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:

GeneDx XomeDxSlice Tool: https://www.genedx.com/xomedx-slice-tool/

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/

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Autoinflammatory Genetic Panel Comparisons; Genes Tested by Lab

	GeneDx XomeDxSliceTool	MNG Laboratories	Fulgent	Invitae	Blueprint Genetics	Prevention Genetics	ARUP Laboratories and GeneDx
Number of Genes	Up to 150. 150+ with review	36	27	26	25	13	7
Cost (Pricing is as of May 2017. Contact the lab for updated pricing.)	Price Unlisted. Compassionate care/payment plans available. Will bill insurance.	$1995. Only bills institutions contracted with. Self-pay available.	Price unlisted. 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.	Price unlisted. Will bill insurance.
Test Name	XomeDxSlice Tool	Fever Syndromes	Periodic Fever/Autoinflammatory Disorders NGS Panel	Autoinflammatory Syndromes Panel	Autoinflammatory Syndrome Panel	Periodic Fever Syndromes Sequencing Panel	Periodic Fever Syndromes Panel
Gene/Syndrome							
MEFV – Familial Mediterranean fever (FMF)	X-per doctor request	X	X	X	X	X	X
LPIN2 – Majeed syndrome	X-per doctor request	X	X	X	X	X	X
MVK – Mevalonate kinase deficiencies (MKD): HIDS and MA	X-per doctor request	X	X	X	X	X	X
NLRP3 – Cryopyrin associated periodic syndromes (CAPS): MWS, FCAS, and NOMID	X-per doctor request	X	X	X	X	X	X
PSTPIP1 – Pyogenic sterile arthritis, pyoderma gangrenosum, and acne syndrome (PAPA)	X-per doctor request	X	X	X	X	X	X
TNFRSF1A – Tumor necrosis factor (TNF)–associated periodic syndrome (TRAPS)	X-per doctor request	X	X	X	X	X	X
ELANE - Cyclic neutropenia	X-per doctor request	X	X	X	X	X	X

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ABCB1 – Inflammatory bowel disease 13 and colchicine resistance	X-per doctor request						
ACP5 - Spondyloenchondrodysplasia with immune dysregulation	X-per doctor request				X		
ADAM17 - Inflammatory skin and bowel disease, neonatal 1	X-per doctor request			X			
Aicardi-Goutieres syndromes (AGS) - TREX1 (AGS1), RNASEH2B (AGS2), RNASEH2C (AGS3), RNASEH2A (AGS4), SAMHD1 (AGS5), ADAR (AGS6), and IFIH1 (AGS7)	X-per doctor request				X		
AP1S3 - Pustular psoriasis (PSORS15)	X-per doctor request		X				
ATG16L1 – Increased risk for Crohn's disease	X-per doctor request						
CARD14 – Familial psoriasis (PSORS2)	X-per doctor request	X	X	X	X	X	
CECR1/ADA2 – Deficiency of adenosine deaminase 2 (DADA2), aka Fever with early onset stroke (FEOS)	X-per doctor request	X	X	X			
COPA - COPA syndrome, aka Autoimmune interstitial lung, joint, and kidney disease	X-per doctor request			X			
DDX58 - Singleton-Merten syndrome	X-per doctor request				X		
FOXP3 - IPEX syndrome	X-per doctor request	X					
GCH1 - Dopa-responsive dystonia, Tetrahydrobiopterin deficiency	X-per doctor request	X					
HAX1 - Severe congenital neutropenia	X-per doctor request		X				

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HLA-C - Psoriasis susceptibility 1, Human immunodeficiency virus type 1, susceptibility to	X-per doctor request						
HTR1A - Periodic fever, menstrual cycle-dependent (PFMC)	X-per doctor request	X					
IL10, IL10RA, IL10RB – Early-onset inflammatory bowel disease (EO-IBD25, EO-IBD28, and EO-IBD with Il-10 deficiency)	X-per doctor request	X	X	X			
IL1RN – Deficiency of interleukin-1ß (IL-1ß) receptor antagonist (DIRA)	X-per doctor request	X	X	X	X		
IL23A – Risk for inflammatory demyelinating diseases	X-per doctor request						
IL23R – Risk for: ankylosing spondylitis, Crohn's, psoriatic arthritis, ulcerative colitis, psoriasis	X-per doctor request						
IL36RN – Deficiency of interleukin-36-receptor antagonist (DITRA)	X-per doctor request	X	X	X	X	X	
IL6 – Risk for juvenile rheumatoid arthritis, type 1 diabetes, inflammatory bowel disease 1	X-per doctor request						
IRF5 – Risk for autoimmune disorders: lupus, rheumatoid arthritis, systemic scleroderma, ulcerative colitis	X-per doctor request						
IRGM – Risk for Crohn's disease	X-per doctor request						
ISG15 - Immunodeficiency 38 with basal ganglia calcification (IMD38)	X-per doctor request				X		

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NCSTN – Hidradenitis suppurativa	X-per doctor request						
NEFL - Charcot-Marie-tooth disease	X-per doctor request	X					
NFAT5 - NFAT5 haploinsufficiency	X-per doctor request			X			
NLRC4 – NLRC4 Macrophage activation-like syndrome (NLRC4-MAS)	X-per doctor request		X	X			
NLRP7 - Recurrent hydatidiform mole	X-per doctor request	X	X				
NLRP12 – Familial cold autoinflammatory syndrome 2 (FCAS2)	X-per doctor request	X	X	X	X	X	
NOD2 – Blau syndrome	X-per doctor request	X	X	X	X	X	
ORAI1 - Myopathy, tubular aggregate, 2 (TAM2), Immunodeficiency 9 (IMD9)	X-per doctor request	X					
PLCG2 – PLCG2-associated antibody deficiency & immune dysregulation (PLAID) or Familial atypical cold urticaria (FACU) or FCAS3 and APLAID	X-per doctor request	X	X	X	X		
PSENEN - Hidradenitis suppurativa	X-per doctor request						
PSMB8 – Chronic atypical neutrophilic dermatosis with lipodystrophy and elevated temperature syndrome (CANDLE)	X-per doctor request	X	X	X	X	X	
PTPN2 – Risk for ulcerative colitis	X-per doctor request						
RBCK1 - Polyglucosan body myopathy 1 with or without immunodeficiency (PGBM1),	X-per doctor request	X	X	X			

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HOIL-1 deficiency							
SCO2 - Myopia 6 (MYP6), Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1 (CEMCOX1)	X-per doctor request	X					
SH3BP2 – Cherubism	X-per doctor request	X	X	X			
SLC19A3 - Biotin-thiamine-responsive basal ganglia disease	X-per doctor request	X					
SLC25A19 - Amish lethal microcephaly	X-per doctor request	X					
SLC29A3 – SLC29A3 Spectrum disorder, aka H. syndrome; Pigmented hypertrichosis with insulin-dependent diabetes mellitus (IDDM)	X-per doctor request	X	X	X			
TLR3 - Human immunodeficiency virus type 1, susceptibility to	X-per doctor request	X					
TMEM173 – STING-associated vasculopathy with onset in infancy (SAVI)	X-per doctor request	X	X		X		
TNFAIP3 - Haploinsufficiency of A20 (HA20), aka Behcet-like familial autoinflammatory syndrome	X-per doctor request					X	
TNFRSF11A – TNFRSF11A-associated hereditary fever disease (TRAPS11)	X-per doctor request	X	X				
TPK1 - Thiamine metabolism dysfunction syndrome 5, episodic encephalopathy type (THMD5)	X-per doctor request	X					
TRAF3 - Herpes simplex	X-per doctor	X					

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encephalitis, susceptibility to	request						
TRAF3IP2 - Psoriasis susceptibility 13, Candidiasis, familial, 8	X-per doctor request						
TRNT1 - Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay (SIFD)	X-per doctor request	X		X			

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