Patient

View this article online at: patient.info/doctor/genetic-counselling-a-guide-for-gps

Genetic counselling - a guide for GPs

What is genetic counselling?

Genetic counselling is not primarily "counselling" in the psychological sense. Genetic counselling is non-directive and aims to explain the facts as clearly as possible, giving the person or family accurate information on their options in a way which they can understand, and helping them to make up their own minds.

Genetic counselling should be regarded as an integral part of the genetic testing process and should be offered and strongly recommended in most genetic testing situations. If an individual insists on having a test without genetic counselling, the medical facts and possible consequences should be discussed by the clinician ordering the test.

It is now possible to sequence the whole genome of an individual. This type of testing could provide a large amount of information about a range of disease susceptibilities for individuals, rather than the targeted use of genetic testing currently.[1] Genomic medicine is a strategic priority for many healthcare systems and therapeutic conversations with genomic data may be more common in the future for GPs.[2] The NHS is introducing whole genome sequencing through the NHS Genomic Medicine Service.[3]

Types of genetic testing

- **Diagnostic testing**: genetic testing performed in a symptomatic individual to confirm or exclude a genetic condition.
- **Predictive testing**: genetic testing in a healthy high-risk relative for a specific later-onset monogenic disorder.
- **Susceptibility testing (risk profiling)**: a genetic test of a marker or several genetic markers with the aim to detect an increased or decreased risk for a multifactorial condition in a healthy individual.

- **Pharmacogenetic testing**: testing for a genetic susceptibility for adverse drug reactions or for the efficacy of a drug treatment in an individual with a given genotype.
- **Carrier testing**: a genetic test that detects a gene mutation that will generally have limited or no consequence to the health of that individual.[4]
- **Prenatal testing**: a genetic test performed during a pregnancy, where there is increased risk for a certain condition in the fetus.[5]
- **Pre-implantation genetic diagnosis**: testing the presence of a mutation, linked haplotype or chromosomal change in one or two cells of an embryo in a family with a previously known risk for a Mendelian or chromosomal disorder, in order to select the unaffected embryos to be implanted.
- **Genetic screening**: testing where the target is not high-risk individuals or families, but where the test is systematically offered to the general population or a specific group (eg, newborns, young adults, an ethnic group).

Guidelines for genetic counselling

- Genetic counselling has to be provided or supervised by a healthcare professional appropriately trained for genetic counselling.
- Non-genetics healthcare professionals have a responsibility to recognise their abilities and limitations with regard to provision of genetic services.
- Healthcare professionals should not agree to testing without pre-test counselling in circumstances where doing so would go against their professional judgement.
- Predictive tests for future severe illnesses with no options for treatment or prevention should not be performed without pre- and post-test genetic counselling, psychosocial evaluation and follow-up.
- Before actual testing takes place, there should be free and informed consent.

- In situations where testing children or other persons who are not able to give informed consent is considered, those individuals should be involved in genetic counselling and in the decision-making process, according to their capacities.
- Testing for adult-onset conditions in children should only be considered when treatment or surveillance would begin in childhood.

Pre-test genetic counselling

Individuals are informed about the purpose of the test, including:

- Up-to-date, reliable description about symptoms and natural history of the disease.
- Prospects of prevention or early diagnosis and treatment.
- Inheritance pattern.
- The risk of disease, available reproductive choices, reliability and limitations of the test concerned, and possible psychological impact and other consequences of the test result to the person and their relatives.
- Privacy and confidentiality of the results, as well as possible consequences related to its disclosure to third parties, such as insurance companies and employers, are discussed, when appropriate.[6]
- Pre-test counselling includes discussion about the rights to know and to decide including the right not to know.
- Possible uncertainties due to present lack of knowledge are declared.
- Discussion about the need to inform relatives about the test result, as well as the best ways to do this, are initiated, especially in conditions where early diagnosis may improve the prognosis.
- Written materials and/or reliable Internet addresses related to the subject should be offered when available.
- A written summary of the discussion should be offered.

- European guidelines on genetic counselling for pre-symptomatic testing have been developed. These include general principles governing the offer of testing (eg, autonomous choice of the patient), objectives of genetic counselling in this context (eg, facilitation of decision making), logistical considerations (eg, use of trained staff) and topics to be included during counselling discussion with the patient (eg, consequences of both positive and negative outcomes).[7] There are also European guidelines available for prenatal diagnostic testing, incorporating the counselling which should be a part of this, in order for parents to be able to make informed decisions.[8]

Post-test genetic counselling

- After disclosure of test results, the first focus is on the emotional impact on the person and others involved.
- If necessary, follow-up contacts with the genetic counselling unit should be offered, and/or a consultation with a psychologist.
- The possibility to contact a social worker and patient support organisations should also be offered.
- A written summary of the test result and issues discussed should be given.
- Implications to the individual (including a follow-up plan, when relevant) and their near relatives should be discussed
- A strategy to inform relatives should also be discussed. Written material to help the counsellee to spread the information in the family should also be offered.

Role of primary care in genetic counselling

GPs are now frequently asked about inherited diseases in the context of both:

- The possibility of an individual patient having an increased risk of a condition which already affects a close member of their family.
- The risks of a couple having a child affected by a particular disorder that may or may not appear in the family.

The GP may need to:

- Explain mechanism of inheritance of a disease to patients.
- Dispel unnecessary fears in patients without significantly increased risk of disease.
- Advise on lifestyle changes for patients who may have inherited an increased susceptibility to a disease - eg, coronary heart disease.
- Provide advice and support to people where consultation with a clinical geneticist may be clinically indicated - eg:
    - People with relatives or offspring with recognisable inherited diseases - eg, cystic fibrosis, Down's syndrome, beta thalassaemia.
    - Ethnic groups at special risk - eg, Ashkenazi Jews.
    - People with a clear family history of high levels of certain forms of cancer occurring at an early age - eg, breast, colorectal, ovarian and endometrial cancers, familial adenomatosis polyposis.[9] [10]
- Help people come to terms with a diagnosis or a high risk in themselves, their baby or a family member. Explain conditions and their inheritance to them when a relative has informed them they need testing. This may require some reading and another appointment, in order to be able to be adequately informed about a condition in order to explain it to somebody else
- Advise on antenatal screening for disorders where there is a strong family history such as muscular dystrophy, cleft lip, spina bifida, congenital heart defect. There is a need to ensure that parents are aware of the alternatives for the pregnancy when screening is positive.
- Advise on the alternatives to normal conception - eg, in vitro fertilisation with embryo selection, adoption, sperm or egg donation.

A number of resources are available for GPs wishing to learn more about genetic testing, counselling, implications and communicating genetic information, including the NHS e-learning hub and the NHS England Genomic Education Programme. [11] [12] The NHS National Genetics Education and Development Centre contains information and resources on learning and teaching genetics for health professionals, including fact sheets on genetic conditions. [13]

Counselling the person, couple or family

- Counselling should be carried out in a relaxed atmosphere with sufficient time to absorb the initial shock of diagnosis, or bereavement.
- Advice should include the clinical presentations of the disease, treatment, natural history, prognosis, complications and a clear explanation of the genetics. The risk to the individual of developing symptoms, the risk to future offspring, and the way in which the disease is transmitted.
- All information must be given in simple, easy to understand language.
- Carefully assess the understanding of the couple about the problem: establish whether they have any misconceptions which need rectifying, or any misplaced guilt.
- The reproductive options of the couple must be discussed in an unbiased manner. If required, contraceptive measures should be arranged and the possibility of in vitro fertilisation or adoption investigated.

As a GP, good communication is key in all consultations. The added issues involved in communicating genetic information include:

- Impact on others (and subsequent confidentiality issues, and guilt feelings that may arise if a person feels they have passed on a genetic condition to a relative).

- Language of genetics, which may not be understood. Terms such as "risk", "mutant", "disease" are better avoided and replaced with more neutral words such as "chance", "variant" and "condition" respectively. Terms such as "carrier" may be misinterpreted as "infectious". Numbers and percentages may not be understood.
- Cultural or ethical differences.
- Uncertainty.

It is even more important in this area to establish what people understand and what their beliefs, concerns and priorities are, to double-check they have understood what has been explained and to involve them fully in decision-making.

This is just as important for children and for adults who do not have capacity, even where others are making decisions on their behalf. An explanation should be provided at a level appropriate to their comprehension ability.

Indications for referral to a genetics centre

Common reasons include:

- A person with a known genetic condition in the family, wanting to know their own risks and/or those of their children.
- Parents of a child with difficulties which may be due to a genetic condition, referred to see if a diagnosis can be made.
- A person with a strong family history of cancer, wanting to know if they are at increased risk and, if they are, what options they have.[14] There are specific National Institute for Health and Care Excellence (NICE) guidelines for indications for referral for genetic testing for women with a family history of breast cancer.[15]
- A person with a known genetic condition wanting specialist advice about the condition.
- A couple whose baby has been diagnosed with a genetic condition following routine newborn screening.
- A person with a possible genetic condition in the family wanting to know if a diagnosis can be made and, if so, their risks and options.

- A pregnant couple told that a test has given an abnormal result, wanting to talk about what the result means and what options are available.
- To help with diagnosis with a child with learning delays or physical features for which a genetic condition is suspected to be the cause.

Further reading

- Genetic testing and counselling; Macmillan Cancer Support
- Albada A, Werrett J, Van Dulmen S, et al; Breast cancer genetic counselling referrals: how comparable are the findings between the UK and the Netherlands? J Community Genet. 2011 Dec;2(4):233-47. Epub 2011 Aug 18.
- Resources for healthcare professionals; The British Society for Genetic Medicine (BSGM)
- Jamal L, Schupmann W, Berkman BE; An ethical framework for genetic counseling in the genomic era. J Genet Couns. 2020 Oct;29(5):718-727. doi: 10.1002/jgc4.1207. Epub 2019 Dec 19.

Disclaimer: This article is for information only and should not be used for the diagnosis or treatment of medical conditions. Egton Medical Information Systems Limited has used all reasonable care in compiling the information but makes no warranty as to its accuracy. Consult a doctor or other healthcare professional for diagnosis and treatment of medical conditions. For details see our conditions.

Authored by:	Peer Reviewed by: Dr Krishna Vakharia, MRCGP	
Originally Published: 20/11/2023	Next review date: 21/08/2023	Document ID: doc_2186

View this article online at: patient.info/doctor/genetic-counselling-a-guide-for-gps

Discuss Genetic counselling - a guide for GPs and find more trusted resources at Patient.

Patient Access

To find out more visit www.patientaccess.com
or download the app

Download on the App Store

GET IT ON Google Play

Follow us