AUTOINFLAMMATORY ALLIANCE

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Genetic Testing and Other Helpful Diagnostic Resources for Autoinflammatory Diseases

Systemic Autoinflammatory Diseases Database

Edited and Co-authored by Autoinflammatory Alliance President Karen Durrant, RN BSN and leading experts on autoinflammatory diseases, this free online database describes the notable symptoms of currently known autoinflammatory syndromes. Since these diseases can be very similar and have overlapping symptoms, the chart can help narrow down which SAIDs to consider and can help guide further testing.

Website: Autoinflammatory-search.org

Eurofever Classification Criteria

This diagnostic questionnaire by the EUROFEVER project helps doctors evaluate the possibility that a patient may have CAPS, FMF, TRAPS, or HIDS/MKD based on symptoms, length of fever days, and family history.

Website: printo.it/eurofever/scoreCriteria.asp

Genetic Testing

Several labs in the U.S. have periodic fever syndrome genetic panels. Below are the commonly used panels. All use next generation DNA sequencing and are very accurate tests. In addition, the Genetic Testing Registry provides information on genetic tests available around the world.

The larger panels by MNG Laboratories, Blueprint Genetics, Prevention Genetics, Avantra Genetics, and Fulgent test for several fever syndromes and some other conditions that may have similar symptoms, recurring fevers, recurring symptoms, and/or involve cytokine dysregulation. Some genes included in these panels are related to immune deficiencies and GI disorders.

Often insurance will cover the cost of genetics, however if testing is denied, some of these labs offer payment plans, and/or patient assistance, and/or compassionate assistance. Contact the lab to find out more information on these programs.

Many genetic testing labs no longer report all variant or novel mutations found on the initial report provided to doctors. To know if any additional variant or novel mutations were found, doctors must make a separate request for this variant/novel report from the lab.

Website Links:

MNG Laboratories: https://mnglabs.com/fullpanel/uploads/files/mng_ngs319e_testreq_201508ngs319form.pdf

Avantra Genetics: http://avantragenetics.com/ (Avantra is a division of Courtagen: www.courtagen.com)

Fulgent: https://fulgentdiagnostics.com/test/Periodic-Fevers-Syndrome

Blueprint Genetics: http://immunology.blueprintgenetics.com/panels/autoinflammatory-syndrome-panel/

Prevention Genetics: www.preventiongenetics.com (Search for test name "Periodic Fever Syndromes Sequencing Panel)

ARUP: http://ltd.aruplab.com/Tests/Pub/2007370

GeneDx: www.genedx.com/test-catalog/available-tests/periodic-fever-syndromes-panel-7-genes/

Genetic Testing Registry: www.ncbi.nlm.nih.gov/gtr/

Autoinflammatory Genetic Panel Comparisons; Genes Tested by Lab

	MNG Laboratories	Avantra Genetics	Fulgent	Invitae	Blueprint Genetics	Prevention Genetics	ARUP Laboratories and GeneDx
Number of Genes	36	33	27	26	25	13	7
Cost (Pricing is as of May 2017. Contact the lab for updated pricing.)	$1995. Only bills institutions contracted with. Self-pay available.	$350 Self-pay only.	Pricing not listed on website. Will bill insurance. Patient assistance/ payment plans available.	Patient-pay price of $475 per clinical area. Will bill insurance. Copay assistance available.	$1400. Will bill insurance. Financial assistance available.	$1440. Will bill insurance.	Pricing not listed on website. Will bill insurance.
Test Name	Fever Syndromes	Autoinflammatory Panel	Periodic Fever/Autoinflammatory Disorders NGS Panel	Autoinflammatory Syndromes Panel	Autoinflammatory Syndrome Panel	Periodic Fever Syndromes Sequencing	Periodic Fever Syndromes Panel

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	MNG Laboratories	Avantra Genetics	Fulgent	Invitae	Blueprint Genetics	Prevention Genetics	ARUP Laboratories and GeneDx
						Panel	
Gene/Syndrome							
MEFV – Familial Mediterranean fever (FMF)	X	X	X	X	X	X	X
LPIN2 – Majeed syndrome	X	X	X	X	X	X	X
MVK – Mevalonate kinase deficiencies (MKD): HIDS and MA	X	X	X	X	X	X	X
NLRP3 – Cryopyrin associated periodic syndromes (CAPS): MWS, FCAS, and NOMID	X	X	X	X	X	X	X
PSTPIP1 – Pyogenic sterile arthritis, pyoderma gangrenosum, and acne syndrome (PAPA)	X	X	X	X	X	X	X
TNFRSF1A – Tumor necrosis factor (TNF)–associated periodic syndrome (TRAPS)	X	X	X	X	X	X	X
ELANE - Cyclic neutropenia	X		X	X	X	X	X
ABCB1 – Inflammatory bowel disease 13 and colchicine resistance		X					
ACP5 - Spondyloenchondrodysplasia with immune dysregulation					X		
ADAM17 - Inflammatory skin and bowel disease, neonatal 1				X			
Aicardi-Goutieres syndromes (AGS) - TREX1 (AGS1), RNASEH2B (AGS2), RNASEH2C (AGS3), RNASEH2A (AGS4), SAMHD1 (AGS5), ADAR (AGS6), and IFIH1 (AGS7)					X		
AP1S3 - Pustular psoriasis (PSORS15)		X	X				

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ATG16L1 – Increased risk for Crohn's disease		X					
CARD14 – Familial psoriasis (PSORS2)	X	X	X	X	X	X	
CECR1/ADA2 – Deficiency of adenosine deaminase 2 (DADA2), aka Fever with early onset stroke (FEOS)	X		X	X			
COPA - COPA syndrome, aka Autoimmune interstitial lung, joint, and kidney disease		X		X			
DDX58 - Singleton-Merten syndrome					X		
FOXP3 - IPEX syndrome	X						
GCH1 - Dopa-responsive dystonia, Tetrahydrobiopterin deficiency	X						
HAX1 - Severe congenital neutropenia			X				
HLA-C - Psoriasis susceptibility 1, Human immunodeficiency virus type 1, susceptibility to		X					
HTR1A - Periodic fever, menstrual cycle-dependent (PFMC)	X						
IL10, IL10RA, IL10RB – Early-onset inflammatory bowel disease (EO-IBD25, EO-IBD28, and EO-IBD with Il-10 deficiency)	X	X	X	X			
IL1RN – Deficiency of interleukin-1ß (IL-1ß) receptor antagonist (DIRA)	X	X	X	X	X		
IL23A – Risk for inflammatory demyelinating diseases		X					

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IL23R – Risk for: ankylosing spondylitis, Crohn's, psoriatic arthritis, ulcerative colitis, psoriasis		X					
IL36RN – Deficiency of interleukin-36-receptor antagonist (DITRA)	X	X	X	X	X	X	
IL6 – Risk for juvenile rheumatoid arthritis, type 1 diabetes, inflammatory bowel disease 1		X					
IRF5 – Risk for autoimmune disorders: lupus, rheumatoid arthritis, systemic scleroderma, ulcerative colitis		X					
IRGM – Risk for Crohn's disease		X					
ISG15 - Immunodeficiency 38 with basal ganglia calcification (IMD38)					X		
NCSTN – Hidradenitis suppurativa		X					
NEFL - Charcot-Marie-tooth disease	X						
NFAT5 - NFAT5 haploinsufficiency				X			
NLRC4 – NLRC4 Macrophage activation-like syndrome (NLRC4-MAS)		X	X	X			
NLRP7 - Recurrent hydatidiform mole	X		X				
NLRP12 – Familial cold autoinflammatory syndrome 2 (FCAS2)	X	X	X	X	X	X	
NOD2 – Blau syndrome	X	X	X	X	X	X	
ORAI1 - Myopathy, tubular	X						

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aggregate, 2 (TAM2), Immunodeficiency 9 (IMD9)							
PLCG2 – PLCG2-associated antibody deficiency & immune dysregulation (PLAID) or Familial atypical cold urticaria (FACU) or FCAS3 and APLAID	X	X	X	X	X		
PSENEN - Hidradenitis suppurativa		X					
PSMB8 – Chronic atypical neutrophilic dermatosis with lipodystrophy and elevated temperature syndrome (CANDLE)	X	X	X	X	X	X	
PTPN2 – Risk for ulcerative colitis		X					
RBCK1 - Polyglucosan body myopathy 1 with or without immunodeficiency (PGBM1), HOIL-1 deficiency	X		X	X			
SCO2 - Myopia 6 (MYP6), Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1 (CEMCOX1)	X						
SH3BP2 – Cherubism	X		X	X			
SLC19A3 - Biotin-thiamine-responsive basal ganglia disease	X						
SLC25A19 - Amish lethal microcephaly	X						
SLC29A3 – SLC29A3 Spectrum disorder, aka H. syndrome; Pigmented hypertrichosis with insulin-dependent diabetes mellitus (IDDM)	X		X	X			

	MNG Laboratories	Avantra Genetics	Fulgent	Invitae	Blueprint Genetics	Prevention Genetics	ARUP Laboratories and GeneDx
TLR3 - Human immunodeficiency virus type 1, susceptibility to	X						
TMEM173 – STING-associated vasculopathy with onset in infancy (SAVI)	X	X	X		X		
TNFAIP3 - Haploinsufficiency of A20 (HA20), aka Behcet-like familial autoinflammatory syndrome		X				X	
TNFRSF11A – TNFRSF11A-associated hereditary fever disease (TRAPS11)	X		X				
TPK1 - Thiamine metabolism dysfunction syndrome 5, episodic encephalopathy type (THMD5)	X						
TRAF3 - Herpes simplex encephalitis, susceptibility to	X						
TRAF3IP2 - Psoriasis susceptibility 13, Candidiasis, familial, 8		X					
TRNT1 - Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay (SIFD)	X			X			