clinical practice are still in the early stages, and as a result, many questions surround the current state of this translation.\(^1\)–\(^3\) Physicians often lack the knowledge of genetics relevant for daily practice,\(^4\) lack the ability to oversee genetic testing and handle concerns about privacy and discrimination, and report inadequacy to deliver genetic services.\(^5\) For genomics to have an effect on clinical practice that is comparable with its impact on research, advances in the genomic literacy of health-care providers will be required.\(^5\)

In the age of genomics, both genetics of common disorders and large-scale applications in screening will become increasingly important, and primary-care health workers will have to be prepared to discuss these issues with their clients. General practitioners (GPs) may become more involved in preventive checkups and develop a more flexible way to deal with patients’ requests for genetic tests, in addition to their original role in an open-access full-time service for every patient.

Defining genetic core competences for non-genetic health-care workers was considered a prerequisite for implementing genetics education for general practice.\(^1,3,6,7\) Such education programs should be based on an educational needs assessment of GPs referring to the three domains of educational activities: cognitive (knowledge), psychomotor (skills), and affective (attitude).

Recently, a focus-group study among participants from a variety of disciplinary backgrounds explored the genetic educational needs of GPs in the Netherlands.\(^8\) The results showed an urgent need for a genetics curriculum for postgraduate and continuing general practice education. Four overarching themes were identified with regard to educational needs: genetics knowledge, family history, ethical dilemmas, and the role of clinical genetics services. These themes clarified genetics in general practice with implications for education.

The aim of this study was to obtain consensus on prioritization of GPs’ educational needs regarding genetics, as identified in focus groups; the study focused on “knowledge,” “skills,” and “attitudes.” The results are aimed at informing the development of effective genetics education for GPs.

MATERIALS AND METHODS

We used a Delphi method to operationalize the findings from our earlier focus-group study and to obtain consensus on the prioritization of topics for GP genetics education. The Delphi...
technique has been widely used and is an accepted method for gathering data and achieving consensus from respondents within their domain of expertise. The technique was mainly developed by Dalkey and Helmer in 1963 at the Rand Corporation.

A panel of experts e-mailed their responses to a questionnaire about GPs’ educational needs to the researchers in three rounds. The responses were fed back anonymously to all panel members in order to share answers and arguments, thereby enabling the participants to reflect on different views and modify their own.

Panel selection
Eighteen purposively selected experts from the Netherlands responded to an invitation to participate in the study sent to 24 experts (response rate 75%). Of the invited experts, three did not participate due to time constraints and three did not respond at all. Recruitment was guided by the researchers’ network, and a snowball method was used. Through the authors’ network and work in general practice, and clinical genetics (Netherlands Association for Community Genetics and Public Health Genomics), we were familiar with key persons eligible for recruitment to our expert panel. Representatives from patient advocacy groups were asked whether they were interested in participating or could refer someone else. We established a heterogeneous panel of experts, comprising six practising GPs who were also involved in research, five GP trainers, four clinical genetics professionals (one genetic counsellor, three clinical geneticists), and three representatives from patient advocacy groups, all of whom participated anonymously (see Supplementary Table S1 online). The participants were considered to represent a complete overview, from different perspectives, of the importance of genetics core competences for general practice and the need of genetics education in general practice, i.e., what is needed, what works, and what does not work. Eleven experts (61%) were female, and the average age was 51.4 years (SD 9.1). Seven panelists also took part in our previous focus-group study.

The Delphi procedure
The initial questionnaire consisted of 29 topics describing GP educational needs. To arrive at these needs, we first transformed all previously identified educational needs within four overarching identified themes in focus groups into learning outcomes. We then refined the list based on the proposed learning outcomes from the suggested core competences for GPs in Europe. Topics relating to three domains of primary-care genetics were presented to the participants: “knowledge” (7 topics), “skills” (12 topics), and “attitudes” (10 topics).

The flowchart in Figure 1 shows the phases and the (anonymous) process of the three consecutive Delphi rounds. After analyzing the responses to each round (E.J.F.H., L.H., and M.W.) and discussing them with the other researchers (S.J.v.L., M.C.C., G.J.D., and C.v.d.V.), the researchers reworked the responses into a new questionnaire. The Delphi study was conducted between December 2009 and March 2010. At the start of the study, all experts were asked to complete at least three Delphi rounds. Each participant received 100 Euros upon completion of the whole procedure.

Criteria for consensus
The research group discussed criteria for consensus on genetics education needs before the actual study was undertaken. The purpose of the study was to obtain consensus on and prioritize genetics education needs in primary care. For this purpose, in the first two rounds, the experts were asked to prioritize the topics by ranking their importance and to give their top three (Top 3) topics for inclusion in educational modules. In the third and final round, 10 items on which consensus was established in the first two rounds were judged. The definition of the inclusion criteria in a Top 3 in favor of a topic became more rigorous in the following second round because in the first round, at least two experts had to agree with a topic in the Top 3, whereas at least three experts had to agree in the second round. This will be explained in more detail below.

Round 1. In the first round, the experts were asked to rate the educational urgency for GPs of each of the 29 topics on a 7-point
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scale: “I believe that GPs have a strong need for education on [topic],” totally disagree (1) to totally agree (7). Experts were asked to comment on the topics they had given the lowest (1) and the highest (7) rating or about which they had doubts. The responses were converted into importance-based clusters of categories (low- (1–2), medium- (3–5), and high- (6–7) importance categories). The experts were also asked to indicate 3 (Top 3) of the 29 educational needs that they thought GPs most urgently wanted to be incorporated in an educational program to be delivered within the next 12 months. In the first round, consensus in favor of a topic was defined as ≥75% agreement regarding the “importance category” and/or inclusion among the Top 3 by at least two experts.

Round 2. The questionnaire for the second round consisted of the 16 topics that had survived the first round. Some small adjustments were made to clarify topics that had been shown to be somewhat unclear. The inclusion criteria for the next round were more rigorous: ≥75% agreement on importance category and/or inclusion in the Top 3 by at least three experts. An exception was made for topic no. 15 (“educating patients on the possibilities and limitations of genetic tests”), which despite 76% agreement was rejected in the second round because the experts thought there was too much overlap with topic no. 4 (“knowledge of possibilities and limitations of genetic tests”), which did pass the round.

Round 3. For each topic, the experts received a summary of the comments from the previous two rounds with the number of Top 3 ratings in round 2. The experts were asked to list their Top 10 genetics education needs for GPs, and Kendall’s W (coefficient of concordance assessing agreement among raters) was computed for these rankings.

RESULTS

After three Delphi rounds, 29 topics (Table 1) were reduced to 10 priorities regarding genetic education needs (Table 2). All 18 participants completed all three rounds. Response was high with many comments per round (Table 1), indicating strong involvement of the experts. Of the 29 initial topics, 10 remained after three rounds (Boxes 1 and 2). Of the 29 initial topics, 3 were modified after comments.

High agreement on a topic did not always imply high frequency in the Top 3. In fact, the reverse was true for some topics, which led to some unexpected results. Topic 1 (“Refreshing knowledge of basic genetic principles”), for example, showed only 39% agreement, but nevertheless made it through to the third round because four experts placed it in their Top 3. In support of topic no. 1, some experts commented, “I think there are great differences in basic knowledge for young and older GP generations” (active GP) and “without a proper knowledge basis, everything else will be futile.” These comments underscored the notion that improving genetics knowledge will pave the way for successful improvement of skills and attitudes for all GPs.

An example of a topic that was accepted in the first round (76% agreement, N = 1 in Top 3) but rejected in the second round (72% agreement, N = 1 in Top 3) is topic no. 18 (“Explaining the consequences of a genetic test for a patient and his or her family”). According to a clinical genetics professional, “this task should be specifically assigned to the genetic counselor. GPs should be able to generally evaluate whether a patient should be referred” and “the consequences of genetic test results are diverse. Generalization would be dangerous and might lead to misinformation. It seems therefore wiser for this kind of specific information to be delivered by a clinical genetics professional.”

After round 2, there was consensus on 10 topics, which increased for most after modification of the wording. In the end, it was not difficult to distinguish between accepted and rejected topics. The list of prioritized topics at the end of round 3 supports the development of educational modules with the main focus on skills and knowledge (Kendall’s W = 0.43, P < 0.001).

Although Kendall’s W showed significant agreement among the respondents, there were also differences of opinion between subgroups of experts on different topics. For example, participants from the active GP subgroup and clinical genetics professionals subgroup commented differently on topic no. 10 (“Discussing genetic risks with patients (risk communication”). Active GPs (mean rank order 6.6) commented, “Risk communication is difficult, certainly in the case of genetic diseases” and “Risk communication is becoming more important, most GPs are not educated on this topic.” Clinical genetics professionals’ comments, however, were less supportive of adding this topic to the Top 10 of educational topics (mean rank order 10). “I think it depends on the [genetic] disease and the degree of difficulty. I prefer the GP to leave this up to the clinical geneticist” and “if risk communication is meant as a means to support the patient in handling their genetic risk, this could be a GP’s responsibility. However, if it is meant the GP should be capable of calculating a certain genetic risk and discuss this with the patient, additional education would be necessary.”

Relatively high agreement was found within the subgroups of GP trainers (Kendall’s W = 0.60, P = 0.002), clinical genetics professionals (Kendall’s W = 0.78, P = 0.001), and representatives of patient organizations (Kendall’s W = 0.92, P = 0.003), whereas agreement was relatively low among practicing GPs (Kendall’s W = 0.27, P = 0.101).

DISCUSSION

Our study generated consensus on a Top 10 list of prioritized topics for GPs’ genetics education. The highest-ranking topics were concerned with skill and knowledge competences: “Recognizing signals that can indicate a hereditary component of a disease,” “Evaluating indications for referral to a clinical genetics centre,” and “Knowledge of the possibilities and limitations of genetic tests.” These priorities could, in particular, be met by case-based education.
Strengths and limitations of the study

The Delphi procedure included 18 selected experts, who completed all rounds. Despite the experts’ differing backgrounds, it remains to be investigated if the results have relevance beyond the Dutch health-care system as the sample was drawn from this particular health-care system. General practice in the Netherlands is an open-access full-time service for every patient with any medical complaint, request, or question. The service includes a list system, implying that every person (with or without a disease) is on the list of one GP, thus guaranteeing optimal continuity of care. The GP handles more than 90% of all presented complaints and diseases. If genetic counseling as a primary-care service is available in their particular region, the GP manages most referrals to this service as to most other primary-care services and to all secondary-care services. Therefore, the GP is the first health-care professional to whom a patient will turn when he/she has questions on prevention and treatment of disease.

The results of this study are in line with some studies and differ from others with regard to the need for increased genetics knowledge for GPs.8 This difference may be due to differences between health-care systems. However, our previous focus-group results8 are supported by the outcomes of the aforementioned studies regarding deficiency in skills (e.g., taking a

| Topic no. | Round 1 | Round 2 |
|-----------|---------|---------|
|           | n       | %       | N in Top 3 | n    | % | N in Top 3 | Result | In round |
| Knowledge |         |         |           |       |    |           |        |          |
| 1         | 18      | 39      | 4         | 18    | 39 | 4         | Accepted |          |
| 2         | 17      | 53      | 1         | 0     | 0  | 0         | Rejected 1 |
| 3         | 17      | 76      | 5         | 18    | 83 | 4         | Accepted 1 |
| 4         | 18      | 89      | 10        | 18    | 89 | 8         | Accepted 1 |
| 5         | 16      | 69      | 5         | 17    | 71 | 3         | Accepted 1 |
| 6         | 17      | 65      | 2         | 18    | 72 | 5         | Accepted 1 |
| 7         | 16      | 50      | 0         | 0     | 0  | 0         | Rejected 1 |
| Skills    |         |         |           |       |    |           |        |          |
| 8         | 18      | 78      | 2         | 18    | 100| 6         | Accepted 1 |
| 9         | 16      | 56      | 1         | 0     | 0  | 0         | Rejected 1 |
| 10        | 17      | 71      | 4         | 18    | 50 | 3         | Accepted 1 |
| 11        | 16      | 75      | 3         | 17    | 71 | 7         | Accepted 1 |
| 12        | 18      | 61      | 1         | 0     | 0  | 0         | Rejected 1 |
| 13        | 16      | 44      | 3         | 18    | 39 | 3         | Accepted 1 |
| 14        | 18      | 61      | 0         | 0     | 0  | 0         | Rejected 1 |
| 15        | 18      | 61      | 2         | 17    | 76 | 2         | Rejected 2 |
| 16        | 17      | 53      | 0         | 0     | 0  | 0         | Rejected 1 |
| 17        | 16      | 56      | 2         | 18    | 61 | 0         | Rejected 2 |
| 18        | 17      | 76      | 1         | 18    | 72 | 1         | Rejected 2 |
| Attitude  |         |         |           |       |    |           |        |          |
| 19        | 18      | 56      | 0         | 0     | 0  | 0         | Rejected 1 |
| 20        | 18      | 33      | 2         | 18    | 33 | 3         | Accepted 1 |
| 21        | 17      | 29      | 0         | 0     | 0  | 0         | Rejected 1 |
| 22        | 16      | 50      | 0         | 0     | 0  | 0         | Rejected 1 |
| 23        | 16      | 56      | 0         | 0     | 0  | 0         | Rejected 1 |
| 24        | 18      | 61      | 1         | 0     | 0  | 0         | Rejected 1 |
| 25        | 18      | 61      | 2         | 17    | 71 | 1         | Rejected 2 |
| 26        | 17      | 71      | 2         | 18    | 56 | 1         | Rejected 2 |
| 27        | 17      | 41      | 0         | 0     | 0  | 0         | Rejected 1 |
| 28        | 18      | 67      | 0         | 0     | 0  | 0         | Rejected 1 |
| 29        | 17      | 76      | 1         | 18    | 72 | 2         | Rejected 2 |
family history, referral to appropriate regional genetics services, and nondirective counseling). It may be problematic for primary-care providers to take appropriate steps in response to the perceived shift in the importance of genetics in primary care, such as taking enough time to discuss the family history or to perform nondirective counseling.

Another possible weakness of our study is regression to the mean, although this is inherent to this consensus method: experts are inclined to adjust their opinions during a consensus process. Nevertheless, there was a high degree of agreement on the 10 final topics, whereas 19 topics were not accepted, despite several adjustments. The procedure started with topics based on our earlier focus-group research, and some experts were aware of these results, although they were unaware of each other’s identities. Although this may have biased their opinions, the validity of the focus-group results was checked by comparing them with the results of the consensus procedure, a process commonly referred to as triangulation. As compared with the results of the focus groups, this study strengthened the prioritization of genetic educational topics for general practice. Also, the transparency of the way we dealt with comments and ratings and adjusted or rejected topics is expected to have improved the validity and reliability of the resulting consensus.

Comparison with existing literature
Our study resulted in a Top 10 list of genetic educational topics in primary care through consensus and prioritization, building on earlier studies. A previous paper described the absence of genetics educational objectives for Dutch non-genetic healthcare providers. This Delphi study has laid a firm foundation, supported by experts’ opinions, for the development of more appropriate genetics education for GPs.

In addition to the perceived inadequacy of primary-care workers to integrate genetics into daily practice, Scheuner et al. identified deficiencies in primary-care workers’ basic genetic knowledge and ability to interpret familial patterns. This is in line with our prioritized educational topics, which include knowledge of basic genetic principles, the most common genetic disorders, and family history skills. Taylor et al. stated that primary care should be encouraged to invest more time in family history data. However, they also stressed identified barriers (e.g., time constraints) and the need to develop strategies to overcome difficulties preventing GPs from routinely obtaining family history information. These barriers and strategies are still under construction and may explain why topic no. 13 (“Taking and interpreting a family history”) ended ninth in the Top 10 list of genetic education priorities. In this study, we did not elaborate on these difficulties, but they should definitely be considered during the development of education modules concerning the integration of family history skills according to referral criteria.

Table 2: Overview of mean rank order (Top 10) and Kendall’s coefficient of concordance assessing agreement among experts

| Topic no. | Mean rank order (general) | Mean rank order (active GPs) | Mean rank order (GP trainers) | Mean rank order (clinical genetic professionals) | Mean rank order (representatives of patient organizations) |
|-----------|--------------------------|------------------------------|-----------------------------|-----------------------------------------------|----------------------------------------------------------|
| 11        | 2.9                      | 3.0                          | 4.4                         | 1.5                                           | 2.3                                                      |
| 8         | 3.1                      | 3.7                          | 3.8                         | 1.8                                           | 2.7                                                      |
| 4         | 3.2                      | 4.7                          | 1.2                         | 4.3                                           | 2.3                                                      |
| 6         | 4.6                      | 5.3                          | 4.4                         | 5.0                                           | 2.7                                                      |
| 3         | 5.2                      | 4.9                          | 5.6                         | 4.5                                           | 6.0                                                      |
| 1         | 6.1                      | 5.3                          | 6.6                         | 7.5                                           | 5.0                                                      |
| 5         | 6.6                      | 6.0                          | 6.0                         | 7.5                                           | 7.0                                                      |
| 10        | 7.3                      | 6.6                          | 5.0                         | 10                                            | 8.7                                                      |
| 13        | 8.0                      | 7.5                          | 8.6                         | 7.3                                           | 9.0                                                      |
| 20        | 8.1                      | 8.0                          | 9.2                         | 5.8                                           | 9.3                                                      |
| N         | 18                       | 6.0                          | 5.0                         | 4.0                                           | 3.0                                                      |

Kendall’s W 0.433 0.271 0.595 0.779 0.919
Chi-square* 70.208 14.660 26.760 28.036 24.818
d.f. 9 9 9 9 9
Significance 0.000 0.101 0.002 0.001 0.003

GP, general practitioner; Kendall’s W, Kendall’s coefficient of concordance. A lower score in mean rank order, results in higher ranking in the Top 10.

*Comparing score for topics within the group.
These results support the implementation of genetics education aimed at enhancing effective referral indications and options.

The results of this Delphi study differ from those of the GenEd study of 2004 by Calefato et al.\(^ {12}\) and of previous focus groups.\(^ {8}\) Competencies relating to attitudes, such as “ethical, legal, and public health issues” and “psychosocial and counseling issues,” received more attention in these studies. This difference may be attributed to the fact that the GenEd study was not limited to the Netherlands but encompassed five European countries with differing health-care systems. In our study, the experts commented that genetics education should first focus on “knowledge” before moving on to “attitudes.” Some comments on this issue were rather ambivalent: “Attitude is not specific to genetics” and “A good attitude should be an intrinsic component of the GP’s role.” Therefore, although it seems the “attitude” domain is considered essential for genetics education of GPs (i.e., case-based learning with medical ethical problems), effective implementation of genetics education may be jeopardized if too much attention is paid to this area.

The higher than expected number of topics in the Top 10 referring to “knowledge” (5 of 7 topics) exceeded the number of “skill” topics (4 of 11 topics). This may be explained by the experts’ perceptions that genetic knowledge should be brought up to date before related skills can be learned. This unexpected result needs further research and probably explains a relatively low agreement among practicing GPs, as some may find their genetic knowledge or skills sufficient and others not.

**Implications for future research and clinical practice**

Unless a scientific, logistical, and ethical framework for the appropriate and effective use of genomic information is in place, the primary-care workforce is unlikely to be adequately prepared\(^ {1,3,5–7,12,20,22–24}\) to provide such information in general practice. If GPs remain uneducated in genetics and therefore incompetent related to the use of genomic information in

| BOX 1: ACCEPTED TOPICS |
|------------------------|
| **Knowledge** |
| 1 Knowledge of basic genetic principles (round 1) |
| 2 Refreshing knowledge of basic genetic principles (rounds 2 and 3) |
| 3 Knowledge of the most important genetic disorders in the Netherlands (round 1) |
| 4 Knowledge of the possibilities and limitations of genetic tests |
| 5 Knowledge of the wide array of referral possibilities concerning genetics |
| 6 Knowledge of the most important sources of genetic information |
| **Skills** |
| 7 Evaluating indications for referral to a clinical genetics center |
| 8 Discussing genetic risks with patients (risk communication) |
| 9 Recognizing signals potentially indicative of a hereditary component of a disease |
| 10 Taking and interpreting a family history |
| **Attitude** |
| 11 Being aware of the fact that many symptoms/conditions can have a genetic component (round 1) |
| 12 Being aware of the possibility of a genetic/hereditary component of symptoms and disorders (rounds 2 and 3) |

| BOX 2: REJECTED TOPICS |
|------------------------|
| **Knowledge** |
| 2 Knowledge of different hereditary patterns |
| 7 Knowledge of the consequences of genetic testing for obtaining a mortgage and insurance |
| **Skills** |
| 9 Discussing with patients how to cope with (an increased risk for) a genetic disorder |
| 12 Explaining genetic information in a way that is adapted to the patient’s level of knowledge |
| 14 Drawing and interpreting a pedigree |
| 15 Educating patients on possibilities and limitations of genetic tests |
| 16 Explaining the genetic aspects, except lifestyle, of multifactorial disorders |
| 17 Informing parents about the possibilities and limitations of prenatal and neonatal screening |
| 18 Explaining the possible consequences of a genetic test for a patient and his or her family |
| **Attitude** |
| 19 Recording a family history in such a way that it can be easily retrieved |
| 21 Guiding patients with genetic issues in a nondirective way |
| 22 A medical practitioner’s role in (actively) suggesting the possibility of having a genetic test |
| 23 Demarcating tasks in the field of genetics in comparison with other caregivers |
| 24 A general practitioner’s role in decisions about discussing (the chances of) a genetic disorder with the patient’s family |
| 25 Offering support to patients with (an increased risk for) a genetic disorder |
| 26 Dealing with the choices relating to genetics made by people from different cultures |
| 27 Dealing with sensitivities surrounding genetic disorders in families |
| 28 Informing patients about genetic risks in consanguine marriages |
| 29 Dealing with ethical dilemmas in genetics |
general practice, individual genetic medical care provided will likely be unhelpful and possibly even harmful. We believe the results of this study should be used in the near future to guide the implementation of genetics education in the Netherlands and perhaps even internationally. Although the majority of the issues investigated cover genetics-related knowledge, skills, and attitudes essential for every medical-care provider, further studies will have to determine whether the results are relevant to other medical specialties as well.

We are currently working on developing genetics education for GPs in collaboration with the Dutch College of General Practitioners. This entails a written educational module aimed at improving genetics knowledge, an informative website specifically aimed at genetics in general practice in the Netherlands, and a live Continuing Medical Education module aimed at improving genetic skills and attitude. Hopefully, this will improve (genetic) medical care in the Netherlands and will meet the needs expressed by GPs and experts in our previous work and the Top 10 of genetic educational topics presented in this paper.

Preparation of health-care providers for the future of genetic medicine, with personalized genomic information and education, will lead to effective use of genetics in daily primary care.

**SUPPLEMENTARY MATERIAL**

Supplementary material is linked to the online version of the paper at http://www.nature.com/gim

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All authors have made a substantial contribution to the concept and design of the study. E.J.F.H., M.W., and L.H. carried out the Delphi consensus procedure, analyzed the data, and wrote the first draft of the paper. E.J.F.H., L.H., S.J.v.L., C.v.d.V., G.J.D., and M.C.C. were involved in the design and coordination of the study.

**DISCLOSURE**

The authors declare no conflict of interest.

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