CONGENITAL ABSENCE OF THE VAS DEFERENS

Cavd

A condition in which the vasa deferentia reproductive organs fail to form properly prior to birth.

Diagnosis   Male

Related Diagnoses:
Obstructive azoospermia

About Congenital absence of the vas deferens

Congenital absence of the vas deferens (CAVD) means that the vasa deferentia (Pic. 1) which connect the sperm-producing testicles to the penis are not formed at birth. The vas deferens derives from the Wolffian (mesonephric) duct and shares a common origin with the kidney. Intrinsic Wolffian duct defects may result in failure of the vas deferens to develop, a condition that can occur in isolation or combined with renal agenesis (a medical condition in which one or both fetal kidneys fail to develop; URA) or malformations.

A missing vas, that is, unilateral absence of vas deferens, albeit relatively uncommon may be found by urologists performing vasectomies or evaluating men with fertility problems. Therefore, those who are missing both vas deferens are typically able to create sperm but are unable to transport them appropriately. Their semen does not contain sperm, a condition known as azoospermia. It may either be unilateral (CUAVD) or bilateral (CBAVD).

Scrotal ultrasonography and transrectal ultrasonography (TRUS) are useful in detecting uni- or bilateral CBAVD, which may be associated with visible abnormalities or agenesis of the epididymis, seminal vesicles or kidneys.

There are two main populations of CAVD; the larger group is associated with cystic fibrosis and occurs because of a mutation in the CFTR gene, while the smaller group (estimated between 10 and 40%) is associated with unilateral renal agenesis. The genetic basis of this second group is not well understood.

Cystic fibrosis

Cystic fibrosis (CF) is a genetic disorder that affects mostly the lungs but also the pancreas, liver, kidneys, and intestine (Pic. 2).

If both parents have the cystic-fibrosis gene, and pass it onto child, the child will get cystic fibrosis. A cystic-fibrosis gene is needed from each parent. The parent might not have cystic fibrosis but still might have the gene.

It causes the body to make thick, sticky mucus, which builds up in the lungs, the digestive system and other parts of the body. Long-term issues include difficulty breathing and coughing up mucus as a result of frequent lung infections. Other signs and symptoms include sinus infections, poor growth, fatty stool, clubbing of the fingers and toes, and infertility in males among others. Different people may have different degrees of symptoms.

CAVD is one of the most consistent features of cystic fibrosis as it affects 98-99% of individuals in this CF patient population. In contrast, acute or persistent respiratory symptoms present in only 51% of total CF patients.
There is no cure for cystic fibrosis. Inhaled antibiotics are used to keep bacteria from growing in the thick mucus. Inhaled salt-water helps keep the lungs moisturized.

**Renal agenesis**

Renal agenesis is a medical condition in which one (unilateral) or both (bilateral) fetal kidneys fail to develop. This absence of kidneys causes oligohydramnios, a deficiency of amniotic fluid in a pregnant woman, which can place extra pressure on the developing baby and cause further malformations.

Males are more commonly affected and most infants that are born alive do not live beyond four hours.

Unilateral renal agenesis is much more common, but is not usually of any major health consequence, as long as the other kidney is healthy.

Adults with unilateral renal agenesis have considerably higher chances of hypertension (high blood pressure). People with this condition are advised to approach contact sports with caution.

**Associated diseases**

- cystic fibrosis
- renal agenesis

**Complications**

- chronic infections
- damaged airways
- growths in the nose
- pneumothorax (a collapsed lung)
- respiratory failure
- nutritional deficiencies
- diabetes
- blocked bile duct
- intestinal obstruction
- thinning of the bones (osteoporosis)
- electrolyte imbalances and dehydration

**Risk factors**

Cystic fibrosis is inherited, so it runs in families. Also it is most common in white people of Northern Europe ancestry.

The renal agenesis is frequently, but not always the result of a genetic disorder, and is more common in infants born to one or more parents with a malformed or absent kidney.

**Impact on fertility**

At least 97% of men with cystic fibrosis are infertile but not sterile, due to condition called obstructive azoospermia (OA), and can have children. Many azoospermic men maintain sperm production at varying levels within the testes. Although selected cases of OA may be surgically correctable, treatment options for most couples with azoospermia-related infertility will ultimately include assisted reproductive techniques (ART).

The main cause of infertility in men with cystic fibrosis is congenital absence of the vas deferens, but potentially also by other mechanisms such as causing no sperm, teratospermia (a condition characterized by the presence of sperm with abnormal shape), and few sperm with poor motility. This all lead to inability to produce sperms capable of fertilization.

Many men found to have congenital absence of the vas deferens during evaluation for infertility have a mild, previously undiagnosed form of CF.
Prevention

If someone in family suffer from cystic fibrosis it is possible to undergo genetic testing before having babies.

Symptoms

Cystic fibrosis

The main signs and symptoms of cystic fibrosis are salty-tasting skin, poor growth, and poor weight gain despite normal food intake, accumulation of thick, sticky mucus, frequent chest infections, and coughing or shortness of breath.

Males can be infertile due to congenital absence of the vas deferens.

Renal agenesis

In case of bilateral renal agenesis, symptoms occur in utero during pregnancy. A deficiency of amniotic fluid in pregnant women is called oligohydramnion. An extra pressure on the developing baby can cause other malformations. Very often the child will not survive more than few days after delivery.

Adults with unilateral renal agenesis have considerably higher chances of hypertension.

Therapies

Self therapy

While not well supported by evidence, many people use airway clearance techniques such as chest physiotherapy in case of cystic fibrosis.

Conventional medicine

Both conditions (cystic fibrosis and renal agenesis) which are associated with congenital absence of vas deferens are not curable. There are several methods, which can cease the symptoms and help in future life. Pharmacotherapy helps to infections and inhalation drugs for better breathing can be helpful.

Surgical therapy is the option in very severe cases of lung and pancreatic damage.

Pharmacotherapy

There is no cure for cystic fibrosis. Lung infections are treated with antibiotics which may be given intravenously, inhaled, or by mouth. Inhaled hypertonic saline and salbutamol may also be useful. Pancreatic enzyme replacement and fat-soluble vitamin supplementation are important, especially in the young.

Surgical therapy

Lung transplantation may be an option if lung function continues to worsen. Lung transplantation is considered when lung function declines to the point where assistance from mechanical devices is required or someone’s survival is threatened.

Although single lung transplantation is possible in other diseases, individuals with CF must have both lungs replaced because the remaining lung might contain bacteria that could infect the transplanted lung.
A pancreatic or liver transplant may be performed at the same time in order to alleviate liver disease and/or diabetes.

Other therapy

Gene therapy

Gene therapy has been explored as a potential cure for CF. Results from clinical trials have shown limited success as of 2016, and using gene therapy as routine therapy is not suggested.

The focus of much CF gene therapy research is aimed at trying to place a normal copy of the CFTR gene into affected cells. Transferring the normal CFTR gene into the affected epithelium cells would result in the production of functional CFTR protein in all target cells, without adverse reactions or an inflammation response. The treatment has little effect.

Assisted reproduction

Individuals with CAVD can reproduce with the assistance of modern technology with a combination of testicular sperm extraction and intracytoplasmic sperm injection (ICSI). Men who ejaculate no sperm require some form of surgical sperm retrieval to enable ICSI to take place. Epididymal sperm obtained by microsurgical aspiration (MESA) or percutaneous sperm aspiration (PESA) and testicular sperm obtained by surgical excision (TESE) or percutaneous aspiration (TESA) are used in ICSI treatment. Alternatively, the retrieved sperm can be cryopreserved for use in future sperm injection attempts. If all efforts to extract vital sperm cells fails, then donated ones may be recommended.

Couples who are pregnant or planning a pregnancy can have themselves tested for the CFTR gene mutations to determine the risk that their child will be born with cystic fibrosis. Testing is typically performed first on one or both parents and, if the risk of CF is high, testing on the fetus is performed.

The preimplantation genetic screening (PGS)/preimplantation genetic diagnosis (PGD) allows studying the DNA of eggs or embryos to select those that carry certain damaging characteristics.

Find more about related issues

Diagnoses

Obstructive azoospermia
Absence of sperm in the ejaculate despite normal spermatogenesis, caused by an obstruction of the genital tract.
Learn more at: www.fertilypedia.org/thrapy/diag/obstructive-azoospermia

Therapies

Egg donation
Process by which a woman donates eggs for purposes of assisted reproduction or biomedical research.
Learn more at: www.fertilypedia.org/edu/therapies/egg-donation

ICSI
A micromanipulative fertilization technique in which a single sperm is injected directly into an egg.
Learn more at: www.fertilypedia.org/edu/therapies/icsi

Sperm donation
The procedure in which a man (sperm donor) provides his sperm for fertility treatment.
Learn more at: www.fertilypedia.org/edu/therapies/sperm-donation
Standard IVF
A process in which an egg is fertilised by sperm outside the body: in vitro. Own or donated gametes may be used.
Learn more at: www.fertilitypedia.org/edu/therapies/standard-ivf

Gallery

**Pic**
Testis, spermatic vessels and vas deferens.

**Pic**
Figure A shows the organs that cystic fibrosis can affect. Figure B shows a cross-section of a normal airway. Figure C shows an airway with cystic fibrosis. The widened airway is blocked by thick, sticky mucus that contains blood and bacteria.

Sources

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