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**Message from our New President!**

Dear AISSGA members and friends,

It is with much excitement that I write this message to you after recently being elected President at the July AGM. I am really honoured that I was asked to do this role and I hope that I will be able to make the most of the opportunity.

I want to first of all thank Phoebe for all her hard work as the previous President. We are indebted to her for her work in raising the profile of AIS in the media, her revival of our dAISy newsletter and website, as well as getting our email system on to GMail. She has many talents, which we are very fortunate to benefit from, and I am glad she is continuing to be involved with the group as Qld Rep and Secretary. Thank you Phoebe!

I would just like to share with you a personal reflection on my AIS journey up to this point. Like many of us, I have had a difficult road to travel in discovering the truth about myself. I was told by my doctor at 17 that my “ovaries” didn’t work properly, I didn’t have a uterus, and that the ovaries had to be removed. Both my family and myself were left to deal with



Gerbera daisy, red, heart thereof by Martin LaBar, 2008. CC.

this virtually on our own. Not only were we deprived of the truth, but also of support.

I spent the next 10 years in the dark about my body, but finally found out at 27 from a kind endocrinologist that I had CAIS. Once I knew the truth then I could start to heal. My search led me to the AIS Support Group in 1997. At last I could now see that I wasn't a freak and that others knew how I felt. At first I was very stressed at meetings. Telling my story was really emotional and difficult. With time, I integrated my AIS self into the rest of my life. Reading others' stories and attending meetings helped me to feel whole and "normal".

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My hope is that if any of you are reading this and are feeling alone and isolated that you might overcome your fear, and reach out to someone, whether it is a family member, doctor or support group member. We are all here to help you. The group has been a tremendous source of support and help for me, and I hope that you might find whatever it is you need also, whether it is information, support or just someone to talk to.

Thanks to all of you who are doing your bit for the AIS cause. I hope that together we can raise more awareness of this condition and that in the future society will see us as

normal, healthy people who have something special and unique to contribute.

Best wishes,  
Sandra



### **Message from the Outgoing President**

Hello all, I hope this newsletter finds you all in terrific health and forging ahead in your chosen fields.

The past six months have certainly been quite eventful, with a number of victories for the group and for people with AIS in general.

Particularly, the legal precedent set in Queensland earlier this year, whereby the parents of a 13-year-old girl with the 46XY genotype and inguinal testes (presumably AIS) successfully petitioned the Family Court in Queensland for permission for their daughter to have surgery to remove her gonads. All parties were in agreement with this outcome – parents, doctors and the patient – and thus the girl's feelings were taken in to account. News outlets reported that she said, 'Sometimes I get angry, even though I know why and how it happened.' Here, the truth was not withheld, nor was the person with the condition left out of the

decision-making process. To quote our Treasurer, Mr Tony Briffa, the judgement is cause for 'much satisfaction' as 'it's the result of the AISSGA's work over the last 10 years.

To see children with intersex conditions finally treated with respect and dignity, and having their human rights upheld so that a court has to decide about irreversible, non-therapeutic surgeries is extremely satisfying.'

Good news also that Caster Semenya, the champion runner from South Africa, has been allowed to compete as a woman. From all reports, it seems that Ms Semenya also has AIS – so welcome to the club, darling! Of course, it can be that you only discover that you have AIS later in life, often when concern about the lack of periods prompts a visit to the doctor. Perhaps it is little wonder that, as elite, teenage athlete, Ms Semenya might not have worried unduly if menarche had not yet commenced. It is good to know that the governing bodies that have the power to say whether someone can or cannot compete have acknowledged that having AIS would not give any competitive advantage against other women, and to restrict anyone with AIS from competition solely for this reason would be terribly unfair.

Finally, I'm pleased to report that the leadership of the AISSG Australia is now in the very capable hands of Sandra after the

election of a new committee at the conference in Melbourne this month. I'll still be around in the as Secretary of the group, and I'll also be bringing you our biannual newsletter, too.

All the best,  
Phoebe



### **AISSGA Conference July 2010 – Melbourne: A Report by Sandra**

It was with much excitement that I traveled to Melbourne recently for the AISSGA Annual Meeting. It was a chilly, rainy day but 8 people came along for a great day of friendship and heartfelt sharing.

We began the day with the AGM and several people were elected for the new committee for this year. It was great to see newer people volunteering for positions on the committee, so I am excited that we have a great group of people to lead the group this year.

The committee for this year is:

President:	Sandra
Vice-President (Media)	Bonnie
Vice-President (Medical)	Andie
Secretary:	Phoebe
Treasurer:	Tony

## dAISy

The Newsletter of the Androgen Insensitivity Syndrome (AIS) Support Group of Australia  
July 2010 Edition

Victoria/Tas Reps:	Sam and Jocelyn
SA/NT Rep:	Carol
NSW/ACT Rep:	Sandra
Queensland Rep:	Phoebe
Parent's Reps	Annette and Ruth
Men's Rep:	Tony
Public Officer:	Andie

Congratulations to everyone elected! It's great to see all the positions being filled. If anyone in WA is interested in being a Rep. please let me know!

The rest of the morning was taken up with animated conversation on all things AIS, including diagnosis, disclosure, parent's issues, and public awareness of AIS and intersex generally. After a grand lunch at a local café we shared our personal stories in greater depth, with each person being offered encouragement, support and empathy for their particular AIS journey.

It was great to have a range of ages and backgrounds at the meeting, and fascinating to hear how different people have coped with their challenges in their own unique way. It was very clear to me once again that these meetings provide a really valuable outlet for people to be heard and to give voice to what is often difficult for them to express with others. Our shared experiences give us a common bond and foundation on which we

can re-build a more whole and "normal" way of seeing ourselves, which I know for me, is such a relief!

Thank you very much to Tony who took time out of his busy schedule to organize the meeting, and to Andie for her kind hospitality in giving me a place to stay. I thoroughly enjoyed my visit and will particularly remember my night tour of Altona and Williamstown in Tony's car, with Andie and Tony pointing out places of interest such as the Town Hall, Council Chambers, lights of Melbourne and several churches!

### **Musings on the AISSG Conference, July 2010 by Bonnie**

After meeting at the nearby café for an informal catch up prior to the conference, we ran through the business side of things – the election of the new committee – first up. Once that bit of fun was done with, we really got talking! An assortment of people with different intersex conditions was present (mostly AIS women), plus parents of children with intersex. I felt there was a lot of love in the room, as everyone had a great deal of compassion for one another in the safe space we had created. It was definitely a cathartic experience to release all these ideas and issues that had been rambling through our heads... occasionally working oneself into a lather! There was actually a lot of laughter,

too. Everyone shared his/her particular story, which was terrific.

There were some points raised with regards whether medical practices and procedures have really changed over the years, and whether doctors and surgeons would direct new parents with a infant with AIS or an intersex condition to a support group or peer counselling service, such as ours. An interesting point in case was the birth of a child a few years ago now with both cleft palate and AIS. Within a week, a cleft palate support group was recommended and a member of that group had contacted the parents, but, at the same time, the same consideration was not given to the AIS aspect, which was ignored, overlooked or just considered 'too hard' or unworthy of similar attention. Support groups are not, as some doctors may believe, a group of malcontents with an axe to grind, but a bunch of people who genuinely want to help others to avoid hurt and pain, and where's the harm in that?

On reflection afterwards, I began to also consider something my partner had said at dinner the night before the conference – that perhaps the partners of men and women with AIS need support as much as men and women with AIS do! Surely, his suggestion bares more consideration. Many who have an intersex condition and have experienced

surgical intervention know that physical and emotional intimacy can be extremely challenging, as such interference strips away libido and physical sensation, and often times our partners certainly have their work cut out for them working with and helping their partners through these difficulties.

I look forward to catching up with my new and old friends again soon!

### **Caster Semenya can run with the Women; It's Official**



South African running phenomenon Caster Semenya, who blew past her competition at the Berlin world championships last August, has been cleared to compete as a woman - nearly a year after controversial gender tests put her career on hold.

Semenya, 19, has not run competitively for 11 months. During that time she underwent a battery of psychological, gynecological and endocrine tests.

"I am thrilled to enter the global athletics arena once again and look forward to competing with all the disputes behind me," Semenya said in a statement released by her lawyers.

Semenya first grabbed the world's attention as an 18-year-old, when she ran 800-meters in 1:56.72 at the African Junior Championships in Mauritius. She clocked the fastest time of the year.

It was then that speculation about her gender began, according to an article in Time.

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Soon after, Semenya clocked 1:55.45 in Berlin - again, the fastest time of the year. Now people really began to talk.

Semenya has an extremely muscular build, and a few of the women she left in the dust shared scathing assessments of her physical makeup with the press. Elisa Piccione of Italy, who came in sixth place, said that Semenya is "not a woman. She's a man." Mariya Savinova of Russia, who finished in fifth place said to just "look at her," according to the article.

The mudslinging angered politicians and activists, and Semenya became a South African household name. The country's ruling

ANC party made a public statement Tuesday, calling the decision "a vindication of the ANC, her family, our government and all progressive forces who stood behind her during her time of need," said national spokesman Jackson Mthembu.

"It has always been our long-held view that Caster is a woman and she should have been allowed a long time ago to participate in athletics as a woman," added Mthembu. "We don't believe that any aspersion should have been cast on her gender as woman."

Yet IAAF, the governing body for track and field, insisted that no insult had ever been intended, and that they did not think that Semenya was deliberately concealing her gender. Nick Davies, IAAF spokesperson called it a "medical issue," ESPN reported.

The medical issue Davies was referring to is called androgen insensitivity syndrome. People with this rare condition appear to be female, and they are, for all practical purposes. Yet in actuality, they have one X chromosome and one Y chromosome in each cell, the pattern normally found in males, according to WebMD.

Some women don't find out that they have the Y chromosome until they try to conceive and

end up getting the news from a doctor in a fertility clinic.

The IAFF was heavily criticized for asking Semenya to take the gender tests, some say for good reason. It was widely reported that the International Olympic Committee stopped testing in 1996, when eight female athletes tested positive for the Y hormone. Experts say that testing serves little purpose, as having androgen insensitivity syndrome does not necessarily give an athlete an advantage.

Semenya's father told Reuter's television that he felt that his daughter was treated badly. The lightning-fast young runner, however, seems to have her sites set squarely on the next race.



### **Voices of the 'other' that need to be heard**

By Goya Bennett, July 10, 2010

HOBSONS Bay deputy mayor Tony Briffa is thought to be the only person to describe his sex as "other" in the Victorian Local Governance Association survey of the numbers of males and females in local government.

The Altona ward councillor was born with an intersex condition, which caused him to be incorrectly assigned as female as a child.

Asked why he described himself as "other" when he fought so hard to be male, Cr Briffa sets the record straight.

"I fought to be me. It wasn't so much about me becoming Tony, a male; it was more about finding out who I was meant to be had doctors not interfered.

"I wanted to understand my true nature and be the person I should have been before doctors castrated and modified me through surgeries and hormone treatment.

"I was born ambiguous in terms of my sex and doctors weren't sure what my sex was at birth. As a child, then even as a teenager, I questioned whether I was a boy or a girl.

"I was always a feminist; I always believed in affirmative action and equal opportunity.

"It's about being true to myself and that whole journey about discovering who I would have been had it not been for the medical intervention, and just being the person that nature made me."

As vice-president of the Androgen Insensitivity Syndrome Support Group

Australia, Cr Briffa has long lobbied for childhood gender assignments not to be surgically reinforced.

"Thanks to the work that we've done and the support group that I run at the Royal Children's Hospital, they've done a 30-year follow-up study of children with these conditions treated at the Royal Children's Hospital," he said.

"If a person's got my condition, for example, if they were raised as a female it would be unlikely they would then do irreversible surgeries like castration as a child.

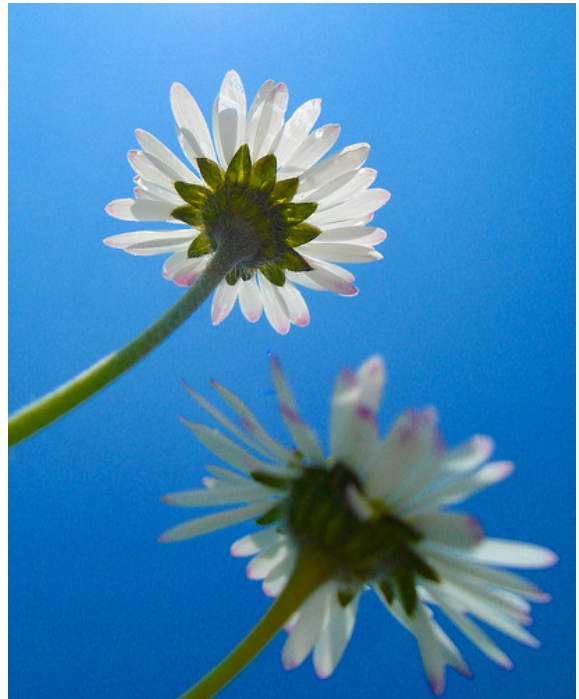
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"The records at the Royal Children's speak for themselves - 8 per cent of children in that 30-year study period were raised in the wrong sex, which is a horrible statistic given that they reinforce that sex surgically."

Cr Briffa said the VLGA survey, marking the Year of Women in Local Government, specifically asked for councillors' sex.

"I just thought, to be true to myself and to have the record accurately reflect the situation, let's put 'other'."

<http://www.theage.com.au/national/voices-of-the-other-that-need-to-be-heard-20100709-1046h.html>



Pushing up the Daisies by James Michael Hill, 2006. CC.

### **The Child Inside**

By Tryla Brown

I grew up with guilt, but not knowing why;  
A naive child living a lie.

They didn't tell me, but everyone knew,  
My mind was so frazzled, I hadn't a clue.  
I wanted to run, I was filled with self-hate,  
There was nowhere to go, there was no escape.

A child of GOD, a sister of fate,  
I wanted to grow up, oh I couldn't wait.  
I became a woman at the age of fourteen,  
A whole lot to learn, the world seemed so mean.

I thought I knew what I would become,  
It ended up I was still on the run.  
I'm much older now, I can see clear,  
I feel it inside, the truth is so near.



I know what I am, I can't hide the pain,  
But the child inside should feel no shame.  
Come home to me child, I'll take care of you.  
Come home to me child, I've been there too.  
Come home to me child, let me feel through  
you.  
I promise I will come through, I promise I will  
come through.



### **Girl with Rare Condition gets OK to have Testicles Removed**

Tuesday, April 27, 2010: An Australian family court has given the OK for a 14-year-old girl — born with a rare genotype — to have surgery to remove two testicles, the Courier Mail reported.

The parents of the girl, known in the case as "Sally", had to apply to the court for permission for the surgery after they discovered their daughter had androgen insensitivity syndrome, which means she is genetically male, with one X chromosome and one Y chromosome.

People with this condition have the external sex characteristics of females, but do not have a uterus, the National Institutes of Health said on its Web site. Instead,

individuals have male internal sex organs (testes) that are undescended.

The court heard "Sally" had been a healthy girl until she turned 11-years-old and discovered two lumps, one of which was in her abdomen.

"It hurts sometimes, knowing I have this condition," the teenager told the court.

"Sometimes I blame myself, because I feel like I am not normal. Sometimes I get angry, even though I know why and how it happened."

The court also approved other procedures to confirm her gender.

<http://www.foxnews.com/story/0,2933,591569,00.html>



Lots of daisies... by Martin LaBar, 2008. CC.

## The third sex: The truth about gender ambiguity



By Colette Bernhardt, Saturday, 20 March 2010, The Independent UK:

Neither wholly male nor entirely female, there are more than 30,000 'intersexed' individuals living in Britain today. Here, they talk about their lives.

The Greek scholar Diodorus Siculus wrote of the mythical double-sexed Hermaphroditus: "Some say [he] is a god and ... has a body which is beautiful and delicate like that of a woman, but has the masculine quality and vigour of a man. But there are some who declare that such creatures of two sexes are monstrosities, and coming rarely into the world as they do, have the quality of presaging the future, sometimes for evil and sometimes for good."

Throughout history, those born with both male and female physical features have been beset by society's interpretations of them as freaks (The Lancet branded a British hermaphrodite "a disgusting spectacle" in 1834), prophets (the part-male/part-female character Tiresias in Oedipus Rex and Antigone was a clairvoyant), or both. What they've seldom been allowed to do is just get on with their lives.

Even in modern times, tales of "intersexed individuals", as they are now known, are often tinged with melancholy. In 1998, a Sunday newspaper reported on the "desperately lonely existence" of Linda Roberts, who was "spat at and stoned" in a village in Snowdonia for her androgynous appearance.

Last year, the South African track athlete Caster Semenya, then just 18, was subjected to chromosome testing and a humiliating media furore when it was discovered she had no ovaries or uterus, as well as unusually high testosterone levels. The International Association of Athletic Federations remains undecided on whether she is "really a woman", and has suspended her from competing in major sporting events, including last weekend's World Indoor Championships in Qatar, until a verdict is reached.

Semenya has no desire to be a pioneer for the intersex community; she simply wants to concentrate on her running. But there are countless others who are calling for increased recognition of their status, as well as for a moratorium on the practice of "sex reassignment surgery" of intersexed infants.

Intersexuality presents itself in numerous different forms, occurring both in individuals with a standard karyotype (the chromosomal make-up of the body's cells) of 46, XX (female) or 46, XY (male), as well as in those with more unusual karyotype combinations such as 47, XXY or 47, XYY. In one of the most common conditions, Congenital Adrenal Hyperplasia (CAH), overactive adrenal glands can cause a female foetus with XX chromosomes to "over-virilise", developing anything from a large clitoris to a fully formed phallus, while the rarer Androgen Insensitivity Syndrome (AIS) can cause a male foetus with XY chromosomes to "under-virilise", developing only a partially formed penis and testes.

Until recently, the overwhelming response among doctors was to surgically "correct" a baby's ambiguous genitalia on the grounds that he/she would, in later life, be stigmatised by these unconventional appendages. From the 1960s, it became common practice to trim down an enlarged clitoris, and to fashion a

malformed penis into a vagina. The line among surgeons was allegedly: "It's easier to dig a hole than build a pole."

Intersexed babies with XY chromosomes have therefore frequently been "reassigned" as female, with parents advised to raise them as girls, and oestrogen pills administered to induce female puberty. This is largely due to the hugely influential 1960s "optimal gender policy" of psychologist John Money, and his famous assertion that nurture could override nature.

The prevalence of corrective surgery is in part responsible for our general ignorance about intersexuality, which is far more widespread than most of us realise; the number of live births displaying "genital dimorphism" is estimated at approximately one in every 2,000. That means there could be as many as 30,000 intersexed people currently living in Britain, a figure that becomes even greater when taking into account all those who only discover their condition at puberty, or when they try to have children. As the renowned professor of neurology and intersex expert Dr Milton Diamond puts it: "Nature loves variety. Unfortunately, society hates it."

"Our constant pursuit of perfection has left many children infertile, with their gender identity stolen," argues Dr Jay Hayes-Light, a

specialist in child mental health and the director of the campaigning organisation the UK Intersex Association (UKIA), which formed in 2000. "There's this fear that if we have women with large clitorises and men with small penises, it'll be the end of civilisation as we know it. In fact, the individuals who end up most damaged are those who are surgically altered, without their permission, to suit someone else's agenda."

Why? Because as well as the physical downsides – "reducing a clitoris carries the risk of permanent loss of sensitivity, while reducing a phallus and removing the testes destroys the child's fertility" – Hayes-Light insists that gender cannot simply be reconfigured through surgery and hormone treatment.

His own studies of individuals who were reassigned as infants, some of whom have eventually transitioned back to their original gender, and some of whom have committed or attempted suicide, have convinced him that "sex is between the legs while gender is between the ears. Most people are 'hard-wired' to a gender identity, whether this is male, female or something in between."

Hayes-Light, now 48, was himself born with an extremely rare intersex condition, 5-Alpha-Reductase Deficiency (5-ARD), in which the

chromosomes are XY, but the body is unable to convert testosterone to dihydrotestosterone, which is necessary for the formation of full male genitalia. "This was the early 1960s; I was a lab rat, a medical curiosity. What saved me from being poked and prodded – and operated on – was the fact that my mother was a doctor."

Advised that he would be "a very happy little girl", Hayes-Light's mother nevertheless declined surgery for her son. But she agreed to try raising him as female, and did so until he was 10. "I was then asked if I wanted to take oestrogen tablets, to push my body through a female puberty," he remembers. "I refused." Despite this troubled and confusing start to life, Hayes-Light considers himself one of the lucky ones: "I shudder to think what I'd be like now if I'd been forced to transition."

Adele Addams wasn't given such a choice. Born in the late 1970s, she has Klinefelter's Syndrome, so is XXY; chromosomally neither male nor female, in other words. Her parents were encouraged to assign her as male, and surgery was performed immediately. In her mind, this was "the wrong decision". After a difficult childhood and adolescence – "I eventually went into care, as my family couldn't cope with what was supposed to be a boy but looked and sounded more like a girl"

– she began living as a gay man. Yet this didn't feel right either, and three years ago, after "a lot of fighting", she was granted NHS surgery to make her body female. Now 31, she finally feels that "it's all come good", and is passionate about empowering others to define their own gender.

Last year she set up Project Silverfish, a support service for intersex and transgender people, whom she describes as "the most marginalised minority community in London". Named after her friend, the transgender DJ and music producer Alex Silverfish, who committed suicide in 2008 following years of harassment, the project offers its users practical advice and advocacy, and has just launched its first therapeutic support sessions. It also delivers training to charities and social services.

Addams, who last December won a Home Office Community Engagement Award for her work, explains that while transgenderism and intersexuality are not the same (the former is not a medical condition), "it's not uncommon for trans people to discover later in life that they're intersex".

This was what happened to Alexandra Tovey, aged 37. From an early age, she felt she was "born with a female brain, but a male body", yet her intersex condition – Partial Androgen

Insensitivity Syndrome (PAIS) – was only diagnosed last year. She'd already been living as a woman for several years, and the news, she says, "made things make more sense".

Now enjoying "being the 16-year-old girl I wasn't allowed to be when I really was 16", and awaiting a full sex-change operation, Tovey, a singer-songwriter, expresses herself through music. She's released 18 albums since 1992, and many of her lyrics reflect her experiences as both a trans and intersexed person.

The actress and playwright Sarah Leaver is also using art "to expose what lives between the lines". Her play, *Memoirs of a Hermaphrodite*, currently running at London's Oval House Theatre, tells the true story of Herculine Barbin, a 19th-century intersexed Parisian whose musings were published by the French philosopher-sociologist Michel Foucault in 1980. Barbin began life as a girl, but faced misunderstanding and contempt when she fell in love with another woman, and was later discovered to have both male and female genitalia. Forced to become a man, she descended into depression and poverty, and died in tragic circumstances.

Thankfully, Leaver's own tale is far happier. "I've always felt in between the genders," she

says. "As a kid I wore boy's pants, played football, and ran around with my top off." Having supportive parents meant this was rarely an issue, and it was only in her teens and twenties that she started to question her identity: "I didn't feel I was in the wrong body, but I knew there was something that made me different."

Six years ago, after watching a documentary on intersexuality, she asked her GP to check her medical history. It transpired that an operation she'd had as a toddler in 1977 to remove a "hernia" had in fact been to remove a male gonad – but neither she nor her parents had ever been informed. "It was like finding the missing piece in a jigsaw," she recalls. "Part of me was relieved, and part of me was really angry. Why had the doctors hidden this from me?"

Despite her anger, Leaver, who is now 34, regards herself as pretty fortunate. Her condition – she hasn't yet sought a specific diagnosis – is at the mild end of the intersex spectrum, as she has XX chromosomes, ovaries and periods, and has experienced few health problems.

Through the play, she's turned her frustration into "a force for change", and hopes others will do the same: "More and more people are speaking out about being intersexed, and the

time feels right for celebrating and embracing our differences; not hiding them away."

"We need to be able to live as ourselves," agrees Addams, "which means making room for more than two genders." Tovey echoes this sentiment: "I've always felt very female, but I know others who identify with both sides, who feel 'in between'. They need acceptance, too."

Hayes-Light, though respectful of the wishes of those who want to be considered "gender-neutral", stresses that they are a minority: "Although some physically intersex people choose not to identify as either male or female, most do identify as either one or the other."

When it comes to operating, all four of these case histories concur that modifying infants' genitals for anything other than a medical necessity is wrong. And it appears that even the medical experts have started to come round to this view. In 2001, the British Association of Paediatric Surgeons (BAPS) recommended that babies with ambiguous genitals should not be given corrective surgery, and should be left to decide their own gender eventually.

Things seem to be moving on in other fields too. "In the 10 years since UKIA was

founded, we've started to see far fewer cases involving secrecy and cover-up [such as that experienced by Leaver]," says Hayes-Light. "We've also succeeded in getting intersex included in the 2004 Gender Recognition Act. And we've seen major changes in social attitudes, with more balanced media reporting, too."

Yet there's still a long way to go. "Society is becoming more open-minded, but most people still haven't heard of the word 'intersex', and gender variance remains the last taboo in our culture," maintains Addams. Despite the recommendation of BAPS, surgery on intersexed babies still goes on, both in Britain and many other countries.

Leaver looks forward to a time "when intersexed people are no longer viewed with disdain or pity, and the world doesn't cling so much to 'male' and 'female'".

No one can predict the future, but it does seem as though our culture is becoming, albeit slowly, less rigid about gender roles, and more accepting of unconventionality in general. It can only be hoped that it's a sign that we're beginning to respect, and even learn from, the individuals we would once have relegated to circus sideshows, and later to operating theatres.

As the writer and psychotherapist Amy Bloom sums up: "Not monsters, nor marvels, nor battering rams for gender theory, people born intersexed have given the rest of the world an opportunity to think more about the odd significance we give to gender, about the elusive nature of truth, about the understandable, sometimes dangerous human yearning for simplicity – and we might, in return offer them medical care only when they need it, and a little common sense and civilised embrace when they don't."

Or, in the simple, succinct words of Dr Milton Diamond: "Let's see if we can change society, not nature."

*'Memoirs of a Hermaphrodite' showed at the Oval House Theatre, London SE11, until 27 March, and at the Marlborough Theatre, Brighton, from 23-25 May, 2010.*

### **University of Guelph Researchers First to Identify a Family of Intersex Horses**



Mar 29th, 2010: Koko the horse appeared to be a typical mare, but when she became overly aggressive and tried to mount other mares, her owner became suspicious. After a barrage of tests at the Ontario Veterinary College, researchers discovered that Koko was, in fact, a male horse that appeared to be female.

The scientists tested Koko's relatives and found that her sister Sequoia and her cousin Pandora were also intersex.

Researchers Allan King, Tracey Chenier and Daniel Villagomez are the first to identify a family of horses with a rare genetic abnormality called pseudo-hermaphroditism, which causes genetically male horses to appear female on the outside.

This discovery has caught the attention of media across the country and has been featured in Macleans.ca, CBC.ca, Yahoo News, AOL News, CTV.ca and several other radio, print and online media.

Initially, they thought Koko had an ovarian tumour, which can cause mares to exhibit stallion-like behaviour because of heightened levels of testosterone. But when they examined the animal's reproductive system, the researchers found Koko had internal testes.

"It was a very exciting discovery," said Chenier. "To be expecting a tumour but discover an entirely different internal system than anticipated sparked a lot of questions."

The research team then conducted a karyotype analysis, which is a detailed study of an animal's chromosomes, and results showed that Koko had a male genotype (XY).

"Because these horses are from the same family line, the condition is likely a genetically heritable one," said Chenier. "A very similar condition can occur in humans with what is known as androgen insensitivity syndrome. It's linked recessively to the X chromosome and passed through the female line."

The researchers suspect Koko's mother has a mutation on her X chromosome that causes male offspring to appear female. Female offspring show no abnormalities but can, in turn, pass it on to their male offspring.

Currently, the team of researchers is looking for the genetic mutation that would cause androgen receptors – those that respond to the male hormones that are responsible for secondary sex characteristics – to be underactive.



Once the mutation is located, a better tool could be developed to test animals suspected of having the condition.

“We are studying the DNA, molecule by molecule, to try and determine what the mutation is and how we can more easily diagnose this disorder in horses,” said King.

After realizing Koko had internal testes, researchers performed surgery to remove them in the hope that she would become less aggressive.

“Before the surgery she was so aggressive that it was too dangerous to ride her,” said the owner Sam Campbell. “But she is completely different now. She is an incredibly lovely horse.”

Since then both Sequoia and Pandora have undergone the same procedure at the Ontario Veterinary College.

Although sex development disorders are rare, two racing standard bred horses in the United States were recently diagnosed with a similar intersex condition. The two animals turned out to be related to one another, but not to the horses found in Ontario, said King.

The two cases in the U.S. were discovered after repeated drug tests found high testosterone levels.

“It was initially assumed the high levels of testosterone were due to steroid use, but when examined more closely it was determined the horses had an intersex condition.”

Finding related horses with this condition raises questions about how common this genetic abnormality is and whether it is fair for horses with this condition to race, he added.

“Nobody knows who they should race against – fillies or colts.”

*Ed: Maybe the researchers at U of G should take a closer look at the Caster Semenya case for clarity on this last point!!*

<http://www.canadaviews.ca/2010/03/29/u-of-g-researchers-first-to-identify-a-family-of-intersex-horses/>



**Novel (60%) and Recurrent (40%)  
Androgen Receptor Gene Mutations in a  
Series of 59 Patients with a 46,XY  
Disorder of Sex Development**

Journal of Clinical Endocrinology &  
Metabolism , doi:10.1210/jc.2009-2146  
The Journal of Clinical Endocrinology &  
Metabolism Vol. 95, No. 4 1876-1888  
Copyright © 2010 by The Endocrine Society  
L. Audi, et. al

Background: Androgen receptor (AR) gene mutations are the most frequent cause of 46,XY disorders of sex development (DSD) and are associated with a variety of phenotypes, ranging from phenotypic women [complete androgen insensitivity syndrome (CAIS)] to milder degrees of undervirilization (partial form or PAIS) or men with only infertility (mild form or MAIS).

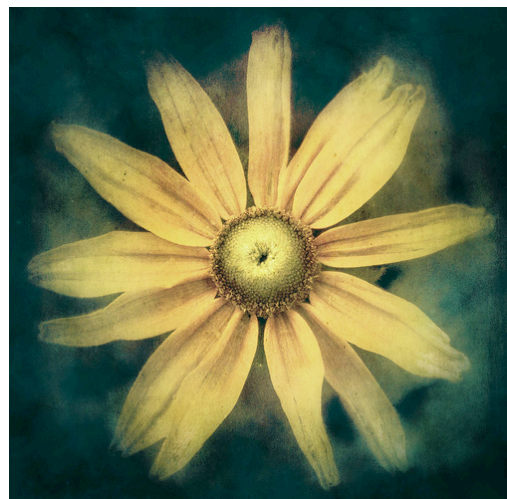
Objective: The aim of the study was to characterize the contribution of the AR gene to the molecular cause of 46,XY DSD in a series of Spanish patients.

Setting: We studied a series of 133 index patients with 46,XY DSD in whom gonads were differentiated as testes, with phenotypes including varying degrees of undervirilization, and in whom the AR gene was the first candidate for a molecular analysis.

Methods: The AR gene was sequenced (exons 1 to 8 with intronic flanking regions) in all patients and in family members of 61% of AR-mutated gene patients.

Results: AR gene mutations were found in 59 individuals (44.4% of index patients), of whom 46 (78%) were CAIS and 13 (22%) PAIS. Fifty-seven different mutations were found: 21.0% located in exon 1, 15.8% in exons 2 and 3, 57.9% in exons 4–8, and 5.3% intronic. Twenty-three mutations (40.4%) had been previously described and 34 (59.6%) were novel.

Conclusions: AR gene mutation is the most frequent cause of 46,XY DSD, with a clearly higher frequency in the complete phenotype. Mutations spread along the whole coding sequence, including exon 1. This series shows that 60% of mutations detected during the period 2002–2009 were novel.



\*Daisy\*.... by pareerica, 2009. CC.

## **Concomitant Mutations in the P450 Oxidoreductase and Androgen Receptor Genes Presenting with 46,XY Disordered Sex Development and Androgenization at Adrenarche**

Jan Idkowiak, et al.

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Context: Undervirilization in males, i.e. 46,XY disordered sex development (46,XY DSD), is commonly caused by either lack of androgen

action due to mutant androgen receptor (AR) or deficient androgen synthesis, e.g. due to mutations in 17 $\alpha$ -hydroxylase (CYP17A1). Like all other microsomal cytochrome P450 (CYP) enzymes, CYP17A1 requires electron transfer from P450 oxidoreductase (POR).

Objective: The objective of the study was to analyze the clinical and biochemical phenotype in a 46,XY individual carrying concomitant POR and AR mutations and to dissect their impact on phenotypic expression.

Methods: We characterized the clinical and biochemical phenotype, genetic identification, and functional analysis of POR missense mutation by yeast microsomal coexpression assays for CYP17A1, CYP21A2 and CYP19A1 activities.

Results: The patient presented neonatally with 46,XY DSD and was diagnosed as partial androgen insensitivity syndrome carrying a disease causing AR mutation (p.Q798E). She was raised as a girl and gonadectomized at the age of 4 yr. At 9 yr progressive clitoral enlargement prompted reassessment. Urinary steroid analysis was indicative of POR deficiency, but surprisingly androgen production was normal. Genetic analysis identified compound heterozygous

POR mutations (p.601fsX12/p.Y607C). In vitro analysis confirmed p.Y607C as a pathogenic mutation with differential inhibition of steroidogenic CYP enzymes.

Conclusion: Both mutant AR and POR are likely to contribute to the neonatal presentation with 46,XY DSD. Virilization at the time of adrenarche appears to suggest an age-dependent, diminishing disruptive effect of both mutant proteins. This case further highlights the importance to assess both gonadal and adrenal function in patients with 46,XY DSD.

*Ed's note: Adrenarche is the onset of androgen dependent signs of puberty in boys or girls, including pubic hair, axillary hair, acne, and adult body odor.*

## Recommended Reading



Bodies in Doubt: An American History of Intersex  
Elizabeth Reis (Author)

240 pages

Publisher: The Johns Hopkins University Press (May 8, 2009)

ISBN-10: 0801891558

What does it mean to be human? To be human is, in part, to be physically sexed and culturally gendered. Yet not all bodies are clearly male or female. *Bodies in Doubt* traces the changing definitions, perceptions, and medical management of intersex (atypical sex development) in America from the colonial period to the present day.

From the beginning, intersex bodies have been marked as "other," as monstrous, sinister, threatening, inferior, and unfortunate. Some nineteenth-century doctors viewed their intersex patients with disrespect and suspicion. Later, doctors showed more empathy for their patients' plights and tried to make correct decisions regarding their care. Yet definitions of "correct" in matters of intersex were entangled with shifting ideas and tensions about what was natural and normal, indeed about what constituted personhood or humanity.

Reis has examined hundreds of cases of "hermaphroditism" and intersex found in medical and popular literature and argues that medical practice cannot be understood outside of the broader cultural context in

which it is embedded. As the history of responses to intersex bodies has shown, doctors are influenced by social concerns about marriage and heterosexuality. *Bodies in Doubt* considers how Americans have interpreted and handled ambiguous bodies, how the criteria and the authority for judging bodies changed, how both the binary gender ideal and the anxiety over uncertainty persisted, and how the process for defining the very norms of sex and gender evolved.

*Bodies in Doubt* breaks new ground in examining the historical roots of modern attitudes about intersex in the United States and will interest scholars and researchers in disability studies, social history, gender studies, and the history of medicine.



Daisy Droplet by Christian Travers, 2008. CC.

## Become a Member

It's not too late to join or renew your membership for this financial year! The Androgen Insensitivity Syndrome (AIS) Support Group Australia Inc. (A0041398U) is

a peer support, information and advocacy group for people affected by AIS and/or related intersex conditions, and their families.

We support members (both in Australia and overseas) that have any grade of Androgen Insensitivity Syndrome, and support any issues relevant to living with AIS. These issues include infertility, disclosure, hormone therapy, gender identity, surgical intervention of children with intersex conditions, sexual intimacy, etc.

We also provide support to those with related intersex conditions like Partial and Complete Gonadal Dysgenesis, MRKH (also known as Vaginal Agenesis), 5a-Reductase Deficiency, 3b-Hydroxysteroid Dehydrogenase Deficiency, 17-Ketosteroid Reductase Deficiency and 17b-Hydroxysteroid Deficiency. (The previous name for AIS was Testicular Feminisation Syndrome).

We acknowledge that people with intersex conditions (including AIS) range from female to male and anywhere in between.

The AISSG Australia believes in a holistic model of health as per the World Health Organisation definition of 'health':

***"Health is a state of complete physical, mental and social well-being and not merely the absence of disease and infirmity".***

## dAISy

The Newsletter of the Androgen Insensitivity Syndrome (AIS) Support Group of Australia  
July 2010 Edition

There are many benefits to becoming a member of the AISSG Australia, including the dAISy newsletter, regular conferences and meet-ups, and up-to-date information and news relevant to our members. Of course, there is also the wonderful benefit of supporting and meeting others who have so much in common and much to share! We are also always looking for people who want to become more involved as representatives and/or committee members.

Since our AGM in July 2010 we are:

President:	Sandra
Vice-President (Media)	Bonnie
Vice-President (Medical)	Andie
Secretary:	Phoebe
Treasurer:	Tony
Victoria/Tas Reps:	Sam and Jocelyn
SA/NT Rep:	Carol
NSW/ACT Rep:	Sandra
Queensland Rep:	Phoebe
Parent's Reps	Annette and Ruth
Men's Rep:	Tony
Public Officer:	Andie

We'd love to have representatives in other states and territories. We also have representatives for parents of children with

AIS and men with AIS... and the word is out that partners of people with AIS need support too!

### **We have a winner!!**

Congratulations Bonnie, who won a beautiful Phoebe lamp by Australian designer Peter Harding at the AISSG Australia conference in Melbourne this month. Bonnie's name was drawn out of a random lottery of people who had taken up a new or renewed membership in the last period.



[www.phoebelamps.com](http://www.phoebelamps.com)

### **The next dAISy**

Next issue is January 2011. The deadline for submissions is 30 December 2010. To submit articles, art, jokes, information, poems, or whatever you would like to share please email [aissgaustralia@gmail.com](mailto:aissgaustralia@gmail.com)

We hope to hear from you again soon!

**Membership Application (2010-2011)**

I wish to apply for membership of the Androgen Insensitivity Support Group Australia Inc. I enclose my annual membership fee which ends on the 1<sup>st</sup> of August 2011. To become a member simply print out the following page and forward it to: PO Box 3239 South Brisbane Q 4101 or email your details to: aissgaustralia@gmail.com

Name/Organisation:.....

Address: .....

State: ..... Postcode: ..... Country: .....

Telephone Number(s): .....

Email address: .....

Age: .....(optional) If organisation name of contact person: .....

Signature: ..... Date: .....

Membership type: (Please tick one)

**Individual Membership:**

**Organisational Membership:**

1 year \$20

1 year \$40

3 years \$50

3 years \$100

Please indicate your membership category:

Person with AIS, Gonadal Dysgenesis or similar condition (Please specify)  
.....

Family / Partner / Friend of someone with AIS or similar condition  
.....

Medical Professional (please state interests in AIS and similar conditions)  
.....

Organisation (please state interests in AIS or similar conditions)  
.....

**Would you like to make a donation to the AIS Support Group Australia?**

(Donations of \$2 or more are tax deductible)

\$10

\$20

\$50

\$100

other \$.....

Please make all cheques payable to the **AIS Support Group Australia**. Direct deposit available on request. **Thank you!**