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SNPs That Affect Inflammation and Autoimmune Disease

A Practitioner's Quick Reference Guide

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Practitioner Education

What is a SNP?

A **SNP (Single Nucleotide Polymorphism)** is a common type of genetic variation that occurs in DNA when a single nucleotide (A, T, C, or G) is altered at a specific position in the genome. The amount of genetic variation—biochemical individuality—is about . 1 percent. This means that about one base pair out of every 1,000 will be different between any two individuals.

Key Points About SNPs:

- **Common Genetic Variants:** Unlike rare genetic mutations, SNPs are widespread and contribute to individual differences in health, traits, and disease susceptibility.
- **Influence on Gene Function:** Some SNPs have no impact, while others can affect how a gene functions, influencing metabolism, detoxification, inflammation, hormone balance, and more.
- **Impact on Health:** SNPs can affect how individuals respond to nutrients, medications, and environmental factors, making them useful in personalised medicine and functional genomics.
- **Location Matters:**
  - **In Coding Regions:** Can alter protein structure and function.
  - **In Regulatory Regions:** May influence gene expression.
  - **In Non-Coding Regions:** Can affect RNA processing or gene regulation.

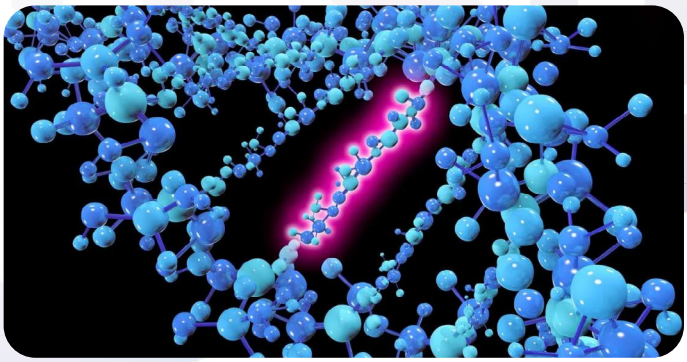
Example in Functional Genomics:

- The **MTHFR C677T SNP** affects the MTHFR enzyme, reducing its ability to convert folate into its active form, impacting methylation and homocysteine metabolism.
- The **COMT SNP (rs4680)** influences dopamine breakdown, affecting mood, stress resilience, and cognitive function.

Why SNPs Matter in Clinical Practice:

- **Nutrigenomics:** SNPs help determine personalised nutritional needs.
- **Pharmacogenomics:** They guide medication selection and dosing.
- **Functional Medicine:** SNPs provide insight into disease risk and health optimisation.

Understanding **SNPs (Single Nucleotide Polymorphisms)** allows health professionals to personalise treatment plans by identifying genetic variations that influence metabolism, detoxification, inflammation, neurotransmitter function, and nutrient processing. By analysing SNPs, practitioners can tailor dietary, lifestyle, and supplement recommendations to optimise health outcomes, improve patient response to treatments, and reduce the risk of chronic disease. This precision approach, known as functional genomics, enables practitioners to move beyond a one-size-fits-all model, offering more targeted and effective interventions based on an individual's unique genetic blueprint.



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SNPs To Assess For Autoimmune and Inflammation

Name	Function	Impact of Variation

(Il-6) Interleukin-6	IL-6 (Interleukin-6) is a **key cytokine** involved in both **protective immune responses** and chronic inflammatory diseases. It signals through the **IL-6 receptor (IL-6R)** and plays a dual role in **immune defence and pathology.**
	
	Physiological (Beneficial) Roles:
	• **Immune Defence:** Helps combat **infections** by promoting **inflammation and acute-phase responses.**
	• **Tissue Regeneration:** Supports **wound healing** and **organ development.**
	• **Immunoregulation:** Helps balance **pro- and anti-inflammatory responses** in controlled immune reactions.
	• **Tumour Suppression:** Can inhibit **tumorigenesis** under normal regulatory conditions.
	
	Pathogenic (Harmful) Roles in Autoimmunity and Chronic Disease:
	• **Chronic Inflammation & Autoimmunity:**
	  • **Elevated IL-6 SNPs** can drive **uncontrolled inflammation**, leading to **autoimmune diseases** like **rheumatoid arthritis, lupus, and Hashimoto's thyroiditis.**
	  • It **suppresses T-regulatory (Treg) cells**, reducing immune tolerance and increasing **autoimmune activation.**
	• **Increased CRP & Systemic Inflammation:**
	  • IL-6 induces **CRP and hepcidin**, contributing to **chronic inflammation, oxidative stress, and iron dysregulation.**
	• **Tissue Damage & Pain Sensitisation:**
	  • Promotes tissue degeneration and **hypernociception (increased pain sensitivity)** in inflammatory conditions.
	• **Metabolic Dysfunction & Cardiovascular Risk:**
	  • Linked to **insulin resistance**, **elevated homocysteine (Hcy), and increased stroke risk.**
	  • Disrupts **lipid metabolism**, promoting **atherogenesis and cardiovascular disease.**
		CC variant is decreased activity
		
		GG variant is increased activity

(TNFa) Tumour Necrosis Factor alpha	TNFa plays a major defence against pathogens.
	• **Beneficial roles:** development of lymphoid organ architecture, tissue regeneration, immunoregulation, inhibition of tumorigenesis.
	• **Pathogenic effect:** inflammation, autoimmunity, tissue destruction, hyper nociception (peripheral neuronal sensitisation), tumorigenesis, atherogenesis. Disrupts both micro and macrovascular circulation, lipid metabolism.
	• **Increased TNF-α activity due to SNP** variations can contribute to **chronic inflammation and autoimmunity.** Elevated TNF-α can:
	  • **Trigger and sustain autoimmune diseases** such as **rheumatoid arthritis, inflammatory bowel disease, psoriasis, and Hashimoto's thyroiditis** by **overactivating immune responses.**
	  • **Induce tissue destruction**, particularly in **joints, gut lining, and thyroid tissue,** leading to progressive autoimmune damage.
	  • **Increase pain sensitivity (hypernociception)** by amplifying **peripheral neuronal sensitisation,** which is often seen in chronic inflammatory conditions.
	  • **Contribute to metabolic and cardiovascular dysfunction** by disrupting **micro- and macrovascular circulation**, promoting **atherogenesis**, and altering **lipid metabolism**, increasing the risk of **insulin resistance and cardiovascular disease.**
		Increased Activity

(CRP) C-Reactive Protein	**CRP (C-Reactive Protein)** plays a key role in the **systemic inflammatory response** and is primarily regulated by **IL-6**, a pro-inflammatory cytokine. **Increased CRP SNPs**, which lead to higher baseline CRP levels, can contribute to **chronic low-grade inflammation**, a key driver of **autoimmune disease progression**. **Elevated CRP can:** • **Amplify the inflammatory cascade** by promoting cytokine release, exacerbating immune dysregulation in autoimmune conditions such as **Hashimoto's, rheumatoid arthritis, and lupus.** • **Affect immune tolerance**, potentially triggering or sustaining an overactive immune response. • **Influence energy metabolism and insulin resistance**, as CRP interacts with **leptin and hypothalamic signalling**, which can impact weight regulation and metabolic health—factors linked to autoimmunity. **Disrupt gut barrier integrity**, increasing the risk of **intestinal permeability ("leaky gut")**, which is a well-known factor in autoimmune pathogenesis.	Increased Activity

(IL-10) Iinterleukin-10	**IL-10 (Interleukin-10)** is a key **anti-inflammatory cytokine** that plays a crucial role in **immune regulation and inflammation control.** It primarily acts to **suppress pro-inflammatory cytokines** such as **IL-6**, **TNF**-α, and **IFN**-γ, helping to maintain immune balance and prevent excessive immune activation.
	
	Key Functions of IL-10:
	
	• **Suppresses Inflammation:** IL-10 **inhibits the production of inflammatory cytokines**, reducing excessive immune responses that can contribute to **autoimmune and inflammatory diseases.**
	• **Regulates Immune Tolerance:** It promotes the activity of **regulatory T cells (Tregs)**, helping to prevent **autoimmune reactions and unnecessary immune activation.**
	• **Protects Against Autoimmune Damage:** Adequate IL-10 levels help **prevent tissue destruction** caused by chronic inflammation.
	• **Supports Gut and Mucosal Immunity:** IL-10 helps maintain gut homeostasis, protecting against **inflammatory bowel diseases (IBD) and gut dysbiosis.**
	
	Pathogenic Effects of IL-10 SNPs:
	
	• **Reduced IL-10 Expression** (due to SNP variations) is linked to:
	  • **Increased autoimmunity** (e.g., rheumatoid arthritis, lupus, Hashimoto's, IBD).
	  • **Excessive inflammation and tissue damage** in chronic inflammatory conditions.
	  • **Heightened immune responses** that can lead to prolonged or exaggerated disease states.
	  • **Elevated IL-10 Expression** can sometimes lead to **immune suppression**, making the body more vulnerable to **chronic infections and reduced tumour surveillance.**
		Reduced Activity

Vitamin D Metabolism DHCR7 CYP2R1 VDR-FOK1 CYP24A1	Several key enzymes regulate **vitamin D metabolism**, and **genetic SNPs** affecting these enzymes can lead to **reduced vitamin D activation, transport, or receptor function**, increasing the risk of **autoimmune diseases.** **1. CYP2R1 (Vitamin D 25-Hydroxylase) – Liver Conversion** • **Function:** Converts **vitamin D3 (cholecalciferol) → 25(OH)D (calcidiol)** in the liver. **2. CYP27B1 (1-Alpha Hydroxylase) – Kidney Activation** • **Function:** Converts **25(OH)D → 1,25(OH)2D (calcitriol, the active form of vitamin D)** in the kidneys. **3. VDR (Vitamin D Receptor) – Cellular Action** • **Function:** Binds active **1,25(OH)2D**, allowing it to regulate gene expression in immune cells. **4. GC (Vitamin D Binding Protein) – Transport** • **Function:** Binds and transports **vitamin D metabolites** in the bloodstream. **Overall Impact on Autoimmunity** Vitamin D SNPs impair its activation, transport, or receptor function, leading to **chronic low-grade inflammation, poor immune tolerance, and increased risk of autoimmunity.**	Decreased Activity
BCOM1 (Beta Carotene Monooxygenase 1)	Codes for an enzyme that converts beta carotene into retinal. Impaired BCOM1 activity due to genetic variations can lead to reduced vitamin A availability, affecting **immune regulation, inflammation, and mucosal barrier integrity**—all critical factors in autoimmune diseases. In thyroid health, adequate vitamin A levels are necessary for **thyroid hormone receptor function, T3 activation, and modulating immune responses**, helping to balance autoimmune activity in conditions like **Hashimoto's and Graves' disease.** Individuals with BCOM1 SNPs may require **preformed vitamin A (retinol) instead of relying on beta-carotene conversion** to maintain optimal levels.	Decreased Activity
(FUT2) Galactoside 2-alpha-L-Fucosyltransferase 2	Involved in the production of secretor-status antigens, which influence **gut microbiome composition, immune function, and vitamin absorption.** It determines whether an individual is a **secretor (expressing blood group antigens in mucus and bodily fluids) or a non-secretor** (lacking these antigens). **Function of FUT2** • Produces **fucosylated glycans** in the gut lining, supporting the growth of **beneficial gut bacteria**, such as **Bifidobacteria.** • Plays a role in **mucosal immunity**, reducing pathogen adherence to the gut lining. • Affects **vitamin B12 absorption** via interactions with intrinsic factor.	

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