



University of the
West of England

bettertogether

**The psychological and social impact of
Klinefelter's Syndrome:
Report for the
Klinefelter's Syndrome Association**

Authors: Marianne Morris, Sue Jackson
& Jude Hancock
University of the West of England

September 2009

eXtra

Contents

Executive Summary	page 2
Introduction	page 8
Study 1: Interviews	page 10
Aim	page 10
Method	page 11
Results	page 11
The Themes	page 13
Theme 1: Diagnosis & Management of KS	page 13
Theme 2: The “Self”	page 17
Theme 3: Me, KS & Others	page 21
Core theme: Emotional impact	page 30
Study 2: Questionnaires	page 33
Aim	page 33
Method	page 33
Collected data	page 33
Results	page 36
Part 1: Whole group	page 36
Part 2: Group differences	page 45
Conclusions and implications	page 50
Recommendations	page 52
Dissemination	page 53
Acknowledgements	page 53
References	page 54
Appendix 1 List of medical conditions reported by research participants	page 56

Executive Summary

Aims:

In collaboration with the Klinefelter's Syndrome Association (KSA) we agreed to plan and deliver an impact study. This comprised two elements: an interview with KSA members and a questionnaire aimed at helping us to address the following areas:

- Participant's experiences of having KS, and where they feel they have been most in need of information and support.
- The impact of KS as a condition affecting the whole of life, including quality of life issues such as the impact of KS on an individual's ability to work, their finances, relationships, etc.
- Identifying the most appropriate services and approaches needed in delivering support to individuals with KS and their carers in response to their identified needs.
- To disseminate the results of the research, on behalf of people with KS, with a view to influencing policy and/or improving service provision.

How we did it:

We carried out two research studies. The study materials were designed working in collaboration with the KSA Committee (ie study 1: interview schedule; study 2: questionnaire selection and elements to cover in the demographic and physical profiling sections).

Study 1 was a qualitative project exploring the participants' subjective experiences. Thirteen participants took part in one-to-one interviews, and six took part in a focus group. Data from the questionnaire study were also included. These comprised the 41 answers to the open ended questions from the questionnaire and 36 critical incidents (where study participants wrote about a time when KS had been a significant problem for them).

Study 2 was a questionnaire project designed to objectively measure the psychological distress and quality of life of the participants. As well as demographic information on study participants, we also undertook physical profiling and collected medical information relating to the symptoms of KS they were experiencing and any other medical conditions they might have. Four standardised questionnaires were employed: the Hospital Anxiety & Depression Scale, the Derriford Appearance Scale (measuring levels of social anxiety and social avoidance, the short form of the World Health Organisation Quality of Life measure

(WHOQoL-Bref) and the Rosenberg Self Esteem Scale. We also included an amended version of the Honey & Mumford Learning Styles questionnaire. In order to explore the impact of KS and its treatment on psychological adjustment and daily functioning more broadly, five Visual Analogue Scales were included. Three hundred people from the Members list at the KS Association were contacted by letter and sixty-two people responded, giving a response rate of 21%.

What we found:

Study 1: The experiences of the men with KS that we interviewed were unique, however there were some similarities in their accounts, for example, their experiences in relation to low levels of testosterone. Differences appear to relate to when they received a diagnosis; the greater the delay (40+ years of age) the greater the risk of long term health problems and disability; the earlier a diagnosis the sooner the access to testosterone and the greater the understanding of KS. Equally their personalities, the understanding and reactions of significant others (for example, parent's and siblings), and life events appeared to influence the impact KS had on each of their lives. Seven minor themes emerged from the data, which were common to all stories. These were: 1. *Diagnosis*; 2. *Testosterone/The Treatment*; 3. *Health care/health problems*; 4. *Appearance*; 5. *Self identity*; 6. *Relationships*; and 7. *School and Education*. Three major themes were identified: 1. *Diagnosis and Management of KS*; 2. *The Self*; and 3. *'Me, KS and others'* with all three relating significantly to the effects that low testosterone has had. During the analysis a core theme emerged of "***Emotional Impact***" which was central to each of the three major themes. The emotional impact of KS, for some men, has been significant and as such has defined them as a person and influenced the life choices they have made.

Study 2: 60% of study participants were reporting clinical levels of anxiety and 34% had clinical levels of depression (as measured by the Hospital Anxiety and Depression Scale). Depression was significantly worse if men reported it as a symptom of KS. Individuals with low level self-esteem had increased levels of general anxiety and depression as well as raised levels of social anxiety and social avoidance and reduced quality of life. The results also suggested that levels of worry about KS and the individual's perceived severity of their condition may have a part to play in the wider impact of the condition. However, it should be remembered that KS is a syndrome and that for those individuals who have very few, or no symptoms, then the reverse pattern would be true. So, high levels of self-esteem were associated with low levels of general anxiety and depression, low levels of

social anxiety and depression and increased quality of life. The key may be the extent to which an individual perceives themselves to be affected by the condition.

This may change as men with KS age since the data suggests that older men reported more symptoms associated with having KS. It was interesting to note the difference between the symptoms the study participants recognised as being part of their KS and the list of symptoms that the medical community recognise. For instance, mood swings, headaches and fatigue are not usually mentioned in the medical research literature. Data suggests that the symptoms associated with KS can have a significant negative impact on levels of psycho-social functioning, particularly in relation to quality of life. These data suggest that the impact that KS can have on men's genitalia in terms of size inhibition may have a significant psychosocial impact, in particular in relation to penis size.

Results from the learning styles questionnaire indicated that half of the study participants preferred to have time to reflect on new information (ie they had a preferred reflective learning style) and 63% preferred information presented to them in a written format.

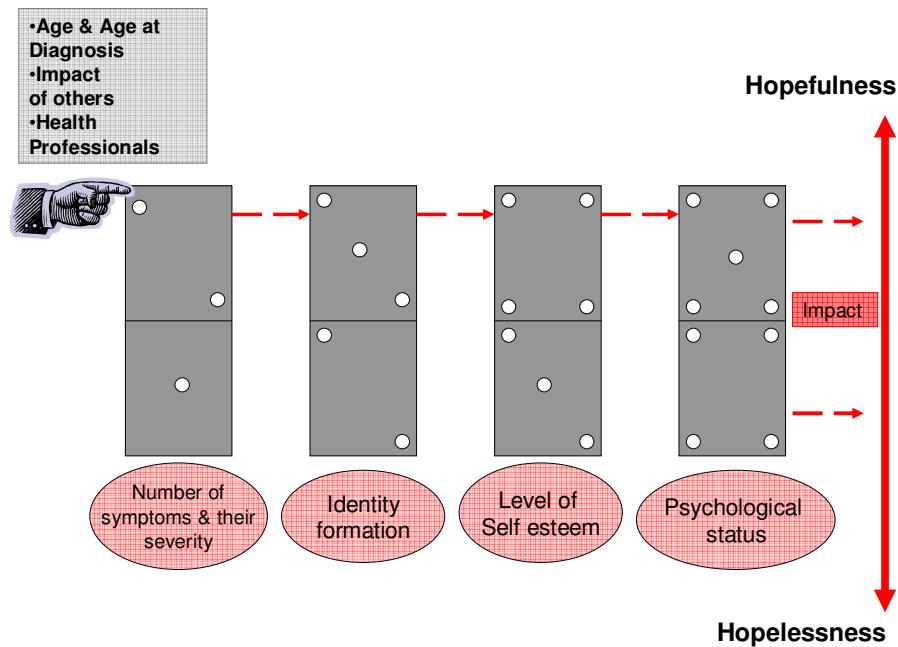
Conclusion and Implications:

Findings highlighted a number of concerns and identified the unmet psychosocial needs of men with KS.

- Diagnosis can take a long time and the prolonged lack of testosterone can have far-reaching negative effects for the individuals concerned.
- There were a significant number of people with KS who might do better in the healthcare system if information was presented to them in a written format, with time allowed for them to consider it before talking it over with a healthcare professional. It would certainly suggest that the current NHS climate of short appointments and limited written information may be disadvantaging some of the individuals who took part in this study.
- The psychological impact of the condition seems to hinge around the number of symptoms and the degree to which an individual perceives that they are affected.
- There are more symptoms associated with KS than the medical community necessarily recognise and these can have a profound impact on the psycho-social functioning of the affected individuals.
- Identity formation seems to be difficult for some of the men with KS and support in coming to terms with this seems to be largely absent.

- Low self-esteem was associated with increased levels of general anxiety and depression, raised levels of social anxiety and social avoidance and reduced quality of life. The reverse was also observed, where high self-esteem was associated with reduced levels of general anxiety and depression, low levels of social anxiety and social avoidance and high ratings of quality of life.
- Depression and anxiety in men with KS needs treating appropriately. Depression was significantly worse if the men reported it as being a symptom of KS. Attribution of poor psychological functioning matters, possibly because if it is a symptom of a permanent condition then there is no hope of avoiding its impact.
- KS is clearly a syndrome since experiences were unique and significant for each of the men who took part in the research. The emotional impact of the condition lay along a continuum from hopefulness to hopelessness where an individual's age, degree of family support, the attitudes and understanding of others, level of self-esteem, time of diagnosis and implementation of testosterone replacement influenced the men's location along the continuum.
- The impact of KS can be described in terms of a "domino effect" (see Figure 1 below) where "symptoms", "identity", "self-esteem" and "psychological status" are the dominos. These dominos may start to topple as a result of age, reactions from other people or as a result of experiences with healthcare professionals. As an illustration, if your first sexual experience is not good, then this might start to topple the dominos. You might suddenly realise that your penis size is a problem, which then might start you questioning your future identity as someone's partner, this can make you wonder if you have any value to others which can lead to a severe reduction in self-esteem. Low self-esteem is strongly associated with poor psychological status which can lead to a general feeling of hopelessness and thoughts such as, "Why should I bother?"

Figure 1: The domino effect



Recommendations:

- Men with KS would benefit from emotional support and one to one counselling at specific stages throughout their journey, beginning at diagnosis continuing through treatment and on to the continued and constant daily management of the condition.
- Depression and anxiety in men with KS requires treatment. It should be noted that depression is significantly worse if men see it as a symptom of KS.
- A review of group support is recommended to address the specific emotional needs across the lifespan, for example, fertility and sexual relationship issues for young adults.
- Increase awareness and understanding of the condition and its psychosocial impact both within the general population and across the medical community, in particular all non-endocrine specialist staff. Social workers and staff involved with employment and education services at all levels should also be targeted as part of any awareness-raising activities.
- KSA to generate a one page information sheet that men with KS can give to healthcare professionals.
- The provision of information to men with KS requires careful consideration. The majority of men in this study would probably do better with information presented in a written format with time to consider it before talking it over with a healthcare professional.

- Training for healthcare professionals to enable them to better understand and communicate with patients on certain sensitive issues that may need to be discussed such as size of genitalia and breasts, infertility and the impact of these.
- To lobby for GP training to include genetic problems and how to recognise these.
- To get the NHS website updated to show how the impact of KS is very different for each person.

Introduction

Background

A recent literature review identified a paucity of research exploring the psychosocial needs of individuals with Klinefelter's Syndrome (KS) even though biomedical researchers and condition specialists continually acknowledge the psychological impact these conditions have and the need to address these issues (Simm & Zacharin, 2006; DeLisi et al, 2005; Geschwind & Dykens, 2004; Swanson & Stipes, 1969).

The medical reports and reviews infer individuals affected by these conditions are visibly different and likely to be affected by their appearance (Visootsak & Graham, 2006; Ratcliffe, 1999). This visible difference results in increased social stigma, the potential for bullying in young people and increased psychological morbidity (Rumsey & Harcourt, 2005; Walters, 1997). Adolescence is a time of identity formation, when the individual strives towards personal autonomy (Durkin, 1995), the presence of social and physiological challenges at this time have the potential to generate greater disturbances in self-concept than at any other point in the life-cycle (Sweeting & West, 2003). Klinefelter's Syndrome can pose an additional threat to an individual's already fragile self esteem, potentially increasing the risk of social isolation, and resulting in psychological distress (Belzeaux & Lancon, 2006; Ablon, 2000; Rickert et al, 1996; Walters, 2007; Rumsey & Harcourt, 2005).

As well as resulting in visible difference, KS is reported as having the potential to affect intelligence, behaviour and personality (Visootsak & Graham, 2006). Individuals with KS tested using standardised measures of intelligence are reported as having a broad band of scores, from well above average to well below average (Rovet et al, 1995 in Visootsak & Graham, 2006). In terms of personality, Visootsak & Graham note that most men with KS are described as "quiet, sensitive and unassertive" and attribute language difficulties as contributing to difficulties in social situations.

The person with KS does not live in social isolation and the impact on their families may also be significant and may involve a high emotional and social cost. How then do individuals with KS negotiate these demands, and what are their priorities regarding their condition and its management?

Concerns identified by the KSA include:

1. Diagnosis is often slow, simply because a GP will see so few cases during the course of his or her career. Even when diagnosed, many patients remain isolated and distressed and find that their quality of life is greatly diminished. In addition, associated hormonal changes frequently cause psychological and psychiatric problems.
2. Infertility and its management, access to which is controlled by the GP.
3. Gynaecomastia which, like infertility, can affect body image and self-esteem.
4. Impact of the condition on relationships, in particular sexual relationships.

Aims

In collaboration with the Klinefelter's Syndrome Association (KSA) we agreed to plan and deliver an impact study. This comprised two elements: an interview with KSA members and a questionnaire aimed at helping us to address the following areas:

- Participant's experiences of having KS, and where they feel they have been most in need of information and support.
- The impact of KS as a condition affecting the whole of life, including quality of life issues such as the impact of KS on an individual's ability to work, their finances, relationships, etc.
- Identifying the most appropriate services and approaches needed in delivering support to individuals with KS and their carers in response to their identified needs.
- To disseminate the results of the research, on behalf of people with KS, with a view to influencing policy and/or improving service provision.

This report will present the findings from the two research studies, identify the implications of the analysis and finally provide recommendations to promote and develop support for people with KS.

Study 1: The Interviews

Aim: To explore, using qualitative methods, the subjective experiences of individuals with KS; particularly the impact of the condition on identity and the related psychosocial impact of living with the condition.

A search of the relevant databases (Medline, PsychInfo, AMED, BNI) suggested no research had been conducted specifically to explore the psychosocial needs of individuals with KS as detailed by themselves, nor had any attempt been made to listen to the lived experiences of this population. Research had focused more on medical diagnosis, treatment and medical problems associated with the condition even though biomedical researchers and condition specialists continually acknowledged the psychological impact these conditions have and the need to address these issues. Thus a qualitative approach to this study was adopted in order to fully explore the subjective experiences of individuals with KS.

Qualitative research can provide descriptions of the phenomena of interest by uncovering and ascribing meaning to issues missed through quantitative methods where assumptions have to be made about the nature of the issues to be examined (Grbich, 1999; Willig, 2008) This type of research also recognizes the complex and dynamic social world in which understanding is constructed and multiple realities exist (Banister et al, 2002). It considers that every person is an individual and as such holds a different perspective on the world, will react differently to events, and will hold different opinions on how the world should be. So, to acknowledge this, individual interviews were the primary method used to collect data to allow the voices of those with KS to be heard. Semi-structured interviews are a powerful and flexible method of data collection guided by a schedule of questions to address areas of interest. However, the schedule acts *only* as a guide and does not dictate the interview. This allows for any interesting, important or unanticipated, unexpected issues that are brought up by an interviewee to be followed up and explored. This can facilitate the opportunity for interviewees to talk about/iterate issues, ideas and experiences that have special salience for them.

In addition one focus group interview was also conducted. This method has been widely used in the field to explore people's experiences of, and knowledge about, disease (Barbour & Kitzinger, 1999). Focus groups provide a forum for the communication of experiences and are particularly suited for this research as they create an environment where sensitive issues can be discussed and explored by the participants in a way that

perhaps the researcher would not feel appropriate to explore. It is a method where the researched become the researchers (Wilkinson, 1998).

Method:

Qualitative design

- The interview schedule was designed in collaboration with the KS Association Committee.
- 1 Focus group: Group size n=6 participants; lasting 1 hour.
- 13 semi-structured 1:1 interviews (mode length 1 hour; range: 45 mins to 2 hours).
- All interviews were audio taped and transcribed verbatim.
- Data from the questionnaire study were also included. These comprised the 41 answers to the open ended questions from the questionnaire and 36 critical incidents (where study participants wrote about a time when KS had been a significant problem for them).
- The data were analysed using inductive thematic analysis by 2 researchers, independently of each other (Attride-Stirling, 2001; Ely et al, 1991; Hayes, 1997). The resulting analyses were compared and discussed and an agreed interpretation will be presented. This process ensures rigor within the analytic process.

Results:

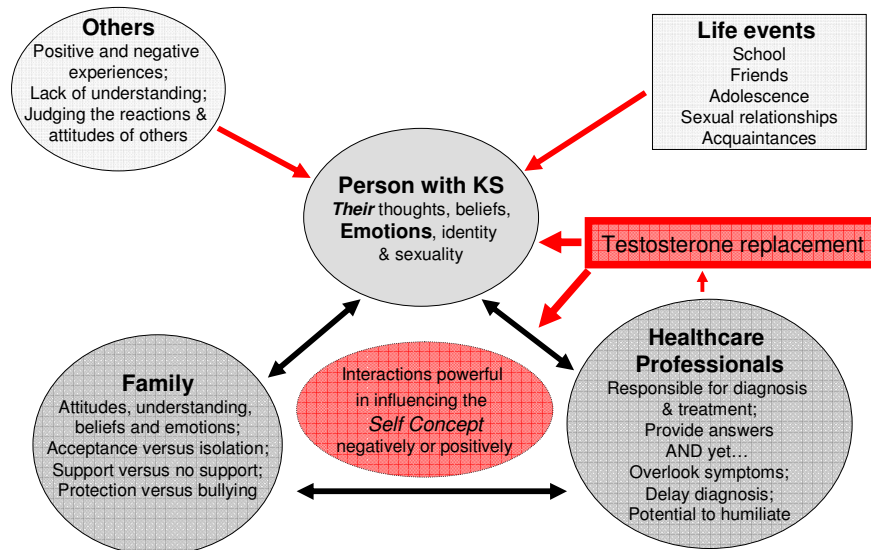
Qualitative research accepts that people are complex individuals and as researchers we can learn a lot about their experiences and the meanings that they attribute to those experiences by talking to people, asking questions, and allowing their voices to be heard. What follows is our interpretation of the lived experiences of men with KS. These experiences were unique and yet there were aspects of living with KS, which were similar for all the men we interviewed (for example, infertility, testosterone replacement). Differences appear to relate to when they received a diagnosis; the greater the delay (40+ years of age) the greater the risk of long term health problems and disability. Equally their personalities, the understanding and reactions of significant others (parent's and siblings), and life events appeared to influence the impact KS had on each of their lives.

The stories that the men had to tell were unique to them, which made pulling the data together challenging. However, seven minor themes emerged from the data, which were

common to all of the men. These are shown in the diagram below. These themes were then grouped under three major headings: 1. *Diagnosis and Management of KS*; 2. *The Self*; and 3. *'Me, KS and others'* with all three relating significantly to the effects that low testosterone has had. During the analysis a core theme emerged **Emotional Impact** which is central to each of the three major themes. The emotional impact of KS, for some men, has been significant and as such has defined them as a person the life choices they have made. (See Figure 2 for an illustration of the themes and how they link together.)



Figure 2: Thematic Framework: Me, KS and others



Other small categories emerged but were not experienced by all the participants reflecting the very individual experiences of KS as a syndrome. These areas were concerned with insight into the illness, the actual interview process, the KS Association, and destruction

through alcohol misuse. These will be alluded to under the three main themes as individual examples of concern.

The Themes:

Theme 1: Diagnosis and Management of KS

There was a huge variability in the age at which diagnosis occurred; for some it was as a teenager and for others it didn't occur until they were in their 40's. This meant there was significant variability in the impact the diagnosis had; for some this was a life changing event, whereas for others it actually meant no change. Diagnosis for some impacted on family relationships, where some families had a complete lack of understanding of the syndrome and others felt controlled by their families since their diagnosis. For some men the diagnosis led to an explanation of some of their problems in particular their difficulties in learning. Each man had a different experience of how their diagnosis was communicated to them and by whom, in most cases with shocking insensitivity. Most men commented that they had to learn to cope and adapt to having Klinefelter's syndrome and how they did this was a very personal experience. In common with other long term conditions, a diagnosis of KS can have a significant emotional impact which is often reported in depth and clarity (see *Emotional Impact* later in this report). Talking about KS and its impact helps to achieve some sense of meaning and eventually can help people adjust to the diagnosis.

The following quotes reflect the diversity in diagnosis experiences:

- *“ diagnosis, um I felt quite sort of wind blown I suppose but um over next couple of years I sort of grew in knowledge of what it was all about and how it was affecting me, and at the time it wasn't a big deal..”* 5: P1, L3
- *“Um, I remember it was about 13, I had, I was, uh, diagnosed with it, it all started out um, I was taken down to a PE lesson, um, swimming, and I, I didn't notice it at the time, but someone my age, colleagues, my ages said um, C, you've got some boobs growing, and I thought, eh, I've not got any boobs, and then I, I, I looked at myself, and I didn't see any boobs there, but they were hard, so uh, and then I, .. Uh, um, and then um, my mum took me to the doctor, and then the doctor, he, he did everything, he even liked touched the boobs and the nipple ...He was quite painful actually...Um, and then um, he did everything else, and then that's when I realised that I um, I had XXY um, because um, I, I thought my testicles were um, normal”* 2: P1; L16

- *“And um just went to the doctor, routine check-up and um, they’d just computerised their records and I saw it on my records and I queried it, as it was my record, and uh and, and they said it was my record and well from there I found out...”* 11: P1; L21
- *“So you get the diagnosis and you feel great and all of a sudden you don’t feel great you feel totally, totally shattered, its like having a glass wall and its fantastic and all the pieces fit together and then you drop it and things just .. your whole life has got in a worse mess than it was before the you got the diagnosis...”.* 13: P1; L20
- *“...have you got KS, it went through a whole list of, and I thought, good god, some people are really unlucky, thank god I haven’t got that, (laughs) of course to find out that’s exactly what I did have, low intelligence, low span of attention, osteoporosis and all this stuff, and I, I don’t know that’s me...”* 6; P5 L1
- *“Well yeah, because actually, he, he had a letter from another doctor, to say, no-one could see my testicles, you know, but at least, in, in my mind, it tells me at least he should have said, well, I’ll send you to such a person, there’s nothing wrong with you.....Nothing, nothing was done, nothing was done, forty-nine I was when I got diagnosed, it was in endocrinology, testing for diabetes, and I was sat asking everybody, because you ask everybody don’t you, and no-body’s heard of it, you know, when it comes back to get this, letter through the post, and it says, carried type (karyotype) A and then XXY, and you try to get all the information you can off the internet and you, and even, even the, the text series on the uh (TV)”* 3: P13; L17
- *“My doctor came up to my bedroom cos my mother and father had had one hell of a row with him, and he said, you’re XXY, and that’s all he said.....”.*9: P3; L30
- *“And you get to a point where you think, I wish I was never diagnosed.”* 9:P51; L30

Table 1: A KS diagnosis

Pre-birth or early childhood	Adulthood
Parental involvement greater understanding and support	No, or limited parental involvement
Treatment decision-making: the parent's or the young person's?	Delayed testosterone treatment
Early preparation for testosterone replacement	Physical consequences of no/low testosterone already apparent and irreversible
More time to prevent the consequences of KS: education, support and low testosterone	More likely to have experienced bullying and/or humiliation because of appearance
	Greater emotional impact (fear----anger)

With understanding comes the reality of what KS is, how it will affect them, and it's management through testosterone replacement.

Testosterone

The absence or presence of testosterone significantly impacted on body image, self identity, emotional stability and relationships. Some men viewed testosterone as the key to feeling normal, having a libido, being assertive and not frightened of others, with a more positive mood and greater confidence. Yet for many access to testosterone replacement was often postponed to the "right age" or delayed (as in the case of those diagnosed at a later age). After being prescribed testosterone, regular monitoring was uncommon. Equally there was often a lack of accurate information regards the benefits and risks of Testosterone replacement, which often led to misunderstandings for many men, and mistrust of the medical profession.

On a practical level most men had tried various testosterone preparations to identify the one which had the least side effects and was the easiest to administer. They also commented on the physical and emotional side effects of testosterone replacement, which for some influenced their decision to stop the replacement. For one man, because of the significant psychological and emotional changes, the use of oestrogen as well as testosterone was discussed. So for some testosterone wasn't the magic cure they had hoped for, and life with KS became more problematic. For the older men they felt that

starting testosterone mid-life was too late, the physical consequences of low testosterone (osteoporosis, diabetes) were acutely evident. Equally the immediate effects of testosterone had brought on feelings of puberty, which they felt were confusing and difficult to deal with.

The following quotes reflect the varied experiences of the men interviews:

- *“Why, why, do we have to have testosterone, I don’t think it’s the miracle cure.”*
“It’s not a miracle cure um, and it, its, it comes with a lot of side effects.” 13:
P25; L31
- *“and the other thing you see, they introduced me to testosterone, when you’ve been passive before, I’m suddenly aggressive, and I went to seek my tormentors, and I went to a particular place, where I knew they were, and I sought my aggressors, I’m bigger than them now, and I just sat staring at them, and they don’t remember me, and I just sat staring, and I put that man in as much fear as he put me in when I was younger, and at the end of the night I just went out, and I seen him, and I gave him a pop, and I was never violent before, never, never.”* 3: P8; L12
- *“It makes me feel wonderful, its my payback for how I was treated when I was younger....It helps, when I initially get the injection, for two weeks you feel like superman, like burns down after a while, but initially for the first two weeks I’m up on libido*
And you feel alive....You know, you don’t, I said to the endocrinologist, before I was living, now I’m alive.” 3: P8; L23
- *“No, um, I all ready think that I’m at that stage where it’s something I do automatically in the morning, um, you, you know, I, I have my shower, I slap this gel on and I brush my teeth, you know, its all that sort of thing, its all part of the general morning routine, and um, I don’t think to myself I’m putting this on because I’ve got Klinefelter’s syndrome, I’m just thinking, this is what I do in the morning, um, when I, when I toddle off for work I always have my weeks supply of tester gel with me, in my suitcase, its just, its just part of the normal morning routine, um”* 5: P10; L21
- *“The injections are much, much better, yeah... Yeah the gel packs are a bit, the problem with them is uh, that you know they’re a constant reminder of your condition so... your not meant to have contact with children, and make contact*

four hours after and all this sort of stuff, and its just yeah, its just a pain in the arse.” 11: P2; L26

- “I’m quite wary of these injections, I, I don’t really particularly like needles in my arm....” C2: P8; L21
- “ Um so I’ve had a variety of different types of Testosterone, I mean it wasn’t until I got Testergel that my body really accepted it, really accepted it” 10: P12; L18
- “It clashes with other medication, I feel nowadays the drug companies are messing about with genetics and they all do drugs with genetics.. and here they are banging me with the most powerful hormone in the body, which is testosterone.” 10: P12; L27
- “I don’t have any at all (Testosterone), I don’t have any problem on that side of it...or I don’t feel I do, um never had any complaints (laughs) if that makes sense?.” 3: P16; L7
- “I wanted not to be tired in the morning, I wanted energy”. 9: P7; L28

**Table 2: Testosterone replacement:
What it means?**

No testosterone	With testosterone
Something missing <ul style="list-style-type: none"> • Physically different • No libido Who am I? <ul style="list-style-type: none"> • Different, not normal • Male, female, both? Emotionally labile <ul style="list-style-type: none"> • Angry and aggressive, anxious and tearful Socially anxious for fear of being bullied and/or persecuted.	Brings hope and change <ul style="list-style-type: none"> • New life, new me • Become more confident and assertive • Have a libido, become sexually active Who am I? <ul style="list-style-type: none"> • Improved self esteem • More masculine appearance
<p style="text-align: center;">However Testosterone replacement is not for all...</p> <ul style="list-style-type: none"> • Side effects • Extreme changes in personality • A perception that it is destroying a part of the self 	

Theme 2: The ‘self’

Appearance

Several men felt that they were quite different in appearance to other men especially during school where they saw themselves as very tall and thin with long arms and legs.

Lack of muscle definition and strength was common to the older men, which was a source of distress and of humour:

- *“got a 1 pack, not a 6 pack...”* FG 6; 3
- *“It’s the six pack, but the plastic, somebody’s nicked the plastic”* 4: P7; L42.

Gynaecomastia (having breasts) was a common occurrence and was discussed at length during the interviews. However, attitudes towards the fact they had breasts was unique to each man; some felt strongly that this was a part of them whereas others expressed a desire to be rid of them, or had undergone or were contemplating a double mastectomy. The management of their appearance and the reactions of others to their breasts will be discussed later.

Most of the older men discussed the fact that they had a small penis and felt as they aged this was becoming smaller still, with a few having considered a prosthesis. Those participants who currently had partners all reported how their partners were comfortable with their appearance even though, in some cases, the men weren’t comfortable themselves. Like men without KS (Cash et al?; Hewstone et al, 1996) some were more self-conscious and highly distressed about their appearance, while others were not, these varied experiences appeared to be dependent on the support they had from significant others and their level of resilience and confidence to deal with the attitudes and reactions of others. Here are some quotes from the interviews to reflect the appearance related concerns:

- *“I was very self conscious for a long time.”* 6: P6; L26
- *“Its strange, they’ve changed with the testosterone, suppliers I don’t know, yeah, but from the point of view of L she just says I’m fine, so its not as important, but it would be nice just to um, uh, you know, uh, when it’s a hot date just take ones shirt off and not mind, uh, cos I’m still thinking its uh, bluff myself, if I’m lying down you wont notice, but....I’ve got to stand up sooner or later, so, I might have a bit of liposuction on the tummy as well (laughs), and a facelift”* 6: P22; L12
- *“I, even today I don’t think I’ve grown up, but sometimes people say to me I look so youthful for my age, um, that’s another thing that’s good about it, that you stay, you stay youthful (laughs) Um, uh, which is nice, but as I say there are things that have happened to uh, my body that are showing age”* 7: P17; L2

- *“I know it’s easier to um, out on weight than it is to reduce it, and I know it’s hard with KS....” 7: P43; L8*
- *“I feel comfortable at home, and even when I was at school, I kept trying to do some after school activities, I did swimming, but um, as soon as I had those breasts it went away, I didn’t really do swimming at all um, I tried to do football, that’s all, I did that quite well but, as I was so thin, I got injured and I was injured for a long time, so that I never took on board, so I, I, I don’t really have anything um, that” 2: P4; L11*
- *“..and then the (testosterone replacement), yeah, and then I started to grow hair on my body, um, I couldn’t shave, I didn’t shave in those days....Eventually, I mean I wear a full beard and a moustache, its not brilliant, but its pretty adequate” 10: P4; L3*

Self identity

Confidence in their own identity was observed in a few of the men, however, for those who had appearance concerns and/or had received negative reactions from others this had reduced their self-esteem and self-confidence. Several older men talked about being different and unsure of their identity as being either male, female, or both. So, while some embraced the female element of themselves in terms of having an extra x sex chromosome (sometimes perceived as a female chromosome) and breasts, others saw themselves only as male. Yet their masculinity was challenged by the fact that they were infertile and could not father children naturally. Some men talked about the need of reassurance from others to confirm their identities. As such those with poor self esteem were diagnosed late, unsupported by their families, school and the health service, and experienced frequent episodes of abuse and humiliation and tended to withdraw from society to protect their fragile identity.

For the majority greater self-knowledge and understanding of KS promoted their confidence and ability to deal with others. As a man with KS you can be seen as ‘Mr Nice Guy’, someone who is sensitive and supportive of others.

The self concept of men with KS varied significantly however KS or its consequences could significantly impact on their self esteem, the following quotes illustrate this:

- *“I suppose it made me a nice guy.” 6: P12; L22*

- *"I find it very, very hard to talk, to talk to girls, um, its very uh, even boys I have, with my own group I'm, I mean I'm in a small group anyway, uh, but, when I go my, what I call out, I, I don't go out at all, I, I stay in my room, um, play video games, so I'm still like a kid really, that's, that's how I feel, I feel like a 12 year old kid but in a 24 (mans body)." 2: P2; L36*
- *"I don't have that (something in common), and, and that's what I think um, I think that separates me from everyone else, and I do like that, I do like to be different, I don't like to be, have um, like different, in some instances, like everyone else, and I, my, my friends actually like me being different, and I'm actually getting used to it, that I'm not like you and I am different, and uh, I'm, I'm special, I, I, I, I use that a lot to my friends, I tell them, I'm special, you're just normal, I'm special, so, I do like that, I'm, I'm, I'm not, and it makes me feel better um, I'm, I'm the only one with it (KS), and, unless when I'm here, and then everyone's got that, but I think I'm like that as well" 2: P14; L25*
- *"I mean, I can understand that counsellor saying, well, forget the past, but, like I said to them all, how I can forget the past, when I have a shower everyday...It's constantly there, it's a constant reminder, you know, and god did me a turn there you know, otherwise I would have had children, so, you know, to feel like a man you've got to have the appropriate equipment, like a woman, you know, you've got to have the appropriate equipment, and if its lacking you don't feel like a man" 3: P30; L2*
- *"..I think that I've sort of developed in stages, so I've gone from being the lonely child sufficient unto itself that it just did its own thing, regardless of what anyone else thought, or whatever, um, uh, from that stage I was a sort of um, I joined all sorts of organisations according to my age, I joined the scouts, the venture scouts, um, ranger act, things that involved me getting involved with other people, doing things and learning new skills, and all that sort of thing, um, probably um, probably, venture scout and ranger act and work, taught me the sort of organisational skills, and the sort of um, people interaction skills...um, holding my own in a group, persuading people to do what I wanted them to do in a group, um, so that, I, I, I think probably the will power part developed out of that." 5: P7; L25*
- **Researcher:** *with the infertility, do you think that's undermined your confidence, or how you see yourself, your, your self esteem?*

“No, if anything my self esteem had improved, I mean I, it has since a result of the treatment, but I, I, only perhaps um, more likely it’s allowed me to gain a better understanding of um, here I am in a way....So it’s allowed me to, it’s allowed me to stay focused a bit more on what I want to do” 11: P11; L15

- *“trapped (laughs), no, I, I, I don’t know how I see myself now, um, I know a bit more what’s wrong with me, but whether that’s going to help me or not, its feedback I want now, more than anything else, I need to know that I’m not, not going round the twist, do you know what I mean, that I am normal, even though I’m not normal in an un-normal sort of way like.” 4: P14; L25*
- *“ It’s to do with my makeup, I.....I define Klinefelter’s syndrome as being unsure, obese,....” 9: P27; L12*

Theme 3: Me, KS and others

Sexual relationships

Some men had been married and were either now divorced or widowed. Those that disclosed that they had had sexual relationships reported that Klinefelter’s had impacted significantly on the relationship. Experiences varied from having a third person in the relationship (affairs), to partners not understanding that because a man can ejaculate does not mean they produce sperm and are fertile. For some their sexuality had either still to be decided, or they had been through a period in their lives when they believed they were homosexual and developed a relationship with a man. All the men felt the need to be in a relationship. For some this need was very intense and often resulted in rejection from potential or actual partners.

- *“47 (age at diagnosis) and un she says (Health professional) she says this probably why your such an emotional guy and all this sort of thing, um, and we, we can give you information, and a directive and I wasn’t too worried about it, but when I told my partner, oh, oh, that means I won’t be able to have any children with you...” 7: P2; L15*
- *“You know, and that’s the other aspect isn’t it, I give her a cuddle, its nice isn’t it, always, you know, I’ve lived my life without sex, so it won’t matter if I live the rest of it without it” 3: P12; L1.*
- *“Yeah, sometimes you feel like a boy trapped in a man’s body, a.....Yeah, from a sexual point of view, times I’ve slept, I mean, my missus thinks I’m oversexed, I don’t think I’m oversexed but she thinks I am, and I’m oversexed in my head but*

its just the rest of it is a bit knackered, with everything else that is, its what you want to do in your head and what actually happens are two different things” 4: P10: L9 and 33

- *“ and she wanted children, um, unfortunately I couldn’t father children, but, because I produce fluid (semen) and she had mistaken that for sperm.” 9: P17; L25*
- *“Every doctor I met, you can’t have children, so I thought it, although I went out with a lot of women.....And I had sex.....I didn’t marry because I felt it was unfair....It’s not, getting married and knowing you won’t have children is one thing, knowing you can’t have children is another” 10: P3; L7-13*
- *“Some older people think inter sex, that’s a hurtful thing.” 9: P30; L32*
- *“I went to XX Hospital and they said you’ve got KS, it as fantastic as I had a reason for all the time of trying to dress as a female of trying to live as female, of trying to live as man, ticked of all....(the symptoms)”. 13: P1; L15*
- *“I was just a little lost lamb really, a very little lamb I suppose, um, decided then to get married, wife number one, um, prior to getting married there was an acceptance of S(f), obviously that’s part of me, um, but when you, when I got married, then everything changes, because you have a third person in the relationship with two people living together, and that doesn’t work, so um, wife number one, get a divorce, second one comes, in fact I got engaged while I was still married, which is probably a bad thing to do really (laughs), but there you go, um, wife number two comes along, same situation, accepts it, accepts it, gets married, doesn’t accept it, um, she had an affair, probably because I couldn’t, I, I wasn’t interested in sex, not in the way that she, you know, raised my interest in sex, S(f) interest in sex, but it either was me wife, I don’t really want to name whatever else, but anyway, um, but she, she um, found um, the same again, third person in the relationship, so she had an affair” 13: P3; L16*
- *“...and she wasn’t bothered about my body shape.” 6: P7; L10*
- *“I noticed other things, with the sexual side of it, is that it’s prolonged, It, it takes a along time..to uh complete”. 7: P20; L11*

Relationships with Family

There was a considerable range in family backgrounds and dynamics. For some their backgrounds had been quite abusive both physically and mentally. Others had very supportive families, but at times it was felt that support could tip over into overprotection

and being controlled by family members. Interaction with siblings varied from a reliance on them to assist in day-to-day life, to siblings disassociating from the individual with KS possibly due to their lack of understanding of Klinefelter's, or related to blaming the person with KS for any dysfunction and distress within the family. This highlighted often significant discrimination and stigma regard men with KS.

- *“I remember my father saying to me when I was at, a youngster, and he, he catch me out in the corridor and he said to me um, you’ll never be um, you’ll never be um, you’ll never be able to carry on the family name cos you haven’t got it in ya, and I thought, well were did that come from, you know, I didn’t”* 7: P22; L8
- *“Although I’ve got Klinefelter’s he doesn’t understand that, so then she went and told him that I’ve got Klinefelter’s cos I couldn’t find anything about it, he said, oh well we could find it on the internet cos that’s what’s his thing, and he found some things for me, but then he just switched off, and all my family, excluding my partner, apart from my cousin St, all my family cannot accept it, not one of them.”* 13: P2; L11
- *“I went to XXX last week, for a family gathering, I went to XXX, and I always hated this, and I’d shy away and go into another room, when they all talk about kids and, like my brothers and sister they’ve all got children, and then I get the old question.....Why have you never had any children, and I can’t say to them that’s the reason why, I’ve never told them, cos, cos its like a hidden shame that you hold within yourself, you know, how, how can I say to them, well, you know, I’ve told me father, he can’t grasp it, my mothers got Alzheimer’s, thank god, because she doesn’t need to know, and my father as I said, I tried to explain to my dad and he’s an old man, and he doesn’t understand, and he said, I’m sorry son, but, and I explained to my brother and he can’t grasp it, the thing is though they can’t..”* 3: P12; L25
- *“when they found out I was Klinefelter’s and couldn’t have children, my sister and my brother decided, and with my half brother that I was to look after my mother”* 10: P18; L23

KS and Work

It was evident in the majority of men that their shyness and learning difficulties had negatively impacted on their working lives. It meant that some men had not been able to work for a considerable number of years which some believed made others view them as lazy. For some men their working life was unaffected and they had achieved a good

education and career, however, in one case it was interesting how work colleagues were reported as viewing the person with KS as being much stronger than he viewed himself. The following quotes illustrate the diversity of experiences the men reported, but in considering their responses it is easy to recognise how their self esteem and a lack of testosterone can affect their ability to acquire and perform their jobs.

- *“And I can’t um, if someone talks to me over the phone and then someone comes and talks to me, I’ll forget what they’d told me, and I won’t know what they’d told me, just doesn’t work, so um, consequently I had a lot of support in the beginning, but then the company went to India anyway and I decided to live, and in desperation I needed money I went to a job in, in a motor spares shop, found that I couldn’t do it, just couldn’t do the lifting in that work, I went to see my doctor and they said you’ve got to give up work.” 13:P 5; L1*
- *“And jobs, well it’s been an office environment, I think more, more, I suffer in an office environment because um, I’m not very good at the politics, and probably not very good at politics because I haven’t got much experience of politics....And, and I don’t like it very much, and everything else, I mean, I know I suffer quite a lot from mood swings at work as well, and I can tell that its me, the politics I’m not good at it...” 11: P6; L7*
- *“I supposed I was the middle man, and so I went to them and said, oh, how do I get promoted to the senior research officer, and he said, well the way you’re going on you’ll be lucky if you have a job in twelve months time, a year, what ever, uh, let alone promoted, and I went to personnel about that, and they said, oh, we’ll sort that out for you, and don’t worry about it, and we’ll input into that, and it turned out that its all an automatic thing, next year it came through, but I uh, I went and saw the head of department about it, and, and next year with staff development I put, however, I want this self assertiveness course, so I thought I could learn to handle it better, but she refused it because she said I was assertive, and I think that’s the most assertive I’d ever come across (laughs), and so every, every time after that went to get on an self assertiveness course, but I never got one, but when I went to uh, when I went to uh, C, I uh, got on my first assessment, I want to go on an self assertiveness course, and they said, nah, you’re self assertive anyway, and so I came over as, as quite er, um, quite controlled.” 6: P15; L34*
- *“I want to go to high school, and go to university, um, but, I’m the type of person who say, say if I’m doing a course, it can’t be a course where it is just*

sitting behind a desk and learning stuff, I need to be active.....Yeah, that's why I'm, I, I'd like to find a real job, my mum says, oh you should go out and get a real job, and I say, well its very easy for you to just say that, its hard for me to actually do it and, how I told my mum that a real job is, for me, not working behind a desk, counting numbers and answering telephones, because I will just get bored of that very quickly, I need to be active." 2: P4; L30

- *"Its still out of my comfort zone, and that's when I feel, when I um, when I'm trying to meet people, and that, unless I'm not close to my home is, is, I find it very, very hard, and that's why I, I probably can't get a real job, um*

Interviewer: So what's a real job, what would be your ideal job?

2 Working for a living, paying rent, I don't pay rent at all." 2: P4; L 4

- *"..as I say my father knocking me down as well, when I was a youngster, um, in, into not believing in myself, you know, um, loosing, I lost um, my self-esteem, my ability to carry on with work, I couldn't cope with work, I had to give it up."*

7: P7; L5

- *"And then I went to my time at an apprenticeship, um, you used to do six months in those days, I was going to do panel beating, working on the local bus, or tram....Um, my father got me that job, in the meantime I decided I wanted to go to technical college, cos I, I hadn't heard of the apprentices, it was a choice, cos I was working on the body section and I wanted to learn carpentrySo um, I went and done a carpentry, but also at night time I used to do welding class, and that was three nights a week, uh, that was from six o'clock to nine o'clock, and welding in those days was a proper trade."* 9: P23; L8

- *"Back to normal for me, yep, so I mean in, in terms, in terms of did uh, my work suffer during that period, no it didn't um, but I, I imagine that's sort of um, in built personal resolve and drive and all the rest of it, that, that takes care of that, um, so no, I achieved all my deadlines, I achieved all my aims, all this sort of thing, um, I suppose that having sort of got the treatment in to me, uh, probably the things that improved were my attention to, to detail and that sort of thing, which perhaps had not been so finely tuned for a while, uh, but as I say no-one noticed."* 5: P5; L34

KS and School and Education choices

Some of the men had no adverse experiences of school but many of the older men reported in detail the bullying they experienced at this time. Individuals that it had occurred

for a variety of reasons, for example, as a result of their appearance, or the fact that they spent more time with the girls than with the boys. Men with KS are known to experience a range of learning difficulties and these men were no exception with several reporting learning difficulties while at school. This may have been a generational effect as schools were yet to acknowledge dyslexia and while some were given extra help or worked extra hard to keep up, many gained their education out of school with either college or apprenticeships. All the men interviewed gave the impression of being very creative with making things, and most reported problems with their concentration that had always impacted on their lives and not just with learning. For some of the older men there was a strong sense of frustration at not achieving more through their education: without a diagnosis of KS their learning difficulties were ignored and they were considered thick or stupid, labels that undermined their self esteem and confidence. During adolescence they reported that physically they appeared and felt different to their peers and the experience of communal changing rooms had to be negotiated. However there were men who did not experience a learning difficulty or bullying and school was described as an OK experience. The following quotes illustrate the different experiences described by the men:

- *“I when I was in primary school, they said like um, they mentioned that like I had a learning disability but I never had any help until I went to the middle school, then when I went to middle school I got pulled out of a lot of lessons, and again I missed out on them lessons to get my reading and spelling where it should be, but I’m, I’m not quite there now, I’m, when I’m talking I stumble a lot, my, my spelling’s atrocious....Oh yes, yes, I have dyslexia.”* 2: P5; L6
- *“Being in school I had plenty of mates, never fell out with people,....”.* 9: P19; L16
- *“At school I was knocked, and you loose your, your self-esteem,...”.*7: P2; L15
- *“I mean I wasn’t educated, I told you I was bad, I had no education at all, I’ve picked up my education and intelligence, etc, either through going to college,...”.* 7: P17; L30
- *“..the learning difficulties, I’ve no doubt that um, I remember having uh, one to one tuition, reading, read it, but I’ve no doubt that I wouldn’t mind doing that, at most it was, I didn’t recognise it, so cos I couldn’t recognise it I was ridiculed, I remember being ridiculed at class many times, and I remember being ridiculed at sports because I didn’t you know, uh, team sports I didn’t take, take part in..”* 3: P18; L20

- *“I left school with nothing, I can’t even write properly as you can see, my signature, I’ve never been able to write properly, its beyond me, I, um, I, I, I, I, mess me big letters and me small letters up, and me, me brain capacity lately is, is next to zero, I, I, I mean, don’t get me wrong, I’m not thick or say I was nothing, but I wouldn’t say I was thick, I’m into the thing, I know about things I’m interested in” 4: P23; L31*
- When asked if KS had affected him educationally participant 6 responded:
“No, uh well, when I was at um, St B boys school just uh, in C we had a system where you went to a higher school until you were fourteen, and then the last two years you went to, into another school, and uh, my sister she was two years older, and she, I remember her, going on her O levels and A levels when I was at school, but she acted, done so badly that mum and dad said, oh well, we’ll, we’ll uh, send you to private school, and get you the teaching, so I went to private school, small classes, brothers in, uh, brothers and that, and corporal punishment was the thing, you got the strap, that nice leather thing, where you hold your hand out, and I got a few of those, um, once for, because I didn’t answer a question, (laughs), um, and um, and so um, yeah, I sort of um, it was good, it was an all boys school, so um, I suppose, but I didn’t get any chance to mix with women, sort of thing, um, uh, but I was quite shy.” 6: P4; L10
- *“..at school, at private school, I found it easier to associate with the girls, so consequently I got bullied for associating with the girls, because, it’s the wrong thing to do isn’t it, also being head and shoulders above everybody else that didn’t help, I found that um, it was easier to stay off school um, so I was, I would be ill, more than I was well, because I didn’t want to go to school, um, if I was bullied at school um, and if I um, got out of school, my brother would then come along, who was six years older, he would beat up the person that had hit me, when I got home my brother would beat me, so I, I had double bullying basically, um.” 13: P2; L29*

Healthcare professionals and managing Health Problems

Most men reported that when they went to see healthcare professionals they had to continually explain their Klinefelter’s syndrome to each new person they met. Some had experiences of being ridiculed and even laughed at by healthcare professionals. Experiences with GPs varied hugely; some had very supportive GPs whereas others reported how their GP had both failed to recognise and had mismanaged the care they

required as a result of the syndrome. Many men felt that their medication (testosterone) had been forced upon them without a full explanation and this was considered to be a particular issue for parents who have to make the decision for their children. Overall throughout the contact with health care services there had been a huge misunderstanding of Klinefelter's syndrome, the problems it can present and the consequences of low testosterone. There was a reference to an NHS website citing vague information about Klinefelter's which was misleading. Various health services had been accessed by those interviewed including primary care, secondary care, acute care (various departments), mental health including inpatient units, counsellors and speech therapists. However, most men reported that the emotional and psychological impact of KS and low testosterone was never acknowledged let alone addressed. The majority of the older men suffered from several health problems including pain, osteoporosis, fatigue, depression and diabetes all as a consequence of delayed testosterone treatment. Some men felt that part of the difficulty with having health problems and Klinefelter's, is that the Klinefelter's itself is not recognised as a disability in terms of receiving state benefits. This prompted one man to compose a one page sheet of information to give to each health professional he came across in an attempt to increase their awareness and knowledge. What follows are some quotes which reflect some of the experiences men with KS have had, but also their understanding of what is needed to improve future interactions with healthcare professionals:

- *“Yeah, cos um, the other thing is I, I, I find what, like I say with, with um, going back to the KS is information, lack of it....” .7: P29; L3*
- *“I think it would be a good thing, the GPs to know a bit more,....” 7: P47; L22*
- Participant 11 attempts to explain the lack of understanding and knowledge of the GPs and makes suggestions how this might be rectified.

“I don't know, look, I mean the condition is one in a thousand people basically isn't it, twenty-five hundred men, twenty-thousand people estimate, its not massive, and so you know, any, any individual practice is probably going to have four or five maybe, um, cases....I'd like doctors to be more educated about it, to know more about it, but GPs have a huge amount of things to do, and its difficult to you know, I mean what can you do, you can, you can send them a pack I guessIt would be good you know, most, if, if every practice had one person that had to, had maybe gone the one day to training, or been, not necessarily training but one, one, spend one day uh, with a group of KS people just to have an idea of the spectrum of um, the spectrum of people with the, within the personal

experience, so if somebody does come with a diagnosis, one person in the practice would be able to help or, you know.” 11: P10; L23

- Here Participant 10 discloses a need for the emotional impact of low testosterone and having a KS to be addressed.

“..it (KS) can’t be cured, but ...At least, in the no time in the forty-two years that I’ve been on medication has any doctor in any country turned round to me and said, how do you feel, how do you get on with this medication.” 10: P14; L22

- However this lack of support frustrates and angers many men..

“But they’re the con artists, so when they come across a problem staring them in the face, they walk away from it. Here’s your prescription, go away, they don’t say it, but that is what the implication is, if you ask them a question they can’t answer, they, they ignore you. You’re just ignored.” 10: P5; L1-5

- While others felt they were kept in the dark, but knew they were different..

“I remember being in primary school and having a medical, and the doctor turning round and to my dad or something, or, and having the medical and them examining me and then me dad leaving the room and something, and something about then saying, oh they’ll grow, you’ll be normal, so I knew I wasn’t normal, from primary school,” 4: P1; L11

- *“I think the only thing that I keep sort of thinking back to, is how little the GP knew, uh, and how little I suspect the GP knows even now, um, and, also the attitudes of GPs in general to the condition, um, I mean what they tend to say, certainly on my experience in the past is that its not life threatening, I’ve got, I’ve got bigger things to worry about, you know, um, its um, its something that if you need treatment, if you need testosterone, we can, we can sort that out for you, um, and yeah, just generally just go out and get on with it, you know, pull yourself together, that sort of lark, and I think uh, um, I, I think to some extent, um, to a large extent, that they’re the professional that worries me most, um, because I, I still think they’re probably still doing the same as they were doing back in nineteen ninety-six when I was first, identified as a problem, um, so I, I, I think if anyone needs to be concerned about um, the provision of NHS service then that’s where they really need to be looked to be starting, I think to some extent we could appear to be society friendly, that we’re teaching them to suck eggs, um, I think that at some, its really the primary care side that you, its, tension, because uh, there’s plenty of anecdotal evidence out there that there are GPs who uh, try to manage it themselves without referring onto a specialist,*

um, whether they don't want to disturb these specialists, or whether, whether they, they, there's a financial implication and they don't want to get near, I don't know, but, um, I think if, if anyone is being diagnosed now, and has gone through the GP network, then its probably a case of history repeating itself, uh, unless they get a GP who knows which specialist to refer them onto." 5: P12; L22.

- "I would like um, the doctor I was to see, whether it was endocrinologist or not....Actually understand what Klinefelter's syndrome isUm, I got to see a doctor and I've got to explain to him, or her..And they've done a medical degree" 9: P36; L18.

Core Theme: Emotional Impact

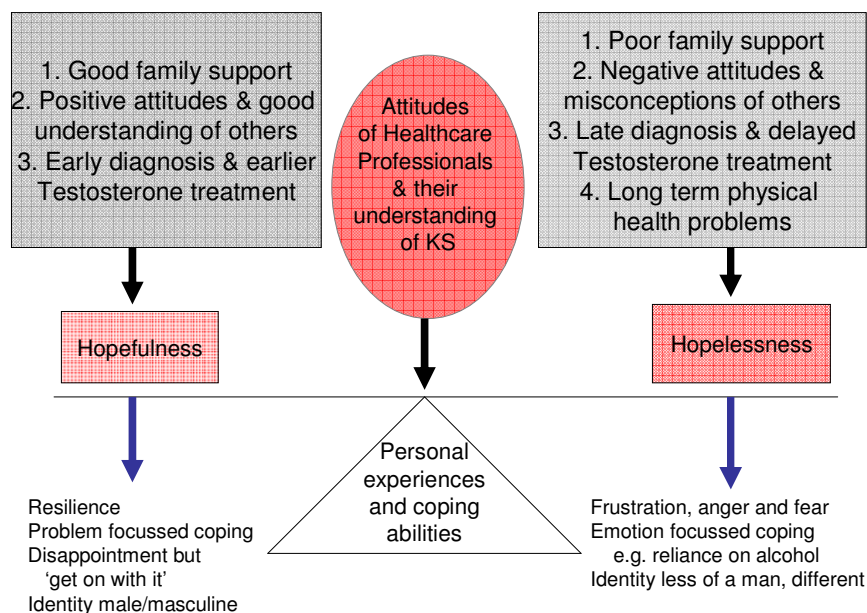
The extent of their personal experiences and living with KS affected their sense of well-being and meaning of life. Many men were disabled by the physical and metabolic consequences of low/no testosterone and others by their confused gender identity. Several men described themselves as caring and sensitive to others with two men describing themselves as having feminine characteristics. These qualities attracted the stigma of others particularly where they were labelled as "gay", "deformed", "transsexual". The stigma within families often resulted in abuse ("to knock it out of me") or rejection. During some interviews, men disclosed that they were unsure of their sexuality and it was often after Testosterone replacement that they perceived themselves to be truly masculine. However for one participant they described gender confusion, living sometimes as woman and the rest as a man, here testosterone was considered destructive as they felt their female self would be destroyed. These experiences resulted in depression and for some sectioned as a risk to themselves and others.

However this core theme (*Emotional impact*) clearly reflects the fact that KS is a syndrome, since experiences were unique and significant for each of the men. The emotional distress experienced by many, included feelings of self-loathing, fear and anxiety, frustration and anger, a sense of loss and isolation, and yet for some it was a challenge to be overcome. Those men diagnosed later in life were significantly affected by a lack of testosterone, physically (osteoporosis; disability and pain). As children and young adults they had been victimised by family, friends and strangers; and now as adults were wary of others but engaged in a constant battle to be heard and understood, particularly by health professionals. Their delayed diagnosis often underpinned their anger and frustration

at doctors especially GPs and insensitive professionals, who could not offer any insight or explanation for the symptoms they experienced. For some there was a sense of hopelessness about their long term future, while others continued their battle with the medical profession, in the hope that they would achieve some sense of recognition of the long term physical impact of KS and hypogonadism. To cope, some men chose to use alcohol as way of reducing their social anxiety and fear of “*persecution*” and “*rejection*”.

A sense of hopefulness was apparent for several men across the age range, who talked about KS not defining their identity. Equally the choices and decisions they made appear to suggest that they have high self esteem and success in life through work, education, and business provided a secure and confident identity. They appeared to demonstrate more adaptive coping styles, to promote adjustment to the limitations imposed by KS, and a strong sense of resilience. However, a low libido, being infertile significantly impacted on them and their partners; equally fluctuations in testosterone levels were recognised as affecting their levels of fatigue, concentration and mood, which at times created some degree of anxiety.

Figure 3: Hopelessness versus Helplessness: the emotional impact of KS.



The following quotes illustrate the two extremes of the hopelessness/helplessness continuum:

- “*Lets get on with this and live the best way I can.*” 11: P25; L31

- *“When he gets chased down the station, that happened to me, but it, about sixteen lads, because I, I, I wasn’t concerned, and I didn’t, I wanted to be angry but I couldn’t, cos some days it doesn’t appear, and of course the other factor kicks in, the shakes, and I didn’t realise that its associated with testosterone, and because I didn’t have testosterone I was shaking, unbelievably, so, the bullies recognised it, and they all chased me down this corridor, and they stripped me off, and I had to make my own way all through school*
And you know what they named me, J no balls, and I had to carry that all through my youth” (gets upset)
To deal with this distress and manage his social anxiety, 3, turns to alcohol:
“I didn’t deserve that, um, life is difficult, life is difficult for me, alcohol is my saviour” 3: P3; L2
- *“But I have been, I have been um, I say alcohol, I did turn to the alcohol for a while, um, and it brought me down, a hell of a way...Um, but I was never an alcoholic, I just drank a lot, got upset, over, overwhelmed myself a bit, um, and then the opposite, I got angry, but never, ever hurt anybody, I know that I couldn’t never do that, ever” 7: P31; L13*
- *“It’s (fear and shame) lessoned with the testosterone, it’s lessoned, I mean before that, I remember being at a public place, and everybody went to me, well say something, and I apparently sat there all night and I didn’t say anything, cos I didn’t feel safe in the company, and it’s fear of being found out, I think it’s the fear of being found out, if I could” 3: P5; L16*

Study 2: Questionnaires

Aim: To understand the psycho-social impact of living with KS in adult men using standardised questionnaires.

Method:

Quantitative design

- The questionnaire was designed in collaboration with the KS Association Committee.
- 300 people from the Members list at the KS Association were contacted by letter.
- 62 people responded, giving a response rate of 21%; a good response for a postal questionnaire.
- All questionnaire packs were scored using the instructions provided by the questionnaire authors.
- The questionnaire scores were entered into an SPSS database. All analyses were done using SPSS version 15.

Collected data

1. Demographic information

We collected data on demographics information such as gender, age, ethnicity, living arrangements and employment status.

2. Medical background

Medical background information related to the symptoms of KS experienced by participants, other medical conditions, the healthcare professional who diagnosed the KS and the time since diagnosis.

3. Physical profiling

These data comprised information on the physical characteristics of study participants such as height, waist measurement, chest circumference, arm span and ratings of the size of participant genitalia.

4. Standardised questionnaires

We used several standardised questionnaires to enable us to objectively measure the psycho-social impact of living with KS.

- The Hospital Anxiety and Depression Scale (HADS) (Zigmond & Snaith, 1983) is a widely used scale measuring general levels of distress using two subscales – anxiety and depression – where higher scores indicate greater distress.
- The Derriford Appearance Scale (DAS-24) (Carr, Moss & Harris, 2005) measures social anxiety and social avoidance. The higher the score on the DAS-24 the more the person is experiencing social anxiety and is using social avoidance as a coping strategy.
- The WHOQoL-Bref (The WHOQoL Group, 1998) measures four quality of life domains: physical, psychological, social and environmental. Higher scores on this questionnaire indicate greater satisfaction with quality of life.
- Rosenberg Self-Esteem Scale (RSE) (Rosenberg, 1965) measures self-acceptance or a basic feeling of self-worth. The measure is brief, applies to all and measures only self-esteem. It has been widely used in a variety of settings. Men usually score higher than women and results should be unaffected by age.

5. Amended questionnaire

The Honey & Mumford Learning Styles questionnaire (Honey & Mumford, 1986) was adapted to be included. It measures preferences for particular learning styles, useful in determining what format information might best be presented to individuals with KS. After discussion with the KSA committee, a subset of questions was included. An additional question was added (q21) to determine if individuals with KS have a preference for written over verbal instructions.

6. Visual Analogue Scales

In order to explore the impact of KS and its treatment on psychological adjustment and daily functioning more broadly, Visual Analogue Scales (VAS) were also included in the study. VAS scales are easy for respondents to complete and are often used in clinical assessments (Carr, 1997).

Questions and responses for the Visual Analogue Scales

1. Lifestyle: scored from +10 (very positively) to -10 (very negatively) and where 0 means no impact on lifestyle
2. Worry: scored from 0 (not at all) to -10 (very negatively)
3. Noticeability of KS: scored from +10 (noticed positively by others) to -10 (noticed negatively by others) and where 0 means the KS is not noticeable to other people
4. Severity: scored from 0 (not at all) to -10 (very negatively)
5. Impact on confidence: scored from +10 (very positively) to -10 (very negatively) and where 0 means no impact on confidence levels.

7. Open ended questions

Study participants were also given space on the questionnaire to write about any other issues they felt were important but that hadn't been covered by any of the questions in the standardised questionnaires, or in the interview project questions. Responses were incorporated into the data and report for Study 1.

Open-ended questions:

1. Would you like to add to or explain any of the answers you have given in previous sections?
2. Is there anything you would like to tell us which has not been covered by the various questionnaires included in this pack?

8. Critical incident analysis (Flanagan, 1954)

In healthcare research use of the critical incident technique (CIT) can be a good resource in identifying the experiences of participants in relation to their illness or condition (Butterfield, Borgen, Amundson & Maglio, 2005). It is a flexible method which can help to identify important events that might be missed when focussing on the everyday experiences of individuals. It can be very useful in determining the cause and severity of reported problems. However, it is vulnerable to misreport since it relies on participants being able to recall events accurately and being willing to relate them, and it has a built-in bias to incidents that happened most recently since these are easier to recall. In this study the critical incident exercise comprised the four components listed below. Responses were incorporated into the data and report for Study 1.

Critical incident analysis questions

- Can you think of a particular situation when KS has been a significant problem for you?
- a) Please describe what happened on this occasion
 - b) How did it make you feel?
 - c) What did you learn from this experience?
 - d) If you were in the same situation again, would you behave, the same or differently? If differently, please explain.

Results

Part 1: Whole group

1. Demographic data

Age range: 18-74 years (mean age 43.7 ± 15.21 years).

Gender: participants identified predominantly as being male (59 participants, 95%); one participant identified as being female, and two participants identified themselves as being intersex.

Ethnicity: 55 participants (89%) identified themselves as being white British, 5 participants (8%) as being white non-British (ie, Irish, Scottish or Welsh), and 2 participants (3%) identified themselves as belonging to another ethnic group.

Living arrangements: 55 participants (89%) were living in England, 3 participants (5%) were living in Wales, 3 participants (5%) were living in Ireland, and 1 participant was living in Scotland. Seventeen participants (27%) reported living alone, 15 (24%) lived at home with their parents and siblings, 18 (29%) reported living with their partner, while 5 (8%) were living with their partner and children. Seven participants (11%) reported other living arrangements (for example, living in rented accommodation with friends).

Employment: 24 participants (39%) reported being in full-time employment and 10 participants (16%) were in part-time employment. Seventeen participants (27%) reported being unemployed while 6 (10%) had retired from work. Two participants were students. Twenty-six participants (42%) did not answer this question.

2. Medical information

100% of participants identified themselves as having Klinefelter's Syndrome.

Close to three quarters of those taking part in the study (73%, 45 participants) reported having other medical conditions as well as KS, while 17 participants (27%) identified no additional medical condition. The three most common medical conditions reported were diabetes (6 participants), arthritis (6 participants), and asthma (6 participants). Pain was reported by 5 participants. The full list of medical conditions reported by the study participants is provided in Appendix 1.

Participants were asked to identify the healthcare professional (HCP) who diagnosed them as having KS. The results are shown in Table 3 below. Endocrinologists were most commonly credited with making the diagnosis, with urologists in second place. The category "other" included two references to a gender clinic, one reference to a surgeon, an

IVF doctor and a fertility specialist, a back specialist, plus references to consultants where the speciality was not able to be identified by the study participant. These data suggest that KS is not easy to diagnose and that individuals with KS have contact with a wide range of healthcare professionals.

Table 3: Healthcare professionals (HCP) making diagnosis of KS

<i>HCP</i>	<i>Frequency (n=)</i>	<i>Percentage</i>
Endocrinologist	25	40%
Other	9	15%
Urologist	8	13%
GP	7	11%
Geneticist	6	10%
Paediatrician	4	7%
Neurologist	2	3%
Psychologist	1	2%

Time since diagnosis was recorded in the following groupings where it can be seen that the greater majority of individuals taking part in this research had had a diagnosis of KS for quite some time.

- recent diagnosis, ie some time during the last 2 years = 2 participants (3%);
- more than 2 years but less than 5 years = 6 participants (10%);
- more than 5 years but less than 10 years = 16 participants (26%);
- more than 10 years ago = 38 participants (61%).

Age at diagnosis revealed a wide variation in study participant experiences. The calculated average age at which study participants received a diagnosis of KS was 26 years of age (\pm 14 years). More detailed consideration showed 10 participants (16%) received a diagnosis during childhood, ie before the age of 10. Seven participants (11%) reported receiving a diagnosis as a teenager (ie between the ages of 11 and 17 years of age). Eighteen participants (29%) reported being given their diagnosis as a young adult (ie between the ages of 19 and 28 years). Twenty-four participants (39%) received their diagnosis during their mid-life years (ie between ages 32 and 55). Three participants did not provide this information.

3. Physical profiling

We tried to collect information on the physical characteristics of study participants such as height, waist measurement, chest circumference and arm span, however, the majority of these data were not provided by the study participants.

The great majority of study participants completed the section relating to the ratings of the size of their genitalia. These data are shown in Table 4 below.

Table 4: Frequency (and percentages) for ratings of penis and testicle size

	<i>Miniscule</i>	<i>Much smaller</i>	<i>Slightly smaller</i>	<i>Same</i>	<i>Slightly bigger</i>	<i>bigger</i>
<i>Penis</i>	0	24 (39%)	10 (16.1%)	12 (19.4%)	9 (15%)	3 (5%)
<i>Testicles</i>	4 (7%)	44 (71%)	8 (13%)	2 (3%)	1 (2%)	1 (2%)

Missing data: n=4 for penis comparisons; n=2 for testicle comparisons

Thirty-four participants (55%) rated themselves as having a much smaller or slightly smaller penis compared to someone else of their age and build, while 56 participants (90%) rated themselves as having miniscule, much smaller or slightly smaller testicles than someone else of their age and build. The psychological implications of these data are considered later in the report.

4. Standardised questionnaires

The first step in understanding the data from the standardised questionnaires is to look at the mean scores for the whole group and make comparisons with a non-clinical “normative” sample. Normative data exist for the HADS, the DAS-24, and the RSE questionnaires but not for the other measures used in this study. A clinical sample of individuals with pituitary conditions was used as an additional comparison group. This was because pituitary conditions affect endocrine function in relation to the production of hormones and also affect appearance. Table 5 shows the scores from the KS participant group compared to scores from the normative and clinical (pituitary) samples.

Table 5: Means (and standard deviations) plus normative values from relevant populations for all study variables and a comparison group of pituitary patients

<i>Variable</i>	<i>KS group (n=62)</i>	<i>Normative values (n=1792)</i>	<i>Pituitary group score (n=134)</i>
HADS Anxiety	9.13±3.86	6.14±3.76 (n=1792)	8.16±4.31
HADS Depression	6.45±4.41	3.68±3.07 (n=1792)	6.39±4.02
DAS-24	47.93±15.47	26.63±11.40 (n=457)	45.51±15.49
QOL Physical	12.88±4.05	Under review†	12.65±3.55
QOL Psychological	12.36±3.58	Under review†	13.07±3.02
QOL Social Relations	11.42±4.32	Under review†	12.91±4.28
QOL Environment	13.98±3.10	Under review†	15.21±2.63
Rosenberg Self-Esteem	16.73±7.38	15-25	n/a

† Under review by WHOQoL-Bref authors

- General anxiety and depression levels (as measured by the HADS questionnaire) in the KS study population were raised compared to the normative data and the pituitary group scores although the variability of scores (the standard deviation) were not much different.
- DAS-24 scores for the KS study group were raised compared to the normative data and those recorded by the pituitary group. This indicates that the participants with KS were experiencing more social anxiety and were using more social avoidance techniques than either the normative group or the group with pituitary conditions. Both the KS study group and the pituitary group had a broad range of scores reflecting the wide range of individual variation in the degree to which people were affected.
- “QOL” in the table above refers to the scores from the Quality of Life measure used in this study (the short form of the WHOQoL). The maximum score possible on the four WHOQoL domains of physical, psychological, social relationships and environment is 20. KS study group participants were scoring well below that especially in relation to the physical and psychological domains. They were also scoring below the pituitary group in relation to the psychological, social relationships and environment domains. These data suggest compromised quality of life for individuals with KS.
- The maximum score possible on the Rosenberg Self-Esteem scale is 30, scores between 15 and 25 are within the normal range, while scores below 15 suggest low self-esteem. Male study participants usually score quite highly on this questionnaire. The KS study group were scoring at approximately the half-way point which suggests that the study group as a whole had reasonable levels of self-esteem. However, the standard deviation indicated a greater variability in the range of scores and therefore in levels of self-esteem in this group.

The next step in analysing the data from the questionnaires is to look at the levels of response on the HADS questionnaire. Since the HADS can be used for making clinical diagnoses of anxiety and depression it was important to get an understanding of where the KS study group were scoring.

Thirty-seven participants (60%) had clinical levels of anxiety, 18 (29%) at levels where some form of psychological support would be helpful/advised

19 (31%) scored 8-10 (mild)

11 (18%) scored 11-15 (moderate)

7 (11%) scored 16+ (severe)

Twenty-one participants (34%) had clinical levels of depression, 9 (15%) at levels requiring treatment

12 (19%) scored 8-10 (mild)

6 (10%) scored 11-15 (moderate)

3 (5%) scored 16+ (severe)

The next step in understanding the data is to look at the relationships between the various questionnaire responses by the KS study group. Pearson's correlations were carried out which provided evidence of a life limited by inherent problems in self-esteem, since:

Low levels of self-esteem (RSE) were associated with

- significantly increased levels of anxiety (HADS Anx, $p < .001$);
- significantly increased levels of depression (HADS Dep, $p < .001$);
- significantly raised levels of social anxiety and social avoidance (DAS-24, $p < .001$);
- significantly reduced ratings of physical quality of life (WHOQoL physical domain, $p < .001$);
- significantly reduced ratings of psychological quality of life (WHOQoL psychological domain, $p < .001$);
- significantly reduced ratings of satisfaction with social relationships (WHOQoL social relations domain, $p < .001$);
- significantly reduced ratings of satisfaction in relation to study participants' environment (WHOQoL environment domain, $p < .001$).

Individuals with low level self-esteem had increased levels of general anxiety and depression (HADS Anx & Dep) as well as raised levels of social anxiety and social avoidance (DAS-24). This is a normal human reaction to difficult situations. We minimise the impact by avoiding unnecessary stress. We hypothesise that as life becomes more restricted participants' quality of life significantly deteriorates (hence the lower scores on the WHOQoL domains).

5. Amended questionnaire

After discussion with the KSA committee, the Honey & Mumford Learning Styles questionnaire was adapted to be included in the study. An additional question was added (q21) to determine if individuals with KS have a preference for written over verbal instructions. Most people have a variety of learning strategies, however, some people have a main, or predominant, style of learning. The four learning styles outlined by Honey & Mumford are as follows:

- **Theorists** – Need to learn in a structured and orderly way, they enjoy theories and need time to look at situations. They become bored easily if the information they are provided with is at too low a level.
- **Pragmatists** – Need to link experience with learning, they like to see the immediate relevance to what is being taught. They prefer to find information from someone who has had relevant experience. They like to put theory into practice straight away.
- **Reflectors** - Like to ponder over new learning and like to look at problems objectively. They do not like to be hurried or told there is only one way to do something.
- **Activists** – Like to be given tasks to perform, and prefer to learn from experience. They like to be involved with other people and hate repetitive tasks.

Table 6 shows the results from the learning styles questionnaire. Forty of the study participants (65%) had scores on the amended learning styles questionnaire which resulted in a clear identification of one learning style. These are shown in the second section of the table. The bottom half of the table shows the numbers of participants where the learning style was split between 2 or more of the options. This applied to 20 participants (32%). There were 2 participants where no learning style preference was evident (ie they scored the same for all the learning styles). By adding together the number of single style scores and combined learning style scores, a total frequency count was achieved for each of the four learning styles. This is shown at the top of the table.

We also asked participants to identify the form in which they preferred to receive information. They were asked to choose between written or verbal presentation of information. Thirty-nine participants (63%) preferred written to verbal presentation, with 20 (32%) choosing verbal. Three participants did not answer this question.

Table 6: Frequency of learning styles

<u>Learning style</u>	<u>Frequency</u>	<u>Percentage</u>
Reflector	31	50%
Theorist	20	32%
Activist	19	31%
Pragmatist	19	31%
<u>Single learning style (n=40, 65% of study participants)</u>		
Reflector	22	35%
Activist	8	13%
Theorist	5	8%
Pragmatist	5	8%
<u>Combined learning style (n=22, 35% of study participants)</u>		
Activist/Pragmatist	5	8%
Reflector/Theorist	5	8%
Theorist/Pragmatist	4	6%
Activist/Theorist	3	5%
Reflector/Pragmatist	2	3%
Activist/Theorist/Pragmatist	1	2%
No preference detected	1	2%

With half of the participants opting for a reflective style at least some of the time these data seemed to suggest was that were a significant number of people in this study group of participants who might do better in the healthcare system if information was presented to them in a written format with time for them to reflect and consider it before talking it over with a healthcare professional. It would certainly suggest that the current NHS climate of short appointments and limited written information may be disadvantaging some of the individuals who took part in this study.

6. Visual Analogue Scales

In order to explore the impact of KS and its treatment on psychological adjustment and daily functioning more broadly, Visual Analogue Scales (VAS) were also included in the study. The first step in understanding the results of the VAS data is to consider the mean scores for each item. Not all participants completed the visual analogue scales. The number of responses column in the table below indicates where there were missing data. These results are summarised in Table 7.

Table 7: Means (and standard deviations) for all study Visual Analogue Scales

<u>VAS line</u>	<u>Number of responses</u>	<u>VAS score</u>
Impact on lifestyle	59	-2.93±4.77
Worry about KS	62	-5.77±3.04
Noticeability of KS	60	-1.31±4.26
Severity of KS	61	-5.97±2.78
KS Impact on confidence	58	-2.85±4.89

These data suggest that KS was having a largely negative effect on the study participants. There was a great deal of variability in the data (the standard deviations for some of the scales were relatively large) possibly reflecting that as a syndrome KS presents a wide variation in symptoms which may or may not affect all KS men. For example, these data suggest that there were some individuals taking part in the study for whom KS was having a very negative impact on their life and others where it was having a more minimal impact. This also seemed to be the case in relation to the data on how noticeable the men reported the condition to be. However, levels of worry about KS and how severe the study participants reported their KS to be were high.

As with the standardised questionnaires, the next step in considering these data is to examine the possible relationships between the scoring of the scales. As before, Pearson's correlations were carried out. These suggest limited relationships between these variables with few significant correlations being observed.

- Higher ratings of the severity of KS were associated with significantly increased levels of worry ($p < .04$).
- Positive ratings of the impact of KS on lifestyle were significantly associated with positive ratings of the impact of KS on confidence levels; similarly, negative ratings of the impact of KS on lifestyle were significantly associated with negative ratings of the impact of KS on confidence levels ($p < .001$).

The final step in understanding these data is to look for relationships between the VAS scale scores and the standardised questionnaires. Once again, Pearson's correlations were carried out. These again suggested limited relationships between these variables.

Three of the VAS scales achieved no statistically significant relationships with any of the standardised questionnaires. These were: VAS for impact on lifestyle, VAS for noticeability of KS to others, and VAS for impact of KS on confidence levels. This may not be surprising when the low scores achieved on these scales are considered (as shown in Table 5).

Two of the VAS scales did have a range of statistically significant relationships with the data from the standardised questionnaires. These were: VAS for levels of worry in relation to KS and VAS for the perceived severity of the KS.

High levels of worry about KS (as recorded by the VAS scale) were associated with

- significantly lowered levels of self-esteem (RSE, $p < .001$)
- significantly increased levels of anxiety (HADS Anx, $p < .005$);
- significantly increased levels of depression (HADS Dep, $p < .001$);
- significantly raised levels of social anxiety and social avoidance (DAS-24, $p < .001$);
- significantly reduced ratings of physical quality of life (WHOQoL physical domain, $p < .001$);
- significantly reduced ratings of psychological quality of life (WHOQoL psychological domain, $p < .001$);
- significantly reduced ratings of satisfaction with social relationships (WHOQoL social relations domain, $p < .005$);
- significantly reduced ratings of satisfaction in relation to study participants' environment (WHOQoL environment domain, $p < .001$).

High ratings of the severity of the KS (as recorded by the VAS scale) were associated with

- significantly lowered levels of self-esteem (RSE, $p < .027$)
- significantly increased levels of depression (HADS Dep, $p < .019$);
- significantly reduced ratings of physical quality of life (WHOQoL physical domain, $p < .008$);
- significantly reduced ratings of psychological quality of life (WHOQoL psychological domain, $p < .004$);

So, before we stated that individuals with low level self-esteem had increased levels of general anxiety and depression (HADS Anx & Dep) as well as raised levels of social anxiety and social avoidance (DAS-24) plus significantly reduced quality of life (WHOQoL). The VAS data add to that picture. They tend to suggest that levels of worry about KS and the individual's perceived severity of their condition may have a part to play in the wider impact of the condition. However, it should be remembered that KS is a syndrome and

some individuals will have very few, or no symptoms, while others have significant symptoms. So, high levels of self-esteem are associated with low levels of general anxiety and depression, low levels of social anxiety and depression and increased quality of life. The key may be the extent to which an individual perceives themselves to be affected by the condition.

7. Open ended questions

Two thirds of study participants (41, 66%) took the opportunity to write about any other issues they felt were important but that hadn't been covered by any of the questions in the standardised questionnaires. These responses have been incorporated into the data and report for Study 1.

8. Critical incident analysis

Thirty-six study participants (58%) wrote about a critical incident where KS had been a significant problem for them. These responses have also been incorporated into the data and report for Study 1.

Part 2: Group differences

Finally, we can explore the questionnaire data in a variety of ways, for example, looking at differences in impact of symptoms, physical appearance, age, and time since diagnosis.

There were some issues we could not explore, for example, geography (the sample was not big enough), ethnicity (the sample was biased, with 89% of participants being White British), employment status (42% of participants did not provide this information), and gender (95% identified themselves as being male). Since the sample is too small for us to put all the group factors into one calculation the results of these inferential analyses can only be considered indicative rather than definitive.

1. Demographic factors

The first demographic factor to be considered was age. The first step was to see if there was any relationship between age and the responses to the standardised questionnaires and VAS scales included in the study. Pearson's correlations were carried out and only one statistically significant result was obtained. This suggested a limited relationship between age and the study variables.

- Increasing age was associated with significantly decreased ratings of satisfaction with physical quality of life (WHOQoL physical domain, $p < .02$).

Since age was associated with decreased satisfaction with quality of life, this led to the question as to whether there was any association between age and the symptoms of having KS. Once again, Pearson's correlations were carried out indicating that as men age certain symptoms of KS seem to be reported more frequently, as well as the number of symptoms of KS increasing.

- With increasing age significantly more men were reporting lowered libido (sex drive) ($p < .001$), problems with fatigue ($p < .005$), and a lack of facial or body hair ($p < .046$).

2. Medical factors

Table 8 shows the main symptoms associated with KS identified by the study participants. The second column shows the number of individuals identifying the symptom as being part of their experience of KS and the final column shows the frequency as a percentage of the whole research group of 62.

Table 8: Frequency of KS symptoms

<u>Symptom</u>	<u>Frequency</u>	<u>Percentage</u>
Infertility	52	84%
Mood swings	41	66%
Lack of facial/body hair	38	61%
Gynaecomastia	37	60%
Fatigue	37	60%
Depression	30	48%
Low libido	29	47%
Weight gain	29	47%
Headaches	23	37%
Osteoporosis	19	31%
Impotence	19	31%
Generally unwell	13	21%

It is interesting to note the difference between the symptoms the study participants recognised as being part of their KS and the list of symptoms that the medical community recognise. For instance, mood swings, headaches and fatigue are not usually mentioned in the medical research literature.

We examined the impact of the various symptoms by comparing the scores on the standardised questionnaires for those reporting a symptom to those reporting being free of the same symptom. We used Mann Whitney U tests for all except symptoms with the

exception of depression where t-tests were used. Non-parametric tests were predominantly used to take account of the uneven numbers within the study group reporting symptoms, for example, 52 participants (84%) reported being infertile, meaning that the comparison group comprised 10 participants who were not infertile.

- Being infertile did not seem to impact on psychosocial functioning with no significant differences on any of the psychosocial variables for those who were infertile (52 participants) compared to those who were not.
- Experiencing mood swings was associated with a significant reduction in ratings of physical quality of life (WHOQoL, $p < .011$).
- For those affected, a lack of facial and/or body hair was associated with increased social anxiety (DAS-24, $p < .034$).
- Men with gynaecomastia reported increased social anxiety (DAS-24, $p < .003$), and reduced quality of life (WHOQoL physical domain, $p < .004$; social relations domain, $p < .034$; and environment domain, $p < .001$).
- Fatigue was associated with increased social anxiety (DAS-24, $p < .04$) and reduced physical quality of life (WHOQoL, $p < .024$).
- Attributing depression to having KS seemed to result in increased general anxiety (HADS anx, $p < .02$), reduced self-esteem (RSE, $p < .03$), increased social anxiety (DAS-24, $p < .04$) and reduced psychological quality of life (WHOQoL, $p < .03$). However, it is interesting to note the lack of significant difference in the HADS depression scale between those who identified themselves as having depression as a symptom of KS and those that did not.
- Having low libido was associated with reduced physical quality of life (WHOQoL, $p < .012$).
- Weight gain did not seem to impact on psychosocial functioning with no significant differences on any of the psychosocial variables for those who were reporting weight gain (29 participants) compared to those who were not.
- Experiencing headaches reduced physical (WHOQoL, $p < .006$) and environment quality of life (WHOQoL, $p < .006$).
- Osteoporosis as a symptom of KS increased social anxiety (DAS-24, $p < .01$) and reduced physical quality of life (WHOQoL, $p < .02$).
- Being impotent did not seem to impact on psychosocial functioning with no significant differences on any of the psychosocial variables for those who reported being so affected (19 participants) compared to those who were not.

- Feeling generally unwell was associated with significantly reduced ratings of physical quality of life (WHOQoL, $p < .016$).

These data suggest that the symptoms associated with KS can negatively impact on levels of psycho-social functioning, particularly in relation to quality of life.

3. Physical factors

As previously reported, men were asked to rate the size of their penis and testicles making a comparison to someone of the same age and build as them. The results of these comparisons were reported in Table 4 earlier in the report.

In order to explore the impact of the reported ratings of penis size for the study participants between group comparisons for the scores on the standardised questionnaires using Kruskal Wallis tests were undertaken.

- Penis size (across the five groups of slightly smaller, smaller, same size, slightly bigger, and bigger) significantly affected levels of depression (HADS Dep, $p < .002$), self-esteem (RSE, $p < .001$), social anxiety (DAS-24, $p < .01$), physical and psychological quality of life (WHOQoL, $p < .03$ and $.02$ respectively)

Since we were, in effect, dividing the study participants into five or six different groups based on the ratings of their genitalia, the next step was to see which groups were most significantly affected in terms of psycho-social impact. Post-hoc testing allows us to compare the groups to determine where the significant differences lie.

- For self-esteem, significant differences were achieved between the group of participants rating their penis as much being smaller and those rating it as being slightly smaller (RSE, $p < .001$), and those in the much smaller and slightly bigger penis rating groups (RSE, $p < .004$).
- In terms of depression, significant differences in the scores on the HADS depression subscale were observed between those rating their penis as much smaller and those rating it as being slightly smaller (HADS Dep, $p < .003$). Similarly, significant differences in HADS depression scores were observed between those in the much smaller group and those in the same size group (HADS Dep, $p < .01$).

- The DAS-24 results (measuring levels of social anxiety and social avoidance as a result of an aspect of appearance) showed two significant results. These were between those who rated their penis as being much smaller and those who rated it as being slightly smaller (DAS-24, $p < .036$), and between those with a much smaller and slightly bigger penis (DAS-24, $p < .009$).
- Only one significant result was obtained in relation to ratings of physical quality of life. This was between those who rated their penis as being much smaller and those who rated it as being slightly smaller (WHO physical, $p < .037$).
- More significant results were observed in relation to ratings of psychological quality of life. Those rating their penis as being much smaller were reporting significantly less satisfaction with psychological quality of life than those who rated their penis as being slightly smaller (WHO psychological, $p < .045$). Similarly, those who rated their penis as being much smaller were reporting significantly less satisfaction with their psychological quality of life than those who rated their penis as being slightly bigger than someone of the same age and build as them (WHO psychological, $p < .034$).

Table 9 below shows the mean scores achieved on the study variables where penis size was indicated as having a significant impact on psycho-social functioning. A consideration of the mean scores achieved by the five groups on the standardised questionnaires certainly suggests that those men who rate their penis as being much smaller have lower self-esteem (RSE), increased levels of depression (HADS Dep), increased social anxiety and social avoidance of others (DAS-24), and reduced quality of life (WHOQoL physical and psychological domains) than those in the other groups. This especially seems to be the case when compared to the means scores achieved on the questionnaires for those who rate their penis as being slightly smaller where levels of self-esteem are higher (RSE), levels of depression (HADS dep) and social anxiety (DAS-24) are lower, and ratings of quality of life are better (WHOQoL physical and psychological domains).

Table 9: Showing mean \pm standard deviation scores on study variables for KS study participants grouped by ratings of penis size

<i>Study variable</i>	<i>Much smaller n=24</i>	<i>Slightly smaller n=10</i>	<i>Same n=12</i>	<i>Slightly bigger n=9</i>	<i>Bigger n=3</i>
RSE	11.7 \pm 6.0	21.5 \pm 5.4	16.9 \pm 6.5	20.7 \pm 6.2	20.0 \pm 9.17
HADS dep	9.0 \pm 4.1	3.5 \pm 2.0	4.4 \pm 3.3	5.7 \pm 5.0	6.7 \pm 2.08
DAS-24	56.0 \pm 13.2	40.3 \pm 17.5	48.8 \pm 15.4	37.0 \pm 10.2	46.0 \pm 7.1
QoL Physical	11.1 \pm 3.6	15.2 \pm 3.8	13.7 \pm 3.5	14.7 \pm 4.2	12.17 \pm 4.2
QoL Psychological	10.3 \pm 3.2	13.9 \pm 3.5	13.4 \pm 3.3	14.1 \pm 3.0	13.3 \pm 4.1

A similar exploration of the impact of testicle size could not be undertaken as there were two groups (slightly bigger and bigger) with only one participant in each.

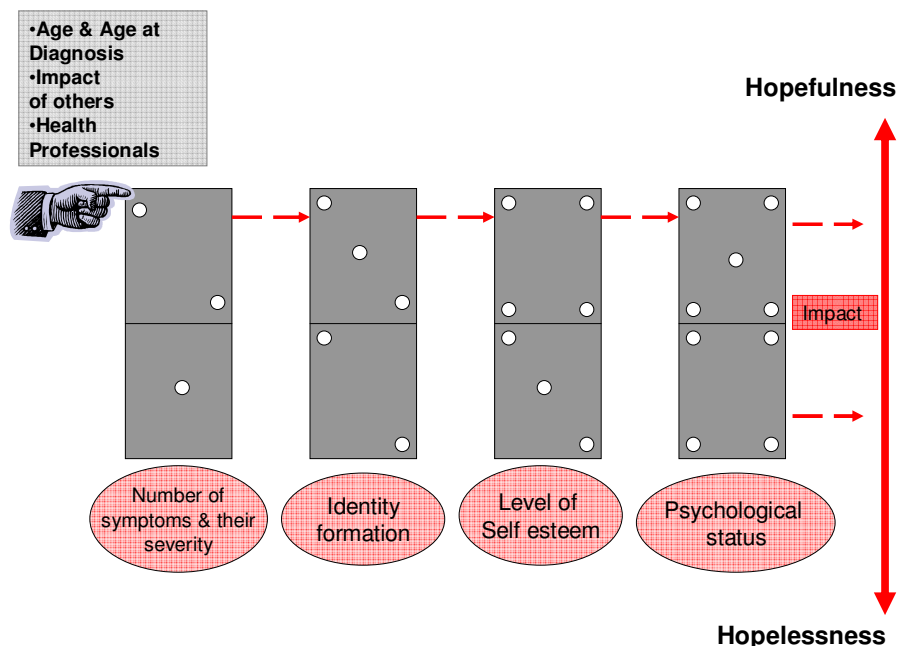
These data suggest that the impact that KS can have on men's genitalia in terms of size inhibition may have a significant psychosocial impact, in particular in relation to penis size where those who rate themselves as having a much smaller penis than someone of their age and build fare less well psychologically than those with a slightly smaller penis.

Conclusion and Implications

Findings highlighted a number of concerns and identified the unmet psychosocial needs of men with KS.

- Diagnosis can take a long time and the prolonged lack of testosterone can have far-reaching negative effects for the individuals concerned.
- There were a significant number of people with KS who might do better in the healthcare system if information was presented to them in a written format, with time allowed for them to consider it before talking it over with a healthcare professional. It would certainly suggest that the current NHS climate of short appointments and limited written information may be disadvantaging some of the individuals who took part in this study.
- The psychological impact of the condition seems to hinge around the number of symptoms and the degree to which an individual perceives that they are affected.
- There are more symptoms associated with KS than the medical community necessarily recognise and these can have a profound impact on the psycho-social functioning of the affected individuals.
- Identity formation seems to be difficult for some of the men with KS and support in coming to terms with this seems to be largely absent.
- Low self-esteem was associated with increased levels of general anxiety and depression, raised levels of social anxiety and social avoidance and reduced quality of life. The reverse was also observed, where high self-esteem was associated with reduced levels of general anxiety and depression, low levels of social anxiety and social avoidance and high ratings of quality of life.

- Depression and anxiety in men with KS needs treating appropriately. Depression was significantly worse if the men reported it as being a symptom of KS. Attribution of poor psychological functioning matters, possibly because if it is a symptom of a permanent condition then there is no hope of avoiding its impact.
- KS is clearly a syndrome since experiences were unique and significant for each of the men who took part in the research. The emotional impact of the condition lay along a continuum from hopefulness to hopelessness where an individual's age, degree of family support, the attitudes and understanding of others, level of self-esteem, time of diagnosis and implementation of testosterone replacement influenced the men's location along the continuum.
- The impact of KS can be described in terms of a "domino effect" (see diagram below) where "symptoms", "identity", "self-esteem" and "psychological status" are the dominos. These dominos may start to topple as a result of age, reactions from other people or as a result of experiences with healthcare professionals. As an illustration, if your first sexual experience is not good, then this might start to topple the dominos. You might suddenly realise that your penis size is a problem, which then might start you questioning your future identity as someone's partner, this can make you wonder if you have any value to others which can lead to a severe reduction in self-esteem. Low self-esteem is strongly associated with poor psychological status which can lead to a general feeling of hopelessness and thoughts such as, "Why should I bother?"



Recommendations

- Men with KS would benefit from emotional support and one to one counselling at specific stages throughout their journey, beginning at diagnosis continuing through treatment and on to the continued and constant daily management of the condition.
- Depression and anxiety in men with KS requires treatment. It should be noted that depression is significantly worse if men see it as a symptom of KS.
- A review of group support is recommended to address the specific emotional needs across the lifespan, for example, fertility and sexual relationship issues for young adults.
- Increase awareness and understanding of the condition and its psychosocial impact both within the general population and across the medical community, in particular all non-endocrine specialist staff. Social workers and staff involved with employment and education services at all levels should also be targeted as part of any awareness-raising activities.
- KSA to generate a one page information sheet that men with KS can give to healthcare professionals.
- The provision of information to men with KS requires careful consideration. The majority of men in this study would probably do better with information presented in a written format with time to consider it before talking it over with a healthcare professional.
- Training for healthcare professionals to enable them to better understand and communicate with patients on certain sensitive issues that may need to be discussed such as size of genitalia and breasts, infertility and the impact of these.
- To lobby for GP training to include genetic problems and how to recognise these.
- To get the NHS website updated to show how the impact of KS is very different for each person.

Dissemination

- The report will be available to download as a pdf from the KSA website.
- The report will be sent to various medical organisations, for example, the British Medical Association, the Royal College of Nursing, Society for Endocrinology, Royal College of Physicians, Department of Health, NICE, Royal College for GPs etc.
- The report will be sent to various other relevant professional bodies, for example, Department for Work & Pensions, Department for Education etc.
- The report will be sent to various other charitable organisations, for example, Jeans for Genes, Unique, Contact a Family, Gay & Lesbian Foundation, Stonewall.

Conferences:

- 2 x posters accepted for Society of Endocrinology meeting in Harrogate (March 09)

Jackson, S & Morris, M (2009) Attribution matters: the impact of Klinefelter's Syndrome on psychosocial-functioning. Poster presented at the Society for Endocrinology BES 2009 conference, Harrogate.

Morris, M, Hancock, J & Jackson, S (2009) "It's your hormones". A qualitative exploration of men's experiences of Klinefelter's syndrome. Poster presented at the Society for Endocrinology BES 2009 conference, Harrogate.

- Paper at European Congress of Endocrinology, Istanbul, Turkey (April 2009)

Jackson S & Morris M (2009) Appearance Matters: the impact of perceived altered appearance as a result of Klinefelter's Syndrome on psycho-social functioning. Oral presentation at 11th European Congress of Endocrinology, Istanbul, Turkey.

- Further submissions to conferences are planned as follows:
 - Appearance Matters 4, June 2010, Bristol, UK
 - Symposium for Society for Endocrinology BES 2011 conference
 - World Congress on Men's Health, October 2011

Journal articles:

We are planning a strategy for disseminating the research through relevant professional journals and more general media publications.

Acknowledgements

Our thanks go to all the people who have participated in this study and in particular to the KSA Committee who have supported us in the development, execution and dissemination of this project.

References

- Ablon J (2000) Parents' responses to their children's diagnosis of neurofibromatosis 1. *American Journal of Medical Genetics* vol 98, pp136-142.
- Attride-Stirling J. (2001) Thematic networks: an analytic tool for qualitative research'. *Qualitative Research* vol 1(3): pp385-405
- Bannister P, Berman E, Parker I, Taylor M, & Tindall C (2002) *Qualitative Methods in Psychology. A Research Guide*. Buckingham: Open University Press
- Barbour RS & Kitzinger C. (1999) *Developing focus group research: Politics, theory and practice*. London: Sage Publications
- Belzeaux R & Lancon C (2006) Psychiatric disorders in neurofibromatosis type 1: From clinical features to neurobiology, case report and review. *Annales Medico-Psychologiques* vol 164(2) pp 141-144
- Butterfield LD, Borgen WA, Amundson NE & Maglio AT (2005) Fifty years of the critical incident technique: 1954-2004 and beyond. *Qualitative Research*, 5(4), pp475-497
- Carr T (1997) Assessment and measurement in clinical practice in Lansdown, R, Rumsey, N, Bradbury, E, Carr, T & Partridge, J (Eds) (1997) *Visibly Different*. Oxford: Butterworth-Heinemann
- Carr T, Moss T & Harris D (2005) The DAS24: A short form of the Derriford Appearance Scale DAS59 to measure individual responses to living with problems of appearance. *British Journal of Health Psychology*, 10(2), pp285-298
- DeLisi LE, Maurizio AM, Svetina C, Ardekani B, Szulc K, Nierenberg J, Leonard J & Harvey PD (2005) Klinefelter's syndrome (XXY) as a genetic model for psychotic disorders. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, vol 135B(1) pp 15-23
- Durkin K. (1995). *Developmental social psychology: From infancy to old age*. Oxford: Blackwell.
- Ely M, Anzul M, Friedman T, Garner D, McCormack Steinmetz A. (1991) *Doing qualitative research: Circles within circles*. London: The Falmer Press.
- Flanagan JC (1954) The critical incident technique. *Psychological Bulletin*, 51(4)
- Hayes N. (1997) Theory-led thematic analysis: social identification in small companies in Hayes N. (Ed) *Doing qualitative analysis in psychology*. Psychology Press Erlbaum Taylor & Francis Ltd. pp93-114
- Geschwind DH & Dykens E (2004) Neurobehavioural and Psychosocial Issues in Klinefelter Syndrome. *Learning Disabilities Research & Practice*, Vol 19(3) pp 166-173
- Honey P & Mumford A (1986) *A Manual of Learning Styles*. Maidenhead: Peter Honey.
- Ratcliffe S (1999) Long term outcome in children of sex abnormalities. *Archives of Disease in Childhood*, vol 80 pp192-195

Rickert VI, Hased SJ, Hendon AE, Cunniff C (1996) The effects of peer ridicule on depression and self-image among adolescent females with Turner Syndrome. *Journal of Adolescent Health* vol 19 pp 34-38

Rosenberg M (1965) *Society and the adolescent self-image*. Princeton, NJ: Princeton University Press

Rovet J, Netley C, Bailey J, Keenan M & Stewart D (1995) Intelligence and achievement in children with extra Z aneuploidy: a longitudinal perspective. *American Journal of Medical Genetics*, vol 60 pp 356-363 in Visootsak J & Graham JM (2006) Klinefelter syndrome and other sex chromosomal aneuploidies. *Orphanet Journal of Rare Diseases* vol 1, 42. Published online 2006 October 24: doi: 10.1186/1750-1172-1-42

Rumsey N & Harcourt D (2005) *The psychology of appearance*. Maidenhead, Berkshire: Open University Press

Simm PJ & Zacharin MR (2006) The psychosocial impact of Klinefelter syndrome – a 10 year review. *Journal of Pediatric Endocrinology & Metabolism*, vol 19(4) pp 499-505

Swanson DW & Stipes AH (1969) Psychiatric Aspects of Klinefelter's Syndrome. *American Journal of Psychiatry*, vol 136 pp 814-822

Sweeting H & West P (2003) Sex differences in health at ages 11, 13 and 15. *Social Science & Medicine*, 56(1) pp31-39

Visootsak J & Graham JM (2006) Klinefelter syndrome and other sex chromosomal aneuploidies. *Orphanet Journal of Rare Diseases* vol 1, 42. Published online 2006 October 24: doi: 10.1186/1750-1172-1-42

Walters E (1997) Problems faced by children and families living with visible differences. In R Lansdown, N Rumsey, E Bradbury, T Carr & J Partridge (Eds) (1997) *Visibly Different*. Oxford: Butterworth-Heinemann.

WHOQOL Group, The, (1998) Development of the World Health Organisation WHOQOL-BREF Quality of Life Assessment. *Psychological medicine*, 28, pp551-558

Wilkinson S. (1998) Focus groups in health research. Exploring the meanings of health & illness. *Journal of Health Psychology* vol 3(3): pp329-348

Willig C (2008) *Introducing qualitative research in psychology*. Buckingham: Open University Press

Zigmond AS & Snaith RP (1983) The Hospital Anxiety and Depression Scale. *Acta Psychiatrica Scandinavica*, 67, pp 361-370

Appendix 1: List of medical conditions reported by research participants

Medical condition	Frequency (n=)
Diabetes	6
Arthritis	6
Asthma	6
Pain	5
Hypertension	3
Intention tremor	3
Platelet Aggregation Defect	2
Factor XII deficiency	2
Panic attacks	2
Memory loss	2
Epilepsy	2
Osteopenia (low bone mineral density)	2
Ehlers Danlos Syndrome	2
Scoliosis (curved spine)	2
Cellulitis	1
IBS (Irritable bowel syndrome)	1
Calcium deficiency	1
Prolapsed disk	1
Hiatus hernia	1
Migraine	1
Tennis elbow	1
Poor circulation	1
Coeliac disease	1
Spondilitis	1
Parkinsons Disease	1
Angina	1
Primary Lymphedema	1
Heart murmur	1
Sleep apnoea	1
Polycythemia	1
Sciatica	1
Weak teeth	1
Underactive thyroid	1
Tinnitus	1
Vasovagal syncope	1
SIJ dysfunction (Sacroiliac joint dysfunction)	1
Vascular disease	1
Psoriasis	1
Hidradenitis suppurativa	1