

# Family Matters

coping with life

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## Rare condition wasn't diagnosed until she was seven

**CHROMOSOMES ARE the DNA molecules that contain our genes.**

**Most humans have 23 pairs, but chromosome abnormalities — like an atypical number or a structural defect in one or more chromosomes — can occur.**

**One in every 1000 babies worldwide is born sick or disabled because of a rare chromosome disorder. And at least one in 200 more is born with a chromosome abnormality that won't be recognised until later in life.**

**Kathryn McKerracher, from Twechar, near Glasgow, was born with Chromosome 18q-, meaning a piece of the long arm of her 18th chromosome is missing. Despite showing symptoms at an early age, it took seven years for her to be diagnosed.**

**ON February 25, 1980, and after a 17-hour labour, Bonnie McKerracher gave birth to her second daughter, Kathryn.**

By Tracey Bryce

Her first daughter, Jeni, had arrived without complications but all was not well with Kathryn.

"Immediately after the birth I heard the midwife on the phone saying Kathryn had a twisted foot and a swelling around her belly button," Bonnie recalls.

"Doctors assured us both could be fixed."

The swelling was caused by an umbilical hernia, where part of the bowel pushes through a weak spot in the surrounding muscle wall.

### Surgery

At five months Kathryn underwent two operations, one to push the bulge in her stomach back into place and strengthen the abdominal wall, and another to release the Achilles tendon in her left foot which bent upwards at a 90 degree angle.

The surgery was successful in correcting the hernia, but despite a further three operations on her foot, by the time she was two Kathryn was still struggling to walk. Bonnie and husband, Ken, noticed she was also slow in starting to talk.

Tests suggested she was suffering from mild hearing loss, but not enough to affect

her speech development.

But her continuing lack of development resulted in referrals to speech therapists, orthopaedic doctors, ophthalmologists and specialists at the ear, nose and throat clinic.

They found she had impaired vision, poor muscle tone and severe delayed development but no-one could say why she had so many ailments.

"By the time she was three, Kathryn was being checked in and out of clinics like a library book," Bonnie says. "But nobody put it all together or even suggested the problems might be related."

"We put Kathryn's struggle with walking and talking down to her foot and her hearing and visual difficulties."

"But when her little brother Michael, who is 20 months younger, overtook her in development milestones, we realised something much more serious must be wrong."

"By the time she went to school she'd started speaking, but wasn't clear. The teachers and other children couldn't understand her."

"The fact she presented a bit of a challenge to them meant she was considered a problem child."

At home, however, Bonnie,



■ Kathryn and her mum Bonnie travel regularly to the US for meetings which have helped change Kathryn's life.

## Kathryn's parents knew something was badly wrong when she missed development milestones

a former teacher, was patient enough to realise Kathryn's potential.

"When the kids played at games like shops and hospitals, Kathryn integrated better," she smiles.

"To encourage her to talk, I made outfits so the children could dress up like doctors, policemen, firemen and lollipop patrol people. Before long, I'd started my own business, making playwear for playgroups and playboxes for Action for Sick Children."

"One day, one of the mums came to the house to order outfits. When she saw Kathryn, she was intrigued by her almond-shaped eyes, narrow lips and a button nose with a flat bridge."

"The woman explained her husband was a plastic surgeon

who was researching a form of dysmorphia, where the bones in the face aren't properly formed, and she thought Kathryn might have it."

It turned out Kathryn didn't have the syndrome he was looking into, but he suggested further tests. The result was Kathryn being referred to the Institute of Medical Genetics at Yorkhill, Glasgow.

### Tests

There, further tests revealed she had a chromosomal abnormality. And six weeks later, aged seven, she was diagnosed as suffering from Chromosome 18q-.

"It all made sense then," Bonnie recalls. "One of the aspects of the syndrome is autistic tendencies, which Kathryn had been showing."

Since then she's achieved two Masters in Medical Genetics and Bioinformatics and a qualification in web design.

"When I was younger I wanted to be a scientist," reveals Kathryn. "Now I have the qualifications to be one. Some teachers used to say I wouldn't make it, so I guess I've proved them wrong."

Ten years ago, Kathryn came across a charity in the US called the Chromosome 18 Registry and Research Society.

"Its annual family conference in Texas was only six weeks later," she says.

"Mum and I decided to go. It was amazing — the first time I'd ever met other people with Chromosome 18. The condition is so rare people with the disorder never meet another affected person."

Bonnie and Kathryn have travelled to the US for the past nine years to attend the annual event.

### Marriage

It's changed Kathryn's life in many ways. In 2006, at the annual family conference in California, she met Australian Martin Bridge (24) who has the same condition. The couple are due to get married in Perth, Australia, next month.

"Because he has Chromosome 18q- too, we really understand each other," Kathryn beams.

At the first conference they went to Bonnie and Kathryn met Chris and Judy Wilkinson, the grandparents of an Aberdeen boy who has the condition.

The families have kept in touch and in March last year launched their own charity — Chromosome 18 Europe — which now has more than 100 members in 15 countries.

On July 30, the charity will stage the first European conference on Chromosome 18 in Glasgow.

Experts from the US, including Dr Jannine Cody, founder and president of the Chromosome 18 Registry and Research Society, and health professionals from the UK are expected to attend, along with delegates from across Europe.

But £22,500 is needed to fund the conference. Already, £11,500 has been raised and the organisers will be working hard over the next few months to secure the remainder of the cash.

"There's no way the conference isn't going to happen," Kathryn smiles. "My mum will make sure of it."

● To find out more about Chromosome 18 Europe log on to [www.chromosome18eur.org](http://www.chromosome18eur.org)