

1. Talking with families about genetics

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1.1 Introduction

Individuals with genetic conditions look to their GPs to perform a number of different roles. These include case-management of both the individual and their family, advocacy for the family and support for family decisions (Hurworth, Robins & Metcalfe 2000; Metcalfe et al. 2002). To effectively perform this work, families highlight the need for GPs to have good communication skills (Hurworth, Robins & Metcalfe 2000).

GPs have identified the need to develop communication skills relating to genetics (Hurworth, Robins & Metcalfe 2000; Metcalfe et al. 2002). Counselling about genetics depends on good communication, but includes certain issues specific to genetics. This section aims to raise awareness of these issues and suggest possible approaches to talking with families about genetics.

Advice about specific cases is available from genetic services.

1.2 Collecting a family history

- Drawing a family tree is a rapid method of documenting family history information, genetic relationships and medical information. For a guide to drawing family trees, see Appendix 1.
- A three-generation family history should be collected, where possible, including children, siblings, parents, grandparents and grandchildren.
- Information collected should include the age at diagnosis of any medical conditions in the family.
- The medical information collected will depend to some extent on the condition or concerns of the individual. Information might include:
 - ✦ Specific inherited conditions present in the family.
 - ✦ Miscarriages and still births.
 - ✦ Blood relationship between partners (consanguinity).
 - ✦ Cancer diagnoses (age at diagnosis, site of primary cancer).
 - ✦ Intellectual and physical disability.
 - ✦ Neurological conditions.
 - ✦ Common adult onset conditions, such as diabetes, dementia, osteoporosis, asthma, cardiovascular conditions, glaucoma.
- An individual's concept of family and relationship to other people may be influenced by their cultural background.
- Ways of asking questions regarding the family and their health include
 - 'How many pregnancies did (name of relative) have?'
 - 'How many [for example] brothers and sisters do you have altogether? Have any died? How is their health?'
- **Family histories should be dated and updated regularly.**

Under the Commonwealth's Privacy Act 1988, a Temporary Public Interest Determination (No. 2001-1) allows health information to be collected from an individual about another individual (see http://www.privacy.gov.au/act/public_interest) in certain circumstances that include family history taking. Under the Victorian Health Records Act 2001, a proposed regulation allows collection of family history information by health service providers, if reasonably necessary for service provision.

1.3 Talking about genetics

General strategies

Everyone has preconceived ideas about genetics and the inheritance of conditions and traits in their family (Chapple, May & Campion 1995). Sometimes this includes misconceptions that may affect their understanding of genetic explanations. Furthermore, genetic information is complex and people can easily become confused. The following strategies can assist an understanding of genetics.

Common misconceptions about genetics

- An individual's physical similarity to other relatives will indicate their risk of developing a condition; for example, 'My father has the condition, but I look like my mother, so I'm OK'.
- Conditions affecting women (for example, breast cancer) can only be inherited through the maternal line.
- A condition only affects one gender in a family; for example, when only men in a family have Huntington disease, the women are not at risk.
- Tests are available for all inherited conditions.
- The presence of an altered gene means that an individual will definitely develop the condition, even though the risk is not 100% (incomplete or reduced penetrance). For example, 'I have the breast cancer gene, therefore I am going to get cancer'.
- A 'one in four' risk means that after one child is affected, the next three will not be.
- If a condition is dominantly inherited, then the altered copy is 'stronger' and will be passed on more often, so more people will be affected than unaffected.

Assess understanding

Ask about the individual's understanding of their situation before providing information. Use their understanding as a starting point for the discussion; that is, start with what the family know or believe, then introduce new concepts or correct misunderstandings.

As you explain genetic concepts, check an individual's understanding of what they have been told before continuing.

Ways to assess understanding include:

- Ask what they believe caused the condition (in the family).
- Ask what they imagine a gene (or chromosome) is.
- Ask how they might explain the information to another individual or family member.

Use strategies to aid recall and understanding

- Actively encourage questions.
- Write down key medical terms.
- Make use of simple diagrams and pictures where appropriate
- Repeat and summarise important information
- Recall after receiving complex information can be improved by:
 - ✦ Writing down relevant information.
 - ✦ Sending the individual a summary letter as a follow-up.
 - ✦ Providing brochures and fact sheets.
 - ✦ The presence of a support person at the appointment.

Deliver genetic information in plain English

- Give clear, specific information. Try to provide information about one issue completely before talking about the next issue. For example, when talking about testing during pregnancy, completely address one type of test (for example, screening) before talking about the next.
- Explain medical terms or genetic terms and avoid jargon. Many people are familiar with words such as genes and chromosomes but may not have a complete understanding of what they mean.
- Feedback from genetic support groups shows that language commonly used to describe genetics can sound emotional and judgemental to people who are not familiar with genetic concepts. Avoid using terms that convey unintentional negative messages.

Some terms with negative connotations

Bad, faulty or abnormal genes, mutation
 Mental retardation
 Genetic disease
 Sufferer, victim, afflicted with ...
 Wheelchair-bound or confined
 Normal
 An achondroplast
 A Downs, a Downsie

Alternatives

Altered gene or gene change¹
 Intellectual disability
 Genetic condition
 Affected by
 Uses a wheelchair
 Unaffected by
 Person with achondroplasia
 Person with Down syndrome

Specifics

Common genetic terms

Families are often confused by genetic terms. A simple explanation for some common terms is given below. See Glossary for more extensive definitions for medical professionals.

Gene

A piece of information that influences the growth, development and/or functioning of the body. A gene can be likened to an instruction or sentence. Together, all the genes form an individual's genetic blueprint. Usually, an individual has two copies of each gene, one inherited from each parent.

Mutation (gene alteration)

An alteration in a gene that causes it to function differently or cease to function. It can be likened to a spelling mistake in an instruction.

Genetic condition

A medical condition that is caused by inherited changes in genes or the presence of changes in chromosomes. The chance of other family members or pregnancies having the same condition will depend on the type of inheritance.

Genetic predisposition/susceptibility

Genetic predispositions are caused by alterations in genes that contribute to the chance of developing an adult-onset condition (for example, cancer). Other factors may also contribute to risk. An individual who has a genetic predisposition may never develop the condition (that is, reduced penetrance).

¹ Note: we have used the terms 'mutation' and 'alteration' and 'altered gene' interchangeably throughout this document.

Chromosome

A threadlike structure made of genetic material (DNA) that is present in almost every cell of the body and is visible under a microscope. A chromosome has different genes along its length, somewhat like a list of instructions. People usually have two copies of each chromosome, one from each parent. Usually, people have 46 chromosomes, 22 pairs of 'ordinary' chromosomes (autosomes) and one pair of sex chromosomes.

Chromosome translocation

A rearrangement of the genetic material, where part or all of one chromosome is attached to another chromosome. The rearrangement is called by the names of the chromosomes involved. If all the genetic material appears to be present, the rearrangement is called 'balanced'. If part of the genetic material has been lost or gained in the rearrangement, it is called 'unbalanced'. This is usually detected microscopically.

Carriers

A carrier is an unaffected individual who has either:

- A chromosome rearrangement.
- or
- One copy of an altered gene for a recessive or X-linked condition and one unaltered (normal) copy.

They will not develop the genetic condition.

This term is also sometimes used for individuals who are known to have a genetic alteration causing dominantly inherited adult-onset conditions (for example, cancer) but are not (yet) affected.

Dominant inheritance

A genetic condition with dominant inheritance occurs when only one copy of a gene is altered. The other copy is normal. The altered copy is 'dominant' over the normal copy. However, when an individual with a dominantly inherited condition has a child, the child has an equal chance of inheriting the condition and not inheriting it (that is, 50% or one chance in two – like the toss of a coin).

Recessive inheritance

A genetic condition with recessive inheritance occurs when both copies of the genes are altered. For a child to have a recessive condition, both parents must have at least one altered copy.

If two carriers are having a child, in each pregnancy they each have a 50% chance (one in two chance) of passing on the altered copy. This means for each pregnancy, there is a 25% chance (one in four chance) of the child being affected, a 50% chance (one in two chance) of the child being a carrier, and 25% chance (one in four chance) that the child will have two normal copies of the gene.

If an unaffected individual has a sibling with a recessive condition, then they have two chances in three of being a carrier.

² In some conditions, skewed X inactivation (see Glossary) may result in varying degrees of symptoms in females. Rarely, a female may be affected if she has an affected father and a carrier mother.

X-linked inheritance

A genetic condition with X-linked inheritance is caused by an alteration in a gene on the X chromosome and usually only affects males.² This is because males only have one X chromosome and therefore only one copy of the gene. If that copy is altered, there is no normal gene and so the male is affected. Females usually have two X chromosomes. If one copy of the gene is altered, they also have a normal copy and therefore are unaffected carriers.

Risk

Discussing genetics can involve explaining many different types of risk, including the risk of:

- Receiving an 'increased risk' result from a screening test.
- Having a baby with a chromosome anomaly (based on maternal age or screening test results).
- Being a carrier of a genetic condition.
- Having a child affected by a genetic condition.
- Having a genetic predisposition to an adult onset condition.
- Developing a condition when the person has a genetic predisposition.

The term 'risk' can imply a negative outcome and sometimes 'chance' may be a more appropriate term.

Many people find concepts of chance or risk difficult to understand and find numbers or percentages confusing. Some people do not realise that risk gives an idea of how likely or unlikely a situation is, not that the situation will definitely happen. Describing risk in terms of individuals can assist this (see box below).

Risk figures can be misunderstood and can be seen as being 'used up'. For example, a one in four risk of an affected pregnancy is often (wrongly) interpreted as meaning once a child is affected, the next three will not be.

Conversely, an individual who has experienced an uncommon event, such as the diagnosis of pregnancy with a fetal abnormality, can feel heightened vulnerability and find it difficult to believe that event is unlikely to happen again.

The significance of a risk varies between individuals. The same risk figure may sound unacceptable (high risk) to one individual, but acceptable (low risk) to another. For this reason, it is important to clarify the individual's reactions to a risk assessment and explore any apparent contradictions with open-ended questions or reflection (for example, 'You seem to feel these are pretty good odds' or 'It seems like too much of a risk to you').

Ways of explaining prenatal screening risk figures

Using a number of ways of describing risk can be helpful.

For example, a 29-year-old woman receives an increased risk result for Down syndrome after a serum screen test. The risk is one in 100. Prior to the test her risk was based on maternal age alone and was one in 1010.

1. Comparison to other women getting same test result.

Of 100 women with this test result, on average, one will have a baby with Down syndrome and 99 will have babies who do not have Down syndrome.

2. Risk relative to age-related risk.

Your risk has increased 10-fold.

3. Risk relative to maternal age.

Your risk is now similar to that of a 39 to 40 year old who has not had any screening. Women who have this level of risk are offered further testing to determine the karyotype of the fetus.

1.4 Counselling issues

Individuals often have emotional reactions to their genetic situation that cannot be resolved by simply providing information and facts. Addressing these reactions can require the use of counselling skills such as active listening and a non-judgemental approach (see Appendix 3 for more detail on counselling and interview skills).

As with all counselling, avoid stereotyping or making assumptions about the people's level of understanding or emotional ability to deal with the situation.

Counselling in consultations about genetics may address breaking bad news, anxiety, uncertainty, grief, guilt and blame. Counselling can assist individuals adjust to their situation; however, in some instances people may have unresolved issues. Referral for specialist counselling³ may be considered.

This section refers to a person or individual, however in situations where two parents are involved, both should be considered and may have very different reactions and needs.

Breaking bad news about genetics

See also Appendix 4 and *Patient and further information* (further reading).

Bad news in genetics can include:

- A higher than anticipated risk shown by screening or clinical assessment.
- Diagnosis of a genetic condition, by testing or clinical assessment.
- Lack of a definitive diagnosis.
- Uncertainty regarding prognosis.
- Uninterpretable test results (for example, no mutation found in cancer genetic testing).
- News that is different from anticipated.
- Identification of carrier status.

It is never easy to break bad news; however, there are sensitive ways to break bad news that facilitate coming to terms with the news over time. A caring and empathic manner is important. The news should be given in person simply and honestly, allowing uninterrupted time. Silence enables the person to absorb the news and respond when they are ready.

Emotional reactions are normal and can take the form of distress, blankness or denial. Sometimes people may appear to listen and understand; however, they actually do not take in the information. In some situations, people respond at an intellectual level and block the underlying emotions. These should be explored.

Individuals need the opportunity to ask questions at the appointment and afterward. A plan should be put in place for the next contact and provision of ongoing support in the interim, if necessary.

The importance of self-awareness in breaking bad news cannot be underestimated. Breaking bad news is difficult for many reasons, most relating to the newsgiver's past experiences, emotional state, uncertainty, acceptance of own mortality and identification with the individual. These impact on the newsgiver's ability to best assist the person.

³ For example, grief counselling, family therapy, relationship counselling, psychotherapy.

Anxiety and living with uncertainty

There are many potential areas of uncertainty in genetics (see box below). These diminish a sense of control over events and life. Anxiety and fear are often the result, or may be exacerbated.

Potential areas of genetic uncertainty

An increased chance (risk) of occurrence of a genetic condition.

An increased risk of a pregnancy affected by a chromosome abnormality.

A risk that future pregnancies will be affected by a genetic condition.

Clinical identification of a child with unusual physical features but no definite diagnosis.

The diagnosis in a child of a condition where the level of ability and future health may be unknown.

An increased risk of developing a condition at some time in the future.

In an attempt to obtain certainty, people may be motivated to search the Internet, visit many doctors, travel overseas for medical advice, and question the expertise of the original doctor.

Family members may expect a genetic consultation or diagnosis to resolve uncertainty and decrease anxiety. This expectation is not always realistic and may lead to new uncertainties.

Counselling for anxiety and uncertainty may be facilitated by:

- Discussing the individual's expectations prior to tests being conducted.
- Enabling the individuals to verbalise their concerns and fears may assist them to identify the underlying source of their anxiety.
- Discussing past experiences that involved uncertainty and their resolution.
- Being self-aware. Anxious individuals can unconsciously 'transfer' their feelings of anxiety and helplessness to the practitioner (transference). Practitioners need to be aware that their own feelings may reflect the emotional state of their patient.
- Avoiding false reassurance or expectations.
- Avoiding jargon.
- Discussing the individual's expectations of a referral (for example, to a geneticist).

Decision making

In many health settings, people expect advice and direction. In genetics, best practice is often unclear and the person may be faced with a range of options. Ultimately, the decision must be made by the person themselves.

Choices that arise in genetics:

- Choosing between medical options (that is, to have a test and which test).
- Deciding whether to terminate a pregnancy.
- Deciding on a prenatal diagnosis or PGD or no testing.

The 'best' decision often rests on personal factors such as values, coping style and circumstances. Individuals may perceive that family, friends, society or the medical profession are applying pressure to make a particular decision.

Individuals may ask 'What would you do?' This question may arise from uncertainty or from a desire to check the acceptability of a decision. It is not appropriate to offer a personal opinion as this can imply that other choices are not valid or acceptable.

An appropriate response to the question 'What would you do?' might be to acknowledge that the decision is a difficult one and to offer support in reaching a decision. Highlight that there is no right or wrong decision, and it is important that the person consider what is best for them.

Decision making may be facilitated by:

- Using counselling skills that allow value free and non-judgemental discussion of the issues and factors impacting on the decision.
- Providing correct, up-to-date and unbiased information from a variety of reliable sources of information.
- Allowing time and the opportunity for consultation with different sources of information.
- Asking the individual to consider different scenarios and the impact these may have on their life.
- Asking the individual to consider past decisions, including how they were made and what helped.

Grief and loss

People given genetic information may grieve the loss or change of an expected lifestyle for themselves or another family member, grieve the loss of a pregnancy, grieve the anticipated loss of a child, or grieve the loss of expectations for their child or themselves.

Grief is not an illness but a normal response to loss, and a healthy process of adjustment over time. An understanding of the physical, emotional and social reactions of grieving people is essential. All people do not grieve in the same way. A person may benefit from some outline of the grief process to realise that their reactions (current and future) are normal. Referral for grief counselling may be appropriate.

Guilt, shame and blame

Emotions commonly experienced after the diagnosis of a genetic condition or predisposition include guilt, shame and/or blame. These reactions are not confined only to parents or affected individuals, but can be experienced by other family members.

Guilt is a very common reaction and can take the form of questions such as 'What did I do wrong?' 'Is this a punishment?' These questions may not always be verbalised. Instinctive rejection of a child or pregnancy affected by a genetic condition can also be a source of guilt. Guilt is a reflection of a feeling of responsibility for the condition and can be experienced by grandparents as well as parents.

Shame is an expression of the (self-) perception that an individual or couple has failed to live up to their own or society's expectations (for example, by not having a 'normal' baby).

Blame can be a defence against a potential threat to self-image, a way of stating 'it can't be me'. Family members may 'blame' the 'other side of the family' for a problem directly or by highlighting the absence of any 'defects' in their family history. Blame directed at specific individuals or between a couple may be a symptom, or the start, of underlying problems in the relationship.

An intellectual understanding of genetics is not always sufficient to counteract these emotions.

Guilt can be alleviated if, at the time of diagnosis *or once feelings of guilt have been expressed*, the fact that the individual (couple) is not responsible is stated or reinforced. Permitting the individual/couple to talk about their feelings and using skills such as normalisation ('Many couples feel guilty when ...', see Appendix 3) may be helpful.

It is common for parents to look for a reason why their child has a condition. Taking a history can unintentionally convey that the parents contributed in some way. When taking a history, offer reassurance where possible that a factor has not contributed to the condition. For example, social drinking during pregnancy has not caused a chromosomal disorder.

Shame may be expressed as the desire to hide the source of shame, from the eyes of a judging world (Kessler 2000). Ridicule and rejection are anticipated, so it is important that they feel the practitioner is not judgemental. Helpful skills include enabling the individual to talk about their feelings, accentuating aspects in which the individual is doing well and bolstering self-esteem. Demonstrate that an affected child or baby is not rejected by referring to them by name, and by being inclusive of the affected child (for example, holding the baby, or talking to the child).

Chronic illness

The issues relating to chronic illness for genetic conditions are similar to those for other chronic conditions, but may be complicated by the other aspects of genetics such as guilt.

More information is available from the Chronic Illness Alliance (*see Patient and further information*).

Cultural issues

Cultural background and ethnicity can influence people's response to genetic conditions and risk, however it is important to individualise and avoid stereotyping. Options discussed should not be limited or choices anticipated on the basis of ethnicity or religion.

Genetic conditions are considered stigmatising in some cultures and familiarisation with some common cultural beliefs can assist in counselling. Ethnic agencies can help with this, and useful information regarding cultural beliefs, multilingual education material and services is available online (see *Patient and further information*).

Whenever possible, medically trained interpreters should be used to avoid burdening family members with translating medical information and the risk of misunderstanding or selective disclosure. Generally, people that have been in an English speaking country for less than two years will require an interpreter; however, language skills can be affected by distress and emotional situations. A telephone interpreting service may be helpful.

1.5 Implications for other family members

Family members inherit their genes from common ancestors and therefore have genes and genetic changes in common. Consequently, information provided to one individual about a genetic condition may also be relevant to other family members.

Conversely, assessment of an individual's risk may require information from other family members.

Informing other family members

It is good practice to discuss with the individual the implications of a genetic diagnosis for other family members, including which family members should be informed and how they might be approached. In fact, individuals may often be concerned for other family members and want this information. Genetic services can provide guidance about which family members should be informed. If the individual has been seen by a genetics service, this is likely to have been discussed with them.

Informing family members can be difficult and it is preferable that other family members contact genetic services or their own GP for a thorough explanation of their situation. Where possible, providing a letter or simple written information with contact details can be useful.

Helpful techniques include asking how the individual will tell other family members and preparing them for possible reactions to the news (for example, denial, fear and anger).

If other family members are also patients of your practice, consent could be obtained to discuss this issue with them at their next appointment.

Asking for information from other family members

Information may be required from other family members to assess an individual's risk. People may feel uncomfortable asking for this information.

- Discuss how they may approach the person (in person, phone call, letter).
- Explain the reasons for needing the information and give the individual some idea of how essential (or otherwise) the information is. They can then weigh the benefits of gaining this information against the difficulties of making contact.
- Consider providing a letter supporting the request to be given to family members.
- Ask genetic services to seek written consent from other family members to access the relevant medical information.

1.6 Support for families

Families and individuals can need different levels and types of sources of support at various times.

In addition to support from the GP, referral to genetic services or to professional counselling and support organisations can be beneficial.

Support groups can be an important source of peer support and empowerment, practical information and advice about living with a genetic condition. Families can benefit from contact with other people in similar situations, regardless of their level of coping or need for support. The Genetic Support Network of Victoria has contact details of support groups for genetic conditions.

Some individuals and carers will be eligible for financial support through Centrelink benefits. Families may not be aware of this and should contact Centrelink for information.

1.7 Insurance issues

The presence of a genetic condition in the family may affect an individual's ability to obtain travel, life, income and/or disability insurance.⁴ Pre-existing life, income and disability insurance cannot be cancelled if there is deterioration or change in the policy holder's health (Investment and Financial Services Association 2002).

At this time, insurance companies have agreed not to require people to undertake genetic testing when requesting insurance cover; however, existing test results must be made available to insurance companies upon request.

1.8 Ethical issues

The ethical principles that guide all medical care apply in genetics. However, ethical dilemmas arise when there is tension or conflict between the rights of different family members.

Key ethical principles include:

- Respect for autonomy (the right of an individual to self-determination, including privacy and confidentiality).
- Non-maleficence (do no harm).
- Beneficence (taking positive action to do good).

There can be tensions when these principles are considered with respect to the right of an individual to:

- Know, or not to know, information relevant to their own health (autonomy).
- Disclose, or not, personal information (privacy).
- Make an informed decision regarding genetic testing.

Genetic counselling emphasises that an autonomous choice be made; that is, a choice that is informed, reflective of the individual's own values and made freely (without coercion). However ethical dilemmas arise, for example, when:

- During testing, an individual will disclose the genetic status of another family member who has not had testing (and may not wish to).
- An individual refuses to disclose to other family members that they are at risk.
- Parents request that their child (under 18 years) be tested for an adult onset condition, thus affecting the child's future autonomy.

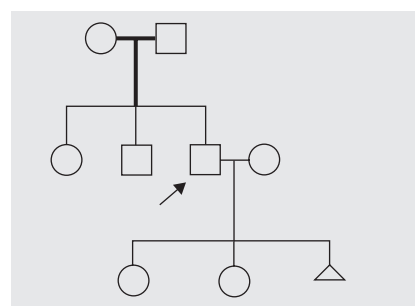
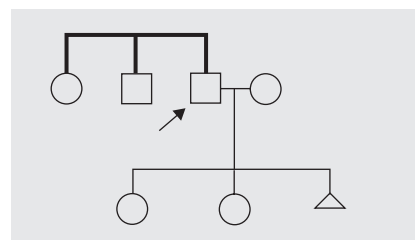
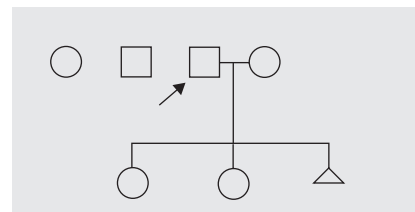
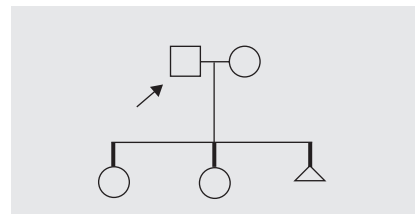
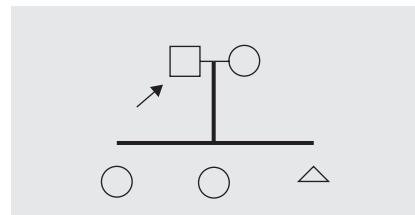
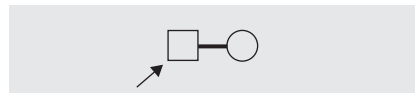
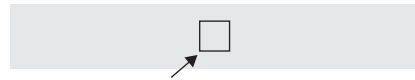
In these situations, it is important to explore the potential harms and benefits with the individual and their reasons for their request. Referral to genetic services for counselling is strongly recommended.

Appendix 1: Constructing a family tree

Reference for symbols: Bennett RL et al. 1995, 'Recommendations for standardized human pedigree nomenclature', J Genet Counsel, 4:267.

At each step ask about the health of the family members being discussed


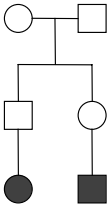
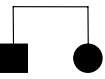
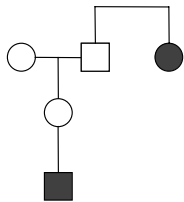


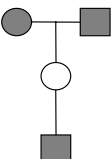
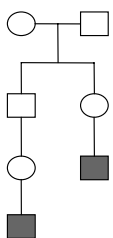
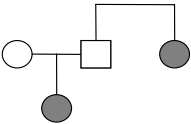
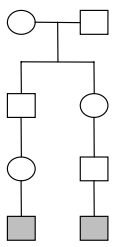
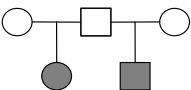
1. Draw symbol for the family member you are seeing. Indicate the person with an arrow.
2. If the individual is married, draw a line across directly to a symbol for the spouse.
3. Ask about the number of pregnancies. Draw a reverse T from the marriage line and add the symbol for each child and pregnancy.
4. Add a line from each child to the reverse T.
5. Ask about brothers and sisters (siblings). Add the symbols.
6. Indicate the relationship between siblings by drawing a line above the symbols and joining to the symbols by vertical lines.
7. Add a vertical line and parents.
8. Deceased family members are indicated by a line through the symbol.
9. Repeat steps 4-6 for each parent to include the aunts, uncles and grandparents of the family member you are seeing.



Appendix 2: Standard Symbols used in drawing a family tree

	Male	Female	Sex unknown	Pregnancy	
Individual					
Affected individual					Key: eg. cystic fibrosis
Two or more conditions					colour blindness
Multiple individuals					Number inside symbol
Deceased individual	 d. 35yr	 d. 4mo	 d. 1936		
Stillbirth	 SB 28wk	 SB 30wk	 SB 34wk		
Consultand (Individual seeking genetic counselling)					
Proband (First affected member coming to medical attention)					
Carrier (Will not develop condition)					
Spontaneous abortion					Record any information e.g. age of onset of condition, age of death.
Affected spontaneous abortion	 8 wk				
Termination of pregnancy					
Affected termination of pregnancy					
Partnership		 Previous			
Twins	 Monozygotic (identical)	 Monozygotic (identical)	 Unknown		
	 Dizygotic (not identical)				

Appendix 3: Degrees of relationship

First degree		Third degree	
	% Genes shared		% Genes shared
	Parent-child 50		First cousins 12.5
	Siblings 50		Great-aunt - nephew 12.5
	Dizygotic (non-identical) 50		
	Monozygotic (identical) twins 100		
Second degree		Fourth degree	
	Grandparent-grandchild 25		First cousins once removed 6.25
	Aunt (uncle) - niece 25		Second cousins 3.125
	Half-siblings 25		

Appendix 4: Counselling skills and interview techniques

Effective communication is facilitated by a congenial physical environment. Consider the positioning of the chairs, direction of sunlight, location of the desk, and the absence of interruptions.

In this section, techniques are suggested that may enhance communication. Although phrases are given as examples of these techniques, individual style develops over time.

Be client-focused

- Enable the individual to set the agenda for the interview and work at their pace. Ensure the individual also knows your agenda.
- Listen actively by concentrating and observing verbal and non-verbal language. Appropriate posture, eye contact and attention are indications that the individual has the practitioner's attention.
- Acknowledge what the person has said.
- **Show respect by:**
 - ✦ Being honest. It is not helpful to avoid giving potentially upsetting information.
 - ✦ Providing information about all of the choices available, and encouraging consideration of opposing points of view; for example, in prenatal testing when a fetal abnormality is detected.
 - ✦ Respecting the right of the individual to make their own decision, based on the information, even though that decision may be contrary to the beliefs of the practitioner.
 - ✦ Offering to seek out further information if required.

Establish rapport and trust

Effective communication requires a mutually trusting and respectful relationship with the family member. This needs to be built and can take time.

Use open-ended questions.

Encourage further discussion by inviting the person to continue a theme, rather than questions that can be answered 'yes' or 'no'. Use questions beginning with 'what' or 'how'; for example, 'What was that like?'

Individualise.

Avoid stereotyping on religious, racial, educational or socioeconomic grounds. Equally, avoid predicting or expecting reactions or decisions.

Express empathy by:

- Reflecting back emotions you observe; for example, 'You seem sad'.
- Using and tolerating silence.
- Checking the emotions of yourself and the individual, then reflecting these using 'I' sentences; for example, 'I sense that you are still uncertain ...'
- Summarising; for example, 'From what you have said, it seems there is a lot happening in your life ...'

Facilitate mutual understanding

Make direct statements.

Use statements that are clear and unambiguous, while conveying appropriate empathy and concern.

By giving clear honest information, the counsellor is showing respect: 'I know that you have been looking for a diagnosis for John for a year now. We have the results of the tests, and we can make a diagnosis. The diagnosis is serious, and I realise that this is not what you had been hoping for ...' The use of appropriate tone of voice, posture and facial expression of the practitioner is important to reflect concern.

Avoid 'You should' statements.

Clarify and re-iterate

Mutual understanding should be constantly checked verbally and non-verbally. This may involve the practitioner asking for clarification; for example, 'I don't understand what you mean by ...'

Difficult concepts may need to be rephrased, perhaps using diagrams, or words that have more meaning or are more familiar.

When summarising the content of the interview, major points of discussion can be repeated; for example, 'We discussed four areas today about the condition in your family called ...'

The counsellor can provide a framework for the confused person to re-think a difficult issue. 'I don't know what I would do in your place, but these are some things to consider ...'

Assist adjustment

Normalise and generalise

Use phrases such as, 'In my experience, people often feel ...' This reinforces the authority and knowledge of the counsellor, while acknowledging to the person that their reaction is common and not 'odd'.

Re-frame

Interpret a negative thought or reaction of the individual, to give a more positive understanding; for example, 'You talk about your guilt at passing the gene to your daughter. However you were not doing this deliberately, as you were not aware of the gene, we all have genes that don't work properly, and your concern shows your responsibility and love for your daughter' (Kessler 2000).

Offer ongoing support

Assess the level of family or social support available to the individual. Discuss sources of support and provide the names and contact details of relevant people or organisations to obtain more information.

Offer or suggest another appointment. Indicate your availability to answer questions that may arise after the consultation.

Appendix 5: Breaking bad news

Breaking bad news is difficult due to:

- The practitioner's own emotional state.
- The degree of identification with the individual.
- Acceptance of the practitioner's own mortality.
- The burden of truth telling.
- Continuing commitment to the individual.
- Uncertainty.

When to tell the individual

- Prepare the individual for the possibility of bad news as early as possible in the diagnostic/testing process.
- Plan a consultation for the time when all of the results will be available.
- Tell the individual as soon as the final result or diagnosis is available.

Prior to giving news

- Ensure the news is given in person.
- Allow enough uninterrupted time in a comfortable place.
- Encourage a second person or family member to be present, if appropriate.
- Ensure you have the necessary information and appropriate referrals.

When giving the bad news

- Assess the individual's understanding of their situation.
- Define the nature of the session. 'You've come today to get the results ...'
- Warn of the news.
 - Warn: 'Unfortunately I have bad news for you.'
 - 'You may not hear or remember all that I will tell you.'
 - 'I will repeat it all later.'
 - 'You can ask questions.'
- Provide the news simply and honestly. Use lay terms and avoid euphemisms.
- Work at the pace the family can cope with; however, avoid holding over further bad news to other appointments.
- Be aware of the types of reactions individuals experience when receiving bad news.
- After giving the news, allow silence to allow the individual to absorb and respond to the information.
- Work from what the individual knows and understands. At each stage, check understanding.
- Be aware of unhelpful self-protection strategies when giving bad news such as suppression of feeling, authoritarian style, therapeutic zeal.
- Be aware of your own body language, actively listen and avoid closed questions.

Afterward

- Express empathy and the ability to help; however, avoid expressions such as 'I know how you feel'.
- Prepare the individual for what they may experience after the appointment; for example, a feeling of disbelief, physical reactions, a sense of unreality, an inability to concentrate, sleeplessness, moodiness, increased thoughts about the situation and anxiety.
- Provide a structure for the next step. This does not mean making a decision but providing support, another appointment or referral or other plan for next contact.
- Provide appropriate written information because recall of information presented in the consultation is often poor.

Appendix 6: What is genetic counselling?

Genetic counselling is a communication process that aims to provide information and supportive counselling to members of families regarding problems in growth, development and health that may have a genetic basis. This process usually involves an appropriately trained individual, working as part of a team to help the individual or family.

The process of genetic counselling involves:

- Comprehending the medical facts – including the diagnosis, probable course of the disorder and the available management.
- Appreciating the way heredity contributes to the disorder and the risk of recurrence in specified relatives.
- Understanding the options for dealing with the risk of recurrence.
- Choosing the course of action that seems appropriate in view of their risk and their family goals, and acting in accordance with that decision.
- Making the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.

Genetic counselling is a relatively new profession that is practised by qualified genetic counsellors, but many health professionals may perform aspects of genetic counselling.

Genetic counsellors are health professionals with a tertiary qualification in fields such as education, nursing, psychology, science or social work. They have additional training in human genetics and counselling and have been certified by the Human Genetics Society of Australasia after a minimum of two years experience and demonstration of competence. Associate genetic counsellors are in the process of becoming certified.

Medical geneticists are medical practitioners with a specialist qualification and training in clinical genetics.

Elements of a consultation about genetics

The elements of a genetic counselling session will depend on the nature of the condition or issue. They may include:

- Collecting a family history.
- Performing a clinical examination.
- Providing genetic information.
- Explaining genes and genetics.
- Discussing genetic testing.
- Discussing the implications for other family members.
- Providing support and counselling.

Preparing families for a genetic consultation

The usefulness of a consultation in a genetics service for a family can be maximised if the family are aware that:

- They should gather information about their family history (including causes of death and ages) and take it to the consultation. If there is the indication for referral for diagnosis, taking photos of relatives to the consultation can be useful.
- They should make a list of questions they wish to ask before the appointment and take it with them.
- Consultations can be an hour long. If tests are required, the family may be at the service for a total of two hours.
- The consultation may not provide definitive information.
- Blood tests may be required.
- The presence of another family member or friend can provide moral support and help recall.
- A letter is sent to the family after a consultation, as well as to the referring doctor.

1.9 Patient and further information

The Genetic Health Services Victoria booklet Frequently asked questions is attached.

Child Health Information Centre
The Royal Children's Hospital
Flemington Road, Parkville 3052
Call: (03) 9345 6429
Fax: (03) 9345 6120
Email: chic@rch.unimelb.rch.org.au
Website: <http://www.rch.org.au/chic/>

Support Groups (General)

Genetic Support Network Victoria
10th Floor, The Royal Children's Hospital
Flemington Road, Parkville 3052
Call: (03) 8341 6315
Fax: (03) 8341 6390
Email: info@gsnv.org.au
Website: <http://www.gsnv.org.au>

The Genetic Support Network Victoria has a complete list of Victorian support groups relevant to families with genetic conditions.

Chronic Illness Alliance
818 Burke Road, Camberwell 3124
Call: (03) 9805 9134
Fax: (03) 9882 7159
Website: <http://www.chronicillness.org.au>

Association for Children with a Disability
590 Orrong Road, Armadale 3143
Call: (03) 9500 1232
Fax: (03) 9500 1240
Email: mail@acd.org.au
Web: <http://www.acd.org.au>

Ethnic Agencies

Centre for Culture Ethnicity and Health
23 Lennox Street, Richmond 3121
Call: (03) 9427 8766
Fax: (03) 9427 8363
Email: admin@ceh.org.au
Web: <http://www.ceh.org.au>

Action on Disability within Ethnic Communities (ADEC)
13 Munro Street, Coburg 3058
Call: (03) 9383 5566
Fax: (03) 9383 5185
Freecall: 1800 626 078
Email: info@adec.org.au
Website: <http://www.adec.org.au>

Further reading

Counselling Skills

Bolton, R 1986, *People skills*, Simon & Schuster.

Egan, G 2002, *The skilled helper: a problem-management and opportunity-development approach to helping*, Brooks/Cole.

Grief and loss

McKissock, M & McKissock, D 1995, *Coping with grief*, ABC Books, Sydney.

Psychological aspects of genetics

Kessler, S & Resta, R (eds) 2000, *Psyche and helix - psychological aspects of genetic counselling*, Wiley-Liss, NY.

Marteau, T & Richards, M (eds) 1996, *The troubled helix. Social and psychological implications of the new human genetics*, Cambridge University Press.

Weil, J 2000, *Psychosocial genetic counselling*, Oxford University Press, Oxford.

Breaking bad news

Girgis, A & Sanson-Fisher, RW 1998, 'Breaking bad news. 1: Current best advice for clinicians', *Behavioral Medicine*, 24:53–59.

Walsh, RA, Girgis, A & Sanson-Fisher, RW 1998, 'Breaking bad news. 2: What evidence is available to guide clinicians?', *Behavioral Medicine*, 24:61–72.

Websites

Better Health Channel: <http://www.betterhealth.vic.gov.au>

Relevant topics:

- Genetic services in Victoria
- Carers helped by specialist services
- Children experience loss and grief
- Depression is a constant feeling of sadness
- Family break up – how to cope
- Family conflict – how to cope
- Grief support centre helps people cope with loss
- Stress trauma fear – questions answered
- Talking about your problems can help

Genetic Health Services Victoria: <http://www.genetichealthvic.net.au>

Genetic Support Network Victoria: <http://www.gsnv.org.au>

Cultural information

Action on Disability within Ethnic Communities (ADEC): <http://www.adec.org.au>

Centre for Culture, Ethnicity and Health: <http://www.ceh.org.au>

Queensland Government: <http://www.health.qld.gov.au/hssb/cultdiv/home.htm>

Financial services

Investment & Financial Services Association Limited: <http://www.ifsa.com.au>

1.10 Where to refer

Genetic services and genetic counselling

Genetic Health Services Victoria
The Royal Children's Hospital
Flemington Road, Parkville 3052
Call: (03) 8341 6270
Fax: (03) 8341 6390

Monash Medical Centre
Clinical Genetics/Genetic Health Services Victoria
Clayton Road, Clayton 3168
Call: (03) 9594 2026
Fax: (03) 9594 2022

Mercy Hospital for Women
Genetics Department
Clarendon Street, East Melbourne 3002
Call: (03) 9270 2394
Fax: (03) 9270 2498

Non-metropolitan services

(Ballarat, Bendigo, Frankston, Geelong, Mildura, Sale, Shepparton, Traralgon, Warragul, Warrnambool)

Genetic Health Services Victoria
Call: (03) 8341 6270
Fax: (03) 8341 6390

Royal Women's Hospital (prenatal diagnosis)
Genetic Health Services Victoria
Grattan Street, Carlton 3053
Call: (03) 9344 2121
Fax: (03) 9344 2066

Albury/Wodonga Genetics Clinic
Genetic Health Services Victoria
78 Vermont Street, Wodonga 3690
Call/Fax: (02) 60560451

Peter MacCallum Cancer Institute
(cancer genetics)
Familial Cancer Centre
10 St Andrew's Place, East Melbourne 3002
Call: (03) 9656 1199
Fax: (03) 9656 1539

The Royal Melbourne Hospital
(adult and cancer genetics)
Clinical Genetics, 1North
C/- RMH Post Office, Parkville 3051
Call: (03) 9342 7151
Fax: (03) 9342 4267

Postal:
Locked Bag 1, A'Beckett Street
Melbourne 3000

1.11 References

Chapple, A, May, C & Campion, P 1995, 'Lay understanding of genetic disease: A British study of families attending a genetic counselling service', *J Genetic Couns*, 4:281–300.

Hurworth, R, Robins, R & Metcalfe S 2000, *Genetics education for GPs in Victoria: a needs assessment*, Royal Australian College of GPs.

Investment and Financial Services Association Limited 2002, *Life insurance and genetic testing in Australia (fact sheet)*.

Kessler, S & Resta, R (eds) 2000, *Psyche and helix*, Wiley-Liss.

Metcalfe, S, Hurworth, R, Newstead, J & Robins, R 2002, 'Needs assessment study of genetics education for general practitioners in Australia', *Genetics in Medicine*, 4:71-77.

