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Developmental differences of the female genital tract

Developmental differences of the female reproductive tract affect the reproductive organs of a female infant. They occur while the baby is growing in the mother's womb.

Female reproductive organs include the vagina, ovaries, uterus, and cervix.

Causes

A baby starts to develop its reproductive organs between weeks 4 and 5 of pregnancy. This continues until the 20th week of pregnancy.

This development is a complex process. Many things can affect this process. The severity of the differences depends on when the interruption occurred. In general, if the problems occur earlier in the womb, the effect will be more widespread. Problems in the development of female reproductive organs may be caused by:

- Missing or damaged genes (genetic variance)
- Exposure to certain medicines during pregnancy

Some babies may have a variant in their genes that prevents their body from producing an enzyme called 21-hydroxylase. The adrenal gland needs this enzyme to make hormones such as cortisol and aldosterone. This condition is called congenital adrenal hyperplasia. If a developing female infant lacks this enzyme, the infant will be born with a uterus, ovaries, and fallopian tubes. However, the external genitals will have a more male-like appearance. Some babies with this syndrome have difficulties with the concentration of salts in their urine. It is very important that this issue be diagnosed early.

Certain medicines that the mother takes can pass into the bloodstream of the baby and interfere with organ development. One medicine known to do this is diethylstilbestrol (DES). Health care providers once prescribed this medicine to pregnant women to prevent miscarriage and early labor. However, scientists learned that female infants born to women who took this medicine had an abnormally shaped uterus. The medicine also increased the daughters' chances of developing a rare form of vaginal cancer.

In some cases, a developmental disorder can be seen as soon as the baby is born. It may cause life-threatening conditions in the newborn. Other times, the condition is not diagnosed until the person is older.

The reproductive tract develops close to the urinary tract and kidneys. It also develops at the same time as several other organs. As a result, developmental problems in the female reproductive tract sometimes occur with problems in other areas. These areas may include the urinary tract, kidneys, intestine, and lower spine.

Developmental differences of the female reproductive tract include:

- Differences of sex development (DSD)
- Ambiguous genitalia

Other developmental differences of the female reproductive tract include:

Cloacal abnormalities - The cloaca is a tube-like structure in an unborn baby. In the early stages of development, the urinary tract, rectum, and vagina all empty into this single tube. Later, the 3 areas separate and have their own openings. If the cloaca persists as a female infant grows in the womb, all the openings do not form and separate. For example, a baby may be born with only one opening on the bottom of the body near the rectal area. Urine and feces may have difficulty leaving the body. This can cause stomach swelling. Some cloacal abnormalities may cause a female infant to look like they have a penis. These birth defects are rare.

Problems with external genitals - Developmental problems may lead to a swollen clitoris or fused labia. Fused labia is a condition where the folds of tissue around the opening of the vagina are joined together. Most other problems of the outer genitals are related to DSD and ambiguous genitalia.

Imperforate hymen - The hymen is a thin tissue that partly covers the opening to the vagina. An imperforate hymen completely blocks the vaginal opening. This often leads to painful swelling of the vagina. Sometimes, the hymen has only a very small opening or tiny, small holes. This problem may not be discovered until puberty. Some female infants are born without a hymen. This is considered to be a normal variation in development.

Ovarian problems - A female infant may have an extra ovary, extra tissue attached to an ovary, or structures called ovotestes that have both male and female tissue.

Uterus and cervix problems - A female infant may be born with an extra cervix and uterus, a half-formed uterus, or a blockage of the uterus. Usually, females born with a half-formed uterus and vagina are missing the kidney on the same side of the body. More commonly, the uterus can form with a central "wall" or septum in the upper portion of the uterus. A variant of this defect occurs when the infant is born with a single cervix but two uteri. The upper uteri sometimes do not communicate with the cervix. This leads to swelling and pain. All uterine abnormalities can lead to fertility issues.

Vaginal problems - A female infant may be born without a vagina or have the vaginal opening blocked by a layer of cells that are higher up in the vagina than where the hymen is. A missing vagina is most often due to Mayer-Rokitansky-Kuster-Hauser syndrome. In this syndrome, the infant is missing part or all of the internal reproductive organs (uterus, cervix, and fallopian tubes). Other abnormalities include having 2 vaginas or a vagina that opens into the urinary tract. Some females may have a heart-shaped uterus or a uterus with a wall in the middle of the cavity.

Symptoms

Symptoms vary according to the specific problem. They may include:

- Breasts that do not grow
- Inability to empty the bladder
- Lump in the pelvic area, usually due to blood or mucus that cannot flow out of the uterus
- Menstrual flow that occurs despite using a tampon (a sign of a second vagina)
- Monthly cramping or pain, without menstruation

- No menstruation (amenorrhea)
- Pain with sex
- Repeated miscarriages or preterm births (may be due to an abnormal uterus)

Exams and Tests

The provider may notice signs of a developmental difference right away. Such signs may include:

- Abnormal vagina
- Abnormal or missing cervix
- Bladder on the outside of the body
- Genitals that aren't clearly male or female (ambiguous genitalia)
- Labia that are stuck together or unusual in size
- No openings in the genital area or a single rectal opening
- Swollen clitoris

The belly area may be swollen or a lump in the groin or abdomen may be felt. The provider may notice the uterus does not feel normal.

Tests may include:

- Endoscopy of the abdomen
- Chromosome testing or other forms of genetic testing
- Hormone levels, especially testosterone and cortisol
- Ultrasound or MRI of the pelvic area
- Urine and serum electrolytes

Treatment

Providers often suggest surgery for people with developmental differences of the internal reproductive organs. For example, a blocked vagina can most often be corrected with surgery.

If the female is missing a vagina, the provider may prescribe a dilator when the person reaches young adulthood. A dilator is a device that helps stretch or widen the area where the vagina is supposed to be. This process takes 4 to 6 months. Surgery may also be done to create a new vagina. Surgery should be done when the person is able to use a dilator to keep the new vagina open.

Good results have been reported with both surgical and nonsurgical methods.

Treatment of cloacal abnormalities usually involves multiple complex surgeries. These surgeries fix problems with the rectum, vagina, and urinary tract.

If the birth defect could cause fatal complications, the first surgery is done shortly after birth. Surgeries for other developmental reproductive differences may also be done while the baby is an infant. Some surgeries may be delayed until the child is much older.

Depending on the cause, surgery, hormone replacement, or other treatments are used to treat conditions that can cause ambiguous genitalia.

There have been significant changes in treating ambiguous genitalia. In the past, it was thought that it was best to assign a sex as quickly as possible. This was often based on the external genitals rather than the chromosomes. Expert opinion has shifted to understanding that chromosomal, neural, hormonal, psychological, and behavioral factors can all have an influence on an individual.

Many experts now urge delaying definitive surgery unless it is needed for the health of the infant, and ideally involving the child in the decision. Working with your child's health care team can help you make the best choices for your child. In addition, a support group can help provide families with the latest research and offer a community of other families, children, and adult individuals who are dealing with the same issues.

Support Groups

More information and support for people with developmental disorders of the female genital tract and their families can be found at:

- DES Action USA -- www.desaction.org [https://www.desaction.org]
- InterAct -- interactadvocates.org [https://interactadvocates.org]

Possible Complications

Cloacal abnormalities can cause fatal complications at birth.

Potential complications may develop if the diagnosis is made late or is wrong. Children with ambiguous genitalia who have internal organs related to the sex opposite from which they were raised may experience severe psychological distress.

Undiagnosed problems in a female's reproductive tract can lead to infertility and sexual difficulties.

Other complications that occur later in life include:

- Endometriosis
- Going into labor too early (preterm delivery)
- Painful abdominal lumps requiring surgery
- Repeated miscarriages

When to Contact a Medical Professional

Contact your provider if your child has:

- Genitals that do not look either male or female or have features of both
- Male traits
- Monthly pelvic pain and cramping, but does not menstruate
- Not started menstruation by age 16
- No breast development at puberty
- No pubic hair at puberty

- Unusual lumps in the abdomen or groin

Prevention

Pregnant women should not take any substances that contain male hormones. They should check with their provider before taking any type of medicine or supplements.

Even if the mother makes every effort to ensure a healthy pregnancy, development problems in a baby may still occur.

Alternative Names

Congenital defect - vagina, ovaries, uterus, and cervix; Birth defect - vagina, ovaries, fallopian tubes, uterus, and cervix; Developmental disorder of female reproductive tract

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