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PERSPECTIVE

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Screening of Autoimmune Polyendocrine Syndromes and its Types, Risk factors and Causes

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Description

A heterogeneous group of rare diseases known as autoimmune polyendocrine syndromes (APSs), also known as polyglandular autoimmune syndromes (PGASs) or polyendocrine autoimmune syndromes (PASs), are characterised by autoimmune activity against multiple endocrine organs, though non-endocrine organs may also be affected. There are three different forms of APS, and endocrine autoimmunity is also present in a variety of other disorders.

Screening

In order to diagnose and treat the autoimmune polyendocrine syndrome (APS) early, screening is crucial. When an endocrine problem first manifests as APS, other endocrine and non-endocrine organs may not get involved for years or even decades. High clinical suspicion should be maintained in APS patients for the presence of additional autoimmune diseases:

• In APS type 1, it can take up to five years from the onset of mucocutaneous candidiasis to develop hypoparathyroidism, and it can take another ten years for adrenal gland involvement to develop. Therefore, patients with a single autoimmune endocrine illness must be treated with a high level of clinical suspicion.

•A patient should be screened for the presence of other auto-antibodies such as 21-hydroxylase, 17-hydroxylase, thyroid peroxidase, parietal cell, anti-intrinsic factor, and islet cell antibodies once they have been diagnosed with a single autoimmune endocrine disorder.

• Recent research has shown that autoantibodies can develop in APS at any age, and there is insufficient evidence to suggest an ideal interval between testing. For instance, celiac disease patients are frequently asymptomatic and are only identified after being tested for transglutaminase autoantibodies. Therefore, even if their first autoantibody tests are negative, people with a particular autoimmune ailment should be re-screened for autoantibodies for other autoimmune conditions at suitable intervals.

Types

• Type 1 autosomal recessive autoimmune polyendocrine syndrome which is characterised by hypoparathyroidism, adrenal insufficiency, hypogonadism, vitiligo, candidiasis, and other conditions.

• Type 2 autosomal dominant autoimmune polyendocrine syndrome which is characterised by hypothyroidism, type 1 diabetes, and adrenal insufficiency as a result of multifactorial gene involvement.

• The *FOXP3* gene mutation on the X chromosome causes the X-linked recessive immunodysregulation polyendocrinopathy enteropathy syndrome, or IPEX syndrome. Due to autoimmune activity against multiple organs the majority of patients develop diabetes and diarrhoea and many pass away. While girls are carriers and could get a slight illness boys are the ones that are impacted.

Risk factors

A known risk factor for autoimmune polyendocrine syndrome does not exist. Patients with a single autoimmune illness, however, are more likely to develop a second one. The list of autoimmune conditions that may enhance a patient's risk for developing autoimmune polyendocrine syndrome is shown below.

- Addison's illness
- Type 1 diabetes mellitus
- Autoimmune thyroiditis
- Hypogonadism (typically caused by autoimmune oophoritis),

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- Vitiligo
- Pernicious anaemia
- Chronic atrophic gastritis
- Chronic active hepatitis
- Idiopathic thrombocytopenic purpura
- Myasthenia gravis are some of the conditions that fall under this category.

Causes

A mutation in causes autoimmunity polyendocrine syndrome.

• **APS type I:** On chromosome 21, the *AIRE* (autoimmune regulator) gene is defective in people with APS type 1.

1. The genetic locus is located at 21p22.3 on the short arm (p) of chromosome 21.
2. The *AIRE* gene's typical function is to confer immunological tolerance for bodily antigens.
3. The loss of self-tolerance, which occurs when developing T lymphocytes with potential for responsiveness to self-antigens are wiped out during early differentiation in the thymus, is caused by the defective *AIRE* gene.
4. A Finnish study found that 82% of APS type 1 cases in Finland are caused by the mutation *R257X* (in the *AIRE* gene).

• **APS type 2:** APS type 2 has a complex inheritance pattern and is not a single gene condition.

1. The HLA locus is the genetic region with the highest risk for APS type 2. The low risk genes *CLTA4* and *PTPN22* are among others.
2. A mutation in the *FOXP3* gene on the X chromosome causes

• **APS type 3 or XPID:**

1. Chromosome Xp11.3-q13.3 contains the FOXP3 gene.
2. *FOXP3* is essential for CD4+CD25+ T regulatory cells to operate.