Helpline: 0300 111 4748
Website: www.ksa-uk.net

Klinefelter's Syndrome isn't rare……but it is rarely diagnosed

Annual Report by Trustees of the Klinefelter's Syndrome Association 2023 to 2024

Objectives and activities of the KSA

The KSA exists to offer support and information about KS/XXY, to raise awareness of KS/XXY particularly within the medical profession and to promote and support research into KS/XXY.

Activities during the year

Raising awareness and research

Exhibiting at conferences

To help raise awareness of KS/XXY, KSA trustees have exhibited at several medical Conferences and Awareness/Information Days throughout the year. These include:

British Endocrine Society (BeS) conference in Glasgow in November and Royal College of General Practitioners (RCGP) conference in Glasgow in October.

Both were attended by trustees Chris Breen and Ken Scott.

Endocrine Clinical Update in Birmingham in April attended by Chris Breen and Alison Bridges.

European Society of Paediatric Endocrinology in the Hague in September, attended by Chris Breen.

Rare Diseases Day in February at St Thomas KS/XXY clinic attended by Sylvia-James Yates and at the Glasgow Children's Hospital attended by Ken Scott.

Information Day at Glasgow University attended by Ken Scott.

Podcasts and webinars

Chris Breen has taken part in two very interesting podcasts. The first in April 23 is Genetic diagnosis of azoospermia: from chromosomes to whole exome analysis webinar: https://endo-ern.eu/event/webinar-genetic-diagnosis-of-azoospermia-from-choromosomes-to-whole-exome-analysis/

The second, in April 24 is more specific to KS/XXY - Understanding Klinefelter Syndrome: The Chromosome Anomaly: https://podcasts.apple.com/gb/podcast/understanding-klinefelter-syndrome-the-chromosome/id1657370014?i=1000653683578

KS/XXY Awareness Day

Our first KS/XXY Awareness Day campaign on 11th May 2023 was organised by trustee AJ Howard with assistance from trustee Ken Scott and volunteer Ed Dobie. Another trustee managed to get a little airtime on local radio to talk about the Awareness Day. AJ also designed the 2024 Awareness Day Advert which featured a series of posts and a short video. We hope this will become an annual occurrence with events to compliment it. Any suggestions/assistance for 2025 would be welcome.

Research and Poster Presentations:

Gender Identity and KS/XXY research, University of Derby:

PhD Studentship - Following Claire Harkin's Psychology MSc that focused on Gender Identity and KS/XXY, the University of Derby was able to secure funding for a PhD studentship on gender identity among people with Klinefelter syndrome (KS/XXY). Athina Tripli was the successful candidate and will be conducting the PhD research, with members of the KSA NEC sitting on the Advisory Panel, as well as other key medical professionals in KS/XXY from across the globe supporting Athina on this journey. The research will assess the perspectives of people with KS/XXY, healthcare professionals and KS/XXY experts to improve clinical practice and treatment/support policy. Athina Tripli will talk more about the studentship and the research at the KSA Conference.

Systematic Review – The School of Psychology, University of Derby.

This is a systematic review of evidence about gender identity and KS/XXY. The proposed review question is: How does gender identity develop among people with Klinefelter syndrome/XXY? The members of the team conducting the review are Professor James Elander, Claire Harkin, Dr Elaina Taylor and Athina Tripli.

Fertility Conference 2024, Edinburgh, Scotland

Claire presented a Rapid-Fire Poster Presentation on "Experiences of infertility among people with Klinefelter syndrome/XXY" to 50 fertility specialist delegates. The abstract was

co-authored with Professor James Elander and Dr Bryan Woodward (Scientific Director, X&Y Fertility and Executive Committee Member of the British Fertility Society).

BPS Health Division Conference 2024, Stirling Scotland

In June Claire will be delivering a Poster Presentation on "Klinefelter syndrome (XXY) and infertility: qualitative insights into the experiences of those affected". The abstract was co-authored with Professor James Elander and Dr Bryan Woodward.

UK Fertility Preservation Conference, Edinburgh, Scotland

Having seen Claire's presentation in Edinburgh, Dr Bryan Woodward will be presenting a Poster Presentation on "*Diagnosing the XXY karyotype:* an audit of the profiles of people referred to the endocrinology department of an NHS state-funded hospital in the UK". The abstract was co-authored with Dr AlJumaah (Higher Specialist Doctor), Dr Levy (Consultant Physician and Endocrinologist), Joella Wormleighton (Admin Manager), Dr Narendra (Consultant Physician and Endocrinologist), Dr Masato (Higher Specialty Doctor), Claire Harkin, and Professor James Elander, Dr. Dr Bryan Woodward to talk more about this at the KSA Conference.

THANKS Study – Huddersfield University

At the 2022 KSA Conference we were lucky to have Dr. Jenny Retzler talking about Executive Function problems often associated with KS/XXY. She is now hoping to conduct some research to understand the interplay between fatigue, cognition and neurodiversity, and the role of testosterone. Dr Retzler is currently applying for a grant to conduct this study and once this is approved the KSA will support the study with Claire representing the KSA as co-researcher for Public and Patient Involvement (PPI).

At the KSA Conference Dr. Retzler will be talking about this research which will be of great interest to the KS/XXY community.

Autism and XXY, Autism Research Centre, University of Cambridge:

The Autism Research Centre are particularly interested in the role of biological sex and steroid hormones in brain development. This research is being led by Dr Alexandros Tsompanidis and Professor Simon Baron-Cohan, who will talk more about the study at the KSA Conference.

RedCap Database

The KSA continues to support financially the setup of a RedCap Database, which is situated at the KS Clinics at Guy's and St Thomas'. This holds anonymized data about KS/XXY patients at the clinic which will inform the clinicians and help them to see trends. The KSA will continue to fund the management of the database going forward.

Support and information.

European Reference Networks (ERN) Activities:

ERNs are virtual networks which allow member clinicians to review a patient's diagnosis and treatment, exchange information and share knowledge with other members in their network. This sharing of information/expertise is particularly useful for Rare Diseases. Although KS/XXY isn't rare (or a disease), it qualifies because of its poor diagnostic rate. These networks also have patient representation. Chris Breen and Claire Harkin are representatives on the urology (UROGEN) and endocrine (ENDO) ERNs respectively and they will be leading with the creation of the KS/XXY Patient Journey.

What is a Patient Journey:

Medical professionals usually look at a patient's experiences/needs in a different way to the patient. The professional will rarely consider such things as the difficulty of attending multiple appointments, seeing different specialists at different hospitals on different days. They are unlikely to consider how bewildering the system can be. They often don't think that patients may need more or different support at specific times in their patient journey.

Patient Journeys are overviews designed from the patient's perspective. They will help medical professionals to visualize the whole process from the patient's viewpoint, allowing clinicians to address the needs of rare disease patients more effectively. Patient Journeys reflect the patients' personal experiences, which may vary depending on the person, clinic and country. In addition, Patient Journeys are a useful resource for patients, families, non-specialist clinicians, and the general public to help them understand the care needs of people living with KS/XXY.

As UROGEN and ENDO European Reference Network Patient Advocate representatives, Claire Harkin and Chris Breen will be leading with the creation of the KS/XXY Patient Journey.

We are delighted to have had the opportunity to contribute to the medical book **'Rare and Complex Urology'** which has been published on Elsevier. Claire Harkin and her fellow UROGEN Patient Advisory Group advocates were asked to co-author Chapter 4: The Patient view on the ERN programme: Share, care, cure and pathways.

This was a brilliant joint effort. Trustee AJ Howard made an important contribution to this chapter on Patient Journeys. We hope his powerful account of his experience of the psychological impact of a late KS/XXY diagnosis will help current and future clinicians understand the importance of early diagnosis and appropriate psychological and medical support for those with KS/XXY.

This has been a great opportunity to highlight the KSA and KS/XXY and the negative effects when correct care, from the correct person isn't available early in life.

A copy of the book will be available at the Conference for members to view.

In April 23 Chris Breen attended the General Assembly of the Endo Ern in Amsterdam.

Website

The main source of information provided by the KSA is their website which is often praised by people who contact us. However, I wonder when you last looked at it to see what is available. The Members' Handbook, information about events, recordings from previous KSA Conferences, webinars. It is all there!

Activity Weekends

KSA Activity Weekends were introduced in 2021 as a way of getting people who are affected by KS/XXY together in a relaxed environment. They provide a unique opportunity to meet others, of all ages, who are affected by KS/XXY. By the end of the weekend people seem to have grown in confidence and self-esteem and many say that they feel part of the wider KS/XXY community.

The April 2024 Activity Weekend was the biggest event we have ever had with 46 attendees – some of whom had to stay in local B&Bs because there was no more room at the Duke's Barn. As usual there was great interaction between parents, adults, young people and children and it was lovely to see many new faces there. We hope to be able to arrange another in September 2024. If you haven't already attended an Activity Weekend, do consider coming along. You don't even have to do the activities if you prefer not to. See our website for information https://www.ksa-uk.net/activity-weekend-2024/

Newsletter

The KSA sends out frequent mailings to its members via Mail Chimp. These contain information about events which have been organised by the KSA, news about current research, updates about fund raisers, opportunities to take part in research and any other information about the KS/XXY world which is likely to be of interest to our members. These mailings are very useful because they can be sent at short notice – unlike a printed newsletter. An annual compilation is made available on the website for members who like to have a printed copy. A limited number of printed copies are sent to members who are unable to access the internet.

Social events

Because our members are scattered across the country (and, indeed, the world) it is difficult to organise social events. To help plug this gap, two regular virtual chats have been arranged. The first, for all KSA members was initiated in January 2022 by trustee AJ Howard who usually hosts it. In November 2023, Vicechair, Alison Bridges started one for parents and carers. Members who join in have said that they value the opportunity to 'meet' and chat with others who understand their experiences. If you haven't already dropped in for a chat, why not give it a go even if you initially just listen?

There was one In-person social meeting in Derbyshire in August 2023. It was attended by 3 dogs and 8 members who enjoyed a chat, a walk and some sustenance in the sunshine. If anyone would like one of these events near them and can suggest a suitable venue, the trustees would be pleased to hear from them.

KSA AGM and Conference

This was again a hybrid 'in-person'/virtual event. It was held in May 2023 at the Stuart Hotel in Derby. This wasn't an ideal venue as there was not a proper breakout room but, having had to relocate and reschedule the conference 3 times due to a clash with a local festival, then a takeover of the hotel by the Home Office and finally a conflict with the date of King Charles' coronation, we were lucky it was available.

Despite the initial difficulties the Conference was very enjoyable. Amongst the speakers were representatives from both London KS/XXY clinics who ran 3 workshops for our delegates. Another very useful workshop on Advocacy was run by two of our members. The Conference helps to educate or members while developing peer support. Much on the content is available to members on the website.

Helpline and website contact form

In the last year the KSA have received and responded to nearly 250 Helpline calls and contacts through the website. The subjects of these vary enormously and include seeking a diagnosis, questions about behaviour or an elderly parent worried about how their adult son will manage when they can no longer support him. Often calls will take an hour or more because people are so grateful to find a sympathetic and supportive listener. Website contacts also often precipitate calls. A single contact can often result in multiple conversations. Alison Bridges and Ken Scott respond to the Helpline. Alison replies to website-initiated contacts but may involve other trustees as appropriate.

Virtual and in-person support at Guy's and St. Thomas' KS Clinic

KSA trustees have joined the team at Guy's and St Thomas' to virtually (and, when possible, in-person) to support patients and families at the Adults and Young Person's KS Clinics. They talk to patients and signpost them to support available outside of the clinics.

KS/XXY as an intersex condition.

People who have KS/XXY have atypical chromosomal pattern or karyotype. Because the chromosomes are neither XX- typical female or XY – typical male, KS/XXY is classed as an intersex condition. This can have serious ramifications when it comes to healthcare but that is often not understood by those affected by the condition or their medical professionals.

Intersex people often suffer from discrimination because their bodies may differ from the norm.
Prospective KSA Chairman, Paul Dutton, continues to work with intersex organisations to raise the profile and support the rights of all people affected by intersex conditions such as KS/XXY. Chris Breen and Paul Dutton attended the Interconnected UK conference in March where Paul ran a workshop on how to navigate everyday life when you don't quite look the same as everyone else.

In October 23 Chris joined the working group for the development of a KS module (I-KS) in sdmregistries (I-DSD). In November they took part in a webinar - Creating welcoming and inclusive spaces for intersex and transgender people in Higher Education https://podcasts.apple.com/gb/podcast/creating-welcoming-and-inclusive-spaces-for-intersex/id1694281354?i=1000636661831.
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In February 24 they took part in a seminar by the Healthy Aging Research Group - Intersex: Inclusion within LBGTQI+ https://harg.blogs.lincoln.ac.uk/2024/01/29/healthy-ageing-research-group-harg-lgbtqi-seminar-series/.

Membership of other organisations

The KSA is a member of, or works with, several useful organisations as detailed below:

- Ken Scott is a patient representative on the I-DSD steering committee and on the Rare Conditions' Patient Advisory Group.
- Chris Breen is a member of Mental Health Partnership Network for Rare Diseases.
- AJ Howard and Claire Harkin are patient representatives on the NICE* forum, discussing the proportionate approach to technology appraisals and improvements to the NICE* guidance concepts.
- the National Council for Voluntary Organisations (NCVO)
- the Prescription Charges Coalition
- EURORDIS-Rare Diseases Europe
- ENDO European Reference Network
- UROGEN European Reference Network
- Genetic Alliance and Rare Disease

- Rare Conditions Network
- the KSA is affiliated to the British Endocrine Association which gives it some authority when corresponding with medical professionals.
- the KSA has representation on the planning board for the new KS Pediatric Clinic at Guy's and St Thomas'. There will be more information about this at the KSA Conference.

Structure, Governance and Management

The KSA has been a charity since 1996 and is run by trustees who form the National Executive Committee (NEC). They are elected at the AGM or can be co-opted at any time. The KSA generally recruits from its membership but would be happy to recruit from outside if the opportunity presented itself.

Trustees

It is with great sadness that I must report the death of Barry (Badger) Duplock who was Chair of the KSA from 2019 until his sudden death in May 2023. By his own admission, Badger struggled with admin and organisation but, when it came to giving friendship and support, he shone. He loved people and made everyone he knew feel special. He is sorely missed.

After 4 years as Membership Secretary, Jim Harkin is stepping down due to pressure of work and trustee David Adams resigned in February 24. Alison Bridges is standing down as Vicechair after 5 years in the role. We would like to thank them for their time and support.

Sylvia James-Yates was co-opted as trustee and Honorary Secretary in June 23 and Sarah Gordon as trustee and Honorary Treasurer in October 23.

Paul Dutton has been nominated for the position of Chairman. Although Paul is not currently a trustee, he has been involved with the KSA for many years and previously served for 6 years as a very effective Chairman.

Chris Breen who became a trustee last year has already proved their capability to take on the role of Vicechair for which they have been nominated.

Melanie Perrins has been nominated as Membership Secretary and Phillipa Reed as trustee. Like Sylvia and Sarah, both prospective trustees have excellent skills which will be of use to the KSA.

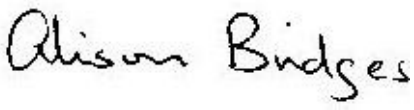
This promises to be the strongest and most effective NEC for many years.

Volunteers

We now have a very small group of volunteers who cannot commit to being trustees but who help in the everyday running of the KSA. Their support really makes a difference.

Miscellaneous

I would like to thank everyone who donated, told someone else about the KSA (or KS/XXY), fund raised, organised a meeting, contributed to the Newsletter or helped the KSA community in any way. A little help from many people adds up to a great deal. You really make a difference. Personally, I would like to thank my fellow trustees, over the years, past and present for their endless support, encouragement and understanding. They were, and are, simply wonderful.



Alison Bridges

Vicechair of Trustees Klinefelter's Syndrome Association (KSA)

* National Institute of Health and Care Excellence (NICE)