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# Family physicians' self-perceived importance of providing genetic test information to patients: A cross-sectional study from Slovenia

Authors' Contribution:
Study Design A
Data Collection B
Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
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**Background:** Management of patients with genetic problems, including provision of genetic testing, is increasingly becoming a part of primary health care. The aim of this study was to determine the family physicians' (FPs) self-perceived importance of providing genetic test information to their patients.

**Material/Methods:** This was an observational cross-sectional postal study in the whole population of Slovenian family physicians (N=950). Its main outcome measure was the perceived importance of providing genetic test information on each of 10 items on a 5-point Likert scale.

**Results:** There were 271 (27.1% response rate) FPs that completed the questionnaire, out of which 205 (75.6%) were women. Mean age of the sample was 45.5±10.6 years. More than 90% of Slovene FPs felt that it was their professional duty to discuss genetic testing issues with their patients. They were particularly prone to discuss clinical implications of positive and negative test results, as well as giving the patients information about the risk of passing a mutation onto children.

**Conclusions:** Most Slovene family physicians feel responsible and willing to offer and discuss genetic testing and implications with their patients. Additional education should be provided to empower them for this task.

**Keywords:** Genetics • Family Medicine • Education

**Full-text PDF:** http://www.medscimonit.com/download/index/idArt/890013







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## Background

Family physicians (FPs) are usually the first physicians that make contact with people at risk for developing various diseases [1]. They also perform preventive activities that are already based on genetics (i.e., taking family history) [2,3]. Also, people in need of a professional advice commonly turn to FPs because their level of trust to their FPs is very high [4].

Patient management from a genetic point of view is increasingly becoming a part of various parts of health care, especially of primary health care [5–7]. This highlights the great need for FPs to gain appropriate knowledge about genetic tests, their indications, and interpretation of results and about ethical issues associated with genetics in medicine. It also highlights the need for FPs to gain specific skills such as communication about early genetic tests in healthy individuals at risk and genetic test interpretation adjusted to patient needs and level of understanding [8–11]. FPs should also be able to provide objective information about pros and cons of early genetic testing to parents of children at risk.

So far, few studies have dealt with the importance of providing genetic information and information on genetic tests by FPs [12–14]. However, previous studies showed that the interest of family doctors in including genetics in everyday management of their patients and the readiness to provide the information on genetic testing to their patients are the main factors influencing the success of integration of genetics into primary health care level [13,15,16]. Therefore, the aim of this study was to determine the FPs' self-perceived importance of providing genetic test information to their patients.

## Material and Methods

This was an observational, cross-sectional, postal study conducted in Slovenian FPs. The study was approved by the Slovenian Ethics Committee (No. 40/09/12).

The study population consisted of all Slovenian FPs. According to the internal data of the Slovenian Medical Chamber, there were 950 working FPs in Slovenia at the time of the study. As the membership in this chamber is obligatory, this number represents the whole population of Slovenian FPs.

Data was collected by a postal survey sent by the Slovenian Medical Chamber in March 2013. The mailing consisted of the questionnaire (described below), the invitation letter, and a pre-stamped return envelope.

We used an internationally validated questionnaire [5,6,8,12] consisting of demographic questions and questions on the self-perceived importance of providing genetic test information. There were 10 questions on the self-perceived importance of providing genetic test information. Each question could be answered on a 5-point Likert scale (1 = not important, 5 = extremely important).

We analyzed the data by SPSS version 13.0 (SPSS for Windows, Chicago: SPSS Inc.) and performed univariate analysis.

**Table 1.** Demographic and professional characteristics of family physicians in a sample.

| Characteristic | Number of family physicians | Percentage of family physicians |
|---|---|---|
| Sex | | |
| Male | 66 | 24.4 |
| Female | 205 | 75.6 |
| Education | | |
| Family medicine specialist | 216 | 79.7 |
| Family medicine resident | 50 | 18.5 |
| Specialist of other specialties | 4 | 1.5 |
| Without any specialization | 1 | 0.4 |
| Education in genetics | | |
| None | 39 | 14.4 |
| Genetic content during undergraduate studies | 220 | 81.5 |
| Genetic content in specialist training | 6 | 2.2 |
| Genetic content in courses | 1 | 0.4 |
| Genetic content in postgraduate studies | 4 | 1.5 |
| No. of inhabitants living in practice catchment area | | |
| Less than 5,000 | 49 | 18.1 |
| 5,000–20,000 | 88 | 32.5 |
| 20,000–100,000 | 63 | 23.2 |
| More than 100,000 | 70 | 25.8 |
| Frequency of contacts with patients with genetic diseases in everyday practice | | |
| Daily | 79 | 29.9 |
| Weekly | 116 | 43.9 |
| Monthly | 46 | 17.4 |
| Several times per year | 12 | 4.5 |
| Less often | 11 | 4.2 |

## Results

There were 271 (27.1%) FPs who completed the questionnaire, out of which 205 (75.6%) were women (Table 1). Mean age of the sample was 45.5±10.6 years, mean working period was 17.3±11.6 years, and mean time from graduation was 19.6±10.9 years.





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**Table 2.** Scores of the questionnaire on self-perceived importance of providing genetic tests' information.

| Item | Mean score ± standard deviation | Not important (%) | Rather important (%) | Important (%) | Very important (%) | Extremely important (%) |
|---|---|---|---|---|---|---|
| Information on what sample needed and what genetic test will be performed | 3.5±0.8 | 1.5 | 8.6 | 39.6 | 41.4 | 9.0 |
| Clinical implications of a positive and negative result | 4.1±0.8 | 1.1 | 0.4 | 18.7 | 51.9 | 28.0 |
| The sensitivity and specificity of the test | 3.7±0.9 | 1.9 | 6.7 | 29.5 | 48.1 | 13.8 |
| Options for giving risk estimates without having genetic testing | 3.5±0.8 | 1.5 | 5.6 | 43.1 | 38.2 | 11.6 |
| Information on the risk of passing a mutation onto children | 3.8±0.8 | 1.1 | 1.9 | 27.6 | 53.4 | 16.0 |
| Psychosocial impact of test results on self and relatives | 3.8±1.0 | 1.5 | 8.2 | 25.5 | 41.6 | 23.2 |
| Confidentiality issues | 3.8±1.0 | 1.1 | 8.6 | 29.1 | 36.2 | 25.0 |
| Options and limitations of medical surveillance following tests | 3.9±0.9 | 1.1 | 3.4 | 28.8 | 42.4 | 24.2 |
| The patient has a right to remain in ignorance | 3.5±1.0 | 3.0 | 10.3 | 35.4 | 33.5 | 17.9 |
| Information about if the test is covered by the insurance or of patients will have to pay for it themselves | 3.5±1.0 | 4.5 | 10.8 | 33.6 | 33.6 | 17.5 |

More than 90% of Slovene FPs felt that it was their professional duty to discuss genetic testing issues with their patients. They were particularly likely to discuss clinical implications of positive and negative test results: 98.6% of FPs felt that this was important or very/extremely important). The majority of them (97.0%) also felt that it was important or very/extremely important to give the patients information about the risk of passing a mutation onto children. They were slightly less likely to discuss risk estimates for a genetic disorder without genetic testing: 92.9% felt that this was important or very/extremely important. Similarly, 90.3% felt that confidentiality issues were important or very/extremely important and 86.8% felt that it was important or very/extremely important that the patient has a right to remain in ignorance (Table 2).

## Discussion

In general, Slovenian FPs perceived that providing genetic test information to patients is important or very/extremely important. This highly perceived importance came as a surprise, as previous studies showed that FPs were interested in genetic medicine topics [10,15,17] but perceived genetics as a low practice priority [16]. Also, FPs identified a clear distinction between the routine use and function of family history in their clinical decision-making versus the conceptualization of genetics and genetic conditions [18]. In our study, FPs actually expressed their clear role in genetics, especially in terms of a comprehensive approach. Specifically, our study showed that FPs' perceived importance of providing genetic test information was the highest for the items associated with practical management of their patients, such as providing information about clinical implications of positive and negative genetic test results to patients and giving information on the risk of passing a mutation onto children. Another study showed that FPs had a high level of uncertainty about genetic test results [9]. This confirms the findings of our study that FPs seemed to think a lot about the practical management of patients. FPs are known to be practically oriented and recognize the implications and the benefits of using some knowledge and tests in their everyday management of patients [13,15,19]. The high perceived importance of giving information about the risk of passing a mutation onto





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children points to the basic feature of family medicine – the inclusion of family features in the management of patients [20].

Lower interest was found in items concerning ethical issues (confidentiality and patient right to remain in ignorance). As ethical issues are an inevitable part of genetic testing, it seems clear that FPs need additional education on this theme.

The majority of FPs in our study received education on genetics at the undergraduate level. On the other hand, more than 74% reported having contacts with patients with genetic diseases at least weekly. Since the mean age of FPs was almost 50 years, it is obvious that they are in need of additional education in genetics.

This study was performed in a representative sample of Slovenian FPs and its findings can therefore be generalized to the whole population of FPs in Slovenia. Another strength of this study is the use of a previously validated questionnaire, which gives us confidence in the reliability of data. This was a cross-sectional study, thus it is impossible to detect any causal relationship between variables. The response rate in this study was as expected because a 20% response rate is usual for postal surveys [21]. Nevertheless, it can be a source of selection bias.

## Conclusions

Most Slovene family physicians feel responsible for and willing to offer and discuss genetic testing and its implications with their patients. Additional education should be provided to empower them for this task.

### Conflicts of interest

The authors declare no conflict of interest.

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## References:

1. Ten Kate LP, Al-Gazali L, Anand S, Bittles A et al: Community genetics. Its definition 2010. J Community Genet, 2010; 1(1): 19–22
2. Rich EC, Burke W, Heaton CJ et al: Reconsidering the family history in primary care. J Gen Intern Med, 2004; 19: 273–80
3. Guttmacher AE, Collins FS, Carmona RH: The family history – more important than ever. New Engl J Med, 2004; 351(22): 2333–36
4. Softic N, Smogavec M, Klemenc-Ketis Z, Kersnik J: Prevalence of chronic diseases among adult Slovene population. Zdrav Var, 2011; 50: 185–90
5. Calefato JM, Nippert I, Harris HJ et al: Assessing educational priorities in genetics for general practitioners and specialists in five countries: factor structure of the Genetic-Educational Priorities (Gen-EP) scale. Genet Med., 2008; 10(2): 99–106
6. Challen K, Harris H, Kristoffersson U et al: General practitioner management of genetic aspects of a cardiac disease: a scenario-based study to anticipate providers' practices. J Community Genet, 2010; 1(2): 83–90
7. Brodziak A, Brewczynski A, Bajor G: Clinical significance of knowledge about the structure, function, and impairments of working memory. Med Sci Monit, 2013; 19: 327–38
8. Julian-Reynier C, Nippert I, Calefato JM et al: Genetics in clinical practice: general practitioners' educational priorities in European countries. Genet Med, 2008; 10(2): 107–13
9. Abad-Perotín R, Asúnsolo-Del Barco A, Silva-Mato A: A survey of ethical and professional challenges experienced by Spanish health-care professionals that provide genetic counseling services. J Genet Couns, 2012; 21(1): 85–100
10. Trinidad SB, Fryer-Edwards K, Crest A et al: Educational needs in genetic medicine: primary care perspectives. Community Genet, 2008; 11(3): 160–65
11. Acheson LS, Wiesner GL: Current and future applications of genetics in primary care medicine. Prim Care, 2004; 31(3): 449–60
12. Nippert I, Harris HJ, Julian-Reynier C et al: Confidence of primary care physicians in their ability to carry out basic medical genetic tasks-a European survey in five countries-Part 1. J Community Genet, 2011; 2(1): 1–11
13. McCahon D, Holder R, Metcalfe A et al: General practitioners' attitudes to assessment of genetic risk of common disorders in routine primary care. Clin Genet, 2009; 76(6): 544–51
14. Blaine SM, Carroll JC, Rideout AL et al: Interactive genetic counseling role-play: a novel educational strategy for family physicians. J Genet Couns, 2008; 17(2): 189–95
15. Robins R, Metcalfe S: Integrating genetics as practices of primary care. Soc Sci Med, 2004; 59(2): 11
16. Suther S, Goodson P: Barriers to the provision of genetic services by primary care physicians: a systematic review of the literature. Genet Med, 2003; 5(2): 70–76
17. Tomatir AG, Sorkun HC, Demirhan H, Akdağ B: Genetics and genetic counseling: practices and opinions of primary care physicians in Turkey. Genet Med, 2007; 9(2): 130–35
18. Mathers J, Greenfield S, Metcalfe A et al: Family history in primary care: understanding GPs' resistance to clinical genetics – qualitative study. Br J Gen Pract, 2010; 60(574): 221–30
19. Poplas Susic T, Kersnik J, Kolsek M: Why do general practitioners not screen and intervene regarding alcohol consumption in Slovenia? A focus group study. Wien Klin Wochenschr, 2010; 122(Suppl.2): 68–73
20. Allen J, Gay B, Crebolder H et al: The European definition of general practice/family medicine: Wonca Europe, 2011
21. Klemenc-Ketis Z, Kersnik J, Ojstersek J: Perceived difficulties in managing ethical problems in family practice in Slovenia: cross-sectional study. Croat Med J, 2008; 49: 799–806





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