

## **Case study: life with Kallmann Syndrome**



*Neil Smith, a biomedical scientist working in the blood transfusion service in East Surrey, has a rare condition called Kallmann Syndrome, also known as congenital hypogonadotropic hypogonadism (CHH), a rare genetic disorder that stops a person starting or fully completing puberty.*

At the age of 23, after he'd finished University, Neil Smith was finally diagnosed with Kallmann Syndrome.

Having been told by every doctor up until then that he was a 'late bloomer' and to 'wait and see', he happened to meet an endocrinologist in the course of his first job working at the Royal Free Hospital in London.

'The first question he asked me after I told him about the lack of puberty was 'can I smell?', says Neil. 'It was the first time a doctor had asked me that question. I just happened to be working at one of the few hospitals in the country that specialise in Kallmann syndrome and CHH.

'From that point I was put on the right form of treatment and eventually began to look my age and to start shaving. I am currently on fertility treatment.'

Neil has made it his mission to improve awareness of Kallmann Syndrome, travelling the world to attend conferences, meeting with clinicians and researchers, co-authoring papers, and advocating for patients.

He knows how vital early diagnosis is, and the heavy psychological toll of a late diagnosis. Neil believes ongoing global research, including at centres in Newcastle and London, could make early postnatal diagnosis possible.

'When he I was first diagnosed in 1990 they knew about five genes; now about 30 genes have been linked with Kallmann Syndrome and congenital hypogonadotropic hypogonadism,' he adds.

'I think research is working towards a positive genetic diagnosis. The number of genes we are aware of is increasing, and while 50% of cases have an unknown genetic cause, the more genes are studied the more hope there is.

'With next generation sequencing, where the whole genome is sequenced, there is an even better likelihood of finding out what is happening.

'Once you have that diagnosis, the treatment is pretty straightforward hormone replacement therapy.

'But early diagnosis and treatment makes all the difference. The earlier the treatment the better the prognosis and the fewer psychological problems.'

He says Rare Disease Day provides a chance to raise the profile of his condition, as well as gain an of understanding other rare diseases.

'There are only around 5,000 people with Kallmann syndrome in this country – but when you put them together with all the other people with rare diseases you are talking about millions,' he adds. 'That gives us all a bigger voice.'

### **Kallmann Syndrome**

The primary symptom is a failure to start puberty.

It affects both men and women and leaves patients infertile and with little or no secondary sex characteristics.

It is also associated with a lack of sense of smell.

A range of other physical symptoms affecting the face, hands and skeletal system can also occur.

The only other major health concern is osteoporosis in both men and women due to lack of androgens.

The underlying cause is a failure in the correct production or activity of GnRH hormone by the hypothalamus. This results in low levels of the sex hormones testosterone in males or oestrogen and progesterone in females.

Diagnosis normally occurs during teenage years when puberty fails to start.

Treatment is hormone replacement therapy, normally life-long.