

Disorders of Sex Development: When to Tell the Patient

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LWPES/PAS Mini Course

Baltimore, MD

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WHEN to TELL the PATIENT?

ALWAYS



HOW?

SLOWLY

Outline:



- Why is “disclosure” such an issue?
- Disclosure vs. explanation
- Critical role of the parents
- General approach to the discussions
- Value of patient support groups





Why is "Disclosure" Such an Issue?

- It's hard to give bad news
- Concern about emotional reactions of patient
- Concern about potential to harm gender identity or self esteem
- Concern about effect on patient/parent relationship
- Physician discomfort
- The subject of disclosure is **SEX**



Historical Attitudes Toward Disclosure

- *Morris J 1953*: "It goes without saying that it would be unwise to inform the patient of the **true state of affairs** . . . it seems only necessary to state that childbearing is impossible."

Why not Tell the Truth? What is the Stigma?



- Y chromosome/testes = "MALE"
- Female body with male "parts"
- Carryover from Victorian attitudes: **sexual variations** are taboo ("don't ask, don't tell")



What is the Stigma?

- **Ambiguity**: is this person "really" a male or a female, or something in between?



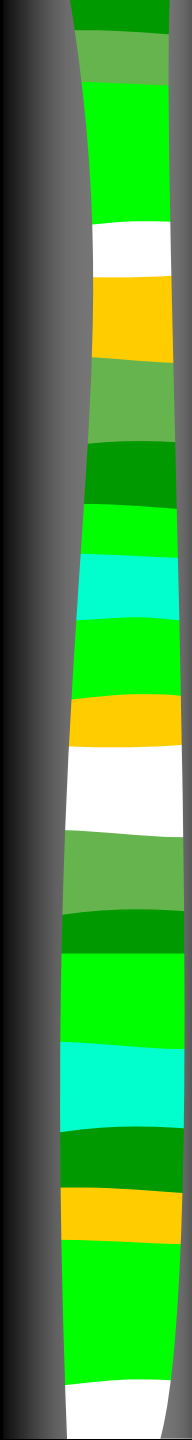
Consequences of Withholding the Truth

- Shroud of secrecy
- Feelings of exclusion, isolation
- Fears regarding the gravity of what is **NOT** being revealed
- Sense of "freakishness" or "repugnance"
- Feelings of shame



Disclosure

- Morris J 1953: "It goes without saying that it would be unwise to inform the patient of the true state of affairs . . . it seems only necessary to state that childbearing is impossible."
- **Conn J et al 2005:** "The argument that disclosure would lead to unacceptable harm is no longer valid. Unquestionably, the greatest harm would result if the patient found out her diagnosis by chance in an unsupported environment." *BMJ* 331:628



Disclos(ur)e, Definitions

- Make known (especially something secret or concealed); e.g. "*the disclosed purpose of their wicked plan*"
- The act or process of revealing or uncovering
- Something uncovered; a revelation

Disclosure vs. Explanation

- Explain the facts rather than revealing a secret



General Approach

- Get to know the family
- Understand their fears, issues and concerns



Parents' Issues

- Loss of "normalcy"
- Grief
- Confusion
- Fear and anxiety
- Depression
- "Genetic guilt"
- Shame
- Overprotection

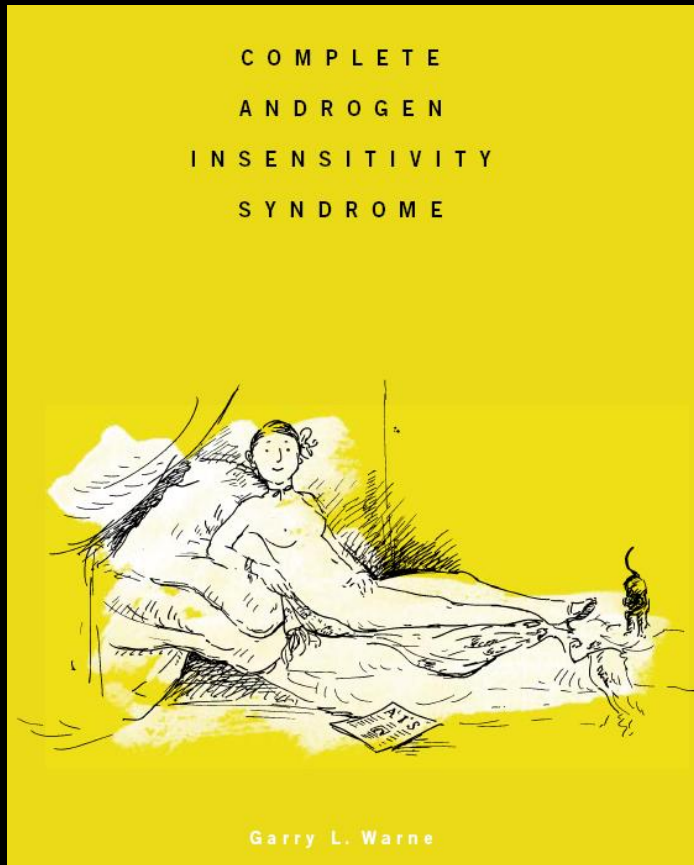




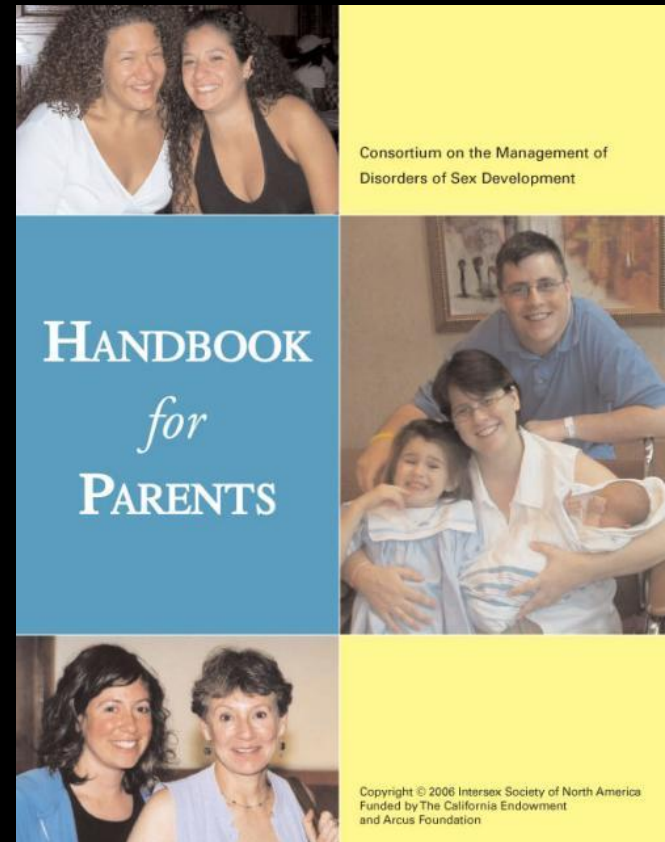
General Approach

- Get to know the family
- Understand their issues/concerns
- Correct misinformation/confusion
- Provide family with information and words they need
- Provide resources (e.g. Garry Warne's booklet; ISNA handbooks; Toronto Sick Kids' Hospital website)
- Support group information
- Contact with other families

Resources

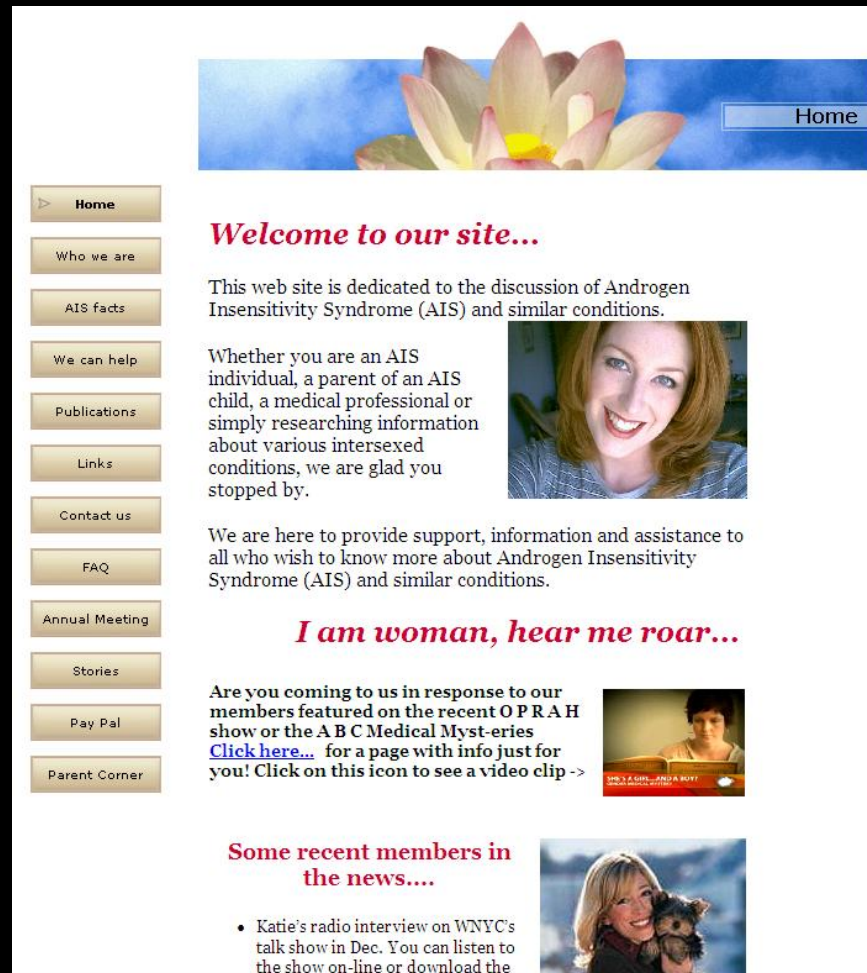


<http://www.rch.org.au/publications/CAIS.pdf>



<http://www.dsdguidelines.org/>

Resources: AIS Support Group




The screenshot shows the homepage of the AIS Support Group website. At the top, there is a banner image of a pink lotus flower with a blue sky background. A "Home" button is visible in the top right corner of the banner. Below the banner is a vertical navigation menu on the left side with buttons for: Home, Who we are, AIS facts, We can help, Publications, Links, Contact us, FAQ, Annual Meeting, Stories, Pay Pal, and Parent Corner. The main content area features a "Welcome to our site..." section with a paragraph about the website's purpose and a photo of a smiling woman. Below this is a section titled "I am woman, hear me roar..." with text about members featured on Oprah and ABC Medical Mysteries, accompanied by a small video player icon. The final section is "Some recent members in the news...." with a bullet point about a radio interview and a photo of a woman holding a dog.

[Home](#)

Welcome to our site...

This web site is dedicated to the discussion of Androgen Insensitivity Syndrome (AIS) and similar conditions.


Whether you are an AIS individual, a parent of an AIS child, a medical professional or simply researching information about various intersexed conditions, we are glad you stopped by.



We are here to provide support, information and assistance to all who wish to know more about Androgen Insensitivity Syndrome (AIS) and similar conditions.


I am woman, hear me roar...

Are you coming to us in response to our members featured on the recent O P R A H show or the A B C Medical Myst-eries [Click here...](#) for a page with info just for you! Click on this icon to see a video clip ->



Some recent members in the news....

- Katie's radio interview on WNYC's talk show in Dec. You can listen to the show on-line or download the





<http://www.aissgusa.org>

Resources: AIS-DSD-Parents' Group

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New User? Sign Up

at&t Get home phone, high speed Internet, and wireless or TV under **\$100**/mo.   [Learn More](#)

AIS-DSD-Parents Search for other groups... Search

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- Messages
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- Promote
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Info Settings

Group Information

Members: 106
Category: Care Giving
Founded: Dec 20, 2004
Language: English


Stay up to speed on the latest Groups news and updates, visit the [Groups blog](#) today!

Home [Join This Group!](#)

Activity within 7 days: **66** New Messages - [New Questions](#)


Description

A support group for parents/family members of children with Androgen Insensitivity Syndrome (CAIS, PAIS) and other Disorders of S*x Development including gonadal dysgenesis, ambiguous genitalia and others. (Note: use of asterisk (*) above necessary as the original word was disallowed as a descriptor; please refer to DSD Manual link on left for proper terminology).



Message History

	Jan	Feb	Mar	Apr	May	Jun	Jul	Aug	Sep	Oct	Nov	Dec
2009	23	91	51	94								
2008	78	249	176	220	73	116	145	237	55	29	40	35
2007	32	82	102	127	77	44	46	103	165	278	97	138
2006	117	175	91	94	38	112	91	125	51	37	37	29
2005	93	53	72	41	190	129	94	111	53	222	51	74

 [Yahoo! Answers](#) What's this?

<http://groups.yahoo.com/group/AIS-DSD-Parents>

Resources

AboutKidsHealth
Trusted answers from The Hospital for Sick Children April 27, 2009

2007 WSJ GOLD WINNER

SickKids

HOME RESOURCE CENTRES HEALTH A-Z NEWS HOW THE BODY WORKS JUST FOR KIDS

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Female Genital Anatomy

Male Genital Anatomy

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Chromosomal Sex

Gonadal Sex

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Sex Development

// Sexual Differentiation

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Sexual Differentiation

Sexual differentiation (becoming male or female) is a multi-step process.

There are key 'branch points' during embryonic and fetal life that shape a baby's sexual chemistry and anatomy.

Chromosomal sex comes first, at the moment of **conception**.

The formation of **gonads** (ovaries or testes), **internal ducts**, and **external genitals**, and the changes that occur at **puberty**, all follow in a 'cascade' of cause and effect.

Please choose a topic from the menu at left to continue.

E-mail Print Comment Share

Role of the Parents



- Parents should be the primary providers of information to the child
- Empower the parents to help their child
- Slowly build information to the child over time
- Age-appropriate details and words
- Other parents can provide guidance



Strategies

- Focus on simply explaining the facts, rather than “disclosing” a secret
- Demystify the chromosomes, gonads and genitalia
- Emphasize the similarities (rather than differences) between male and female development
- Explain concept of different pathways to similar endpoints
 - Some children take the direct route, others take a detour



Sharing the Facts with Parents

- Your daughter has a condition called Androgen Insensitivity Syndrome (AIS)
- AIS is caused by alteration of a gene involved in development of sex - the androgen receptor gene or AR
- Because of the alteration in this gene your daughter has some differences in her sex development compared with average girls

Sharing the Facts

- Most girls/women have 2 X chromosomes . . .
- But some do not
 - ~1 in 2,000 has only one X (Turner)
 - ~1 in 20,000 has 1 X and 1 Y (AIS, other)
- Most boys/men have 1 X and 1 Y chromosome
- But some do not . . .
 - ~1 in 500 has 2 Xs and a Y (Klinefelter)
 - ~1 in 100,000 has 2 Xs and no Y (XX male)

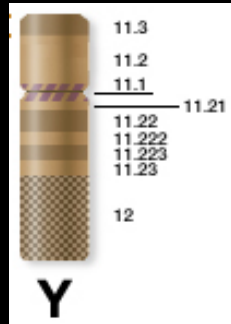




Sharing the Facts

- Because she has AIS, your daughter has an X chromosome and a Y chromosome
- AIS occurs in about 1 in 40,000 people
- There are about 7,000 girls/women in the US with AIS
- *What does it mean for a girl to have a Y chromosome?*

Demystifying the Y Chromosome



- ❑ Chromosomes are just packages to store genes (DNA)
- ❑ Y chromosome is basically a "shrunk" X chromosome
- ❑ Y carries the same critical genes as the X



Clarifying the Genetics of Sex Determination

- It's not just about the X or the Y chromosome
- At least 20 genes are involved in development of sex (probably 100s)
- 3 known "sex genes" on X
- 1 known "sex gene" on Y
- At least 1 "sex gene" on each of chromosomes 2, 3, 4, 5, 8, 9, 10, 11, 12, 17, 19 etc.



Clarifying Sex Development

- **ALL** babies, no matter what sex they are going to be, begin life with the same internal and external structures
- "Gonads" are initially neither testes nor ovaries
- Genitalia are undeveloped, neither male nor female
- Whether the child develops as a boy or a girl depends on the effects of a whole lot of genes, not just having an X or Y chromosome

Sharing the Facts

- Most girls/women have ovaries
- But some do not
 - Turner, Swyer syndrome: "streak" ovaries
- Girls and women with AIS have testes instead of ovaries
- Testes and ovaries are structurally and functionally similar





Sharing the Facts

- Because her body doesn't respond to male hormones, she developed physically as a girl
- However, her testes blocked the development of the uterus
- Because the cells can't use testosterone, the body converts it to estrogen
- Estrogen causes female body shape to develop at puberty

Sharing the Facts

- *How will this affect her?*

- She will be generally healthy
- Her puberty may be late
- She will not menstruate
- She will not be able to get pregnant
- She will have reduced pubic hair and armpit hair
- Her vagina may be smaller than average (needs to be checked when she's older), but this can be helped



Guidance to Parents for Explaining to the Child



- Toddler years
 - introduce the concept of differences
"everyone's body is different in many ways"
 - provide easy examples
- Childhood (various ages)
 - explain that not everyone has children the "ordinary" way
 - some families adopt children
 - explain sexual/reproductive organs and differences between individuals



Guidance to Parents for Explaining to the Child

- Late childhood (especially if the child has started learning biology in school)
 - some girls don't have periods for various reasons; she will not have periods
 - some women can't have children "the typical way"
 - like many other women, she will not be able to become pregnant
 - there are other ways to have a family



Explaining to the Child/Teen

- Late childhood/early teens
 - like all girls/women with AIS, she has a Y chromosome
 - Y chromosome caused testes to develop
 - testes blocked uterus from developing
 - testes produce "boy hormones" and "girl hormones" (just as ovaries do)
 - her body can't use the "boy hormones" but can only use the "girl hormones", so she developed as a girl

Explaining to the Child/Teen

- Late childhood/early teens
 - Avoid stereotypes or references to movie stars (not all women with AIS look like models)
 - Don't minimize the importance of the patient's losses
 - Don't be afraid of tears
 - Check for understanding at each step, by asking patient to tell you what she knows



Roles of Parent/Patient Support Groups



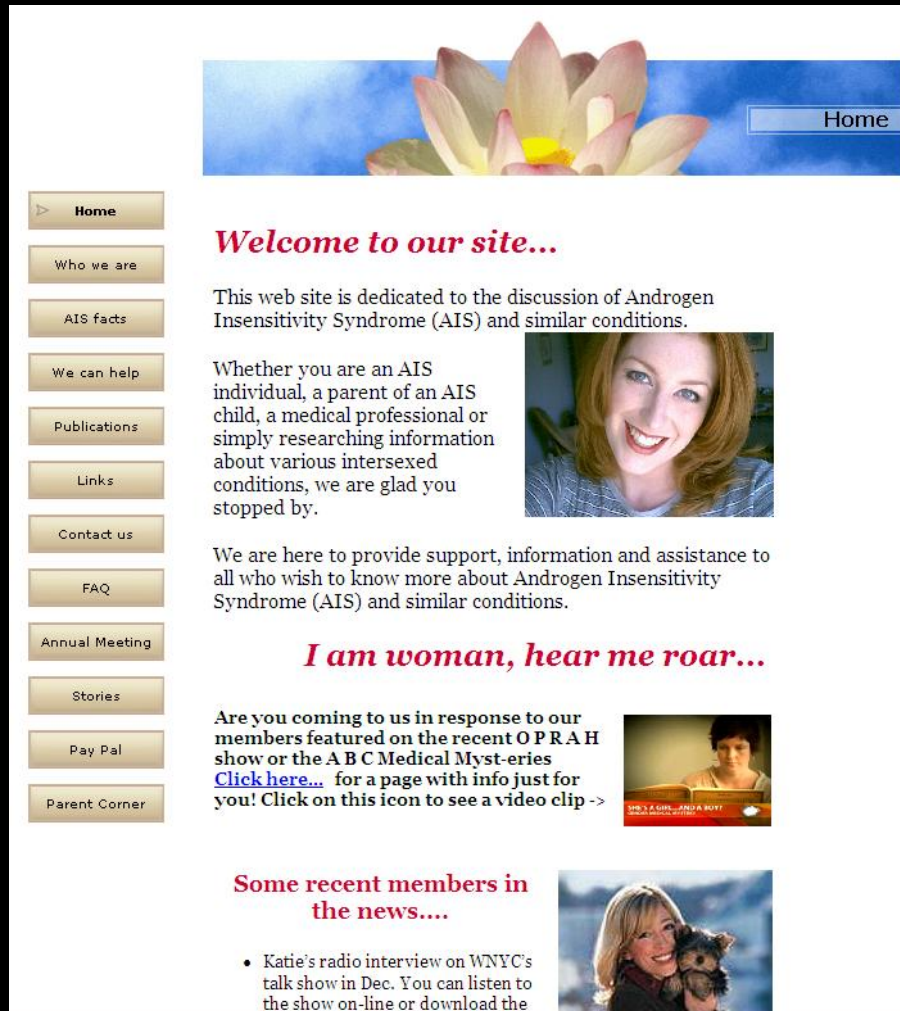
- Other parents/patients know **EXACTLY** how they feel → bonding
- Affected girls/women share a "sisterhood"
- Safe place to discuss their issues and concerns
- Provide strategies for coping with emotional and practical issues
- Share successes and failures
- Provide role models



Parents' Support Group

- Parents "offer emotional support but not medical advice"
- From one parent to the mother of a newly-diagnosed patient:
- "You know how Oprah has her '*this I know for sure*' insights? Well, what I know for sure is that being able to explain AIS will get easier over the years because you'll know more about it, you'll be more comfortable with it, you'll know from experience what things make people wince when you say them, and your growing daughter will be evidence that this isn't as awful as it sounds. . . .What we live with it isn't as awful as it first sounded."

Resources



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