

The Fragile X Society

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The Importance of Genetic Counselling

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What is Genetic Counselling?

Many of you will have had 'genetic counselling' whether you realise it or not! Genetic counselling does not entail lying on a couch while someone asks "and how does it feel to have fragile X in the family", but is a process where information is given about the condition and families have the opportunity to ask questions. This information is usually given by genetic counsellors or genetic doctors working at a Genetic Centre.

The information given might vary depending on a number of factors, for example whether the person receiving the counselling is the parent of a recently diagnosed child, or perhaps a more distant family member with no experience of fragile X. Information is tailored to the individual's needs but could include a description of fragile X and how it affects people, its inheritance pattern (how it is passed on in a family), the availability and reliability of genetic tests, including prenatal tests, and discussions about other family members who might need to be informed of the situation. People will be made aware of the different choices available to them, and helped to carry out their decisions. Genetic counsellors will not tell people what to do, or what they think they should do, even if asked their opinion.

You will notice that I used the word "process" in the first paragraph. Genetic counselling is very much a "process"; and with a condition as complicated as fragile X, it is rare for all the information to be given in one session. Genetic counsellors will often work with a family for several months, seeing different family members as appropriate.

In most families there are relatives for whom genetic counselling is not appropriate at the time of the diagnosis in the family but who might need genetic counselling in the future. An example might be young (or as yet unborn) siblings or cousins of affected individuals. Although policies differ, there is general agreement that ideally young people should be given the opportunity to make contact with the genetic service at around the age of 16.

This can be easier said than done. For siblings of affected individuals, there is not usually a problem. The subject of fragile X and its inheritance will arise within the family, and the advisability of genetic counselling can be discussed. Families should have access to the genetic centre where they were originally seen, or if they have moved to a different area they can be seen at a centre in their new area.

Difficulties can arise for more distant relatives. Parents might be aware that their child could be a carrier of fragile X, but find it hard to raise the subject. It can be hard to find

the right opportunity, or the right time, and if the affected relative is distant (either in the family or geographically) the subject of fragile X is unlikely to arise by chance. It is easy to let things go: "I'll talk to him after his GCSE's results/A levels/when she's done her degree/after her wedding/oops – too late". For these families, genetic counsellors can be an invaluable help.

Parents can approach the genetic centre for advice about how to raise the subject with their children. A letter from the genetic centre along the lines of "We have been reviewing our files and notice that Jane and John have reached an age where they could be offered genetic counselling if they wish" might just provide the right opening, and a genetic counsellor will usually be happy to write such a letter at the family's request. Some families assume that the genetic centre will automatically approach them at an appropriate time, but this is not usually the case.

If a family is already known to a genetic centre, it is perfectly in order to contact the centre directly and ask for an appointment, or to speak to a genetic counsellor for advice. If the family needs a referral to a different centre, it is usual to ask the family doctor (GP) for a referral. If any family experiences difficulty in arranging this, the Fragile X Society will be able to help.

Finally, genetic counsellors are nice people! Well, I would say that wouldn't I, being one myself?! Even so, sometimes you will meet a genetic counsellor with whom you do not hit it off, and find it difficult to talk to. If this should be the case, do not be afraid to ask if you can see someone else in the department. It should be possible to arrange this and it will not affect the care you are given.

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