

## A Parent's View

Kylie, my second daughter was born on a sunny, June afternoon. I was delighted; a beautiful baby girl. Following emergency surgery after an earlier miscarriage, I was not sure if I would ever have another baby, so this was a very special time for the family.

When she was born I noticed that her hands and feet were very puffy and swollen. She was also a very grey colour. I was told this was due to poor circulation in the womb and that she would 'pink up'. During the first hours of her life the swelling got worse and a paediatrician was called. The paediatrician said he wanted to do some tests.

The next day when all the tests were complete my husband and I went to see the consultant. We were given the diagnosis of Turner syndrome (TS). We were confused; we had never heard of TS. To discover that your child has a chromosomal abnormality is traumatic enough, but when it is something you have never heard of before it is even worse.

Kylie was transferred to the special baby care unit because of her feeding difficulties. I was in a ward without my baby. My husband was at home. I felt totally isolated and devastated.

Kylie and I were discharged a week later and she had regular check ups with the paediatrician. I began to learn more about TS. When Kylie was six weeks the doctor gave me the address of a support group and it was a relief to contact them. When I read the booklet I was sent a lot of things made more sense, but many questions remained unanswered. Why me? Did I do something wrong? Could I have done something to prevent this happening? If I had other children would they have Turner syndrome? I wasted so much time feeling distressed and missed out on much of the joy of being a new mum.

It was suggested that I see a genetic counsellor. This was a turning point for all of us. During the appointment we were given a full explanation of how chromosomes work, what is thought to happen in TS and an explanation of the difference between Classic and Mosaic TS. Professor Michael Connor explained the condition fully; he took time to answer our questions and concerns. He drew diagrams and showed us photos. Everything became much clearer. He was genuinely interested in our concerns.

I was encouraged by Professor Connor to start a support group in Scotland and he said he would help. We held the first Scottish TS meeting when Kylie was six months old. Thirteen years on the Scottish group continues to meet four times a year and Professor Connor spoke at our tenth anniversary meeting, where Kylie was able to answer questions about her chromosomes. Her understanding of her condition has come from the understanding that we gained from our genetic counselling.

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Further information about Turner Syndrome can be obtained from:

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# Genetic Counselling

FACTSHEET TEN

# Genetic Counselling

Being told that your baby or child has a genetic condition is extremely distressing and worrying. You will have so many questions that you want to ask about Turner syndrome. You and your family will need information, advice and support. This leaflet outlines just one source of excellent information and support - genetic counselling - and explains how you can obtain such help.

## Role of genetic counselling?

Any genetic testing should include genetic counselling as an integral part of the process, and should be offered and strongly recommended in most genetic testing situations. It should also be a pre-requisite to any prenatal diagnostic procedure.

Genetic counselling aims to explain the facts as clearly as possible. It helps people who are faced with the diagnosis of genetic disease to understand the factual information about the condition and the effect it will have on their lives.

Genetic counsellors are specially trained professionals, most of whom come from a medical or nursing background. They have first-hand knowledge of genetic diseases and their practical impact. The counsellors are trained to help people through the inevitable emotions that a diagnosis arouses and they are sensitive to the fact that a diagnosis can have different meanings to different people.

A genetic counsellor can provide accurate, up to date information in a way that people can easily understand. They aim to bridge the gap between the often complex and fast moving field of genetics and the everyday world. Counsellors help people to understand the nature of the condition and what having it will mean in practical terms, the possible risks or otherwise of recurrence and the implications for family members.

## Requesting a referral

Genetic counsellors work as part of a healthcare team which includes consultants, nursing and primary care teams. If you think you would benefit from talking to a genetic counsellor and it has not already been suggested to you, then raise it with your GP or consultant and ask to be referred.

The following article, 'A Parents View' by Arlene Smyth, Executive Officer of the Turner Syndrome Support Society is an edited version of her own personal experience of genetic counselling. The article was originally published in the British Medical Journal (2001;322:1005-1006).

