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

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

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

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

• 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) 999-4428

• Living with Klinefelter Syndrome [47,XXY], Trisomy X [47,XXX], and 47,XY: 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

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

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

- Behavioral symptoms (female)
  - Masculinized behaviors
  - Higher risk of gender dysphoria
  - Anxiety
  - Depression
  - Aggression
  - Disruptive behaviors
  - ↑ suicidal ideation

- Behavioral symptoms (male)
  - 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

• 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.aisdsd.org
• Local support group
  • www.succeed-clinic.org

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


