7 March 2014

Attention:
GLBTIconsultations@health.vic.gov.au
community.affairs.sen@aph.gov.au
Professor Christopher Baggoley, Chief Medical Officer, Australian Government, GPO Box 9848, MDP 84, Canberra ACT 2601

Re: Regarding Community engagement – Gay, lesbian, bisexual, transgender and intersex (GLBTI) health and wellbeing 2014: The Senate: Community Affairs References Committee: Involuntary or coerced sterilisation of intersex people in Australia, October 2013

Response from Australian Klinefelter XXY Support Group

The Australian Klinefelter XXY Support Group brings together state support groups for parents and individuals affected by 47,XXY (Klinefelter Syndrome), 47XYY, XXX, XYY and other sex chromosome aneuploidies (SCAs). We are supported by KS&A / AAKSIS (USA). We are parents of children with XXY / SCAs, and XXY adults.

We use XXY, 47,XXY and Klinefelter Syndrome interchangeably.

1. We are aware that XXY and other sex chromosome aneuploidies are classified with the group of medical conditions known as “disorders of sex development”. The medical diagnosis of disorders of sex development is different from the gender self identification of an older individual as male, female or intersex. The vast majority of 47,XXY boys and men identify as male and not as intersex.

   • 47,XXY boys have a sex chromosome variation; this does not typically affect their appearance (phenotype) or gender identity
   • Ambiguous genitalia is not a feature of 47,XXY
   • 47,XXY adults occasionally identify as intersex or other genders later in life, and we welcome and support this diversity, but in no way should this have impact on terminology used, care or treatment for 47,XXY boys

Follow-up studies of XXY individuals diagnosed through newborn screening surveys would suggest that almost all of these babies will be phenotypically male and identify as male... However, although probably only a small minority, individuals with XXY who do not identify as male do indeed exist. Helihy and Gillam

Our position includes recognition that XXY individuals who, at the age of puberty, exhibit signs of gender confusion or clear gender identity that is not male should receive gender counseling before considering testo therapy... When there is no doubt, gender counseling would be a waste of money, and it could be traumatic for a young boy to be subjected to questions about his gender...implying that he's not male enough. Imagine a pre-pubescent young boy who already may have body image issues and social and developmental delays being subjected to this kind of
Gender dysphoria applies to a tiny minority of individuals with XXY, and it is ridiculous to thwart proven therapies that work for the vast majority in deference to advocates for a tiny minority. - Jim Moore, KS&A

The report examines care for people who later identify as intersex and surgical considerations for children with ambiguous genitalia, and extends its recommendations to 47,XXY and other sex chromosome aneuploidies. This is incorrect. 47,XXY has a well researched and accepted treatment schedule and should not be wrapped up in the recommendations for intersex.

There is a big difference between stating that some adults with XXY consider themselves to be “intersex” and stating that XXY is an intersex condition. – Virginia Isaacs

2. We are particularly concerned about recommendations 3, 5, 6 and 7:

Recommendation 3… The guidelines should favour deferral of normalising treatment until the person can give fully informed consent, and seek to minimise surgical intervention on infants undertaken for primarily psychosocial reasons.

These recommendations may lead to 47,XXY boys being denied appropriate medical treatment, or support for learning and psychosocial difficulties.

Effects of 47,XXY may include

• General motor and neuro development delays
• Emotional, social and behavioural challenges
• Speech and language and learning difficulties
• Cognitive processing problems, Auditory Processing Disorder
• Autism Spectrum Disorder traits
• ADHD traits
• Low muscle tone and gross and fine motor skills challenges
• Hormone imbalance and lack of testosterone creating fatigue, increased height, hypogonadism, gynecomastia, sterility
• Health risks including anxiety and mood disorders, osteoporosis, cancers

With this complex mix of challenges, it is key that 47,XXY individuals have access to appropriate medical treatments through their lifespan.

Specifically, adolescents commonly require appropriately timed testosterone therapy to develop emotional stability and secondary sexual characteristics (facial hair, deep voice, male muscle and fat distribution) and normal bone density. Lack of testosterone leaves

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1 Email correspondence with Monica Bray, 4 March 2014. Refer www.genetic.org
2 Email correspondence with Susannah Bowen, 1 March 2014. Also refer Living With Klinefelter Syndrome
individuals fatigued, depressed and anxious. Latest research investigates benefits of testosterone for newborns and at primary school age.³

**Worldwide research agrees that best practice for 47,XXY includes testosterone therapies.** This is supported by individuals, families and the medical professions in Australia, USA and worldwide. If 47,XXY is included under these recommendations, there is the risk that essential treatments such as testosterone therapies may be withheld.

Many 47,XXY men who have not had access to testosterone treatment due to late diagnosis suffer from social stigma, bullying and depression as well as physical health impacts such as osteoporosis. Our boys are already at risk of illness, marginalisation and exclusion. Denying appropriate medical treatment raises risks unjustifiably. In a first-world country such as Australia, linking early medical intervention to approval by the Family Court is incomprehensible.

*Klinefelter Syndrome and other sex chromosome aneuploidies... should not be considered in way ‘intersex’ and should not be included in any policy regarding the management of ‘intersex’ disorders. Klinefelter boys and men almost exclusively identify as male and Turner Syndrome girls and women unanimously identify as female. To include them in any complicated management ‘intersex’ management strategy will lead to delays in their appropriate care. - Professor Paul Hofman, Chair of the Disorders of Sexual Development and immediate past president, Australasian Paediatric Endocrine Group⁴*

*Removing the option of testosterone prior to the age of 18 when one can "consent" would do a disservice to those boys who either fail to enter puberty or whose puberty stalls. If they are very distressed about this, and many are, testosterone treatment would be precluded even where the young man, his parents and his doctors believe that testosterone treatment would benefit him. Holding off on T treatment until age 18 can result in early osteoporosis as well as body disproportion because the growth plates may fail to close at the appropriate time. It can also negatively affect academic performance and progress toward work. - Virginia Isaacs Cover⁵*

We ask that recommendations be amended to specify that individuals with sex chromosome aneuploidies such as 47,XXY are not included in recommendations for people who identify as intersex.

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⁴ Statement from Professor Hofman to Susannah Bowen by email, 4 March 2014
⁵ Email correspondence with Susannah Bowen, 1 March 2014. Also refer Living With Klinefelter Syndrome
3. We call for multidisciplinary medical care for these complex conditions.

47,XXY individuals may require support from

- Endocrinology
- Occupational therapy,
- Psychology and psychiatry
- Neuropsychology
- Speech and language therapy
- Genetic counsellors

Best practice overseas sees multidisciplinary clinics provide holistic care, become centres for family support and drive research, such as the eXtraordinarY kids Clinic at Children’s Hospital, Colorado; eXceptional Kids Clinic at EmorY, Atlanta; Johns Hopkins Klinefelter Syndrome Centre.

One of the authors of this letter, Susannah Bowen, has a 9 year old son who is XXY and has a cleft lip.

- For his cleft condition, Leo attends Cleft Clinic at RCH Melbourne. He sees a plastic surgeon, paediatrician, speech therapists, and specialist dentists on the same visit. The team discuss his needs and treat him holistically.
- For XXY, Leo individually sees endocrinologist, speech and language therapist, OT, psychologist, special education support, and paediatrician. These specialists work in isolation and rely on Leo’s mother to communicate information between them about his treatment.

There is no specialist multidisciplinary clinic for children or adults with SCA anywhere in Australia.

Australia requires a multidisciplinary support service for XXY and SCA individuals, and a diagnostic and management protocol that reflects current global knowledge and expertise.

Thank you

Susannah Bowen, Melbourne
Monica Bray, Sydney
Bec Waters, Sydney
Leisa Hislop, Adelaide
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References

Bojesen and Gravholt: Klinefelter syndrome in clinical practice, 2007 (Nature)

Cover, Virginia Isaacs: Living with Klinefelter Syndrome, Trisomy X and 47XY, 2012 (Cover)


KS&A / AAKSIS www.genetic.org

Radiocioni et all: Strategies and advantages of early diagnosis in Klinefelter’s Syndrome, 2010 (Molecular Human Reproduction)


Multidisciplinary clinics for 47,XXY and sex chromosome aneuploidies:

The eXtraordinarY Kids Clinic at Children’s Hospital Colorado. Children see a developmental pediatrician, endocrinologist, speech therapist, occupational therapist, clinical psychologist, neuropsychologist and a genetic counsellor. http://www.childrenscolorado.org/conditions-symptoms/conditions/x-y-chromosome-variations

eXceptional Kids Clinic at EmorY, Atlanta

Johns Hopkins Klinefelter Syndrome Centre (for adults) http://klinefelter.jhu.edu/