



HUMAN GENETICS SOCIETY OF AUSTRALASIA

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Guideline

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1.0 INTRODUCTION

Genetic counselling is a communication process, which aims to help individuals, couples and families understand and adapt to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions. This process integrates the following:

- Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
- Education about the natural history of the condition, inheritance pattern, testing, management, prevention, support resources and research.

- Counselling to promote informed choices in view of risk assessment, family goals, ethical and religious values.
- Support to encourage the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder (1).

The process of genetic counselling is complex and information must be presented in a meaningful and non-judgemental way and be relevant to the needs of the clients. The goal is to facilitate the clients' ability to use genetic information in a personally meaningful way that minimises psychological distress and increases personal control (2). The diagnosis and information discussed in genetic counselling may have implications for other family members and this can impact on relationships and family dynamics. The process of genetic counselling may involve multiple phases, including pre-clinic contact with clients, clinical consultation, follow-up and review. It also involves record keeping, literature and database searches.

These guidelines have been developed to ensure that health professionals, who provide genetic counselling, have access to the current *best practice standards* in Australasia. It is also considered important to encourage and maintain consistency of practice among genetic health professionals throughout Australasia, as relatives of the same family may be seen in different genetic units. It is acknowledged that other health professionals may be involved in the provision of genetic counselling, particularly complex genetic conditions that require a multidisciplinary focus. However, the HGSA recommends that the core provision of genetic counselling be conducted by appropriately trained professionals to maintain high standards and consistency. Guidelines for the training of clinical geneticists and genetic counsellors can be found on the HGSA website: www.hgsa.com.au. It is also acknowledged that different clinical genetics units and/or services may function differently and are administered by various public and private health facilities.

It is also important to note that, in Australasia, genetic counsellors, who must complete an approved post-graduate program, work in partnership with clinical geneticists or other medical specialists. Medico-legal responsibility for genetic consultations will vary between services (public and private) and may rest with the medical specialist, the institution or the individual's private liability cover.

2.0 REFERRALS

2.1 Receipt of referral / Intake

This will usually be a referral letter or phone call from the person referred or from the referring health professional. Written referrals are preferred as this facilitates continuity of care following the consultation, although self-referrals may be acceptable to some units. Referrals should be assessed regarding their urgency and appropriateness and appointments arranged at the next intake meeting or earlier as required.

All clinical genetics units should have a regular intake meeting where decisions regarding referrals will be determined. For genetics units Levels 1 and 2 (see HGSA document: [Structure of Clinical Genetics Units in Australasia](#) (1999 GL01)), regular email or phone communication may be substituted. Regular intake meetings provide an opportunity for the genetics team to:

- review planned appointments;
- allocate referrals to a clinical geneticist or a genetic counsellor as appropriate;
- make decisions regarding referrals which may have been inappropriately received and;
- assess the urgency of referrals.

2.2 Assessment of appropriateness of referral against criteria

The appropriateness of the referral is assessed against the following criteria:

- client has, or is suspected of having a genetic condition;
- client has an undiagnosed condition or birth defect;
- client is a carrier for a genetic condition or is concerned about the possibility of being a carrier;
- client is from a population known to be at increased risk for a particular genetic condition;
- client has a relative with a genetic condition and may be at increased risk for, or concerned about, developing the disorder;
- fetal abnormality has been detected in pregnancy or there is concern about risk of a fetal abnormality, including risk associated with exposure to potentially teratogenic agents;
- client has advanced maternal age, high risk prenatal screen result, unexplained infertility or recurrent miscarriages;
- individual or family requires additional information about, or interpretation of, a genetic test result i.e. newborn screening result, prenatal screening results;
- client is in consanguineous relationship.

The genetic counsellor or clinical geneticist should notify the referring health professional if a referral is deemed inappropriate.

2.3 Recommendations for Clinic Waiting times

Urgent Referrals

- Local and specialty clinics – next available appointment depending on degree of urgency.
- Outreach clinic - next available clinic or urgent referrals may need special arrangements. Client may need to be referred to a major genetics unit or seen by the local genetic counsellor. Outreach genetic counsellors who co-ordinate clinical geneticists' clinics are advised to leave one clinic appointment unfilled until the week prior to clinic, in order to meet the demands of urgent unplanned referrals.
- Alternatively, some clinics / health professionals may utilise telemedicine consultation

Non-urgent referrals should be seen by a genetic counsellor and / or clinical geneticist within the following time frames:

- local clinic – Approximately 6 weeks
- specialty clinic – Approximately 12 weeks
- outreach clinic - Next available clinic, usually less than 8 weeks

2.4 Management of non-urgent referrals

- ensure that the referring health professional has sent a referral letter;
- identify the information needed for the clinic appointment ;
- allocate cases to the most appropriate clinic eg. general or specialty clinic at clinical genetics unit or at an outreach site;
- allocate cases to the most appropriate health professional eg. clinical geneticist and / or genetic counsellor, genetic counsellor only, or social worker / psychologist specialised in genetic conditions.

2.5 Notifying clients of clinic appointment

- date, time and place of appointment
- attending health professional(s)
- required information to bring to appointment
- general information about genetics clinic appointments

3.0 THE PROCESS OF GENETIC COUNSELLING

3.1 Pre-consultation contact

The clinic co-ordinator and / or genetic counsellor should contact the client prior to the scheduled clinic appointment in writing, face to face, or by phone. Where possible, contact should be made by a genetic counsellor, and the genetic counsellor may:

- confirm that appointment details were received and that the client is planning to attend;
- outline or review the purpose of the consultation as well as explain the consultation process and aims, including the possibility of a physical examination so that concerns that the client may have regarding the nature and conduct of the consultation may be addressed;
- ascertain the needs and expectations of the client / family (their agenda), identifying any special requirements eg. wheelchair access or interpreter services. Social and/or cultural issues which may impinge on the consultation may be identified by the counsellor;
- gather pedigree information (see HGSA Guidelines: [Certification and Training of Genetic Counsellors](#) (2002 GL01), Section 3 p.37) and identify necessary documentation, such as post-mortem reports, pathology results, and names of attending specialists and hospitals where treatment was received. Signed consent forms may need to be obtained from the client or other relatives to facilitate release of this information and;
- clarify unrealistic expectations of the clinical consultation.

3.2 Preparation for consultation

- check that all information needed for the consultation has been obtained;
- review relevant health unit record and/or clinical genetics unit records;
- discuss the case with supervisor or with relevant colleagues as necessary;
- review relevant medical literature;
- prepare information that will be given to the client, including appropriate support group information and fact sheets as applicable.

3.3 The consultation

The physical setting for the genetic counselling consultation is important. The consultation room and examination facilities should ensure total privacy for the client, as the matters discussed may be confidential. Children should be examined in the presence of a parent or guardian, and females should have another female present if the examining doctor is male.

There should be a limit on the number of professionals present during the consultation. Rapport with the family should not be compromised by the educational needs of professionals and trainees. Consent should be obtained from the client for other health professionals, such as students and trainees, to be present at the consultation.

Elements of the counselling process, which may be addressed over more than one consultation, include:

- setting the agenda of client and counsellor;
- gathering specific health information;
- informing the client about how their information will be stored and who will have access to it;
- making or verifying the diagnosis by history taking, physical examination and use of information obtained before or during the consultation;
- providing information about the condition, which may include its cause, pattern of inheritance, natural history, complications, and treatment options;
- providing information about the risk of a condition affecting the client, their children and/or other relatives;
- discussing the medical, emotional and social implications for the individual and family;
- considering, and discussing with the client, implications for genetic relatives;

- presenting options, including genetic testing and reproductive options and assisting with informed decision making in a non-judgemental / non-coercive manner;
- arranging genetic tests after obtaining informed consent, which may include carrier testing, predictive testing, screening, research testing;
- conveying and explaining test results;
- referring to other health professionals as needed;
- addressing the interests of third parties;
- providing educational material and / or appropriate references;
- offering contact with community based support groups or persons;
- contracting for further genetics appointments if necessary.

3.3.1. Setting the agenda

The agenda for the consultation should be set early, taking into consideration the needs and expectations of the client or family, as well as the responsibilities of the professionals. It is important for the counsellor not to proceed with her / his own agenda without establishing the individual's, couple's or family's needs and perceptions.

3.3.2. Collecting information

Information may be gathered in several ways. Information about the client / family medical history may be available via the referring doctor and hospital / medical records. Results of previous tests, X-rays and other investigations may also be accessible via the referring doctor, hospital / medical records, or directly from the laboratory. The genetic counsellor must be aware of the confidential nature of medical information and always obtain consent from the client before accessing this information.

The construction of a family pedigree is an important part of the genetic counselling process and as family information can be highly sensitive, great care needs to be taken to maintain confidentiality. Health professionals are entitled to record a family medical history if it is necessary for the care of the client.

3.3.3. Access to and disclosure of the clients' information

The client should be informed about the possible uses of their information including their right to access their own information and that their information may be given to another medical practitioner in the course of their management (if relevant) only after consent is given to do so.

3.3.4. Verification of the diagnosis

Clinical geneticists may need to verify a diagnosis by clinical examination, arranging appropriate tests, or by the examination of available records / reports, which may include test reports on relatives, including deceased relatives, and /or post-mortem examination reports. Information gathering that requires access to pathology / post-mortem reports, registry records or genetic test results of relatives will need the consent from the appropriate individuals, or from the next-of-kin or legally responsible person / agency.

3.3.5. Providing information

All genetic health professionals have a responsibility to obtain the most up-to-date information prior to the consultation wherever possible. Information on genetic conditions should be given in a non-biased way, using language and concepts that clients understand and should include:

- the natural history of the condition, range of prognosis, treatment or management options;
- discussion of the limitations of current information and / or tests and the offer to review clients, as appropriate;
- information given to families over a number of sessions;
- referral to more appropriate expert treating professional if required (facilitated via liaison with the referring medical practitioner), as most genetic consultations provide a clinical geneticist's opinion or diagnosis, or are for education and counselling support and rarely involve ongoing management or treatment, with the exception of metabolic genetics cases.

3.3.6. Estimation of risk

Once a diagnosis is made in a family, individual risk can be estimated using empirical data, inheritance patterns, pedigree information, clinical expertise and test results. The counsellor should counsel the client about risk interpretation and the limitations of risk calculations.

3.3.7. Emotional responses and implications for clients

Genetic information often has a profound impact on an individual and their family. This should be acknowledged in the counselling process and clients must be given a safe environment in which to express their emotional / psychological responses. As well as dealing with genetic issues, counsellors should offer emotional support to clients during the consultation and / or in follow-up contact sessions.

Genetic information, if given to other family members or outside agencies such as insurers or employers, has the potential to result in discrimination and stigmatisation so this issue should be discussed during the consultation.

3.3.8. Implications for genetic relatives

A genetic diagnosis or test result may have direct risk implications for other family members and genetic counselling should be made available to them, as appropriate. Support and assistance in conveying genetic information to 'at risk' relatives should be offered by the counsellor.

3.3.9. Presenting options and informed decision making

It is important for the genetic counsellor to check the client's understanding throughout the consultation. To enable the client to make an informed decision:

- all available options need to be presented without bias;
- the advantages and disadvantages of each option discussed and understood;
- the client's reaction to each option and consequences should be explored;
- the client's informed decision should be supported by the counsellor.

3.4 Genetic tests

There are a variety of genetic tests that may be available to clients. The choice of which test is the most appropriate for the client's needs should be determined as part of the genetic counselling process. The client / family should be informed about the nature of the sample required, appropriateness of the test, the information the test is seeking, the limitations of the test, and the possible implications of the result. It is important that clients have some idea of how long results will take and the details of any associated financial cost to the client, and a contract should be agreed for arrangements to give the results. In some cases results might be obtained faster if the test is undertaken via a private pathology service and the client pays for the test. Clients should be informed of this option. The appropriate request forms should be completed and a system of follow-up or tracking of outstanding results must be set in place for efficient reporting and action.

After the client is fully informed about all aspects of the testing process, the appropriate consent forms must be signed and witnessed as appropriate. The types of tests that are available include carrier, screening, diagnostic, predictive, presymptomatic, and donation of samples for research purposes. When a client needs to choose which test they would like to have eg. screening or diagnostic, they should be given all of the information available to enable them to make an informed choice.

3.4.1 Diagnostic test

A diagnostic test is usually ordered by a clinical geneticist to make, or confirm, a suspected diagnosis, or to exclude a differential diagnosis. A clinical diagnosis or ordering a genetic test is reliant upon the judgement and expertise of the clinical geneticist and is within his / her responsibility. Geneticists may delegate the responsibility of arranging a genetic test to a genetic counsellor with appropriate supervision. A prenatal test may be a diagnostic test when carried out on a sample from a developing fetus.

3.4.2 Genetic carrier testing

A carrier test is primarily used to determine if an adult, with or without symptoms, has a genetic mutation, which increases the chance that his / her children will have the disorder in question. Minors should only have carrier testing performed when the resulting information will be used to help with their health management in the immediate future.

3.4.3 Screening tests

Screening tests are usually non-diagnostic, population-based tests, which provide the client with a personalised risk assessment. Newborn blood-spot screening is a public health activity aimed at the early identification of infants who are affected by certain congenital disorders. Timely intervention in these disorders significantly reduces morbidity, mortality and associated disabilities. Other genetic screening may be available to specific 'high risk' groups in order to clarify individual carrier status. When performed prenatally, screening tests, such as ultrasound examination, may identify fetal abnormality and ultrasound +/- biochemical analysis may reveal an increased risk of fetal abnormality. Support and counselling should be made available to persons receiving a high-risk result so that future options are understood.

3.4.4 Predictive tests

These tests are performed on an individual who usually has no symptoms of a specific disorder at the time of testing, to determine whether he / she carries a genetic mutation. If the genetic mutation is present, the individual is at an increased probability of developing symptoms of the disorder. (See HGSA guidelines: [Presymptomatic and Predictive Testing for Genetic Disorders 2005 PO2](#)) In some situations the individual may already have symptoms but be unaware of them.

3.4.5 Pre-symptomatic tests

These tests are performed on an individual who currently shows no symptoms of the disorder. If the specific genetic mutation is present, the individual is almost certain to develop the condition during their lifetime, if they live a normal lifespan. Genetic counselling for this type of test may involve adherence to a widely established protocol. (See HGSA guidelines: [Presymptomatic and Predictive Testing for Genetic Disorders 2005 PO2](#)).

3.4.6 Research tests

These are tests carried out as part of a research study supported by special funding and approved by an institutional ethics committee. The NHMRC's National Statement on [Ethical Conduct in Human Research \(2007\)](#) provides guidelines for genetic testing which is performed as part of a research study.

3.4.7 Conveying test results

Information about test results should be delivered and explained to clients in a non-biased manner. Results must be explained clearly to the client using language that they are able to comprehend.

3.5 Referral to other health professionals

Genetic services may facilitate referral (by the supervising medical practitioner or via liaison with the referring general practitioner or specialist) of an individual or family for:

- a second opinion;
- emotional and psychological support;
- treatment or management options considered to be outside the expertise of the genetics unit.

3.6 Implications for third parties

Clinical genetics health professionals need to be mindful of the implications to third parties of a genetic diagnosis, or the result of a genetic test. Third parties may benefit from the information obtained and this should be discussed with the client. Support in conveying genetic information to 'at risk' relatives should be offered.

Australasian health professionals must comply with the current federal and /or local government privacy laws at all times and may disclose confidential information only as documented by the client (written consent) and / or required for medical management or as permitted / required by law.

All human genetic research requires approval from a Human Research Ethics Committee(s). Some research samples are collected on the understanding that no results will be given to individual participants, and this must be made clear to participants in advance of the consent process. Where individual results are to be disclosed to participants, research groups would benefit from the involvement of a genetic counsellor, preferably before consent to the research is obtained, but especially in association with result disclosure.

Third parties outside the family who may have an interest in genetic test results / diagnoses may include life insurance companies, health insurance companies and employers. Where insurance companies may have an interest, it is important to explain to the client (in advance of the consent process) the likely requirement to disclose to these companies (on their application forms) the genetic diagnosis or genetic test result.

3.7 Educational material

Clients should be provided with educational material or the means of obtaining educational material (eg. fact sheets, leaflets, brochures, internet addresses), which is linguistically and culturally appropriate.

3.8 Community support

Genetic counsellors and/or social workers are able to arrange contact with community based support groups or appropriate individuals or families according to the client's requirements.

3.9 Client follow-up

Client follow-up is an important component of the genetic counselling process. This ascertains that the client understands the information, allows an opportunity for further questions to be addressed and provides further support to the client. It is the responsibility of the health professional to offer re-contact from the client after a clinic appointment in order to address any further concerns. It is the geneticist's responsibility to convey test results and associated implications to the client, however, this role may be delegated to the genetic counsellor involved depending upon the level of counsellor's experience. Some genetic specialist clinics are involved in the treatment and management of clients and provide long-term follow-up.

3.10 Audit

Clinical genetic units should be involved in the appropriate regular review process to ensure that clients consistently receive the highest standard of care.

Further Reading:

For an in-depth review of the process of genetic counselling refer to current literature, for example the following texts:

- Psychosocial Genetic Counseling, (Weil, 2000)
- A Guide to Genetic Counseling, (Baker, 1998)
- Practical Genetic Counselling, (Harper, 2004)
- Genetic Counselling: A Psychological Approach (Evans, 2006)

4.0 RECORD KEEPING

4.1 Client record

Clinical genetics units should have client files that are separate from records of the health unit to which they are attached. This is recommended because of the:

- confidential nature of genetic information;
- need to maintain the record for future generations;
- need to have a complete and comprehensive file for each family;
- need to have access at central clinical genetics units to the records of clients seen in outreach areas.

Local health unit requirements for record keeping should be followed, with the proviso that the confidentiality guidelines of the clinical genetics unit are not breached. With the client's consent, copies of relevant correspondence and reports should be sent to their current healthcare provider(s).

4.2 Face-to-face consultations (including telemedicine consultations)

The information to be documented and retained includes:

- genetics file number, relevant hospital UR numbers as appropriate;
- identifying information - name, date of birth, address, phone number.
- date and place of consultation;
- names of health professionals and other individuals present at consultation;
- name of referring health professional and other health professionals involved in their care;
- reason for referral;
- client history, test results and reports from other health professionals / hospitals;
- pedigree – minimum three generations;
- diagnosis and documentation to confirm as appropriate;
- relevant information about members of the extended family;
- summary of information given to client and the counselling issues addressed;
- agreed process for providing client with results of investigations as appropriate;
- copies of consent forms for accessing medical records, DNA testing or sample storage;
- copies of correspondence to client and health professionals;
- results of investigations, including documentation that the result has been sighted by the appropriate health professional;
- copy of completed birth defects register form if applicable;
- arrangements for follow up.

4.3 Phone consultations

Records must also be kept of all telephone consultations and letters about identified clients. For clients who have a genetics file, any further contact should be well documented. In situations where the individual does not have an existing file, genetics units must decide on the documentation and storage policy for such contact records.

4.4 Client databases and electronic records

Clinical genetics units should have a client database, which, at a minimum, contains the following information about the client and the consultation:

- clinical genetics unit file number;
- other health unit UR numbers;
- identifying information eg: name, date of birth, sex;
- contact details eg. address, telephone numbers;
- consultation details eg: date, place, and name of health professionals;
- diagnosis.

The database should also have the properties to enable:

- searching for specific information;
- editing of records;
- linking branches of the same family;
- audit ;
- access by outreach clinics;
- security in terms of password access, systems for back-up and virus protection (with regular upgrade).

4.5 Storage of hardcopy records

Genetic client files should be stored in a separate place to other hospital / health records. Records must be:

- securely stored;
- accessible to authorised staff only;
- accessible for client management at all times, with a mechanism in place for tracing records.

4.6 Confidentiality of records

- client records are confidential and subject to relevant professional standards or legislative requirements related to privacy and release of information;
 - in general, information about a person must not be released to a third party without the written or documented verbal consent of the person / guardian;
 - if information about a deceased individual needs to be obtained, consent may be sought from the next of kin or the executor of the individual's will;
 - if information about an individual who is intellectually disabled needs to be obtained, consent may be sought from the recognised legal guardian. If the individual is under the care of a guardianship board, a written request must be submitted;
 - if information about a client is to be conveyed to other health professionals involved in their care, consent must be obtained from the client;
 - if client records are subpoenaed, the Director of the genetics unit must also comply with requirements of their relevant State health service;
 - it is the responsibility of the Director of the clinical genetics unit or Director of the associated health unit to ensure that all staff have an adequate understanding of the relevant, current federal and local government privacy laws / codes.
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5.0 COMMUNITY OUTREACH

To facilitate equity of access, genetic counselling services have been established in outreach areas. Community outreach (rural or metropolitan) refers to genetic services, which are usually staffed primarily by genetic counsellors, with administrative support in some instances, and usually involve provision of regular clinics / consultations by a visiting clinical geneticist (See HGSA policy: [Structure of Clinical Genetics Units in Australia](#) 1999 GL01). Each community outreach service is attached to and supported by a major genetics unit.

The following clinical support should be provided to outreach genetic counsellors:

- orientation at the major genetics unit;
- regular visits to the major genetics unit to facilitate supervision, liaison, support, networking and observation of speciality genetics clinics (ideally such visits should be at least bi-annually);
- all outreach genetic counsellors should have access to informatics, including e-mail and internet, as outlined in HGSA [Structure of Clinical Genetics Units in Australia](#) (1999 GL01);
- supervision by a clinical geneticist with at least weekly contact by telephone to discuss referrals, complex clinical issues, and service needs;
- access to a counselling supervisor and support to maintain professional counselling supervision;

- opportunity to update and maintain genetic knowledge, including support to attend relevant in-service and conferences;
 - engagement of the major genetics unit with local administration in discussions about the service, required resources and provision of educational presentations to local groups;
 - provision of a mentor genetic counsellor / associate genetic counsellor from the major genetics unit for support and to facilitate acquisition of journal articles, client file information and resources;
 - at each clinic visit, time should be allocated for genetic supervision with the clinical geneticist to meet the requirements of the Board of Censors for Genetic Counselling.
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6.0 IMPROVING CLINICAL PRACTICE

The continual improvement of clinical practice is paramount. Each genetics service must be involved in quality assurance work practices. All aspects of the process of genetic counselling must be reviewed, including:

- waiting times for appointments: urgent and routine;
- phone consultations and enquiries: documentation and outcomes;
- face-to-face consultation, including pre and post-clinic discussions;
- follow-up phone calls, letters to clients and health professionals as well as referrals to outside agencies;
- client satisfaction questionnaires;
- service liaison with genetic laboratories.

Other important processes include:

- maintenance of professional standards;
 - regular staff appraisal;
 - maintenance and review of database(s);
 - review of protocols and guidelines;
 - review of association with outside agencies;
 - review of adverse incidents or events.
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References

1. Adapted from: NSGC (1995-2006) "Consumer Information: Definitions: Genetic Counseling as a Profession" National Society of Genetic Counselors <http://www.nsgc.org/consumer/definition.cfm> accessed: 28th June 2007
2. Biesecker, B. and K. Peters (2001). "Process studies in genetic counseling: peering into the black box" *American Journal of Medical Genetics* **106**(3): 191-198.