Submission to the Australian Human Rights Commission from the Australian X and Y Spectrum Support (AXYS)

Protecting the Human Rights of People Born with Variations in Sex Characteristics in the context of Medical Interventions

September 2018

Preamble

Australian X and Y Spectrum Support (AXYS) wish to acknowledge the substantial work and achievement of the Australian Human Rights Commission (AHRC) who made available the Consultation Paper (the Paper) - Protecting the Human Rights of People Born with Variations in Sex Characteristics in the context of Medical Interventions. AXYS commends the AHRC for identifying, among other things, the moral, legal and medical imperatives in the Paper that require consultation, and reasonable resolution around complex issues.
AXYS is a group comprising about 350 members around Australia, working in consultation with various health professionals, educators, sponsors and supporters with an interest in, or directly affected by X & Y sex chromosome variations, most commonly Klinefelters Syndrome (KS)/XXY. AXYS was started as a special media interest group about six years ago by concerned parents; desperate for information about X & Y sex chromosome variations as they relate to KS/XXY and other variations e.g.: 48XXXY, 49XXXXY, XYY, 48XXYY and XXX.

AXYS is affiliated to like-minded organisations in Australia and overseas and will remain a much-needed conduit between the public, politicians, Government, administrators, health professionals, educators, employers, sporting, leisure and social groups concerned about X & Y sex chromosome variations.

KS/XXY is the most common sex chromosome condition in men affecting about 1:500-650. The condition is not widely understood by many health professionals, educators, individuals, families, relatives and others. Many individuals experience lifelong challenges that are physical, mental or social impediments. Appropriate support is crucial, especially early interventions.

X & Y sex chromosome variations present as a spectrum with everyone being affected in different ways and with differentiate severity. Individuals may experience;

- Fine and gross motor delay due to hypotonia
- Infertility or significantly lowered fertility
- Hypogonadism (low levels of testosterone)
- Speech delay/expressive language disorder
- Mild autism
- Developmental delays
- Learning challenges
- Attention deficits
- Sensory processing disorders
- Mental health impacts: depression, anxiety, mood swings
- Gynecomastia
- Dental health and taurodontism
- Health impacts: such as testicular cancer, diabetes, osteoporosis, autoimmune diseases, venous conditions, lung disease, sleep apnoea, thyroid insufficiency[1].

AXYS, until recently, was self-funded with a dedicated team of volunteers working to increase awareness and support. It has taken six years to evolve a formal connection between

[1] **Cover, V.I:** ***Living with Klinefelter Syndrome, Trisomy X, and 47, XYY : A Guide for Families and Individuals Affected by X and Y Chromosome Variations,*** **2012, Virginia Issacs Cover, New York.**

those affected by X & Y sex chromosome variants and the wider community of health professionals, educators and others affected by these variants. This is an ongoing pursuit that requires the continued work and dedication of volunteers to manage the significant growth in enquiries and the diversity of information and services that AXYS provides.

AXYS, with financial assistance through the Victorian Government's LBGTI Community Grants program – Organisational Stream, is rapidly moving to fulfil their need for dedicated specialist equipment and an administration that will allow for robust interactions, receipt and processing of information and specialised distribution of our services.
This submission, is limited to directly address pertinent questions in the Paper that may affect individuals with an X & Y sex chromosome variant diagnoses, their parents and health professionals overseeing their care.

The submission is made for and on behalf of the AXYS membership. AXYS submission relates to X & Y sex chromosome variations and should not be taken out of context in dealing with terminology and a debate and resolutions arising from the Paper.
In other words, AXYS do not want the consultation process to inadvertently allow government, whether by legislation or other intervention to make determinations about X & Y sex chromosome variations that impacts the needs, aspirations and imperatives that relate to X & Y sex chromosome variants such as KS/XXY and other variations e.g.: 48XXXY, 49XXXXY, XYY, 48XXYY and XXX.

Considerations relating to intersex variations that are not X & Y chromosome variants are not addressed by the AXYS submission, nor does AXYS seek in any way to offer advice in relation to the experiences or needs of people with those variations and their carers.

AXYS fully supports the underlying human rights principles which require all decisions made on behalf of a minor, to be made in the best interests of the child. To that end, AXYS works tirelessly to support individuals with X & Y sex chromosome variations and strongly encourages parents and carers to work in collaboration and consultation with health professionals taking account of evidence-based research to make informed decisions about the individual treatment options available for their child to ensure they can help minimise the impacts associated with X & Y sex chromosome variations. AXYS also supports age and cognition appropriate involvement of minors in decisions and disclosure of information about their own medical history. There is a large body of evidence which supports early intervention, including medical interventions, that can help ensure that individuals with X & Y sex chromosome variations maximise their prospects to lead a much-improved quality of life: to live life to the fullest as children, adolescents, and adults in the home, school, work, sporting, leisure and social environments.

The Submission: We now turn to the Discussion Questions

Terminology

1. Is the term 'people born with variations in sex characteristics' appropriate, or is there a better way to describe the people who are the subject of this Consultation Paper?
"X & Y sex chromosome differences like 47, XXY (commonly diagnosed as Klinefelter syndrome)" is identified only once in the Paper as falling within the purview of the Paper at section 3 clause 23; being under the 'Intersex' umbrella.

AXYS believe that the use of the term 'people born with variations in sex characteristics' is appropriate for the majority of people who are subject of this Consultation Paper but do not necessarily feel that the terminology accurately encompasses individuals with X & Y sex chromosome variations that AXYS support and advocate for.

Understanding lived experiences

2. Broadly, how would you describe your experiences in the context of medical interventions?

Parents and carers in the AXYS community believe that making considered, individual, child-centred decisions for X & Y sex chromosome variations in consultation with health professionals, should not be viewed as harmful within parameters as set out by the Commission.

As mentioned above X & Y sex chromosome variations are a wide spectrum. Medical intervention needs to be made on an individual basis, and in consultation with medical professionals, drawing on evidence-based research. This process should include medical reviews of an individual's bloods, bone scan, physical signs and symptoms which often include motor and language delays. Psycho-social assessments may also be relevant. AXYS members report, that those who have had a collaborative and informed approach to treatment options have had very positive experiences with decreased health risks if the appropriate treatment methods are used[2].

Medical intervention for individuals with a diagnosed X & Y sex chromosome variation could include:

- Early testosterone boosters in infants: (If deemed necessary in consultation with medical professionals and an individual assessment of the infant). These are very low dose testosterone boosters of 25mg or less, where 1 to 3 doses are given as a stand-alone treatment option to assist in increasing muscle tone, language development, microphallus and to assist in decreasing anxiety. There are research studies available which indicate these benefits, however there is no current evidence-based research identifying that low dose T boosters in infants alters sex characteristics[3].
- Testosterone or other hormone treatment in children nearing puberty: (The majority of individuals with Klinefelter's syndrome will not be able to enter puberty naturally which can result in numerous health implications). The child or teenager is holistically assessed including a clinical review if they are not showing signs of entering puberty naturally e.g. through blood tests, bone scans, bone density tests and through a mental health and general wellbeing check-up.
- Testosterone and hormone treatment in adults: (If deemed necessary in consultation with medical professionals and on assessment of the individual).
- Micro TESE: The majority of males with KS/XXY have hypogonadism and significantly low sperm counts leading to infertility. Micro TESE can open the possibility for these individuals to have biological children of their own. Some research indicates this decision needs to be made sometimes when a child is as young as 15. Unfortunately, due

[2] Turriff A, Macnamara E, Levy HP, Biesecker B. The Impact of Living with Klinefelter Syndrome: A Qualitative Exploration of Adolescents and Adults. *Journal of genetic counseling*. 2017;26(4):728-737. doi:10.1007/s10897-016-0041-z.

[3] Samango-Sprouse C, Stapleton E, Gropman A, et al. Positive effects of early androgen therapy on the behavioral phenotype of boys with 47,XXY. *American Journal Of Medical Genetics Part C-Seminars In Medical Genetics* [serial online]. n.d.;169(2):150-157. Available from: Science Citation Index, Ipswich, MA. Accessed September 15, 2018.

to no Government subsidies, many cannot afford to have this procedure undertaken and suffer depression and mental health concerns due to not being able to have their own biological children[4].

Medical interventions common to the general population

- Orchidopexy surgery: For children in the general population (i.e., without X & Y sex chromosome variations) with undescended testicles, orchidopexy surgery is considered necessary. Children with KS/XXY may also require access to this surgery, as they already carry a higher risk of testicular cancer, decreased fertility and testicular torsion; further increased if they have ascended or retractile testicles, a common occurrence[5].
- Insulin injections due to Type 1 diabetes: Individuals with KS/XXY have increased risks of diabetes[6].

3. What are the current Australian sources of information and education about the experiences of people born with variations in sex characteristics?

AXYS where possible and where appropriate provides education, support, sharing and information to people affected by X & Y sex chromosome variations (XXY, XYY XXX). The AXYS service is all about targeted information management, our relevant web site portal, health professionals, support networks, early intervention, Facebook (public and secret), email contact, help line, annual conference, face to face group engagement, regular Leadership Team meetings and telephone conferences. Nevertheless, urgent government support and resources are required to extend the depth and reach of information available. Although KS/XXY is quite common (1:500-650 males), there remains a greater need for more concise and accurate information about these conditions to be readily available. Also, there is a distinct lack of knowledge and awareness about intersex terminology and why X & Y sex chromosome variations come under the umbrella term "*Intersex".*

AXYS understands the vital need to increase knowledge and awareness. We therefore make representations on behalf of our members to the Australian Human Rights Commission Intersex Reference Group, Victorian Intersex Advisory Group, communicate with health professionals at Royal Children's Hospital, Genetics Support Network Victoria and participate in focus groups, forums and other events about intersex awareness and education.

4. Are there gaps and/or inconsistencies in sources of information and education that are available about the experiences of people born with variations in sex characteristics? If so, what is the impact of this?

AXYS has identified major shortfalls in available resources. The impact for those diagnosed later in life without early intervention and support is devastating in some situations with far reaching consequences for individuals who experience severe symptoms. There is a lack of

[4] Aksglaede L, Link K, Giwercman A, Jørgensen N, Skakkebæk NE, Juul A. 2013. 47,XXY Klinefelter syndrome: Clinical characteristics and age-specific recommendations for medical management. American Journal of Medical Genetics Part C (Seminars in Medical Genetics) 163C: 55–63.

[5] Aksglaede L, Link K, Giwercman A, Jørgensen N, Skakkebæk NE, Juul A. 2013. 47,XXY Klinefelter syndrome: Clinical characteristics and age-specific recommendations for medical management. American Journal of Medical Genetics Part C (Seminars in Medical Genetics) 163C: 55–63.

[6] Jiang-Feng M, Hong-Li X, Liang-Ming L, et al. Original article: Prevalence and risk factors of diabetes in patients with Klinefelter syndrome: a longitudinal observational study. *Fertility And Sterility* [serial online]. November 1, 2012;98:1331-1335. Available from: ScienceDirect, Ipswich, MA. Accessed September 15, 2018

relevant and up to date information for Australians with an X & Y sex chromosome variation. It is evident with our adult members who have had little or no support that the impacts of this lack of knowledge and support can have major negative impacts on their lives often leading to health and mental wellbeing challenges.

Consent

5. How is the consent of a person born with a variation in sex characteristics currently sought prior to a medical intervention?

For many with an X & Y sex chromosome variation it depends on the age of the individual if consent is sought and obtainable. For infants and young children, where age or cognitive appropriate disclosure and discussion about intervention is not possible, responsible parents or carers consent to interventions on behalf of the child. Generally, according to AXYS members, there is a discussion between parents and one or more health professionals and then, based on further professional assessment of the child, it is sometimes deemed that there is a significant need for early intervention in the best interests of the child.

For children and teenagers reaching puberty, the child is usually privy to discussions between the responsible parents or carers and health professionals. Children can give consent if they are deemed Gillick competent. Informed consent is given following consultation conducted on medical grounds with a holistic view of the child; that may include, mental health and well-being checks, blood tests, x-rays and bone density scans. Evidence-based research is often reviewed using this collaborative approach on the best health outcomes for the child.

If a child isn't deemed Gillick competent, or if an adult isn't deemed competent to give informed consent due to delayed or impaired cognitive capacity; in the experience of AXYS members, a parents' or carers' informed consent is sought in consultation with medical professionals.

However, AXYS would encourage a review of the training and continuous professional development offered to medical professionals about X & Y sex chromosome variations, including appropriate discussions around informed consent.

6. How do current guidelines or protocols relating to the medical management of people born with variations in sex characteristics deal with the issue of consent, including the ability to withdraw any consent given at any time?

There are currently very few guidelines available around the management of X & Y sex chromosome variations. The current guidelines/protocols pertinent to hormone replacement outline the indicators for its use but AXYS has been unable to find consent guidelines. Substantial resources must be re-directed to assist parents, professionals and allied health personnel and advisors to ensure that best practice is readily available to parents and their children. In the case of later diagnosis of people with KS/XXY then similar support networks and greater awareness need to be established for the public, politicians, Government, administrators, health professionals, educators, employers, sporting, leisure and social groups.

As few as 25% of people with an X & Y chromosome variant are diagnosed which indicates a great need for better diagnostic tools and indicators to be implemented across the health system[7].

[7] A.F. Radicioni, E. De Marco, D. Gianfrilli, S. Granato, L. Gandini, A.M. Isidori, A. Lenzi; Strategies and advantages of early diagnosis in Klinefelter's syndrome, *MHR: Basic science of reproductive medicine*, Volume 16, Issue 6, 1 June 2010, Pages 434–440, https://doi.org/10.1093/molehr/gaq027

AXYS is aware of the current Victorian Department of Health, 'Decision making principles for care of infants, children and adolescents with intersex conditions' which resource is aimed at achieving the best possible outcomes for infants, children and adolescents with intersex conditions who are cared for in Victorian hospitals. Relevantly this includes:

- assistance for parents about informing their child in stages about their condition, and with seeking their child's consent for any medical or surgical intervention;
- an acknowledgment that hospitals should test decisions against the Victorian Charter on Human Rights, including the right to protection from medical treatment without consent.

The Victorian principles are currently under review and AXYS is contributing to this process through the Victorian Intersex Expert Advisory Group, convened by the Department of Health.
Consequences arising from the term "intersex" as enunciated in 'Decision making principles for care of infants, children and adolescents with intersex conditions' and the 'Victorian Charter on Human Rights' should not, in this submission, be seen as an endorsement for the medical management of people with X & Y chromosome variations.

7. *What practices/safeguards are in place to ensure any consent obtained remains informed?*

AXYS believes that the above-mentioned safeguards are already in place around protecting children with X & Y sex chromosome variations. Decisions are made collaboratively with a holistic approach for these children ensuring intervention is targeted to improve health, physical and mental wellbeing.
AXYS strongly condemns any medical intervention being performed without informed consent of either a competent individual, or the relevant parent/carer where appropriate.

8. *What could enhance the capacity of people born with variations in sex characteristics or their caregivers to provide full and informed consent?*

Enhancements, for the parents or carers of X & Y sex chromosome variants to provide full and informed consent include the following:

- There is a very low incidence of identification of X & Y chromosome variants both pre-natal and post-natal[8], though pre-natal diagnoses are growing significantly due to widespread availability for NIPT and other pre-natal testing; never-the-less, there is a significant cost to testing and individuals and couples are ill-informed of what anomalies can be detected.

  More expansive clinical trials will identify a much larger group that should be the target for early intervention to address age specific needs[9].

- Increased awareness and knowledge about X & Y sex chromosome variations provided to health professionals, those affected and our wider community including

[8] A.F. Radicioni, E. De Marco, D. Gianfrilli, S. Granato, L. Gandini, A.M. Isidori, A. Lenzi; Strategies and advantages of early diagnosis in Klinefelter's syndrome, *MHR: Basic science of reproductive medicine*, Volume 16, Issue 6, 1 June 2010, Pages 434–440, https://doi.org/10.1093/molehr/gaq027

[9] : *Reference: Klinefelter Syndrome in Childhood: Variability in Clinical and Molecular - Findings published June 2018 J Clin Res Pediatr Endocrinol. 2018 Jun; 10(2): 100–107).*
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5985377/

the public, politicians, government administrators, educators, employers, sporting, leisure and social groups. Education is particularly important for reaching stakeholders not directly affected and for normalising the experience of people with X & Y sex chromosome variations.

- Increased and better information that is relevant and pertinent to the individuals needs including expert opinion and sharing with others who already have some experience with X & Y sex chromosome variants.

Consent in the absence of legal capacity

9. To what extent should parents and carers be involved in making decisions on behalf of their child? How can parents and carers be best supported to make these decisions?

No medical interventions should take place on a child without parent or carer informed consent. Medical professionals must involve parents or carers to the full extent and provide them with all the information they require to make an appropriate decision, including the most up-to-date evidence-based research.

AXYS members feel strongly that parents are acting in the best interest of their children to ensure their delays and challenges are addressed and any health concerns are reduced. This is best ensured when parents are equipped.

Parents will invariably seek available information, research and expert opinions. AXYS relies upon the support of government, health professionals, allied health, community and member reassurance and feedback to provide much needed general information to support the decision maker parents and carers.

10. What, if any, legal oversight mechanism(s) should be in place to guide decision-making about medical interventions involving a person born with a variation in sex characteristics where the person does not have the legal capacity to provide consent?

Note: This response is specific to AXYS members for individuals with an X & Y sex chromosome variation. There may be many other varied situations for intersex conditions that are not X & Y sex chromosome variations.

Legal oversight mechanism(s) are not the appropriate forum for making decisions about medical interventions. AXYS is concerned that a legal mechanism, particularly court-based decision making or other adjudicative system, would lead to complications, conflict, issues about interpretation, adversary situations, unnecessary cost and potential harm for individuals and families.

11. If such a mechanism existed

AXYS believes this would have a negative impact on individuals with an X & Y sex chromosome variation requiring the above discussed medical intervention/s specific to care for people with X & Y sex chromosome variations.

Health professionals, guided by appropriate decision-making principles including the best interests of the child, and supported with resources to undertake individual holistic needs-based assessments, are the appropriate place for decisions about the appropriateness and necessity of interventions for people with X & Y sex chromosome variations.

Removing this decision-making capacity from individuals, families and health professionals could have huge health and life long impacts resulting in an individual not being able to achieve their full potential, increasing the risk of social isolation, mental health concerns and other health impacts reducing longevity of life.

- *How could this mechanism adequately address different interventions and different variations?*

While AXYS is not commenting on safeguards which may be required for other variations not addressed in our submission, nevertheless, it is our experience that insufficiently granular definitions lead to conflation of the needs of different intersex variations. Failing to account for valid differences, particularly in relation to supportive interventions for individuals with X & Y sex chromosomes, is one of the additional reasons AXYS does not support the introduction of a legal mechanism.

Medical necessity

12. Would a legal definition of medical necessity or therapeutic treatment be helpful

No, AXYS does not support the creation of a legal definition of medical necessity or therapeutic treatment. In order to provide for differentiated need between intersex variations, flexibly adopt new research or best practice, and allow for individual assessment, these definitions should not be legally enshrined.

and, if so, what should the definition be?

AXYS emphasises its support for appropriate protections, where required, for other intersex variations. However, the capacity of individuals with an X & Y sex chromosome variation and the parents and carers who support them to access interventions where informed consent is given must not be impacted by the introduction of legal definitions. Based on our extensive involvement in discussion in Victoria around defining medical necessity AXYS is concerned that a legal definition would fail to make the necessary clear distinctions to ensure that individuals with an X & Y sex chromosome variation are not adversely impacted.

13. What are the permissible rationales/considerations that should be taken into account when determining whether or not to undertake a medical intervention on behalf of those who lack the capacity to consent?

Above we described how all medical interventions need to be made on an individual basis, in consultation with medical professionals and drawing on evidence-based research. For children, or those without the capacity to consent, a responsible parent or carer must provide informed consent.

Every variation and every individual will have a series of differentiated experiences and reasons why a medical intervention should or should not proceed. An appropriate response would be principles-based approach which requires medical staff to take into account the best interests of the individual, likely outcomes – including the impact of not proceeding, and provision of full and frank information to parents or carers in order that they might give informed consent.

Within the AXYS community, typically parents and carers are required to give informed consent on behalf of children. Supporting the evolution of networks, more and better information and promoting the sharing of resources is crucial to help instil trust in parents and carers to work with medical professionals to make the best possible decision for their child.

Regulation – Legal and policy prohibitions

14. Should all non-emergency and/or deferrable medical interventions that alter a child's sex characteristics, where the child does not have legal capacity to consent, be prohibited by law? If so, should this prohibition be civil or criminal?

AXYS strongly believes there should be no legal prohibition on the medical interventions we describe in our submission for X & Y sex chromosome variations. We are concerned that

discussion around defining non-emergency and/or deferrable medical interventions has indicated that current, evidence-based interventions for X & Y sex chromosome variations may be inadvertently captured under these terms. This may result in unexpected, or potentially harmful outcomes for individuals with X & Y sex chromosome variations.

Regulation – Clinical guidelines

15. What are the current approaches to the management of people born with variations in sex characteristics? What are these based on?

AXYS realises the many varied, sometimes complex and yet to be fully appreciated, evolution of X & Y sex chromosome variation subtleties. There must be an individualised approach. The current approaches are many and varied and tailored to individual needs. AXYS strongly emphasises that the limitations are primarily around resources to deal with important, confidential, private and sometimes privileged information; coupled with much needed professional, allied and service provider advice and assistance tailored to individual needs.

The one clear, urgent need is for interdisciplinary, centralised care for individuals with X & Y sex chromosome variations. Currently most individuals and families must self-manage a vast range of health and education professionals to support best-practice care, including, but not limited to, general practitioners, endocrinology, physiotherapy, occupational therapy, psychiatry, developmental paediatrics, speech pathology, specialised education intervention and others. For those without the capacity, resources, finances or access to these allied professionals, individuals are not afforded the care they need and deserve.

16. Do any medical guidelines exist that are considered best practice in Australia or internationally, either for the general management of people born with variations in sex characteristics, or for specific variations?

AXYS believe that this question is appropriately addressed by individual professional bodies representing specialties working in this area. E.g. GP's, Paediatricians, Endocrinologists, Psychologists, Physiotherapists, Speech Therapists, Occupational Therapists, Education Department, Employer Groups, Employee Groups etc.

17. Should there be national guidelines to guide medical interventions involving people born with variations in sex characteristics?

AXYS does not see any pressing need to introduce guidelines for individuals with X & Y sex chromosome variants as it is a broad spectrum. Each individual needs to be assessed and clinical decisions and treatment options made as a collaboration between parents and health professionals.

18. If so:

- *what factors should the guidelines take into account?*
- *what should be the legal status of the guidelines?*
- *what should be the process, including consultation, for drafting the guidelines?*
- *what should be the oversight mechanism for decisions made under the guidelines?*

Lack of data

19. What are the current Australian sources of data on:

AXYS is of the view that the following data collection and dissemination is the domain of Government and their connected health services. Notwithstanding that, AXYS is well positioned to assist with data collection in future as to X & Y sex chromosome variations, provided we have adequate funding and human resources.

o the number and nature of medical interventions involving people born with variations in sex characteristics?
o long-term outcomes of medical interventions involving people born with variations in sex characteristics?
o long-term outcomes of people born with variations in sex characteristics not undertaking medical interventions?

20. How adequate are the current Australian sources of data for each of these areas?
AXYS see data collection and processing of relevant statistics as a Government responsibility at this time. We understand that there is currently poor statistical data and encourage appropriate resources be deployed to enhance this information.

21. What barriers exist to nationally consistent data collection?
AXYS see data collection and processing of relevant statistics as a Government responsibility at this time.

Privacy
22. How can medical practices best respect the privacy of people born with variations in sex characteristics?
AXYS requests an urgent review of how karyotype information (including data on X & Y sex chromosome variations) is collected and held. For example, we are aware of breaches, where genetic providers have passed on data to researchers. We understand that there is no consistent national approach.

Further, protection from disclosure of private health related information, in this case regarding X & Y sex chromosome variations is uncertain and could lead to discrimination in the provision of related services, including health insurance.

23. Have you faced any difficulties accessing your medical records?
AXYS need more time to explore this question, especially in the context of the MyGov Health Records platform, data security and the intersection of privacy and health professionals support. We understand that other variations have faced significant issues with respect to access of historical records, but we need to undertake further consultation to understand if this has been an issue for people with X & Y sex chromosome variations. AXYS will provide further and better particulars about this matter if we are invited to make a separate submission.

24. How can access to medical records and histories be improved?
AXYS understands that the Federal Government is working toward secure systems to ensure health records are available for individuals following the further roll out of online health records for individuals and health professionals and allied health concerns. This system is also being built around ensuring a holistic and inclusive approach to medical care is achieved.

Access to support services and peer support
25. How can people born with variations in sex characteristics and their families and carers be more adequately supported?
Despite being the most common X & Y sex chromosome variation, KS/XXY remains unknown to many, including many within the health and allied health services. Early diagnosis is the key but information about available support services is not readily at hand. AXYS is working hard to bridge this gap but significant support is required.
Systemic Australia-wide change is required in the following areas:

- interdisciplinary centrally coordinated medical care for both children and adults and appropriate transition support between systems;
- better access to government benefits and support for those who require it;
- specific educational and learning support for children and adults experiencing learning difficulties or delay;
- discrimination protections in state and territory-based laws, including access to appropriate workplace adjustments;
- sustained funding for peer support groups;
- wide community-based education campaigns to increase knowledge and acceptance; and
- continued close consultation in the development of any legal or policy solutions where people with X & Y sex chromosome variations might be affected.

26. How can psychological and peer support be more integrated into decision making processes?

AXYS, as much as we are able, is a pivotal source of information and much needed conduit for X & Y sex chromosome variations allowing people to connect with, evaluate and discover relevant information that will assist the decision-making process for pre-natal, children, adolescent and adults.

Where we have close relationships and referral mechanisms with particular health care providers, we are able to better support individuals and their families. Mandatory inclusion of information on peer support mechanisms at the time of diagnosis would help to drive consistent information and support to individuals. Commensurate resourcing is also required.

27. What barriers exist to connecting individuals to support services?

Resources, both financial and human are the greatest barrier to connection.

AXYS has grown through an evolution over 6 long years. AXYS has received its first much-needed financial support, via a modest government grant, to alleviate some barriers. Nevertheless, the need for more financial support to further develop our capabilities is vital. Resources would enable us to exponentially expand what we offer and to whom. This would include,

- easier access to information via our website, information management services and Facebook site/s;
- the development, distribution and printing of information resources for health professionals including General Practitioners and teachers, physiotherapists etc;
- peer-to-peer support and counselling;
- support for education institutions and workplaces on inclusion and appropriate resourcing for specialised supports;

28. What barriers exist for individuals in accessing support services?

AXYS is aware of major impediments such as late diagnosis, late intervention, inability to find out about available services and a generally mediocre knowledge among health and allied health professional services about X & Y sex chromosome variations and its associated repercussions.

Crucially the following questions need to be addressed in order to break down access barriers:

- How to identify or alert a parent or GP as to the potential diagnoses of an X & Y sex chromosome variation and especially KS/XXY?
- Once an X & Y sex chromosome variation is identified as a potential diagnosis then who should order the tests for diagnosis?
- Should IVF consultants and gynaecologists be better informed?
- Beyond midwives and GPS, which other health care professionals need an early awareness procedure to look for symptoms?
- Should obstetricians and maternal and child health nurses be better informed?
- How can parents access greater support to confront health issues such as speech delays, hypotonia, sensory and auditory processing challenges, anxiety, social interaction etc?
- Are early educators and teachers aware of the commonality and challenges that are faced throughout an individual's education?

29. How can peer support groups and organisations be adequately resourced and supported?

AXYS is much in need of additional financial and human resources to consolidate, reinforce and provide professional services for its management systems and the resourcing, processing and distribution of information. AXYS needs full time staff, office space, additional technological hardware and software and income streams to allow for travel, attendance at conferences and to further promote solutions for X & Y sex chromosome variations; much of which is provided already by volunteers and generous, but limited, in kind contributions. The AXYS experience is one of steady growth and learning about the issues involved for X & Y sex chromosome variations, available support structures and networking arrangements and opportunities around management of X & Y sex chromosome variations. Refinement of the definition and awareness of 'intersex' and the raft of issues confronting other intersex variations that are not X & Y sex chromosome variations are just as important to AXYS as the X & Y sex chromosome variations issues themselves.

AXYS will continue to further develop as a peak Australia-wide organisation with Chapters outside of Victoria comprising similar, if not the same, duplicate arrangements for X & Y sex chromosome variations. Notwithstanding the AXYS role; AXYS itself does not wish to duplicate services provided by others who look after the best interest of intersex variations that are not X & Y sex chromosome variations. At the same time where synergies exist then a peak body such as AXYS would welcome others to access our services.

As few as 25% of individuals with X & Y sex chromosome variations are actually diagnosed. With advancements in prenatal testing there will be greater numbers of prenatal diagnoses resulting in more studies with much more information available to the general public as well as people working in the health profession. This will accelerate the health care and individual needs of these children which will shape their future.

Conclusions

AXYS will continue to strive for vastly improved awareness, better understanding and greater improvements in support networks to ensure individuals with X & Y sex chromosome variations lead positive and fulfilling lives with decreased health implications. We want to ensure all individuals can gain access to medical care and treatment options in collaboration with their health professional team without any barriers. Placing barriers could have detrimental impacts on a child achieving their best quality of life.

AXYS thanks the AHRC for the opportunity to make a submission on behalf of its members and invites the AHRC to come back and seek more clarity and better particulars about any aspect of our submission.