

DISEASE	GENE
Spondyloenchondrodyplasia with immune dysregulation	<b>ACP5</b>
Inflammatory skin and bowel disease, neonatal, 1	<b>ADAM17</b>
Dyschromatosis symmetrica hereditaria AND Aicardi-Goutieres syndrome 6	<b>ADAR</b>
Vibratory urticaria	<b>ADGRE2</b>
New Autoinflammatory Phenotype	<b>AGBL3</b>
Singleton-Merten syndrome 2	<b>DDX58</b>
Type I interferon-mediated autoinflammation due to DNase II deficiency	<b>DNASE2</b>
Singleton-Merten syndrome 1 AND Aicardi-Goutieres syndrome 7	<b>IFIH1</b>
Familial Mediterranean fever	<b>MEFV</b>
Hyper-IgD Syndrome/ Mevalonic aciduria/Porokeratosis 3, multiple types	<b>MVK</b>
Tumor Necrosis Factor receptor-associated periodic syndrome	<b>TNFRSF1A, TNFRSF11A</b>
CINCA syndrome, Familial cold inflammatory syndrome 1, Keratoendothelitis fugax hereditaria, Muckle-Wells syndrome AND Deafness, autosomal dominant 34, with or without inflammation	<b>NLRP3 (CIAS1, NALP3)</b>
Familial cold autoinflammatory syndrome 2	<b>NLRP12 (NALP12)</b>
Hydatidiform mole, recurrent, 1	<b>NLRP7</b>
Blau Syndrome	<b>NOD2 (CARD15)</b>
Pyogenic sterile arthritis, pyoderma gangrenosum, and acne	<b>PSTPIP1</b>
Majeed Syndrome	<b>LPIN2</b>

Interleukin 1 receptor antagonist deficiency	<b>IL1RN</b>
Psoriasis 14, pustular	<b>IL36RN</b>
Proteasome-associated autoinflammatory syndromes	<b>PSMG2, PSMA3, PSMB4, PSMB8, PSMB9</b>
Cherubism	<b>SH3BP2</b>
H Syndrome	<b>SLC29A3</b>
STING-associated vasculopathy, infantile-onset (SAVI)	<b>TMEM173</b>
Autoinflammatory syndrome, familial, Behcet-like	<b>TNFAIP3</b>
Polyglucosan body myopathy 1 with or without immunodeficiency	<b>RBCK1</b>
Autoinflammation, panniculitis, and dermatosis syndrome	<b>OTULIN</b>
Autoinflammation with infantile enterocolitis AND Familial cold autoinflammatory syndrome 4	<b>NLRC4</b>
Autoinflammation, antibody deficiency, and immune dysregulation syndrome AND Familial cold autoinflammatory syndrome 3	<b>PLCG2</b>
Susceptibility to Psoriasis 15, pustular	<b>AP1S3</b>
Pityriasis rubra pilaris AND Psoriasis 2	<b>CARD14</b>
ADA2 Deficiency	<b>CECR1</b>
IL10/R/RB Deficiency	<b>IL10, IL10RA, IL10RB</b>
Autoinflammation with arthritis and dyskeratosis; Palmoplantar carcinoma, multiple self-healing AND Susceptibility to Vitiligo-associated multiple autoimmune disease	<b>NLRP1</b>
Proteasome-associated autoinflammatory syndrome 2 AND Keratosis linearis with ichthyosis congenita and sclerosing keratoderma	<b>POMP</b>
Autoimmune interstitial lung, joint, and kidney disease	<b>COPA</b>
Chilblain lupus, Aicardi-Goutieres syndrome 1, dominant and recessive AND Vasculopathy, retinal, with cerebral leukodystrophy	<b>TREX1</b>
Chilblain lupus 2 AND Aicardi-Goutieres syndrome 5	<b>SAMHD1</b>
Aicardi-Goutieres syndrome 4	<b>RNASEH2A</b>
Aicardi-Goutieres syndrome 2	<b>RNASEH2B</b>

Aicardi-Goutieres syndrome 3	<b>RNASEH2C</b>
Retinitis pigmentosa and erythrocytic microcytosis	<b>TRNT1</b>
Van Esch-O'Driscoll syndrome AND Pigmentary disorder, reticulate, with systemic manifestations, X-linked	<b>POLA1</b>
Pseudo-TORCH syndrome 2	<b>USP18</b>
Autoinflammatory periodic fever, immunodeficiency, and thrombocytopenia (PFIT)	<b>WDR1</b>
Autoinflammation associated with LUBAC deficiency	<b>SHARPIN/RNF31</b>
Amyloidosis, primary localized cutaneous, 1	<b>OSMR</b>
Multiplex Family with $\gamma$ -Secretase Spectrum of Autoinflammatory Skin Phenotypes	<b>NCSTN</b>