Autoimmune polyglandular syndromes

Polyglandular syndrome is a multiple damage of endocrine glands. There are two groups of polyglandular syndromes: autoimmune polyglandular syndrome (APS) and multiple endocrine neoplasia syndrome (MEN). In autoimmune syndrome, the glands become affected by autoimmune inflammation and the hypofunction develops. In multiple endocrine neoplasia syndrome, there is a co-occurrence of tumors in several endocrine glands. Autoimmune polyglandular syndrome is divided into APS-I and APS-II.

Autoimmune polyglandular syndrome type I

Epidemiology

It is a very rare disease. The incidence in men and women is the same.

Etiopathogenesis

It is an autosomal recessive inherited disease with a mutation in the AIRE gene. The AIRE gene encodes a transcription factor that appears to be important in inducing immunological tolerance.

Clinical picture

The syndrome is characterized by a triad:

- **chronic mucocutaneous candidosis**
- **autoimmune hypoparathyroidism**
- **peripheral adrenocortical insufficiency** (*Addison's disease*)

A necessary condition for diagnosis is the presence of two symptoms. A complete diagnostic triad occurs in about 57% of patients. In addition, patients may experience nail dystrophy, enamel hypoplasia, hypogonadism, malabsorption, alopecia, pernicious anemia, thyroditis, diabetes mellitus, and other symptoms.

The syndrome usually manifests itself in childhood. The most common combination is hypoparathyroidism and adrenocortical insufficiency. The clinical picture results from the deficiency of hormones of individual glands. Hypogonadism occurs in up to 60% of women, but only in 14% of men.

Diagnostics

The presence of at least two components from the diagnostic triad is required for the diagnosis of APS-I. Molecular-biological examination of the AIRE gene will provide confirmation.

Differential diagnostics

In addition to the differential diagnosis of individual components of the syndrome, we must distinguish between rarer complex syndromes such as Turner syndrome, DiGeorg syndrome, Kearns-Sayer syndrome, etc.

Therapy

We treat the individual components of the syndrome. It should be borne in mind that the treatment may interact.

Prognosis

The prognosis depends on the presence of individual components and the possibility of influencing them.

Autoimmune polyglandular syndrome type II

Autoimmune polyglandular syndrome type II is called "Schmidt syndrome".

Epidemiology

The prevalence of the disease is 14-20 cases per million people. APS-II is the most common autoimmune disease with associated endocrine glands. The incidence in women is up to 4 times higher than in men.

Etiopathogenesis

It is a disease with polygenic inheritance. An association with some HLA antigens has been identified.

Clinical picture

The syndrome is characterized by a triad:

- **adrenocortical insufficiency**,
- **autoimmune thyroiditis** and
- **type 1 diabetes mellitus**.

The clinical picture is the same as for individual diseases. Addison's disease sometimes occurs with a delay against other components. In addition to this triad, hypogonadism, vitiligo, alopecia, pernicious anemia, myasthenia gravis, rheumatoid arthritis and Sjögren's syndrome are less common. Unlike APS-I, the syndrome manifests itself in adulthood, most often between the ages of 20 and 40.

Diagnostics

The condition for diagnosis is the presence of at least two components of the diagnostic triad, and **one of them must be Addison's disease**. The disease **cannot** be verified by molecular biology.

Differential diagnosis

In the first place, it is necessary to distinguish APS-I. Other differential diagnoses are quite broad and include Down syndrome, hemochromatosis, Kearns-Sayre syndrome, etc.

Therapy

We treat the individual components of the syndrome. If autoimmune thyroiditis is present, the presence of adrenocortical insufficiency should also be considered.

Treatment of thyroid hormones without previous substitution of missing corticoids worsens the patient's condition by suddenly increased metabolic demands!

Prognosis

The patient's prospects are most affected by the presence of type 1 DM. The other components of the syndrome are better controllable.

Links

Related links

- Addison´s Disease
- MEN syndrom

External links

Bibliography

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