LBM/UMAI/0283 V1

Diagnostic des maladies auto-inflammatoires (Panel)										
GENETIQUE					**PHENOTYPE**					
Gène	**HGNC ID**	**OMIM**	**Locus**	**Transcrit RefSeq**	**Hérédité**	**Disease**	**Nom**	**OMIM**	**Aliases**	
ACP5	HGNC:124	171640	19p13.2	NM_001111035.2	Recessive	SPENCDI	Spondyloenchondrodysplasia With Immune Dysregulation	607944		
ADA2	HGNC:1839	607575	22q11.1	NM_001282225.1	Recessive	VAIHS	Vasculitis, Autoinflammation, Immunodeficiency, And Hematologic Defects Syndrome	615688	DADA2	Deficiency of adenosine deaminase 2
					Recessive	SNDNS	Sneddon Syndrome	182410		
					Recessive	PAN	Polyarteritis Nodosa, Childhood-Onset			
ADAM17	HGNC:195	603639	2p25.1	NM_003183	Recessive	NISBD	Inflammatory Skin And Bowel Disease, Neonatal	614328		
ADAR	HGNC:225	146920	1q21.3	NM_001111	Recessive	AGS6	Aicardi-Goutieres Syndrome 6	615010		
					Dominante	DSH	Dyschromatosis Symmetrica Hereditaria	127400		
AP1S3	HGNC:18971	615781	2q36.1	NM_001039569.1	Dominante	PSORS15	Psoriasis 15, Pustular, Susceptibility To	616106		
CARD14	HGNC:16446	607211	17q25.3	NM_024110.4	Dominante	PSOR2	Psoriasis 2	602723	CAMPS	CARD14-mediated pustular psoriasis
					Dominante	PRP	Pityriasis Rubra Pilaris	173200		
CASP1	HGNC:1499	147678	11q22.3	NM_001257118	Candidat					
COPA	HGNC:2230	601924	1q23.2	NM_001098398.1	Dominante	AILJK	Autoimmune Interstitial Lung, Joint, And Kidney Disease	616414		
DDX58	HGNC:19102	609631	9p21.1	NM_014314	Dominante	SGMRT2	Singleton-Merten Syndrome 2	616298		
FAS	HGNC:11920	134637	10q23.31	NM_000043.5	Dominante/ Recessive	ALPS	Autoimmune Lymphoproliferative Syndrome	601859		
FASLG	HGNC:11936	134638	1q24.3	NM_000639.2	Recessive	ALPS	Autoimmune Lymphoproliferative Syndrome	601859		
FBLIM1	HGNC:24686	607747	1p36.21	NM_017556.3	Candidat (AR)	CRMO	Chronic Recurrent Multifocal Osteomyelitis			
IFIH1	HGNC:18873	606951	2q24.2	NM_022168.3	Dominante	AGS7	Aicardi-Goutieres Syndrome 7	615846		
					Dominante	SGMRT1	Singleton-Merten Syndrome 1	182250		
IL10	HGNC:5962	124092	1q32.1	NM_000572.2	Candidat	IL10 deficiency				
IL10RA	HGNC:5964	146933	11q23.3	NM_001558.3	Recessive	IBD28	Inflammatory Bowel Disease 28, Autosomal Recessive	613148		

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IL10RB	HGNC:5965	123889	21q22.11	NM_000628.4	Recessive	IBD25	Inflammatory Bowel Disease 25, Autosomal Recessive	612567		
IL1RN	HGNC:6000	147679	2q14.1	NM_173841.2	Recessive	OMPP	Osteomyelitis, Sterile Multifocal, With Periostitis And Pustulosis	612852	DIRA	Deficiency of IL1 Receptor Antagonist
IL36RN	HGNC:15561	605507	2q14.1	NM_173170.1	Recessive	PSORP14	Pustular Psoriasis, Generalized	614204	DITRA	Deficiency of IL36 Receptor Antagonist
LACC1	HGNC:26789	613409	13q14.11	NM_001128303.1	Candidat (AR)	SJIA	Systemic Juvenile Inflammatory Arthritis			
LPIN2	HGNC:14450	605519	18p11.31	NM_014646.2	Recessive	MJDS	Majeed Syndrome	609628	LPIN2-CNO	LPIN2-Chronic non-bacterial osteomyelitis
LYN	HGNC:6735	165120	8q12.1	NM_002350.3	Candidat (AD)	LAID	Fever, Small Vessel Neutrophilic Vasculitis And Systemic Inflammation, Neonatal			
MDFIC	HGNC:28870	614511	7q31.1-q31.2	NM_199072.4	Candidat					
MEFV	HGNC:6998	608107	16p13.3	NM_000243.2	Recessive	FMF	Familial Mediterranean Fever	249100		
					Dominante	FMF	Familial Mediterranean Fever, Autosomal Dominant	134610		
					Dominante	PFAD	Periodic Fever With Autoinflammatory Disease			
					Dominante	PAAND	Pyrin-Associated Autoinflammation With Neutrophilic Dermatosis			
MVK	HGNC:7530	251170	12q24.11	NM_000431.3	Recessive	MEVA	Mevalonic Aciduria	610377	MKD	Severe Mevalonate kinase deficiency
						HIDS	Hyper-Igd Syndrome	260920	MKD	Mild Mevalonate kinase deficiency
NCSTN	HGNC:17091	605254	1q23.2	NM_015331.2	Dominante	ACNINV1	Acne Inversa, Familial, 1	142690	PASH	Pyoderma gangrenosum acne suppurative hidradenitis
NLRC4	HGNC:16412	606831	2p22.3	NM_021209.4	Dominante	AIFEC	Autoinflammation With Infantile Enterocolitis	616050		
						FCAS4	Familial Cold Autoinflammatory Syndrome 4	616115		
NLRP1	HGNC:14374	606636	17p13.2	NM_033004.3	Dominante/ Recessive	AIADK	Autoinflammation With Arthritis And Dyskeratosis;	617388	NAIAD	NLRP1-associated autoinflammation with arthritis and dyskeratosis
NLRP12	HGNC:22938	609648	19q13.42	NM_144687.3	Dominante	FCAS2	Familial Cold Autoinflammatory Syndrome 2	611762		
NLRP3	HGNC:16400	606416	1q44	NM_001243133.1	Dominante	CINCA	Cinca Syndrome	607115	NOMID	Neonatal onset multisystem inflammatory disease
									Severe NLRP3-AID	NLRP3-associated autoinflammatory disease (NLRP3-AID)
					Dominante	MWS	Muckle-Wells Syndrome	191900	Moderate NLRP3-AID	

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					Dominante	FCAS1	Familial Cold Autoinflammatory Syndrome 1	120100	Mild NLRP3-AID	
NOD2	HGNC:5331	605956	16q12.1	NM_022162.2	Dominante	Blau	Blau Syndrome	186580		NOD2-associated granulomatous disease
OTULIN	HGNC:25118	615712	5p15.2	NM_138348	Recessive	AIPDS	Autoinflammation, Panniculitis, And Dermatosis Syndrome	617099		
PLCG2	HGNC:9066	600220	16q23.3	NM_002661.4	Dominante	APLAID	Autoinflammation, Antibody Deficiency, And Immune Dysregulation, Plcg2-Associated	614878		
						FCAS3	Familial Cold Autoinflammatory Syndrome 3	614468	PLAID	PLCG2 associated antibody deficiency and immune dysregulation
POMP	HGNC:20330	613386	13q12.3	NM_015932.5	Recessive	KLICK	Keratosis Linearis With Ichthyosis Congenita And Sclerosing Keratoderma	601952		
					Dominante	PRAAS2	Proteasome-Associated Autoinflammatory Syndrome 2	618048	CANDLE	Chronic Atypical Neutrophilic Dermatosis with Lipodystrophy and Elevated temperature
PRF1	HGNC:9360	170280	10q22.1	NM_005041.5	Recessive	FHL2	Hemophagocytic Lymphohistiocytosis, Familial, 2	603553		
PSENEN	HGNC:30100	607632	19q13.12	NM_172341.2	Dominante	ACNINV2	Acne Inversa, Familial, 2	613736		
PSMA3	HGNC:9532	176843	14q23.1	NM_002788.3	Candidat (AR)	PRAAS				
PSMB4	HGNC:9541	602177	1q21.3	NM_002796.2	Recessive	PRAAS3	Proteasome-Associated Autoinflammatory Syndrome 3	617591	CANDLE	Chronic Atypical Neutrophilic Dermatosis with Lipodystrophy and Elevated temperature
PSMB8	HGNC:9545	177046	6p21.32	NM_148919.3	Recessive	PRAAS1	Proteasome-Associated Autoinflammatory Syndrome 1	256040	CANDLE	Chronic Atypical Neutrophilic Dermatosis with Lipodystrophy and Elevated temperature
PSMB9	HGNC:9546	177045	6p21.32	NM_002800.4	Recessive	PRAAS3	Proteasome-Associated Autoinflammatory Syndrome 3	617591	CANDLE	Chronic Atypical Neutrophilic Dermatosis with Lipodystrophy and Elevated temperature
PSMG2	HGNC:24929	609702	18p11.21	NM_020232.4	Candidat (AR)	PRAAS	Proteasome Associated Autoinflammatory Syndrome			
PSTPIP1	HGNC:9580	606347	15q24.3	NM_003978.4	Dominante	PAPA	Pyogenic Sterile Arthritis, Pyoderma Gangrenosum, And Acne	604416	PAPA	PSTPIP1-associated arthritis, pyoderma gangrenosum and acne
PYCARD	HGNC:16608	606838	16p11.2	NM_013258.4	Candidat					
RBCK1	HGNC:15864	610924	20p13	NM_031229.3	Recessive	PGBM1	Polyglucosan Body Myopathy 1 With Or Without Immunodeficiency	615895	PBMEI	Polyglucosan Body Myopathy, Early-Onset, With or Without Immunodeficiency
RIPK1	HGNC:10019	603453	6p25.2	NM_003804.5	Candidat (AD)	CRAF	Cleavage Resistant Ripk1 Associated Fever			
RNASEH2A	HGNC:18518	606034	19p13.13	NM_006397.2	Recessive	AGS4	Aicardi-Goutieres Syndrome 4	610333		
RNASEH2B	HGNC:25671	610326	13q14.3	NM_024570.3	Recessive	AGS2	Aicardi-Goutieres Syndrome 2	610181		
RNASEH2C	HGNC:24116	610330	11q13.1	NM_032193.3	Recessive	AGS3	Aicardi-Goutieres Syndrome 3	610329		

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RNF31	HGNC:16031	612487	14q12	NM_017999.4	Candidat (AR)	HOIP Deficiency				
SAMHD1	HGNC:15925	606754	20q11.23	NM_015474.3	Recessive	CHBL2	Chilblain Lupus 2	614415		
						AGS5	Aicardi-Goutieres Syndrome 5	612952		
SERPING1	HGNC:1228	606860	11q12.1	NM_000062.2	Dominante	HAE1	Angioedema, Hereditary, Type I	106100		
SH3BP2	HGNC:10825	602104	4p16.3	NM_003023.4	Dominante		Cherubism	118400	SDCM	SH3BP2 deficiency with multilocular cystic disease of the mandibles
SLC29A3	HGNC:23096	612373	10q22.1	NM_018344.5	Recessive	H syndrome	Histiocytosis-Lymphadenopathy Plus Syndrome	602782		
TMEM173	HGNC:27962	612374	5q31.2	NM_198282.3	Dominante	SAVI	Sting-Associated Vasculopathy, Infantile-Onset	615934		
TNFAIP3	HGNC:11896	191163	6q23.3	NM_001270508.1	Dominante	AISBL	Autoinflammatory Syndrome, Familial, Behcet-Like	616744	HA20	Deficiency in HA20 protein
TNFRSF11A	HGNC:11908	603499	18q21.33	NM_003839.3	Recessive	OPTB7	Osteopetrosis, Autosomal Recessive 7	612301	TRAPS11	TNFRSF11A-associated hereditary fever disease
					Dominante	FEO	Familial Expansile Osteolysis	174810		
TNFRSF1A	HGNC:11916	191190	12p13.31	NM_001065.3	Dominante		Periodic Fever, Familial, Autosomal Dominant	142680	TRAPS	TNF receptor-associated periodic fever syndrome
TNFRSF9	HGNC:11924	602250	1p36.23	NM_001561	Candidat					
TREX1	HGNC:12269	606609	3p21.31	NM_016381.5	Dominante/ Recessive	CHBL1	Chilblain Lupus 1	610448		
					Dominante	AGS1	Aicardi-Goutieres Syndrome 1	225750		
					Dominante	RVCL	Vasculopathy, Retinal, With Cerebral Leukodystrophy	192315		
TRNT1	HGNC:17341	612907	3p26.2	NM_182916.2	Recessive	RPEM	Retinitis Pigmentosa And Erythrocytic Microcytosis	616959		
					Recessive	SIFD	Sideroblastic Anemia With B-Cell Immunodeficiency, Periodic Fevers, And Developmental Delay	616084		
USP43	HGNC:20072		17p13.1	NM_153210.4	Candidat (AR)	PRAAS				
WDR1	HGNC:12754	604734	4p16.1	NM_017491.4	Candidat (AR)	PFIT	Autoinflammatory Periodic Fever, Immunodeficiency, And Thrombocytopenia			
XIAP	HGNC:592	300079	Xq25	NM_001167.3	XLR	XLP2	Lymphoproliferative Syndrome, X-Linked, 2	300635	X-linked HLD	