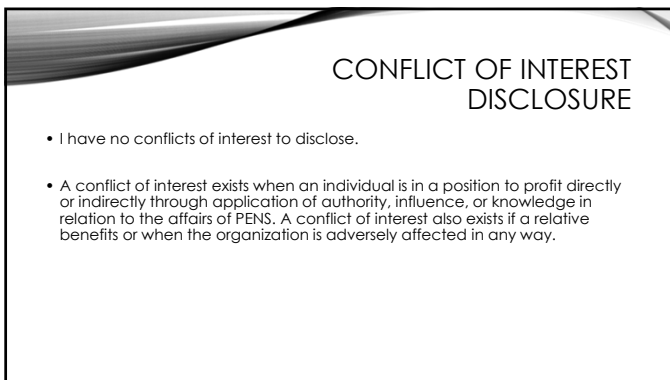


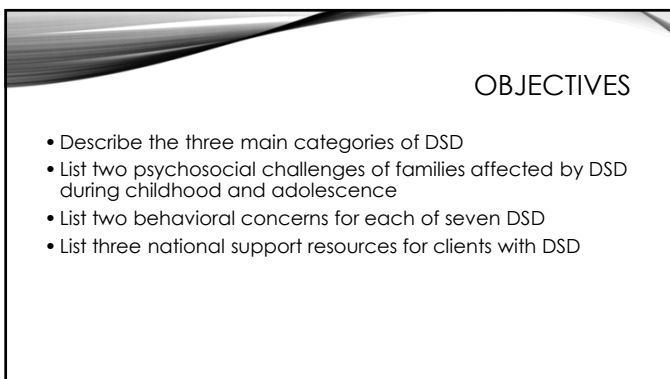
WORKING WITH CLIENTS WITH DISORDERS OF SEX DEVELOPMENT

Traci Schaeffer RN, BSN



CONFLICT OF INTEREST DISCLOSURE

- I have no conflicts of interest to disclose.
- A conflict of interest exists when an individual is in a position to profit directly or indirectly through application of authority, influence, or knowledge in relation to the affairs of PENS. A conflict of interest also exists if a relative benefits or when the organization is adversely affected in any way.



OBJECTIVES

- Describe the three main categories of DSD
- List two psychosocial challenges of families affected by DSD during childhood and adolescence
- List two behavioral concerns for each of seven DSD
- List three national support resources for clients with DSD

OVERVIEW

- Sex vs gender
- Specific needs, management, and resources by diagnosis
- Process of disclosure

The Genderbread Person v2.0 by its pronounced *neTRosexual*

Gender is one of those things everyone thinks they understand, but most people don't. Like fiction. Gender isn't binary. It's not either/or. It means even if it's both/and. A bit of this, a dash of that. This little title guide is meant to be an appetizer for understanding. It's okay if you're hungry for more.

Identity
 Nongendered ↔ Woman-ness
 ↔ Man-ness

Gender Expression
 Agender ↔ Masculine
 ↔ Feminine

Biological Sex
 Asex ↔ Female-ness
 ↔ Male-ness

Attracted to
 Nobody ↔ (Men/Males/Masculinity)
 ↔ (Women/Women/Femininity)

♀ self-refers possible, gnd and abstr conds

read more: bit.ly/1gmg9uq

WHAT ARE DSD?

- Disorders of Sex Development
- congenital conditions in which development of
 - chromosomal,
 - gonadal, or
 - anatomical sex is atypical

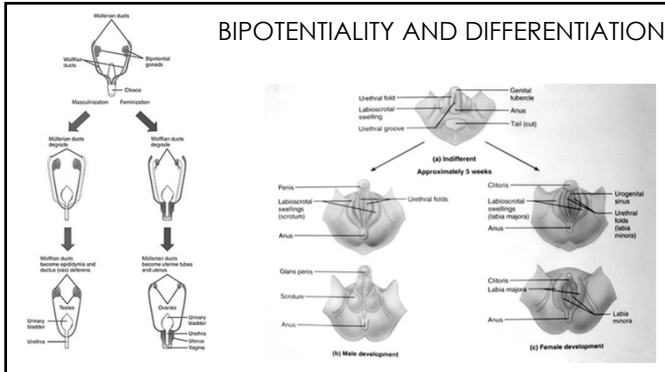
SO WHAT IS GENDER DYSPHORIA?

- Dysphoria – feeling of dissatisfaction, anxiety, and restlessness
- Gender dysphoria – dissatisfaction/anxiety about the fact that your body doesn't reflect your gender
 - Leads to severe distress, anxiety, and depression
- AKA "gender identity disorder"
 - Mismatch between body and internal sense of gender is not the problem
 - Problem is the stress, anxiety, and depression that go along with the mismatch

QUESTION 1

- Disorders of Sex Development are defined as conditions affecting:
 - A – Gender Identity
 - B – Gender Expression
 - C – Biological Sex
 - D – Sexual Attraction

BIPOTENTIALITY AND DIFFERENTIATION



THREE CATEGORIES

- Sex chromosome DSD
 - Turner Syndrome: 45,X
 - Klinefelter Syndrome: 47,XXY
 - Mixed Gonadal Dysgenesis: 46,XY/45,X

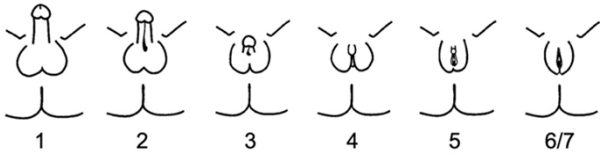
THREE CATEGORIES

- XX DSD
 - Congenital Adrenal Hyperplasia
 - Vaginal Agenesis/MRKH
 - True Hermaphroditism

THREE CATEGORIES

- XY DSD
 - Complete Androgen Insensitivity Syndrome
 - Partial Androgen Insensitivity Syndrome
 - 5- α -Reductase Deficiency
 - Persistent Müllerian Duct Syndrome

AMBIGUOUS GENITALIA- QUIGLEY SCALE



QUESTION 2

- All embryos form with the parts to develop into either a male or a female.
 - A – True
 - B – False

PARENTAL ISSUES COMMON TO MANY DSD

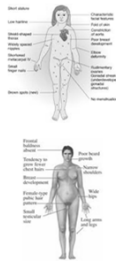
- Parents grieve the loss of the "perfect child"
- Fertility challenges
 - "Will I ever be a grandparent?"
- What to say to friends and family in cases of ambiguous genitalia
- Feelings of shame and guilt, especially in mothers
 - Significant in genetic conditions where parent is a carrier

LET'S GET SPECIFIC!

- Questions before we get into specific diagnoses?

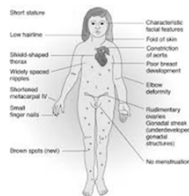
THREE CATEGORIES

- Sex chromosome DSD
 - Turner Syndrome: 45,X
 - Klinefelter Syndrome: 47,XXY
 - Mixed Gonadal Dysgenesis: 46,XY/45,X



TURNER SYNDROME (TS)

- Incidence: 1 in 2500 females (high rate of spontaneous miscarriage)
- Medical symptoms
 - Short stature – treat with GH
 - Premature ovarian failure – treat with estrogen
 - Infertility – donor eggs or surrogacy
 - Heart problems (aortic coarctation, hypoplastic left heart) – if present, contraindication for pregnancy
 - Hypothyroidism – treat with thyroid hormone
 - Webbed neck – can do plastic surgery, most don't



TURNER SYNDROME (TS)

- Behavioral symptoms
 - Depression
 - Anxiety
 - Nonverbal learning disability
 - Lack of executive function
 - Problems with social skills
 - Delayed sexual debut
 - Low self-esteem



TURNER SYNDROME

- Things to know
 - Often drive late
 - Trouble with math
 - Sequencing challenges
 - Usually live independently
 - OT can be very helpful
 - Almost all on GH injections
 - Need IEP throughout schooling
 - 45,X karyotype, female gender identity
 - Normal genitalia



SUPPORT

- Turner Syndrome Society of the US
 - www.turnersyndrome.org
- Local support group
 - www.succeed-clinic.org



KLINFELTER SYNDROME

- Incidence: 1 in 500 males
- Medical symptoms
 - Low muscle tone
 - Thin tooth enamel
 - Delayed puberty (treat with testosterone)
 - Gynecomastia (breast development in males)
 - Low sperm count/infertility
 - Undescended testes



KLINFELTER SYNDROME

- Behavioral symptoms
 - Learning disabilities
 - Anxiety
 - Depression
 - Problems with social skills
 - Developmental disorder
 - Speech delay
 - ADHD
- Things to know
 - Female-typical body habitus
 - Tend to be tall
 - 47,XXY karyotype, male gender identity
 - Normal genitalia, UDT
 - Poor coordination
 - Many need IEP
 - Almost all live independently

SUPPORT

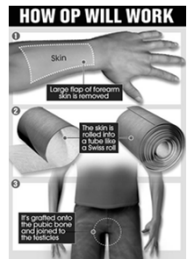
- Knowledge, Support, and Action
 - www.genetic.org
 - (888) 999-9428
- Living with Klinefelter Syndrome (47,XXY) Trisomy X (47,XXX) and 47,XYY: A Guide for Families and Individuals Affected by Extra X and Y Chromosomes

MIXED GONADAL DYSGENESIS

- Incidence: rare, may be assigned male or female at birth
- Medical symptoms
 - Ambiguous genitalia
 - ↑ risk of gonadal tumors
 - Infertility
 - Need hormone replacement at puberty
- Behavioral symptoms
 - ↑ risk of gender dysphoria
 - Depression
 - Anxiety
 - Feelings of inadequacy related to infertility

MIXED GONADAL DYSGENESIS

- Things to know
 - Often have genital surgery
 - 1 in 4 change gender
 - 45,X/46,XY karyotype




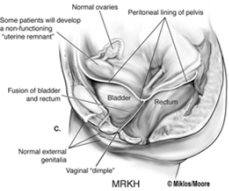
QUESTION 3

- You can always tell a person's gender identity from their karyotype.
 - A – True
 - B – False

THREE CATEGORIES

- XX DSD
 - Congenital Adrenal Hyperplasia
 - Vaginal Agenesis/MRKH
 - True Hermaphroditism





© Mike Moore

CONGENITAL ADRENAL HYPERPLASIA (CAH)

- Incidence: 1 in 15,000, occurs equally in males and females
- Medical symptoms
 - Females have ambiguous genitalia
 - Can't make stress hormone cortisol
 - At risk of adrenal crisis, circulatory collapse, and death
 - Treat with hormone replacement three times a day
 - Tested for on newborn screening
 - Males at high risk of benign testicular tumors

CONGENITAL ADRENAL HYPERPLASIA

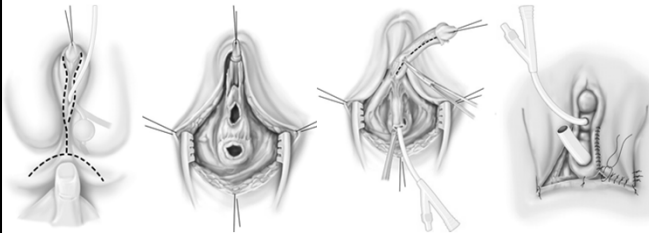
<ul style="list-style-type: none"> • Behavioral symptoms (female) <ul style="list-style-type: none"> • Masculinized behaviors • Higher risk of gender dysphoria • Anxiety • Depression • Aggression • Disruptive behaviors • ↑ suicidal ideation 	<ul style="list-style-type: none"> • Behavioral symptoms (male) <ul style="list-style-type: none"> • Anxiety • ADHD • Disruptive behaviors
---	---

CONGENITAL ADRENAL HYPERPLASIA

- Things to know
 - Increase in homosexuality (female)
 - Need school plan for emergency (IEP or 504)



FEMINIZING GENITOPLASTY



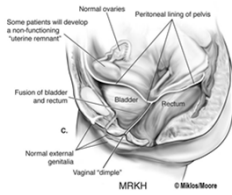
SUPPORT

- CARES Foundation
 - www.caresfoundation.org
- MAGIC Foundation
 - www.magicfoundation.org
- Adrenal Insufficiency United
 - www.aiunited.org
- Local support group
 - www.succeed-clinic.org



VAGINAL AGENESIS/MRKH

- Incidence: 1 in 5000 females
- Medical symptoms
 - Absent vagina, cervix, and uterus – treated with surgery or dilation
 - ↑ risk of kidney abnormalities (missing kidney, horseshoe kidney)
 - ↑ risk of skeletal abnormalities



VAGINAL AGENESIS/MRKH

- Behavioral symptoms
 - Depression
 - Feelings of inadequacy related to infertility
- Things to know
 - 46,XX karyotype
 - Female gender identity
 - Normal female genitalia

SUPPORT

- MRKH Support
 - www.mrkh.org
- Local support group
 - www.succeed-clinic.org

QUESTION 4

- Doctors can always tell a baby's sex based on the genitalia.
 - A – True
 - B – False

THREE CATEGORIES

- XY DSD
 - Complete Androgen Insensitivity Syndrome
 - Partial Androgen Insensitivity Syndrome
 - 5- α -Reductase Deficiency
 - Persistent Müllerian Duct Syndrome



COMPLETE ANDROGEN INSENSITIVITY SYNDROME (CAIS)

- Incidence: 1 in 20,000
- Medical symptoms
 - High levels of testosterone
 - Absence of fallopian tubes, uterus, cervix
 - Infertility
 - Absence of underarm hair, sparse pubic hair
 - ↑ risk of gonadal tumors
 - Treat with estrogen for bone health



COMPLETE AIS

- Behavioral symptoms
 - Depression
 - Feelings of inadequacy related to infertility
- Things to know
 - Tends to run in families
 - 46,XY karyotype, female gender identity
 - Vaginal dilation often required

PARTIAL ANDROGEN INSENSITIVITY SYNDROME (PAIS)

- Incidence: rare, may be assigned male or female at birth
- Medical symptoms
 - Ambiguous genitalia
 - ↑ risk of gonadal tumors
 - Infertility
 - Need hormone replacement at puberty

PARTIAL ANDROGEN INSENSITIVITY SYNDROME (PAIS)

- Behavioral symptoms
 - ↑ risk of gender dysphoria
 - Depression
 - Anxiety
 - Feelings of inadequacy related to infertility
- Things to know
 - Often have genital surgery
 - 1 in 4 change gender
 - 46,XY karyotype, male or female gender identity

SUPPORT

- Androgen Insensitivity Support Group
 - www.aidsd.org
- Local support group
 - www.succeed-clinic.org



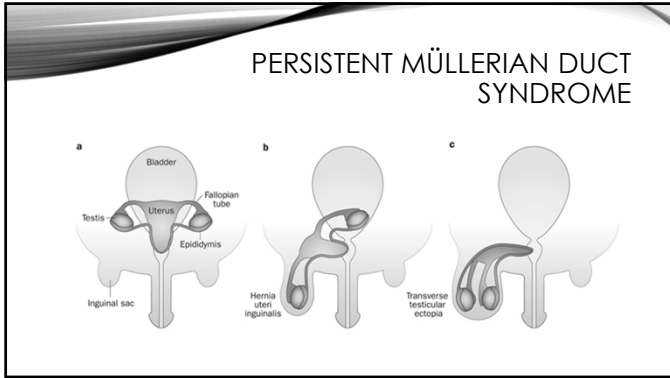
The logo for the AIS-DSD Support Group features a stylized five-petaled flower with a central circle containing a smaller flower. To the right of the flower, the text "AIS-DSD Support Group" is written in a sans-serif font.

QUESTION 5

- Some people can be happy and well adjusted, regardless of which sex they were assigned. Other people feel very strongly about their gender identity, and need to alter their sex if it doesn't fit.
 - A – True
 - B – False

PERSISTENT MÜLLERIAN DUCT SYNDROME

- Incidence: rare, affects males only
- Medical Symptoms
 - Presence of uterus and fallopian tubes (Müllerian ducts) in normally developed XY male
 - Treated with surgical removal
 - Usually associated with undescended testes
 - Testes may be in unusual location
 - Genetic condition – autosomal recessive – but doesn't occur in females



PERSISTENT MÜLLERIAN DUCT SYNDROME

- Behavioral Symptoms
 - Depression – accepting dx, infertility
- Things to know
 - XY karyotype, male gender identity, male genitalia
 - Slight risk of cancer of the abnormal tissue
 - High rate of infertility

QUESTION 6

- What should you call a person who has a uterus and a penis?
 - A – transvestite
 - B – person with a DSD
 - C – hermaphrodite
 - D – their name

THINGS TO REMEMBER

- DSD does not equal ambiguous genitalia
- Patients have often had multiple genital exams (trauma)
- Don't assume anything
 - They may not want to know more info about diagnosis
 - You don't need to know about anatomy
 - Issues may not be related to DSD

DISCLOSURE IS A PROCESS

- This is an example of the process in a patient with PAIS, diagnosed early
- Age 2-5
 - You are healthy and growing
 - You come to the doctor to make sure you keep growing well
- Age 6-8
 - You are healthy and growing
 - Boys and girls look different in their private parts
 - In some boys and girls, their private parts didn't finish growing when they were in their mom. Those kids, like you, had surgery on their private parts to help them finish growing.

DISCLOSURE IS A PROCESS

- Age 9-11
 - You are healthy and growing
 - Puberty starts around this time for most kids. Sometimes, kids like you need help to start puberty because your gonads don't make the hormones they should. (Explain how the brain tells the gonads to make hormones, which cause the body to start puberty, a 2-3 year process.)
 - You will take estrogen in a patch or a pill. (We explain what happens to a body going through puberty and ask if this is ok with the child. This is a good time to check for gender dysphoria.)

DISCLOSURE IS A PROCESS

- Age 12-14
 - You don't make the hormones that you need to go through puberty on your own because you were born with testes.
 - Your body made testosterone, but can't respond to it. You are a girl, because you don't have the receptors to process testosterone.
 - You don't have a uterus and you won't have periods. This is because the hormones produced by your testes suppressed the development of your uterus.
 - You have XY chromosomes, as do many other women. Women who possess a Y chromosome act like women, look like woman and are women.

THANK YOU!

- Please feel free to call our team with questions or concerns
 - Traci-Schaeffer@ouhsc.edu
 - 405-271-6764
 - www.succeed-clinic.org
- Questions?



REFERENCES/RESOURCES

- Auchus, R., Witchel, S., Leighl, K., Asenberg, J., Aziz, R., Bocheho, T., ... , Berenbaum, S. (2010). Guidelines for the development of comprehensive care centers for congenital adrenal hyperplasia: Guidance from the CARES Foundation Initiative. *International Journal of Pediatric Endocrinology*.
- Fedele, D. A., Kirk, K., Wolfe-Christensen, C., Phillips, T. M., Mozer, T., Mullins, L. L., ... , Wisniewski, A. B. (2010). Primary Caregivers of Children Affected by Disorders of Sex Development: Mental Health and Caregiver Characteristics in the Context of Genital Ambiguity and Genitoplasty. *International Journal of Pediatric Endocrinology*.
- Hong, D. S., Dunkin, B., & Reiss, A. L. (2011). Psychosocial functioning and social cognitive processing in girls with Turner syndrome. *Journal of Developmental and Behavioral Pediatrics*, 32-36.
- Huges, L., Houk, C., Ahmed, S., Lee, P., & Group, L. C. (2006). Consensus statement on management of intersex disorders. *Journal of Pediatric Urology*, 148-162.
- Johannsen, T. H., Ripa, C. P., Mortensen, E. L., & Main, K. M. (2006). Quality of life in 70 women with disorders of sex development. *European Journal of Endocrinology*, 877-883.
- Meyer-Bahlburg, H. F. (2014). Psychoendocrinology of Congenital Adrenal Hyperplasia. In M. New, O. LeKarev, A. Pansa, B. O'Malley, & G. D. Hammer, *Genetic Steroid Disorders* (pp. 285-300). Amsterdam: Elsevier.
- Sandobas, D. E., Gardner, M., & Cohen-Kettenis, P. T. (2012). Psychosocial Aspects of the Treatment of Patients with Disorders of Sex Development. *Seminars in Reproductive Medicine*, 443-452.
- Schaeffer, T., Tryggvason, J., Mallappa, A., Hanna, A., Krishnan, S., Chemousek, S., ... , Wisniewski, A. (2010). An Evidence-Based Model of Multidisciplinary Care for Patients and Families Affected by Classical Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency. *International Journal of Pediatric Endocrinology*.
