

The rogue genes that put men off sex

By Rachel Ellis

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Depressed? Low libido? Unable to concentrate? Like thousands of men, you could have a devastating condition even your GP won't have heard of...

After trying to conceive for 18 months, Richard and Jane Brown decided to seek medical help. Because in 40 per cent of infertile couples it's the man who has a problem, Richard was the first to undergo tests.

'I remember thinking there can't be anything wrong with me because I've got such a hairy chest. I thought I must be full of testosterone,' says Richard, 32, a research chemist from Sawley, Nottingham.

But the results showed Richard was, indeed, the problem: his sperm count and motility (the speed at which sperm moves) were ten times lower than normal.

In fact, as he was astonished to learn, Richard has Klinefelter's Syndrome, a genetic condition affecting only men.

It causes a testosterone deficiency; as well as infertility, symptoms can include a low sex drive, depression, poor concentration and learning difficulties.

Between 50,000 and 60,000 men in Britain have Klinefelter's, but perhaps as many as half of those affected will not realise it. This might be because some have a mild form - and the first indication of a problem is having difficulties fathering a child.

The condition takes its name from the U.S. endocrinologist Dr Harry Klinefelter, who first identified it in 1942.

Normally, boys inherit 23 pairs of chromosomes carrying all the genetic information from their parents: one pair has an X sex chromosome from their mother and a Y chromosome from their father (girls inherit two X chromosomes, one from each parent).

Klinefelter's occurs when boys inherit an extra X chromosome - in rare cases, men can even have two or more of these extra chromosomes. As a result, the boy ends up with smaller testicles - and this affects the amount of testosterone they produce.

with the condition have testes that initially develop very quickly, then shrink in childhood,' says Professor Ashley Grossman, a consultant endocrinologist at Barts and the London Hospital.

'By puberty, they have very small testes and, as a result, very low levels of testosterone; that is the reason their fertility is affected.'

The first noticeable signs are nonphysical symptoms, such as a lack of concentration, speech and language problems and shyness; boys with Klinefelter's may also be clumsy and dislike physical games.

The problem is these symptoms are often misdiagnosed as learning difficulties or are missed completely, says Professor Grossman. Getting the correct diagnosis can depend on whether a doctor is familiar with Klinefelter's.

The condition becomes more noticeable as boys go through puberty. Their arms and legs grow quickly and keep on growing so they tend to have extra-long limbs,' says Grossman.

This is thought to be caused by low testosterone levels delaying closure of the ends of the bones.

'They often don't develop male secondary features such as facial hair and remain boyish. Those with the most severe form are not terribly interested in sex and, if they do have sex, often can't perform very well.

'Testosterone sets the threshold for arousal, so men with lower levels can have sex and be potent, but the level of stimulation needs to be higher.

'Between 20 and 30 per cent of sufferers also have behavioural problems such as learning difficulties and speech and language problems. Yet often no one realises these are men without testosterone and they are simply put down as being quiet, unassertive or not interested in girls.

'There is no greater incidence of homosexuality among men with Klinefelter's. It is simply a physical condition caused by their testicles not developing properly.' Other symptoms include the development of breasts - up to a third of affected men develop breasts large enough to embarrass them - and a pear-shaped body.

Symptoms vary widely - some men have no obvious physical signs, others are not affected developmentally. But for all men with Klinefelter's, infertility is inevitable, as Richard discovered.

A low sperm count is a tell-tale sign of the condition; blood tests are then used to detect if an extra chromosome is present.

Richard's tests revealed that he had a 'mosaic' form of Klinefelter's Syndrome - this means not every cell in his body is affected.

'When they gave us the diagnosis, it was a shock because I have none of the other symptoms of Klinefelter's and I'm not a shy and retiring type. You go through life thinking everything is OK and when you find out it is not, it does take you by surprise,' he says.

'When we were told, Jane gave me a big hug and said it was not my fault. Thankfully, they were able to find some sperm - I was so relieved we could try to have a child of our own.'

The couple's best chance was a type of IVF treatment called intra-cytoplasmic sperm injection (ICSI), where a single sperm is injected directly into one of the eggs to fertilise it.

Richard, Jane and Elizabeth Brown

However, before they were able to go ahead with the treatment, the Browns had to see a genetics counsellor to discuss the risks of passing on Klinefelter's Syndrome.

Men with the full-blown condition have a higher risk of passing it on. There is no way of predicting how seriously a child will be affected. Richard was told he had a 1 to 2 per cent chance of this happening (girls can also inherit an extra X chromosome, but it does not cause any problems).

Though the risk was considerably higher than in the normal population - when the chance of a boy having Klinefelter's is one in 600 - the couple decided it was a risk worth taking. So three years after first trying for a baby, the couple started ICSI treatment last July.

The procedure was a success and their daughter Elizabeth was born in April.

Colleen Lynch, an embryologist and molecular geneticist at the CARE clinic, Nottingham, where the couple were treated, says men with Klinefelter's will almost inevitably need in vitro fertilisation in order to father a child.

'In the past, the only option for these men was sperm donation,' she says. 'However, techniques are constantly improving and we are able to help men with low sperm counts by doing surgical sperm recovery and ICSI.'

Meanwhile, for men with physical symptoms of the condition, daily doses of testosterone - in spray, gel or injection form - will be necessary for the rest of their lives.

They are also at greater risk of osteoporosis - low testosterone levels affect their oestrogen levels, which is needed by men as well as women for healthy bones. So they should have bone scans at least every two years. Fortunately, Richard needs no treatment for the condition.

And with two other embryos, there is a real chance he and Jane will be able to have another child.

'We are ecstatic to have our daughter,' says Richard. 'But I do think why did it have to be me that had Klinefelter's.'

'When I was diagnosed, I couldn't help but feel guilty that my wife had to go through all of this. As a man, it stays with you that you can't do your job and father a child naturally.'

'But we are a strong couple and, now we have Elizabeth, all those feelings are drifting away.'

* For more information, contact the Klinefelter's Association: 08452 300047.

Read more: <http://www.dailymail.co.uk/health/article-1202578/The-rogue-genes-men-sex.html#ixzz0MrhSUt6c>

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