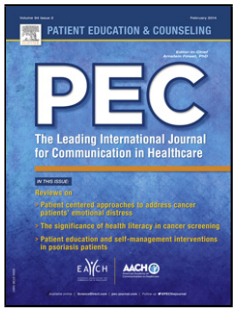

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The Importance of the General Practitioner as an Information Source for Patients with Hereditary Haemochromatosis

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Abstract

Objective: To explore Hereditary Haemochromatosis (HH) patients' perspectives on genetic information, namely the types of sources used, preferred or trusted.

Methods: **A survey online was conducted by the European Federation of Associations of Patients with Haemochromatosis (EFAPH) and applied to members of 9 national associations.**

Results: **From a total of 1019 validated questionnaires, 895 respondents had performed a genetic testing for HH. From these,** 627 self-declared that they were sufficiently informed about the implications of the genetic test to their health. The majority (66%) obtained the information from a specialist doctor, but would like to obtain it from the family doctor. However, the specialist was still the one they trusted more (69%). Regarding the 298 respondents who did not feel sufficiently informed, the majority (78%) also would like to have information from the family doctor although they also trusted the specialist more (75%). A different perspective was reported when patients were asked about the implications of the genetic testing to their family members, where the majority of respondents preferred obtaining information from a specialist (69%).

Conclusion: This study elucidates the patients´ needs for information and identifies the General Practitioner (GP) as the preferred source to obtain information about HH.

Practice Implications: These results may have important implications in future strategies for HH awareness, giving a special emphasis on GPs as the main players.

Keywords

Hereditary Haemochromatosis; Patient Communication; Sources of Information; General Practitioner.

1. Introduction

Patient-centered medicine (i.e., care in which the doctor responds to the patients' needs, including their thoughts or feelings), is a recent a buzz-word [1]. **This concept became most popular after its comprehensive description by Brown and co-workers in 1995 [2] when identified several interconnecting components which included the enhancement of doctor-patient relationships**. It is also increasingly evident that patients want to be fully informed and be part of the treatment decision making. They have a better idea of their requirements, and are able to verbalize their needs and preferences when they are invited to do so [3]. There is evidence that informed patients are better aware of matters relating to their care and therefore they should also be better placed to take an active part in their own care [4] and improve the quality and responsiveness of health care services. Nevertheless, to our knowledge, no sufficient attention has been paid to the patients' perspectives on the sources they use to obtain information on genetic disorders.

HH is one of the most common adult-onset genetic disorders in European-derived populations [5]. It is a chronic disorder which occurs when the normal regulation of iron absorption is disrupted resulting in the accumulation of excessive iron in the liver, pancreas, heart and joints leading to organ damage, and impaired function [6]. In spite of being a potentially lethal disease, early diagnosis of HH and treatment by venesections can restore normal life expectancy, reduce symptoms, and help to prevent organ damage [7]. In contrast, failure to detect HH increases the likelihood that irreversible adverse health effects will occur and increases the future financial burden associated with health care for persons with HH [8]. These findings have led to recommendations for increased case detection and universal screening using phenotypic testing (eg, transferrin saturation) to permit early treatment before the onset of clinical disease [9]. **Moreover, taking into**

consideration the healthy status of early diagnosed HH carriers, the potential use of their blood for transfusion is nowadays an issue of major interest [10].

Early detection and treatment of HH depends on increased awareness and proper information among health professionals and patients themselves, who play an important role in the motivation and spread of information among their relatives. In spite of these recommendations, much of the literature suggests that HH is still largely underdiagnosed and therefore undertreated [11-13], a position that was also recognized by the European Commission in response to a parliamentary question promoted by the European Federation of Patients with Haemochromatosis (EFAPH) (*E-012656/2011*).

In general, there is a paucity of information about the patients' perceptions about HH and, to our knowledge, no studies have ever been performed regarding the sources of information they find useful [8, 14]. This is particularly relevant in Europe where, in spite of a low clinical penetrance, the genetic condition is highly prevalent [5]. That question was addressed in this study in which members of nine patients' associations were invited to participate in an international survey online. The survey was designed to understand which sources are used by patients or subjects at risk to find information about HH, where do they prefer to get it from, where they would like to find it, and which information sources they trust more.

Our expectation is that the results of this study will be useful for the implementation of new strategies on HH awareness and consequently may contribute to increase early diagnosis of the disease.

2. Methods

2.1. The questionnaire

Members from nine National Associations of Patients with Haemochromatosis, all EFAPH members, (France, Germany, Hungary, Italy, Ireland, Netherlands, Norway, Portugal and United Kingdom) were enrolled in the present study regarding genetic testing and the sources of information used to find information about HH. They were invited to answer to a survey posted online by EFAPH, using the *SurveyMonkey* platform. The survey online was preferred as a suitable and economical method for data collection because our target population was large and our measurements focused on patients' own perceptions [15]. A copy of the survey is available from the authors upon request.

Because this is the first survey of its kind, there was no available *a priori* information to guide our strategy to identify the sources of information usually used by the associations' members. Therefore various steps were taken to ensure the validity and reliability of the study. **Initially, the study design was evaluated by a committee composed of the coordinators of the Biosense project, an experimental platform of engagement and collaboration between Portuguese academic and scientific research institutions and the society, inspired by the European concept of *science shop* [16]. Next the questionnaire format and questions were discussed among members of the Scientific Committee of EFAPH and sent to 3 patients' representatives, namely from Germany, Ireland and Norway, for validation through a pre-testing exercise where they responded to the questions and pointed out possible interpretation difficulties or shortcomings. After this validation, the Scientific Committee approved the English version of the questionnaire. This final version was sent to 9 local coordinators nominated for each participating Association. Whenever necessary, i.e., for non English-speaking populations (Dutch, English, French, German,**

Hungarian, Italian and Portuguese) the local coordinators had to translate the questionnaire into their own native language, without editing or modifying its content. Next the coordinators were responsible for pre-testing the survey online format (including detailed answering instructions) before making it public. They were further responsible for contacting all registered members of the respective Associations (without any exclusion), inviting and motivating them to participate in the study. For this purpose, each local coordinator was sent a suggested model letter that could be used to approach the members stating their voluntary participation and anonymous nature of the study. This process started in November 2011 and finished in July 2012. During this period two reminder e-mails were sent to local coordinators.

The cover page of the questionnaire explained the objective of the study and contained elements of informed consent (including an opt-out option), so that participants only answered the survey after acceptance of the conditions.

2.2. Data collection, sampling and contents

Data were mostly obtained from the electronic version of the survey, but in two cases (Germany and Portugal) the questionnaires were sent by post to the coordinator who then transferred the data to the *SurveyMonkey* platform. The local coordinator was also responsible for the translation of the answers. Demographic data were collected in order to characterize the sample, including gender, age, nationality, years of education and occupation. The respondents' answers about age were displayed as six categories (<25; 25-35; 35-45; 45-55; 55-65; 65-75 and >75 years old) and answers about occupation into fourteen categories (according to the International Standard Classification of Occupations [17]).

Participants were first asked if they had ever undergone a genetic test. **Only respondents who answered affirmatively were allowed to continue with the questionnaire and be validated as participants. This strategy was used as a means to optimize the sample in terms of representing a population of patients or patients at risk.** Participants were then asked if they felt whether or not they were sufficiently informed about the genetic testing and its implications to their health. According to their response to this question, respondents were segregated in two groups for further analyses, (i.e., those who answered "yes" and those who answered "no"). They were further asked where they would like to find (or find more) information about HH and which information sources they trusted more. Sources of information displayed included: Family Doctor, Specialist, Scientist, Nurse, Internet - Official Website (ex: Hospital and University), Internet – Other Website (ex: Wikipedia, Blogs, Facebook, Twitter), Books, Papers and Magazines, Associations of Patients, Family and Friends or Other (free answer).

2.3. Analysis

We computed descriptive statistics (means, standard deviations, and proportions) of the variables and examined the distributions of the responses. Differences in distributions were analyzed by the Chi-Square test with IBM SPSS Statistics 19.

2.4. Ethics

The basic principles of research ethics were followed at all stages of the study. All the data were handled confidentially according to the principles stated in the Helsinki Declaration [18].

3. Results

Three thousand nine hundred and seventy three members were invited to participate and 1032 (26%) answered the questionnaire. **From the total population of 1032 respondents, 13 were excluded because they had supplied only partial replies to demographic questions and did not proceed with the questionnaire. In addition, 124 respondents had not had a genetic test and were therefore excluded from the analyses. The remaining 895 respondents who declared to have performed a genetic test were considered for further analyses.** A schematic representation of sample collection and processing is given in Figure 1. The average age of respondents was 55. 7 ± 11.7 years (mean ± standard deviation), 58% were males and 42% were female. Thirty eight percent of the participants were graduates and 30% of the participants were included in major group 2, Professionals**,** and 21% of total number of participants were retired. The distribution of participants by nationality is recorded in Table 1.

When asked if they "*feel sufficiently informed about your genetic testing and its implications to your health?*" 70% answered "yes" and 30% answered "no". The respondents that felt sufficiently informed were aged between 55-65 years and the respondents that did not feel sufficiently informed were 45-55 years old.

Regarding the respondents that answered "yes" to the above question, when asked "*where did you get information?*", the majority reported that their main source of information was the specialist (66%), 39% referred to the family doctor and 38% to Patients Associations (Table 2 and Fig. 2). When data were analyzed by National Associations we observed that they all obtained information mainly from the specialist, except the participants from Irish Haemochromatosis Society, who obtained information mainly from their family doctors.

When asked "*where would you like to find information*?", a higher percentage expressed a preference for obtaining information from the family doctor (66%), while a lower percentage preferred the specialist (59%) or the patient association (44%) (Table 2 and Fig. 2). Nevertheless, when asked "*Of those sources of information, in which do you trust more?*" the majority mentioned the specialist (69%) followed by the Patient Association (41%) or the family doctor (31%) (Table 2 and Fig. 2).

Regarding the respondents who answered "no" to the question "*Do you feel sufficiently informed about your genetic testing and its implications to your health?*", the majority preferred to obtain information from their family doctor (78%), closely followed by a specialist (77%), with the third preference being official websites on the internet (53%) (Table 2 and Fig. 3). Such as in the case of informed participants, these also trusted more in the specialist (75%), rather than the family doctor (35%) or Patient Association (24%) (Table 2 and Fig. 3).

We also asked the participants if they "*feel sufficiently informed about the implications of your genetic testing to other family members*"**:** 78% answered "yes" and 22% answered "no".

Of the respondents who answered "yes" to the question above, when asked "*where did you get information?*", the majority reported that their main source of information was the specialist (72%). The specialist was also the preferred source (69%) and whom they trusted more (68%) (Table 3).

Of the respondents who answered "no" to the question "*Do you feel sufficiently informed about the implications of your genetic testing to other family members"* 77% stated that they would like to obtain the information from the specialist and 72% had more trust in the specialist (Table 3).

4. Discussion and conclusion

4.1. Discussion

This is the first European survey designed to explore patients' perspectives on genetic information. The main motivation to perform this study was the general concern about HH underdiagnosis and undertreatment, suggesting that both clinicians and laypeople have little awareness of the disease. Therefore HH awareness needs to be improved. In order to optimize resources, we did not approach all possible intervenients (such as health professionals, patients in general, etc) but decided to target a potentially "informed" HH population. It is well known that informed patients are better aware of matters relating to their care [3]. For that purpose we chose the members of Patients' Associations who are "a priori" more likely to be informed. We asked them where they obtained the information, namely the types of sources used, preferred or trusted.

Results of the questionnaire revealed that 70% of respondents reported that they felt sufficiently informed about genetic testing and its implications to their health. However, a lower percentage (55%) that they were sufficiently informed about the implications to their family members.

The results reveal that respondents would like to receive more information about HH from the family doctor, despite having more trust in the specialist. This highlights the importance of targeting GPs as an important requested source of information. This is not surprising, since it had been already reported that about half of HH diagnoses are performed by a gastroenterologist, a hematologist and some other specialist physician, as opposed to being diagnosed by a primary care provider [14]. A recent study in Australia [7] showed that of 80% of GPs who reported that they had patients with HH, only 41% managed the condition primarily.

The participants from Irish Haemochromatosis Society were the only ones who answered that they obtained information about genetic testing mostly from the family doctor. This is interesting and probably reflects a commitment of GPs in Ireland to promote the diagnosis, treatment and management of patients with HH in the Primary Care Setting, in collaboration with the Irish Haemochromatosis Association [19]. This could also reflect the very high frequency of HH encountered by GPs in Ireland compared to those in other countries where the disease is not so common. As the GPs in Ireland have more cases of patients with HH they are more informed about the disease and thus their patients receive better treatment.

The results showed that participants also used the internet as a source of information. We are aware that web-based information is a valuable resource for patients with many conditions although its reliability and quality have been questioned [20]. This is reflected in the results by the fact that a higher number of respondents selected official internet official websites rather than other websites. It is important to note that healthcare professionals have an important role in evaluating websites and directing patients and care givers to sites which provide accurate and up-to- date information [21].

In this study the respondents also recognized the specialists' relevance in transmitting the information about genetic testing implications to other family members. In a recent study, Leandro and co-workers reported a general lack of knowledge about the selection of patient cases that should be sent for genetic counseling or for molecular testing of HFE-HH by physicians[22]. The lack of a primordial family-based screening may indirectly compromise the efficiency of disease prevention in terms of early diagnosis and treatment. A partnership between genetic counselors, medical geneticists, and primary care providers is also essential in order to develop effective policies,

educational tools, and practice guidelines, including the appropriate uses of genetic testing [23].

4.2. Limitations

The fact that a high proportion of participants felt sufficiently informed can be explained by them being members of Patient Associations and thus being more motivated to find information about the disease. **It could be suggested that this is a limitation of the study as it does not necessarily reflect the degree of awareness of HH population in general, eventually compromising the results' generalization. However, we felt that by targeting an already informed population we could identify more effectively the focus of future actions to increase awareness about HH.**

The response rate was not as high as desired and this might limit the generalization of the results. Nevertheless, the Patient Associations' members proved to be a good target population in terms of motivation, participation and geographic diversity. In addition, this study was able to identify some opportunities for stimulating and improving cooperation between associations **and to create a feeling amongst the participants of the study that they are making an active contribution to the activities of their own associations.**

4.3. Conclusion

In conclusion, the present study clearly identified the General Practitioner as being the preferred source for delivering specific information to HH patients about the disease and its implications to their health.

4.4. Practice Implications

According to Acton [24] many physicians have **an** inadequate knowledge about HH diagnosis and the results reinforce the importance of GPs as a source of information about HH**.** Consequently these results may have important applications in future strategies for increasing awareness of the disease.

We confirm that all patient/personal identifiers have been removed or disguised so the patient/person(s) described are not identifiable and cannot be identified through the details of this report.

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Figure Legends

Figure 1

Outline of the sequence for subjects' recruitment, data collection, validation and analysis. From a total of 3973 members from 9 European HH patients' associations invited to participate, 1032 answered the questionnaire, and 1019 surveys were validated. Of these, 895 members stated that they had performed a genetic test and their responses were therefore considered for further analyses.

Table 1
Demographic characteristics of the patients who **have done** a genetic test for HH (n=895)

Characteristics	N	Percentage
Gender		
Male	378	42, 0
Female	517	58,0
Age (years)		
<25	3	0,3
25-35	41	4,6
35-45	142	15,9
45-55	243	27,2
55-65	274	30,6
65-75	166	18,5
>75	26	2,9
Nationality		
Dutch	33	3,7
French	26	2,9
Irish	194	21,7
German	165	18,4
Italian	155	17,3
Norwegian	67	7,5
Portuguese	29	3,2
United Kingdom	206	23,0
Other Country	20	2,2
Yr education (n=760)		
Level 0 – 0 years	46	6,1
Level 1 – 1 to 4 years	8	1,1
Level 2 – 5 to 8 years	29	3,8
Level 3 – 9 to 13 years	201	26,4
Level 4 – Graduate Degree	314	45,3
Level 5 – Postgraduate Education	52	6,8
Level 6 – Other	80	10,5
Occupation (International Standard Classification of occupations)		
Major Group 0 - Armed forces occupation	5	0,6
Major Group 1 - Managers	88	10,2
Major Group 2 - Professionals	264	30,6
Major Group 3 – Technicians and Associate Professionals	66	7,6
Major Group 4 - Clerical support workers	31	3,6
Major Group 5 - Service and sales workers	52	6,0
Major Group 6 - Skilled agricultural, forestry and fishery workers	2	0,2
Major Group 7 – Craft and related trades	30	3,5
Major Group 8 - Plant and machine operators, and assemblers	7	0,8
Major Group 9 - Elementary occupations	43	5,0
Other	77	8,9
None	3	0,3
Unemployed	7	0,8
Retired	188	21,8

Table 2
Information sources about genetic testing and its implications to health, selected by respondents divided according to whether they do or do not consider themselves sufficiently informed.

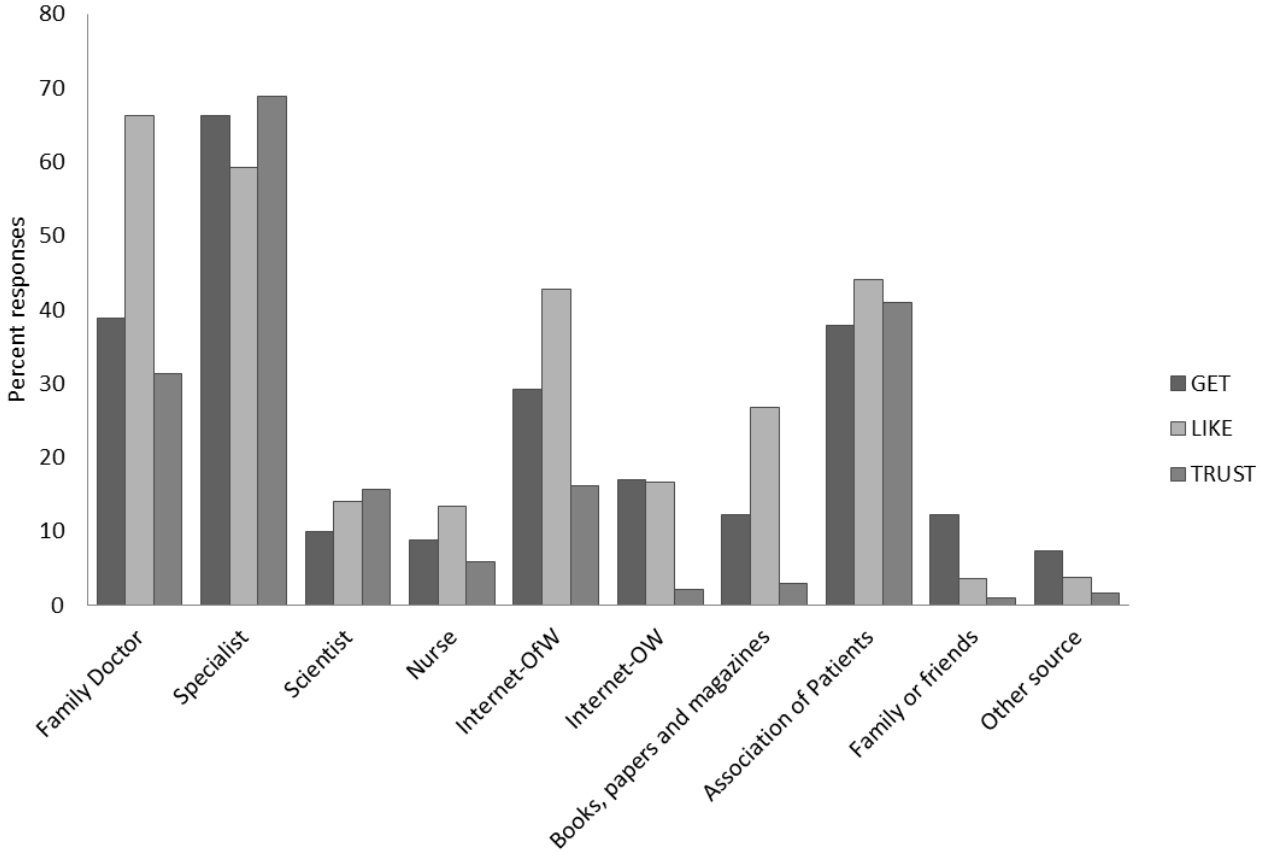
		Sufficiently Informed						Not Sufficiently Informed			
		GET[1] (n=609)		LIKE[2] (n=589)		TRUST[3] (n=579)		LIKE[2] (n=264)		TRUST[3] (n=248)	
Medical Sources	**Family Doctor**	236	**38,8%**	390	**66,2%**	181	**31,3%**	206	**78,0%**	87	**35,1%**
	Specialist	404	**66,3%**	349	**59,3%**	399	**68,9%**	202	**76,5%**	185	**74,6%**
Non Medical Sources	**Internet – Official Websites**	178	**29,2%**	252	**42,8%**	94	**16,2%**	141	**53,4%**	59	**23,8%**
	Internet – Other Websites	103	**17,0%**	98	**16,6%**	12	**2,1%**	38	**14,4%**	11	**4,4%**
	Association of Patients	231	**37,9%**	260	**44,1%**	237	**40,9%**	106	**40,2%**	59	**23,8%**
	Scientist/Researcher	61	**10,0%**	83	**14,1%**	91	**15,7%**	48	**18,2%**	43	**17,3%**
	Nurse	54	**8,9%**	79	**13,4%**	34	**5,9%**	50	**18,9%**	13	**5,2%**
	Books, Papers and Magazines	75	**12,3%**	158	**26,8%**	17	**2,9%**	45	**17,0%**	7	**2,8%**
	Family or Friends	74	**12,2%**	21	**3,6%**	6	**1,0%**	15	**5,7%**	2	**0,8%**
	Other Source	45	**7,4%**	22	**3,7%**	10	**1,7%**	4	**1,5%**	0	**0**

[1] Information sources **used** by respondents to get information about genetic testing and its implications to their health.
[2] Information sources where respondents **would like** to find information about genetic testing and its implications to their health.
[3] Information sources which respondents **trust more** to find information about genetic testing and its implications to their health.

Table 3
Information sources about genetic testing and its implications to other family members, selected by respondents divided according to whether they do or do not consider themselves sufficiently informed.

		Sufficiently Informed						Not Sufficiently Informed			
		GET[1] (n=648)		LIKE[2] (n=624)		TRUST[3] (n=600)		LIKE[2] (n=184)		TRUST[3] (n=177)	
Medical Sources	**Family Doctor**	236	**36,4%**	416	**66,7%**	203	**33,8%**	137	**74,5%**	71	**40,1%**
	Specialist	466	**71,9%**	429	**68,8%**	410	**68,3%**	141	**76,6%**	127	**71,8%**
Non Medical Sources	**Internet – Official Websites**	187	**28,9%**	243	**38,9%**	108	**18,0%**	84	**45,7%**	32	**18,1%**
	Internet – Other Websites	65	**4,2%**	97	**15,5%**	10	**1,7%**	18	**9,8%**	7	**4,0%**
	Association of Patients	273	**17,7%**	295	**47,3%**	240	**40,0%**	83	**45,1%**	45	**25,4%**
	Scientist/Researcher	84	**13,0%**	104	**16,7%**	84	**14,0%**	43	**23,4%**	37	**20,9%**
	Nurse	64	**9,9%**	86	**13,8%**	27	**4,5%**	36	**19,6%**	8	**4,5%**
	Books, Papers and Magazines	67	**10,3%**	129	**20,7%**	17	**2,8%**	25	**13,6%**	2	**1,1%**
	Family or Friends	57	**3,7%**	26	**4,2%**	6	**1,0%**	11	**6,0%**	3	**1,7%**
	Other Source	47	**3,0%**	16	**2,6%**	13	**2,2%**	4	**2,2%**	5	**2,8%**

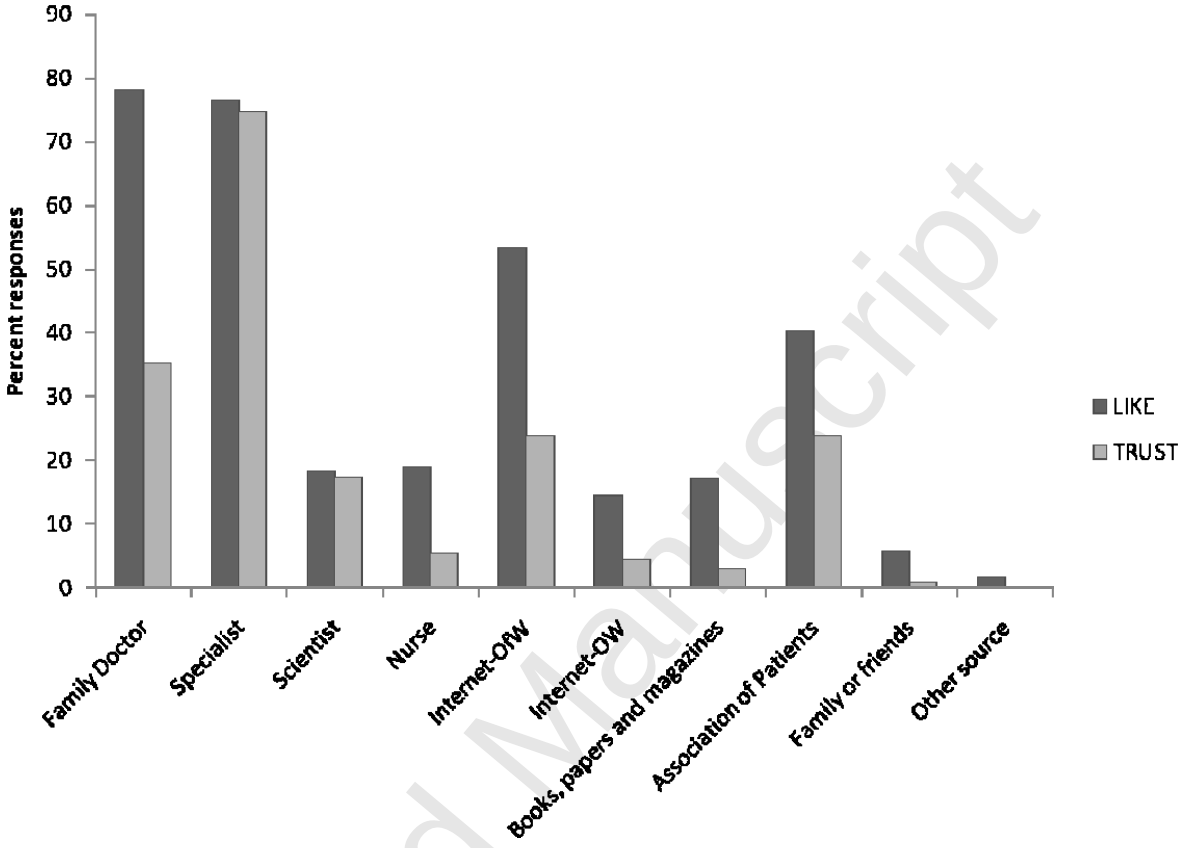
[1] Information sources used by respondents to get information about genetic testing and its implications to other family members.
[2] Information sources where respondents would like to find information about genetic testing and its implications to other family members.
[3] Information sources which respondents trust more to find information about genetic testing and its implications to other family members.



GET = Information sources used by respondents to get information about genetic testing and its implications to their health.

LIKE = Information sources where respondents would like to find information about genetic testing and its implications to their health.

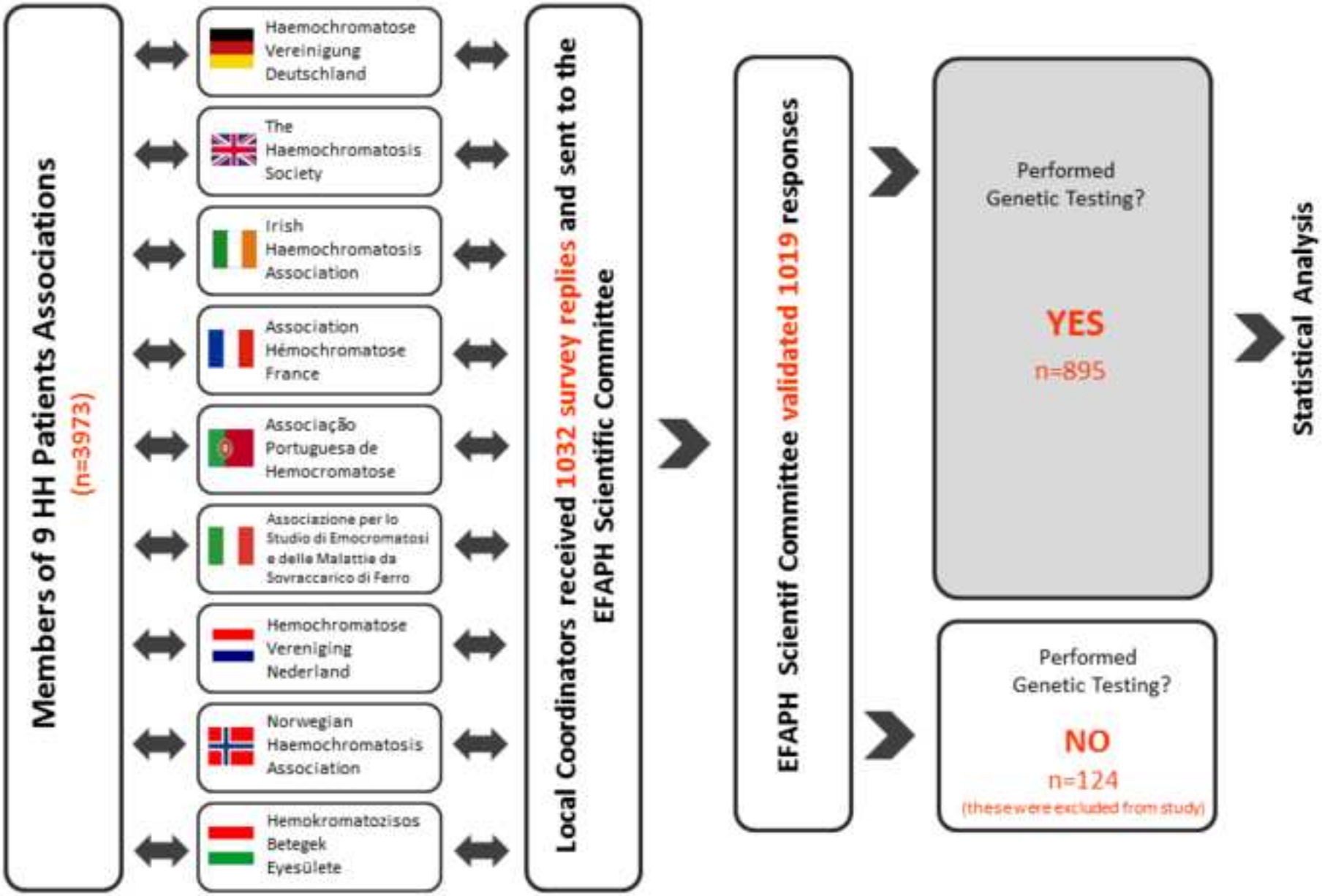
TRUST = Information sources which respondents trust more to find information about genetic testing and its implications to their health.



LIKE = Information sources where respondents would like to find information about genetic testing and its implications to their health.

TRUST = Information sources which respondents trust more to find information about genetic testing and its implications to their health.

Figure 1


Members of 9 HH Patients Associations
(n=3973)
Haemochromatose Vereinigung Deutschland
The Haemochromatosis Society
Irish Haemochromatosis Association
Association Hémochromatose France
Associação Portuguesa de Hemocromatose
Associazione per lo Studio di Emocromatosi e della Malattia da Sovraccarico di Ferro
Hemochromatose Vereniging Nederland
Norwegian Haemochromatosis Association
Hemokromatozisos Betegek Egyesülete
Local Coordinators received 1032 survey replies and sent to the EFAPH Scientific Committee
EFAPH Scientif Committee validated 1019 responses
Performed Genetic Testing?
YES
n=895
Performed Genetic Testing?
NO
n=124
(these were excluded from study)
Statistical Analysis