Article

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[Intervention Protocol]

Interventions to improve patient access to and utilisation of genetic and genomic counselling services.

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ABSTRACT

This is the protocol for a review and there is no abstract. The objectives are as follows:

Primary objective

The primary objective is to assess the effectiveness of interventions to improve patient identification, access to and utilisation of genetic and genomic counselling services when compared to:

i) No intervention;

ii) Usual or current practice; and

iii) Other active intervention.

Secondary objective

The secondary objective is to explore the resource use and costs associated with interventions aimed at improving patient identification, access to and utilisation of genetic and genomic counselling services from studies meeting the eligibility criteria.

We will report on factors that may explain variation in the effectiveness of interventions aimed at improving patient identification, access to and utilisation of genetic and genomic counselling services from studies meeting the eligibility criteria.

Another secondary objective is to explore how interventions which target improved patient identification, access to and utilisation of genetic and genomic counselling services affect the subsequent appropriate use of health services for the prevention or early detection of disease. It is also possible that the genetic counselling interaction itself will contribute to the possible use of preventative services.

BACKGROUND

Description of the condition

In 1963 an expert committee of the World Health Organisation observed the importance of genetics in health by stating, "Genetic considerations add a new dimension to public health work: a concern not only for the health and well-being of persons now living, but also for generations yet to come" (WHO 2015). The Council of Europe recommends that all countries develop a coherent and comprehensive national policy framework for genetic services, making adequate genetic counselling available in an equitable manner (Council of Europe 2015). They state that, "the development of genetics in health care services has a major impact on the organisation of health care, leading to shifting from curative to preventive services, from in-patient to out-patient treatment, from specialised genetic services to genetics as an integral part of general health services".

Genetic and genomic technologies have the potential to provide vital insight to support the accurate prevention, diagnosis and treatment of disease (HGSG 2012). It is estimated that 5.5% of the UK population will develop a genetic disorder by age 25, affecting 2.8 million people (Genetic Alliance UK 2012). Patients and families affected by rare diseases face lengthy delays in accessing a correct diagnosis. For example, almost half (46%, 221/481) of patients in a UK survey had to wait over one year for a final diagnosis following the onset of disease symptoms (Rare Disease UK 2011). Data from the 12,000 voices European survey of rare diseases show that there are inequalities in access to genetic healthcare for diagnosis and on-going treatment (EURORDIS 2009). Research has shown that social, environmental and economic conditions may deter individuals or communities from accessing the benefits of these new technologies (WHO 2010; Burton 2011; Genetic Alliance UK 2012; Bellcross 2013; Delikurt 2014).

Over the past 10 years genetic counselling has started to move away from only being offered in genetic centres of excellence and is now being accessed through new service delivery models. These service delivery models involve mainstream clinical specialties such as cancer or cardiac care (Burton 2011). Battista 2012 reviewed the current organisation of genetic services in Europe, North America and Australia and found that genetic counselling services relied heavily on co-ordination of activities between professionals and new ways of working required the reconfiguration of professional roles and responsibilities. Barriers to introducing new service models included redistribution of roles, sharing of data and the lack of preparedness of non-genetics professionals and healthcare systems (Battista 2012). There is also evidence from professional surveys that variation exists in models of service delivery within the same country (Cohen 2013).

To date there have been few studies aimed at determining the effectiveness of interventions to help patients access genetic services. The body of research evidence is developing rapidly and further studies are currently ongoing or in the feasibility stage (Hodgson 2014). In order for patients and their families to benefit from genomic advances they need to be identified, referred to and then supported to use the services offered. This review is needed to provide a benchmark of existing evidence and aims to describe the effectiveness of interventions aimed at identifying potential patients and enabling them to access and use genetic and genomic counselling services.

Definitions

There remains confusion over how the terms 'genetics' and 'genomics' are used within studies and also by clinicians and scientists. Until the late 1990s, most clinical services used the term genetic services or medical genetics services. After this, the term genomics was applied to some clinical services. The genome has been described as 'an organism's complete set of DNA, including all of its genes and non-coding regions' (Genetics Home Reference 2015). The following definitions reported by the UK Nursing and Midwifery task-force will be used for this review (Nursing and Midwifery Council 2011).

Genetics

Genetics is the study of heredity and variation. In the healthcare setting this has been associated with single gene and chromosomal conditions traditionally managed by specialist genetics services.

Genomics

Genomics is the study of the structure and function of the genome, including the interaction between genes and the environment.

Genomic healthcare

Genomic healthcare involves the use of genomic information and technologies at any stage of the healthcare continuum to determine disease risk and predisposition, diagnosis and prognosis, and the selection and prioritisation of therapeutic options. Genomic healthcare also takes into account the potential ethical, psychological and social implications of genomic information and the application of genomic technologies. The term 'genomic healthcare' incorporates the use of both genetic and genomic information and technologies (Nursing and Midwifery Council 2011).

The process of genetic and genomic counselling

There are varying definitions of the process of genetic and genomic counselling. This review will define the process of genetic and genomic counselling using the definition provided by the United States National Society of Genetic Counselors (Resta 2006).

"Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following:

• Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.

• Education about inheritance, testing, management, prevention, re-

sources and research.

• *Counseling to promote informed choices and adaptation to the risk or condition".*

Genetic and genomic counselling services

This review will define the term genetic and genomic counselling services as services that offer the process of genetic or genomic counselling or both as part of service provision. In this review genetic and genomic counselling services are broadly defined as, patient-facing products or services designed to support or inform the patient regarding the risk of carrying, developing, or being affected by health conditions which have a genetic or genomic aetiology. Genetic and genomic healthcare services can consist of both patient-facing and laboratory elements, employing a multidisciplinary workforce consisting of both clinical health professionals and laboratory scientists.

This review will include studies investigating the effectiveness of interventions aimed at identifying potential patients and enabling them to access genetic or genomic counselling services. These services are available in most countries and patients have a right to access them. However, there is great variation in how they are organised, delivered and funded (WHO 2010; Trepanier 2014). Increasingly, genetic and genomic counselling services are situated within other clinical specialties, such as a breast cancer family history clinic operating as part of an oncology service (Burton 2011; Kirk 2014). This has been termed 'mainstreaming' of work traditionally undertaken by genetic services or the use of genetic knowledge within other clinical specialties. Services are adapting to this challenge by developing new ways of working to meet the demand for genetic and genomic counselling services (Eeles 2007; Middleton 2014).

The genetic counselling interaction

This review is not aimed at determining the effectiveness of the clinical genetic counselling interaction between the health professional and the patient. Interventions which solely target the patient's psychological function or knowledge of the medical aspects of their genetic condition will be excluded from this review. Interventions which target patient identification, access to and use of genetic and genomic services are the focus of this review. Usually these activities occur prior to the genetic counselling interaction event.

Health professionals who provide genetic counselling services

The professional role and training of clinical health professionals providing the genetic counselling interaction vary. Health professionals undertaking genetic counselling include but are not limited to, medical clinical geneticists, genetic counsellors and genetic nurses. In the United States there is a credentialing system which regulates genetic nurses (ANCC 2015). In the United States, Canada, Australasia, South Africa and the United Kingdom genetic counsellors have an established training, certification, and regulatory system (ABGC 2015; CAGC 2015; GCRB 2015; HPCSA 2015). Across Europe, the European Board of Medical Genetics have introduced a European genetic counsellor registration system for health professionals from a variety of backgrounds (Skirton 2010).

For the purposes of this review, studies investigating genetic and genomic counselling services which offer the process of genetic counselling by a range of health professionals will be included. When describing the effectiveness of the interventions included in this review, mediating factors related to professional role, preparation and training will be considered when studies are reported and acknowledged in the data analysis.

Description of the intervention

This review will include studies investigating a variety of interventions that target patient identification, access to and utilisation of genetic and genomic services which provide genetic and genomic counselling. Interventions that address these issues could be targeted at the general population, health professionals (e.g. genetic specialist or non-genetic specialist), clients of health professionals or a combination of these.

Interventions could be categorised in a number of ways. We will consider grouping interventions into those which aim to target potential patients directly (e.g. publicity campaigns) and those which target health professionals (e.g. clinical guidelines). Alternatively, if appropriate within the above categories, we may categorise studies by the mode of delivery (e.g. printed or electronic guidelines) or healthcare setting (e.g. primary, secondary or tertiary care).

Interventions could range from educational interventions (Westwood 2012), to behaviour techniques to improve communication of test results to at-risk relatives (Hodgson 2014). Interventions may be targeted at the organisational level, such as doctor or nurse role substitution (Torrance 2006). The interventions included in this review will represent all those described by the Cochrane Effective Practice and Organisation of Care (EPOC) group taxonomy (EPOC 2002), and will include the following:

- Professional interventions - Those directed to professionals to change their practice or behaviour;
- Financial interventions - These will include both health provider and patient interventions;
- Organisational interventions - Including both health provider and patient orientated interventions;
- Structural interventions - Including where services are delivered; and
- Regulatory interventions - Including regulation or adoption of certain genetic tests (EPOC 2002).

Table 1 shows the possible links between interventions, how these interventions might work, based on the application of the theoretical domains framework (Cane 2012); and the outcomes of interest for this review which include improving identification, access to and use of genetic and genomic counselling services.

Table 1 - Linking interventions, possible behaviour change modes of action and review outcomes

Interventions to improve patient identification, access and use of genetic and genomic counselling services	
Description of interventions likely in genetic services	**Possible Mode of Action - based on the 14 categories of the theoretical domains framework** (Cane 2012)
Health professional attendance at educational workshops aimed at increasing identification of patients at genetic risk	• knowledge • skills • professional role
Reducing the cost of genetic tests to the health professional, patient or organisation	• environmental context and resources • social influences • beliefs about consequences
Revision of professional roles, role substitution	• environmental context and resources • knowledge • skills • professional role
Change of where services are delivered (e.g. from specialist services to primary care)	• knowledge • skills • professional role • environmental context and resources • social influences
Adoption of a genetic test by regulators (e.g. Preimplantation Genetic Diagnosis for *BRCA1*)	• knowledge • skills • belief about capabilities • belief about consequences • social influences

How the intervention might work

Recognising which individuals are at increased genetic risk and who would benefit from access to genetic counselling services is a key facilitator in patients actually utilising services (Delikurt 2014). Many of the interventions targeted at non-genetic health professionals involve educational training and skills development in order for them to recognise at risk individuals. Many health professional organisations and policy groups have published competency standards in genetics, however it is unclear if these have improved patient identification or access to services (Kirk 2003; NCHPEG 2007; ANA 2009). Evidence in the medical education literature show a number of theories relating to the success of delivery of continuing practice development with varied effects on clinical patient care (Schostak 2010). Behaviour change interventions, such as empowering patients to inform relatives who are at genetic risk may facilitate patient access to services. If a study reports a theoretical rational for the mechanism of the intervention, this will be reported (Craig 2008). It is acknowledged by the theory of planned behaviour that intention to perform a behaviour could be a predictor of actual behaviour change. Therefore studies targeting intention to perform certain behaviours as defined in the Primary outcomes section will be included in this review.

Other interventions, such as the introduction of clinical guidelines, or establishing clinical pathways which include referral to a genetic counselling service, will be included but the success of these interventions in other areas of healthcare is variable (Grimshaw 2004). This review will include any interventions targeting any stage of the patient pathway from interventions which aim to improve patient identification, referral to or use of genetic and genomic counselling services or those interventions which target multiple elements (e.g. health professional educational sessions might improve either identification of at risk individuals or referral or both). A service reorganisation might help improve access. It is possible that some interventions may have the adverse effect of re-

ducing appropriate referrals or deter patient utilization of services. These outcomes, and any other adverse events reported in the included studies, will be reported in the review. We will examine study outcomes related to interventions which target the general population, patients, health professionals or organisations.

Why it is important to do this review

Access to genetic services is seen as an essential part of the health care system in most developed countries. However, there are only two Cochrane reviews which address provision of care for genetic conditions. Hilgart 2012 reviewed interventions which helped assess risk in individuals already identified as at-risk for familial breast cancer. Cox 2013 reviewed interventions promoting physical activity in patients with cystic fibrosis. A review regarding the effectiveness of preconception genetic risk assessment for a subset of genetic conditions is currently ongoing (Hussein 2013).
The Delikurt 2014 systematic review of interventions aimed at increasing patent access indicated a number of barriers to patient referral among non-genetic health professionals including lack of awareness of patient's risk factors, failure to obtain adequate family history, lack of knowledge of genetics and genetic conditions, lack of awareness of genetic services, inadequate coordination of referral and lack of genetics workforce. No reviews of the effectiveness of interventions aimed at identifying patients, and enabling them to access and use genetic and genomic counselling services have been carried out.

OBJECTIVES

Primary objective

The primary objective is to assess the effectiveness of interventions to improve patient identification, access to and utilisation of genetic and genomic counselling services when compared to:

i) No intervention;

ii) Usual or current practice; and

iii) Other active intervention.

Secondary objective

The secondary objective is to explore the resource use and costs associated with interventions aimed at improving patient identification, access to and utilisation of genetic and genomic counselling services from studies meeting the eligibility criteria.

We will report on factors that may explain variation in the effectiveness of interventions aimed at improving patient identification, access to and utilisation of genetic and genomic counselling services from studies meeting the eligibility criteria.

Another secondary objective is to explore how interventions which target improved patient identification, access to and utilisation of genetic and genomic counselling services affect the subsequent appropriate use of health services for the prevention or early detection of disease. It is also possible that the genetic counselling interaction itself will contribute to the possible use of preventative services.

METHODS

Criteria for considering studies for this review

Types of studies

We will include randomised controlled trials (RCTs) and cluster-RCTs comparing the intervention to usual practice, a control intervention or other active intervention. In line with EPOC recommendations (EPOC 2015a), cluster-RCT studies will only be eligible if there are at least two intervention sites and two control sites. We will include studies regardless of language or publication status (i.e. abstract, full text, unpublished data).

Types of participants

We will include studies of interventions targeted at the general population, health professionals, health or social care organisations or patients with the aim of improving patient identification of, access to and use of genetic and genomic counselling services in primary, secondary and tertiary care service settings and those transitioning between healthcare settings.
Participants will include:

- the general population;
- patients of all age groups (including but not limited to genetic groupings, 'pre-natal' or 'reproductive', 'adult', 'cancer' and 'paediatrics');
- health professionals including genetic and non-genetic specialists; and
- organisations providing health or social care.

Eligible health professionals include, but are not limited to, doctors, genetic counsellors, nurses, physiotherapists, pharmacists, occupational therapists, social workers, dieticians, psychologists, and dentists if involved in the genetic counselling process. Genetic specialists include consultant medical clinical geneticists, genetic counsellors, or genetic nurse specialists. Studies aimed at multidisciplinary providers and organisations will also be included.
We will exclude studies directed at healthcare scientists involved in non-patient facing services (e.g. the laboratory techniques of genetic testing and studies with no patient facing contact such as a change in laboratory techniques).

Types of interventions

We will consider any intervention to improve potential patient identification, access to and utilisation of genetic and genomic services as defined in the Description of the condition section compared to no intervention, usual care or practice or other active intervention. These active interventions may include professional, financial, organisational, structural or regulatory interventions as described in the EPOC taxonomy (EPOC 2002).

We will include studies of interventions that target the health behaviour of individuals such as patient utilisation of health services outside genetic counselling services, health screening (e.g. mammography or diabetes monitoring) and health management (e.g. prophylactic surgery) only if the patient had previously accessed genetic or genomic counselling services and were made aware of an increased health or reproductive risk. We will include studies of organisational interventions such as displays of leaflets and posters or awareness campaigns.

We will include studies that involve a mixture of interventions. In these situations outcomes will be reported if they are associated with a specific type of intervention alone, otherwise they will be reported under the category of complex interventions. It is expected that most interventions within the area of interest will be complex interventions as described by the Medical Research Council (Craig 2008).

We will exclude studies if the intervention solely focuses on laboratory processes or new technologies. However if the study aims to assess the effectiveness of a complex intervention which is designed to target the patient care pathway, and includes appropriate genetic testing it will be included. We will also exclude studies where the intervention is intended to identify potential patients, their access to and use of somatic genetic testing (e.g. tumour testing to inform treatment), microbial genetics, applied infection genetics or biomedical genetic testing not applied to healthcare. Interventions which aim to target patient level psycho-educational outcomes, such as improved education about the condition or decreased psychological stress will be excluded if they are not assessed using one of the primary outcome measures specified for this review.

Types of outcome measures

Primary outcomes

We will include studies that consider at least one of the five primary outcome measures listed in Table 2. We will include studies that sought to measure one of the primary outcomes even if data were not reported. Studies not targeting these outcomes will be excluded.

Table 2 - Primary Outcomes of interest and examples of possible outcome measures

Primary outcome of interest	Examples of possible outcome measures
1. Increased identification of people who are at genetic risk or who may benefit from genetic or genomic counselling services	• Number or proportion of patients within a population identified at increased risk • Increased communication or intention to communicate genetic risk to relatives • Rate of health professional adherence to guidelines (e.g. NICE familial breast cancer)
2. Increased access to genetic or genomic counselling services	• Reduction in waiting times for clinic • Number or proportion of referrals from primary care to genetic or genomic counselling services • Proportion of minority cultural and linguistic groups within the population (e.g. black and ethnic minority groups, d/Deaf individuals')
3. Increased appropriate utilisation of genetic or genomic counselling services	• Failure to attend rate for genetic or genomic counselling services • Uptake of genetic tests
4. Increased access to or use of other appropriate healthcare services after genetic or genomic counselling services	• Referral rate and utilisation or intention to utilise appropriate evidence based mammography screening • Actual compliance or intention to engage with appropriate preventative health service, such as diabetes management

(*Continued*)

5. Resource use and costs	• Out of pocket patient costs • Direct healthcare resource

Secondary outcomes

We will consider any adverse events and the equity outcomes reported in the included studies as secondary outcomes. We will use an 'equity lens' to determine how factors associated with disadvantage (social stratification) might interact with the hypothesised mechanisms of action of the intervention (Ueffing 2012; Tugwell 2010). We will exclude studies that only report on secondary outcomes.

Search methods for identification of studies

Electronic searches

In consultation with the authors, the EPOC trials search coordinator drafted a sensitive search strategy designed to retrieve studies from electronic bibliographic databases. We will search the following databases:

- MEDLINE via OVID (1946 to July 2015);
- Cochrane Library via Wiley (Issue 7, 2015) including CENTRAL;
- Database of Reviews of Effects (DARE);
- Health Technology Assessment database (HTA);
- EMBASE via OVID (1947 to July 2015);
- CINAHL via Ebsco (1980 to July 2015);
- PSYCINFO via OVID (1867 to July 2015);
- Genetics Abstracts via Proquest (1990 to July 2015); and
- Pubmed related articles search of key papers.

The searches for this review will be developed and carried out in accordance with current guidance from the Cochrane Collaboration (MECIR 2013). The Cochrane RCT Sensitivity and Precision Maximising Filter (Lefebvre 2011), will be used for the MEDLINE and EMBASE searches. We will not apply any study design filters to the other database searches. We will not apply any restrictions based on language, publication type, or publication year. The search strategy will be devised for the OVID MEDLINE interface and then adapted for the other databases.
The MEDLINE search strategy is reported in Appendix 1

We will also search for economic studies using the NHS CRD economics search filter (CRD 2014). This search will be used to identify published economic evaluations and cost analyses meeting the primary objectives and inclusion criteria for the review.
We will pool all titles and abstracts and delete duplicates.

Searching other resources

We will search the reference lists of the included studies and any relevant systematic reviews to identify additional studies. Relevant individuals and organisations will be consulted for information about unpublished or ongoing studies. We will search the clinicaltrials.gov study register to identify ongoing studies.

Data collection and analysis

Selection of studies

The titles and abstracts of the studies identified by the literature search will be independently screened by two members of the review team to assess which studies meet the inclusion criteria. One screener will be a review author (CB, JC, LT, HS, KC, SG, RB). Next, we will retrieve full-text copies of all potentially relevant papers and these studies will be independently assessed by two review authors (CB, JC, LT, HS, CP, KC, SG, RB) to assess eligibility. Disagreements will be resolved through discussion and consensus or by input from a third review author as necessary.
We will report data on the number of retrieved references, the number of obtained full-text papers and the number of included and excluded studies based on the Preferred Reporting Items for Systematic Reviews and Meta-Analysis (PRISMA) flowchart guidelines and include a 'Characteristics of Excluded Studies' table (Moher 2009). Studies will be managed in Endnote X7 (Endnote 2014), and Review Manager (Review Manager 2014).

Data extraction and management

Two authors (CB, JC, LT, HS, CP, KC, SG, RB) will independently extract data using a standardised data extraction sheet (Appendix 2), based on the generic EPOC data collection checklist (EPOC 2015b), including:
• **Mode of delivery** e.g. one-to-one face to face or group based face to face or web-based face to face sessions or web-based self

study (video/ DVD); type of health professional delivering the intervention (e.g. doctor or genetic counsellor);

• **Setting and process** e.g. primary care or secondary care; duration of intervention (including length and number of sessions and period over which intervention delivered); incentives or reimbursement of staff;

• **Theoretical basis** classified using the 14 categories of the Theoretical Domains Framework (Cane 2012), and defined by Michie 2010 and Michie 2013; and

• **Content** e.g. genetic counselling or genetic information giving, family history awareness, risk assessment, psychosocial support, family communication, health behaviour change management, screening advice, access to genetic testing, interpretation of genetic tests.

Data will be extracted directly from study reports. Before investigating whether data can be standardized across studies (e.g. number of patients identified per 100,000 population), we will report data in natural units as reported by the investigators. Any unresolved differences in data extraction will be referred to a third author. Where necessary additional information will be sought from the authors of the primary studies. All relevant data will be entered into the RevMan5 software (Review Manager 2014).

We will collect contextual data to consider if any characteristics of the populations (including settings and contexts) could be used as explanatory factors. This is described further in the Assessment of heterogeneity section.

We will identify studies that have reported collecting primary resource use and cost data and summarise these studies in terms of interventions, study population, including the relevant country for the analysis; study perspective, time horizon, method used to identify resource use data, method used to identify cost use data; method used to assimilate resource use and cost data, key findings, key uncertainties and limitations of the analysis. Economic studies will be classified as full economic evaluations (e.g. cost effectiveness analysis, cost utility analysis or cost benefit analysis) or partial evaluation studies (e.g. cost minimisation analysis or cost analysis). Within this classification, we will summarise model, observational and trial-based studies that have identified primary resource use and cost data.

Assessment of risk of bias in included studies

Two review authors (CB, JC, CP, LT) will independently assess the risk of bias for all eligible studies using the criteria described in the EPOC group resources for review authors. The risk of bias will be assessed using the Cochrane risk of bias tool (Higgins 2011a), and in accordance with the EPOC group's 'Guidance on Risk of Bias' (EPOC 2013a). We will compare results and resolve discrepancies by discussion and consensus.

The EPOC group's guidance on assessing risk of bias consists of nine criteria for all RCTs:

- Was the allocation sequence adequately generated?;
- Was the allocation adequately concealed?;
- Were baseline outcome measurements similar?;
- Were baseline characteristics similar?;
- Was the study adequately protected against contamination?;
- Were incomplete outcome data adequately assessed?;
- Was knowledge of the allocated interventions adequately prevented during the study?;
- Was the study free from selective outcome reporting?; and
- Was the study free from other risk of bias?

The results will be reported in a 'risk of bias' table.

We will summarise the overall risk of bias for each study (across outcomes) and for each outcome or class of similar outcomes (across studies) using the following criteria (EPOC 2013a; Higgins 2011a):

- Within each study across domains:
  - Studies with low risk of bias for all key domains or where it seems unlikely for bias to seriously alter the results will be considered to have a low risk of bias;
  - studies where risk of bias in at least one domain was unclear or judged to have some bias that could plausibly raise doubts about the conclusions will be considered to have an unclear risk of bias; and
  - studies with a high risk of bias in at least one domain or judged to have serious bias that decreases the certainty of the conclusions will be considered to have a high risk of bias.
- Across studies:
  - each outcome (or class of outcomes) will be defined as having a 'low risk of bias' if most information is from studies at low risk of bias;
  - as 'high risk of bias' if the proportion of information from studies at high risk of bias is sufficient to affect the interpretation of the results; and
  - an 'unclear risk of bias' if most information is from studies at low or unclear risk of bias.

It is likely for this review that study participants will not be blinded for some interventions (e.g. delivery of an educational intervention). This will be noted in the quality assessment.

Measures of treatment effect

Many of the outcome measures for this review consist of discrete quantitative data (e.g. numbers of patients gaining access to services). For continuous outcomes we will calculate the mean difference (MD) and corresponding 95% confidence interval (95% CI). If the numbers of participants and events are available for dichotomous outcomes (e.g. referral made yes/no), we will calculate the risk ratio (RR) or odds ratio (OR) and corresponding 95% CI. If effect estimates (RR or OR) are reported instead of proportions these will be extracted accompanied by measures of uncertainty (e.g. 95% CI or P value if available).

Unit of analysis issues

Consideration will be given to whether any unit of analysis errors are made in the reported analysis for a study. For example, for cluster-randomised trials we will consider whether the reported results are on the same level as the level of allocation or whether an analysis is adjusted for clustering effect. If a unit-of-analysis issue is identified, the treatment of the study will depend on the type of design and on the available information. Depending on the study, one of the following methods of avoiding unit of analysis issues will be accepted: re-analyse the data if information is sufficient; if there is insufficient information to re-analyse the results the study authors will be contacted to obtain necessary data; if data are not available results of the analysis will be reported in the review in the form of estimates without reporting a measure of uncertainty.

Dealing with missing data

Authors will be contacted if data are missing from the published papers. If missing data are still present then each case will be discussed to determine the most appropriate analysis strategy. We will either report the results based on observed data or use imputation for appropriate types of continuous data (e.g. using the standard error to calculate missing standard deviations).

Assessment of heterogeneity

It is likely that the included studies may show both statistical and contextual heterogeneity. Contextual heterogeneity may include: a range of measured outcomes, differing health systems or health economies, and a wide range of diagnoses and patient populations. We will assess contextual differences by examining these factors. It is possible that due to lack of suitable studies there may not be enough data to draw firm conclusions about the overall effectiveness of an intervention. In addition, there may not be enough data to perform sub-group analysis. In this case we will specify explanatory factors and, if possible, use these factors to guide discussion of the applicability of the findings. Possible explanatory findings might include:

- Differences between insurance based or free point of use services;
- Interventions delivered by non-genetic specialists or genetic specialist health professionals;
- Differences between patient target groups (e.g. pre-natal, adult, cancer or paediatric);
- Intervention setting (e.g. primary care or specialist service); and
- Socioeconomic status of participants.

Information relating to context will be collected and reviewed against the results obtained. It is likely that these factors will help with the interpretation of the results and form part of a narrative description in the final review. The context surrounding the study is important in assessing the effectiveness of the intervention and the overall provision of genetic counselling services. Context will also be important when developing the summary of findings tables (See Data synthesis).

Assessment of reporting biases

If there are a sufficient number of pooled studies (i.e. > 10), we will assess reporting bias by visual inspection of funnel plots.

Data synthesis

Due to heterogeneity in service provision and study design, we expect to find variation across studies in follow-up periods and outcomes. We will report study outcomes irrespective of the range, timing and follow-up periods for the study or how the outcomes were measured. This will minimise selective outcome reporting.
Data synthesis will use a range of effects and plain language summaries following the GRADE guidelines (EPOC 2015c). The results will be presented in a summary of findings table (EPOC 2015c; Higgins 2011b) and make qualitative assessment of the effects of the studies - based on quality, size and direction of effect observed and statistical significance. We will pool data for meta-analysis when studies are reasonably similar in terms of populations, interventions,
characteristics, and outcomes. For dichotomous outcomes, we will calculate the pooled RR or OR and corresponding 95% CI. For continuous outcomes we will calculate the pooled MD and corresponding 95% CI. We will calculate the standardized mean difference (SMD) and corresponding 95% CI when studies utilise different scales to measure the same underlying construct. A fixed-effect model will be used to pool data unless significant heterogeneity is identified.
We will report the following data where available: pre-intervention and post-intervention study outcome in natural units, statistical significance across groups, and variability of outcome.
Resource use will be considered for inclusion in a Summary of Findings (EPOC 2015c), table and a brief economic summary will form part of the review (EPOC 2013c).

Subgroup analysis and investigation of heterogeneity

We will assess statistical heterogeneity by both a visual inspection of forest plots and by calculating the Chi^2 and I^2 statistics. We would consider an I^2 value of greater than 60% as evidence of substantial heterogeneity of a magnitude where statistical pooling is not appropriate. We will use a random-effects model to pool data in situations where there is moderate heterogeneity (e.g. I^2 is less than 60% but more than 25%).
Data synthesis will be structured as recommended by EPOC guidelines (EPOC 2013b). If there are sufficient studies of similar interventions it might be possible to perform a sub-group analysis. Potential subgroup analyses could include:

- Healthcare setting (e.g. primary care or specialised hospital services);
- Who delivers the intervention (e.g. educator or health professional);
- Disadvantaged or advantaged population (e.g. lower socioeconomic groups);
- Ethnicity (e.g. general populations versus minority groups); and
- Income status of the country (e.g. low and middle income countries versus high-income countries).

The above factors could be used to guide discussion around the applicability of the findings. Data extraction will include these key explanatory factors (see Data collection and analysis).

Sensitivity analysis

We will explore the robustness of the results by conducting sensitivity analysis based on:

1. Excluding studies assessed as being at high risk of bias;
2. Excluding studies with missing or imputed data; and
3. Calculating a random-effects model.

Where appropriate, sensitivity analysis will be performed to assess the robustness of conclusions and results will be presented in a summary table. If review results are sensitive to particular assumptions (e.g. study quality) this will be investigated and reported in the review. We will also use sensitivity analysis as appropriate to explore potential explanations for heterogeneity (e.g. by excluding obvious outlier studies).

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* *Indicates the major publication for the study*

APPENDICES

Appendix 1. MEDLINE search strategy

Ovid MEDLINE(R) In-Process & Other Non-Indexed Citations and Ovid MEDLINE(R) <1946 to Present>
Search Strategy:

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1 exp Genetic Counseling/ (11904)
2 exp Genetic testing/ (27106)
3 exp Genetic predisposition to disease/ (94167)
4 ((risk$ or gene$ or geno$) adj3 (service$ or screen$ or inform$ or counsel$ or test$)).tw. (152697)
5 or/1-4 (255462)
6 delivery of health care, integrated/ (8702)
7 exp health services/ (1610829)
8 exp "Referral and Consultation"/ (57906)
9 exp patient-centered care/ (11588)
10 exp health services accessibility/ (85233)
11 (service$ adj3 organi$).tw. (5702)
12 (multi-disciplin$ or multidisciplin$).tw. (51388)
13 "Delivery of Health Care"/ (66464)
14 or/6-13 (1775488)
15 5 and 14 (56030)

16 (randomized controlled trial or controlled clinical trial).pt. or randomized.ab. or placebo.ab. or clinical trials as topic.sh. or randomly.ab. or trial.ti. (935973)
17 exp animals/ not humans.sh. (3986356)
18 16 not 17 [Cochrane RCT Filter 6.4.d Sens/Precision Maximizing] (863154)
19 15 and 18 (2845)

Appendix 2. Data Extraction Form

Interventions to improve patient access to and utilisation of genetic and genomic counselling services.
Data extraction and management - Draft data extraction sheet

- Country where research was carried out
- Study inclusion and exclusion criteria
- Study design (e.g. RCT, cluster-RCT)
- Recruitment method (e.g. self referral, advertisement)
- Target population
  - Condition (e.g. breast cancer, cystic fibrosis, all conditions)
  - Group Category (e.g. prenatal, paediatric, cancer or adult (non-cancer)
  - Risk Category (e.g. low, medium, high, mixed)
  - Ethnicity (e.g. Ashkenazi Jewish)
  - Setting (e.g. population, primary care, secondary care, genetic patients)
  - Socioeconomic (e.g. low or middle income or high economic population)
- Study intervention healthcare setting (e.g. primary care, specialised services)
- Description of usual care
- Intervention details
  - Mode of delivery
  - Setting and process
  - Theoretical basis (e.g. using 93-item BCT taxonomy (Michie 2013) or educational theory)
  - Content
  - Intervention target (e.g. healthcare staff or clients or both),
  - Behavioural target (e.g. communication with family members; utilisation of healthcare services, genetic service, genetic testing, or screening services);
  - Health condition targeted (e.g. diabetes, breast cancer, mixed)
- Healthcare worker details
  - Professional group
  - Qualification level
  - Job title
  - Number
  - Age
  - Socioeconomic status
  - Ethnicity
  - Gender
  - Time since qualification
- Client/Patient details: number, age, socioeconomic status, ethnicity, gender, time since diagnosis (where applicable)
- Other outcomes measured by the study
- Quality criteria (in line with EPOC recommendations (EPOC 2015b) and EPOC Guidance on risk of bias (EPOC 2013a))

CONTRIBUTIONS OF AUTHORS

CB led the writing of the protocol. LT JC SG HS RB CP KC and KP provided comment and feedback. For the full review all authors, supported by the University of Central Lancashire School of Health Research Team will screen the title and abstracts of records for eligibility. CB CP HS CK RB and SG will screen full text for eligibility and will abstract data. ST will perform data analysis assisted by Chris Sutton. The clinical review team, Dr Astrid Weber and Dr Paul Brennan (Consultant Clinical Geneticists), will assist with the interpretation of results and will provide comment on the final review document.

DECLARATIONS OF INTEREST

None known

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