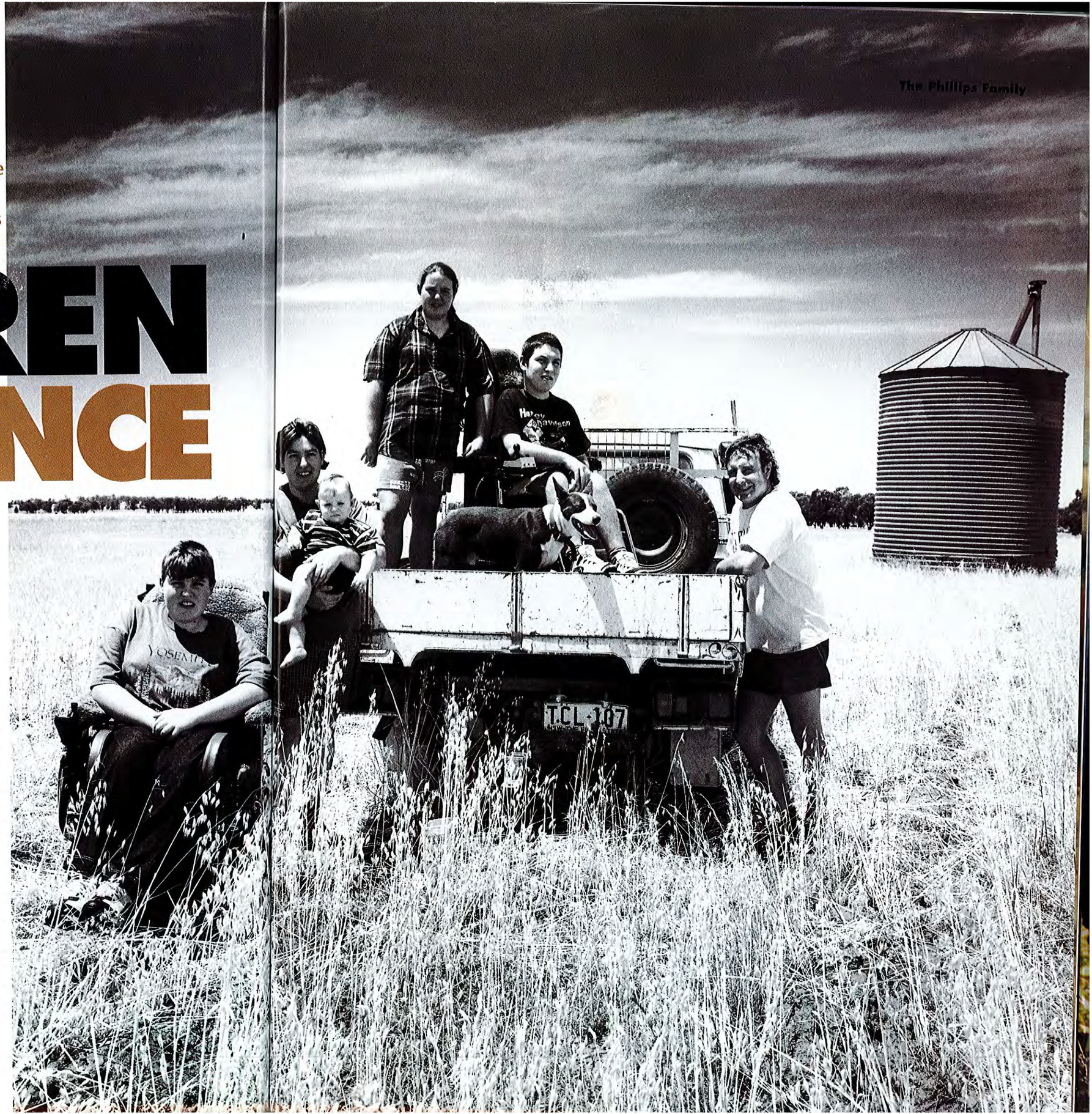
Discovering your baby or child has a life-threatening or debilitating disease is devastating for any parent. But imagine the despair of finding out you were the one who passed on the disorder. Julie Nance talks to three women who have faced this shocking news and who are now full of hope for the future.

CHILDREN OF CHANCE


The Phillips Family

Each year, Australian women give birth to babies with rare conditions, including haemophilia, Duchenne muscular dystrophy and fragile-X syndrome. These X chromosome-linked hereditary disorders are often passed on by women, but affect mostly boys.

With genetic risks, it's just like tossing a coin – all a matter of chance. Women with an X-linked disorder in their family face a 50 per cent chance that any son they have will be affected. If they have a daughter, there is a 50 per cent chance she will be a carrier. Men who have X-linked disorders will pass the gene on to their daughters, who become carriers, but their sons will be unaffected.

For many couples – up to 30 per cent in the case of haemophilia – the genetic disease comes completely out of the blue. They usually have a healthy-looking toddler who goes on to show worrying symptoms, and then the heartbreaking diagnosis is made. Some families unknowingly have more than one child with the same disorder, because the first one is not diagnosed by the time the second or third child is born.

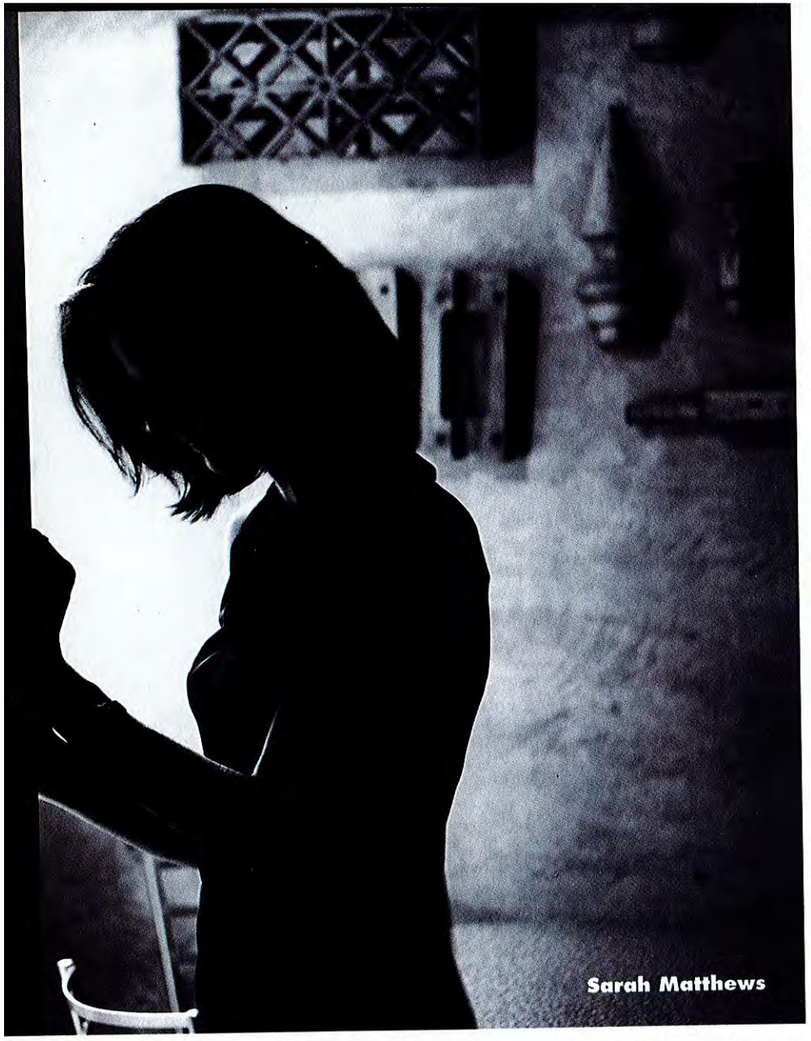
In some cases, DNA testing can determine whether a person is a carrier and whether an unborn baby is affected. The most common prenatal test is chorionic villus sampling at 10 to 12 weeks of pregnancy. Kate Talbot, genetic counsellor in maternal and foetal medicine at Sydney's Royal Hospital for Women, says she has counselled women who've had three or four bad test results and have had to terminate the pregnancies. "There are ethical as well as emotional dilemmas for many parents," Talbot says. "If it has happened a third time, the parents may face the prospect that it could keep on happening. We all assume we have the right to have a normal, healthy baby. It is incredibly traumatic and stressful for couples each time there is a pregnancy."

Duchenne Muscular Dystrophy

As Maria Phillips cradled her new baby son in her arms, she had little idea of the genetic gamble she had taken. The 19-year-old mother had watched the crippling and terminal disease Duchenne muscular dystrophy (DMD) gradually take hold of her elder brother, Geoff – leaving him unable to feed himself or do any other daily tasks. But knowledge of the disorder in the early '80s was scant and there was no reliable test to determine whether Maria was a carrier. So she and her husband Garry took a chance but, sadly, it was one that didn't pay off.

"We were young and naive, and thought surely it won't happen to us," says Maria, now aged 34 and living on a farm at Condobolin in central western New South Wales. "At three months of age, Nicholas was assessed and we were told there was a fifty/fifty chance he would be affected. All we could do was wait."

At age four, Nicholas, who by then had a two-year-old sister, Sonia, began to display worrying signs. He waddled when he walked and was unable to run properly. A paediatrician diagnosed DMD and told the Phillips that, apart from physiotherapy and exercise to strengthen Nicholas' muscles and help delay symptoms, there was nothing anyone could do. The family was dealt another blow when Nicholas' cousin Tim, who is 10 months younger, was also diagnosed >



Sarah Matthews

> with DMD. "Although I began to accept Nicholas had the disease before he was diagnosed, I was still shocked and upset," says Maria, whose brother died at the age of 30. "We started thinking about how we could help him and give him a good quality life. You go through a period of denial and grieving, but eventually we got over that."

DMD affects approximately 450 Australian males, but rarely affects females. Most sufferers survive into their 20s, but a minority only make it into their late teens.

Today, Nicholas is an easygoing 15-year-old in Year 10 who is planning to complete his HSC and do an office course at TAFE. He has been wheelchair-bound since he was seven, his muscles having gradually deteriorated over the years. He can only just lift his arms enough to feed himself and has little strength in his neck and upper torso.

Tim has suffered similar muscle decline. He moved in with the Phillips' three years ago because his divorced parents were unable to care for him. "Garry and I thought about it for a long time then decided: we've got one, we may as well have two," says Maria, whose family has home care assistance for 16 hours each week. "I have days when it all seems too much and I sit down and cry, but then I get up again."

Although the Phillips haven't fully discussed the implications of the genetic disease with their 13-year-old daughter, she has already asked, "are my kids going to have this?" Future tests will determine whether she is a carrier like her mother.

Nine months ago, the family was blessed with a new addition, baby Dean. Unlike his elder brother, Dean's health status was determined in the womb through prenatal testing. After enduring "one hell of a week" waiting for the results, Maria and Garry were given the good news – their unborn baby had been spared.

"It was the best news anyone could ever have, apart from finding a cure, of course," Maria says. "We had already decided, if we were going to have another boy with muscular dystrophy, we were going to terminate the pregnancy. There was a fifty/fifty chance – it all depends on how the dice lands. I'm glad we took the chance and went through with it. We didn't feel our lives were complete."

CHILDREN OF CHANCE

Haemophilia

When Sarah Matthews made the agonising decision to terminate her pregnancy, it went against her personal beliefs and her Catholic faith. But uppermost in the 30-year-old's mind were the years of pain endured by her brother and uncle as a result of severe haemophilia. Sarah and her husband Robert learned through prenatal testing that their unborn foetus was male and would also be affected by the blood-clotting disorder. Despite advances in treatments, the mother-of-one did not want to inflict the disease on another human being.

"Although we discussed it with each other and we were counselled, you still wonder if you are doing the right thing," says Sarah, who had the termination two years ago. "We didn't have to go through this with our first baby because, thankfully, the test results showed she was a girl. I was pregnant last year and miscarried. I felt like it was God punishing me. I was always told abortion was a wrong thing – that it is murder."

Haemophilia affects approximately 1,600 Australian males to varying degrees. Haemophiliacs' blood clots very slowly, so they experience prolonged bleeding following any injury or wound. Women who carry the haemophilia gene generally show no symptoms of abnormal blood clotting, but some carriers, called symptomatic carriers, have low levels of clotting-factor activity. Sarah is a symptomatic carrier and needs a clotting agent even for minor surgery.

Several generations of Sarah's family have been affected by haemophilia. Her 40-year-old uncle, Peter, is crippled with arthritis as a result of years of bleeding in his joints. Her earliest memory of her 17-year-old brother, Andrew, is as a toddler being held down by his parents and three nurses while they tried to infuse him after a bleed. A couple of years ago, he was diagnosed with hepatitis C as a result of infected blood he received years previously. Before screening was introduced in the early '80s, around 30 per cent of haemophiliacs were infected with HIV. Sadly, half have since died.

"I vividly remember all the times Andrew cried in pain, the late nights in hospital being given treatments, and our family on edge when he was around other children in case he was hurt," says Sarah. "If Andrew was running and knocked himself, he would bleed into the joints, and his ankle, or whatever it was, would swell within 20 minutes. He would often just lie in his room and rock, in a bid to block the pain."

During the difficult decision-making process, made easier by the "wonderful counsellors and doctors", Sarah says she had to consider the impact on her seven-year-old daughter and extended family of bringing >

What's her name?
VM
veronika maine
Available at selected boutiques Australia & New Zealand. For your closest stockist call 1800 06 07 06

> a child into the world with haemophilia. Although they stand by their decision to terminate at that time, the Brisbane couple has recently decided to try for another baby in two years and forgo prenatal testing.

"People may be confused by this but, after the trauma we have been through, we realise any child is a gift," says Sarah, who will have her daughter tested as a possible carrier when she is nine. "There are worse things than haemophilia, and in a way we are lucky because we are familiar with it. I could torture myself by looking back at the termination and wondering if it was the right thing to do. It was at the time, but there are a lot of moral issues to consider."

Names in this case study have been changed to protect the subjects' identities.

CHILDREN OF CHANCE

Fragile-X Syndrome

When James Hook was three years old, he hated being cuddled and found comfort in banging his head against a wall. He would often flap his hands in an autistic way, and was so driven by jealousy in kindergarten that he hit a girl over the head with a shovel. Understandably, his parents, Sue and Graham, were concerned by their child's unacceptable and often dangerous behaviour. But it wasn't until two years later that the reason for their toddler's inner torment was revealed. James was diagnosed with the most common form of inherited mental retardation – fragile-X syndrome. James mainly vented his anger and frustration on the person who unwittingly gave him the disorder – his mum.

"Before we knew what it was, we were tearing our hair out, thinking we were really bad parents who were bringing up this monster," says Sue, 45. "A DNA test was carried out on James' blood on the off-chance it was fragile-X. I have since found out I am a carrier and so is my brother. My mother gave it to us."

A Sydney Children's Hospital study has found fragile-X affects one in 4,000 males and one in 2,000 females, with the major feature of the syndrome being developmental delay. About 90 per cent of males are in the moderate to severe range, while 10 per cent have the condition in a mild form. Females are not usually as severely affected as males, but about 30 per cent of female carriers have learning difficulties. Unlike other X-linked disorders, males can be carriers of fragile-X.

Sue had a completely normal and healthy pregnancy, oblivious to the fact she was carrying fragile-X. "I know that there are women who are extremely upset they were the ones who passed the disorder on," says Sue. "But it didn't affect me that way. I was just concerned about what the consequences would be for James."

James is now 10 and, with personal assistance, attends a normal school in the lower Blue Mountains of New South Wales. His biggest problem is the storage of information in his brain – what he knows today, he forgets tomorrow. Sue says her son is improving socially, but he finds it difficult to make friends and, once he does, he is "all-encompassing on that one person", not wanting them to play with anyone else.

Sue and Graham look to the years ahead with optimism, but all future plans include their son. "We hope James will get a job and hold it down, but we don't think he'll ever be able to live out in a flat on his own," Sue says. "I've been told I'd have a moderate chance of having another child affected by fragile-X. The odds are too high. I don't think I would cope very well with it again." □

Who can you call?

There are a number of support groups around Australia with the specialised knowledge to help anyone with questions about genetic disorders:

- Phone the Fragile-X Association of Australia Incorporated on (02) 9450 2158.
- DMD – Look for the Muscular Dystrophy Association of Australia in your state.
- Phone the Haemophilia Foundation Australia on (03)9572 5533.
- Phone Support After Foetal Diagnosis of Abnormality (SAFDA) on (02)9382 6670.

The Hook Family