

Part 3

Guidance for rating each question

Question 1 - Are the aims clear?

What the question is about and why it is important

A good quality publication will have clear aims. A publication should include an overview indicating what it is about, what it intends to cover and who it is meant for. Clear aims in a publication are important because they indicate what aspects will be addressed and help you to judge if the publication is likely to contain the information you need. It is particularly important to know what may *not* be included, as you may need additional information before you can make an informed decision about genetic testing.

Rating the question

Examine the opening paragraphs for a description of the content, scope and target audience of the publication. Although the publication's title may be descriptive, clear aims should still be outlined if the publication is to get a good rating. The aims should relate specifically to the information provided by the publication, which may not necessarily be covered by more general goals or purpose such as an organisation's *Mission Statement*. In the case of a Website or online information, you may need to search some of the background sections (e.g. *About This Site*) for aims - if they are hard to find, you should be cautious about giving a high rating to this question.

Guidelines for rating the question:

5: yes - the publication has clear aims

2 – 4: partially - the publication has aims but they are unclear or incomplete

1: no - the publication does not include any indication of its aims

Examples:

We have devised simple examples here to demonstrate the quality criterion underlying Question 1. However, the information rated by this question should also have clear sources and referencing where appropriate as outlined in Question 16. For further guidance and examples, please refer to the section on Question 16 in the Handbook

5 rating:

Example (i) Inherited cancer

A national cancer charity booklet includes the following paragraph on the opening page:

Some people are more likely to develop this form of cancer because they have inherited the faulty gene that causes it. If you have relatives with this form of cancer, you may be at risk. We have written this leaflet to provide some information which might help you

decide whether or not to be tested. Its main aim is to help you estimate your chances of having the mutated gene. We also provide guidance on some of the personal and practical issues surrounding testing, although more detailed information about what you can do about your increased risk if you test positive is provided in our supplementary leaflet “Living with a Positive Result”. Many things remain unknown, but we hope the leaflet clarifies some of the issues and provides you with questions about testing which you may like to discuss with your healthcare providers.

Example (ii) Huntington’s Disease

An international health foundation factsheet includes the following on its opening page:

This is a hereditary disease caused by a faulty gene. This factsheet has been developed for patients and their families to explain:

- *The genetic mutation*
- *How the disease is passed on*
- *The chances of developing the disease*
- *Genetic testing, including antenatal testing*

In many cases, a diagnosis is confirmed by additional tests such as a neurological examination and a brain scan. More information about these tests is available from The Foundation’s publications department. The Foundation also provides advice about this condition and treatment, and support services for families.

Example (iii) Antenatal screening and testing

A government health department leaflet distributed to all pregnant women through their family doctor commences:

During your early pregnancy, you may have a screening test for detecting Down’s Syndrome in your baby. This is a routine test offered to all women during pregnancy, but there are many things you and your partner may want to consider beforehand. This leaflet contains the most important things you need to know about screening and testing for Down’s Syndrome. It provides a brief description of the condition and what causes it, and explains the screening procedure and any followup testing that may be recommended. It also outlines your options if you receive a positive result at any stage during screening and testing, and includes sources of further information and support.

Partially rating:

Example (iv) Inherited muscle disorder

A website produced by a hospital genetics department is entitled *Inheriting Muscular Dystrophy*. The home page commences with:

This website has been produced by the Clinical Genetics department. Muscular Dystrophy is an inherited muscle disorder. Genetic testing is available in this department. The decision to be tested is a highly personal issue, and our counsellors are here to help you.

Additional rating notes: There is too little introductory text - although inferred, it is not clear whether providing information about testing is the main aim of the website or who the readers might be.

Example (v) Fragile X Syndrome

A consumer health company Web page entitled *Screening Tests for Fragile X Syndrome* commences with the statement:

There have been significant advances in detection of Fragile X in recent years and there is now an accurate test available.

Additional rating notes: The content is implied from the title and opening, but there is no mention of any particular focus or omissions, including what type of test process *Screening* refers to.

1 Rating:

Example (vi) Carrier screening – Sickle cell disease

An international charity website provides information about Sickle cell disease. The Home Page provides the following menu:

About Us

Education

Health Professionals

Fundraising

Further help

A *Mission Statement* under *About Us* describes the aims of the charity as *dedicated to supporting research on treatments for Sickle Cell Disease and supporting all those whose lives are affected by this condition.*

Screening and testing information is provided in a brief paragraph in a subsection entitled *For Parents* under *Education*. More detailed information is provided under *Diagnostics* in the *Health Professionals* section.

Additional rating notes: The content is implied, but none of the pages or subsections commence with any indication of what is included in the text or who it is written for. Users interested in carrier screening and testing would have to work hard to find the information they need.

Question 2 - Does it achieve its aims?

What the question is about and why it is important

A good quality publication will cover the information it has aimed to provide.

Question 2 is designed to help you assess whether the information that was outlined at the beginning of the publication has actually been provided. This question is important because a publication that does not achieve its aims is incomplete and you may need more information before you can make an informed decision about genetic testing.

Rating the question

After reading the publication, refer back to the aims and consider whether all the information you were led to expect has been provided. If aims were outlined but were not clear or were incomplete (rated 'partially' on Question 1), you need to use your judgement to decide what expectations the aims had raised and then rate to what extent those expectations have been fulfilled.

Guidelines for rating the question:

5: yes - all the information you were expecting from a description of the aims has been provided

2 – 4: partially –some of the information you were expecting from the aims has been provided

1: no - *none* of the information you were expecting from the aims has been provided

N/A - the publication did not have clear aims (rated '1' on Question 1). Question 2 is irrelevant and should be rated "Not applicable".

Examples

We have devised simple examples here to demonstrate the quality criterion underlying Question 2. However, the information rated by this question should also have clear sources and referencing where appropriate as outlined in Question 16. For further guidance and examples, please refer to the section on Question 16 in the Handbook.

5 rating:

Example (i) Inherited cancer - rated '5' on Question 1 (Example (i))

In accordance with its aims, the publication provides a detailed explanation as to who may be at risk and why a genetic test may or may not be recommended. Details of the test

includes a description of what it involves and how the results are interpreted. It also outlines in detail many personal and practical issues that need consideration before being tested (as covered by later DISCERN Genetics questions). Although the implications of a positive test result are mentioned in these sections, readers are referred to a separate leaflet providing more detailed information about living with a positive result and the condition itself.

Example (ii) Huntington's Disease - rated '5' on Question 1. (Example (ii))

In accordance with its aims, the publication provides detailed information about the genetic basis of the condition, its transmission and presentation, and genetic tests for the condition.

Additional rating notes for (i) and (ii): In both examples, the aims were very specific and additional information such as other testing procedures (Huntington's example) or care options and quality of life issues were not intended to be included. It is good practice to list publications and organisations providing this additional information (see Question 15), although these examples would still get a high rating on this question without this information.

Partially rating:

Example (iii) Fragile X Syndrome - rated 'Partially' on Question 1 (Example (v))

A consumer healthcare company Web page entitled *Screening Tests for Fragile X syndrome* and commences with the statement:

There have been significant advances in detection of Fragile X in recent years and there is now an accurate test available.

The website is a scientific document outlining the development of a new blood testing kit and comparing its reliability and commercial potential with an existing kit.

Additional rating notes: The publication does not provide any information about the condition or the actual testing procedure, including decisions about testing, which might be expected from the title and opening statement and which would provide a context for testing.

1 rating:

Example (iv) Haemochromatosis

A University hospital web page entitled *Haemochromatosis* includes an opening statement that it *aims to provide information about this rare, inherited blood disorder*. The publication describes the genetic fault underlying the condition and the technicalities of a genetic test.

Additional rating notes: There is no information about the background, effects or treatment of the condition which would be expected from the stated aim.

Question 3 – Is there an explanation on the background and effects of the condition?

What the question is about and why it is important

A good quality publication will provide a clear description of the condition. This description should cover possible causes (including details of the genetic basis of the condition), inheritance patterns, symptoms, course, and prognosis. This information is important for deciding about testing, as it helps you understand the condition and what being identified “at risk for” or having the condition might mean for you.

Rating the question

Rate the question according to the information it provides on the background and effects of the condition. This information does not need to be very detailed, but should provide you with a clear overview of the nature of the condition.

Guidelines for rating the question:

5: yes - the publication provides a clear explanation of the background and effects of the condition

2 – 4: partially - background and effects are mentioned but the information is unclear or incomplete

1: no - there is *no* information provided on the background and effects of the condition

Examples

We have devised simple examples here to demonstrate the quality criterion underlying Question 3. However, the information rated by this question should also have clear sources and referencing where appropriate as outlined in Question 16. For further guidance and examples, please refer to the section on Question 16 in the Handbook

5 rating:

Example (i) Inherited cancer

A national cancer charity booklet provides information about genetic testing for an inherited form of cancer. The opening sections include a description of the symptoms and progress of the condition, an explanation and diagram of the genetic fault underlying the condition, details of who it affects (prevalence, age range and sex ratios), how it is

inherited and how genetic variations of the disease produce different symptom profiles, and areas of scientific uncertainty regarding risks and outcomes for the condition.

Partially rating:

Example (ii) Tay Sachs disease

A hospital website provides information about screening for Tay Sachs disease. The condition is described as follows:

Tay Sachs Disease is a fatal, inherited neurodegenerative disorder. It usually begins in infancy. There is no treatment or cure, and affected infants usually die before age 5 (cases of later onset usually die by the age of 15). Tay Sachs occurs most frequently in descendants of Jewish families from Central and Eastern Europe (Ashkenazi Jewish ancestry). It describes the prevalence of the condition in Jewish and non-Jewish groups. It is a recessive disorder, which means that both parents have to be carriers to pass on the faulty gene.

Additional rating notes: The description of the disease provides minimal information on symptoms and features of the condition. Clear details of transmission patterns and risks are not provided. Other omissions include an indication of whether any affected children survive into adulthood, or whether being a carrier has any physical or emotional effects.

Example (iii) Turner syndrome

A charity website provides details of Turner Syndrome, including symptoms, features and testing. The section on *Causes* consists of the following:

Turner syndrome is caused by a missing or abnormal chromosome normally found in women.

Additional rating notes: This description of causes is incomplete as it is not clear whether the genetic mutation is inherited or sporadic and there is no link between the chromosome abnormality and symptoms and features of the condition.

1 rating:

Example (iv) General

A file downloaded from a University website describes genetic testing for an inherited condition. The publication gives a detailed description of the genetic markers for the disease and methods of testing. However, there is no discussion of the condition, its symptoms, prevalence, inheritance patterns or related risk factors.

Question 4 – Are treatment and management choices for the condition described?

What the question is about and why it is important

A good quality publication will include a description of treatment and management choices for the condition. This information is important when deciding about testing, as it helps you understand what options are available if you are diagnosed with the condition or found to be “at risk”. Treatment and management choices can include preventing or eradicating the condition or its symptoms, slowing progress of the condition, and maintaining quality of life with the condition.

Rating the question

Rate the question according to whether there is a clear description of treatment and management choices for the condition. This information should be explicit but can be brief, as many publications appraised using DISCERN Genetics will be primarily concerned with testing and diagnosis and a detailed analysis of the quality of the information about treatment choices is beyond the scope of the DISCERN Genetics tool. To fully appraise the quality of the information on treatment choices, please refer to DISCERN Treatment (see Part 6 for details).

Note that this question is only concerned with the treatment described in the publication. The question cannot be used to assess whether *all* possible treatment choices for the condition have been described as this would involve checking against other sources.

Guidelines for rating the question:

5: yes - the publication provides a clear description of treatment and management choices for the condition

2 – 4: partially - treatment and management choices are mentioned, but the information is unclear or incomplete

1: no - there is no information on treatment and management choices for the condition.

Examples

We have devised simple examples here to demonstrate the quality criterion underlying Question 4. However, the information rated by this question should also have clear sources and referencing where appropriate as outlined in Question 16. For further guidance and examples, please refer to the section on Question 16 in the Handbook

5 Rating:

Example (i) Inherited cancer

A national charity booklet on genetic tests for an inherited form of cancer outlines a “journey” from testing to management which covers the role of various health professionals, packages of care, living with the condition and reproduction issues. The section on care outlines monitoring and care following a positive test result, plus options for treatment if symptoms develop. The role of lifestyle factors in managing disease progress and symptoms is outlined, and information on treatment choices consists of a table summarising the risks and benefits of various drug treatments and surgery. Throughout the publication, readers are guided to resources providing more detailed information on specific treatments and outcomes which are fully listed under *References* and *Further Information*.

Partially rating:

Example (ii) Antenatal screening and testing – Cystic Fibrosis

A health authority booklet on screening and testing in pregnancy includes a section on genetic tests for cystic fibrosis. It includes brief descriptions of diagnosis, symptoms and progress of the disease, plus risk statistics and testing issues. Treatment and management information consists of: *At present there is no cure for CF, but symptoms can be reduced through physiotherapy and drug treatments. Research into new treatments is ongoing.*

Additional rating notes: There is insufficient detail on treatments, and the process of management and referral following a positive test result and quality of life issues are not mentioned.

Example (iii) Neonatal screening and testing

A maternity hospital leaflet provides information for parents on neonatal screening for a range of conditions (through a blood spot test). Screening is recommended *because of the benefits of early detection and treatment of these serious conditions*. For each condition, it provides a brief description of its effects, states whether or not it is inherited and describes how the symptoms of each condition are managed, highlighting that treatment is more effective if commenced in infancy. Complications and choices associated with treatments are not mentioned. The possibility that a baby may be found to be a *carrier* for a condition is also mentioned without any explanation or discussion of any management options. Screening results are described as either indicating *your baby is normal*, or *thought to have a condition which may require further action*, but the process of referral is not described, and quality of life issues are not mentioned anywhere in the leaflet.

Additional rating notes: There is insufficient detail on treatments, particularly risks and choices, and the process of management and referral following a positive test result and quality of life issues for affected children and carriers are not mentioned.

1 rating:

Example (iv) Antenatal screening and testing – Down's Syndrome

A popular book on pregnancy and early childhood includes information about antenatal screening and testing for Down's Syndrome. It provides a detailed description of the condition, the screening test and interpretation of results, and the diagnostic tests most commonly used - amniocentesis and chorionic villus sampling. It does not include any information or additional references on management options during pregnancy if Down's Syndrome is detected (such as the process and impact of having a termination or of continuing with a pregnancy), or after giving birth (such as managing the physical and mental health of a child with Down's or any other aspects of living with Down's Syndrome).

Question 5 – Is risk explained in simple terms?

What the question is about and why it is important

A good quality publication will describe the risk of developing, carrying or passing on a genetic condition in a way that is easy to understand. It is important that the publication enables you to understand clearly why *you* might or might not be at risk, and what your *risk* (*chances* or *likelihood*) of different outcomes might be. The explanation should help you to understand why testing might or might not be recommended for you and what it may reveal about the type and level of risk you may confront.

Rating the Question

The question focuses on the risks of developing, carrying or passing on a genetic condition, and not risks associated with the test procedure (Question 7), test accuracy (Question 8), or treatment choices (Question 4). This question is only concerned with the risk or risks described in the publication and cannot be used to tell you whether *all* of the risks have been described, as this would involve checking against other sources.

Risk information is based on studies of disease patterns in large groups of people with the genetic condition and in the general population. An individual's risk is an estimate of the *chance* or *probability* or *likelihood* of a particular outcome, and is often described using statistics such as 1 in 100, 1%, and so on. An important part of explaining these risks is also acknowledging uncertainty where aspects of the condition and its development are poorly understood.

You may find it helpful to consider 2 main issues when rating this question – (i) the risks involved and (ii) the way the risk is *explained*. Terms used below are described fully in Part 1 and Glossary.

1. Risks

(a) Do I have the faulty gene? Will I pass it on?

Information about *who* the mutation and condition affects is important for understanding your risk. You may be at greater risk for a condition with a genetic cause compared with other people in the general population because the condition runs in your family. This may be linked to your ethnicity or geographical roots - there may be a higher rate of carriers or people with the condition in families from your background than in families from other ethnic groups.

Risk may not be related to family history but to other factors such as your age (e.g. the chance of having a child with Down's Syndrome) or unpredictable individual circumstances (e.g. a new, sporadic mutation). Here, a description of how the mutation and condition may arise and the reasons why you may be at risk are important. The

random nature and lack of scientific knowledge surrounding some of these events should also be described honestly.

Family history

If a condition with a genetic cause runs in your family, there is a chance you have inherited the mutation and will pass it on to your children. You may be affected yourself or you may be an unaffected carrier. To understand risks here, you need information about patterns of inheritance and the likelihood or chance of individual family members having or not having the mutation (e.g. “if both partners carry the faulty gene, then your child has a 1 in 4 chance of inheriting this gene - so a 3 in 4 chance of not inheriting it”). This can also be presented in diagram form as a family tree or *pedigree* using symbols for different types of risks and relationships (e.g. affected and unaffected siblings; carriers; types of twins etc). We have included an example in Part 1 and in Example (iii) below.

Risk information presented this way can help you understand your own risks and those of your relatives, and can clarify the possible reasons for testing and likely outcomes of testing.

(b) If I have the mutation, will I become ill?

The chances that an asymptomatic individual with a mutation will go on to develop disease varies with each condition. For some diseases, *everyone* who has the mutation will develop the disease (e.g. Huntington’s disease). A positive predictive test result in such cases means you will certainly develop symptoms at some stage. For many other conditions, a result indicating you have a mutation does not necessarily mean you will develop symptoms or the condition, and even a confirmed diagnosis can be associated with different outcomes. If you are a carrier for a recessive disorder, you may not be affected by the condition, but it is possible that one or more of your offspring will be. The publication should link information about symptoms and disease progression with the likelihood of these outcomes in individuals with the mutation.

Information about the risk of developing symptoms or a condition is often described as a “life-time” risk. For example, if your lifetime risk is 70%, it means that 7 out of 10 people with this genetic mutation develop the condition at some point in their lives. See the references resources in Part 6 for more information about lifetime risk estimates.

Antenatal and neonatal screening and testing

Many screening tests in pregnancy and at birth provide useful information about risks that may need further investigation. Genetic screening tests commonly offered in pregnancy (e.g. cystic fibrosis) and at birth (e.g. sickle cell disease) may indicate that a baby is at *increased risk*, but a further *diagnostic* test would be needed to confirm or reject this result. A publication should provide an explanation of risk arising from each of these stages.

2. Explanation of risk

Understanding risk can be difficult, as it often involves imagining and comparing possible outcomes in the future with no certainty about what will happen to *you*. Our reactions to different types of risk estimates and ways of explaining them can also be unpredictable.

There has been a lot of research in recent years into ways of explaining risk. Ideally, the risk estimates and language most meaningful to *you* should be agreed within the consultation. However, information can also be helpful to you and a wide range of other users if it follows some good practice principles. The hints accompanying Question 5 in the DISCERN Genetics tool outline some suggestions. Further details are provided in the Appendix at the end of the handbook, and by the resources listed in Part 6.

Guidelines for rating the question

5: yes - the publication describes the risks of developing, carrying or passing on the condition and presents the information in a meaningful and useful way

2 – 4: partially - the publication describes these risks, but the information is unclear or incomplete

1: - no the publication does not provide *any* meaningful information about the risks of developing, carrying or passing on the condition

Examples

The information needed to rate this question can be very detailed and will usually be provided with information about the condition (Question 3) and the nature of the test (Question 6). We have devised simple examples here to demonstrate the quality criterion underlying Question 5. However, the information rated by this question should also have clear sources and referencing where appropriate as outlined in Question 16. The sources of the real data used in the examples below are provided in Part 6. For further guidance and general examples of referencing, please refer to Question 16 in the Handbook

5 Rating:

Example (i) Inherited cancer (fictitious risk estimates)

A family doctor's website provides the following information in a section about a new genetic test for a cancer that has received a lot of media coverage:

This cancer is not common, affecting approximately one in every 10,000 people – so it would probably only affect 1 person from our small town. Most cases (9 out of 10 or 90%) develop as a result of chance and the reasons are not yet clearly understood. Fewer than 1 of every 10 cases of this form of cancer (less than 10% of cases) are thought to be inherited. You would therefore need to discuss your family history and health with your

doctor to assess your risk before testing could be recommended. If testing shows you have inherited the mutation for this cancer, you will not necessarily develop the disease - 6 in 10 (60%) people with the mutation develop symptoms (so 4 in 10 don't), which usually happens by the age 50.

Example (ii) Huntington's Disease (based on real risk estimates)

An international charity leaflet provides the following details about inheritance of Huntington's disease:

Huntington's disease is an autosomal dominant genetic condition, which means you only need to inherit the faulty gene from one of your parents to get the disease. So, if you have a parent with Huntington's disease, you have a 50:50 chance of inheriting and an equal chance of not inheriting the faulty gene. If you have inherited the gene, each of your offspring will also have the same risk. Males and females are affected equally. Everyone who inherits the faulty gene will, at some point, develop the disease. Unfortunately, because it is a late onset disorder, you may not know you are at risk until you have had children and passed on the condition. Support and counselling for you and your family is therefore an important part of the test procedure.

Example (iii) Tay Sachs Disease (based on real risk estimates)

The patient information section of a hospital genetics department website provides the following information about Tay Sachs disease:

Tay Sachs Disease is an inherited neurodegenerative disorder. It usually begins in infancy. There is no treatment or cure, and affected infants usually die before age 5 (cases of later onset usually die by the age of 15).

Tay Sachs occurs most frequently in people with Ashkenazi Jewish ancestry (descended from the Jewish populations of Central and Eastern Europe). The disease is approximately 100 times more common in infants of Ashkenazi Jewish ancestry (about 1 per 3,000 births) than in other infants (about 1 in 300,000 births in other ethnic groups). Each sex is equally affected.

Tay Sachs is a recessive disorder, which means that both parents have to be carriers to pass on the faulty gene. 1 in 30 people with Ashkenazi Jewish ancestry are carriers for Tay Sachs, and if you and your partner share this background, there is a 1 in 900 chance that you are both carriers and risk passing the condition on to your children. Each pregnancy will have 4 possible outcomes: a 25 in 100 chance that the baby will have the disease, a 50 in 100 chance that the child will be a carrier, and a 25 in 100 chance that the child has not inherited the faulty gene. The following diagram shows how the condition runs in families:

INSERT Tay Sachs Pedigree here

Access to a national screening programme is available through this hospital for all young adults and pregnant women from this background.

Example (iv) Antenatal Screening – Down's Syndrome (based on real risk estimates)

A UK obstetrics department leaflet provides the following explanation of risk estimates arising from a screening test for Down's Syndrome:

The risk of having a baby with Down's syndrome increases with the mother's age. It is not clear why this happens, although it is likely that chromosome abnormalities occurring during the formation of the egg prior to conception become more common as women get older.

The screening test for Down's syndrome provides a clearer estimate that your baby might be at risk than your age alone. It involves taking a sample of your blood and conducting biochemical tests. If your baby has Down's syndrome, you will have raised levels of some biochemical markers for Down's syndrome in your blood. This information is combined with information about your age and stage of pregnancy to produce a numerical estimate of risk for Down's Syndrome. A cut-off risk estimate of 1 in 250 is usually used, and results above this figure are described as "screen positive". If your results come back with a figure over 1 in 250, this does not mean that your baby definitely has Down's Syndrome. It means that 1 person with this result will have an affected baby, but about 249 people will not. There is therefore a 1 in 250 chance (about 4 in 1000 or 0.4%) that your baby may be affected and you will be offered a more accurate test (see the following section) to confirm how likely this is.

If your risk is less than 1 in 250, you will be described as "screen negative". This level of risk is interpreted as meaning you are very unlikely to be carrying a baby with Down's Syndrome although it does not mean your risk is zero: for example, a risk of 1 in 500 this still means that 1 in 500 pregnancies with this result will carry a baby with Down's syndrome). Because there are risks associated with further diagnostic testing, we do not recommend it for women with this low level of risk.

Our healthcare team are here to support you and will be happy to discuss the screening tests, results and options with you in more detail.

Details of the diagnostic test procedures are then provided.

Partially rating:

Example (v) Inherited muscle disorder

A family medical health reference book includes the following background in a section on sex-linked disorders and genetic testing:

This is an inherited degenerative disorder, and symptoms usually develop in adult life. However, there is a probability that some children born with this condition will develop very severe physical disability at an early age, but it is not currently possible to predict who this might be or when this will happen.

Additional rating notes: there is no description of inheritance patterns or clarification of the terms "usually" or "probability" or "some children". However, the reference to uncertainty is a positive feature.

Example (vi) Inherited cancer

An article on breast cancer in a women's magazine contains the following:

If there is a history of breast cancer in your family, you may also be at risk. You should discuss this with your doctor and investigate options for genetic testing.

Additional rating notes: the nature of the family history is unclear, and the link between family history and genetics is not explained. The size and nature of the risk is not specified.

1 rating:

Example (vii) Neonatal screening

A maternity hospital leaflet provides information for parents on neonatal screening. It includes a brief description of the conditions being detected and the test procedure, which is a blood spot test. The conditions are described as *rare* but there is no information on prevalence or incidence. Some conditions are also described as *inherited* with no further explanation of how they are passed on, or whether the parents, older children or future pregnancies would also be affected if the test is positive. Screening is described as *aiming to detect babies more likely to have these conditions*, and results are described as either indicating *your baby is normal*, or *thought to have a condition which may require further action*. The possibility that a baby may be found to be a *carrier* for a condition is also mentioned without any explanation of this term. No further information or statistics are provided on each stage of testing.

Additional rating notes: the risk language and descriptions are too general, brief and imprecise to give parents meaningful information for understanding risks and for making an informed choice about genetic testing.

Example (viii) General

A commercial online health encyclopedia has the following entry for a genetic condition and testing:

This is an inherited disorder. People with this condition have numerous symptoms (described) and reduced life span. A number of tests, including DNA testing, are used to detect the condition.

Additional rating notes: There is no mention of any risk or chance associated with any aspect of the disorder or testing.

Question 6 – Is the nature of the test clear?

What the question is about and why it is important.

A good quality publication will provide clear information about the type of test available and its purpose. Such information will highlight what you can and cannot find out from being tested. This information can help you make an informed choice about testing by clarifying what the test is for, why it is being done and what the consequences might be. These details may help you decide whether testing is appropriate for you. It is important to remember that a blood or tissue sample may be obtained for a variety of tests, in some cases this may be for a genetic test and in other cases it may be for another type of test.

Rating the question

A full description of the nature of genetic tests is outlined in Part 1 and the Glossary. Rate the question according to whether it provides clear information about the nature of the test, including the reasons for testing and the type of test available. Note that this question is not about the actual testing *procedure* (Question 7) or the *accuracy* of the test (Question 8).

Guidelines for rating the question:

5: yes - the publication provides clear information about the nature of the test

2 – 4: partially - the nature of the test is described, but the information is unclear or incomplete

1: no - there is no information about the nature of the test

Examples

We have devised simple examples here to demonstrate the quality criterion underlying Question 6. However, the information rated by this question should also have clear sources and referencing where appropriate as outlined in Question 16. For further guidance and examples, please refer to the section on Question 16 in the Handbook

5 Rating:

Example (i) Huntington's Disease

A hospital neurology department patient handout provides the following background to genetic testing for Huntington's Disease (a separate section outlines details of the test procedure, including an explanation of the genetic mutation and test accuracy):

Testing is done in a laboratory on DNA taken from your blood sample. The test is to find out whether or not you have inherited the faulty gene that causes Huntington's disease. This test is being offered to you for one of the following reasons:

i. you have a family history of Huntington's and you may also be affected - even if you are not showing any signs or symptoms at present

or

ii. you are experiencing symptoms which suggest that you may have Huntington's disease and a test is needed to confirm (or reject) this diagnosis.

Example (ii) X-linked recessive condition

A clinical genetics department website provides the following information about testing for an X-linked disorder:

The pattern of inheritance for this condition is called X-linked recessive. This means that the genetic fault responsible for this condition is carried by females but only male offspring develop the condition. This is because females carry 2 copies of the X sex chromosome and one will therefore function normally. Males only have one X chromosome and so if this is faulty, they will show the disease.

We offer 2 types of genetic test to families affected by this condition. Female family members can be tested to estimate the likelihood that they are carriers of the mutation. Tests can also be done during pregnancy to find out whether the foetus is affected. In both types of test, we send a small sample of blood to the laboratory where it will be analysed for the genetic fault which causes this condition. The accuracy of these tests and the interpretation of results are then discussed in detail.

Example (iii) Antenatal testing – Down's Syndrome (following on from Question 5, example (iv))

A UK hospital obstetrics department leaflet entitled *Diagnostic Tests for Down's Syndrome* provides the following information:

You are being offered further tests in your pregnancy because you have received a "screen-positive" result for Down's Syndrome from your screening test. This result means there is at least a 1 in 250 chance (0.4%) that your baby is affected, and a diagnostic test is recommended if you want more precise information about your risk. There are 2 diagnostic tests available at this hospital – amniocentesis and chorionic villus sampling (CVS). These tests involve taking a sample of material that contains genetic information from the fetus (either amniotic fluid or placental tissue) and examining it in the laboratory for evidence of the chromosome abnormality that causes Down's syndrome. Full details of each of these test procedures, accuracy and issues to consider before testing are described later in the leaflet.

Partially rating:

Example (iv) Inherited muscle disorder example

A Web page produced by a disability self-help group includes a section on an inherited muscle disorder. The main focus of the page is a description of the symptoms, treatments

and support available for those affected. There is a brief section on inheritance patterns which concludes *Recent research breakthroughs have ensured that a genetic test is now available for those that need it.*

Additional rating notes: The nature of the test has to be inferred from information about inheritance - there is no clear description of what the test detects or who it is meant for.

Example (vi) Neonatal Screening (Q5 Example vii)

A maternity hospital leaflet for parents provides information on neonatal screening. It includes a brief description of the conditions being detected and the test procedure, which is a blood spot test. The conditions are described as *rare* but there is no information on prevalence or incidence. Some conditions are also described as *inherited* with no further explanation of how they are passed on, or whether the parents, older children or future pregnancies would also be affected if the test is positive. The term *screening* is used but is not defined and is described as *aiming to detect babies more likely to have these conditions*. Screening results are described as either indicating *your baby is normal*, or *thought to have a condition which may require further action*. The possibility that a baby may be found to be a *carrier* for a condition is also mentioned without any explanation of this term. No further information or statistics are provided on each stage of testing.

Additional rating notes: The reasons for testing are unclear due to a lack of discussion about risk (prevalence, inheritance factors) and no clear description of “screening”. There is no distinction between genetic testing and other forms of test performed on the blood sample. There is no clear explanation of what the screening results mean, particularly if not “normal”, and no clear reference to the possibility of further testing if a result is “abnormal” and requires action.

1 rating:

Example (v) Haemochromatosis

A University hospital web page entitled *Haemochromatosis* provides detailed information about the genetic mutation underlying the condition and the technicalities of the genetic test. Although the condition is described as inherited, no other context for testing is given.

Additional rating notes: There is no indication of who the test is for or what the results are meant to indicate.

Question 7 – Is the testing procedure described?

What the question is about and why it is important

A good quality publication will provide details of the testing procedure.

Information about all aspects of the testing procedure is important when deciding whether or not to be tested. It helps you to understand what testing involves and what to expect during testing.

Testing procedures for genetic conditions can vary according to the nature of the test (see Question 6) and the way health services are organised. For example, some are quick, simple and painless procedures, whereas others may involve some physical discomfort, or are associated with additional risks. Some are conducted through local primary care or family medicine services, whereas others may only be available through hospitals or specialist centres. Some are publicly available whereas others involve payment or private care. Most involve counselling before and after testing, but how and where this is done and the time taken to receive results may vary. The decision-making process itself can prove stressful and time-consuming, regardless of whether the actual procedure is simple or complex.

Rating the question

Rate the question according to whether it provides a clear description of the testing procedure.

Guidelines for rating the question:

5: yes - the publication provides clear information about the testing procedure

2 – 4: partially - the testing procedure is described, but the information is unclear or incomplete

1: no - there is no information on the testing procedure

Examples

We have devised simple examples here to demonstrate the quality criterion underlying Question 7. However, the information rated by this question should also have clear sources and referencing where appropriate as outlined in Question 16. For further guidance and examples, please refer to the section on Question 16 in the Handbook

5 Rating:

Example (i) Inherited cancer

A national cancer charity website provides the following information on a genetic test for cancer:

Background to testing

Genetic testing is only recommended for people with a strong family history of this form of cancer. The probability that you have the genetic mutation is first assessed by examining your family history – this is usually done during a consultation with your family doctor. If you are at risk, you will be offered a referral to a special clinic for testing. The test is only available free at certain clinics. Your doctor can provide you with information about the different providers, test availability and test performance.

The test procedure

Going to the clinic does not mean you are obliged to take the test. You will meet with a counsellor who will discuss all aspects of this condition and testing procedure with you. All clinics offering this testing procedure free to the public follow agreed guidelines which means that the counselling is spread over several sessions to give you time to decide, and you can withdraw from the procedure at any time.

If you decide to have the test done, a nurse at the clinic will take two separate blood samples. The samples are then sent straight to the hospital laboratory where DNA is extracted and analysed. Two samples are tested to enable laboratory staff to check the results.

The blood test itself is very quick and is similar to having an injection, so you should not experience any discomfort. Following the test the counsellor at the clinic will support you and keep you informed, and will contact you for an appointment and further counselling when your results become available.

Example (ii) Antenatal testing – Down’s syndrome (based on real statistics – see Question 16 Example (i) and Part 6 for references)

A government health department website has a section on diagnostic tests in pregnancy. It includes the following information about the amniocentesis procedure for detecting Down’s syndrome:

Amniocentesis is a diagnostic test which can confirm whether your baby has Down’s syndrome. It can also detect a number of other genetic and congenital conditions described elsewhere on this website. It is available free in all health regions to all pregnant women who receive a positive screening test result, are aged over 35, or have other risk factors.

The liquid in the womb is called amniotic fluid. It contains cells from the fetus which carry its genetic information (DNA on chromosomes). Amniocentesis is performed by an obstetrician at your local hospital at around the 15th week of pregnancy and involves extracting a sample of amniotic fluid and sending it to the hospital laboratory for testing. In the laboratory test, the chromosomes in the baby’s cells are identified and counted

(this is a cytogenetic technique). If your baby has Down's syndrome, it will have an extra copy of chromosome 21.

On the day of the test, you will need to arrive at the hospital with an empty bladder, and it may be helpful to have arranged for someone to drive you home and spend time with you afterwards. The doctor uses ultrasound to guide a hollow needle through your abdomen to take a small sample of the amniotic fluid surrounding your baby. You should only feel slight discomfort, and some hospitals offer an injection with a local anaesthetic. The procedure only takes a few minutes but before leaving the hospital, your baby's heartbeat will be checked and you will be asked to stay for an hour's observation. You should then go home and rest for the day. A cramping feeling afterwards is normal, but you should contact the hospital if you start to leak blood or fluid from the vagina.

If you have a rhesus negative blood group, the procedure may cause rhesus positive blood cells from your baby to enter your blood stream and your body may develop antibodies which could harm the baby. You will be given an injection into the muscle of your arm or leg after the test to prevent the antibodies developing Full details of this treatment are provided in another section ([link to another section of the site](#)).

Results

Additional cells are usually grown from the amniotic sample to provide enough material for the laboratory analysis. The growth of these cell cultures can take several weeks. A preliminary result may be available within 48 hours but a firm result will not be available for two to three weeks. In a small number of cases, the culture growth fails and the test may need to be repeated.

Risks and Disadvantages

Amniocentesis during the second trimester of pregnancy is associated with a slightly increased risk of miscarriage (approximately 1% or 1 in 100 amniocentesis procedures are associated with miscarriage). This risk is higher earlier in pregnancy which is why amniocentesis is not usually offered before 15 weeks. When deciding about having amniocentesis, you therefore need to balance the advantages of having information about the baby's chromosomes against the small risk of losing the pregnancy.

The timing of the test during the second trimester and the waiting time for test results also means that if your results are positive, you may be making decisions about a termination at a fairly advanced stage of pregnancy. You will have opportunity to discuss the procedure and any concerns you have with your obstetrician and healthcare team before the test.

Additional issues

In some cases, such as multiple pregnancies or HIV positive mothers, amniocentesis may present additional risks or may not be possible. Your healthcare team will discuss your options with you.

Before deciding whether to have an amniocentesis test, you might like to compare this information with the benefits and risks of another diagnostic test - [chorionic villus sampling](#) ([link to CVS section](#)).

Partially rating:

Example (iii) Antenatal screening – Cystic Fibrosis

The procedure simply involves you and your partner providing a mouthwash sample early in your pregnancy. The sample is sent to a local hospital laboratory for analysis. If the results come back positive, you will be called back for further testing.

Additional rating notes: There are no details of when, where, or how the sample is provided or who collects it, what the test results are based on, or waiting time for results.

Example (iv) General example

Testing is available to determine whether or not a person has inherited the genetic mutation that causes this condition. The testing can be done on a blood or tissue sample, and usually takes between 2 to 4 weeks for results.

Additional rating notes: There are no details of where or how the sample is provided or what the test results are based on.

1 rating:

Example (v) Cancer example

A hospital Web page entitled *Genetic testing for breast cancer* describes a genetic mutation linked to breast cancer and the risks associated with having this mutation. It mentions a *simple test* for detecting mutations in these genes, and describes in detail the consequences of a positive result.

Additional rating notes: There is no information on the actual testing procedure.

Question 8 – Does it describe how accurate the test results are?

What the question is about and why it is important

A good quality publication will provide details of the accuracy of genetic tests.

In order to make an informed choice about testing, you need to know about the reliability and limitations of testing, including the possibility of incorrect or unclear test results.

Very few genetic tests are 100% accurate. There are two main causes of test inaccuracy:

- i. incomplete scientific knowledge, which means that currently available tests cannot identify all types of genetic mutation underlying the condition.
- ii. technical problems or errors occurring at different stages of the testing process, including sample collection, laboratory analysis, and interpretation of results.

Describing Test Accuracy

Test accuracy is usually described in terms of the ability of the test to detect true cases of disease and is based on data from the development of the test amongst people with the condition. If a genetic test is described as being 80% accurate, it means that 8 out of 10 people who have the mutation and are tested will be detected and receive a positive result. This is the rate of *true positive* test results, which is often described as *test sensitivity*. The remaining 2 out of 10 or 20% of cases receive an inaccurate *false negative* test result – these people have the mutation but it is not detected by the test.

Test accuracy may also include information about the detection rate amongst samples of people without the genetic mutation. This is particularly important information for developing reliable screening programmes. Information may be provided about the rate of *true negative* test results, which is the number of people who do not have the mutation and who test negative (often described as *test specificity*), or *false positive* results, which is the rate of positive test results in people who do not have the mutation.

Variations in Accuracy

The varied nature of genetic conditions and tests is associated with different levels of test accuracy. In some cases, repeated or additional testing may be necessary to confirm or clarify test results. Some forms of genetic mutation may be hard to find or are as yet unknown, and a conclusive result may not always be possible. Different laboratories using the same test can also produce conflicting results due to variations in skills, standards and equipment. Some test procedures are technically difficult and may fail even in the most skilled hands. Technical failures can be due to difficulties or errors occurring during collection and storage of blood or tissue samples, cell culture growths in the laboratory, and interpretation of DNA sequences and karyotypes.

These are all areas where scientific research is ongoing, and scientists are constantly working to improve the accuracy of genetic tests.

Consequences of Inaccuracy

Inaccurate or unclear test results can have a significant physical and emotional impact. Rather than providing reassurance or clear answers, testing may actually *increase* the amount of uncertainty you have about your health. A *false negative* result may mean that early opportunities to treat a genetic condition or prevent it being passed on have been missed. *False positive* results may produce anxiety and lead to unnecessary treatment and life changes (e.g. not having children). In each case, a prolonged process of repeat testing, referral and consultation may be involved. An *inconclusive* test result may mean you have to live permanently with uncertainty about your status.

The availability of clear and honest information about these issues is important when deciding about testing, as it will help you understand what the test can and cannot tell you and what can go wrong during testing. If you decide to be tested, this information can help you understand and prepare for the results and any further testing that may be recommended.

Rating the question

Rate the question according to whether it provides clear information about the accuracy of *all* the genetic tests described. This may also be described as *reliability* or *certainty*. Note that this question cannot tell you whether the reported accuracy is true and does not recommend an acceptable level of accuracy or format for reporting accuracy (there are no currently agreed standards, but there is growing international collaboration to address these issues).

Guidelines for rating the question

5: yes -the publication provides clear information about accuracy and areas of uncertainty in testing

2 – 4: partially - test accuracy and uncertainty is mentioned, but the information is unclear or incomplete

1: no - there is no information on the accuracy or uncertainty of the test

Examples

We have devised simple examples here to demonstrate the quality criterion underlying Question 8. The information rated by this question should also have clear sources and referencing where appropriate as outlined in Question 16. For further guidance and examples, please refer to the section on Question 16 in the Handbook

5 Rating:

Example (i) An antenatal screening – Fictitious data

Here is some fictitious data from research about a new genetic screening test being offered nationally to all pregnant women. The text demonstrates how these data are used to provide information for the public about the test's accuracy. We have used a baseline figure of 1000 pregnancies to provide a simple numerical demonstration:

	Does the baby have the genetic mutation?		
	Yes	No	Totals
Number of pregnant women receiving a <i>Positive</i> Test Result	<p>(a) 9 <i>9 out of 10 (90%) are true positives</i></p>	<p>(c) 20 <i>20 out of 990 (2%) are false positives</i></p>	<p>29 <i>29 out of 1000 receive positive test results</i></p>
Number of pregnant women receiving a <i>Negative</i> Test Result	<p>(b) 1 <i>1 out of 10 (10%) are false negatives</i></p>	<p>(d) 970 <i>970 out of 990 (98%) are true negatives</i></p>	<p>971 <i>971 out of 1000 receive negative test results</i></p>
<i>Totals</i>	<p>(e) 10 <i>10 in 1000 (1%) babies have the mutation</i></p>	<p>990 <i>990 in 1000 (99%) babies don't have the mutation</i></p>	<p>1000 <i>pregnancies are tested</i></p>

This genetic condition affects approximately 1% or 1 in 1000 of all babies born every year (box e). A screening test for the condition is now offered to all pregnant women and consists of a simple blood test taken at the 12 week antenatal check. The test involves analysing the mother's blood sample in the laboratory for a marker of the genetic mutation that causes this condition.

The test is very accurate, picking up the majority of affected babies– 90% of cases in all health regions (box a). This means that 9 out of 10 babies with the condition are detected at the 12 week antenatal screening. However, the test cannot detect all forms of the genetic mutation, and this means that 10% of pregnancies producing a negative screening test result are actually affected - the screening test produces a 1 in 10 false negative rate (box b).

Due to problems with the interpretation of test results, there is also a chance that some women receiving a positive screening test result actually have babies who are not affected - around 2% of all pregnancies tested will give false positive results (box c). This still means that 98% of unaffected pregnancies will receive a negative test result (box d).

Counselling before the test will help you clarify these issues with your healthcare team and will enable you to work through the possible outcomes and implications of testing. If you receive a positive screening test result, you will be contacted for a repeat test to check the result and if this is also positive, you will be offered a diagnostic test and further support. If you receive a negative test result you will also be able to discuss your results with the healthcare team. Full details of the test procedure, results, and some of the issues you may need to consider before and after testing are outlined later in this leaflet.

Some independent clinics offer testing earlier in pregnancy for a small fee. However, tests conducted before 12 weeks may be associated with higher rates of false-negative results. You should discuss these issues with your doctor or midwife if you are considering earlier testing.

Example (ii) Inherited cancer

A hospital oncology department handout provides the following information on a genetic test:

This test reliably detects the condition in 75 to 85 per cent of cases. However, it is associated with a false positive result (positive result when the condition is absent) in 3 to 10 per cent of cases. Accuracy is higher in public laboratories but waiting times for testing and results can be many months longer than other providers. If you decide to be tested at this clinic, we will take 2 blood samples so that we can check our results, and you may be asked back to provide further samples if the test results are unclear. You will be called back for counselling and treatment if your results are positive. If you receive a negative or inconclusive test result, you will still be monitored regularly and recommended for further testing if you develop symptoms of this disease.

Partially rating:

Example (iii) General

A hospital genetics department handout includes the following information:

This test is very accurate, detecting the majority of cases where there is a faulty gene.

Additional rating notes: Test accuracy is mentioned but not clearly explained.

Example (iv) General

A commercial online health encyclopaedia includes the following information about a genetic test:

It is important that you know that there is no method of detection or prevention that has been proven to be completely effective.

Additional rating notes: Test accuracy issues are implied but not clearly explained.

1 Rating:

Example (vi) Neonatal screening

A maternity hospital leaflet for parents provides information on neonatal screening. It includes a brief description of the conditions being detected and the test procedure, which is a blood spot test. The conditions are described as *rare* but there is no information on prevalence or incidence. Some conditions are also described as *inherited* with no further explanation of how they are passed on, or whether the parents, older children or future pregnancies would also be affected if the test is positive. Screening is described as *aiming to detect babies more likely to have these conditions*, and results are described as either *indicating your baby is normal*, or *thought to have a condition which may require further action*. The possibility that a baby may be found to be a *carrier* for a condition is also mentioned without any explanation of this term. No further information or statistics are provided on each stage of testing.

Additional rating notes: The blood test is used to screen for a number of genetic conditions which would involve several different genetic tests with varying reliability. However, there is no mention of test accuracy anywhere in the publication.

Question 9 – Does the information explain what happens after the test?

What the question is about and why it is important

A good quality publication will describe the procedure following testing. Information about what happens after testing is important when deciding whether to be tested, as it helps you understand how the test results are handled and can prepare you for any further action that may be required.

Rating the question

Rate the question according to whether it provides clear information about what happens after the test. Topics that should be covered include how you obtain your results (whether you are contacted or have to chase them up), how they are given (by phone, letter, in person) and whether this procedure differs for positive and negative results. This information should also mention the possibility of further testing (giving reasons outlined in Question 6) and describe the process for notifying other family members (although full implications of a positive result are dealt with more fully in Question 14). Note that the question does not recommend a standard procedure for providing test results, but counselling before taking the test and when receiving positive test results are considered best practice.

Guidelines for rating the question:

5: yes -the publication provides clear information about what happens after the test

2 – 4: partially - the publication provides information about what happens after the test, but the information is unclear or incomplete

1:no - there is no information about what happens after the test or

Examples

We have devised simple examples here to demonstrate the quality criterion underlying Question 9. However, the information rated by this question should also have clear sources and referencing where appropriate as outlined in Question 16. For further guidance and examples, please refer to the section on Question 16 in the Handbook

5 Rating:

Example (i) Inherited cancer

A national cancer charity website provides the following information in a section on testing:

The tests are complicated, and interpretation of their results can be difficult, so testing is only available at specialist clinics. The results can have enormous implications for you and your family, and expert advice and support from your family doctor or a counsellor at the clinic before and after taking the test is a vital part of the test procedure.

When the test results are available, they will be sent to your doctor or counsellor who will contact you in writing to offer an appointment. The results will be discussed during this appointment – they are never posted out or given over the telephone. If the result is positive, your doctor or counsellor will discuss a care plan and referral for specialist treatment with you. They will also give you a leaflet to pass on to family members which will help you explain your test result and issues they may want to consider regarding testing. If your result is negative, you may still be at risk of developing the condition as the test is not particularly sensitive to all possible mutations (as outlined in an earlier section on test procedure and accuracy) and a plan for monitoring and future testing will be discussed.

Example (ii) Antenatal screening -Down's Syndrome

A government health department leaflet distributed to all pregnant women contains the following information on the antenatal screening procedure for Down's Syndrome (after a section on the test procedure and the meaning of positive and negative results and test accuracy).

Screening test results are normally available within 2 weeks of the test procedure. If your results are negative, your baby is unlikely to be at risk of Down's Syndrome and further testing is not recommended. If a midwife does not contact you within two weeks of taking the test, you can assume that you have a negative result. However, you are welcome to contact the midwife at any time if you have any concerns.

If your screening test results are positive, your midwife will contact you by telephone within two weeks after the test to give you your results and to offer you further information and an appointment for counselling. You should make sure that the telephone number you have provided will reach you at a time and place where you feel comfortable taking this call. The midwife will not leave messages. At the followup appointment, your midwife will discuss the meaning of the screening test result and will explore your options with you, including further diagnostic testing to confirm or reject the screening result (outlined elsewhere in the leaflet).

In all cases, your results will be filed with your patient record and will only be routinely accessible to you, your family doctor and midwife. They will contact you if they feel there is a need to share this information with any other professionals involved in your care, and all your decisions and preferences will be respected by your healthcare team and noted in your record.

Example (iii) General

A flyer provided by a private clinic offering genetic testing includes the following:

We will give you a phone number to call to find out when your results are ready for collection and to book an appointment with the counsellor who will give them to you. Receiving your test result can be an emotional and stressful time, regardless of whether they are positive or negative. Think carefully about timing this appointment. You may need to make sure you have people around to support you, and it is advisable to avoid doing it when you are in the middle of other stressful events. You may also change your mind, and after issuing one reminder, we will not pursue you if you do not keep your appointment to collect your results. However, they will be stored safely and you will be able to collect them at a future date.

Partially rating:

Example (iv) Inherited cancer

A commercial online health encyclopedia provides the following information about a genetic test for people at risk of an inherited cancer. It states: *The test is technically difficult and it takes a long time for results to become available (up to 2 years at present).* There is no mention of support in the meantime or how results are collected. Options following a positive result are outlined: *You can talk with your doctor about the options available to you. Some people with an inherited mutation may consider prophylactic surgery, and various studies are underway into possible drug treatments that may prevent the disease.* It mentions that *close relatives can be informed of their risk and offered counselling, screening and genetic testing* although how this is done and by whom is not described.

Additional rating notes: Procedures following testing are unclear because the information is very general and non-specific.

Example (v) An antenatal testing example – Down's Syndrome

A Web page developed by a local women's health action group provides information for pregnant women. A section on diagnostic tests states that *results for both tests are usually available within a week or two, although we recommend counselling before collecting the result.*

Additional rating notes: There are no details of where or how the results and counselling services are provided.

Example (vi) General

A hospital genetics department handout includes the following information:

The testing can be done on a blood or tissue sample, and usually takes between 2 to 4 weeks for results. The section immediately following deals with the impact of results.

Additional rating notes: There are no details of where, how or by whom the results are delivered.

1 Rating:

Example (vi) Neonatal screening

A maternity hospital leaflet for parents provides information on neonatal screening. It includes a brief description of the conditions being detected and the test procedure, which is a blood spot test. Screening is described as *aiming to detect babies more likely to have these conditions*, and results are described as either indicating *your baby is normal*, or *thought to have a condition which may require further action*. No further information about the test or results is provided.

Additional rating notes: there is no information about when, where, how or by whom the results are delivered. Further action is mentioned but there is no indication as to what this would be.

Question 10 – Does the information state who will have access to the test results?

What the question is about and why it is important

A good quality publication will provide details of how your test results are handled and stored. The results of tests for genetic conditions can be highly sensitive as they may have implications for other family members and for your insurance and employment. Details of who will have access to your results is important - concerns about confidentiality may influence your decision to be tested or may encourage you to specify your preferences about who can access your results.

Rating the question

Rate the information according to whether it states explicitly who will have access to the test results. Topics to consider include how the information is handled (e.g. who gives you the test results – see Question 9 – and who receives copies) and stored (e.g. electronic databases, paper copies). The question cannot be used to tell you whether the procedures outlined are acceptable, but may encourage you to review or request changes to your existing arrangements e.g. disclosure to insurers - see Question 12.

Guidelines for rating the question:

5: yes - the publication provides clear information about who has access to the test results

2 – 4: partially - the publication provides information about who has access to the test results, but the information is unclear or incomplete

1: no - there is no information about who has access to the test results

Examples

We have devised simple examples here to demonstrate the quality criterion underlying Question 10. However, the information rated by this question should also have clear sources and referencing where appropriate as outlined in Question 16. For further guidance and examples, please refer to the section on Question 16 in the Handbook

5 Rating:

Example (i) Inherited cancer

A hospital handout for patients has the following information about handling the results of a genetic test for cancer:

Our genetic counsellor will arrange the test and a date for you to return to discuss your results, which the hospital laboratory will send directly to the counsellor. The sample sent to the laboratory and the written results file have coded labels so that your name is not identifiable. The results file is posted back to the clinic counsellor using a special hospital mail service. If the result is positive, the counsellor will outline your care plan with you. If you decide to have treatment following this counselling session, the counsellor will also send your results to the consultant in the hospital who will be involved in your care. The counsellor will also seek your consent to send a copy of the report to your family doctor to be filed with your notes, which you are entitled to see at any time. All information is treated as strictly confidential and the results cannot be released to anyone other than these named professionals without your consent. The procedure for sending and storing this information conforms to the hospital's confidentiality protocol regarding genetic test results. A copy of the protocol is available from the clinic on request. We do not give your results to insurers or employers without your consent, although you need to be aware that withholding this information may affect your insurance policy or employment contract.

Example (ii) Antenatal testing

A government health department leaflet on antenatal testing distributed to all pregnant women includes the following:

In all cases, your results will be added to your electronic patient record and will only be routinely accessible to you, your family doctor and midwife. They will contact you if they feel there is a need to share this information with any other professionals involved in your care, and all your decisions and preferences will be respected by your healthcare team and noted in your record.

Partially rating:

Example (iii) General

A national patient support group website provides the following information on storage of results:

The result cannot be released to others without the formal consent of the person tested.

Additional rating notes: No further information is provided on who initially handles the result, where the results “reside”, how they are accessed or how your consent is obtained.

1 Rating

Example (vi) Neonatal screening

A maternity hospital leaflet for parents provides information on neonatal screening. It includes a brief description of the conditions being detected and the test procedure, which is a blood spot test. Screening is described as *aiming to detect babies more likely to have these conditions*, and results are described as either indicating *your baby is normal*, or

thought to have a condition which may require further action. No further information about the test or results is provided.

Additional rating notes: There is no mention of how results are delivered, stored, handled or who has access to them.

Question 11 – Does it provide support for shared decision making?

What the question is about and why it is important

A good quality publication will provide information that enables you to participate in decisions about genetic screening and testing. The information should help you prepare for a consultation with a health professional to discuss genetic testing options. It should also highlight issues to discuss with family, friends or carers regarding your decision about genetic testing. The specific consequences of the test results are rated separately under *Psychosocial Issues* (Question 13) and *Impact on Partner and Relatives* (Question 14).

Rating the question

Consider the publication as a whole and decide whether it encourages shared decision-making – perhaps by enabling you to prepare a list of specific issues to discuss regarding the best test choices for *you*. These issues should be made clear throughout the publication, rather than merely being queries arising from its deficiencies and gaps, and should be presented in the context of support and discussion with healthcare providers and family. How high you rate the publication will depend on your judgement of how well it supports you in sharing decisions about test choices. If you do not wish to share in decision-making about testing, rate the publication from the point of view of a carer or relative who wants to know more about your choices.

Guidelines for rating the question:

- 5: yes** - the publication provides good support for shared decision-making
- 2 – 4: partially** -the publication provides some support for shared decision-making
- 1: no** - the publication does not provide any support for shared decision-making

Examples

We have devised simple examples here to demonstrate the quality criterion underlying Question 11. However, the information rated by this question should also have clear sources and referencing where appropriate as outlined in Question 16. For further guidance and examples, please refer to the section on Question 16 in the Handbook

5 Rating:

Example (i) Huntington's Disease

A national charity website for people affected by Huntington's Disease outlines the testing procedure and the implications of the results. It lists the various people who may be affected by the test result – your partner, children, family, friends, employers - and provides a prompt list of issues you may want to consider and share with them when deciding whether or not to be tested. It also lists the role of various health professionals involved in testing and treatment for this condition, and highlights the types of support and information each can provide. In each case, decision-making issues relating to technical aspects of the testing procedure, interpretation and impact of results, and followup care are highlighted, but the publication also encourages readers to share emotional and practical worries (anxiety, relationships, family planning) with their healthcare team. All of this information is reinforced with endorsement to decide what is best for *you* regarding testing.

Example (ii) Inherited cancer

A local cancer patient support group newsletter provides the following information in a section on genetic testing:

The decision to be tested may be one of the most important issues you ever have to deal with. Individual reactions differ, and there are many things you might want to consider. These include the positive medical and psychological benefits to testing as well as some more negative outcomes, and available alternatives to testing such as regular symptom monitoring. These issues are outlined in detail in the following section. It may help to think about testing for a while before making a decision – perhaps keep a notebook of your thoughts for a few days and make a list of the pros and cons of testing for you. You may also find it helpful to discuss these issues with those close to you, bearing in mind that members of the same family may have very different feelings about testing. The counselling process before the test will also help you work through all these issues with your doctor or counsellor before finalising your choice.

Example (iii) Antenatal screening and testing

A government health department leaflet on antenatal screening and testing provides the following information:

Before deciding upon screening and diagnostic tests, you and your partner need to consider the risks and benefits of each stage of testing and what difference being tested (or not being tested) would make to your pregnancy and plans for a family. Take time to make sure you understand what the tests involve and what they can and can't tell you. The final decision rests with you and your partner, but healthcare staff are keen to support you in your decisions and are available to provide expert advice and information.

Partially rating:

Example (iv) Inherited cancer:

A hospital genetics department handout for patients testing positive for a mutation for an inherited cancer provides the following information about referral to specialist care:

Your oncologist will explain the test results in more detail and will also outline your treatment plan.

Additional rating notes: Specific choices and issues for patients to discuss with the doctor are not specified. Information-giving appears valued, but patients are not encouraged to be actively involved in decisions and discussions about their care.

Example (v) General

A popular healthcare book provides the following information in a section on genetic tests:

You may find taking a partner or close friend to appointments can be reassuring and can help you get more out of consultations.

Additional rating notes: There is no detail of the actual decisions or topics you might discuss, and relationships with healthcare professionals are not mentioned.

Example (vi) General

An online patient support group notice-board provides the following in a section on genetic tests:

The final decision must be taken by you and should not be the result of pressure from family or your healthcare provider.

Additional rating notes: No further guidance is provided on what types of issues may arise or how healthcare professionals and family could help with your decision.

1 Rating

Example (vii) Turner Syndrome

A public health genetics database provides information about antenatal and diagnostic testing for Turner Syndrome. It gives a detailed description of the features of the condition including the underlying genetic cause, antenatal screening and diagnostic testing procedures. It states that *there is still debate amongst clinicians about the ethics of testing amongst certain groups*, but the issues are not outlined and there is no explicit discussion of the implications of testing for patients which would support decision-making.

Example (viii) Neonatal screening

A maternity hospital leaflet for parents provides information on neonatal screening. It includes a brief description of the conditions being detected and the test procedure, which is a blood spot test. Some of the conditions are described as *inherited* with no further explanation of how they are passed on, or whether the parents, older children or future pregnancies will also be affected if the test is positive. Screening is described as *aiming to detect babies more likely to have these conditions*, and results are described as either indicating *your baby is normal*, or *thought to have a condition which may require further action*. The possibility that a baby may be found to be a *carrier* for a condition is also mentioned without any explanation of this term. The leaflet states that *screening is*

strongly recommended because of the benefits of early detection and treatment of these serious conditions. The only mention of a role for parents is how to comfort their baby whilst the blood sample is being taken.

Additional rating notes: The leaflet does not present testing as an issue involving options and choices for parents, and does not refer to any of the wider issues or anxieties that parents might have. Parents are not encouraged to seek further information or discuss any specific issues with the healthcare team.

Question 12 – Are issues of discrimination discussed?

What the question is about and why it is important

A good quality publication will alert you to the fact that being tested for a genetic condition can lead to discrimination. It is possible that receiving a positive test result may make you potentially risky and expensive in the eyes of insurers and employers. You need to have clear information about the implications of taking a test and disclosing the results to such bodies in order to make an informed choice about testing.

The following section outlines the main issues. More detailed information is provided by the References listed in Part 6. Note that this question is primarily about financial and employment discrimination - social issues arising from testing are covered by Questions 13 and 14.

In the UK, insurers are allowed to collect details of your current health status and medical history, including family history, when issuing health or life insurance policies. At present, there are legal limits regarding the genetic testing information that insurers can request but this is a changing situation and varies according to the particular condition or disease. In some circumstances, there may be advantages to taking a test and revealing the results. For example, insurers take family history into account when setting premiums, so a *negative* test result for a hereditary condition may *reduce* your assessed risk and premiums (e.g. Huntington's Disease).

Good quality information about genetic testing should include an overview of these issues. A detailed analysis is more likely to be found in specialised publications, and is therefore linked to the need for additional resources and information (Question 15). As legislation and practice can vary from country to country, you also need to know whether the information you are using is relevant to your locale (see Question 16) - particularly on the web, which is a largely international forum. The information should also incorporate the latest developments (Question 17).

Rating the question

Rate the information according to whether issues of discrimination are discussed. This question is not relevant for some genetic conditions and tests (e.g. routine antenatal and neonatal screening) - you will need to use your judgement to decide whether it is appropriate for the publication you are rating.

Guidelines for rating the question:

5: yes the publication provides a clear discussion of issues of discrimination surrounding genetic testing

2 – 4: partially the publication refers to discrimination, but the information is unclear or incomplete

1: no the publication does not provide any discussion of issues discrimination

N/A – this question is not appropriate for this publication and should be rated “Not applicable”.

Examples

We have devised simple examples here to demonstrate the quality criterion underlying Question 12. However, the information rated by this question should also have clear sources and referencing where appropriate as outlined in Question 16. For further guidance and examples, please refer to the section on Question 16 in the Handbook

5 Rating:

Example (i) General

A Website produced by a patient support group attached to a local hospital provides the following information:

If a mutation is found that increases your risk of developing this condition, it may have an adverse effect on your personal finances. The terms or costs of your existing mortgages, pension plans and insurance policies may change or you may have difficulty obtaining new ones. Terms and benefits linked to your employment may also become an issue. It is wise to review your financial and employment arrangements before testing. You may find it helpful to compare different policies or to seek out independent advice. You should also discuss the accessibility of your medical records with your doctor, and check whether your existing financial arrangements will still be valid if you do not disclose test results. It is also worth finding out whether there are any benefits arising from disclosure. There are many helpful organisations providing confidential, independent legal advice and employment rights information - we have listed the main national agencies at the end of this leaflet. The healthcare team can also provide guidance on these matters.

Partially rating:

Example (ii) Inherited muscle disorder

A booklet produced by a national charity provides information about an inherited muscle disorder which includes the following:

If you have the faulty gene, this information may affect your financial security.

Additional rating notes: No further details are provided to explain how financial security is affected.

1 Rating

Example (iii) General

An international health foundation website provides detailed information on testing for genetic conditions, including many personal and decision-making issues. However, it does not mention financial or legal discrimination.

Question 13 – Does the information acknowledge the psychosocial consequences of being tested for the condition?

What the question is about and why it is important.

A good quality publication will alert you to the emotional, psychological and social impact that genetic testing may have on you. This information is important for making an informed choice about testing because it highlights personal issues and emotions you may experience before, during and after testing. These can include anxiety about being tested and when waiting for results, and reactions such as guilt, anger and relief following both positive and negative results. Positive test results may also have an important impact on life decisions such as your career and your plans for a family. The decision not to be tested or inaccurate and inconclusive test results may also mean you have to deal with uncertainty about your health status, perhaps indefinitely. Throughout this process, your relationships may be affected and day to day life may be difficult. Information about these issues will help you understand the consequences of investigating your risk for a genetic condition and will help you prepare if you decide to have a genetic test.

Rating the question

Rate the publication according to whether the psychosocial consequences of testing for the condition are acknowledged. How high you rate it will depend on your judgement of how well these issues are presented. Topics to consider include your experiences and emotions when deciding whether to be tested and when waiting for results, and possible reactions to both positive and negative test results and any uncertainty arising from the testing process. It should also provide some reassurance that such reactions are normal and that seeking support may be beneficial. Note that the consequences for your *partner and family* are considered separately in Question 14.

Guidelines for rating the question:

5: yes - the publication provides a clear acknowledgement of the psychosocial consequences of being tested for the condition.

2 – 4: partially - the publication acknowledges the psychosocial consequences of being tested for the condition, but the information is unclear or incomplete.

1: no - the publication does not provide any acknowledgement of the psychosocial consequences of testing.

Examples

Discussions of psychosocial consequences may occur at various points throughout a publication. The following are simple examples where information has been organised

within a single section of the publication to demonstrate the quality criterion underlying Question 13. Information rated by this question should also have clear sources and referencing where appropriate as outlined in Question 16. For further guidance and examples, please refer to the section on Question 16 in the Handbook

5 Rating:

Example (i) Huntington's Disease

An international charity website provides the following information on the consequences of testing:

Deciding about testing

Knowing you may be at risk can be very worrying. You may feel you would prefer to know for certain whether or not you have the faulty gene, and many people feel a sense of relief once they have decided to be tested. However, the process of being tested and dealing with the results can also be very worrying and disruptive. It can be helpful to have support from a loved one, but involving them may also make you feel guilty or anxious - particularly if they put pressure on you (often unintentionally) to make decisions that you are not comfortable with.

Testing should only take place when the time is right for you. If you can't deal with a decision or testing now, it can be helpful to delay it until after a specific date or event in the future. This can help you keep the test in perspective and will give you time to prepare. Some people decide they do not want to be tested or reveal their results, but they also plan to review their decision on a regular basis. Knowing that you can change your mind in future can make such decisions and their consequences less stressful.

If you decide to be tested, each test outcome is associated with a range of reactions that can vary from person to person. Here are some examples:

Positive test results

People react in different ways when results confirm they have inherited the mutation for or have a definite diagnosis of Huntington's Disease. Negative emotions such as anger, disbelief, guilt and anxiety, are all normal reactions. It is very common to feel regret about not knowing sooner or about passing on the gene. It can also be stressful knowing you are at risk or have a condition which may not be cured, and you may feel anxious not knowing when symptoms will develop or how they will progress. Your relationships may change and you may face difficult decisions about who to tell, whether to get married or have a family, and what career choices are open to you. Joining a support group or having regular meetings with a genetics counsellor can help. Remember that there can be benefits to receiving a positive result, such as a loss of uncertainty, early prevention and treatment of symptoms, job reclassification or financial aide. Most people eventually come to terms with their results and use the information to help them make plans for the future.

Negative result:

Most people feel happy and relieved when they receive a negative test result but some negative emotions are also common. You may feel guilty about affected family members

and concern for their care. You may feel emotionally drained or annoyed that you were so anxious and preoccupied about the test. It may be hard to get back into daily life. If you have symptoms of the condition, a negative result may raise questions about the accuracy of the result and the need for further testing. Most people find these issues improve with time, and your counsellor, healthcare team and family can help you work through any difficulties.

d) Inconclusive results:

Testing may not provide any clear answers and you may have to live with uncertainty. This can be extremely frustrating, and you may feel that the emotional and time-consuming testing process has been pointless. Counselling and support is available which can help you deal with this outcome.

Example (ii) Antenatal testing for Down's Syndrome

An article in a parent and baby magazine provides the following information about the consequences of antenatal diagnostic tests for Down's Syndrome:

Amniocentesis is a reliable and widely used diagnostic test to confirm a positive screening test result for Downs' syndrome. However, it can be an anxiety-provoking process and you will face some difficult decisions which you need to be aware of before you are tested. The test may provide useful information about your baby but is also associated with a small risk of miscarriage. A positive test result means you will have to decide whether to terminate a well-advanced pregnancy or whether to give birth to a baby with Down's Syndrome without knowing how severely disabled they will be.

You are likely to have a range of reactions during this process. The choices may seem impossible and overwhelming, and you may feel anxious about the impact of your decisions on the rest of your family. Relationships can become strained, particularly if those close to you have differing views about how to proceed. If you receive a positive result, you may feel angry and your religious or moral beliefs may be challenged by the decisions you face. You may feel grief that your plans for a family have been let down. You may be fearful about future pregnancies or worried about your ability to raise a child with Down's syndrome. If you receive a negative result, you may feel relieved but sad for women whose babies are affected, or you may not necessarily feel reassured that your baby is all right.

All these reactions are entirely normal and your healthcare team should be able to offer support and advice. You may also benefit from learning more about the procedure or the experiences of other women – we include a list of organisations and publications at the end of this article which may also be helpful.

Partially rating:

Example (iii) General

Genetic testing can raise anxiety and disrupt family dynamics.

Additional rating notes: There are no further details on why or how genetic testing has these effects.

Example (iv) General

Deciding to be tested can be stressful and the results can have a strong impact, even when they are negative.

Additional rating notes: There are no further details on why or how genetic testing has these effects.

1 Rating:

Example (v) Inherited muscle disorder

The website of a commercial genetic testing company provides details of a test for an inherited muscle disease. The site gives a comprehensive description of the genetic test and procedure, including the underlying mutation, test accuracy and waiting time for results. However, there is no mention of the personal consequences of being tested.

Question 14 – Are the consequences of genetic testing and screening for the relatives and partner of the person being tested discussed?

What the question is about and why it is important

A good quality publication will highlight the practical, ethical and emotional impact of genetic testing on relatives and family. This information is important for making an informed choice about testing because it will help you understand what your test could mean for other family members. It can promote mutual support and help family members prepare if you decide to be tested.

Rating the question

Rate the publication according to whether the consequences of genetic testing for the relatives and partner of the person being tested are discussed. This information may be provided at various points throughout the publication, and how high you rate it will depend on your judgement of how well these issues are presented. Topics to consider include the impact of both positive and negative test results such as emotional reactions, relationship stress, the discovery of non-paternity, questions of marriage and family planning, and caring responsibilities. Note that the emotional and social consequences for *you* are considered separately in Question 13, and that specific issues for shared decision-making about *your* test are covered by Question 11.

Guidelines for rating the question:

5: yes - the publication provides a clear discussion of the consequences of being tested for relatives and partner

2 – 4: partially - the publication discusses the consequences of being tested for relatives and partner, but the information is unclear or incomplete.

1: no - the publication does not provide any discussion of the consequences of testing for relatives or partner.

Examples

Discussions of consequences of testing for relatives and partners may occur at various points throughout a publication. The following are simple examples where information has been organised within a single section of the publication to demonstrate the quality criterion underlying Question 14. Information rated by this question should also have clear sources and referencing where appropriate as outlined in Question 16. For further guidance and examples, please refer to the section on Question 16 in the Handbook

5 Rating:

Example (i) Inherited cancer

A hospital genetics department leaflet for patients having a genetic test for an inherited cancer includes the following:

Genetic testing can affect other family members. Your decision about testing and any results you receive will have implications for your partner's life and relationship with you. Test results may also provide important information about the health status of your biological relatives, including your offspring, which in turn could affect the way you interact with each other.

If you receive a positive result for this condition, your relatives may need to be told that they are also at risk, and you will have to decide how this should be done. The clinic will be happy to help you, but if you would prefer to inform them personally, you will need to decide who to tell, and how and when. Reactions and decisions amongst your relatives may be varied. Many will be pleased to have this information because it removes uncertainty and provides them with the opportunity to investigate their own risk and take a test for the disease. Others may be upset and could blame you for depriving them of the right not to know. Your parents and other family may feel guilty that they were unaware they had passed on the condition or did not reveal their own positive result. Your partner may be glad to have this information so that you are both able to act and plan, but may also have concerns about your health and shared circumstances including family planning and financial security. Your children may become worried about their own future, relationships and offspring. You may need to make arrangements for your care if you are diagnosed or start to have symptoms, and those close to you may need to feel involved and prepared. Sensitive information may also be revealed - for example, that someone is adopted or not a child's biological father. Decisions to terminate pregnancy may also arise, which could reveal strong feelings and conflict amongst those around you.

The consequences for your family are not confined to positive test results. If you receive a negative test result, mixed reactions may arise amongst family members who have had a positive test result or diagnosis. An inconclusive result can also be very stressful, as many practical and emotional issues could remain unresolved and it may be difficult for you and your partner and family to see a way forward.

As you can see, any decision to take a genetic test has implications not only for you but for a number of people in your life. In some cases, existing family tensions and rifts may worsen, although these situations can also bring people closer together. Of course, you are not required to take the test or disclose the results, and the final decision should be yours alone. However, you also need to think about the consequences for your family and partner if you decide not to investigate your risk or pass on information about your health status. Counselling before and after the test should help you and those involved in your decisions to work through all these issues.

Partially rating:

Example (ii) Huntington's Disease

A popular family health compendium includes the following in a section on Huntington's Disease:

If you are shown to be at risk due to a strong family history and a positive test result, a wide range of extended family members will be offered testing. It is important that each one is fully counselled before taking the test.

Additional rating notes: There are no further details on how or why the test has an impact on others.

Example (iii) General

An online public health genetics database includes the following in a section on testing:

The genetic test can have an impact on your family, relationships, reproductive decisions and insurance.

Additional rating notes: There are no further details on how or why the test has an impact on others.

1 Rating

Example (iv) Neonatal screening

A maternity hospital leaflet for parents provides information on neonatal screening. It includes a brief description of the conditions being detected and the test procedure, which is a blood spot test. Some of the conditions are described as *inherited* with no further explanation of how they are passed on, or whether the parents, older children or future pregnancies will also be affected if the test is positive. Screening is described as *aiming to detect babies more likely to have these conditions*, and results are described as either indicating *your baby is normal*, or *thought to have a condition which may require further action*. The possibility that a baby may be found to be a *carrier* for a condition is also mentioned without any explanation of this term. The leaflet states that *screening is strongly recommended because of the benefits of early detection and treatment of these serious conditions*. No further detail is provided.

Additional rating notes: There is no discussion of the consequences of neonatal test results for the baby's parents, siblings and other family members.

Question 15 – Does it provide details of additional sources of support and information?

What the question is about and why it is important

A good quality publication will enable you to find further information about testing for a genetic condition. This is important because the publication may not provide you with all the information you need for making an informed choice, and you should be able to trace further information easily.

Rating the question

Additional sources of support and information can be reading materials or organisations such as charities and research institutes. They are often included under headings such as *Useful addresses, Resources, Contacts* and *Further Reading*. A *Bibliography* or *Reference List* also provides additional information (as well as providing the sources of the information for the publication – see Question 16). In an online publication, access to other online information may be provided through links within the text or under the above headings on the site menu or map.

The details provided should enable you to find additional information easily. If an online link is provided, this should be functional and take you to another website or publication where clear details of the information producer are available. If organisations are listed, contact details should be included. If printed publications are listed, the details should include the title, and the author, producer and publisher (and date and ISBN if it is a book).

Many information producers such as national charities or government health departments list the contact details of their own organisation's branches. Whilst these may be useful for information about local services and support groups, they are unlikely to provide different information or perspectives on testing choices, and you should be cautious about giving a high rating to a publication that only provides these details.

Please note: if the publication lists *any* additional sources, it will rate above 1 on this question. There is no “gold standard” for a minimum number of additional sources listed.

Guidelines for rating the question:

5: Yes – the publication provides full details of *any* additional sources of information other than local branches of the same organisation

2-4: Partially – the publication provides details of additional sources, but the details are incomplete or only consist of local branches of the same organisation.

1: No - No additional sources of information are provided.

Examples

5 rating:

Example (i) Antenatal testing- Down's Syndrome in the UK (a fictitious leaflet using real references & organisations)

A health authority leaflet about genetic testing for Down's Syndrome during pregnancy concludes with two sections – *References* and *Advice and Support Services* as follows:

References:

1. Royal College of Obstetricians and Gynaecologists (RCOG). *Amniocentesis and chorionic villus sampling* Greentop Guideline No.8 (3rd Edition). RCOG: London, January 2005. Available free from the RCOG Website: www.rcog.org.uk/resources/Public/pdf/amniocentesis_chorionicjan2005.pdf
2. Alfirevic Z, Sundberg, K. Brigham S. *Amniocentesis and chorionic villus sampling for prenatal diagnosis*. The Cochrane Database of Systematic Reviews 2003, Issue 3. Art No: CD003252. DOI: 10.1002/14651858.CD003252. Available free online - The Cochrane Library 2006 Issue 1 www3.interscience.wiley.com/cgi-bin/mrwhome/106568753/HOME. Review abstract and Plain English Summary available at <http://www.cochrane.org/reviews/en/ab003252.html>

Advice and Support Services

Antenatal Results and Choices (ARC): A national charity providing non-directional information and support to parents on all aspects of antenatal screening and testing. 73 Charlotte Street, London W1T 4PN. Helpline: 0207 631 0285, Tel: 0207 631 0280, www.arc-uk.org.

Testing for Down's Syndrome in Pregnancy: A booklet produced by UK National Screening Committee, Institute of Health Sciences, Old Road, Headington, Oxford OX3 7LF. Tel: 01865 226 666. www.screening.nhs.uk/downs/home.htm (the booklet can be downloaded free from the website).

Contact a family: A national charity providing support to families with disabled children, including those with genetic conditions. 209-211 City Road, London EC1V 1JN. Helpline: 0808 808 3555, Tel: 020 7608 8700. www.cafamily.org.uk.

Down's Syndrome Association: A national charity providing information and support for living successfully with Down's Syndrome. Langdon Down Centre, 2a Langdon Park, Teddington, TW11 9PS. Tel: 0845 230 0372. www.downs-syndrome.org.uk

Partially rating:Example (ii) Inherited cancer

A website produced by a national cancer charity provides information on genetic testing procedures. Under a section entitled *Further Support*, it lists contact details of its branches throughout the country, including web links where available.

Additional rating notes: No independent resources are listed.

1 RatingExample (iii) General

A leaflet produced by a genetic testing company is on display in a local pharmacy. It provides detailed information about several genetic conditions with onset in adulthood, including background effects, causes and available treatments. It describes the tests the company produces that are on sale direct to the public and gives the company's contact details. However, there are no additional sources of support or information mentioned anywhere in the leaflet.

Question 16 – Is it clear what sources of information were used to compile the publication?

What the question is about and why it is important

A good quality publication should make it clear where the evidence for the information about genetic testing has come from. Information about genetic testing and screening choices should be accurate and based on the best available research evidence. Although DISCERN Genetics cannot be used to tell you whether the information you are reading is true or based on sound evidence (as this would require systematic checking against other sources), it *does* enable you to check whether the sources of information are explicit. Details of the sources are important, as they enable you to check the evidence or may encourage you to seek further information. Sources of evidence can include research articles and books (including online publications), and the opinions of experts such as clinicians and representatives from charities and self-help organisations

Rating the question

The hints guide you to look for details of the sources used to compile the text. Ideally, evidence presented in the text and details of its source should be linked. So, there are two main features to look for when rating this question:

- a) in the text, a main statement or “fact” about genetic testing or cited research should be accompanied by a reference to the source of evidence for that information. This could be a name, quote, superscript number, or online link which will lead you to ...
- b) details of that source in a Bibliography or Reference list, Contacts or Additional Information list, or the actual source online

Other important points to note:

- c) the author or producer is not considered a source for this question, as this information is nearly always provided and will not necessarily help you discriminate between good and poor quality publications. You need to know where the author or producer got their information from.
- d) resources and publications rated as *additional* sources of support or information (Question 15) are not as the sources of *evidence* used to compile the publication unless they are acknowledged explicitly as a reference in the text as outlined in (a) above.
- e) it is possible that the publication is based on a single source of information - it can rate highly on this question provided this is clear. Concern about the bias or balance of a publication based on a single source is a separate issue that is addressed more broadly in Question 18.

- f) rating the quality of the sources or evidence is a separate issue beyond the scope of DISCERN Genetics

Guidelines for rating the question:

5: Yes – the sources of evidence are very clear and the publication enables you to link the source mentioned in the text to full reference details.

2-4: Partially – the sources of evidence are clear to some extent. Partial ratings would apply in any of the following cases:

- sources are provided for some but not all of the main statements or facts (you may have to use your judgement to decide when reference to a source would be expected).
- the relationship between the text and the sources is unclear
- full details of the sources are not provided (so it would be difficult to trace them)

1. No: No sources of evidence are mentioned.

Examples

5 rating:

Example (i) Antenatal diagnostic testing – Amniocentesis (Leaflet): (A fictitious leaflet using real data and references - see also Question 7 Example (ii))

Amniocentesis during the second trimester of pregnancy is associated with a slightly increased risk of miscarriage (approximately 1% or 1 in 100 amniocentesis procedures are associated with miscarriage^{1,2}). This risk is higher earlier in pregnancy which is why amniocentesis is not usually offered before 15 weeks.^{1,2}

The end of the publication includes a reference list as follows:

References

1. Alfirevic Z, Sundberg, K. Brigham S. *Amniocentesis and chorionic villus sampling for prenatal diagnosis*. The Cochrane Database of Systematic Reviews 2003, Issue 3. Art No: CD003252. DOI: 10.1002/14651858.CD003252. Available free online - The Cochrane Library 2006 Issue 1 www3.interscience.wiley.com/cgi-bin/mrwhome/106568753/HOME. Review abstract and Plain English Summary available at <http://www.cochrane.org/reviews/en/ab003252.html>

2. Royal College of Obstetricians and Gynaecologists (RCOG). *Amniocentesis and chorionic villus sampling* Greentop Guideline No.8 (3rd Edition). RCOG: London, January 2005. Available free from the RCOG Website: www.rcog.org.uk/resources/Public/pdf/aminiocentesis_chorionicjan2005.pdf

Example (ii) Inherited cancer (A totally fictitious example):

An international cancer charity website provides the following information on genetic testing:

A review of the possible causes of this form of cancer published by A Foundation suggests that only about 1 in 100 cases are inherited. In 2003, scientists identified a genetic mutation amongst a large sample of families with a history of the disease which has led to the development of A Genetic Test. The test can detect the mutation with 85% accuracy and is now widely available. Research is ongoing to develop more sensitive and appropriate tests for the remaining 15% of cases that cannot be detected using this technique.

The underlined text links to the named organisation's websites and to online research journals and reports.

Partially rating:

Example (iii) Antenatal testing

A leaflet ends with a list entitled *References/Further Reading*. The list consists of 10 books and journal articles. There is no mention of these publications in the text.

Additional rating notes: It is unclear which of these publications are sources and which are additional publications providing supplementary (and possibly different) information.

Example (iv) Neurological condition

A research charity website provides details of a new genetic test for an inherited neurological condition. Facts about the test – including test accuracy, procedure and risks – are attributed to a leading neurologist at a research institute. The name of the doctor and organisation are given, but there is no online access or printed details of the contact address or any publications.

Additional rating notes: There is not enough information to locate the source.

Example (v) General

A leaflet on genetic testing for an inherited disorder produced by a hospital genetics department consists of several sections:

About this condition

Testing for the condition

Treatment and management of the condition.

Each section is several paragraphs long.

Two references are quoted in the text: one relates to the discovery of the genetic mutation in *About this condition* and one describes a trial of a new drug therapy under *Treatments*. A *Bibliography* at the end of the leaflet provides the full details of the research articles for each of these references. The text also describes guidelines on testing for this condition, a global survey of families at risk, and several other therapies, but no references in the text or bibliography are provided relating to these issues.

Additional rating notes: Sources are provided for some but not all of the main statements or facts included in the text.

1 Rating

Example (vi) Neonatal testing

A maternity hospital leaflet for parents provides information on neonatal screening. It includes a brief description of the conditions being detected and the test procedure, which is a blood spot test. The final page ends as follows:

This information was compiled by staff at The Maternity Hospital using research evidence.

© *The Maternity Hospital 2004*

Additional rating notes: details of the research evidence and other sources used are not provided.

Question 17 – Is it clear when the information used or reported in the publication was produced?

What the question is about and why it is important

A good quality publication will make the date of the information about genetic testing explicit. Evidence and policies for genetic testing can change and it is important that information includes the latest developments in knowledge and practice. Although DISCERN cannot be used to assess how “up-to-date” the information is (as the rate of change will vary with each genetic condition and test), it *does* enable you to assess whether the age of the information is explicit. This may lead you to question whether the information is current and to seek further information about the most recent developments.

Rating the question

Rate the publication according to how clearly it indicates the age of the information. The hints guide you to look not only for the age of the publication itself, but also for the age of its *content*. The publication cannot be older than the sources of evidence used, whereas the sources of evidence can be much older than the publication. Therefore, in order to score highly on Question 17, the dates for the sources identified in Question 16 should be clear - you should be cautious about giving a high rating to a publication that has rated **1** on Question 16.

Hint 1: Sources - Dates will be found either in the text or in the details of the bibliography or reference list (see Examples for Question 16). Sources such as journal articles should provide the date of the issue when the article was first published. If the source is a website or online reference, you will need to look for an indication of the age of the content – by applying similar questions to that source. If tracking down a date for the information proves difficult, this may lead you to conclude that it is unclear.

Hint 2: Books usually list the date of publication (**copyright** date ©) on one of the title pages. Leaflets should also provide a publication date. Websites should provide a publication date for the site (often found on the home page), and should also give the date of any updates on relevant sections within the site.

A **revised** publication has been substantially changed or updated since the publication (copyright) date and may therefore include more recent information. A **reprinted** publication will not contain new information and should therefore be rated according to publication (copyright) date.

Be wary with **online information**, as **revisions and updates** often only involve changes in presentation or mechanics (e.g. graphics, navigation, links). A Web page that was “updated” yesterday or appears very different from the last time you visited may still contain *content* that has not changed in a very long time! For these publications, you will

need to consider whether there is any additional detail about the scope of revisions and updates, including any updating policy as outlined in Hint 3.

Hint 3: an updating policy will tell you how *new* material is incorporated within a publication, and what it consists of. At present, there is no consistent method for presenting this detail. It is often found in the producer's editorial policy in printed and online information. Websites may also include this information under descriptive or legal sections relating to the producer or site, including *About Us* or *Terms and Conditions*. It is also becoming common to publish a date for a scheduled review alongside the copyright or recent review date on a web page: this provides some clues to the information currency and the producer's updating policy.

Guidelines for rating the question:

5: Yes – the date of the information used or reported in the publication is clear (there are dates for all acknowledged sources)

2-4: Partially – the date of the information used or reported in the publication is unclear or incomplete (only the date of the publication itself is given, or dates for some but not all acknowledged sources have been given).

1: No: No dates have been given.

Remember: although the dates of the sources may be clear and the publication gets a high rating on this question, it is possible that the information on which the sources is based is not “up-to-date”. Question 17 will enable you to judge whether the dates are explicit, but not whether the information is current.

Examples

5 Rating:

Example (i) Antenatal Testing (Leaflet) – Question 16 Example (i) (fictitious leaflet with real academic references):

The end of the publication includes a reference list as follows:

1. Alfirevic Z, Sundberg, K. Brigham S. *Amniocentesis and chorionic villus sampling for prenatal diagnosis*. The Cochrane Database of Systematic Reviews 2003, Issue 3. Art No: CD003252. DOI: 10.1002/14651858.CD003252. Available free online - The Cochrane Library 2006 Issue 1 www3.interscience.wiley.com/cgi-bin/mrwhome/106568753/HOME. Review abstract and Plain English Summary available at www.cochrane.org/reviews/en/ab003252.html

2 Royal College of Obstetricians and Gynaecologists (RCOG). *Amniocentesis and chorionic villus sampling* Greentop Guideline No.8 (3rd Edition). RCOG: London, January 2005. Available free from the RCOG Website: www.rcog.org.uk/resources/Public/pdf/aminiocentesis_chorionicjan2005.pdf

The bottom of the back page of the leaflet also displays the following:
© 2006 *The Health Authority. Due for revision February 2008.*

Example (ii) Inherited Cancer (Website) – continuation of Question 16 Example (ii) (all details fictitious).

Sources in the text:

- underlined text provide links to an external website or journal article providing more detailed information which also contains a copyright date and update information.

The Website itself:

- © 2003 appears at the foot of each Web page within the site.
- The organisation's *Updating* policy is a clearly labelled section within the *Editorial Policy* in *About Us*. It describes how and when the content of all its publications – printed and online – is reviewed. The review dates are represented as a *Content last reviewed* date on all its information. This appears alongside Copyright date and Page Modification date on each web pages as follows:

© 2003

Content last reviewed 30th May 2005.

Page last modified 15th March 2006

Partially:

Example (iii) Neonatal screening (rated '1' on Questions 15 and 16)

A maternity hospital leaflet for parents provides information on neonatal screening. It includes a brief description of the conditions being detected and the test procedure, which is a blood spot test. The final page ends as follows:

This information was compiled by staff at The Maternity Hospital using research evidence.

© *The Maternity Hospital 2004*

Additional rating notes: there are no sources mentioned anywhere in the leaflet. The only date available is the copyright date indicating when the leaflet was first published.

Example (iv) General (Website)

The home page of a local charity website contains a footnote *Copyright 2001, Last updated March 2006*. There is no detail anywhere on the site to indicate what the updating involved. The only source mentioned is a news article from an international health bulletin board - a link takes you to the online article whose only date details are *Posted 13 January 2005*.

1 rating

Example (v) General (Leaflet)

A leaflet produced by a genetic testing company is on display in a local pharmacy. It provides detailed information about several genetic conditions with onset in adulthood, including background effects, causes and available treatments. It describes the tests the company produces that are on sale direct to the public and gives the company's contact details. It states that people with these conditions *can now live longer thanks to recent breakthroughs in testing and treatment*. There are no cited references and no dates given anywhere in the leaflet.

Question 18 – Is it balanced and unbiased?

What the question is about and why it is important

A good quality publication will provide fair and impartial information. It is important that information about genetic testing is presented in a way that enables you to choose what is in *your* best interests. A publication should be honest and informative. It should not influence you by “promoting” particular tests or by using “shock tactics” to influence your decision about testing.

Rating the question

Your rating should be based on your impression of the information about genetic testing as a whole. The hints will help you develop a “feel” for the balance and bias of the information, but your own judgement will also be important. Here are some additional points to help you:

- you should judge the information on its own merits, using the appraisal system outlined in DISCERN Genetics. Try not to be influenced by **what you know about the author or producer** - well-respected individuals (including doctors, researchers and charities) can produce poor quality information, and an unknown author or producer can produce information that meets high standards.
- the information about the test should be drawn from a **range of research and experience**. You should be cautious about giving a high rating on this question to a publication that has not revealed any sources or relies solely on a single source of evidence (Question 16).
- ideally, a publication should be **independently reviewed and approved** by experts or professional organisations and by patient groups. Evidence of an external assessment provides readers with some assurance that the information is unprejudiced. The assessment may be acknowledged in a declaration alongside copyright and production information, or in an Editorial Policy. However, this is not yet common practice and there are no consistent standards as to how this information should be collected and displayed.

Guidelines for rating the question:

5: Yes - the information is balanced and unbiased

2-4: Partially - some aspects of the information are unbalanced or biased

1: No - the information is completely unbalanced or biased

Examples

We have devised simple examples here to demonstrate the quality criterion underlying Question 18. However, the information rated by this question should also have clear sources and referencing where appropriate as outlined in Question 16. For further guidance and examples, please refer to the section on Question 16 in the Handbook

The simplest way of demonstrating this question is to provide fictitious examples of high and low scoring publications:

5 rating:

A commercial online family health encyclopedia (funded mainly through onsite advertising) includes a section on antenatal testing for a variety of genetic conditions. The information is attributed to an editorial team and their details are provided under *About Us* alongside information about the producer organisation and the editorial process. The information consists of an overview of each genetic condition, management and treatment options, and an analysis of the advantages and disadvantages of several diagnostic tests, including their accuracy, risks and interpretation. The authors also refer to the possibility of variation according to healthcare provider and locale. The implications of testing and the consequences of different decisions following a positive result are outlined and linked to personal testimonies from parents who chose a termination and from parents who chose to continue the pregnancy. The site provides links to online scientific sources and to sources of additional support information. The Home Page includes a statement of the producer organisation's subscription to international health publisher standards and an endorsement from an independent public health research foundation. An online feedback page is also included with access to comments and reviews of the website from individual users and patient organisations.

1 rating:

An international genetic testing company website describes its testing service for an inherited neurological condition. The service is provided by two doctors, and a list of their medical qualifications is provided under *About Us*. They describe their service as *internationally recognised for excellence in genetic testing and preventive healthcare* although no external endorsement, accreditation or scientific record is provided to support this claim.

The site includes a brief outline of the condition, describing it as having *unpleasant, degenerative symptoms which are distressing for sufferers and their families*.

Information about testing consists of the following:

We are at the forefront of genetic testing for this condition. We believe our clients have a right to the outstanding benefits offered by the astonishing breakthroughs in modern genetic medicine that will protect their health and that of their loved ones.

If this genetic condition runs in your family or you suspect you have symptoms, we strongly recommend testing to avoid a devastating fate. Early detection means you can benefit from effective prevention and treatments. Clients using our service also enjoy

expert guidance and rapid turnaround time. Our tests are reliable and provide all the facts and reassurance you need. Read our client testimonial and see how you too can prevent this terrible disease becoming your destiny.

Brief descriptions of the test procedure and the cost are given, but no further information is provided about the test such as personal risk assessment, test accuracy or limitations, implications of results, alternative tests, or broader personal issues. No sources of evidence or sources of additional support and information are listed, and there are no independent endorsements.

Question 19 – Is information provided on local availability of services and test performance?

What the question is about and why it is important

A good quality publication will highlight geographical variations in testing services. Genetic testing often involves rare genetic conditions and procedures that are still highly experimental or expensive. Tests and treatment may not be routinely available throughout your area – you may need to travel to a specialist unit or pay for services privately. It is also possible that testing standards (such as accuracy, procedural risks and waiting times) vary because of differences in the availability of special equipment and skilled staff. It is important to be aware of these issues when deciding about testing.

This question is only relevant to information about access to services in a specified area - the clinics or hospitals *you* may have access to. Information intended for a more general or international audience should not be rated.

Rating the question

Rate the question according to whether it provides details of local availability of services and test performance. How high you rate it will depend on your judgement of how well the relevant information has been covered.

Guidelines for rating the question:

5: Yes – details of local availability of services and test performance are provided

2-4: Partially details of local availability of services and test performance are provided, but the information is unclear or incomplete

1: No – no details of local availability of services and test performance are provided (where they would be expected)

N/A – this question is not appropriate for this publication and should be rated “Not applicable”.

Examples

We have devised simple examples here to demonstrate the quality criterion underlying Question 19. However, the information rated by this question should also have clear sources and referencing where appropriate as outlined in Question 16. For further guidance and examples, please refer to the section on Question 16 in the Handbook

5 Rating:

Example (i) Inherited cancer

A national charity website provides information about genetic testing for an inherited cancer:

A genetic test for this condition is currently available free at all local hospitals to everyone with a strong family history of this cancer (you will need assess your risk with your family doctor first). However, the test was developed at a time when all variants of the underlying genetic mutation had not been identified, and can only detect 7 in 10 people with the mutation. Recently, a more accurate test which can detect a wider range of variants of the genetic mutation (picking up 9 in 10 cases) has been developed. Not all laboratory staff have undergone training to conduct this genetic analysis, so it is only available through a few laboratories nationally for a fee.

However, there is also a national research programme into this cancer which is aimed at providing clearer information about risk factors and disease progression in people with the mutation. Volunteers are offered the new genetic test free of charge at a number of regional hospitals. Transport and accommodation is provided. For more details, speak with your family doctor or contact the research manager at [The University Hospital](#) (link to hospital website).

Partially rating:

Example (ii) General

Most hospitals offer the test, although some charge a fee. Waiting time for results ranges from 2 to 10 weeks.

Additional rating notes: there is no information about which hospitals do and don't provide the test or charge a fee. There is no explanation of why waiting time for results varies, or where these variations occur

1 rating:

Example (iv) Cystic fibrosis

A popular book on child health produced by a national organisation of health professionals and distributed through local pharmacies includes details of a range of screening testing procedures for detecting cystic fibrosis both before and after birth. It provides a detailed description of the condition, including current understanding about its genetic causes and the ways in which different tests detect the condition. However, there is no mention of local availability or performance of any of the tests described.

Question 20 – Based on the answers to all of the above questions, rate the overall quality of the information as a source of information about genetic testing and screening

What the question is about and why it is important

Question 20 is an ‘intuitive summary’ of your responses to the preceding 19 questions. All publications will have deficiencies, and it is unlikely that any one publication will rate high on all of the questions. However, after completing all of the questions on DISCERN Genetics, you should have developed some feeling for the overall quality of the publication which will help you decide whether it is a useful and appropriate source of information about your choices about genetic screening and testing.

Rating the question

The guidelines below should help you to rate this question, but your judgement is also very important. It may encourage you to know that during the development and testing of the DISCERN Genetics tool, users independently reached very similar conclusions about the overall quality of the publications they were rating even though we did not provide very specific instructions for this question. DISCERN Genetics has been designed to help you develop your critical skills, so trust your own opinion!

Guidelines for rating the question:

- **High (5)** - The publication rated high (4 or above) on the majority of questions. A high overall quality rating indicates the publication is “good” quality - it is a useful and appropriate source of information about choices for genetic testing.
- **Moderate (3)** - The publication rated high and low on a similar number of questions, or the majority of questions rated in the mid ranges (3). A moderate overall quality rating indicates the publication is “fair” quality - it is a useful source of information about genetic testing but has some limitations. Additional information or support would be needed.
- **Low (1)** - The publication rated low (2 or below) on the majority of questions. A low overall quality rating indicates the publication is “poor” quality - it has serious shortcomings and is not a useful or appropriate source of information about genetic testing.