Important points

- Consanguinity describes a relationship between two people who share a common ancestor: a 'shared blood' relationship
- The most common form of a consanguineous relationship or marriage is between first cousins and in some societies, can account for a large proportion of relationships
- Traditionally, some cultures have practised and continue to practise marriage between relatives such as cousins as a means of strengthening family ties and retaining property within the family
- We all carry several harmful faulty gene copies on our chromosomes but have a working copy on the other partner chromosome to provide the information for our bodies
- Usually two unrelated people will not carry the same faulty gene copy
  - Children of unrelated parents are at low risk of inheriting from each of their parents a copy of the same faulty gene that could result in a genetic condition
  - They have a risk of between 2% and 3% (2 to 3 out of every 100 births) of having a child with a birth defect or disability, many of which will be genetic
- People who are blood relatives share a greater proportion of the same genes than unrelated people do because they have a common ancestor such as a grandparent from whom they inherited their genes through their parents
- The closer the biological relationship is between relatives, the more likely that they will have the same faulty gene in common
- Children of parents who are blood relatives generally have a small increased risk over that of unrelated parents of inheriting from each of their parents a copy of the same faulty gene that could result in a genetic condition
  - **For example, if parents are first cousins**, the risk is a little higher (about twice) ie. 5% to 6% (5 to 6 out of every 100 births). Looked at another way, where parents are first cousins, there is about a 95 chance out of 100 (95%) that they will have a baby unaffected by a condition due to the parents' faulty genes
  - The chances of having a baby with a problem would be higher, and parents more likely to share the same faulty gene copy, if their parents and/or grandparents are also close blood relatives
- In most families where the parents are close blood relatives, there will be no history of a specific condition and there are no tests that can usefully be carried out to see if the baby is at risk for being affected by a particular genetic condition
- Where there is a family history, or where the parents' ancestry suggests their risk for having a faulty gene for a condition is increased eg. thalassaemia, genetic testing may be possible to determine if the parents are carriers of the same faulty gene

'Consanguinity' comes from two Latin words: *con* meaning shared and *sanguis* that means blood. Consanguinity describes a relationship between two people who share a common ancestor: a 'shared blood' relationship; for example, a relationship between two cousins.

The most common form of a consanguineous relationship or marriage is between first cousins and in some societies, can account for a large proportion of relationships.

Traditionally, some cultures have practised and continue to practise marriage between relatives such as cousins as a means of strengthening family ties and retaining property within the family.

Genes and families

The cells of the body contain the genes or set of instructions for the cell to make all the necessary proteins (chemicals) for our bodies to grow and work normally (see Genetics Fact Sheet 1).

If a gene contains a variation which means that it does not work properly, the gene is described as being faulty (i.e. there is a gene *mutation* present). The result is that either a protein is produced that is faulty, produced in limited quantity or is not produced at all (see Genetics Fact Sheets 4 & 5).

Genes come in pairs: one copy of each gene inherited from the mother and one copy from the father. Since there are two copies of the genes in the cells, a variation in one of the gene copies that could be potentially harmful will generally have no direct effect on an individual's health: the working gene copy will usually override the faulty copy of the gene.

These variations that make the gene faulty (mutations) are called 'recessive' (or hidden) to the working copy and the individual is said to carry the faulty gene.

Carrying a single faulty gene copy out of the 20,000 genes in the cell does not usually cause a problem. But if an individual has two copies of the same faulty gene containing a recessive mutation, the cell will not receive the right instructions to enable normal function and may result in the individual having a genetic condition (Genetics Fact Sheet 2).

Importantly, everyone carries several faulty gene copies without having any impact on their health or development.

There are thousands of possibly harmful faulty gene copies but usually two unrelated people will not carry the same faulty gene copy.

- Children of unrelated parents are at low risk of inheriting from each of their parents a copy of the same faulty gene that could result in a genetic condition
- They have a risk of between 2% and 3% (2 to 3 out of every 100 births) of having a child with a birth defect or disability, some of which will be genetic

While everyone usually has the same number and type of genes, there are small differences in the genetic information between individuals that make us unique. Since our genetic information is passed down to us from our parents and grandparents and so on, family members will have more similarities in their genetic information than differences.

- People who are blood relatives share a greater proportion of the same genes than unrelated people do because they have a common ancestor such as a grandparent from whom they inherited their genes through their parents
- Children of parents who are blood relatives generally have a small increased risk over that of unrelated parents of inheriting from each of their parents a copy of the same faulty gene that could result in a genetic condition
- **For example, if parents are first cousins**, the risk is a little higher (about twice) ie. 5% to 6% (5 to 6 out of every 100 births). Looked at another way, where parents are first cousins, there is about a 95 chance out of 100 (95%) that they will have a baby unaffected by a condition due to the parents' faulty genes

Relationships between blood relatives

Geneticists have classified how 'close' relationships are considered, based on the proportion of their genes that they share (Table 16.1). The closer the biological relationship is between relatives, the more likely that they will have the same faulty gene in common.

- Identical twins have all of their genes in common while, statistically, first cousins would have one in eight of their 20,000 or so genes in common
- These figures could be higher, however, in families and communities where there is a higher degree of relatedness between individuals

Relationship to each other	Relationship type	Proportion of genes they have in common
Identical twins (monozygotic)		All (100%)
Brothers and sisters, non-identical (dizygotic) twins, parents and children	First degree relatives (1^0)	Half (1/2, 50%)
Uncles and aunts, nephews and nieces, grandparents and half brothers and half sisters	Second degree relatives (2^0)	Quarter (1/4, 25%)
First cousins, half-uncles and aunts and half-nephews and nieces	Third degree relatives (3^0)	Eighth (1/8, 12.5%)

Table 16.1: *Proportion of genes shared between close blood relatives*

What is the risk for having children with a problem when parents are close blood relatives?

It is not true that those having a child with a blood relative have a very high chance that their children will have birth defects or a disabling condition.

Where the parents have no family history of a specific condition and their parents and perhaps grandparents are not blood relatives:

- If parents are unrelated, their risk for having a child with a birth defect or disability is between 2% and 3% (2 to 3 out of every 100 births)
- If parents are first cousins, the risk is a little higher (about twice) ie. 5% to 6% (5 to 6 out of every 100 births). Looked at another way, where parents are first cousins, there is about a 95 chance out of 100 (95%) that they will have a baby unaffected by a condition due to the parents' faulty genes

In general, parents being close relatives has no effect on the number of babies born with genetic conditions that are due to a faulty gene on the X chromosome (Genetics Fact Sheet 10) or a faulty gene that is 'dominant' to the working copy of the gene (Genetics fact Sheet 9).

It seems likely, however, that when parents are close relatives there will be some increased risk for a child inheriting conditions that are due to a number of different genes acting together (polygenic) or where there is an interaction between genes and the environment such as spina bifida and some forms of congenital heart disease (Genetics Fact Sheet 11).

Unfortunately, what this increased risk actually is for parents in their individual situation is difficult to calculate.

A cautionary note

In societies with a tradition of first cousin marriage, many couples are often more closely related than first cousins are and consequently their risk may be significantly higher.

- **The risk is much higher** than 5% to 6% when both the parents of the first cousin couple -and their grandparents - are also blood relatives
- The actual risk that parents in this situation have needs to be worked out for each family during genetic counselling (see Genetics Fact Sheet 3)

What tests are available when parents are close blood relatives?

Where there is a family history of a genetic condition, the risk of having a baby affected by that condition depends on the pattern of inheritance of the condition in the parent's family (see Genetics Fact Sheets 8, 9 & 10).

In most families where the parents are close blood relatives, there will be no history of a specific condition. In this situation there are no tests that can usefully be carried out, as there is no indication for which condition the baby may be at risk.

In situations where a known condition exists in a family, it may be possible to determine whether either or both partners are carriers of the faulty gene known to cause the condition (see Genetics Fact Sheet 21).

In addition, a few genetic conditions are more common in some population groups than in others. If the ancestry of the parents is clear, genetic testing may be possible to determine if they are carriers of the faulty gene for one of these few conditions (see Genetics Fact Sheet 21).

For example, people where their background is from:

- Europe (including the United Kingdom), have a 1 in 25 chance of being a carrier of the faulty gene that causes cystic fibrosis, a condition that affects the respiratory and digestive system (see Genetics Fact Sheet 33)
- Southern Europe, the Indian sub-continent, the Middle East, Africa and Asian countries have a similar chance of being a carrier of the faulty gene that causes one of the forms of thalassaemia, a condition that affects the blood (see Genetics fact Sheet 34)

Getting correct and up to date information

It is important for couples who are close blood relatives and thinking about becoming parents, to seek genetic counselling (see Genetics Fact Sheet 3).

Some relationships are complex, involving a number of generations where parents are closely related. The specialised knowledge that the genetic counselling team has is required to estimate any possible risk to children resulting from the couple's family relationship.

Other Genetics Fact Sheets referred to in this Fact Sheet: 1, 2, 3, 4, 5, 8, 9, 10, 11, 21, 33, 34

Information in this Fact Sheet is sourced from:

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Edit history

May 2012
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Previous editions: 2007, 2004, 2002, 2000, 1998, 1996
Acknowledgements previous editions: Prof Alan Bittles; Bronwyn Butler; Amanda O'Reilly; Gayathri Parasivam; Mona Saleh;

