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Dsd with internal testes and ovaries

Disorders of sexual development (DSDs) are a range of conditions in which a person has characteristics of both sexes. These characteristics can happen due to chromosomal, gonadal (ovaries or testes) or genital differences — and they can appear at birth, during puberty or later in adulthood. Examples include people born with:Male (XY) chromosomes but genitals that appear female (vulva).Female (XX) chromosomes but genitals that appear male (penis or enlarged clitoris).Both ovarian and testicular tissue. This can cause the genitals to appear male, female or a mixture of both.Typical sex organs but an abnormal chromosome arrangement. This can disrupt growth and development during puberty.Healthcare providers used to call DSDs “intersex” conditions. Intersex refers to people who have chromosomes, genitals or reproductive organs that don’t fit into the male/female sex binary. Some people with DSDs prefer the term “differentiation” rather than “disorder.” Having a DSD doesn’t mean there’s something “wrong” with you. It just means you developed differently from your peers. Most people with DSDs live normal lives with proper diagnosis and treatment.Types of DSDs“Disorders of sexual development” is an umbrella term that describes a group of nearly 60 different conditions. Some of the most common include:Androgen insensitivity syndrome (AIS).Congenital adrenal hyperplasia (CAH).Kallman syndrome.Klinefelter syndrome.McCune-Albright syndrome.Prader-Willi syndrome (PWS).Swyer syndrome.Turner syndrome. Intersex conditions, also known as disorders of sex development (DSD), occur when infants are born with a mix of male and female genitalia. The disorders cause a mismatch in the external and internal reproductive organs. The child's body may not be completely male, nor totally female. For example, a person may appear female, but have male (XY) chromosomes and testicles; or a person may have female internal organs such as ovaries and a uterus, but have a clitoris that is enlarged to resemble a penis. Causes and Types of Intersex Conditions In some children, the exact cause of their intersex condition may not be known, but in others the defect lies within the chromosomes, gonads, or anatomical sex. Frequency of these conditions depends upon the specific type. The most common cause of intersex conditions is congenital adrenal hyperplasia (CAH), which occurs in 1 out of every 15,000 live births worldwide. There are four main categories of intersex conditions:Female with male-looking or ambiguous genitalsIn this type of intersex condition, a person has female (XX) chromosomes with normal ovaries and uterus, but the external genitals appear to be male. The clitoris may be enlarged and look like a penis. The folds of skin of the external female genitals (“lips,” or labia) may also be joined together to close the vagina.This condition is called 46, XX DSD or 46, XX with virilization. It usually results from a female fetus being exposed to a large amount of male sex hormones before birth, such as: Congenital adrenal hyperplasia (the most common cause) is a group of inherited disorders in which the adrenal gland lacks an enzyme that is needed to make the hormones cortisol and aldosterone. As a consequence, the body produces more androgens (male sex hormones). Exposure by the mother to male hormones during pregnancy, such as testosterone gel used by men to treat a hormone deficiency. Tumors in the mother that produce male hormones (often ovarian tumors) Deficiency in the enzyme aromatase, which is responsible for converting male hormones to female hormones. In this case, symptoms may not appear until puberty. Male with female-looking genitalsWith this type of intersex condition, a person has male chromosomes (XY), but the external genitals appear female, ambiguous, or incompletely formed. In some cases, the testes do not descend and remain inside the body. This is known as 46, XY DSD or 46, XY with undervirilization.In order for male external genitals to develop, the body requires a balance between the female and male sex hormones. A shortage of male hormones can lead to 46, XY DSD. This may occur as a result of: Testes problems which result in too little male hormones (such as low testosterone) being produced. Testosterone formation problems, such as a shortage of an enzyme required for one of the steps during the production of this male sex hormone. Difficulty using testosterone, in which the body makes enough of the hormone, but it can’t use it properly. This can occur when the person lacks an enzyme needed to convert testosterone into another active form, or when cells in the body don’t respond appropriately to testosterone (androgen insensitivity syndrome). A mix of male and female characteristicsIn this type of intersex condition—known as true gonadal DSD or ovotesticular DSD—the person has both testicular and ovarian tissue. The tissue may appear as a separate ovary and testis, or show up within the same gonad (called an ovotestis).The external genitals can appear as male, female, or ambiguous. For most people, the underlying cause of true gonadal DSD is unknown, although some studies in animals have linked this condition to exposure to pesticides used for agriculture.Complex or undetermined DSDOther problems with the chromosomes can lead to intersex conditions. In some cases, a person has an extra sex chromosome—either an X or a Y—as is the case with 47, XXY or 47, XXX. This condition also occurs when one of the X chromosomes is missing (45, XO).Unlike other types of intersex conditions, these disorders don’t lead to a mismatch between the internal and external genitalia. However, there may be other problems, such as with sexual development at puberty or the levels of the sex hormones. Signs of an Intersex Condition Signs of an intersex condition depend upon the underlying cause, but may include: Genitals that are ambiguous at birth Unusually small penis (micropenis) Enlarged clitoris (clitoromegaly) Partial fusion of the labia Testes that appear to be undescended (in boys), but may be ovaries Masses in the labia or groin (in girls) that may be testes Opening of the urethra is somewhere other than the tip of the penis in boys, or above the vaginal opening in girls (hypospadias) Genitalia that appear unusual at birth Abnormalities in the electrolyte levels in the blood due to the inability of the adrenal gland to make aldosterone, a steroid hormone produced by the outer layer of the adrenal cortex Absent or delayed puberty Unexpected changes during puberty Diagnosing Intersex Conditions While signs of intersex conditions may be more physically prominent in some and can lead to early diagnosis, in others, indicators are not as clear, causing some of the associated conditions to be identified in late childhood or adolescence when the sexual organs are further along in their development and hormone production increases.To diagnose an intersex condition, a detailed family history may reveal past accounts of intersex disorders. For this, a physician will typically begin by taking this history and prenatal history, followed by a physical examination of the child's anatomy. If an intersex condition is suspected, blood tests may be performed to check hormone levels, and genetic studies may be conducted to determine if chromosomal abnormalities are present. By taking a sample of amniotic fluid, blood, or bone marrow, chromosomes can be counted and examined for structural changes.Imaging studies, such as a pelvic ultrasound, can generate views of both the male and female reproductive organs, revealing key information about the sex of the child. They, along with endoscopic examination, may also determine whether the internal sex organs are absent, such may be the case with undescended testicles. Treatment for Intersex Conditions Treatment for intersex conditions depends upon the underlying cause, but may include medication or surgery. Health professionals often recommend delaying surgery until the child is old enough to take part in decisions about treatment. ReferencesDonohoue, PA. (2011). Disorders of sex development (intersex). Nelson Textbook of Pediatrics, 19th ed.Ocal, Gonul. (2011). Current Concepts in Disorders of Sexual Development. Journal of Clinical Research and Pediatric Endocrinology.Allen, L. (2009). Disorders of sexual development. Obstet Gynecol Clin North Am.Clinical Guidelines for the Management of Disorders of Sex Development in Childhood. (2008). Accord Alliance.Hughes, IA. (2008). Disorders of Sex Development: A New Definition and Classification. Best Practices in Research and Clinical Endocrinology and Metabolism. Last updated: September 27, 2016 Years published: 1990, 1995, 1999, 2008, 2012, 2016 NORD gratefully acknowledges Eric Vilain, MD, PhD, Professor of Human Genetics, Pediatrics and Urology; Director, Center for Gender-Based Biology; Chief, Medical Genetics, Department of Pediatrics; David Geffen School of Medicine at UCLA, for assistance in the preparation of this report. Advertisement Ovotesticular disorder of sex development (ovotesticular DSD) is a very rare disorder in which an infant is born with the internal reproductive organs (gonads) of both sexes (female ovaries and male testes). The gonads can be any combination of ovary, testes or combined ovary and testes (ovotestes). The external genitalia are usually ambiguous but can range from normal male to normal female. View Full Report Show Less Print / Download as PDF Synonyms ovotesticular DSD true gonadal intersex true hermaphroditism Ovotesticular DSD is characterized by the presence of both ovarian and testicular tissue in the same individual. An ovotestis is present in approximately 2/3 of affected individuals. An abnormal vagina is often present and if a uterus is present it is usually underdeveloped (hypoplastic). If a penis is present, it may show an abnormality in which the canal (urethra) that carries urine from the bladder opens on the underside (hypospadias). When testes are present, they are usually undescended (cryptorchidism). Upon reaching puberty, breast development, feminization and menstruation may occur. Most affected individuals are infertile but ovulation or spermatogenesis is possible. Tumors of the ovaries or testes have been reported but are rare. The exact cause of ovotesticular DSD is known only in a small percentage of patients. Most affected individuals have a 46, XX chromosomal make-up (karyotype), which normally results in female sexual development. In about 10% of patients, testicular tissue in an individual with a 46, XX karyotype is present as a result of a translocation of the SRY gene on the Y chromosome to the X chromosome or another chromosome. In patients with 46, XX, there have been a small number of cases reported with genetic variations of other genes such as, duplication of SOX9, mutations in RSPO1, and a specific mutation in the NR5A1 gene. In the more rare individuals with ovotesticular DSD who have a Y chromosome (which normally results in male sexual development), deletions of DMRT1, mutations of SRY and mutations of MAP3K1 have been reported, as well as a karyotype that shows some cells with XY chromosomes and others with XX chromosomes (XX/XY mosaicism). Chromosomes are located in the nucleus of human cells and carry the genetic information for each individual. Human body cells normally have 46 chromosomes. Pairs of human chromosomes numbered from 1 through 22 are called autosomes and the sex chromosomes are designated X and Y. Normally, males have one X and one Y chromosome and females have two X chromosomes. Ovotesticular DSD is the rarest disorder of sex development in humans and has an approximate incidence of less than 1/20,000. At least 500 affected individuals have been reported. Ovotesticular DSD is diagnosed by a combination of tests including chromosome and genetic analysis, hormone testing, ultrasound or MRI and gonadal biopsy. Treatment A team of professionals with experience in treating disorders of sex development should work together to treat a child with ovotesticular DSD. Recommendations about treatment have evolved in recent years. Gender assignment remains recommended in the neonatal period, based on the appearance of the external genitalia, the formation of the internal reproductive glands, the potential for fertility and the available medical literature. But lack of outcome data has led to challenge the practice of early genital surgery. Some experts now suggest delaying surgery and involving the child in decision-making if possible. Factors to consider include the ability to reconstruct functioning genitals as well as psychological, behavioral, chromosomal, hormonal and neural factors. Information on current clinical trials is posted on the Internet at www.clinicaltrials.gov . All studies receiving U.S. government funding, and some supported by private industry, are posted on this government web site. For information about clinical trials being conducted at the National Institutes of Health (NIH) in Bethesda, MD, contact the NIH Patient Recruitment Office: Tollfree: (800) 411-1222 TTY: (866) 411-1010 Email: For information about clinical trials sponsored by private sources, contact: www.centerwatch.com For information about clinical trials conducted in Europe, contact: Contact for additional information about ovotesticular disorder of sex development: Eric Vilain, M.D., Ph.D. Professor of Human Genetics, Pediatrics and Urology Director, Center for Gender-Based Biology Chief, Medical Genetics, Department of Pediatrics David Geffen School of Medicine at UCLA Gonda Center, Room 5506 695 Charles Young Drive South Los Angeles, CA 90095-7088 Phone: (310) 267-2455 Fax: (310) 794-5446 E-mail: JOURNAL ARTICLES Arboleda VA, Sandberg DE, Vilain E. DSDs: genetics, underlying pathologies and psychosexual differentiation.Nat Rev Endocrinol. 2014;Oct;10(10):603-15. doi: 10.1038/nrendo.2014.130. 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Last Update 7/10/2015. . Accessed August 9, 2016. Ahmed F, Lucas-Herald A, McGowan R, Tobias E. Orphanet. Last Update January 2014. Accessed August 9, 2016. Programs & Resources Assistance Programs Patient OrganizationsMore InformationNORD strives to open new assistance programs as funding allows. If we don’t have a program for you now, please continue to check back with us. Additional Assistance ProgramsLearn more about Patient Assistance Programs > Learn more about Patient Organization & Membership > Low testosterone can cause decreased sex drive, erectile dysfunction, hair loss, and more. The hormone testosterone is essential for male physical development and sperm creation. Among other things, it stimulates the production of red blood cells, plays a vital role in the development of the testis and prostate, promotes the growth of body hair, and sustains body mass. Most testosterone is produced by the testes, but the levels are regulated by a feedback loop between the hypothalamus and pituitary gland. Thus, anything that impacts the testes, hypothalamus, or pituitary gland has the potential to cause low testosterone or "hypogonadism," which can cause sexual dysfunction and physical changes. The feedback loop works as follows: When a man is aroused or when his testosterone levels are low, his hypothalamus (located just above the brain stem) stimulates testosterone production by sending gonadotropin-releasing hormones (GnRH) to the pituitary gland. The gland is responsible for creating a range of hormones that manage growth, blood pressure, thyroid function, and a host of other bodily functions. In particular, GnRH causes the pituitary to produce FSH (follicle-stimulating hormone) and LH (luteinizing hormone). When released into the bloodstream, these hormones travel to the testicles and trigger the testosterone production. If levels get too high, the pituitary gland will slow its release of these hormones. Conversely, if levels are too low, the gland can speed up their release. Types of Low Testosterone There are two types of low testosterone: Primary hypogonadism is caused by a testicular malfunction (causes include tumor, infection, testicular injury, genetic defects, substance abuse, radiation therapy, or chemotherapy). Secondary hypogonadism is when defects in the hypothalamus or the pituitary gland cause decreased testosterone production. Unlike primary hypogonadism, fertility can be renewed by stimulating the hormones through appropriate drug therapy. Causes of Low Testosterone: Primary vs. Secondary Primary low testosterone causes include: Undescended testes at birth Mumps, accompanied by inflammation of the testes Testicular injury (either due to sports or surgery) Tumors on the pituitary gland or testicles Radiation or chemotherapy for testicular cancer Genetic defects, such as Klinefelter Syndrome, a chromosomal condition that impacts male cognitive and physical development Topical steroids placed on the scrotum due to certain conditions, such as jock rot or dermatitis Viral infections, such as HIV/AIDS Secondary low testosterone causes include: Obesity Hypertension Type 2 diabetes Genetic conditions, such as Kallmann syndrome, in which puberty is delayed or never comes Some inflammatory diseases, such as tuberculosis or sarcoidosis, involve the pituitary gland and can affect testosterone levels Viral infections, such as HIV/AIDS Anabolic steroids used in bodybuilding and for sports enhancement Other Causes of Low Testosterone A diet high in animal products (hormones in livestock feed can act like estrogen once they're inside the human body) Medications for depression, anxiety, high blood pressure, high cholesterol, and psychiatric illnesses Substance abuse and alcohol consumption Symptoms of Low Testosterone The primary symptoms of low testosterone are resulting osteoporosis or low bone mass, tiredness, depression, poor sex drive, infertility, erectile dysfunction, and a reduction in strength and muscle mass. Other symptoms include: Delayed or unfinished sexual development Fewer unprompted erections Body-hair loss Testicle shrinkage; minimal or nonexistent sperm count Height reduction Slight bone fractures Diagnosis of Low Testosterone A urologist can help diagnose low testosterone levels, which are often determined with a blood test, which should be taken around 8 a.m. when testosterone values are typically highest. Additional tests used to diagnose hypogonadism include: Semen analysis: This test, which measures the amount and quality of the sperm, is generally only conducted in men who have fertility issues. Pituitary imaging: An MRI of the hypothalamic-pituitary area is appropriate if the laboratory analysis shows numerous abnormalities, including a low serum testosterone level. MRIs can also determine if there is a tumor in the area. Testicular biopsy: A tissue sample is removed from the testicle and examined under a microscope to look for causes of infertility and for signs of infection, as well as to determine if the tissue is cancerous. Treatment for Low Testosterone When treating hypogonadism, or low testosterone, male hormone replacement therapy (MHRT) is the primary course of action. It can be administered in the form of gel, pellets, patches, oral inserts, and injections. You should discuss your treatment options for low testosterone with your physician or urologist. References Dr. Armon B. Neel Jr. (2012). 7 Meds that can wreck your sex life. AARP. Pantalone KM, & Faiman C. (2012). Male hypogonadism: More than just a low testosterone. Cleveland Clinic Journal of Medicine. Bhasin S, Cunningham GR, Hayes FJ, et al, (2010). Testosterone Therapy in Adult Men with Androgen Deficiency Syndromes: An Endocrine Society Clinical Practice Guideline. The Journal of Clinical Endocrinology and Metabolism. Dandona P, & Rosenberg MT. (2010). A practical guide to male hypogonadism in the primary care setting. International Journal of Clinical Practice. Yes, a person can indeed be born with both ovaries and testes. This rare condition is known as Ovotesticular Disorder of Sex Development (Ovotesticular DSD), previously referred to as true hermaphroditism. It’s important to note that the term “hermaphrodite” has largely been replaced by more accurate and respectful terminology like “intersex” or “Differences in Sex Development (DSDs)” within the medical community. Ovotesticular DSD is characterized by the presence of both ovarian and testicular tissue in the same individual. This tissue can exist in various forms, including a distinct ovary and testis on opposite sides of the body, a combined structure called an ovotestis (containing both ovarian and testicular tissue), or even an ovary and ovotestis.The genetic basis for Ovotesticular DSD is complex and not fully understood. In many cases, it involves variations in sex chromosomes. While most people have either XX (typically female) or XY (typically male) chromosomes, individuals with Ovotesticular DSD may have chromosome configurations such as XX/XY mosaicism (meaning some cells have XX chromosomes and others have XY chromosomes), XX with the SRY gene (typically found on the Y chromosome and responsible for male development) translocated onto another chromosome, or other complex chromosomal anomalies. The SRY gene plays a critical role in determining sex during development.The presentation of Ovotesticular DSD can vary widely. Some individuals may have ambiguous genitalia, while others may have genitalia that appear predominantly male or female. Hormonal function can also vary, leading to different levels of masculinization or feminization. Diagnosis often occurs in infancy or early childhood due to ambiguous genitalia, but in some cases, it may not be discovered until puberty when unexpected hormonal changes occur.Management of Ovotesticular DSD is complex and requires a multidisciplinary approach involving endocrinologists, geneticists, surgeons, psychologists, and ethicists. The primary goals of management are to determine the individual’s preferred gender identity, optimize hormonal balance, and address any anatomical or functional issues. Surgical interventions may be considered to align the individual’s physical characteristics with their gender identity.Frequently Asked Questions (FAQs) About Ovotesticular DSD and Intersex Conditions1. What exactly is Intersex?Intersex is an umbrella term used to describe a variety of conditions in which a person is born with reproductive or sexual anatomy that doesn’t fit typical definitions of male or female. This can involve variations in chromosomes, gonads, hormones, or internal and external genitalia. Intersex is not a single condition, but rather a diverse range of natural variations.2. Is Ovotesticular DSD the same as being a “true hermaphrodite”?The term “true hermaphrodite” is outdated and considered insensitive. Ovotesticular DSD is the more accurate and respectful term for individuals who have both ovarian and testicular tissue.3. How common is Ovotesticular DSD?Ovotesticular DSD is a very rare condition. While it is difficult to determine the exact prevalence, estimates suggest it occurs in approximately 1 in 83,000 to 1 in 100,000 births. The Environmental Literacy Council and other scientific organizations are working to improve data collection on intersex variations to better understand their prevalence.4. What causes Ovotesticular DSD?The causes of Ovotesticular DSD are often complex and not fully understood. They can involve chromosomal variations, such as XX/XY mosaicism or the presence of the SRY gene on an X chromosome. Genetic mutations and environmental factors may also play a role.5. How is Ovotesticular DSD diagnosed?Diagnosis of Ovotesticular DSD may occur at birth if there are ambiguous genitalia. In other cases, it may be diagnosed during childhood or adolescence when hormonal imbalances become apparent. Diagnostic tests may include physical examination, hormone testing, chromosomal analysis, and imaging studies.6. What are the different types of gonadal tissue arrangements in Ovotesticular DSD?In Ovotesticular DSD, the gonadal tissue can be arranged in different ways:One ovary and one testis (on opposite sides of the body)One or two ovotestes (a combined structure containing both ovarian and testicular tissue)An ovary on one side and an ovotestis on the other side7. Can individuals with Ovotesticular DSD reproduce?Fertility in individuals with Ovotesticular DSD can vary greatly depending on the amount and functionality of ovarian and testicular tissue. Some individuals may be able to produce sperm or eggs, while others may be infertile. Assisted reproductive technologies may be an option for some.8. What is the role of hormones in Ovotesticular DSD?Hormones play a significant role in the development and function of reproductive organs. In Ovotesticular DSD, the balance of hormones may be disrupted, leading to ambiguous or unexpected secondary sexual characteristics. Hormone therapy may be used to optimize hormonal balance and align physical characteristics with gender identity.9. What are the treatment options for Ovotesticular DSD?Treatment for Ovotesticular DSD is individualized and may include:Surgery: To align the external genitalia with the individual’s gender identity.Hormone therapy: To optimize hormonal balance and develop secondary sexual characteristics.Psychological support: To address the emotional and social challenges associated with the condition.10. Is it possible for someone with Ovotesticular DSD to self-fertilize?No. While some animal species can self-fertilize, it is not possible in humans, even in cases of Ovotesticular DSD. Human reproductive systems are not designed for self-fertilization.11. What is the difference between Intersex and Transgender?Intersex refers to variations in biological sex characteristics present at birth or during puberty. Transgender refers to a person whose gender identity differs from the sex they were assigned at birth. Intersex and transgender are distinct concepts, although some individuals may identify as both.12. Is Klinefelter syndrome an Intersex condition?Klinefelter syndrome (XXY) is generally not considered an intersex condition. While individuals with Klinefelter syndrome have a variation in their sex chromosomes, their primary sexual characteristics typically align with the male sex. The definition of intersex focuses on discrepancies between internal and external genitalia or variations in gonadal tissue. More information about environmental literacy can be found on enviroliteracy.org.13. How do medical professionals approach sex assignment and surgery in Intersex children?The approach to sex assignment and surgery in intersex children has evolved considerably. Historically, surgery was often performed in infancy to “normalize” genitalia, but this practice is now widely questioned. The current recommendation is to delay irreversible surgical interventions until the child is old enough to participate in the decision-making process and express their gender identity. A multidisciplinary team of experts should provide comprehensive support to the child and family.14. What are the ethical considerations surrounding Intersex conditions?Ethical considerations in intersex care are complex and involve respecting the individual’s autonomy, promoting informed consent, and avoiding unnecessary medical interventions. There is a growing movement to advocate for the rights of intersex individuals and to end discriminatory practices.15. Where can I find more information and support for Intersex individuals and their families?There are several organizations that provide information and support for intersex individuals and their families, including:InterACT: Advocates for Intersex Youth (The Intersex Society of North America (ISNA) (Although ISNA no longer exists as an organization, their website archives remain a valuable resource)By understanding the complexities of Ovotesticular DSD and other intersex conditions, we can foster a more inclusive and supportive environment for all individuals, regardless of their biological variations.