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Genetic epidemiology literature in Europe—an overview

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Background: Genetic epidemiology deals with the aetiology, distribution and control of disease in groups of relatives, and with inherited causes of disease in populations. The main goal of this overview, part of the collaborative study *SPHERE* (Strengthening Public Health Research in Europe) was to have an up-to-date, detailed summary of the available information for epidemiologists and researchers on the present status of genetic epidemiology in Europe. **Methods:** The PubMed literature search engine was used to recruit papers published in Europe (EU15, EU + 10 and non-EU countries) in this field restricted to the time period of 1 January 1987 and 31 December 2004. **Results:** The number of publications increased significantly in Europe in the period analysed, however, the publication activity was restricted mainly to EU15 countries, with only sporadic papers from EU + 10 countries. Research areas studied are slightly different in Europe and in the USA with a larger emphasis on cancer, mental disease and behavioural disease genetic epidemiology in Europe. **Conclusion:** The aim must be to develop research to support policy in this important field as is already seen in the United States.

Keywords: bibliometry, Europe, genetic epidemiology, literature overview, public health research

Genetic epidemiology is a science that studies the role of genetic factors and their interaction with environmental factors in the occurrence of diseases—typically non-communicable chronic diseases—in human populations.[1] Genetic epidemiology is in close relationship with population genetics, the branch of genetics that deals with frequencies of alleles and genotypes in populations and also deals with selective influences on the genetic composition of the population.

Public health genetics creates an interface between genetics and public health. Public health genetics is directive, achieving health gains for the population as a whole, using genetic epidemiology and focusing on improving population health by collectively organized interventions. Public health genetics is a rapidly emerging field that requires competence in genetics, medical genetics, clinical genetics, epidemiology and public health.

Until now, there has been a lack of information on the situation in the field of genetic epidemiology in Europe. This overview, undertaken within the collaborative study *SPHERE* (Strengthening Public Health Research in Europe) was to produce a detailed summary of the available information for epidemiologists and researchers on the present status of genetic epidemiology in Europe.

Materials and methods

A literature search was undertaken using the PubMed database (which included all papers found in Embase). The keyword 'Genetic epidemiology' was used, as shown in table 1. The search was extended by using the names of particular diseases (table 1). Limitations were made for countries of Europe, for papers with an abstract written in English, including both reviews and original papers, and where an abstract was available. The algorithm for scientific paper recruitment is shown in figure 1.

Databases were searched until 31 December 2004, with no specified start date. All publications yielded by the searches are included in the results. There were no further inclusion or exclusion criteria. Country of corresponding author (or first author if separate corresponding author was not identified) was used to allocate publications to countries. Results are presented by EU countries in 2005 (EU25), EU in 2003 (EU15) and the 2004 accession countries (EU + 10) where appropriate, and for specific non-communicable diseases. Comparisons are made with the USA.

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Results

Keyword combination 'genetic epidemiology' appearing anywhere in the text

The total number of peer-reviewed publications produced by this search was 2518. This included all those published in the journal Genetic Epidemiology until 31 December 2004, of which only 625 (24.8%) were published by European research groups. Almost all these European papers (603, 96.5%) were published from EU15 countries.

Six hundred and four (96.6%) of the European papers were written in English, only one paper was published in Russian, one in Serb, one in Spanish and there were seven in French. Five hundred and ninety-four (95%) of the European peer-reviewed papers had an abstract available in PubMed. Just under half (46.6%) of papers in English were published between 1 January 2000 and 31 December 2004, i.e. in the last 5 years of the analysed period. Approximately 20% of the publications were reviews, of which just under half were written between 1 January 2002 and 31 December 2004. The UK published 223 (35.6%) of the European papers in this field, and France 116 (18.6%) of the papers, followed by Finland, the Netherlands and Denmark with 57 (9%), 39 (6%) and 32 (5%) papers, respectively.

The number of studies carried out in Europe steadily increased. However, there were very few papers from the EU + 10 countries and outside of EU even by the end of the analysed period. For many countries from outside the EU and in the EU + 10, there were no papers at all on genetic epidemiology using this search and only one research group from the former USSR published a paper in this field.

Keyword combination 'genetic epidemiology' appearing in the title or abstract of the paper

Papers with the phrase 'genetic epidemiology' appearing in the title or abstract were collected and analysed separately.

A total of 647 papers were published from all over the world by 31 December 2004, with 160 of those (24.7%) produced by European countries. Most of the papers (95.6%) were produced by EU15 countries, mainly by research groups from the UK (34.6%), France (18.3%), the Netherlands (9.1%) and Germany (7.8%). Again, the proportion of publications of EU + 10 countries was very low (only 1.9% of the total European papers). (One paper was published from the Czech Republic, and two from Slovenia). Of the total European articles, 94.4% were written in English, one in Russian, one in Spanish and seven in French. The first paper was published in

Table 1 Keyword combinations used in the literature search

1	'genetic epidemiology.
2	'genetic epidemiology' [TIAB][a]
3	'genetic epidemiology' AND 'population based'
4	'Genetic epidemiology' [TIAB][a] AND (cardiovascular OR 'coronary heart disease' or 'coronary heart diseases')
5	'genetic epidemiology' [TIAB][a] AND (cancer OR malignant OR tumour OR tumor OR carcinogenesis)
6	'genetic epidemiology' [TIAB][a] AND diabetes
7	'genetic epidemiology' [TIAB][a] AND (mental OR mood OR psychiatric OR Alzheimer OR schizophrenia OR dementia OR depression)
8	'genetic epidemiology' [TIAB][a] AND ('respiratory disease' OR 'respiratory diseases' OR asthma OR COPD OR emphysema)
9	'genetic epidemiology' [TIAB][a] AND (alcohol[b] OR drug OR 'drug abuse' OR smoking)

All search were restricted to human studies
a: [TIAB]; search is done only in the title and/or abstract
b: In any form

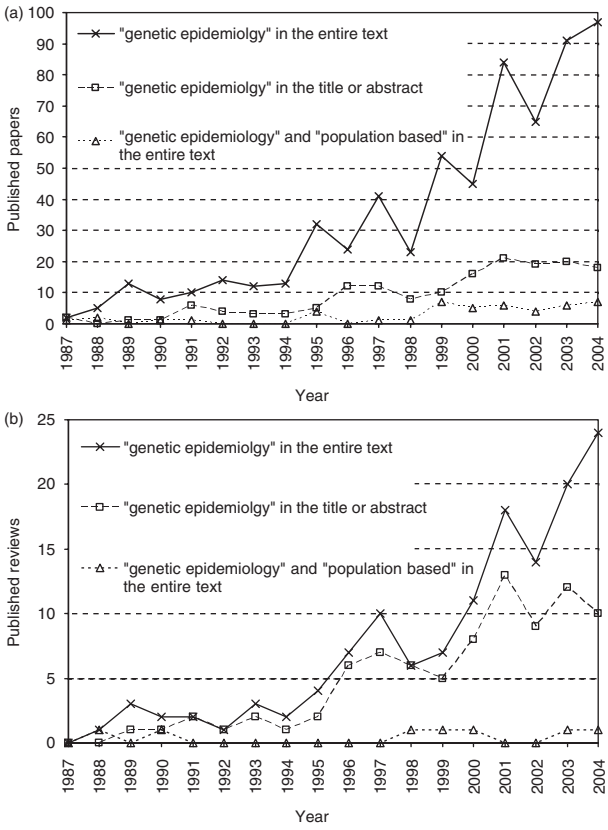
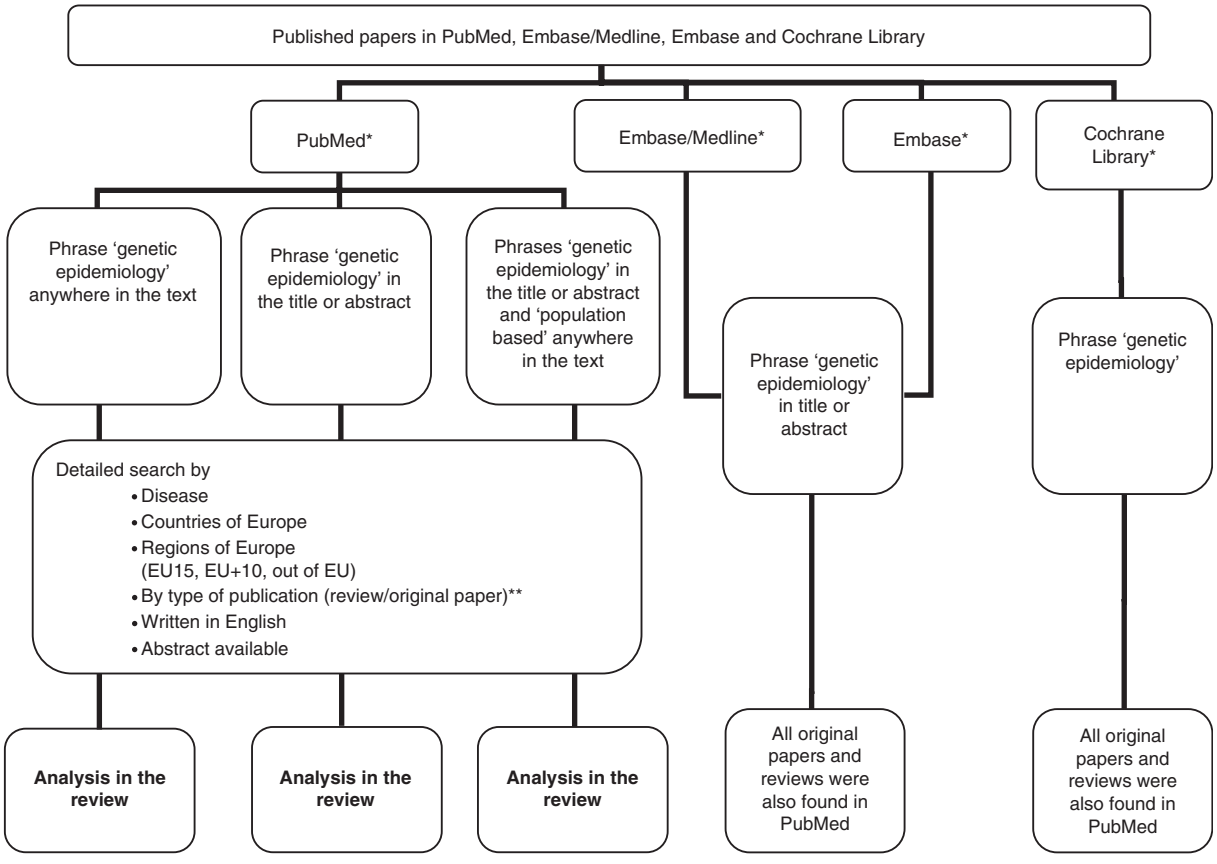


Figure 2 Trends of publishing of original articles (a) and reviews (b) on genetic epidemiology in Europe, 1987–2004



Legends: *Restricted to human studies ** applied filters: 1) all published papers until 31 Dec, 2004, 2) only reviews published until 31 December 2004, 3) reviews published between 1 Jan 2002 and 31 Dec 2004 (last 3 years of the analysed period)

Figure 1 The algorithm of the scientific paper recruitment for the literature overview

1987 in Europe, 91.7% of the English-written articles had an abstract and 47% were published between 2000 and 2004. Of the publications, 53.6% were reviews or tutorials with just over one-third of these published very recently (2002–04). Again, of the reviews, most European countries from the groups of EU + 10 and non-EU countries had no publications in this field, and there were no publications from Luxembourg or Austria.

Although the number of papers recruited in this second search was lower, the main trends are similar to those found in the initial search: published original papers are mainly affiliated to EU15 countries; and less than 5 papers were produced from EU + 10 countries or in European countries outside the EU. No reviews concerning genetic epidemiology were published by any European country outside the EU15 until the end of 2004 (figure 3).

Keyword combinations 'genetic epidemiology' appeared in the title or abstract of the paper and 'population based' anywhere in the text

There were 170 papers with the keywords 'Genetic epidemiology' appearing in the title or abstract and 'Population based' appearing anywhere in the text. Of these, 46 (27%) were published in the European region. Again almost all were published in the EU15 (figure 3). The UK had the highest number of publications for this search (14 papers; corresponding to a third of all papers published in EU15). Apart from a French paper, all 170 papers were written in English, all had an abstract available in PubMed and all were published between 2001 and 2004. The proportion of reviews in this search was relatively low: 23 papers (13.3%).

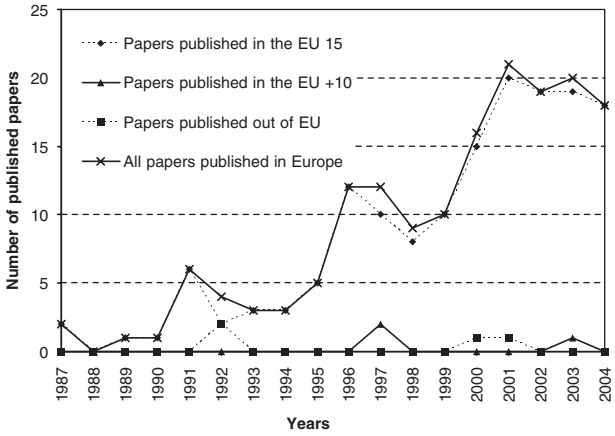


Figure 3 Trends of publishing original articles with the keyword 'Genetic epidemiology' appearing in the title or abstract by European regions between 1 January 1987 and 31 December 2004

Comparison by disease areas

Figure 4 shows the findings on disease areas for research on non-communicable disease genetic epidemiology. Most of the papers were on cancer epidemiology in Europe and mental disease epidemiology in the USA. In second place was mental disease genetic epidemiology in Europe and cancer epidemiology in the USA. Behavioural disease epidemiology and papers on the genetic epidemiology of cardiovascular diseases, diabetes and respiratory disorders also ranked highly in both the USA and Europe.

Comparisons by country

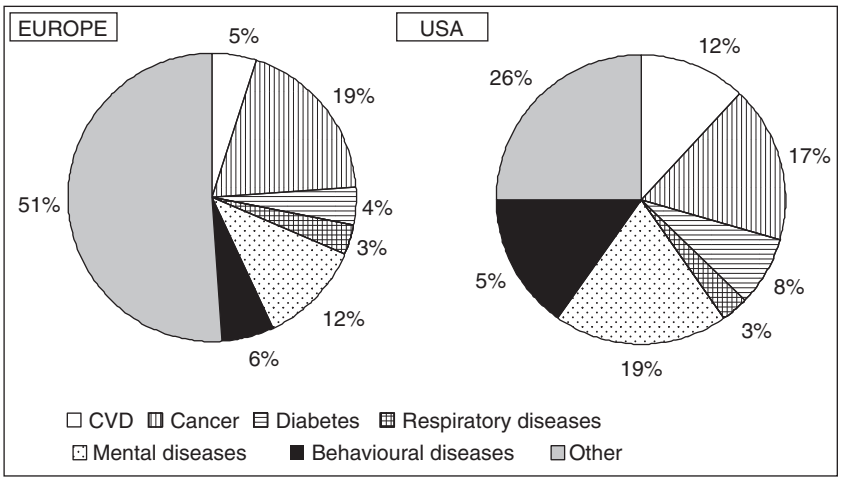
EU15 countries had a dominant role in successful publishing for almost every specific field of genetic epidemiology in Europe. Almost all countries had at least one published paper on genetic epidemiology from this group. Norway seems to be the most active from the non-EU countries, with publications on the genetic epidemiology of almost every disease type involved in this overview. In the EU + 15 regions Czech Republic and Slovenia published the most.

However, some countries had no publications at all on genetic epidemiology: Luxembourg and Austria from the EU15 group, Cyprus, Estonia, Hungary, Latvia, Lithuania, Malta, Poland, Slovakia from the EU + 10 subgroup and from the non-EU member states only Norway and Russia were active in publishing.

Discussion

Public health implications

This overview of genetic epidemiological research using PubMed spans 1987 (when the first paper was published in Europe) to 2004. The number of publications increased significantly in Europe over the period analysed. However, publication was almost entirely from EU15 countries.



Legend: CVD, cardiovascular disease

Figure 4 Diseases in genetic epidemiology research published in Europe ($n = 160$) and in the USA ($n = 201$) between 1 January 1987 and 31 December 2004

One might conclude that either there is no genetic epidemiological research in these absent countries, or that the lack of publication was due to the disadvantaged situation of the new EU member states, with low GDP. There are only a few journals with a significant impact factor that publish papers in languages other than English. Epidemiological projects mainly depend on external funds, EU grants were of limited availability before the extension of the EU in 2004. Equally, EU10 governments put their priority on basic and applied research in previous years.

Moreover, research groups from western countries have had a high success rate in publishing, even recently. The hard and rigorous reviewing process of publishing is challenging to the research groups that are not native English speakers. According to the data derived from the official website of British Medical Journal (a journal that may be considered Euro-centric), the general acceptance rates of central and eastern European countries are extremely low.[2] The most success in publishing is by research groups from the American continent and from western Europe.

It seems obvious that European research groups are more successful in publishing in Euro-centric journals than in journals published in the USA. However, the scientific impact factors of the American edited journals are generally higher in this field, so that European research teams in genetic epidemiology are disadvantaged in publishing original papers from this point of view too. Research activities on genetic epidemiology have a dominant focus on the non-communicable diseases, which reflects their public health importance all over the world. Research areas studied are slightly different in Europe than in the USA, with a higher emphasis on cancer, mental disease and behavioural disease genetic epidemiology in Europe, as compared to the USA.

Implications of the overview

More recently, European genetic epidemiological research teams have published papers describing the strength and association of certain inherited and/or acquired gene alterations predisposing to any of the non-communicable chronic diseases in this field. Gene therapy has become of higher impact in genetic studies, as an optional and available therapy for high-risk groups. Methods of genetic studies are changing, with traditional genetic studies becoming insignificant as genomic investigations have come to the fore. For example, microarray techniques can provide a genome-wide complex analysis to establish genetic predisposition for certain diseases.

The increasing number of the EU grants available for European research teams, and other financial possibilities to support genetic/genomic investigations are welcome. The aim should be to develop research support for these important and valuable specialized research activities similar to that already seen in the USA.

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Conflict of interest: None declared.

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