

# Rare Autoimmune Disease in Women

Autoimmune diseases occur when the body's immune system mistakenly identifies its own healthy cells as foreign and attacks them. There are more than 80 known autoimmune diseases, and roughly three out of five patients are women.<sup>1</sup> There are approximately 15 million people diagnosed with at least one autoimmune disease, and many of these are classified as rare diseases (diseases which affect fewer than 200,000 people).<sup>1</sup> Rare autoimmune disease research lags in understanding causes, treatments, and remission options.

Rare autoimmune diseases are often invisible, misunderstood, and delayed in diagnosis, especially for women.

## Rare Autoimmune Vasculitis & Hematologic Disorders

Vasculitis and hematologic disorders refer to disorders of the blood vessels, blood cells, plasma, or bone marrow.

### Cold Agglutinin Disease (CAD):

CAD is a rare autoimmune hemolytic anemia caused by specific cold-reacting antibodies. Normally, antibodies are protective proteins that find and neutralize harmful invaders like viruses and bacteria. In CAD, these antibodies mistakenly attack the body's own red blood cells when exposed to the cold. This causes red blood cells to clump together, leading them to be destroyed faster than the bone marrow can replace them and creating a shortage of red blood cells (anemia).<sup>2</sup> Approximately 1 in 300,000 individuals are diagnosed with CAD per year.<sup>3</sup> Symptoms include fatigue, jaundice, anemia, and Raynaud's phenomenon, worsening in cold environments. Disease management includes avoiding cold environments and newer emerging targeted therapies, such as complement inhibitors.



Rare autoimmune symptoms in women are too often minimized as stress, hormones, or “normal” aging, which can delay life-saving care.

### Evans Syndrome:

Evans syndrome is characterized by the simultaneous or sequential loss of both red blood cells (autoimmune hemolytic anemia) and platelets (immune thrombocytopenia). More common in women, it has an average onset at age 50.<sup>4</sup> The condition is frequently associated with lupus and other autoimmune disorders. Its tendency for disease relapse makes long-term management challenging, often requiring immunosuppressants and regular blood health monitoring.

Women are affected by CAD nearly twice as often as men.<sup>3</sup>



Rare autoimmune diseases can affect women across every stage of life, including menstruation, fertility, pregnancy, postpartum health, and menopause.

## Rare Dermatologic & Mucocutaneous Autoimmune Diseases

Dermatologic and mucocutaneous diseases refer to diseases of the skin, hair, and nails, as well as the mouth, genital, and anal areas (referred to as the mucous membranes).

### Lichen Sclerosus:

Lichen sclerosus is a chronic, non-contagious inflammatory skin disease that predominantly affects women, particularly in postmenopausal years.<sup>5</sup> The anogenital (genital and anus) region is commonly impacted, causing intense itching, white patches, and thin skin. It carries a small risk of squamous cell carcinoma (a form of skin cancer) at affected sites. Potent topical steroids, which are super-strength medicated ointments, remain the main treatment, and long-term monitoring is recommended.

### Autoimmune Progesterone Dermatitis (APD):

APD is a unique condition exclusive to women, triggered by the body's hypersensitivity to its own progesterone. Cyclical skin conditions, including eczema, urticaria (hives), and vesiculobullous lesions (fluid-filled blisters on the skin or mucous membranes), appear during the luteal phase of the menstrual cycle and resolve after menstruation. Diagnosis is made via intradermal progesterone testing. Management includes stopping ovulation or, in severe cases, oophorectomy (ovary removal surgery).<sup>6</sup>

Some rare autoimmune diseases, like APD, are uniquely tied to female hormones and reproductive cycles.

## Rare Endocrine & Metabolic Autoimmune Diseases

Endocrine and metabolic diseases refer to conditions linked to hormone imbalances or other chemical processes that relate to energy and growth.

### Addison's Disease (AD):

AD (also referred to as autoimmune adrenal insufficiency) results from the body's immune system attacking and destroying its own adrenal cortex (outer layer of the adrenal glands).<sup>7</sup> This leads to lowered cortisol and aldosterone production. Women are affected at nearly twice the rate of men.<sup>8</sup> Symptoms include chronic fatigue, weight loss, hypotension (low blood pressure), and characteristic hyperpigmentation of the skin. Adrenal crisis can be life-threatening. Lifelong hormone replacement therapy is necessary, and patients must be educated about stress management during illness or surgery.

Pregnancy and postpartum immune changes can increase risk for rare pituitary autoimmune disorders in autoimmune hypophysitis.

### Autoimmune Hypophysitis:

Autoimmune hypophysitis is an inflammatory condition of the pituitary gland that is three to eight times more common in females than males, depending on subtype.<sup>9</sup> Lymphocytic hypophysitis, the most common form, frequently occurs in late pregnancy and postpartum. Patients may present with headaches, visual disturbances, and varying degrees of hypopituitarism (when the pituitary gland fails to produce sufficient amounts of one or more hormones). Diagnosis often requires an MRI showing pituitary enlargement. Management ranges from hormone replacement to steroids; rarely, surgical decompression is considered.<sup>10</sup>

## References

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