**The KS Story**

*You are not alone*

**AN INFORMATIVE GUIDE & FURTHER STUDY**

*The KS Story* is the culmination of twelve years ongoing study and an emotional journey through the workings of the congenital chromosome condition Klinefelter’s Syndrome (KS).

The book was created primarily as a conduit to respected support groups, medical specialists and research units, with a chapter devoted to further study, including a comprehensive support address and online section and a selection of books, research papers, medical journals and scientific magazines.

The book also offers a glimpse into the various aspects of Klinefelter’s Syndrome; showing how it affects individuals and those who love, care and treat them.

*The KS Story* has been compiled by a person with Klinefelter’s Syndrome (karyotype: 47,XXY), from reputable medical, research and support group sources, and with the help and support of several UK and international scientists in the fields of biology, cytogenetics, endocrinology, neuroscience and sexology.

To balance the overwhelming amount of data there is a degree of comic relief in the writing, with a sprinkling of cartoons. All that is asked, is that you please put aside any preconceived ideas or stereotypes and view the guide with a respectful and open mind.

**The KS Diaries**

*LIVING WITH KLINEFELTER’S SYNDROME*

Sensitive, fearful, powerful, touching, forgiving, loving...

A planned second book (pdf) will present personal histories of childhood, adolescence and adulthood titled: *The KS Diaries* and will give you a glimpse through the eyes of those who live and cope with Klinefelter’s Syndrome every day.

However, this book is subject to you and people like you. If you wish to contribute your own KS story, please see page 80.

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**WARNING**

*The KS Story You are not alone*

contains frank and sensitive information of a gender & sexual nature, relating to Klinefelter’s Syndrome and the effects it has on an individual.

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Another page shows a blue background with the text: "Klinefelter’s Syndrome. AN INFORMATIVE GUIDE & FURTHER STUDY. Compiler: Iain W McKinlay 47,XXY. Foreword by: Prof Milton Diamond Ph.D. A reference book for healthcare professionals, counsellors and anyone who works with children and young people."
It is strange to think that a condition which was discovered in 1942 is still basically unknown to the general population, even though it is now classed as a “common genetic condition” in males (1:750-1000).

And the reason? It comes down to Invisibility and Complexity. ‘Invisibility,’ in that generally no characteristics (phenotype) are present at birth, so diagnosis is not usually made at this time, unless the mother has undergone specific karyotype tests, possibly for aneuploidy conditions in older mothers; generally over 35 years old, examples: trisomy 13, 18, 21 (Down’s Syndrome), or X & Y (Klinefelter’s Syndrome).

‘Complexity’ also plays a part, as those with Klinefelter’s Syndrome can also be diagnosed at any time after birth, generally because of a lack of academic or social skills pre-puberty, including physical aspects at around puberty and through the teenage years, to infertility problems as adults, or as a result of obesity, osteoporosis, diabetes, or other possible linked conditions in later life.

To those with KS, there will be some with mild implications while there will be others who suffer in silence, aware that they are different but oblivious to the condition and support networks and afraid to openly discuss their feelings with their parents, spouse/partner or doctor.

Commenting purely on the UK, I am unaware of any national figure of diagnosed individuals, however, given the current estimate for annual live births of between 400 and 500 for the more frequent 47,XXX karyotype and an upper age of 80, there is a potential for around 40,000 people in the UK walking around with Klinefelter’s Syndrome.

Add to that the rarer male karyotypes and mosaics (please see p23); parents & family members; spouses/partners; relatives... and you have a substantial number of people involved with Klinefelter’s Syndrome.

With only a few hundred registered with UK support groups, I believe there is a need for more education. However, knowing where to look for reputable sources of information is another story.

For the many individuals who know there is something not quite right about themselves and who may feel confused or alone.

For those who have just been diagnosed, feel fine, but want to understand the mechanisms behind their traits and symptoms.

For the aforementioned people and with respect, to doctors & medical staff, health workers, teachers and anyone who works with children and young people – and for society in general, I offer this informative guide to Klinefelter’s Syndrome.

If The KS Story can help just one person take those first steps to finding the truth about themselves and seeking any appropriate treatment, or give a parent, spouse or partner who has noticed changes in their loved one vital information, then the guide will have made a difference and I’ll be more than satisfied.”

Iain W McKinlay 2010.

Phenotype n. The observable characteristics of an individual...
Karyotype n. The chromosome set of an individual...
Aneuploidy n. The condition in which a chromosome number of a cell is not an exact multiple... (e.g. X or XXX).
Trisomy n. An aneuploidy condition where there is an extra chromosome present within each cell of the body... (e.g. XXX).

(Please look at a medical dictionary or online for a full description of the above terms.
See page 13 / M2 in margin, for two online encyclopedia examples you could try. There are a load out there, so find the one that suits you best).
The KS Story has been compiled from medical papers, journals, books and the Internet, from respected research & medical specialists and support groups, by an individual with Klinefelter’s Syndrome (XXY).

The KS Story is not mine alone, but a fusion of knowledge and experience offered by the latter dedicated and compassionate people who have contributed to its content.

This informative guide is offered primarily as a conduit to the above people, to whom you should direct all enquiries for the most appropriate support, advice and literature.

All that I ask, is that you please put aside any preconceived ideas or stereotypes and view the following pages with an open and respectful mind.

Harry Fitch Klinefelter Jr

Image courtesy of The Alan Mason Chesney Medical Archives of the Johns Hopkins Medical Institutions.

Patricia A Jacobs OBE

Image kindly supplied by Prof P A Jacobs.
Foreword

XXY The Most Common Genetic Difference

Milton Diamond Ph.D.
University of Hawaii  John A. Burns School of Medicine  USA
Image kindly supplied by Prof M Diamond.

When Iain McKinlay approached me with questions about Klinefelter's syndrome for a book he was writing on the subject I was surprised. As one of the more common atypical genetic conditions I thought a book on the subject would have been done several times over from not only scientific and medical perspectives but also from different personal points of view. I was quite mistaken. There were none to be found. Certainly many scientific, medical and informative articles covered the topic but nothing existed that attempted to gather all aspects of the subject for the average reader. There was especially nothing generally readily and in detail available for those individuals with the condition or for their family and friends, or even for those just interested or curious who wanted more information.

In this volume you will find exactly that. Iain McKinlay presents, in a candid and direct way, those things he had found useful in his quest to understand his own XXY situation and aspects of himself. Here he presents his findings so both he and the reader become fairly current in ability to answer many personal questions that occur to those living with this circumstance and others trying to come to terms with it.

Despite the relative widespread occurrence of the XXY combination in people the condition has only come out of the medical and scientific community to common attention about a decade or so ago. And the way it has become understood has varied greatly. Some concentrate attention on the genetics involved and others attend more to behavioural or potential medical concerns or consequences of having any of the Klinefelter's combinations e.g., XXY, XXXY, XXYY. Some consider Klinefelter's syndrome and its many variations as intersex conditions, others don't. In any case it is a uniting of typical X and Y chromosomes to form different combinations. This fusion occurs naturally and in different forms for the same reasons that there is variation in just about everything in nature. We all develop with different hair, eye and complexion colors, of different height and weight and variation in different forms for the same reasons that there is variation in just about everything in nature. We all develop with different hair, eye and complexion colors, of different height and weight and variation in different forms for the same reasons that there is variation in just about everything in nature. We all develop with different hair, eye and complexion colors, of different height and weight and variation in different forms for the same reasons.

The book is unique for several reasons in addition to the subject matter covered. It is the product of Iain's individual labor of intense curiosity and creativity, it is a work of Iain's talent as a book designer, and the contained drawings and art work are testament to his artistic abilities. His generosity in offering this book to the public is another mark of this special individual. I think many owe him great and heartfelt thanks.

Without doubt this book can and will serve as a starting place from which many readers will go further. There are all sorts of resources listed.

The book is unique for several reasons in addition to the subject matter covered. It is the product of Iain's individual labor of intense curiosity and creativity, it is a work of Iain's talent as a book designer, and the contained drawings and art work are testament to his artistic abilities. His generosity in offering this book to the public is another mark of this special individual. I think many owe him great and heartfelt thanks.

It was never my intention to profit from this book, merely to 'do my bit' and to stop going stir-crazy while undergoing extensive medical treatment.

However, while I am delighted to offer The KS Story to you as a gift, may I ask one favour? That you please consider a donation to help support the organisation where you obtained the pdf with their educational projects. The amount is your choice; whatever you feel is appropriate.

You will be contributing towards valuable work and furthering the knowledge of Klinefelter's Syndrome in your own country and around the world.

Most support groups have secure payment methods online, or you can send them a cheque or postal order. If you're not sure, just ask them.

That said, there's absolutely no arm-twisting and I'm not expecting anyone on pocket money to contribute!

If you are a professional who has obtained the pdf, then your interest is a donation in itself and I would not expect researchers, consultants and other healthcare professionals to contribute – unless you wish to of course.

Alternatively, you might like to donate your own KS story for the second book (pdf), The KS Diaries (please see p80).

Humongous thanks.
Important Information

PLEASE READ

WARNING

The KS Story – You are not alone
contains frank and sensitive information of a gender & sexual nature, relating to Klinefelter’s Syndrome and the effects it has on an individual.

The Book (pdf)
NOT TAKING ADVANTAGE
The purpose of creating this book is to supply pure information about Klinefelter’s Syndrome and to point you in the right direction to reputable information, advice and support resources.

Other than specific contributors, credited and referenced within The KS Story and links to two free search engines (included for the convenience of the reader), this has been done without commercialism or personal gain.

To this end, no named products will be associated with, or promoted within The KS Story.

If you wish specific information on a product, please contact an advice or support group, or search for specific products online.

Disclaimer Please see page 86.

Further Study (contact listings)

CONTACT DETAILS
Please be assured that within the Further Study chapter all contributors have supplied up-to-date [2010] contact information.

Some contact details, email or web addresses are likely to change before the book pdf is updated in November 2012.

DIFFERING VIEWPOINTS
While there may be differing views within the general population and some support groups, in what Klinefelter’s Syndrome is, or isn’t, (e.g. Intersex), all contributors have approved (in writing) their contact details under a specific category heading.

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The KS Story –

Family & Friends

Chauffeur, welfare visits, food parcels and family gatherings. Just a few of the unselfish acts my mother, sister and brother-in-law have provided in order to ease my passage through the last 14 years of pain, stress and uncertainty. Of course their love and support runs far deeper.

There are no words to express what they mean to me, no words to thank them for their continuing support and understanding. That said, if I can help just one child with Klinefelter’s Syndrome, or the parents of a child or teenager with this condition, perhaps that would go some way.

Family acknowledgments would not be complete without mentioning my father. He died in 1980 after many years of heart disease and 17 years before I was diagnosed with Klinefelter’s Syndrome. With only the traits & symptoms of KS apparent to him our relationship was ‘strained’ at times, but despite our differences I think of him with love.

I am rich in relatives and delight in our evolving love and friendship. I am also grateful for their support and understanding. That said, if I can help just one child with Klinefelter’s Syndrome, or the parents of a child or teenager with this condition, perhaps that would go some way.

I am in relatives and delight in our evolving love and friendship. I am also grateful for their support during recent cancer treatment (2009/10).

I am fortunate to have many good friends, evolving from holidays on the white stuff, Munro-bagging, camping trips and annual BBQs of note.

Staff at the former Princess Margaret Rose Orthopaedic Hospital and the New Royal Infirmary of Edinburgh, 2001 & 2003. Staff at Hay Lodge Health Centre (The Practice); for allowing me to freely moon once a month (Male HRT), Hay Lodge Hospital (WHS Trust), for 6 years of care; on the couch and scraped off the ceiling! Ward 1 for their healing hands and unique humour. Medical & surgical staff at Borders General Hospital and the Borders Macmillan Centre (BGH), for their outstanding bedside manner and interest in The KS Story. 2007-08 & 2009-10.

‘The KS Story’ Advisors

My sincere thanks to the following research advisors for their invaluable and enthusiastic assistance, user-friendliness and patience:

Prof Milton Diamond, University of Hawaii, John A Burns School of Medicine, USA; Prof Patricia A Jacobs OBE, Wessex Regional Genetics Laboratory, UK; Dr Shirley G Ratcliffe, Consultant Paediatrician (Retired), UK; Prof Anne Fausto-Sterling, Biology & Gender Studies, Brown University, USA; Prof Dick F Swaab, The Netherlands Institute for Neuroscience, NL.

Unsung Heroes & Heroines

A huge THANK YOU to the numerous Klinefelter’s Syndrome information and support groups and individuals, and allied organisations in the UK and worldwide who have assisted me with facts and figures for the book. They truly are the heroes & heroines of this book.

Healthcare Professionals

Very special thanks to the following healthcare professionals who have treated me over the last 14 years. Also for reviewing The KS Story; making valuable suggestions and pleasing noises.

Staff at the former Princess Margaret Rose Orthopaedic Hospital and the New Royal Infirmary of Edinburgh, 2001 & 2003. Staff at Hay Lodge Health Centre (The Practice); for allowing me to freely moon once a month (Male HRT), Hay Lodge Hospital (WHS Trust), for 6 years of care; on the couch and scraped off the ceiling! Ward 1 for their healing hands and unique humour. Medical & surgical staff at Borders General Hospital and the Borders Macmillan Centre (BGH), for their outstanding bedside manner and interest in The KS Story. 2007-08 & 2009-10.

Last and never least

My GP, Dr P T Young, for helping me get through the pain and stress of the last decade and for his faith and support in my efforts to create The KS Story.

References within text

Throughout this guide there are Acknowledgments to the many contributors who have taken time out from their furious schedules to substantially assist a mere mortal with his studies. These people have been referenced within the specific chapter margins. References for main advisors are shown below, with small red initials within the appropriate chapter text (e.g. PJ).

If appropriate, contact details and published work for the following people can be found within the Further Study chapter (p74-79).

I would never knowingly plagiarise someone else’s work. With such a large body of information and with an appaling memory, despite making every effort to check my work, there may be information taken from a source from which I have forgotten to reference/credit. To those concerned I sincerely apologise and thank them for their wisdom.

MC Cull, M (AHN Founder), Adrenal Hyperplasia Network, UK.
MD Diamond, M (Professor), University of Hawaii, John A Burns School of Medicine, USA.
AF-S Fausto-Sterling, A (Professor of Biology & Gender Studies), Brown University, Rhode Island, USA.
LG Gooren, LJ G (Professor), Free University Hospital, Amsterdam, NL.
PJ Jacobs, PA (Professor), Wessex Regional Genetics Laboratory, Salisbury, UK.
SR Ratcliffe, S G (Consultant Paediatrician) retired. Previously at MRC Genetics Unit, Edinburgh, UK.
MS Simmonds, M Androgen Insensitivity Syndrome Support Group, Banbury, UK.
DS Swaab, D F (Professor), Netherlands Institute for Neuroscience, Amsterdam, NL.

Steroid Hormones

The mass of the steroid hormones are sufficiently small enough (lipophilic) to pass easily from blood to brain tissue.

Steroid hormones bind with steroid receptors in the cytoplasm of cells in order to regulate gene expression (switch-on). The resultant processes translocate to the cell nucleus Kawata, CF. (1999).

Cholesterol is the precursor for the five major steroid hormones: Androgens, Oestrogens, Progestins, Glucocorticoids and Mineralocorticoids.

The testes secretes three gonadal steroid hormones known collectively as Androgens, including Testosterone, Dihydrotestosterone (DHT) and Androsterone – testosterone being the most important.

There are also three Oestrogens: Oestriodiol, Oestrone and Oestriol. By far the most important of these is oestriodiol, the oestragenic potency of which is 12 times oestrone and 80 times oestradiol. Oestradiol is also the female equivalent of testosterone in males.

Androgens are also secreted in small amounts by the ovaries and adrenal cortex, and oestrogens by the latter, testses and placenta.

Recent studies show sex hormones & receptors exert many actions beyond reproduction, such as: areas of the brain to do with learning and memory, emotions, motor coordination and pain sensitivity.

McEwan, (1999), Behaviour, mood, cognitive function. Also functions which could have important neuropsychiatric and neurodegenerative diseases.

Information overload! – just dabble

The KS Story is two things. Primarily it is a conduit to the various support and scientific experts, but to get you started it also offers an overview of both Klinefelter’s Syndrome and conditions associated with it. Within these pages there is a great deal of information to take in, but the book isn’t a novel; you don’t have to read it from cover to cover. Treat it as a reference book, just taking what you need, that way hopefully you’ll retain more information and you won’t get bogged down in chapters that don’t interest you. You can then contact those groups within the Further Study chapter which can assist with your specific needs. M1

Please read the margin box on the opposite page, titled ‘Important’.

Complexity and Study

Klinefelter’s Syndrome is a highly complex condition and cannot be generalised – everyone is different. As too are the different characteristics of a person (phenotypes/karyotypes), which are also complex and varied. (Further details see p22-23).

The KS Story deals with Klinefelter’s Syndrome generally as a whole entity, though there is a bias towards 47,XXY in some chapters; basically because it’s my own karyotype and the one I studied the most. As mentioned, this book is an overview of KS, so if you have another karyotype it should still be of interest to you.

You can also go to the Further Study chapter, where you will find contacts and websites to support and research groups who can offer you a tremendous amount of information, on ALL karyotypes. These specialists will help you to understand more fully your own particular condition, your symptoms and your feelings, and if you wish, will put you in touch with other KS individuals or families.

Copyright and Copying ‘The KS Story’

You have permission to download the whole pdf or print off specific chapters or pages, or make copies of the pdf or photocopies of specific pages for your own studies; to give to family members, spouse/partner, or to your endocrinologist or GP (MD).

If you are a healthcare professional; counsellor; anyone who works with children or young adults etc., you have permission to copy pdfs or pages for yourself and colleagues.

It is strictly forbidden to make pdfs or photocopies for commercial use or monetary gain.

Unpronounceable mouthfuls

Throughout the guide you will come across new and unfamiliar names and procedures. Don’t be put off by them. If you wish to know more about your condition it will help if you can understand the meaning behind these unpronounceable mouthfuls. And it’s amazing how quickly they just roll of the tongue – honest!

If you plan to study your condition more fully, then I highly recommend the purchase of a good concise general and medical dictionary. Believe me, they are invaluable. If you can’t stretch to the latter, your local library should be able to loan you the books. Alternatively, you could check out any definitions online. M2

Little red references

Throughout this guide you will see a few little red letters and numbers within the text, which relate to additional information, or to original source references. For example:

M3 additional information found within page margins;
MD reference (see p11).

Dictionaries and many medical papers continue to use the words ‘normal’, ‘disorder’, ‘defective’, ‘abnormal’. I’m not abnormal or defective! Within the context of describing Klinefelter’s Syndrome, I have tried to use certain guidelines, laid out to eliminate some of these offensive words. For example, for the word ‘normal’ the words ‘typical’, ‘usual’ or ‘most frequent’ have been used, and ‘condition’ rather than ‘disorder’. Of course there are times when this is not possible, e.g. within a support group name, or when quoting someone.

Who comes first?

So as not to offend anyone; wherever possible, alphabetical order is used or for that matter desired or required by all. There should be sufficient to help you understand the basics and point you in the right direction for further study.
Who should read this book?

There are six groups who hopefully will benefit from reading and using this book.

One Typical kids can seem unwilling to listen and learn and to parents may be seen as simply bone idle. But in some the cause is genetic and is not a fault of the child.

Everyone’s situation is different and sometimes due to embarrassment or a lack of knowledge, there may be an unwillingness by some parents to offer sex education and advice to their children; leaving it primarily in the hands of schools and peer groups. It then becomes doubly difficult for both sides to communicate about the symptoms and feelings of typical teenagers—never mind those with Klinefelter’s Syndrome (KS). With the result that those individuals with KS can go through their adolescence and early adulthood feeling alone and confused; not knowing who to turn to for support.

This guide is primarily offered to those individuals who exhibit some of the KS traits shown on page 22, but who are perhaps too shy to express their concerns and feelings to parents, spouse/partner, or to their GP. From experience, being a generally shy child and teenager, I was unwilling to share my sexual feelings and physical lackings with anyone, particularly to my parents, and certainly not to a family doctor. But then attitudes change with the generations; today’s parents are far more open-minded and knowledgeable of some genetic conditions and GPs too are much more approachable.

Two Following on closely to the latter are those individuals who have just been diagnosed with Klinefelter’s Syndrome and are interested in knowing more about their condition.

It is often the case that if you know that there are others out there with the same condition; who have gone through the same experiences as yourself, it somehow eases the loneliness. And when you discover that these people are willing to listen to you and offer support and guidance; and these are individuals with KS, as well as parents of KS children, it can really lift your spirits.

Three Those with KS will have their own set of circumstances regarding the ‘telling’ of their physical differences and emotions.

The third group who should benefit from reading this book are therefore the parents, family members spouse or partner of individuals with KS. Hopefully it will help them to understand a little better what’s going on in the mind and body of their loved ones.

Four I have tremendous respect for the work all medical personnel do. All of us know the pressures that our local frontline doctors and medical staff are under. It must be hard enough keeping up with all the usual diseases and conditions, and while I’m sure a good number of consultants and GPs know about Klinefelter’s Syndrome, because of the complex nature of KS, (from experience), I have come across many hospital staff, including those working in associated fields, who had never heard of it, or needed ‘reminding’ of some of the finer points.

Five This book may also be useful to family planning counsellors, teachers, nursery staff and other people involved with children and young adults. And of course to anyone interested in understanding this “common genetic condition” in males.

Six With such a complex condition, within this guide I generally treat Klinefelter’s Syndrome as a whole entity. That said, I do highlight 47,XXY, being the one that most people seem to mention and the one which is perhaps the easiest to explain how it occurs.

Many of the karyotypes that until very recently were considered part of the KS ‘family’ are now seen as syndromes in their own right. The Further Study chapter (p70) will help you find specific information and support for your particular karyotype, or offer contacts who may be able to direct you. It also includes a range of linked and sex-determining conditions.

Interestingly, Triple X Syndrome, karyotype: 47,XXX (often called the female version of KS) which is also a result of nondisjunction (see p37), 47,XXY Syndrome and Klinefelter’s Syndrome are often grouped together within research studies. (Please see Karyotypes p23 and Further Study for more information).

And finally If you take only one piece of advice from this book – make it a visit to your family doctor, or to an endocrinologist. Why? See p61.

Of course, there will also be those who never tell. But it should be remembered that none of us have any say in how our blueprint creates us. So why should anyone suffer for a condition over which they have no control?

Within the research statistics and complex medical data lies someone’s child. Above all else, don’t they deserve the same love and support as any person? Klinefelter’s Syndrome, though genetic, is not hereditary, so can’t be predetermined, but if something doesn’t seem right, perhaps for an older couple trying for a family, or to parents worried about their son, knowing some of the KS symptoms might just be beneficial.

Klinefelter’s Syndrome deserves a fair chance at life; to be able to fulfil their ambitions and to contribute to the world around them.

KS creates vastly different traits and symptoms, even within the same karyotype. Medically, some do suffer, some have mind implications, while many have no symptoms at all and can go through life unaware that they have KS, only finding out if they have infertility tests or another linked condition. (See p22 & 58).

According to one study in 2000 there were some obstetric units that had no established protocols for health professionals on how parents should be told about prenatal diagnosis. There were also some obstetric staff who knew very little about the effects of sex chromosome anomalies and when first informed, some parents were given misleading information. Of course this situation may have changed since 2000 – but I can’t stress enough how vital it is that you should NOT consider termination until you have talked to a specialist genetic counsellor and ideally a Klinefelter’s Syndrome support group; who can give you advice, support and firsthand knowledge of growing up with this condition.

At the end of the day, having knowledgeable parents who can love, encourage and support their child through the difficult years of life; such as puberty and adolescence, will make a difference beyond measure.

---

1 Abramsky, L. et al. (2000). What parents are told after prenatal diagnosis of sex chromosome abnormality: interview & questionnaire study. North Thames Perinatal Health Unit, Imperial College of Science, Technology & Medicine, Harrow and Psychology & Genetics Research Group, Guy’s, King’s College and St Thomas’s Hospital School of Medicine, London.
What is Klinefelter’s Syndrome?

The basics

Every human adult contains more than one hundred million million cells (each averaging one hundredth of a millimetre in diameter). The cells vary enormously in shape and size depending on their job within the body, e.g. muscle, nerve, organ, blood etc.

However, they all have the same basic content (see cell diagram on p40). Within the wall of the cell is the cytoplasm, a jelly-like substance and embedded in this is the nucleus. Think of the cytoplasm as the factory floor, producing products for the body, and the components of the nucleus as the managers; issuing instructions to the workforce.

The blueprints relating to specific products are stored within the nucleus in a chemical form (or code), called Deoxyribonucleic Acid (DNA). Organised into groups along a ladder-shaped structure, it makes up the majority of each chromosome (see p41).

The total blueprint to create a typical human are stored on 46 such chromosomes, arranged in 23 pairs, in each cell of the body.

A typical female and male have 46 chromosomes; 23 maternal and 23 paternal (paired), with the last pair (X and Y) determining the sex of the individual, written as: 46,XX and 46,XY respectively.

What causes Klinefelter’s Syndrome?

In life variations happen and sometimes an egg cell (ovum) or sperm cell (spermatozoon) develops which carries one or more extra X chromosomes. Although the cause is currently unknown, it results primarily in a change occurring during meiosis; a process which is undertaken by all cells destined to be sperm or ovum. In this process the 46 chromosomes in the cell divide to make four new cells with 23 chromosomes each.

During meiosis chromosomes pair with their corresponding chromosomes and exchange bits of genetic information; this is called ‘Crossing over.’ In males, the X and Y chromosomes pair and in females, the X chromosomes from each parent pair. After the exchange the chromosomes separate and meiosis continues.

In some cases the X and Y chromosomes or the two X chromosomes fail to pair and exchange genetic material, resulting in them moving independently to the same cell; producing a sperm with both an X and Y chromosome or an ovum with two Xs. This is called ‘Nondisjunction.’ (See table to right).

When a sperm with an X and Y chromosome fertilises an ovum with a single X chromosome, or a typical sperm with a Y chromosome fertilises an ovum with two Xs, an XXY child is conceived. So instead of 46,XY a male with Klinefelter’s Syndrome has 47,XXY. (See diagram on p41).

An extra X can also occur in females – 47,XXX (‘Triple X Syndrome’), or an extra Y in boys (i.e. 47,XYY). Additional Xs and Ys and combinations of the two can also occur. (See karyotype table on p23 and Human Reproduction on p42).

Klinefelter’s Syndrome (karyotype: 47,XXY) is classed as a “common genetic condition” in males. A study in 2007 now shows KS at 1.72 per 1000 male infants (previously 1.3).

It has also been shown that there are 400-500 live male KS births in Britain each year, “though many will not be recognised until adulthood.”SR

As mentioned previously, because of the variable nature of KS, some individuals may go through life blissfully unaware of their condition, others may suffer in silence; never knowing why they are that way, or to be shunned and rejected. Many only find out they have KS if they have medical tests; such as for infertility or associated conditions such as osteoporosis.

It is preconceived by many parents that their son is gay, because he is not interested in girls, when often it is shyness and possible rejection. Research currently shows no more links to homosexuality than is in the general population.

And finally

Amongst other vital components, such as proteins and ATP, chromosomes carry our DNA, which passes on hereditary traits and controls how our body is made and how it functions.

Although classed as a ‘genetic’ condition; because of the additional chromosome(s), KS itself is not an hereditary condition. KS is not passed down from previous generations.

REFERENCES p11 CONTACTS p74
That said, although a large percentage of XXY males do not produce enough sperm to allow them to become fathers, you should not automatically assume you are infertile without further testing. There is evidence which points to a substantial number of XXY males having the ability to father a child.

It should also be noted that a father with Klinefelter's Syndrome may transfer his 47,XXY karyotype to his naturally conceived offspring. If this course of action is chosen, like Intracytoplasmic Sperm Injection (ICSI); counselling should be seriously considered.

(See the ICSI chapter p48, which also includes information on specific prenatal diagnosis and screening procedures).

KS or not KS – that is the question

It seems that karyotype 47,XXY isn’t necessarily Klinefelter’s Syndrome!

Up to now all support and research groups have acknowledged 47,XXY as Klinefelter’s Syndrome. In fact some include XXXY & XXXY to XXXXY.

Now there are some people in the USA that have proposed to call the most common karyotype 47,XXY ‘Klinefelter Disease’ and all the other variants: ‘Klinefelter Syndrome.’

However, other scientists including Prof M Diamond, (John A Burns School of Medicine, University of Hawaii) strongly discourage the use of the word ‘disease’ as “it just adds to the stigma.”

According to a Canadian KS website, if an individual has been diagnosed with an additional X chromosome, but exhibits none of the symptoms, then they do not have Klinefelter Syndrome.

“I never refer to newborn babies as having Klinefelter’s, because they do not have the syndrome.”

Dr Robinson, A. MD, Paediatrics University of Colorado Medical School, USA, and the director of the NICHD-sponsored study into 47,XXY.

The latter quote is mentioned in a paper written by Bock, R. (1993), National Institute of Child Health & Human Development (NICHD).

The paper (13 pages long), does offer a balanced view.

Dr Klinefelter said that, “this was really another of Dr Albright’s diseases. He unselfishly allowed my name to come first on the list of authors.”

Following his time with Prof Albright, Dr Klinefelter served 3 years in the US army during WW2, advancing from First Lieutenant to Major. He then returned to private practice and a member of the faculty at Johns Hopkins University, becoming Associate Professor of Medicine in 1965.

He devoted the rest of his life to studying alcoholism, endocrinology and clinical research in rheumatology, retiring in 1988 at the age of 76. He died in 1990.

Professor Fuller Albright...
The KS Story – You are not alone

Fuller Albright (1900-1969)

Fuller Albright graduated from Harvard Medical School in 1924. In the late 20s he worked with Dr Ellsworth at Johns Hopkins University, before moving to Berlin to work with Dr Zondek, mostly in pituitary gland research.

On returning to the States he set up a kidney stone clinic, and an endocrinology clinic at the Massachusetts General Hospital, specialising in ovarian dysfunction.

Dr Albright discovered many mechanisms of endocrine diseases; mostly in calcium metabolism.

He developed Parkinson’s Disease around 1946 and although he continued to work, in his last years each month a medical student was assigned to him, to take him around the hospital and to learn from him.

He died in 1969 following a neurological attempt to correct the Parkinson’s Disease in 1956.

1949

M L Barr (1908-1995)

Murray L Barr was born in Belmont, Ontario, Canada. He gained his BA in 1930 and MD in 1933 from the University of Western Ontario.

After working as a GP for two years, he returned to the Anatomy Department, specialising in Neurology.

After the Second World War Dr Barr returned to the same university were in 1949 he and a graduate student, Ewart G Bertram began a study into the process of fatigue in the cells of the nervous system.

While examining the sample brain cells (neurons) of their animal specimens, they discovered dark blotches, which looked like a drumstick. It was later discovered that the marks were ‘clumps’ of chromatin.

More surprising, was that when Bertam’s detailed notes were examined they discovered that all the marks came from females.

In 1949 they published their work in a magazine publication of the Royal Canadian Air Force (their funding partners), on the sex chromatin, which later became known as ‘Bertram & Barr Bodies’ or just ‘Barr Bodies’.

In 1951 Dr Barr became Professor of Anatomy.

The Klinefelter’s Syndrome connection came in 1956. Buccal smear tests of men with Congenital Testicular Hypoplasia were reported as having a positive X-chromatin Barr Body.

Under typical circumstances, if an individual has an XX karyotype, one X is put to work while the other is put aside as a clump of chromatin. A woman therefore has a positive X-chromatin, or Barr Body, while a man; who typically has an XY karyotype, does not.

Therefore, the conclusion was, to theorise, that there was an extra X chromosome present in individuals that exhibited the symptoms of Klinefelter’s Syndrome.

1959

Patricia A Jacobs OBE (1934-)

“A CASE OF HUMAN INTERSEXUALITY HAVING A POSSIBLE SEX-DETERMINING MECHANISM.”

Professor Patricia A Jacobs’ first exposure to cytogenetics was in 1955, and except for a brief study by the Praying Mantis and a goat-sheep hybrid; called a ‘Grep’, she has spent her entire career in the study of the human chromosome.

In 1957 Dr Jacobs was appointed to the scientific staff of the newly created Medical Research Council’s group in Edinburgh, by the director Michael Court Brown, for research into general effects of radiation; to study the chromosomes of leukemic cells.

Dr Jacobs spent 4 months at Harwell and Oxford learning techniques of mammalian cytogenetics and culturing human bone marrow, from Dr Charles Ford and Dr Lazlo Lajtha respectively.

With few leukemic patients back in Edinburgh, in 1958, fired up with enthusiasm Dr Jacobs looked for ways to use her newly acquired skills for examining human chromosomes. It was then that she was offered a marrow sample from Dr John Strong, a local endocrinologist. The sample was from a chromatin-positive man with Klinefelter’s Syndrome, and in 1959, with the technical assistance of Miss Muriel Brunton, went on to describe her first sex-chromosome abnormality in humans.

With few leukemic patients back in Edinburgh, in 1958, fired up with enthusiasm Dr Jacobs looked for ways to use her newly acquired skills for examining human chromosomes. It was then that she was offered a marrow sample from Dr John Strong, a local endocrinologist. The sample was from a chromatin-positive man with Klinefelter’s Syndrome, and in 1959, with the technical assistance of Miss Muriel Brunton, went on to describe her first sex-chromosome abnormality in humans.

Immediately recognising the importance of their observations Dr Court Brown encouraged Dr Jacobs to publish a note in Nature (1959; 183: 302-03), (as title), by Patricia A Jacobs and Dr J A Strong. Dr Jacobs was 24.

1959 continued to be an outstanding year with the published work of J Lejeune et al., of an additional chromosome 21 (Down’s Syndrome), and C E Ford et al., regarding the chromatin-negative female; karyotype 45,X (Turner’s Syndrome). That year Dr Jacobs also described the first female with a 47,XXX karyotype. And in 1964 karyotype 48,XXYY was described.

In 1965, following a controversial period regarding studies of XYY patients, both in the UK and US, Dr Jacobs left Edinburgh to be with her American husband and to start a new chapter in her life; taking up an appointment at the newly established John A Burns School of Medicine, in the Department of Anatomy & Reproductive Biology, in Honolulu, Hawaii, studying spontaneous abortions linked to chromosome abnormalities, employing the previously unused, and more precise banding techniques.

Back in Britain, Prof Patricia Jacobs was made a Fellow of the Royal Society in 1993 and was awarded the OBE in 1999.

She continues to work with her beloved chromosomes and with the same enthusiasm for cytogenetics as she had back in the late 50s.

These are just some of the people who pioneered cytogenetics in the 20th Century, and the story continues today, with new advances in the understanding of human chromosomes and their function and effect within the body.

REFERENCES

11. The aforementioned text was kindly supplied by Prof PA Jacobs.

12. Personal text is based on reprints kindly supplied by Prof PA Jacobs.

M1. The aforementioned text was based on information written by Haché D, which was supplied to the support group Klinefelter Syndrome and Associates, USA.

M2. The original Bertram & Barr paper was published in a Royal Canadian Air Force magazine.

Additional information from:


The Lancet, supplied by Dr S G Ratcliffe.
Klinefelter's Syndrome is a variable condition. Some people are seriously affected, while others have mild symptoms.

Behavioral & Personality traits seen more in individuals with Klinefelter's Syndrome (KS):

- Concentration difficulties
- Delayed speech development
- Emotional immaturity
- Impaired gross motor coordination in early childhood
- Impaired memory, particularly short-term
- Increased dependency
- Increased frequency of speech disorders
- Lack of self-assertion in early childhood
- Low self-esteem
- Lowered endurance
- Lowered I.Q., particularly verbal
- Lowered level of attention
- Lowered vigour and drive
- Mood swings
- Passivity
- Poor auditory discrimination
- Poor contact ability and loneliness
- Preference for quiet games
- Shyness & reticence

**Klinefelter's Syndrome**

- 47,XXY
- 48,XXYY
- 49,XXXYY
- 50,XXXXX

**Triple X Syndrome**

- 47,XXX
- 46,XXYY

**Mosaics**

- 46,XX/47,XYY
- 46,XX/47,XXX

**Other Karyotypes**

- 47,XXY (Male)
- 47,XXY (Female)
- 48,XXX (Male)
- 47,XXXXY

Please contact the nearest support or research centre for detailed information. A website has been established for this purpose.

**46,XX Male (De La Chapelle Syndrome)**

Sometimes at early egg formation the SRY gene (Sex determining Region of the Y chromosome) attach themselves to an X chromosome, creating an XX male. 46,XX Males have a similar phenotype to 47,XXY (KS) but there is no increase in height as appears with the latter.

**47,XXY Female**

There has been a case in Germany of an SRY-negative 47,XXY woman, with a son and two daughters (one daughter also has the 47,XXY karyotype).

**Mosaic**

In a Mosaic person different cells have different sex chromosomes; i.e. 46,XY/47,XXY has typical male sex chromosomes in one cell but in another, KS chromosomes. There are some extremely complex mosaic karyotypes, such as 45,X/46,XXY/47,XXXYY, (i.e. Turner’s Syndrome/typical female/Tricle X Syndrome).

**REFERENCES**

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Endocrinology

Hormone replacement therapy

At your first endocrine appointment you will be examined and the consultant will discuss your particular treatment. Everyone will be different in how they are looked after.

The consultant will probably suggest that you obtain a density scan, to evaluate the condition of your bones. The DEXA scan is a simple, non-invasive procedure, similar to a film-producing x-ray, i.e. no tunnels, and you even get to laze for 15 minutes while it checks you out!

Following that, you may be placed on an osteoporosis therapy to boost your bone density. This consists of a course of tablets, taken daily for a minimum of three years, possibly indefinitely. However, as it becomes part of your daily routine you will not find it a chore. The tablets periodically stop bone breakdown, as well as increase your calcium intake. You may be offered some other treatment.

Next comes the hormone replacement therapy. There are several ways to administer the treatment nowadays, and your GP or endocrinologist will advise you on this, as some will not be suitable for everyone.

Please note. The following therapies should only be taken with a diagnosis and support from your endocrinologist or doctor, who will be able to offer you more details and suitability for these and newer methods of treatment. For instance, some products have not been tested on males less than 18, or limited testing on older males (e.g. 65 upwards).

Additionally, some therapy products are only licensed for certain countries.

There are other serious health issues to consider before treatment begins, which will be outlined to you by your GP (MD) or endocrinologist. You also have to ‘work’ with your healthcare professional; keeping them informed of initial and later side effects.

Additionally, testosterone should only be used by males, as masculising will result in women, with changes in menstrual cycles. It will also seriously affect the foetus of pregnant women.

I have included here support group reports on each item’s effectiveness, including my own experiences (italics), but at the end of the day everyone is different and will experience differing results. In discussion with your consultant, it’s up to you to find what suits you best.

Check out the KSA and KO support group websites, in the UK and KS support groups in your own country, for a full list of products and information relating to each.

Gel: This method is set to revolutionise the way some people take testosterone and one which will delight many who miss treatment, or have never begun it, perhaps because they dislike the needle.

Supplied in a pump canister, sachets or a tube, the gel is rubbed into the skin of the upper arm or belly, waiting a few minutes until absorbed before dressing.

Important information to remember:
• wash hands thoroughly with soap and water after applying the gel as it can be transferred to others.
• if having sexual intercourse within a few hours of applying the gel, then you are advised to wear a t-shirt, to protect your partner.

SG report: Because it is so new there is little feedback on use. Like patches the gel may cause aggravation to your skin.

Implant: This consists of a visit to an out-patients clinic every 6 months, for this gradual release alternative. If you prefer this option talk to your consultant who will give you all the facts. M1/M2

SG report: one person reported that the implant forced its way back out, causing round scars.

Contd.

M1 Support Group (SG) report
Based on information from the Klinefelter’s Syndrome Association website. w: www.ksa-uk.co.uk
(See p74 for contact details).

M2 Support Group report
Based on information from the Klinefelter Organisation w: www.klinefelter.org.uk
(See p74 for contact details).

REFERENCES p11 CONTACTS p74
**Injections:** There are now two versions of the deep intramuscular injection. The original jab with an application varying from three weeks to monthly (though it can be more often), and a newer one just taken every three months (10-14 weeks).

Sometimes known as a depot injection, the drug itself is injected into your rump (upper buttocks) and forms a reservoir of testosterone, which will be absorbed over the stated application period. Injections can be given by your GP or practice nurse to ensure you don’t forget your dosage. It takes only seconds and you’re done. Some GPs offer self injection, while other people prefer the professional touch.

**Trust me — The needles are so sharp nowadays that most times I feel only a little pressure on my rump. Sometimes not even that. Occasionally there is a wee nip, and I mean a wee nip... LOOK, IF A WIMP LIKE ME CAN DO IT, ANYONE CAN!**

There are mixed reports on the monthly/three week distribution. Some groups report good results, while others find horrendous libido peaks.

For the first few years after commencing hormone treatment I generally found a strong peak about a week after the jab; which resulted in about 7-14 days of hyperactivity (if you get my drift); when the least stimulus set me off... M3

Then for about a year it very slowly began to level out. 13 years on, throughout the entire month I drift through a kind of generalised medium-grade haze of sexual awareness. Perhaps my brain has got used to the hormone, or perhaps I’ve finally departed my second puberty, ready to responsibly face society – again!

**Patch:** This is a transdermal drug delivery system, which you will have to replace daily. It comes in various dose strengths. Higher body weight can also affect daily dose.

This therapy may not have been tested with children. Discuss other options with your doctor or endocrinologist.

**SG report:** Complications usually take the form of skin reactions and difficulty sleeping with the rustling noise it makes. M1

I tried them a few years ago to see if there was a better hormone balance. The spread was good with no recognisable peaks, however there are few places to position the patches and if you get them on a hairy site — well you know what it’s like removing a sticky plaster where there are hairs — imagine a three inch square plaster!

In my case hairless days are over, 13 years of male HRT have seen to that, so I found I was placing them back on the same place, with resulting irritation to my eczema. Also the patch was made of crackly plastic that drove me nuts; like sleeping on a packet of crisps! And so after 3 weeks of perseverance I gleefully returned to the injections!

That said, if you are new to replacement therapy, and are clear of hair, give them a try.

**Tablet:** You will have to remember to take them as prescribed. Again, this type of therapy may not have been tested with children.

**SG report:** Some people have felt a little depressed as the dose wears off. M1

**Tablet (buccal sustained release):** A very small tablet-like product is placed between your gum and lip, just above one of your upper front teeth, which then becomes gel-like and sticks to the surrounding tissue. It’s called a ‘buccal sustained release system’, as once placed, the tablet gradually releases testosterone which is absorbed directly into your bloodstream. So don’t chew or swallow the tablet.

Dosage is twice daily (12 hours apart). You will need to periodically check to make sure the tablet is still in place, such as after eating, brushing your teeth or using a mouthwash.

There are a number of side effects with this method of application, so let your GP know if you experience anything unusual.
Hormones – the chemical messengers

There are two types of gland within the body. Endocrine glands, which produce (secrete) substances directly into the bloodstream, which then transports them around the body to ‘target glands’. These are called ‘Ductless Glands.’

The other type is the Exocrine gland: which employs ducts to direct their products into the area where they are required, (i.e. salivary glands).

But for this chapter, and for your understanding (hopefully) of Klinefelter’s Syndrome it is the secretions of the Endocrine System that are important here.

The latter secretions are called ‘Hormones’, and can be thought of as chemical messengers; sending instructions all around the body.

Hormones are produced in minute quantities yet their effect on the body can be profound and sometimes long lasting.

A few endocrine glands control themselves, but the majority are controlled by the Pituitary gland; which for this reason is called the ‘Master Gland’ of the endocrine system. This pea-sized body is located in a bony cavity at the base of the skull, just under the Hypothalamus to which it is connected.

The pituitary gland can regulate the production of hormones within a particular target gland (TG), by producing a ‘Stimulating Hormone.’ These stimulating hormones are regulated by specific ‘Releasing Hormone’ which is produced in the Hypothalamus.

The stimulating hormone ‘calls’ the TG to produce its particular hormone. If too much hormone is produced it sets up a ‘negative feedback response’ to the pituitary gland, which produces less of the stimulating hormone, and so the TG produces less of its hormone.

Several glands are controlled in this way, including the testes. (See diagram).

The Endocrine glands

Adrenal gland: Situated on top of each kidney and produces the hormone adrenaline. This prepares the body for ‘fight or flight’ by raising blood pressure; increasing heart-beat and breathing rates; increases the amount of glucose released from the liver and increases the amount of blood to the muscles and reducing the amount to the gut.

Pancreas: Situated below the stomach, one part produces digestive juices, while the ‘Islets of Langerhans cells’ produce the hormones glucagon, somatostatin and insulin, the latter of which increases the rate that the liver releases glucose into the blood; enabling cells to absorb glucose and stimulates the body to change glucose into fat.

If too little insulin is produced the liver releases too much glucose, causing the condition ‘sugar diabetes’ (Diabetes Mellitus · type 1 & 2). This causes the glucose in the blood to increase to a dangerous level.

Parathyroid gland: Four small glands embedded within the thyroid. Parathyroids are sensitive to calcium levels in the blood. If there is too little, the gland releases the hormone parathormone which increases calcium absorption in the intestine; withdraws calcium from the bones and increases re-absorption of calcium by the kidney tubules.

Pituitary gland: Produces hormones which control growth. Other hormones cause the ovaries to release ova (eggs); testes to produce spermatozoa (sperm); the uterus to contract and expel the fetus at birth; and the mammary glands (breasts) to produce milk. Other hormones control the amount of water in urine and the activities of other endocrine glands.

Placenta: Apart from its main function within the uterus this organ also acts as an endocrine gland secreting Progesterone, oestrogen and Chorionic Gonadotrophin, which regulates the maintenance of pregnancy.

Thyroid gland: Situated in the neck in front of the trachea (wind-pipe). It produces the hormone thyroxine, which has a major influence on the mental and physical development after birth, by controlling the rate of chemical reactions in all body cells.

Ovaries: Two organs situated in the lower abdomen, below each kidney. They produce ova (eggs). The ovaries also produce the hormone called Oestrogen which controls the development of female secondary sexual characteristics (breasts, soft skin, female voice); prepares the uterus (womb) to receive a fertilised ovum; stimulates the uterus to protect and nourish a developing baby.

Testes: Two organs situated in the groin within the scrotum and produce spermatozoa (sperm). The testes also produce the hormone Testosterone which controls the development of male secondary sexual characteristics (coarse skin, deeper voice and more body hair than in females).

REFERENCES p11 CONTACTS p74
1997 changed the way I viewed my life; past, present and future. It put a name to 45 years of fears and embarrassment, and to what I felt but could never tell anyone about. It was the year that I was diagnosed with Klinefelter’s Syndrome.

The punchline was somewhat lost on me however, as at the time I was undergoing hospital in-patient tests to discover the cause of acute pain and weakness in the quadriceps of both my legs. And with still no light on the latter problems, I certainly wasn’t expecting to be diagnosed with another medical condition.

Some weeks later on leaving my first visit to the endocrine out-patient clinic, having just discovered that a huge chunk of my life had been affected by this condition, I asked a junior doctor if there was any printed information on Klinefelter’s Syndrome. He handed me a small pamphlet titled: ‘Hypogonadism in Adult Men’, which to be honest confused me even more.

I thought to myself, call me insensitive, but having just been told I had Klinefelter’s Syndrome there was only ONE pamphlet available, amongst the piles of literature on other endocrine conditions – and what the hell was hypogonadism? Being my usual non-confrontational self; I thanked the doctor and left.

It wasn’t until a few months later; when I began to look into the background to KS that I realised the true extent of what it really means to have Klinefelter’s Syndrome.

Amongst the day-to-day business and social struggles and treatment for the crippling leg pain and allied stress, it had not dawned on me at that point, that my search to piece together a clearer picture of my condition had in fact set me on an academic course unparalleled in my life, and one which would last well over eleven years!

It also became clear as I progressed that Klinefelter’s Syndrome involved more than just endocrinology. There were many other fields involved, such as: clinical & surgical medicine, genetics, gender issues, rheumatology, orthopaedics and neurology.

You have to understand that much of my academic years were a disaster, so studying was going to create its own degree of challenge. But here I had a quest for information; something vitally important in my life, something that would help me understand my life and enable me to cope more fully with my symptoms.

As usual, my spelling and grammar went out the window and my English and medical dictionaries became constant companions, but it’s truly amazing what you can achieve when you’re interested in the subject you are learning about.

Of course, while I progressed with my studies into KS the various medical specialists were beavering away trying to discover the original leg condition, with x-rays, electromyography, a nerve induction velocity test and muscle biopsy – joy!

Unfortunately, for the first three years of my study into Klinefelter's Syndrome I had no access to the Internet. I would spend days in feverish letter writing, followed by weeks of non-activity, as I awaited replies. Then in 2001 I discovered that my local library offered Internet access, which transformed the way I studied, and the staff were so helpful.

Believe me, the Internet makes a humongous difference to the speed and ease of collecting information.

Eventually, in 2002 with family support I managed to purchase a secondhand computer upgrade — with a modem!

I guess there will be many individuals that on leaving the endocrine clinic for the first time will be quite happy to leave it at that. Everyone is different - but some (like myself) will want to know more and that is the primary reason for this book.

In my own case, I don’t agree with the adage that too much information turns you into some kind of hypochondriac. After so many years in the dark and a general nature for trying to understand how things work, I found that I was drawn to discover more. But again everyone is different.

Everyone will have their own set of circumstances regarding the ‘telling’ of their physical and/or emotional differences to parents, spouse/partner or to their family doctor. And of course if you have their support, study should be that much easier; as you won’t be resorting to covert tactics.

I plunged into my studies with the enquiring mind of a child learning about the world for the first time. After studying cells and chromosomes I found myself delving deeper; trying to understand DNA, then its components, gene replication, protein production, human reproduction, anatomy, orthopaedic surgical techniques...

For weeks I was obsessed with the mind-blowing design and beauty of the human body – an enthusiasm that exists to this day. Strangely, I can remember the ‘central subdivision of the bed nucleus of the stria terminalis’, but I keep forgetting to water my plants!

It would be fair to say that my grammar and spelling are appaling and in any other written work it would be strongly suggested that I “don’t give up the day job.” However, Klinefelter's Syndrome can affect academia big time, (see p22) that's my excuse and I'm sticking to it! Correcting my scribbles has probably only taken the edge off the worst of it.

Although I have been studying the many facets of Klinefelter's Syndrome for the past 12 years and sound like an expert, I'm not. I have only scratched the surface. It is the research & support groups and medical publications with whom I have obtained study information who are the stars.

Growing out of my own academic flaws and subsequent frustrations in trying to find and understand facts about my condition, my primary objective was to compile a book which would direct the reader to respected UK and international research, support and information resources.
Obviously there are many thousands of websites, papers, journals etc., which I have not yet accessed, but what is important is that with this book you now have a reasonable contact database to commence your own studies.

I have learnt a great deal from my own studies into Klinefelter’s Syndrome. It has put names to feelings and removed the fear of uncertainty. It has taught me the value of talking to and working with my GP and in the importance of seeking advice on endocrine and osteoporosis therapy. But above all, it has shown me that I didn’t have to suffer in silence; that there are others out there just like me, specialists who are ready to help and support. People who have KS, who know what I’m going through and lift my spirit with their honesty and compassion.

When no one knows
People see the ‘normal’ external persona of those with KS and particularly with regard to children, adults may think that their son is just hamming it. They don’t sense the physical and psychological maelstrom that some children might feel within.

Some people believe that because the national diagnosis figures are so low that it follows that the majority of males aren’t severe enough to warrant them seeking help. While there will be many who are not severe, from personal experience and from talking to individuals and support groups it is clear that from an early age many children with KS hide their physical differences and emotions.

With the hassle of everyday life and the trials and tribulations of all family members, without knowledge of KS, children may close off their feelings to those around them, usually because they think no one will believe them, they may be too embarrassed, or perhaps just don’t know how to express their feelings. And so throughout their life they live with their secret – or until they are eventually diagnosed.

Because I felt embarrassed about my chest proportions, since the age of about 10 or 11 I have always worn a top of some kind when in public—even on the beach. And no amount of comments such as “there are far worse bodies out there than yours”, would ever convince me to uncover. Stripping to the waist felt unnatural to me; more akin to a female going topless than a male.

From experience, at 10 years old, the thought of surgery (whether or not it was viable), was extremely frightening so I never let on that I had hard lumps beneath my areola, even though they were painful if prodded.

Quite often diagnoses of Klinefelter’s Syndrome is made in childhood or early teens as a result of discovering glandular tissue (gynaecomastia).

Puberty is a period of extreme physical and emotional change and stress for any child, whether or not they have KS. For some individuals with KS puberty can be a minefield of lies, embarrassment and loneliness. When lumps appear; whether gynaecomastia or fat deposits, or a son’s body shape doesn’t match up to that of their peers, it is unlikely they will confide in their parents. However, if a parent is overly concerned about their child, there are non-confrontational ways to discover whether there is an underlying reason; by seeking their GP’s advice.

It is important to look out for other traits; such as low self-esteem, passivity, inability to concentrate... (see p22).

You could also talk to other parents who have gone through the same thing. (Please see Further Study p74, for family related support groups).

When parents know
The latter family groups are of particular value when the situation arises where a son has been diagnosed in childhood, and it comes to the inevitable point when the parents have to decide how, or if, they should tell their child the truth.

When the truth is withheld, the child may think that their parents are hiding something from them, though sometimes holding back to a later age is more preferable; for instance, if the child is immature.

Some clinicians believe that when a boy is about 10 or 11 years old, parents should inform them that they have body cells which are slightly different from other people, and that is why they are attending hospital clinics. Then around 12-18, more detailed information can be given; including the fact that he might be sterile.

That said, parents know their child and at what age they can be told certain aspects of the condition. So it is vitally important that their child’s emotional maturity is evaluated before embarking on heavy-duty details. The scenario being that at 12 years old it could be so easy for him to casually pass on his ‘secret’ to a pal, only for them to thoughtlessly pass it on to someone else, mistaking ‘infertility’ for a ‘sexual disorder’, leading ultimately to possible bullying tactics.

So you can see the value of talking to other families and to professionals who can give you help and support.

Possible gender issues
I would like to say at this point that everyone is different and I talk from my own experience and stress that currently medical research sees no direct link between KS and gender dysphoria, though to gender support groups there are a good many people who exhibit both.

However, don’t automatically assume anything, until you get specialist advice.

Even having been diagnosed with Klinefelter’s Syndrome, there can still be other issues and powerful emotions which some will find hard to express – to anyone.

I am blessed with a loving and supportive family, yet through puberty and into adulthood, with no idea of KS, I was unable to open my feelings to them. I had physical differences, but I had something else – something so sensitive I said nothing to anyone until I was in my early fifties.

Since around puberty I have felt partially feminine. It’s hard to express the amount, but I guess it’s around 15%.

I have had no interest in changing sex or gender role; that is there has never been any
confusion over being anything other than a *man/male* and heterosexual.

After 35 years of silence, I really needed to know about my gender issues, but I was embarrassed, unable to talk to anyone and no idea who to contact for information.

Thinking that one of the endocrine doctors would know, I plucked up the courage. Although he made a gallant effort to answer my questions it was an utter disaster, for he knew nothing of this subject and of course it resulted in tangible embarrassment for both of us and inner anger towards myself for having put both of us through that cringing ordeal.

When I eventually contacted one of the gender support groups I was staggered by their vast experience and understanding of emotions such as mine. I can’t express in words how I felt that day, listening to the confidential contact on the phone.

Having studied KS for so long I became confident enough to eventually talk to my GP and it was fine, not the big deal I thought it was going to be. And he was so supportive and understanding I wished I had talked to him sooner.

I have recently discovered that I am ‘Androgyne.’ This is different from being a Transsexual or Transgendered person. *(See the chapter The Sex/Gender Spectrum – Terminology & Usage, ‘Androgyne’, p67).*

**You are not alone!**

From talking to various support groups the most widely felt emotion expressed by those with Klinefelter’s Syndrome is the feeling of being alone. Often not aware of what is happening to them, they withdraw into themselves; unable to talk to family members, spouse/partner or to their GP.

And of course nobody around them knows what they are going through, making assumptions – such as that they are gay; because they are not interested in girls/women, or a wimp because they won’t participate in physical education or rough competitive contact sports.

Though in general children with KS are passive, there can be mood swings. This is particularly true of adults, with disruptive outbursts, usually due to frustration.

Throughout my life I have been a pussycat— to a point. Then, through building frustration I would suddenly snap, totally losing the plot. Thankfully, it has mostly been inanimate objects that have received my unswerving attention.

Over the years I have learnt to reduce the ferocity of the outbursts; otherwise I would tend to destroy whatever fiddly thing I was trying to create or mend.

Back in 1997, eager to commence my studies into Klinefelter’s Syndrome, it seemed then that it was a straightforward and relatively easy to reach goal.

*How wrong I was!* It would have been a waste to just file away all the knowledge I have collated and learned over the past eleven years, and so I offer it to you, in the hope that it helps you or someone close to you to understand more fully the complex nature of Klinefelter’s Syndrome.

As I have said, I may not be the world’s greatest writer, but the work is – to the best of my ability, honest.

**So please trust me.** There is someone out there right now, waiting to hear from you, someone with experience of Klinefelter’s Syndrome – and the many other facets that accompany this condition, who is ready to listen, to offer advice, and to ease your mind. And if you’re too embarrassed at this stage to talk to someone, go to one of the support group websites.

Whatever you decide to do— please remember,

**You are not alone!**

Left: AAAAAAAAAAAAAAAAAAAAAAAAAAAA!
One of those fiddly jobs that didn’t quite go to plan! And yes – that might be me!
Loneliness and fear
Klinefelter’s Syndrome may not be life threatening, but it can cause untold suffering. Many experts will tell you that with early diagnosis and with the appropriate treatment children and later adolescents with Klinefelter’s Syndrome can lead a ‘normal’ life.

On the other hand, researchers will point out that because of the complex nature of Klinefelter’s Syndrome the majority of individuals will not be diagnosed until adulthood. So, what happens if you are into your 30s, 40s, or even 70s, and have gone through your life with both the physical and psychological symptoms, but not the diagnosis - how ‘normal’ a life is that?

There can be the sense of loneliness and fear associated with those symptoms, when you feel you’re the only one in the world like that and there is no one to talk to.

For those in their early teens, from experience, it can turn school days; supposedly ‘the happiest days of your life’, into a minefield of lies and deceptions, as a teenager with Klinefelter’s Syndrome finds ways not to participate in rough competitive games, or change and shower for physical education.

In sport, often it is a case of having to endure being left as a spectator, or picked last, usually in goal or a place where you don’t have major participation. And afterwards in the shower-rooms, it is common for boys of that age to compare genital size, which for those with KS is an absolute no-no.

It is often the case that the teenager will be bullied by boys and ridiculed by the girls for being weak and will cry at the drop of a hat, which also incurs the wrath of their peers.

Additions and Losses
There is another element to the physical aspects of KS — that of breast tissue (‘Gynaecomastia’). Again this is a point of ridicule and also happens to thousands of typical males. Figures range from 30-60% of the general population, with most cases starting and ending during puberty. Generally, those with breast tissue have hard, often painful lumps behind the areola (the pink or brown area surrounding the nipple).

While many individuals with KS are ‘pleasantly plump’ to obese, gynaecomastia is not caused by being over-weight, but by glandular tissue surrounded by fat deposits; similar to a woman’s breasts and can’t be ‘cured’ by diet or exercise. The cause is generally a hormone imbalance, which is why it can affect those with Klinefelter’s Syndrome.

Some males hate their breasts and want them removed. Some tolerate them, while others grow to love them, though this normally takes many years. Those affected go through all manner of emotions, sometimes even questioning whether they are male. Breasts can range in size from small buds to double Ds.

To the individuals involved there can be severe embarrassment and low self esteem, and like KS, many teenagers find it impossible to talk to their parents about it. Often the person will wear loose fitting clothes to try and hide their lumps and will stay covered in public places, such as the beach.

As mentioned, usually gynaecomastia is temporary but in some cases it can persist into adulthood. If this occurs, often doctors will suggest surgery. Prior to the 80s surgery was basic; in some cases brutal and generally disfiguring. However, the latest procedures employ liposuction through a tiny hole to remove the fat deposits. To remove any glandular tissue (the hard lumps), small incisions are made at the lower edge of the areola to minimising the appearance of scaring.

While many individuals will be delighted with the surgery and adjust well to having a flat chest, some males who have lived with
Changing from female pattern hair to male can also result in loss of hair. This can be profound and in some men can happen within two years of the commencement of testosterone therapy. But everyone is different and like the general population scalp hair loss is variable. While a large percentage of individuals with KS appear to be happy to be a man/male and to maintain that gender role, other individuals see themselves quite differently. This is a highly complex subject and one which is explained in full in The Sex/Gender Spectrum – Terminology & Usage chapter, starting on p63.

**Gender and sex issues**

The physical embarrassment is only part of the story as on rare occasions the individual can also start to experience ‘feelings’ or a sense of being other than their outwardly perceived gender – that is a boy who feels partially or totally feminine. This can happen from around puberty onwards. Worst of all, it creates anxiety, loneliness and in some cases even thoughts of suicide. “Gonads produce hormones that affect the brain and it’s our brains that tell us whether we’re male or female. In most cases, there’s a physical reason why individuals might be unsure about their sex.”

Additionally, according to research carried out in the 90s, if you have KS you are no more prone to sexual orientation changes than the general population.

**Other Karyotypes**

While not wishing to detract from the grief you may be going through, like so much in life, there are always others with different concerns. There are other more severe karyotype conditions, with very rare cases reaching three additional X chromosomes (i.e. 49,XXXXY). There are also mosaics (e.g. 46,XY/47,XXY) and the 47,XXX Syndromes (see Karyotypes p23) and of course other genetic and hormone related conditions.
The KS Story –

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You are not alone

AG

TC

Genetics

DID YOU KNOW?

It was the American cytogeneticist Nettie Maria Stevens (1861-1912), who discovered through her fertility experiments with mealworms that there were distinct differences between the sex and the chromosome type of females and males, calling them “X and Y.” Her work was published in May 1905.

Based on information from Net Industries, LLC and Advameg Inc websites.

Chromosomes

What they are and where they are in the body

The Cell

Perhaps the best place to start is within the cell; the basic unit of all living organisms. A human adult has a hundred million million of such cells; making up the bones, tissue and organs of the entire body.

Each cell is contained within a cell membrane of lipids and protein, which controls the passage of substances in and out of the cell. Cells contain cytoplasm, in which are suspended a nucleus and other structures (organelles), specialised to carry out particular activities in the cell.

Laying within the nucleoplasm of the cell nucleus is the chromatin; the material which contains DNA (Deoxyribonucleic Acid) and protein. The substance of which chromosomes are made. (Diagram above, Red circle).

The Chromosome

The chromosome is a thread-like structure in the cell nucleus that carries the genetic information in the form of genes.

It is composed of a long double filament of DNA coiled into a helix, with the genes arranged in a linear manner along its length, together with associated proteins and ATP; for energy.

The nucleus of each typical human somatic cell contains 46 chromosomes, 23 of which are of maternal and 23 of paternal origin. The sex cells (sperm & ova = gametes) always contain half the number of chromosomes of all the other cells of the body; at fertilisation a sperm and ovum combine to form a cell with a complete set of chromosomes that will develop into the embryo.

The last two chromosomes determine the sex of the person; XX for a typical female and XY for a typical male.

Each chromosome is a different part of DNA, or sequence of the four bases.

Structure of a Chromosome

Meiosis - reduction division (part of process). At the start of gamete cell division each chromosome, which consists of DNA strands, protein and ATP, shortens and thickens; by repeated coiling. At Interphase the chromosome duplicates itself, creating two chromatids joined by a centromere... (See also Meiosis and Mitosis in a medical dictionary).
Human Reproduction

The Female

There are four ways of recognising a baby’s sex;

- **Genetic sex** (its karyotype – e.g. 46,XY);
- **Biological sex** (according to internal organs);
- **Biological sex** (according to external sex organs);
- **Brain sex** (structures which are believed to be linked to a particular sex).

At fertilisation, within a Fallopian tube the chromosome type is established (Genetic sex) i.e. XX typical female; one X paternal, one X maternal. The cells of the developing zygote begin to divide and within a few days implants itself within the the uterus. The overall term for the products of conception is an ‘embryo’. At the 8th week it is known as a fetus. (See p44-45).

**From the 3rd to 7th week**

Tissue around the developing kidneys and gonads changes into a variety of tubes and ducts; the beginnings of the internal organs of reproduction.

Although all the main organs of the body develop at the embryonic stage, the external sex organs for both males and females start on the same basic path, that is identical ‘Biological sex’, resembling neither female nor male. (See diagram to left).

**The 8th to 12th week**

At this point the sex organs are formed. If there is an absence of androgen (testosterone), the genitals are pre-programmed to become female, regardless of the genetic sex, (e.g. XX), and alters the developing gonads into ovaries.

An XX foetus will typically have no masculinising hormone (testosterone), and so the male structures called Wolffian ducts begin to die under the influence of Mullerian Inhibiting Hormone (MIH), and the external structures take on the female form of the: clitoris 1 labia majora (outer labia) 3 vagina 4.

There are also various genes which assist in the process by preventing the development of male hormone producing structures; such as Leydig cells.

After this point the ovaries then produce oestrogens which instructs most of the rest of the development, including assisting the development of the Mullerian ducts into the uterus, fallopian tubes, cervix uteri and upper part of the vagina.

Sexual orientation, sexual identity and gender expression are wired in stages throughout pregnancy. As with the sex organs, differentiation of the female brain takes place due to a lack of testosterone during this period.

In both females and males, currently at birth, social gender is assigned strictly based on external genitalia.

**Not quite the typical**

There are many ways in which the aforementioned 46,XX karyotype development can be altered. A change in Meiosis can cause Triple X Syndrome (47,XXX), as well as mosaics. (See p23).

The absence of anti-male genes can alter the internal anatomy, while the presence of testosterone (secreted from the adrenal glands), can masculinise the external organs (as in the condition Congenital Adrenal Hyperplasia – CAH), and some research suggests even the brain.

**Ongoing research**

It is currently believed that for both sexes, the feeling of having a particular gender identity comes predominantly from Brain sex (physiological), while, as previously mentioned, Nurture plays a part in the formation of our ultimate gender identity.

Recent research in the Netherlands points to a fundamental relationship between physical structures in the brain and gender identity and behaviour. However, “we don’t know yet whether the sexual dimorphic structures described so far in the brain are the cause, or just a symptom of gender dysphoria and I don’t think we will know soon.”

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**DID YOU KNOW?**

There is a higher risk of a child developing a genetic condition, e.g. Cystic Fibrosis, Down’s Syndrome or Klinefelter’s Syndrome, if a woman conceives later in life. Generally, clinicians see this as over 35. Also recent research points generally to a dramatic fall in fertility after 28 years of age. (contd. on p46)

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**REFERENCES p11 CONTACTS p74**

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**M1**

Prof Dick F Swaab, Netherlands Institute for Neuroscience, Amsterdam. (See Further Study chapter for details, p76).

See also an article on Prof Swaab and other gender related topics, in the New Scientist, (12th May 2001, No. 2290, p31), Gender.

**Brain Sex related:**

‘IT MAY BE YOUR BRAIN NOT YOUR GENITALS THAT DECIDES WHAT SEX YOU REALLY ARE.’

Full story, see New Scientist, (9th October 2002, No. 2365, p17).

‘Our brains could be hard-wired to be male or female long before we grow testes and ovaries.’

Contrary to current understanding of early human development (read this chapter), research carried out in the USA on rodents back in the late 90s, showed differences between males and females in the expression (switching on) of a staggering 50 genes, well before the SRY gene gets a look-in.

This of course is sexual differentiation of mice, not humans. So the next step will be to show that these genes truly influence the brain - and not just in mice. It is then hoped that in the future, through a simple blood test, doctors will be able to accurately establish the Brain Sex of new born babies who have ambiguous genitalia, taking the guesswork out of it for clinicians, and the possible emotional turmoil to parents and ultimately the child.

**Continued p44 margin...**

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**M2**

(See The Sex/Gender Spectrum - Terminology & Usage, starting on p63).
The Male

3rd to 7th week
Males start off in similar mode to females, however around the 6th week if the embryo is genetically male, genes on the sex chromosome, in this case Y, (SRY gene) signals the creation of testicular cells, called Sertoli cells. With the assistance of another gene these cause other cells to become testicular cells.

8th to 12th week
Leydig cells in the gonadal ridge start to produce the masculinising hormone testosterone, which triggers the development of the male reproductive tract from the Wolffian ducts, and the Sertoli cells make a hormone called anti-mullerian hormone (AMH), which destroys the Mullerian ducts, which would otherwise become the female reproductive tract.

After this point, the chromosomes have only a secondary role in sexual development. The foetus converts testosterone into dihydrotestosterone which masculinises the external genitalia: the penis (Glans) 1 the urethra moves to top of Glans from its central position 2 the scrotum 3 (see diagram on page 42).

20th week to 3 months postnatal
While sex organs differentiate up to the 12th week of gestation, in males testosterone influence upon the brain will typically occur later; in two peaks, the first at mid gestation (around 20 weeks), and the second around birth — up to a few months postnatally (about 3 months).

“These are probably the peaks that programme sexual differentiation of the brain. Because of the time lag between sexual differentiation of the sex organs and the brain, they can follow a different course (e.g. in transsexuals).” DS

Puberty
In both males and females, it is at puberty that the secondary sex characteristics are defined by the same hormonal messengers.

Not quite the typical
As with females, male development doesn’t always go to plan. An XY embryo can become female if the SRY gene or testosterone is missing or reduced, or if the foetus doesn’t respond to testosterone, as in individuals with Androgen Insensitivity Syndrome (AIS). (See p23 for the various KS karyotypes).

Pre-natal diagnosis
Chorionic villus sampling (CVS) at about 10 weeks or amniocentesis at about 16 weeks is available for older mothers; generally defined as over 35 years of age. However, it should be pointed out that there is a risk of miscarriage associated with the latter tests, currently about 1:100. Your consultant can advise you on this, and any other questions you may have.

(Important: Please also read the ICSC chapter p48, which outlines ‘Preimplantation Genetic Screening’ (PGS) for aneuploidy conditions, which occurs at around 2 days after fertilisation).

(Also, see the Further Study chapter contacts, p75 for:
• Androgen Insensitivity Syndrome
• Congenital Adrenal Hyperplasia.

**DID YOU KNOW?**
Klinefelter’s Syndrome is classed as a genetic condition. However, KS is not an hereditary condition. It is a change in Meiosis, and is only known as a genetic condition because of the additional X chromosome(s).

**DID YOU KNOW?**
Meiosis (reduction division), creates 4 daughter cells with only half the chromosomes of the mother cell (i.e. 23). It occurs before the formation of ova and sperm. While in body cell reproduction, Mitosis produces 2 cells, with the full 46 chromosomes.
I have included the following human reproduction section to outline not only the beauty of design, in all its forms, but to indicate the various stages (approximate weeks) at which genetic and hormonal events take place; primarily relating to Klinefelter’s Syndrome (in dark blue).

The First Trimester (first 3 months) of development sees the main genetic and hormonal changes. After fertilisation the zygote makes its way along the fallopian tube, dividing as it goes, until after about 3 days it reaches the uterus. By now it is a tiny ball of cells called a morula. The ball of cells floats within the uterus for a further 3 days or so until it forms into a blastocyst (a hollow clump of cells), which is just visible to the eye.

**Week 2** The blastocyst embeds itself into the wall of the uterus. Chorionic villi projections burrow into the wall lining. The outer layer of the blastocyst (trophoblast), begins to develop into the placenta. Blood cells and the first heart cells begin to form.

**Week 4** The heart is already beating and the spine and nervous system are starting to form in the embryo, which is about 7mm long.

**Week 6** The development of the internal parts of the ears and eyes continues under the external depressions, the latter covered with skin which will form the eyelids. Holes that will become nostrils begin to form, as do the mouth and jaw. The brain and spinal cord are nearly formed. The urinary and digestive tract continues to form and the hand and feet digits are almost recognisable as webbed fingers and toes. **Week 6-7 genetic influence takes place; as the embryo’s karyotype forges the gonads into testes or ovaries.**

**Week 8** By now the eyes are almost formed and the foetus’s face continues to develop with a rudimentary nose. The umbilical cord has almost fully formed. The toes and fingers begin to separate. The appropriate hormones now start to play their part; influencing the sexual organs, continuing up to the 12th week. 

**Week 12** The foetus, now around 10cm in length (4”), is not yet conscious, and although it begins to move around; thanks in part to fully developed inner ears, the mother will still not sense her child’s movements.

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**DID YOU KNOW?**
One study on KS, showed that the risk at maternal age 40 is 2-3 times that at age 30. 
MRC, Western General Hospital, Edinburgh, UK.

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**REFERENCES p11 CONTACTS p74**
M1  ‘One at a time’ is a professionally-led site aimed at reducing the risks of multiple pregnancies from fertility treatment. It also offers a good patient perspective.

More information on this and other aspects of ICSI at:

- National Institute of Clinical Excellence (NICE)
  - [www.nice.org.uk/CG11](http://www.nice.org.uk/CG11)
- NICE policy covers England, Scotland and Wales. Other guidance for the NHS in Scotland is developed by:
  - [www.rcogs.ac.uk/guidelines](http://www.rcogs.ac.uk/guidelines)

M2  The HFEA produces a variety of statistics on the treatment licensed centres provide and have a comparison of birth rates following ICSI and IVF at:

- [www.hfea.gov.uk/2588.html](http://www.hfea.gov.uk/2588.html)
- [www.oneatatime.org.uk](http://www.oneatatime.org.uk)
- [www.dhsspsni.gov.uk](http://www.dhsspsni.gov.uk)
- [www.nhsb.fs.nhs.uk/icsirisks.html](http://www.nhsb.fs.nhs.uk/icsirisks.html)

More information on and genetics

Intra-cytoplastic Sperm Injection

For those men in the past with non-mosaic Klinefelter’s Syndrome, who wished to have a family, life was bleak. But nowadays there may be the option of ICSI (as in *Pixie*). It basically depends on whether you have live sperm.

ICSI was discovered by Belgian scientists in 1991 and is one of the assisted reproductive technologies (ARTs) that can be particularly useful in cases of male infertility. Here it can literally be true that it takes only a single sperm to bring about fertilisation and hopefully a successful pregnancy.

Sperm retrieval will be by ejaculate or testicular fine needle aspiration (FNA) from the epididymis of the vas deferens (PESA or Per-Epididymal Sperm Aspiration) from the testicles themselves (TESA or Testicular Sperm Aspiration) or extracted from a sample of biopsied testicular tissue (TESE or Testicular Sperm Extraction).

In both ICSI and IVF (In Vitro Fertilisation), embryologists will attempt to fertilise as many eggs as they are able to recover (this could be up to 20). That said, following public consultation, in 2007 the HFEA introduced a policy to reduce the risk of multiple births and attendant problems from patients identified as most likely to conceive twins. Now one embryo (max) should be transferred per cycle.

The exception to this is for women over 40 using their own eggs who may have up to 3 embryos transferred. During the fertilisation process some eggs may fail to fertilise or fail to develop after fertilisation. While the HFEA guidance on the maximum number of embryos transferred has not changed, because of the complexity of this procedure, the latter policy statement may also be relevant.

If there are embryos that are surplus to immediate transfer requirements that are of suitable quality, these can be frozen and stored for future use. It must however be remembered that not all embryos will survive freezing and subsequent thawing when they come to be used at a later date. Sadly, sometimes none of the stored embryos will survive freezing and thawing.

The difference between the two methods lies with the fertilisation process. With IVF, eggs and sperm are placed in culture medium in vitro (‘in glass’ – not actually a glass testube but a plastic culture dish) and fertilisation occurs ‘naturally’. With ICSI a single sperm is injected into the cytoplasm of each mature egg.

Signs that fertilisation has occurred are usually present 16-20 hours later. 2, 3 or 5 days after eggs retrieval, embryo transfer into the woman’s uterus can be carried out. (See diagrams).

Although ICSI improves fertilisation rates compared to IVF, once fertilisation has been attained the pregnancy rate is no better than with IVF.

There have been some follow-up studies on ICSI, the associated risks are outlined on the HFEA website.

When genetic problems have been detected by chromosome analysis (Karyotyping), genetic counselling is essential so that the risk of passing on a genetic problem to a child are completely understood.

Diagnosis and Screening procedures

In conjunction with ICSI and IVF the most common screening procedures to assess the genetic ‘normality’ of embryos are PGS (Preimplantation Genetic Diagnosis) and PGS (Preimplantation Genetic Screening).
The KS Story – You are not alone.

M4 The PGD & PGS procedures are based on information kindly supplied by the Human Fertilisation & Embryology Authority (HFEA). (obtains results from PubMed, OMM and other databases), or search direct from PubMed at: www.ncbi.nlm.nih.gov/pubmed

M5 HFEA licensed clinic directory: www.hfeagov.uk/guide/advancedSearch.aspx

Pre-implantation Genetic Screening (PGS) (also known as aneuploidy screening) involves checking the chromosomes of embryos conceived by intra-cytoplasmic sperm injection (ICS) or in vitro fertilisation (IVF), for common anomalies. For more details visit: www.hfeagov.uk/pgs

Clinics licensed to undertake PGS: England 5; Northern Ireland 4; Scotland 1; Wales 1. (For more details see M5 above). There are no clinics (in the UK) licensed to screen for Klinefelter’s Syndrome. However, centres using PGs will identify KS as a matter of course.

Genetic Screening). PGD is used to test for specific genetic conditions and characteristics. PGS is used to identify the number of chromosomes present in a cell, by analysing them about 2 days after fertilisation, when the morula has within it around 8 cells, called ‘Blastomeres’ (see diagram on p46).

This involves a technique called ‘Fluorescence in-situ Hybridisation’ (FISH); which uses small pieces (probes) of DNA, which are labelled with different coloured fluorescent dyes, which attach themselves to the various chromosomes of each type in the cell; enabling the geneticist to count them.

While both these procedures are generally defined as a means to identify ‘abnormal’ cells and to select ‘normal’ embryos for transfer, this often refers to inherited genetic conditions within a family history.

However, as I mentioned on page 15 (margin); Klinefelter’s Syndrome is a highly complex condition which affects people in different ways. A minority will be seriously affected, others will struggle, while the vast majority will overcome earlier difficulties or will be totally unaware of any symptoms. After all, who wrote and designed this book on Klinefelter’s Syndrome!!

With regards to Klinefelter’s Syndrome, unless there are serious complications, termination should not be the overriding objective in seeking any kind of screening.

For the number of clinics in the UK licensed for PGD and PGS please see the HFEA website. There are no centres currently licensed in the UK to carry out PGS specifically for Klinefelter’s Syndrome [2011], however, most centres carrying out PGS for chromosomal ‘anomalies’ will use probes for sex chromosomes and will, as a matter of course, identify embryos with Klinefelter’s Syndrome. In addition to other aneuploidy conditions those licensed for PGS use probes for trisomies: 13, 18, 21 and X & Y.

‘Polar Body’ testing. A developing ovum will produce two small cells called ‘Polar bodies’, which do not develop into functioning egg cells; degenerating after fertilisation.

Examination of these cells provides an indication of the number of chromosomes within the ovum.

After removal of a polar body by micropipette, the ovum is placed in an incubator. This procedure is usually only carried out where there is thought to be a risk of maternal age-related aneuploidy, (e.g. KS), where maternal meiosis stage 1 errors are a factor.

It is now possible to carry out PGS tests on a Polar body thereby avoiding the need to remove a blastomere from the embryo.

There is strong opposition to PGS and other procedures of this kind from some groups who see a single cell blastomere as a potential human being. That said, it is not for me to make judgements within this guide, but to leave you to make up your own mind on the ethics.

Your doctor or consultant can give you more up-to-date information on everything mentioned here, including success rates, clinical geneticists and genetic counsellors. There are also information sites on the web on the clinical, ethical and moral aspects of prenatal screening and diagnosis.

REFERENCES p74 CONTACTS p74

Stop Smoking Suggestions

Breathing through and nourished by the lifestyle that is the umbilical cord, the embryo or fetus is a part of the mother. Smoking during pregnancy can increase the chances of producing a child with Klinefelter’s Syndrome. Men who smoke reduce their chances of fathering a child by two to three times. The latter study which relates to both IVF and ICSI, also points to a marked reduction in retrievable eggs and fertilisation, if the mother is the smoker.

A study by Dr Zitzmann, M. (2002), Smoking damages IVF chances. Institute of Reproductive Medicine, Münster, Germany.

If you live outside the UK: I suggest you either obtain a recommendation from your MD, or do an online search for reputable government based or supported sites. Just type into the search field: “Stop smoking” followed by your own country’s name (obviously in your own language). I tried it, and from the search lists found USA: www.smokefree.gov/ This site also includes a link to: http://women.smokefree.gov/ For Australia: www.quitnow.info.au/. I also tried India, New Zealand and Sweden and got similar results. If you’re not sure if the site is government based, look for an official crest or read their About Us link. The aforementioned sites are just suggestions. I strongly recommend that you do your own search.

My “In terms of information the UK National Screening Committee does not provide details about this condition [KS]... as there is no programme to screen for this condition.”

Department of Health: Richmond House, 29 Whitehall London SW1A 2NS

That said, if parents have had previous experience of having a child with a genetic condition amniocentesis will be offered by the NHS. With the support of a genetic counsellor, parents can then decide whether to proceed or not.

As mentioned, KS may also be identified indirectly during PGS for other genetic conditions. Parents will then be informed and referred to the regional genetics team.

UK National Screening Committee (NSC): www.mcr.nhs.uk/index.htm

For “Code of Practice & Guidance on Genetic Paternity Testing” and “Guiding Principles for Commissioners of NHS services”. For the UK: Visit the NHS Choices’ SMOKEFREE website. You can also Chat to an adviser online by clicking the link on the Home page, top centre, under the phone number.

The website also has numbers for Asian language advisors.

NHS Stop Smoking Helpline: 0800 022 4332

NHS Pregnancy Smoking Helpline: 0800 169 0 169

Lines open Mon to Fri 9am to 8pm, Sat & Sun 11am to 5pm

Isle of Man Smoking Helpline: 001624 642 404

Northern Ireland Smoking Helpline: 0800 85 85 85

Scotland Smoking Helpline: 0800 84 84 84

Wales Smoking Helpline: 0800 160 169

NHS Pregnancy Smoking Helpline: 0800 169 0 169

Lines open Mon to Fri 9am to 8pm, Sat & Sun 11am to 5pm

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**Important personal note**

This chapter was completed in 2005 and does not take into account the Consensus Statement on Management of Intersex Disorders, published in 2006.

(Please see the end of this chapter (p55) for more details).

I have been incredibly fortunate to have had the opportunity to ask questions of so many important scientists, both in the UK and abroad, who’s pioneering work spans half a century in the fields of biological and cytotgenetic research. Without their support and patience, The KS Story would now be so many megabites in a bulging hard drive folder.

I appreciate that professional feelings run high regarding this subject, and that I am dealing with lifelong careers and passions. I don’t want to rub anyone up the wrong way; particularly those who are trying to help me, but I am no academic, I know only the facts that are supplied by the experts — but I don’t know who is right.

And so the consequences of asking the above question has troubled me greatly; should I include ‘intersex’ within the guide. And having decided, trying to obtain a balanced consensus on the response. Then ultimately afraid of how everyone involved would react to the final chapter.

To this day I can’t understand how this confused situation hasn’t arisen many times before. Relating to Klinefelter’s Syndrome, all articles and documents I have found to date, state one explanation or the other and never the twain. Routinely, intersex is never mentioned.

Of course, I could simply have avoided the whole issue and moved on to another non-confrontational subject, but I felt that I would be letting the reader down if I didn’t at least try to obtain some facts so I decided to investigate further, and to include all viewpoints.

Continued in opposite margin...
Viewpoint C

It is clear from the supplied document that Klinefelter's Syndrome is included within this study of intersex. However, this advocate intimated in the covering letter that the 'question' “depends on how you define intersex.” Further, it is pointed out that the study paper “is a practical and philosophical question of gender really, and there is no correct answer, only practical, personal and philosophical choices.”

“Abstract The belief that Homo sapiens are absolutely dimorphic with the respect to sex chromosome composition, gonadal structure, hormonal levels, and the structure of the internal genital duct system and external genitalia, derives from the platonic ideal that for each sex there is a single, universally correct developmental pathway and outcome. We surveyed the [International] medical literature, from 1955 to the present, [2000], for studies of the frequency of deviation from the ideal male or female.'

“We defined the typical male as someone with an XY chromosomal composition, and testes located within the scrotal sac. The testes produce sperm which, via the vas deferens, may be transported to the urethra and ejaculated outside the body. Penis length at birth ranges from 2.5 to 4.5 cm (Flatau et al., 1975); an idealised penis has a completely enclosed urethra which opens at the tip of the glans. During fetal development, the testes produce the Mullerian inhibiting factor, testosterone, and dihydrotestosterone, while juvenile testicular activity ensures a masculinising puberty.”

Within this viewpoint, intersex is defined as the following:

• “Sex” chromosome composition. Individuals with XXY, XO, XYY, XXXX, and XXX females...
• Androgen insensitivity in XY individuals... (Complete and Partial)
• Congenital adrenal hyperplasia... (Classic and Late-onset)
• Vaginal and penile agenesis...
• Hormone-producing tumors and exogenous sex hormones...
• True hermaphrodites and idiopathic mixed genitalia...
Children and Young People

ChildLine

ChildLine is the free 24-hour helpline for children and young people in the UK.

- Children and young people can call about any problem, at any time – day or night.
- ChildLine is confidential, which means they won’t tell anyone about your call unless you want them to talk to somebody for you, or you are in danger.
- If lines are busy, please keep trying and you will get through.

ChildLine is the free helpline and online service for children and young people in the UK. Children and young people can call 0800 1111 or visit www.childline.org.uk to talk to a trained counsellor about any problem, or talk to other young people on the message boards.

Bullying

In general, bullying is a harrowing and relentless experience, with individuals subjected to verbal threats and actual physical abuse.

At school...

Individuals of school age who are bullied often feel alone and unable to tell anyone in authority; particularly teachers; in case of reprisals. Even if the bully responsible is punished or expelled there is still the real risk; in the victim’s mind, that the bully will ‘get them back’ somehow, at some point in the future; perhaps when the person feels safe.

For children and teenagers with Klinefelter’s Syndrome there is already an aura of low self-esteem and a sensitive nature. They’re generally unwilling to fight and to participate in rough contact sports – all traits the bully can reinforce to their advantage. Kids with KS also make ideal victims since they are easily brought to tears by the merest of threats of gang beatings or extortion.

Often with a reluctance to involve parents, bullied individuals with KS may not get the full support they need and will struggle within the abuse/fear cycle until they either crack under the pressure and seek help, or resort to truancy to avoid any confrontations. In extreme instances, some only see one way out.

I experienced bullying at primary and secondary school, though thankfully it was sporadic. Occasionally I even managed to interact and even receive praise from ‘the enemy’; those boys who saw themselves as far superior to myself, which brought forth a kind of pride that I had been accepted. But all too often it was short-lived.

Given enough harassment I occasionally snapped into a kind of damage limitation frenzy, but with blind rage and limited fighting abilities the fracas was usually over relatively quickly, with me nursing my wounds – and usually in tears. So more often than not I gained a reputation for swift strategic withdrawals; preferring flight rather than fight.

Kids can be cruel at the best of times and bullies know all too well that they can always have another go at the ‘soft-touches’ who seem unable to challenge their authority.

There is nowadays the added fear of Cyber bullying; the use of information & communication technology, particularly mobile phones logged-on to social networking sites (SN), which penetrates even the perceived safety of the victim’s home. Cyber bullying can also take place between children and adults; such as a teacher, which can cause great stress and depression.

Apart from accidentally uploading personal details or images which can later cause upset, there are trends on certain SN sites, such as the ‘Like’ button (2010), generally intended to embarrass a person, possibly with crude comments about their sexuality.

Usually close school friends have each other’s mobile phone number, so if pals fall out it can be extremely harrowing to receive text message abuse and threats, particularly as x-pals will probably know some very private and possibly sensitive information about the other person.

Many schools today have set up peer support groups and there are specific helplines available for children and teenagers in this situation, but it is still vital that teachers and parents play their part in listening and supporting all vulnerable individuals of bullying; whether they have Klinefelter’s Syndrome or not.

Often it should be stressed that often there are underlying reasons why a child resorted to bullying, such as conflict elsewhere, resulting in low self worth, and that by informing a teacher about an incident it may also bring to light these problems; allowing counselling and support.

...college, university and work

Bullying doesn’t stop when a teenager leaves school. Further education and in the workplace, unchallenged; those who seek power; at whatever level of authority, will seek out and create victims. And it’s not just women who are victimised at work.

As a well practiced coward it is easy for me to say that workmates should try to support their colleagues and wheedle out those people who bully, but sadly in the big world it’s not that easy. A victimised employee hasn’t even the luxury of truancy. And if the bully is a director of the company – what then? Perhaps talking to a trusted family member, close friend or one of the adult support networks, such as those mentioned in the margin or within the Further Study chapter, may be the answer.

People of all ages

There are many good samaritans out there waiting to listen to you and offer confidential support.

Use a search engine and type: ‘Adult bullying in (type in your own country)’, or ‘talk to someone’, or experiment with text yourself.

It may be that your website browser has a search button specifically for results from your own country (e.g. ‘pages from the UK’). Clicking on it.

Whatever you decide to do, go carefully. Choose a well known and trusted organisation.

Children, Young People and Parents

Use one of the popular search engines to access several other valuable sites, including:

- BullyingUK
  - Formally Bullying Online w: www.bullying.co.uk
  - Kidscape w: www.kidscape.org.uk

Further details and Legal Support

Please see the Further Study chapter p78 for information on all contacts mentioned in this chapter, including advice on legal support.

REFERENCES p11 CONTACTS p74
Avascular necrosis, cancer, diabetes, leg ulcers, osteoporosis...

All these conditions have been linked to Klinefelter’s Syndrome in one medical paper or another. But as mentioned several times before, everyone is different. There are also generally applied criteria for having these conditions, for example; current medical thinking states that it is unlikely KS is involved in leg ulcers if there is venous insufficiency, or other underlying causes.

The following will give you a basic background to these conditions and their association with Klinefelter’s Syndrome (KS). By all means study these links, but don’t get bogged down in self-analysis and worry — everyone is different.

Avascular necrosis

If you have KS or have been recently diagnosed, then you should be either under the care of an endocrinologist (in the UK referred by your GP), or be receiving hormone replacement therapy. The sex steroid (hormone) testosterone is necessary to maintain healthy bones.

Avascular necrosis is a disabling condition which, in severe cases, can lead to joint collapse.

The condition can start with few warning signs, perhaps only with a dull ache in an unrelated area. Although it can happen in any bone it most commonly affects the femur. Other common sites include ankle, upper arm, knee and shoulder.

What is Avascular necrosis?

It is caused by the death of blood vessels to a particular area, and in the case of the femur, the joint becomes weakened; through lack of oxygen and nutrients; slowly dying, to the point of its collapse.

What causes this condition?

There are several general causes; from trauma, and damage to blood vessels, fat blockages and embolisms.

Associated conditions:

These include Caisson’s disease, Crohn’s disease, Cushing’s Syndrome, Gauher disease, Sickle Cell disease, Chronic Pancreatitis, Rheumatoid Arthritis, Steroid usage, Systemic Lupus Erythematosus, Alcoholism... and Klinefelter’s Syndrome (allegedly).

In my own case, there is a dichotomy of opinion on this link. Rheumatology sees it as endocrine based, while endocrinology is not convinced that testosterone deficiency is the culprit. To be fair, both consultants agree that there is considerable medical literature linking Klinefelter’s Syndrome to Osteoporosis. However, there seems to be no specific case studies of KS directly linked to Avascular Necrosis (a form of osteoporosis).

What are the symptoms?

In the early stages there may be no symptoms at all, however, as the condition progresses, which can take months, most individuals will experience joint pain, and/or referred pain in areas such as the knee, quadriceps or back. There will be limited motion and stiffness of the joint and weakness within the leg muscles.

The condition can affect just one joint; such as the hip, or both hips, or different joints at different times.

In the later stages the pain and associated weakness can be dramatic. Motion can also be restricted as both bone and cartilage are broken down and the smooth head of the femur becomes jagged and worn within the pelvic socket.

The time between the first symptoms and loss of joint function is different for everyone; ranging from several months to over a year.

How is it diagnosed?

Initially, in the case of the femur, the doctor or rheumatologist will rotate and flex the joint to check for pain and stiffness.

Avascular necrosis diagnosis is often made by X-ray. However, by the time film X-ray tests show up substantial damage can already have been inflicted on the bone (up to 30% bone density loss), therefore this form of X-ray is usually considered for later-stage findings.

To detect early signs, MRI scanning (magnetic resonance imaging) may be used to see the condition of the internal bone structure, and particularly to the neck of the femur. Alternatively, a DEXA scan (Dual Energy X-ray Absorptiometry), also known as DXA, will be used to assess the density of the bone, and the risk of fracture.

What treatment is available?

This depends critically on what stage the avascular necrosis has reached. In the early stages (before changes show up on film X-ray), there are some treatments, such as bone grafts. Later stages typically lead to surgery, e.g. a partial or total hip replacement. This involves replacing the femoral head with a titanium prosthesis and a special polyethylene cup fitted into the pelvic socket. (Try a computer web search for an illustration or photograph of a hip prosthesis).

What about pain management?

To try and maintain a decent quality of life, progressively stronger analgesics can be prescribed, as well as NSAIDs (non-steroid anti-inflammatory drugs). In severe cases, an anti-inflammatory can be injected directly into the joint area; under a local anaesthetic; which gives pain relief for up to 9 months. This usually requires a 2-3 day in-patient stay; to monitor the absorption.

Continued bone maintenance?

Including the testosterone replacement therapy you take for Klinefelter’s Syndrome; to ensure the bone does not deteriorate, you may be placed on a non-steroid osteoporosis therapy, which entails taking a daily tablet, for at least 3 years — possibly life.

This treatment temporarily stops “turnover” (the breakdown of bone) for 14 days, followed by a calcium tablet for 76 days (three month pack).

Cancer

Male breast cancer

Of the very limited research currently available, it is clear that generally, men have a very low risk of...
developing 'breast' cancer, with a possible higher risk for individuals with gynaecomastia, though one KS support group suggests that it may be the hormonal imbalance in KS which causes cells to become cancerous rather than actually having the breast tissue.

A couple of sources and one website puts the risk factor as similar to women, at 50 times that of a typical male. The same source mentions the need for XXY adolescents and adults to practice regular breast self-examination. The Cancer Research UK website suggests a 20 fold increase in a person with KS, but concludes that this is still a very low risk factor.

Germ cell cancer
Those with KS also have a very small risk of developing germ cell cancer, and the risk factor is greatly reduced after the age of 30, the peak being between 15-30. Judging by recent research into KS links to cancer, the incidence of these cancers are very low or inconclusive. (See Obesity.)

Other cancers
In 2005 a study of other possible associated cancers, on behalf of the UK Clinical Cytogenetics Group, suggested that men with Klinefelter's Syndrome may be at substantially elevated risks for non-Hodgkin's Lymphoma and perhaps lung cancer.

Diabetes
"Compared with other males individuals with KS have a slightly higher risk of autoimmune disorders." M7

The most common of these conditions is type 1 diabetes. In a British study, "the increased frequency of diabetes in Klinefelter's Syndrome, Turner's Syndrome, and possibly Down's Syndrome lead to the hypothesis that non-disjunction may, in some way be associated with a predisposition to diabetes." While a Taiwan study states that patients with Klinefelter's Syndrome "have a higher incidence of diabetes mellitus." M8

Leg ulcers
One study from France reported that an individual with chronic leg ulcers, (six years), was diagnosed with Klinefelter's Syndrome. It also stated that there was a higher frequency of venous insufficiency in patients with KS. While a study from The Netherlands stated that leg ulcers, especially in combination with hyperpigmentation or atrophic blanche of the surrounding skin can be a symptom of Klinefelter's Syndrome and are not necessarily to be attributed to venous insufficiency. M9

Both these papers contradict a verbal medical statement that leg ulcers are only likely to be linked to KS if there is no underlying venous insufficiency.

Obesity
There is no doubt that Klinefelter’s Syndrome is linked to obesity, but KS is not the direct cause. Eating the wrong diet is the culprit. Obesity is a very serious condition – one in which I know all too well, having spent most of my adolescent and adult life trying to correct and is linked to cancer, strokes and heart disease.

Those with KS will find it easier to gain weight and harder to lose it, and many with KS will be over-weight or obese.

For a child, sweet things are a favourite and all to easily they are on the slippery slope to weight problems. For this reason, parents of a diagnosed KS child, or teenager, will be advised by their GP or dietician, as to the most appropriate diet for their child – and this should be for life!

Cancer link
One of the causes of breast cancer in males is from high oestrogen levels. All males produce some oestrogen, which helps to maintain our bones, but high oestrogen levels have been linked in the past to breast cancer. Besides chronic liver conditions and genetic conditions obesity is a causal element; in that oestrogen is partly made in fat tissues of the body.

Osteoporosis
There are many people with KS who have an aversion to medical establishments and particularly to injections, dropping vital therapy and perhaps never commencing hormone replacement in the first place. This can lead to extreme osteoporosis.

Yes — you're right, I am trying to scare you. For it's the only way to get the message across. Remember what I said on page 15? 'If you take only one piece of advice from this guide.' Will this is it? No excuses now... GET YOUR TESTOSTERONE TREATMENT!

Osteoporosis worsens silently, and when breaks occur they can be caused by a small amount of force and are painful and debilitating, leading to months of pain and stress.

If you have been without hormone replacement for decades then you should talk to your GP. The procedures will be much like those mentioned in the Endocrinology chapter.

What is Osteoporosis?
It is the loss of bone density, which in advanced stages can leave individuals vulnerable to frequent bone fractures, particularly of the hip, spine and wrist, destroying quality of life.

Generally, osteoporosis is worse in the elderly, and in women following the menopause. While more women are affected by osteoporosis (1 in 3, compared with 1 in 12 men), generally, more men suffer from 'Avascular necrosis.'
What causes this condition?
For those with Klinefelter’s Syndrome, as well as males in general, one of the risk factors is from low levels of testosterone and dihydrotestosterone (called hypogonadism). Because these sex steroids (hormones) play an essential role in the maintenance of bone mass, some studies have been done to investigate males with gonadal dysgenesis, particularly those with Klinefelter’s Syndrome.

Studies have shown a reduced osteoblast function and reduced 5 alpha-reductase activity; which is responsible for partial resistance to androgens, with increased bone turnover, particularly at the femoral neck. In another case it was suggested that bone rarefication in KS is associated with a reduction in hormones, not directly with the KS.

Associated conditions:
These include chronic use of corticosteroid drugs, Crohn’s disease, chronic immobility, gastric surgery, heavy drinking, a history of fractures, low body weight, smoking and specifically to KS; those with low levels of testosterone (hypogonadism). There is also a recognised and well documented link between KS and Systemic Lupus erythematosus, and the latter to osteoporosis.

What are the symptoms?
Warning signs are usually a broken bone resulting from a minor fall, curvature of the spine and height loss.

In itself, osteoporosis causes no pain; it is purely the thinning of bone tissue. However, in advanced cases, if that breakdown is part of the femoral head for example, pain is very much in evidence when exposed ragged bone interacts with the muscle and cartilage of the surrounding pelvic socket and tissue.

How is it diagnosed?
As with avascular necrosis, normal X-rays do not highlight osteoporosis soon enough, and again, DEXA (DXA) scans are employed to measure the bone density. “It is currently the most accurate and reliable means of assessing the density of bones. It is a 10-15 minute painless procedure, that uses very low doses of radiation.”

Treatment, Pain Management and Ongoing bone maintenance
Basically As Avascular necrosis.

As with the latter condition, there is one thing that anyone with KS can do right now, and that is to discuss possible hormone replacement therapy with their doctor or an endocrinologist.

Don’t jeopardise your future health for the sake of a few seconds (possible) discomfort each month.

Of course if you’re suited to the testosterone gels (see p24) there should be no excuse!

Other conditions
There are also (possible) KS links to conditions such as Taurodonism and Radial Ulnar Synostosis, and because of the slightly increased risk of autoimmune conditions, there are links to Asthma and Chronic Thyroiditis.

Understanding and Compassion
As yet there are no controls over how our body’s blueprint creates us. We are born how we are born.

It is strange that the older humanity gets the more confused it becomes. Aren’t we supposed to learn from our experiences? Why can’t humanity abandon outdated Victorian attitudes, put aside sensationalistic notions, flip negative to positive — and celebrate the diverse spectrum of its offspring?!

Why does it have to be difficult; why can’t it be easy? Unacceptance often comes from ignorance of the facts and unfamiliarity. With just a little more understanding and compassion, think how much less grief there would be in the world.

That said, today, more than at any other time, there are those who are willing to cast aside outdated prejudices towards minority physiological and social groups, and to try and understand. But accepting other sex or gender groups is one thing, but what terms do you use without causing offense?

Terminology and Usage
As far as Sexology is concerned, around the globe language is in flux; altering the meaning of well established terminology and definitions. This is compounded by various social communities who periodically interact, incorporating themselves into other groups’ politics, which can cause serious inter-group disputes and confusing terms and definitions. The situation also changes from country to country. And if that wasn’t confusing enough, it also seems that even to those working in sexology on a daily basis sex/gender terminology and usage is confusing!

So opening your mind is one thing, but when you don’t understand the answer, then you’re going to wonder why you bothered; i.e. what is and what isn’t PC. And to cap everything else, terminology is fluid, so what you are about to read will undoubtedly change.

I would be lying if I said the following wasn’t complicated; it took over a year to get this chapter right, but I hope like me, you will be astonished at what you are about to read. And please persevere, for the more we understand and use these terminologies the more we help everyone — and made a difference.

Key words:
Sex, Gender, Sexual Identity, Gender Identity, Gender Role, Gender Dysphoria, Gender Identity Disorder, Transsexualism Transgendered, Androgyne, Sexual Orientation, Intersex, Transvestism.
Sex and Gender

While these two terms are generally used interchangeably by society without any difficulty, it is when medical, research, political, legal and religious aspects arise that this generalisation can lead to confusion and misunderstanding.

‘Sex’ describes the perceived external or anatomical or biological image; ‘phenotype’, while ‘Gender’ is the psychological identification of the individual, it is how individuals experience themselves. Gender is also the social/cultural expression of that identity. Typically, of course, these elements of sex and gender are consistent.

“Sex typically, but not always, represents what is between one’s legs, whereas Gender represents what is between one’s ears.”

Identity – Sexual Identity

As mentioned, terminology and its definitions vary enormously around the world and the above term is no exception. Originally this book was targeted at the UK reader, but now covers other parts of the world. Because of this, the above term varies from country to country. Trying to explain this has caused much confusion, stress and gnashing of teeth.

For my sanity and as there is limited space here to explain it all, the following are the terms and definitions originally supplied to me by Prof M Diamond, University of Hawaii, John A Burns School of Medicine, Hawaii, USA, which relates to a sizable part of the world.

However, the name ‘Sexual Identity’ is an issue in the UK, Austria, Australia, New Zealand and some parts of the USA, where it refers to ‘Sexual Orientation’. For this reason I strongly advise you to contact the Gender Identity Research & Education Society (GIRES) for another gender information group for the current terms. (Please see p76 for contact details).

Sexual Identity & Gender Identity

‘Sexual Identity’ is the way someone sees his or herself as male or female, based on the person’s outward physical appearance (phenotype). ‘Gender Identity’ is the perceived social image of being a boy/man, or girl/woman.

For example, a KS person may see their ‘Sexual Identity’ as male but see their ‘Gender Identity’ (their psychological self) as feminine and prefer to live as a woman e.g., “I am a woman trapped in a male body.”

In the typical individual sexual identity and gender identity are in agreement. It is only when the individual is possibly a Transsexual person that it becomes an issue – and only in certain societies.

It is believed that like Sexual Identity, Gender Identity is hard-wired into our brains, but there is insufficient evidence to be certain. There is likely to be some ‘modification’, which does not show itself until after birth when behaviour patterns are observed.

Gender identity may or may not be in accord with the sex characteristics of the rest of the body. Where there is profound and persistent discord the individual may be said to be a transsexual person.

Nothing stands still in research and some studies have shown a fundamental relationship between Gender Identity and “hormonal changes at puberty, which alter previously set neuronal systems within the hypothalamus of the brain.”

In fact, “Transsexuals clearly show evidence of mixed structures.”

Although it has not yet been determined if this is causal or symptomatic. (These areas are called SDNs, or Sexually Dimorphic Nuclei).

Gender Role

‘Gender Roles’ refer to our idea of how boys and girls and men and women should act and be treated in society.

For example, it used to be that men’s ‘Gender Roles’ were typically associated with strength, aggression and dangerous occupations, while women’s ‘Gender Roles’ related to a gentle nature, childrearing and nurturing. Nowadays, to a certain extent, these roles are more blurred, with women becoming the sole bread-winners while the man becomes the house-husband. Of course these descriptions change around the world, for example, in African societies roles will be completely different to those in the UK.

‘Gender Roles’ on the other hand, could be said to be openly or discretely dictated by society, for example, in the way girls learn to keep their knees together, properly adjust their clothing and apply make-up, while boys tend to sit with their legs spread wide open, memorising the rules of sport.

Throughout life, gender role will be influenced by interactions with family and the rest of society.

Not all transsexual people will decide to change their gender role so that it conforms to their gender identity.

Gender Dysphoria or Gender Identity Disorder (GID)

This relates to the conflict between a person’s gender identity and the sex characteristics of the rest of the body.

“Despite considerable gradations most people are close enough to one end or the other of the gender/sex spectrum that they never have to question whether their gender identity is consistent with their sex. Since this is true for most, it is assumed to be true for all babies - that what you see is what you get. But this is not always so.”

Some people are born male but their brain, as they develop, tells them that they are a girl or woman, or born female and feel that they are a boy or man. It is as natural to them as it is to others who are, for example, born male and know they are a boy or man.

There are cases of gender conflict in children as young as 4.

Whatever the reason, and there are many, it is a powerful and compelling force. We are, what our brains tell us we are.
Transsexualism

Transsexualism arises from a conflict between the brain and the rest of the body. Transsexual people are those who exhibit the criteria for Gender Dysphoria or Gender Identity Disorder (GID). M6

Sometimes distinctions are made as to whether the person is a preoperative or postoperative transsexual person.

To partly remove the distinctions of Gender Identity Disorder and Gender Dysphoria which are made as to whether the person is a ‘Transwoman’ were adopted – later modified to ‘Transsexual’, the terms ‘Transman’ & ‘Transsexualism’ arises from a conflict between the brain and the rest of the body. Transsexual people are those who exhibit transgender behaviour want to change aspects of their gender, but don’t necessarily want to permanently change their sex. That said, in 2010 the UK media still use the same definition for transgendered and transsexualism.

As with different social groups so there are global changes to the definitions. In some states of Australia for instance, they use ‘transgender’ exclusively for ‘transsexualism’, (generally shortened to ‘Tranny’). M7

It seems like the Australians, that some other communities around the world, such as homosexual and bisexual people and also other groups such as intersex, are adopting the term transgender.

About five years ago, in the UK the term ‘3rd G’ (Third Gender) and in the States the term ‘TGV’ (TransGender Variant) were adopted, though it is not clear if these terminologies took hold and became commonplace.

From recent studies it is clear that individuals who exhibit transgender behaviour want to change aspects of their gender, but don’t necessarily want to permanently change their sex. That said, in 2010 the UK media still use the same definition for transgendered and transsexualism.

Some welcome the term owing to its inclusiveness and others abhor it for the same reason.” M6

Androgyne

There are two definitions for this term. One refers to the androgynous (sexual) aspect of males and females (see Intersex p68), the second to the androgynous aspect of masculinity and femininity (gender).

The latter definition is explained here, as it is often used, particularly in the UK and parts of the USA, where this community of people are called ‘Androgynes’. Other terms include: agendered, between genders, bigendered, gender fluid, gender-neutral, intergendered, non-gendered and pangendered.

Society has grown up within a sex and gender binary system. As we welcome the dawn of life we are assigned a gender governed by what genitalia we have. But there are those who see a whole spectrum of genders; individuals on the fringes of both the transsexual and transgender communities.

It relates to a person who’s gender transition isn’t 100%, e.g. a male who feels that they are both genders, a man and a woman at the same time, in comparison to a transsexual person who knows that they are totally the opposite gender, example: a male who feels she is a woman.

This divided gender mix can sit anywhere along the man/woman spectrum e.g. 70% man/30% woman or 30% man/70% woman.

Like any gender related issue, these feelings can be an extremely sensitive and emotional aspect of a person’s life and may not be divulged even to a partner, spouse or close family members.

While I have heard of three individuals with KS with this gender mix, because of my limited study so far, it is not clear how they view their situation (Androgyne, Transsexual or Transgendered person).

Two individuals have a female partner and while one is happy to be a man, the other lives and works as both a man and a woman. So, I can only really comment on the third case history... myself.

I had a slow gender shift, resulting in an 85% masculine/15% feminine shift. I am unsure if the gender shift was caused by a chromosomal or hormonal influence on my brain during gestation caused by the Klinefelter’s Syndrome or some other physical or psychologically unconnected factor.

While this diagnosis was not made by a healthcare professional, it does not detract from how I feel. And although I don't openly discuss my dysphoria, I can say that I have – and will continue to be, happy to maintain my man/male persona and to embrace my feminine side. My only regret was in not telling those closest to me until recently. I am heterosexual and previously married. (See A Personal Message p30).

Maybe others with Klinefelter's Syndrome hide their mixed gender feelings too. It would be intriguing to discover just how widespread this trait is within the KS community and what the mean percentage shift is, but sadly my efforts to find out have, understandably, yielded silence.

Sexual Orientation

This term relates to whether you are attracted to the opposite sex, your own sex, or both. In other words Heterosexual (Straight), Homosexual (Gay male or Lesbian) or Bisexual (Bi). Also, Pansexual and Asexual.

Currently the above terms are used to describe a person (e.g. “He is Bisexual.”).
However, it has been suggested that for clarity the terms should be used to describe behaviour, rather than people and to use non sex/gender terms which avoid the stigma that goes with terms such as ‘homosexual’ etc.

These relate to the erotic/love preference of an individual, that is:

- Androphillic = male loving (attracted to people with male sex organs);
- Gynoecophillic = female loving (attracted to people with female sex organs);
- Ambiphillic = both loving.

**Intersex**

**Please note:** See p52-55 for ‘Intersex’ definitions, or contact the AHN, AIS and CAH support groups for an extensive description of Intersex, relative to their own conditions. (Contact details p77-78).

The majority of this chapter was compiled in 2005 and does not take into account the Consensus Statement on Management of Intersex Disorders, published in 2006. Please see p55 for more details.

“Intersex (IS) groups are against shame, secrecy and non-disclosure. IS groups, adults and teens in particular, are also against surgery without informed consent, unless life-threatening circumstances due to both the psychological and physical damage to sensate and sexual function.

Parents and clinicians though still often opt for early surgery, as society is so unconforming of IS because it’s not understood.”

“Sometimes even specialist clinicians use the term ‘Intersex’ to mean ‘Ambiguous genitalia’ forgetting that they are excluding a large proportion of intersexed patients who have a complete female phenotype (and usually no sexual or gender identity problems). This causes us a lot of problems as the media pick up on this... conveying to the general public that gender identity is, of necessity, an issue in intersex which it is absolutely not.”

You may come across the terms ‘Hermaphrodite’ and ‘Pseudo-hermaphrodite.’

Medicine took on these terms in the 1800s. The name originates from a Greek mythical character which had a complete set of female and male internal and external reproductive organs, “which in reality is totally impossible.”

However, it is possible to be born with one ovary and one testis, or one organ with both ovarian and testicular cells (ovotestis). This is an extremely rare condition and can affect those with KS.

That said, some people (including those with KS) consider themselves – and are considered by others, to be ‘Androgynous’ and call themselves ‘Androgynes.’ This term applies primarily in Australia and parts of the USA.

The term ‘Hermaphrodite’ is very occasionally used in the USA and UK, but ‘Intersex’ is now the preferred term. “Most of our members detest these hermaphrodite terms.” “...we feel these terms should be banned from the medical literature.”

**Transvestism (or Crossdressing)**

Like many of the other terms this one can mean more that one thing.

The original term made by Magnus Hirschfeld (1910) identifies all orientations participating (i.e. straight, gay and bi). However, to most sexologists today the term refers to heterosexuals only.

Up to now the term has meant a person, usually male, who acquires erotic pleasure by wearing clothes of the opposite sex. While this will be true for some, in recent years many heterosexuals have disagreed with the latter definition, stating that autoerotic pleasure is not the motive, but a part of their personality which cannot be expressed in any other way.

That said, for some individuals there can be a sexual element to it; for example, feeling the texture of the clothes, or observing the reaction of other people.

Around the world there are societies in existence specifically for heterosexuals and their wives, with the Society of the Second Self (SSS) being the largest organisation of its kind in the US. The latter shows that the way people dress doesn’t always identify our preferred partner.

To postoperative transsexual people crossdressing is not generally sexually arousing as it is their typical everyday attire.

Females also cross-dress, however, the term transvestite is rarely applied, since females are able to wear male clothing far more easily. For instance, in business circles it is common to see female fashion depict grey pin-striped trouser suits & ties.

There are also those who cross-dress for theatrical purposes and they will be of any orientation.

The term ‘Hermaphrodite’ is very occasionally used in the USA and UK, but ‘Intersex’ is now the preferred term. “Most of our members detest these hermaphrodite terms.” “...we feel these terms should be banned from the medical literature.”

**In-depth information**

This chapter has been compiled with the invaluable assistance of the following people:

**MC** Miss M Cull, Adrenal Hyperplasia Network (AHN), UK.
www.ahn.org.uk

**MD** Prof M Diamond, University of Hawaii, John A Burns School of Medicine, USA. Some definitions used from: “Sex and Gender are Different: Sexual Identity and Gender Identity are Different,” and “What’s in a name? – Some terms used in the discussion of Sex and Gender.”
www.hawaii.edu/PCSS

**BR** Mr B Reed, Gender Identity Research & Education Society (GIRES), UK.
www.gires.org.uk/

**MS** Ms M Simmonds, Androgen Insensitivity Syndrome Support Group (AISSG), UK.
www.aissg.org/

**REFERENCES p11 CONTACTS p74**
The KS Story – You are not alone

Further Study

Addresses and Websites to Support Groups
Medical Specialists and Research Units

I hope that the following contacts help you to find what you are looking for; be it a few explanatory leaflets, someone to talk to on the phone or through correspondence, or understanding and help from another family going through the same things as yourselves.

I wish you every success with your studies, but like so much in life, be sensible about it. Just because there might be a Klinefelter’s Syndrome link to another medical condition, don’t automatically assume you’ve got it too. Even with the same karyotype everyone is different and will have vastly differing symptoms and traits.

If you feel really strongly about a subject, then talk to your GP. But please, remember that doctors have a very heavy workload, so be sure no one else can help you first.

When you do visit your GP take a written note of all your symptoms, explain how you feel, try not to leave anything out; you may regret it later, and if you want to talk about emotional things consider asking to see a counsellor.

Like your doctor, things you say to a counsellor are strictly confidential. Don’t expect quick results though, as it can sometimes take a number of weeks.

You may prefer to initially contact one of the support groups. They are usually run by people with the same condition and offer support and information. Support groups also have a louder voice than just one person, if you want to be heard.

Libraries

Take full advantage of your local library’s resources and the expert knowledge of the staff, who can also acquire published research for you to borrow. There may be a small charge for loans outside your region.

Most libraries offer Internet access. There is usually a maximum time limit (e.g. 1 hour) and there may be a charge for this service.

There is a small list of books and published research papers at the end of the contacts, which may be of interest to you. Support groups will be happy to advise you on publications for your particular condition.

Need to make an international phone call?

If phoning outside your own country, please remember to include the International Access Code of your own country; the Country Code for the place you are calling; the Area Code (if it has one) and the Local phone number.

For instance, to phone the UK, dial 00 44 before the Area Code. Examples: to phone Contact a Family, dial 00 44 020 7608 8700. You will have to drop the first zero of the Area Code (tinted). To phone Canada dial 00 1 first.

Every effort has been made to check out the following contacts. However, occasionally inter-group politics and other situations can arise and you can get the odd bogus organisation that slips through the net – so get into the habit of thoroughly checking out any group or individual; whether it is included here or elsewhere. Try obtaining information from two or three groups, for comparison. A good place to start is the FAQ and About us links on a group’s website.

Names and addresses can also change, as well as websites. I have included some contact names, which are preferable to ‘Madam/Sir’, but please remember people move on. Websites can also be in the process of being upgraded. Don’t give up though, go back to the site another day.

It is possible that like me, you will settle on your own favourite and trusted contacts; giving the best information for your particular condition. I have tried to include as many as possible to aid in your search. And hopefully once the book is circulated, more organisations will wish to add their contact details, which I can include at the next pdf update in 2012.

There are also some contacts which are included as a ‘thank you’ for their invaluable assistance with specific chapter facts and figures, and aren’t necessarily linked to Klinefelter’s Syndrome.

In most countries, particularly the USA, the word Klinefelter is used, rather than Klinefelter’s.
Website search tips

Unused to using the Internet? Here are some tips to get you started on your studies.

When you first access a particular website, quite often you can gain a good deal of valuable information from the FAQ page (Frequently Asked Questions).

If you are unable to access the information you require from a particular site, go to the Home page and if a ‘Search’ facility is available, type in key words within the field: e.g. ‘Klinefelter's Syndrome and Osteoporosis’, or variations. An ‘Advanced Search’ facility will give better results, with less chance of non-related material. Also look out for similar linked information (e.g. ‘Related articles’).

Specific paper titles or the author can be typed into a search field in a general search engine, though you would be better going to a reputable medical or support site. There are also specialised websites for finding specific organisations or people. Have a word with your local library staff for books on the Internet.

If a dialogue box states that the web address you asked for cannot be found, try experimenting e.g.:
.co.com (company) .org (organisation) .gov (government) .ac (academic) .edu .education) .net (network).

You may also have typed a dot instead of a dash.

There are going to be times when a particular site will link you to a related article site, and that to yet another site (ad infinitum). If you wish to return to your original site then add it to your ‘Favourite’ or ‘Bookmark’ folder.

If you have difficulty getting back to your original site, go into ‘History’ and click on the appropriate site.

USING GENERAL SEARCH ENGINES

Here are a more Internet tips for you to try.

‘Plain-English’ searches
The simplest search for beginners. For instance, to find other international Klinefelter’s Syndrome support groups, type a question into the field, e.g. where can I find swedish klinefelter syndrome support groups? Or non-specific: ...klinefelter syndrome support groups?

Note: Plain-English searches don’t work on all search engines.

Key words
To reduce the quantity of non-relevant information, in the search field try to use the most unique key words. Also try swopping around your key words, as some search engines take more notice of the first word.

Multi-step searching
If looking for a specific subject linked to KS, e.g. Diabetes, try a multi-step process. First, give it your best shot, then review the results of the first couple of pages picking out key words linked to diabetes, then use these words within your next search. Narrow the results by using ‘set searching’. To do this, most leading engines offer a ‘search within these results’ field.

Title words
Try using words that might be in the title of the document you seek, for example (starting with the search term): title:“klinefelter syndrome” (note, no space between colon and first quotes, and no apostrophe ‘s’ for US sites). For Google use the search terms allintitle: or intitle: The former term will look for all words in the title, while the latter any words. For Yahoo use t:

Case-sensitive searches
Always use lower-case unless searching for a person, location, book/paper, etc; then use initial capitals.

Spelling
Often a ‘no results’ outcome will be caused by poor spelling. ‘nough said!

FURTHER STUDY pdf LINKS TO WEBSITES

Addresses in two lines
Due to technical aspects beyond my comprehension (grrr), website links on the following pages which are in two or more lines, will not function. For these links, take a note of the address or adjust your web browser window so you can read the address link in the book, then manually add the additional lines of text to your browser’s search field.

Facebook and Twitter also don’t link.

No hash key?
Please note that some addresses use the hash symbol (#). For those people who use UK keyboards, you will not have this symbol, instead, type: Alt + 3.

The end of the KS story?

As long as there are people out there trying to understand Klinefelter's Syndrome; whether for research, support, treatment, or just you and I trying to understand how our mind and bodies function, then the KS story will continue.

Whatever you decide to do. However you decide to do it. I wish you peace of mind — and body.

And as you look through the following pages, remember...

You are never alone
Klinefelter's Syndrome

**UNITED KINGDOM:**

**BBC Health**

Klinefelter's Syndrome  
[UK]

Contact: Mrs Sue Cook  
Founder and National Co-ordinator  
56 Little Yeldham Road  
Little Yeldham  
Halstead Essex CO9 4QT  
All telephone enquiries:  
te: 0845 230 0047 (Local rate UK number)  
Family, child & young people enquiries:  
e: coordinator@ksa-uk.co.uk  
Adult enquiries:  
e: adults@ksa-uk.co.uk  
Information can be posted; please send an SAE (C5 or DL). Membership details on website.  
w: www.ksa-uk.co.uk  
Information and membership are available to those with Klinefelter’s Syndrome or its variants, their families, spouses/partners, friends and carers.

The KSA accepts that it is the individual's right to self-determine if they consider themselves to be intersex or not. This charity supports and respects, regardless of that individual choice, those with KS.

Forum:  
w: www.xxytalk.com

**Klinefelter Organisation**

PO Box 9969  
Colchester CO1 9FQ  
e: ask@klinefelter.org.uk  
Or use the Contact Us link on the website.  
w: www.klinefelter.org.uk

Valuable information and an opportunity to contact other KS people, including wives, partners and parents. Membership is available to all KS individuals from the UK and Ireland.

**Klinefelter’s Syndrome Association (UK)**

Contact: Mrs Sue Cook  
Founder and National Co-ordinator  
56 Little Yeldham Road  
Little Yeldham  
Halstead Essex CO9 4QT  
All telephone enquiries:  
te: 0845 230 0047 (Local rate UK number)  
Family, child & young people enquiries:  
e: coordinator@ksa-uk.co.uk  
Adult enquiries:  
e: adults@ksa-uk.co.uk  
Information can be posted; please send an SAE (C5 or DL). Membership details on website.  
w: www.ksa-uk.co.uk  
Information and membership are available to those with Klinefelter’s Syndrome or its variants, their families, spouses/partners, friends and carers.

The KSA accepts that it is the individual's right to self-determine if they consider themselves to be intersex or not. This charity supports and respects, regardless of that individual choice, those with KS.

Forum:  
w: www.xxytalk.com

**International:**

**Klinefelters Syndrome (CA)**

Contact: Stefan D Schwarz  
e: stefan13@cox.net  
w: www.klinefeltersyndrome.org

This website was created for boys, men and their families to have a resourceful place to get information. The website also acts as information for those learning about the condition for potential diagnosis or for research projects.

**Klinefelter’s Support Group of Western Australia**

14 Shetland Drive  
Morgan Fields Estate  
Henley Brook  
Western Australia 6055  
t: (61) 08 9296 8661  
e: kerrygavey@iinet.net.au  

**Turner Syndrome Support Society (UK)**

Contact: Beverly Searle  
Chief Executive Officer  
PO BOX 2189  
Caterham  
Surrey  
CR3 5GN  
t & f: +44 (0)1883 330766  
e: info@rarechromo.org  
w: www.rarechromo.org

An international group providing support and information to anyone affected by a rare chromosome disorder. Support & information for individuals or families. Rarer karyotypes, not 47,XXY.

**Trip-X Family Network Support Group**

32 Francemary Road  
London SE4 1JS  
t: 020 8690 9445  
e: helenclements@hotmail.com

This is a support network started in 1997, offering support to families of children with karyotype 47,XXX. It publishes a biannual newsletter and has information available on request. There are over 360 families in the UK and abroad in touch with the network.

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**REFERENCES p11**

**Further Study**

An important aspect of the Klinefelter’s Support Group is to provide support, information and resource materials about Klinefelter’s syndrome to affected individuals and families in Western Australia.

It also endeavours to provide health professionals and the community with information about our Support Group and Klinefelter’s syndrome.

**International: 47xxy.com**

Contact: Stefan D Schwarz  
e: 47xxyorg@gmail.com  
w: www.47xxy.com

**Klinefelters Syndrome (CA)**

Contact: Stefan D Schwarz  
e: stefan13@cox.net  
w: www.klinefeltersyndrome.org

This website was created for boys, men and their families to have a resourceful place to get information. The website also acts as information for those learning about the condition for potential diagnosis or for research projects.

**Klinefelter’s Support Group of Western Australia**

14 Shetland Drive  
Morgan Fields Estate  
Henley Brook  
Western Australia 6055  
t: (61) 08 9296 8661  
e: kerrygavey@iinet.net.au  

**Other Sex Chromosome Conditions**

**UNITED KINGDOM:**

**Unique**

Rare Chromosome Disorder Support Group  
Contact: Beverly Searle  
Chief Executive Officer  
PO BOX 2189  
Caterham  
Surrey  
CR3 5GN  
t & f: +44 (0)1883 330766  
e: info@rarechromo.org  
w: www.rarechromo.org

An international group providing support and information to anyone affected by a rare chromosome disorder. Support & information for individuals or families. Rarer karyotypes, not 47,XXY.

**Triple-X Family Network Support Group**

32 Francemary Road  
London SE4 1JS  
t: 020 8690 9445  
e: helenclements@hotmail.com

This is a support network started in 1997, offering support to families of children with karyotype 47,XXX. It publishes a biannual newsletter and has information available on request. There are over 360 families in the UK and abroad in touch with the network.

**Turner Syndrome Support Society (UK)**

Contact: Beverly Searle  
Chief Executive Officer  
PO BOX 2189  
Caterham  
Surrey  
CR3 5GN  
t & f: +44 (0)1883 330766  
e: info@rarechromo.org  
w: www.rarechromo.org

An international group providing support and information to anyone affected by a rare chromosome disorder. Support & information for individuals or families. Rarer karyotypes, not 47,XXY.

**Triple-X Family Network Support Group**

32 Francemary Road  
London SE4 1JS  
t: 020 8690 9445  
e: helenclements@hotmail.com

This is a support network started in 1997, offering support to families of children with karyotype 47,XXX. It publishes a biannual newsletter and has information available on request. There are over 360 families in the UK and abroad in touch with the network.

For XYY Syndrome support and rarer KS karyotypes, some of which are now considered separate conditions, please contact Unique or try a web search.

**REFERENCES p11**

**Further Study**

An important aspect of the Klinefelter’s Support Group is to provide support, information and resource materials about Klinefelter’s syndrome to affected individuals and families in Western Australia.

It also endeavours to provide health professionals and the community with information about our Support Group and Klinefelter’s syndrome.

**Klinefelter Syndrome (AU)**

Support Group (KSSG)

We have phone support, email support, discussing issues around Klinefelters, we give referrals to GPs & Specialists. We offer 1 on 1 chats. We seek out information for clients on other matters to do with their condition.
Society for Endocrinology (UK)
22 Apex Court Woodlands Bradley Stoke Bristol BS32 4IT UK
Healthcare professionals only
General enquiries: t: +44 (0)1454 642200
Publications: t: +44 (0)1454 642220
Conferences & Events: t: +44 (0)1454 642220
Fax: +44 (0)1454 642222
e: public@endocrinology
w: www.endocrinology.org
w: www.yourhormones.info

Please note. In the UK referral to an endocrine clinic is through a General Practitioner.

GENETICS:
Genetic Alliance UK
Unit 4D Leroy House 436 Essex Road
London N1 3QP
t: +44 (0)20 7770 3141 f: +44 (0)20 7359 1447
e: mail@geneticalliance.org.uk
w: www.geneticalliance.org.uk

GENERAL MEDICAL:
Department of Health
Customer Service Centre Richmond House
79 Whitehall London SW1A 2NS
t: 020 7210 4850 textphone: 020 7210 5025
Opening hours: Monday to Friday 8.30am-5pm
w: www.dh.gov.uk

National Institute for Health and Clinical Excellence (NICE)
Level 1A City Tower Piccadilly Plaza Manchester M1 4BD United Kingdom
t: 44 (0)161 870 3133 f: 44 (0)161 870 3133
e: nice@nice.nhs.uk w: www.nice.org.uk
The National Institute for Health and Clinical Excellence (NICE) provides guidance, sets quality standards and manages a national database to improve people’s health and prevent and treat ill health - regardless of where they live in England and Wales. [See p48 for Northern Ireland and Scotland].

Professor Milton Diamond PhD
University of Hawaii John A Burns School of Medicine Department of Anatomy and Reproductive Biology Pacific Center for Sex and Society 1960 East-West Road Honolulu Hawaii 96822 (USA)
t: (808) 956-7400 f: (808) 956-7922
e: diamond@hawaii.edu w: www.hawaii.edu/PCSS
Professor Diamond has published a prolific amount of material on sex and gender issues and lectures widely. More details on website.

MedicineNet
w: www.medicinenet.com
Comprehensive articles on health and medical topics produced by US Board Certified Physicians. Go to: w: www.medicinenet.com/sitemap to read the main article on Klinefelter Syndrome.

National Center for Biotechnology Information
A highly recommended site for information on KS and other associated conditions. For research papers use the Search field to find specific authors or titles (will access PubMed and other databases). For MedlinePlus ('Health Topics' (KS definitions and general health issues), 'Medical Encyclopedia' and 'Medical Dictionary'). Go to National Library of Medicine at: w: www.nlm.nih.gov

REFERENCES p11
You are not alone

INTERSEX contd:
Chris Somers xxy (AU)
M.Ed. by Research (UWA); B.Ed. (Melb); Hons.
Diploma of Creative Photography (Trent Poly, now
Nottingham Trent University, UK)
E: Sarkay@westnet.com.au
A Board member of the Organisation Internationale
des Intersexuels:
Please see p77 for a research thesis and one paper
by Chris Somers xxy.

Congenital Adrenal Hyperplasia
(UK)
Group
Contact: Mrs Sue Elford
CAHG Chairperson
2 Windrush Close Fliitwick Bedfordshire MK45 1PX
T: 01525 717 536
E: webmaster@diningwithcah.com
W: www.cah.org.uk
Provides support, newsletters, information and meetings
for families and people with CAH. They also aim to
raise awareness of the condition with the general
public and the medical profession.

LEGAL SUPPORT:
UK and International
I suggest that for England & Wales, Northern Ireland
and Scotland, that you pop into your local
ChildLine’s Advice Bureau for information on contacting
a law society or a lawyer/solicitor. If you are in another
country, try your equivalent public advice service.

OSTEOPOROSIS:
National Osteoporosis Society
(Camerton Bath BA2 0P)
T: 01761 477771
E: info@nos.org.uk
W: www.nos.org.uk
The National Osteoporosis Society is the only
UK-wide charity dedicated to improving the diagnosis,
prevention and treatment of osteoporosis.

PLANNING A FAMILY:
Please discuss options with your GP.

SOMEONE TO TALK TO:
ChildLine
ChildLine is the free helpline and online service for
children and young people in the UK. Children and
young people can call or visit:
T: 0 800 1111
W: www.childline.org.uk
to talk to a trained counsellor about any problem, or
talk to other young people on the message boards.
(See p56 for more details).

SOMEONE TO TALK TO:
Specifically about Bullying
BullyingUK
Formally Bullying Online
10C Mornington Terrace Harrogate North Yorkshire
HG1 5DH
E: help@bullying.co.uk W: www.bullying.co.uk
Children
Helping to keep children safe from bullying
and abuse
2 Grosvenor Gardens London SW1W 0DH
T: 020 7733 7300 F: 020 7733 7081
E: info@kidscape.org.uk
W: www.kidscape.org.uk
Send an A4 SAE with First Class stamps for free
booklets and leaflets on bullying and personal safety.
Runs one-day ZAP courses for children who have
been severely bullied.

EVERYONE!
For this first edition of the book, I have tried to
include as many of the main information,
advise and support groups as possible, but
there are many more out there in countries
around the world; specifically for
Klinefelter’s Syndrome and its variant
karyotypes, and for associated conditions.
Many of the contacts in this chapter
include on their websites, useful links to
organisations around the world.
They also have resource links to genetic
groups, conference details, support group
campaigns, current research, activities
to get you out the house and meeting other...
If you are a support group committee
and would like your group’s contact details
included in the next pdf update in 2012,
please get in touch:
E: theksstory@btinternet.com

BOOKS & RESEARCH PAPERS:
A case of Human Intersexuality having a possible
XXY sex-determining mechanism
Paper by: Patricia A Jacobs and Dr J A Strong
Androgen Insensitivity Syndrome and Klinefelter’s Syndrome:
sex and gender considerations
Chapter within book
As Nature Made Him - The boy who was raised as a girl
(Perennial, 2001)
Book by: John Calapinto
Edinburgh study of growth and development of
children with sex chromosome abnormalities
Paper by: Dr Shirley G Ratcliffe
John Wiley and Sons, 1991
Contained within a paperback
of research papers, titled: Children & Young Adults
with Sex Chromosome Aneuploidy.
Atypical Gender Development – A Review
Article (International Journal of Transgenderism, to be published
by the Hayworth Press Inc).
Can be viewed on the Gires.com website:
W: www.gires.com
Gynecostasia – A Medical Dictionary, Bibliography
& Annotated Research Guide to Internet References
by: Milton Diamond, PhD and Linda Ann Watson, ME
(© Elsevier Inc. 2004)
Volume by: Dick S Fwaab
Chapter 24.4 deals with ‘Klinefelter’s Syndrome or
Testicular Druagenesis’. Additionally, chapter 24.5 covers
‘Sexual differentiation of the brain and sex behaviour.’
(See please p74 for contact information).
Intersex in the Age of Ethics
(Perennial, 2001)
Book by: Alice Domurat Dreger
Klinefelter’s Syndrome
Paper by: Dr Shirley G Ratcliffe
An Introduction to Klinefelter’s Syndrome, for parents,
adolescents and adults. Available from the Klinefelter’s
Syndrome Association (UK) (See p72).
An important message

2010

It's one thing to spout facts & figures, but another matter to actually live every moment with Klinefelter's Syndrome. And you can help. No matter what your association with Klinefelter's Syndrome, you can make a huge difference to someone's life – by telling your story. You will be communicating directly with those in a similar situation, who can take strength from how you cope living each day with Klinefelter's Syndrome.

Welcome to ‘The KS Diaries’

This second book (pdf) will contain histories supplied by people who have Klinefelter's Syndrome or in some way are involved with this condition. It is their story; their account of life; or a moment in time.

Wherever you live on this blue planet, if you feel your Klinefelter's Syndrome story can make a difference to others, then please think seriously about contributing. And I'm not just talking about those that have Klinefelter's Syndrome. You can be a parent, parents-to-be, spouse, partner, brother or sister, grand-parents, friend, healthcare professional or work with children and young adults, and you too can make a huge difference to ease the mind of those in similar circumstances.

‘The KS Diaries’ will be dedicated to those courteous people who, through strength of spirit, tell their story in the hope of helping others with Klinefelter's Syndrome.

Guidelines for safely creating & sending your manuscript

Whether you experience Klinefelter's Syndrome personally or see KS from a professional viewpoint, if you wish to submit your KS story, it is vital that you please contact me first to receive guidelines (pdf) in safely creating and sending your manuscript. If you do not do this I cannot include your story.

Total anonymity

Authors' stories will NOT include personal names or contact details. Due to this anonymity, and the frank and sensitive nature of some stories, proofs will not be sent to authors for final checking. I will do my utmost to ensure accuracy, but will not be held responsible for errors.

Author's written permission

While all stories within the book will be anonymous, I will still need you to include written permission with your manuscript, stating that you are happy to have your story included in the book (pdf), and that includes those authors who have already sent me their manuscripts.

For Guidelines in safely creating & sending your manuscript (pdf):

theksstory@btinternet.com

Authors’ manuscript, written permissions and any personal details will be filed in a secure location. Please note, I will not include your story on the pdf without your written permission.

Non-automatic inclusion

I cannot guarantee that your story will be accepted for the book (pdf). There will be various reasons for this. For example, extreme language or descriptions which are far too graphic, will be rejected, as individuals as young as 11 could be reading it. I appreciate that you want to get your story across as you experienced and felt it, but please keep the latter example in mind. The guidelines will give you more details on this.

Non-profit venture

Like The KS Story, there is no money involved in this second book (pdf); I and story authors will not receive royalties or other rewards, nor is there a fee for inclusion in the book. Also, like the first book, any donations must be sent directly to specific KS support groups and allied organisations to assist in their educational projects. Do NOT send monies to me.

Please remember, this book will not happen without people like you! If I don't receive enough interest the book will be shelved – and that's not a joke.

Got 5 minutes...

I have already received 10 stories and begun to create ‘The KS Diaries’. Please take a moment to read the following pages. I hope they will galvanise you into action, to open your heart and tell your own KS Story.

Thank you so much.

Iain W McKinlay

Iain W McKinlay

xxy
A book is born

Four years on from my diagnosis, signed off work and gathering dust, at the tender age of 45, I had the bright idea of utilising the data I had collected from my KS studies to create a small pamphlet for my GP’s practice. It was a way to exercise my brain, keep my graphic design skills honed and give something back for all the medical help I was receiving.

As contacts in the UK and around the world began expressing an interest in my wee venture, the pamphlet took on a life of its own; taking on more subjects and more pages. Then it suddenly dawned on me that ‘he who can’t spell’ was now writing and compiling an actually book!

I then thought it would be good to break up the masses of data with snippets of my own KS tale, hence the title: ‘The KS Story’, but one of my advisors strongly advised against mixing facts & figures with personal stories. And so ‘The KS Diaries’ was conceived.

Here we are again!

In 2005, in the midst of finding a publisher for The KS Story a major contributor and supporter of the book suggested that adding personal histories would immeasurably improve it, and so I tentatively asked for stories from UK KS support group members and received ten initial manuscripts. I then developed another health issue and The KS Diaries was placed on hold.

The next Booker Prize?

During my studies into KS I was shocked to read that a substantial percentage of couples around the globe choose termination when given prenatal test results that their child has Klinefelter’s Syndrome.

And that dear reader, for all of us with Klinefelter’s Syndrome, and for couples on the verge of parenthood, for consultants and for those that see us as a waste of space, to take heed. If only one thing is learnt from creating The KS Story, it is that we are all different – even those with the same karyotype and that given the opportunity, the vast majority of us are capable of such amazing feats. Indeed some of the stories I read from other people with KS were eloquent, poignant and powerful pieces, which touched me greatly.

Despite academic shortfalls many people with KS go on to become managers, executives, teachers; gain higher education and degrees; start their own businesses.

Barring acute physical or psychological traits and symptoms that could seriously affect quality of life, we have as much right as any typical person to exist and to contribute to the world around us.

Selfless courage

As the following stories are about Klinefelter’s Syndrome, most contributors are focusing on specific events within a lifetime.

Like myself, I would imagine that a large proportion of these people have had joyous times in their life, with loving parents, siblings, spouse/partner, friends, and have achieved remarkable things to be proud of.

While sadly not for everyone, it is truly inspiring how many people who have been bathed in Klinefelter’s presence; who have run the gauntlet of symptoms, bullying, fear, self-loathing, surgery and more, emerge (perhaps not a stronger person in their own eyes), more forgiving and able to not only talk about their experiences but to help others in need.

It is never easy to bare your soul, even if trying to help; letting closely guarded emotions into the open. Some brave souls are still in pain, yet manage to support and advise others. A selfish act of courage in anyone’s book.

Nearest and dearest

But dealing with a diagnosis of Klinefelter’s Syndrome isn’t just the preserve of the patient. Parents, family members, partner, spouse, also have to cope with this revelation and the associated aftermath.

Whether diagnosis is made prenatally, during childhood, adolescence or in adulthood, parents feel responsible for their offspring (I’m 58 and 16 stone yet I’m still my mother’s wee boy). Even when told the facts of Klinefelter’s Syndrome parents may still blame themselves.

After an amount of time a spouse or partner almost become one entity; you know each other inside out, you finish each other’s sentences, you become comfortable together. Then with diagnosis enters hormone replacement therapy and all hell breaks out!

It’s a kind of puberty all over again; a maelstrom of emotions, with social and sexual experimentation; increased body and facial hair; personal expectations...

We often forget that it is the parents and/or spouse/partner who have to be strong and supportive for their loved one despite a whirlpool of emotions swirling around in their own head. Physical and emotional changes in the person with KS can put a great strain on a family, marriage or partnership, particularly for older couples who may find it difficult to talk about innermost feelings and sexual issues.

As a responsible adult I thought I could cope with male hormone replacement therapy. WRONG! No matter your age, life experience, sensibilities, responsibilities, go sailing out the window when hormones are involved – and those closest to you are generally the first to get the flack.

There is also the prickly question of fertility and how couples cope. Tests; anger; counselling; self-doubt; hopelessness; it can tear couples apart. But thankfully with love, support and a degree of give and take, it can also bring them closer. And remember, KS and sterility aren’t carved in stone.

Peace of mind

Sometimes just knowing that you’re not the only one in the world going through something is often a comfort in itself. And when you can read other people’s accounts of how they reacted and how they dealt with a particular situation I think you’ll agree, it truly brings home just what living with Klinefelter’s Syndrome is all about.
The KS Story –
You are not alone

Closing thoughts

It seems ironic that someone who has such difficulty with academia should end up working with text. However, as a graphic designer I see the beauty and form of letters, words, phrases, from a different angle; a technical perspective, as individual entities – compare and balance the spaces between them. In some cases, components of a story I will never know. I understand and see the true meaning hidden in the characters – someone’s education, someone’s life, in shapes.

Eyes darting back and forth from manuscript to monitor, the contributor’s story in abstract form. Afraid to miss punctuation, afraid to misspell; to, too, bare, bear, never picked out by the spell checker. And ever the perfectionist, tom whether to change the contributor’s single to double quotes – but stopping short, realising in time that it’s their content; their learning, their story.

A pitifully slow reader all my life, packets of words are inwardly repeated, filling my head until the computer keys are struck.

Back and forth from manuscript to monitor; light between the dark, fear and joy behind the words.

OK, perhaps not a poet, but I am a graphic designer to my bones. I trust and revel in my skills as an artist. 43 years of graphics experience, yet I have no confidence selling that work... and I have Klinefelter's Syndrome.

Having read the stories sent to me for this second book, many of whom have the same karyotype as myself, my story does not compare. In a world of pain and suffering, both past and present, to individuals and social communities, my own KS story is nothing. It is the pinnacle of the tip of the iceberg in terms of physical and psychological complexity and severity.

But these pages aren’t about who is worse than the other, but about making known the many facets of Klinefelter’s Syndrome, no matter what the karyotype, or whether you are the person with KS, a parent, parents-to-be, partner, spouse, sibling, grandparent, friend...

It’s about lifting a child from the darkness of loneliness and fear. It’s about shedding light on prejudice and misinformation.

It’s also how everyone associated with Klinefelter’s Syndrome copes with the diagnosis: guilt, anger, fear, doubt, even a sense of bereavement as treatment begins and physical and emotional changes occur which, to varying degrees, can affect an individual’s character, perhaps by growing a beard or by sexual experimentation.

But equally it’s about understanding, love, support, caring. It’s about sharing all those experiences in the hope that others who mirror that life can benefit in some way.

Many with Klinefelter’s Syndrome never tell their own story or discover the stories of others, suffering their lot relative to their own lives; how family, friends and work colleagues interact and treat them. Locked in their own experiences of life.

But often by telling your story it is somehow comforting, often lifting something out into the open is spiritual, a kind of therapy. And that’s not my words, the latter comes from people who have already contributed their KS story.

Whether you look back on 3 score years and ten or look to the future of adolescence and adulthood with trepidation, I hope these personal accounts have helped to ease your worries; to show that you are infinitely more than your atypical parts, to motivate and encourage you to go out there and make your own unique mark on the world.

Throughout the past 12 years of ill-health, when I felt burdened by pain, self-pity, doubt and fear for my future, several lines of text reinforced my will to persevere and gave me courage to overcome those glitches in my life. In a way the words are also relevant to us folk associated with Klinefelter’s Syndrome, as individuals and as a social community...


REFERENCES p11 CONTACTS p74
The person who created this guide is The Compiler.
Any person obtaining this guide is The Reader.
Any individuals or organisations who are involved in medicine, research or studies, support & information or caring etc., are The Specialists.

Disclaimer

To the Reader

General information:
The Compiler is not an expert regarding this condition and does not offer medical advice. He will also not debate the rights and wrongs of any particular medical procedure. Unless in quotes, the content of this book is The Compiler’s opinion or assumptions drawn from other sources during his own studies. Having absorbed and discussed the ethics, risks, advantages etc with your consultant and/or GP, genetic counsellor and ideally a support group, it is for The Reader to choose the most appropriate course of action. The information contained within The KS Story is not a substitute for professional health care and is supplied for guidance only.

If The Reader suspects they may have Klinefelter’s Syndrome, or they know of someone who may have it, they should contact their GP, an endocrine consultant, or one of the appropriate Specialists in the Further Study chapter, for more information relating to their own specific needs.
The Compiler has endeavoured to make this guide as accurate as possible and every effort has been made to verify information supplied. However it should be noted that research is ongoing and facts do change, also subtle differences in research data can occur from various sources, and depending on which country the information is sought. Besides seeking the editorial advice from various research individuals and groups, The Compiler has generally taken the most widely held view or beliefs on current research. But not everything within these pages is so clear cut.

Further Study chapter:
Please remember, Klinefelter’s Syndrome is a highly variable and complex condition (even within the same karyotype), and this guide is but an overview. It is up to The Reader to seek their own viewpoint and up-to-date facts, through further study of their own particular condition.

Every effort has been made to check out the contacts and supplied details within this book and to obtain only reputable sources. However, from time to time inter-group politics and other situations can arise, and very occasionally a bogus organisation can slip through the net creating confusing or conflicting facts. It is therefore strongly suggested that The Reader get into the habit of verifying the qualifications and experience of individuals and support groups. An idea of this can be gained from their websites, from the FAQ or About us links. Try obtaining information from two or three groups and seek recommendations whenever possible and appropriate.

Important:
Please note, all Further Study contacts have kindly given their approval for inclusion within that chapter; in writing.
The inclusion of these contacts are for the convenience of The Reader. The Compiler does not endorse any particular service or product, nor any particular group or individual. He is however, a full member of two British KS support groups. Also, there are some contacts within these pages who do not endorse The KS Story guide.
The Compiler does not take the creation of this guide lightly; in that he appreciates that he is dealing with other people’s lives.
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Further Study chapter:
This guide has been specifically created to act as a conduit to The Specialist. All these extracts and contacts are here to assist and encourage The Reader to seek out such experts, in order to understand their own situation relating to Klinefelter’s Syndrome.

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