AUTOIMMUNE DISORDERS

A condition arising from an abnormal immune response to a normal body part.

診断 Diagnosis ♂ Male & Female

Related Diagnoses:
Endometriosis | Thyroid disorders | Turner syndrome | Repeated implantation failure | Antiphospholipid syndrome |
Premature ovarian failure | Menopause | Sperm autoantibodies | Lupus erythematosus | Hyperthyroidism

ℹ️ About Autoimmune disorders

Autoimmune diseases arise from an inappropriate immune response of the body against substances and tissues normally present in the body. In other words, the immune system mistakes some part of the body as a pathogen and attacks its own cells. Autoimmune diseases are commonly considered complex immune disorders. While many autoimmune diseases are rare, collectively these diseases afflict millions of patients. Despite their clinical diversity, they have one similarity, namely the dysfunction of the immune system.

Some autoimmune diseases attack specific organs, whereas others are more systemic:

- Organ-Specific Autoimmune Diseases

Some autoimmune diseases are considered organ specific, meaning that the immune system targets specific organs or tissues.

- Systemic Autoimmune Diseases

Whereas organ-specific autoimmune diseases target specific organs or tissues, systemic autoimmune diseases are more generalized, targeting multiple organs or tissues throughout the body.

It is suspected that genetic defects play a role in the etiology of these diseases. In contrast to classical inherited genetic diseases, like sickle cell anemia, autoimmune diseases are not caused by the defect of a single gene, but by the dysfunction of the complex interaction of a group of genes. Although no autoimmune disease has been completely analyzed, there has been tremendous success in recent years in identifying major players in the development of autoimmune diseases.

There are over 50 publications that list gene variants that are associated with a certain autoimmune disease. Interestingly, a lot of these genes are located in the same regions on the chromosomes (contains DNA molecule with part or all of the genetic material). This has led to a “common cause hypothesis” of autoimmune disorders. However, defects of one or more of these genes do not cause an autoimmune disease, but only predispose a person for an autoimmune disease.

The environmental influences are very diverse, rendering research in this area extremely difficult. These influences may be toxic substances like mercury in one case and ultraviolet light or even certain nutrients in another. Moreover, several bacteria, viruses, or hormones are among the suspected triggers of autoimmune disorders. The treatment of autoimmune diseases is typically with immunosuppression: medication which decreases the immune response.

Associated diseases

Organ-specific autoimmune diseases:

- celiac disease
Graves disease (the most common cause of hyperthyroidism - excessive production of thyroid hormone by the thyroid gland)
- Hashimoto thyroiditis (thyroid gland is gradually destroyed)
- type 1 diabetes mellitus
- Addison disease (adrenal glands do not produce enough steroid hormones)
- premature ovarian failure (POF; absent menarche (primary amenorrhea) or premature depletion of ovarian follicles before the age of 40 years)
- polycystic ovary syndrome (PCOS; woman’s ovaries or adrenal glands produce more male hormones than normal)
- endometriosis (growth of endometrial tissue outside the uterine cavity)
- unexplained infertility (an infertile couple whose standard investigations (semen analysis, tubal patency, and laboratory assessment of ovulation) yield normal results)
- thyroid disorders

**Systemic autoimmune diseases:**
- multiple sclerosis (damaging of insulating covers of nerve cells in the brain and spinal cord)
- myasthenia gravis (weakness in the skeletal muscles)
- psoriasis (autoimmune disease affecting the skin)
- rheumatoid arthritis (primarily affects joints)
- systemic lupus erythematosus (immune system mistakenly attacks healthy tissue in many parts of the body)

**Complications**

Complications depend on the disease. Medicines used to suppress the immune system can cause severe side effects, such as higher risk of infections.

**Pregnancy complications**

Systemic lupus erythematosus may occur with secondary antiphospholipid syndrome (APS). APS provokes blood clots (thrombosis) in both arteries and veins as well as pregnancy-related complications such as miscarriage, stillbirth, preterm delivery, and severe preeclampsia (pregnancy complication characterized by the onset of high blood pressure and often a significant amount of protein in the urine).

**In vitro fertilization (IVF) failure**

Autoimmunity and the presence of autoantibodies have been invoked as a possible mechanism of IVF failure. There are contradicting data regarding the importance of certain antibodies to damage directly the preimplantation embryo, interfering with implantation process or formation of placenta. Consequently, the overall activation of the immune system in female infertility has been suggested.

**Side effects of immunosuppressant drugs**

A common side-effect of many immunosuppressive drugs is immunodeficiency (weak immune system), because the majority of them act non-selectively, resulting in increased susceptibility to infections and decreased cancer immunosurveillance. There are also other side-effects, such as hypertension (high blood pressure), dyslipidemia (abnormal amount of lipids in the blood), hyperglycemia (high blood sugar), peptic ulcers, lipodystrophy (abnormal distribution of fat), moon face (full rounded faces), liver and kidney injury. The immunosuppressive drugs also interact with other medicines and affect their metabolism and action. Actual or suspected immunosuppressive agents can be evaluated in terms of their effects on lymphocyte subpopulations (white blood cells) in tissues using immunohistochemistry (imaging of antigens in cells of a tissue section).

Some of immunosuppressant drugs can cause birth defects, while others carry milder risks during pregnancy and breast-feeding. In any case, patient should talk to the doctor before taking an immunosuppressant drug if planning to become pregnant.

**Risk factors**

- genetical predisposition
- environmental factors
- female gender
- childbearing years
- infection
Male infertility

Anti-sperm antibodies (ASAs) or immunity to sperm markedly reduce sperm quality. Anti-sperm antibodies interact with numerous sperm antigens that perform functions in fertility, markedly decreasing the chance of pregnancy by reducing sperm count, motility, and viability, leading to male or female "immune infertility".

Female infertility

Conflicting reports have described the presence of antibodies to human chorionic gonadotropin (hCG; hormone produced by the placenta after implantation) and its relation to female fertility and pregnancy outcome. Pathological mechanisms and immunologic pathways, involved in anti-hCG autoantibody formation in women with preserved conception or recurrent pregnancy loss have been noted previously but remain poorly understood. As no consensus exists, various approaches to specific treatment of patients with possible autoimmune factor of infertility have been introduced.

Autoimmune mechanisms as well as an increased production of multiple autoantibodies are involved in such infertility disorders as premature ovarian failure (POF), endometriosis, polycystic ovary syndrome (PCOS), unexplained infertility, and repeatedly unsuccessful IVF attempts and may be responsible for the pathophysiology of preeclampsia or spontaneous abortions.

The impact of a particular autoantibody on the pathogenesis of infertility is not uniformly understood. Antinuclear autoantibodies (ANA) could interfere with early implantation of embryo and smooth muscle autoantibodies (SMA) could alter the fallopian tube function. It is concluded that antiphospholipid antibodies (APA) may be involved in uterine vascular modifications affecting implantation processes. Except antiovarian antibodies (AOA) in ovulatory dysfunctions and disease-specific autoantibodies described in case of endometriosis, autoantibodies detected in infertile patients are usually not specific to infertility or to the gynecological diseases leading to infertility. Furthermore, the number of detectible autoantibodies, in particular, has been proposed to predict the pregnancy rate of IVF treatment. Therefore, some studies suggest lesser importance of specific autoantibodies and stress the key role of overall activation of the immune system in reduced fecundity.

Prevention

This condition cannot be prevented. While pregnant, the autoimmune disease may flare-up, but this can be controlled with medications.

Symptoms

Autoimmune diseases can have a variety of mixed symptoms (Tab. 1) that flare up and disappear, making diagnosis difficult.

In some cases conditions that affect the blood vessels can cause problems, such as erectile dysfunction in men, while women who have autoimmune diseases affecting their glands may have vaginal dryness. Joint pain can also inhibit sexual relationships.

Therapies

Self therapy

There is no self-therapy for this condition.
Ultimately, damage to tissues and organs in the autoimmune disease state comes as a result of inflammatory responses that are inappropriate; therefore, treatment often includes immunosuppressive drugs and corticosteroids.

**Pharmacotherapy**

**Immunosuppressive drugs**

Immunosuppressant drugs are a class of drugs that suppress, or reduce, the strength of the body's immune system. Because immunosuppressant drugs weaken the immune system, they suppress autoimmune reaction. This helps reduce the impact of the autoimmune disease on the body. All immunosuppressant drugs are available only by a prescription. Regular blood tests are done during the treatment to monitor how effective the drugs are.

**Pain-killing and anti-inflammatory medication**

To reduce inflammation and pain due to autoimmune disease, painkillers and anti-inflammatory drugs are used, such as paracetamol and codeine as painkillers and corticosteroids to reduce inflammation.

**Surgical therapy**

Surgery may be opted to replace the damaged joint or to treat intestinal blockage in the case of Crohn's disease (inflammatory disease of the intestines).

**Other therapies**

**Physical therapy**

Physical therapy is used as a supportive treatment to encourage mobility.

**Assisted reproduction**

If infertility persists after the treatment, assisted reproductive technologies (ART) give an option. Assisted reproductive technology is the technology used to achieve pregnancy in procedures such as fertility medication, artificial insemination, in vitro fertilization and surrogacy. It is reproductive technology used primarily for infertility treatments, and is also known as fertility treatment.

If conservative medical treatments fail to achieve a full term pregnancy, the physician may suggest the patient undergo in vitro fertilization (IVF). IVF and ART generally start with stimulating the ovaries to increase egg production. However, the success of attaining pregnancy following IVF depends on the effectiveness of controlled ovarian hyperstimulation (COH). Generally, women with autoimmune disease need two layers of treatment: a first layer to conceive, and a second to prevent pregnancy loss after conception depending on the disease. Disease control should be achieved prior to the institution of fertility drug.

Most fertility medications are agents that stimulate the development of follicles in the ovary. Examples are gonadotropins and gonadotropin releasing hormone (GnRH). It is recommended to use protocols for the stimulation of multiple ovulations with limited use of gonadotropins, aiming at reducing ovarian hyperstimulation syndrome (OHSS; ovaries become swollen and painful) risks, multiple pregnancies, economic burdens and the couple's emotional stress and keeping a good oocyte quality as well as similar success possibilities. This concerns all patients, but mainly those who must avoid or limit their exposure to high estrogen levels, for instance, patients suffering from autoimmune diseases. After stimulation, the physician surgically extracts one or more eggs from the ovary, and unites them with sperm in a laboratory setting, with the intent of producing one or more embryos. Fertilization takes place outside the body, and the fertilized egg is reinserted into the woman's reproductive tract, in a procedure called embryo transfer.
Intracytoplasmic sperm injection (ICSI) is beneficial in the case of male factor infertility where sperm counts are very low or failed fertilization occurred with previous IVF attempt(s). The ICSI procedure involves a single sperm carefully injected into the center of an egg using a microneedle. With ICSI, only one sperm per egg is needed. Without ICSI, you need between 50,000 and 100,000.

Two techniques that enable to some extent the selection of physiologically normal spermatozoa have recently been developed. One of these is termed intracytoplasmic morphology-selected sperm injection (IMSI). Here, spermatozoa are selected for ICSI and analysed digitally prior to the microinjection procedure in order to deselect morphologically abnormal spermatozoa. With this technique, abnormalities not visible in standard ICSI procedures have been observed. IMSI increases the pregnancy rate during ICSI cycles, and some data suggests that the level of pregnancy termination is also decreased. A second technique recently introduced to assisted reproduction is that of sperm selection with hyaluronic acid (HA), e.g. PICSI. In this technique, mature sperm with HA receptors are distinguished from immature and abnormal sperm since these do not express such receptors.

The rate of success for IVF is correlated with a woman’s age. More than 40 percent of women under 35 succeed in giving birth following IVF, but the rate drops to a little over 10 percent in women over 40.

### Diagnoses

#### Endometriosis
A state in which pieces of the tissue alike to the lining of the uterus (endometrium) grow in other parts of the body.
Learn more at: [www.fertilitypedia.org/therapy/diag/endometriosis](http://www.fertilitypedia.org/therapy/diag/endometriosis)

#### Thyroid disorders
A medical condition impairing the function of the thyroid.
Learn more at: [www.fertilitypedia.org/therapy/diag/thyroid-disorders](http://www.fertilitypedia.org/therapy/diag/thyroid-disorders)

#### Turner syndrome
Turner syndrome is a genetic disorder in which a female is partly or completely missing one X chromosome that results in ovarian dysgenesis.
Learn more at: [www.fertilitypedia.org/therapy/diag/turner-syndrome](http://www.fertilitypedia.org/therapy/diag/turner-syndrome)

#### Repeated implantation failure
The absence of implantation after three or more transfers of high quality embryos or after placement of 10 or more embryos in multiple transfers.
Learn more at: [www.fertilitypedia.org/therapy/diag/repeated-implantation-failure](http://www.fertilitypedia.org/therapy/diag/repeated-implantation-failure)

#### Antiphospholipid syndrome
A condition when immune system mistakenly attacks some of the standard proteins in blood.
Learn more at: [www.fertilitypedia.org/therapy/diag/antiphospholipid-syndrome-do-rf](http://www.fertilitypedia.org/therapy/diag/antiphospholipid-syndrome-do-rf)

#### Premature ovarian failure
The loss of function of the ovaries before age 40.
Learn more at: [www.fertilitypedia.org/therapy/diag/premature-ovarian-failure](http://www.fertilitypedia.org/therapy/diag/premature-ovarian-failure)

#### Menopause
The time in most women’s lives when menstrual periods stop permanently, and the woman is no longer able to have children.
Learn more at: [www.fertilitypedia.org/therapy/diag/menopause](http://www.fertilitypedia.org/therapy/diag/menopause)

#### Sperm autoantibodies
Antibodies that bind to sperm, inhibiting their movement, stopping recognition and entry into the egg.
Learn more at: [www.fertilitypedia.org/therapy/diag/sperm-autoantibodies](http://www.fertilitypedia.org/therapy/diag/sperm-autoantibodies)
Lupus erythematosus
Collection of autoimmune diseases in which the human immune system becomes hyperactive and attacks normal, healthy tissues.
Learn more at: www.fertilitypedia.org/therapy/diag/lupus-erythematosus

Hyperthyroidism
Condition that occurs due to excessive production of thyroid hormone by the thyroid gland.
Learn more at: www.fertilitypedia.org/therapy/diag/hyperthyroidism

Reproductive functions

Endometrial receptivity
Period when the womb is receptive for implantation of the free-lying blastocyst.
Learn more at: www.fertilitypedia.org/edu/reproductive-functions/endometrial-receptivity

Implantation
The very early stage of pregnancy at which the embryo adheres to the wall of the uterus.
Learn more at: www.fertilitypedia.org/edu/reproductive-functions/implantation

Therapies

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

Sperm donation
The procedure in which a man (sperm donor) provides his sperm for fertility treatment.
Learn more at: www.fertilitypedia.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

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