Annual Report by Trustees of the Klinefelter's Syndrome Association 2022 to 2023

Objectives and activities of the KSA

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

The KSA also promotes and supports research into KS/XXY.

Achievements and performance

KSA Attendance at Medical Conferences and Shows

To raise awareness amongst the medical profession, the KSA exhibits at medical conferences throughout the year.

The KSA exhibited at the British Endocrine Society (BeS) Clinical Update in Birmingham in April 2022.
The BeS conference in Harrogate in November 2022 was attended by Ken Scott and Chris Breen. It was particularly useful to have KS/XXY adults available because the medical professionals could engage directly with them.

The RCGP did not hold their annual 2-day conference this year and it was decided that attendance at a single day event in London would not be cost-effective.

AJ Howard and Ken Scott both attended Rare Diseases Day events on behalf of the KSA in February 2023.

Activity Weekends

The August 2022 Activity Weekend was well attended with twenty-eight residential members. We were delighted to see so many happy smiling faces. We ran an equally enjoyable one in April 23 with thirty attendees. It was particularly satisfying to welcome five new adult attendees. The Activity Weekends provides an opportunity to meet others, of all ages, who are affected by KS/XXY.

Raising Awareness Day

A small but dedicated team comprising of AJ Howard and Edward Dobie with assistance from David Adams, have spent the last two months preparing a campaign for Klinefelter's Syndrome Raising Awareness Day on 10th May 2023. They have learned new skills and are already looking forward to next year!

Social events

The regular Zoom chats initiated by trustee AJ Howard have proved very popular. They provide a great opportunity to chat, debate or just listen and feel less isolated. Currently, they run every other Thursday.

KSA AGM and Conference

It was so great to be able to hold a hybrid 'in-person'/virtual AGM and Annual Conference in June 2022 in Huddersfield University. This was an excellent venue and we are very grateful to the University for making it available to us. We had excellent speakers including a number from the Klinefelter Clinic at Guys/St Thomas. This was our first attempt at running a hybrid conference allowing us to reach a wider audience. We are repeating the format this year.

Improving the rights of people who have intersex conditions such as KS/XXY.

One of our volunteers, ex-chair of trustees, Paul Dutton, continues to work to with intersex organisations and it was through this association that we were permitted to use Huddersfield University for the KSA Conference.

Research

In 2021/2022 Claire Harkin, KSA Secretary, conducted a qualitative research study for a Psychology MSc. The study is about gender identity among KS/ XXY individuals. The study has since been concluded and a manuscript is being reviewed by the Journal of Genetic Counselling. The University of Derby has recognised the importance of the need for more research in this area and has granted a PhD studentship to take it further over the next three years. Claire and her supervisor, Professor James Elander, will be presenting this at the Annual Conference.

The KSA have been liaising with Sir Simon-Baron Cohen and his research team at the Autism Centre at Cambridge University, where they are currently working on gaining ethical approval to conduct research on the prevalence of Autism amongst those with KS/XXY.

Recently the KSA have become key stakeholders representing the KS/XXY community in the Genomics and Newborn Blood Spot Screening Research project based in the province of Victoria, Australia. For the next three years every child born in Victoria will be screened for a number of rare diseases, including KS/XXY! The project has already started and the evidence generated will be incredibly powerful in helping us to convince NICE* that the same needs to happen in the UK.

RedCap Database

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

Membership of other organisations

Claire Harkin and Chris Breen represent the KSA in two European Reference Networks (Urology and Endocrinology), which work to improve diagnostics and treatment for patients for rare conditions. KS/XXY is considered rare only because it is underdiagnosed.

Ken Scott is a patient representative on the I-DSD steering committee and on the Rare Conditions' Patient Advisory Group.

Claire Harkin and AJ Howard are patient representatives on the NICE* forum, discussing the proportionate approach to technology appraisals and improvements to the NICE* guidance concepts.

The KSA is a member of:

- the National Council for Voluntary Organisations (NCVO),
- the Prescription Charges Coalition
- EURORDIS-Rare Diseases Europe
- Genetic Alliance and Rare Disease
- Rare Conditions Network

The KSA is also affiliated to the British Endocrine Association which gives it some authority when corresponding with medical professionals.

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.

Trustees

There has been no change in trustees during this year. However, according to our constitution, trustees must stand down after 2 years – although they may stand for re-election. Sadly, Roz Heinze has decided to stand down permanently, having been a trustee since the KSA was formed in 1996. Roz has worked tirelessly over the years and will be very sorely missed.

Treasurer Mike Green is relinquishing his post having been treasurer for the maximum term of 6 years. Thankfully, he is standing for re-election as a trustee – as are the other current trustees.

We are delighted to have two new trustees standing for election. Both have worked as volunteers and we know they will both have a positive impact on the KSA.

Volunteers

We now have a 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. We would particularly like to thank Elaine Bennett who organised a Black Tie event in Bristol in May. Thanks to her hard work and the generosity of all involved, she raised over £1,500.

Barry Duplock

Chair of Trustees Klinefelter's Syndrome Association (KSA)

* National Institute of Health and Care Excellence (NICE)