Diagnostic des maladies autoinflammatoires (Panel de 300 gènes)

ACP5	*ADA*	*ADA2*	*ADAM17*	*ADAR*	*ADGRE2*	*AICDA*	*AIM2*	*AIRE*	*ALPI*	*ALPK1*	*ALPL*	*AP1S3*	*AP3B1*	*ARHGEF6*
ARPC1B	*ATAD3A*	*B2M*	*BACH2*	*BTK*	*C1QA*	*C1QB*	*C1QC*	*C1R*	*C1S*	*C2*	*C3*	*CARD11*	*CARD14*	*CARD9*
CARMIL2	*CASP1*	*CASP10*	*CASP8*	*CD27*	*CD3E*	*CD3G*	*CD40*	*CD40LG*	*CD70*	*CDC42*	*CEBPE*	*CFH*	*COG6*	*COL1A1*
COL7A1	*COPA*	*CORO1A*	*CTLA4*	*CYBA*	*CYBB*	*CYBC1*	*DCLRE1C*	*DDX58*	*DEF6*	*DKC1*	*DNASE1*	*DNASE1L3*	*DNASE2*	*DOCK2*
DOCK8	*DPP8*	*DPP9*	*EGFR*	*ELANE*	*ELF4*	*ELP1*	*EPCAM*	*ERBIN*	*ERCC4*	*ERCC6L2*	*F12*	*FAAP24*	*FAS*	*FASLG*
FBLIM1	*FCN3*	*FDPS*	*FERMT1*	*FOXP3*	*G3BP1*	*G6PC3*	*GALNT3*	*GATA2*	*GBP5*	*GSDMD*	*GUCY2C*	*HAVCR2*	*HPS1*	*HPS4*
HPS6	*IARS1*	*ICOS*	*IFIH1*	*IKBIP*	*IKBKB*	*IKBKG*	*IKZF1*	*IL10*	*IL10RA*	*IL10RB*	*IL16*	*IL18*	*IL1RAPL1*	*IL1RN*
IL21	*IL21R*	*IL27RA*	*IL2RA*	*IL2RB*	*IL2RG*	*IL36RN*	*IL6R*	*IL7R*	*IRAK1*	*IRAK4*	*ISG15*	*ITCH*	*ITGB2*	*ITK*
JAK1	*JAK2*	*KPNA1*	*KPNA2*	*KRAS*	*LACC1*	*LCK*	*LIG4*	*LIPA*	*LPIN2*	*LRBA*	*LSM11*	*LYN*	*LYST*	*MALT1*
MAN2B1	*MASP2*	*MCM3AP*	*MCM9*	*MEFV*	*MLKL*	*MPO*	*MRTFA*	*MSN*	*MVD*	*MVK*	*MYD88*	*NAE1*	*NCF1*	*NCF2*
NCF4	*NCKAP1L*	*NCSTN*	*NFAT5*	*NFIL3*	*NFKB1*	*NFKB2*	*NFKBIA*	*NGLY1*	*NLRC3*	*NLRC4*	*NLRP1*	*NLRP12*	*NLRP13*	*NLRP3*
NLRP4	*NLRP7*	*NOD1*	*NOD2*	*NOX1*	*NRAS*	*NSMCE3*	*OAS1*	*ORAI1*	*OTUD7B*	*OTULIN*	*PDGFRB*	*PGM3*	*PIK3CD*	*PIK3R1*
PKN1	*PKN2*	*PLCG2*	*PMVK*	*PNPT1*	*POLA1*	*POLE*	*POMP*	*PRF1*	*PRKCD*	*PSEN1*	*PSENEN*	*PSMA3*	*PSMB10*	*PSMB4*
PSMB8	*PSMB9*	*PSMG2*	*PSTPIP1*	*PSTPIP2*	*PTPN22*	*PYCARD*	*RAB27A*	*RAC2*	*RAG1*	*RAG2*	*RASGRP1*	*RASGRP3*	*RBCK1*	*RC3H1*
RELA	*RELB*	*RHOG*	*RIPK1*	*RIPK3*	*RNASEH2A*	*RNASEH2B*	*RNASEH2C*	*RND1*	*RNF31*	*RTEL1*	*SAA1*	*SAE1*	*SAMD9*	*SAMD9L*
SAMHD1	*SERPING1*	*SH2D1A*	*SH3BP2*	*SHARPIN*	*SKIV2L*	*SLC11A1*	*SLC27A1*	*SLC27A6*	*SLC29A3*	*SLC37A4*	*SLC7A7*	*SLC9A3*	*SLCO2A1*	*SOCS1*
SPINK5	*SPINT2*	*STAT1*	*STAT2*	*STAT3*	*STAT4*	*STAT5B*	*STIM1*	*STING1*	*STK4*	*STX11*	*STXBP2*	*STXBP3*	*SYK*	*TAP1*
TAP2	*TAPBP*	*TBK1*	*TERT*	*TET2*	*TGFB1*	*TGFBR1*	*TGFBR2*	*TNFAIP3*	*TNFRSF11A*	*TNFRSF13B*	*TNFRSF13C*	*TNFRSF1A*	*TNFRSF9*	*TNFSF15*
TRAF3IP2	*TRAP1*	*TREM2*	*TREX1*	*TRIM21*	*TRIM22*	*TRNT1*	*TTC37*	*TTC7A*	*UBA1*	*UBA2*	*UBA3*	*UBA5*	*UBA6*	*UBA7*
UBAC1	*UBAC2*	*UNC13D*	*USP18*	*USP43*	*WAS*	*WASF2*	*WDR1*	*WIPF1*	*WNT2B*	*WNT6*	*XIAP*	*ZAP70*	*ZBTB24*	*ZNF341*

Gène	OMIM	Orphanet	Transcrit (RefSeq)	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
ACP5	171640	259358	NM_001111035.2	Spondyloenchondrodysplasia with immune dysregulation (SPENCDI)	Récessive
ADA	608958	117771	NM_000022.3	Adenosine deaminase deficiency	Récessive
ADA2	607575	396032	NM_001282225.1	Deficiency in Adenosine deaminase 2 (DADA2)	Récessive
ADAM17	603639	299456	NM_003183.6	Neonatal inflammatory skin and bowel disease	Récessive
ADAR	146920	119502	NM_001111.5	Aicardi-Goutieres syndrome, Dyschromatosis symmetrica hereditaria	Récessive/ Dominante
ADGRE2	606100		NM_013447.3	Vibratory urticaria	Dominante
AICDA	605257	119554	NM_020661.3	Immunodeficiency with hyper-IgM type 2	Récessive
AIM2	604578		NM_004833.2		Candidat
AIRE	607358	119562	NM_000383.3	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia	Récessive/ Dominante
ALPI	171740		NM_001631.4	Inflammatory bowel disease, Behçet-like	Récessive

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ALPK1	607347		NM_025144.3	Retinal dystrophy, Optic nerve edema, Splenomegaly, Anhidrosis and migraine Headache syndrome (ROSAH)	Dominante
ALPL	171760	119640	NM_000478.5	Hypophosphatasia and Chronic recurrent multifocal osteomyelitis	Récessive/ Dominante
AP1S3	615781	402648	NM_001039569.1	AP1 deficiency-mediated pustular psoriasis	Dominante
AP3B1	603401	121374	NM_003664.4	Hermansky-Pudlak syndrome	Récessive
ARHGEF6	300267	168322	NM_004840.2		Candidat
ARPC1B	604223		NM_005720.4	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease	Récessive
ATAD3A	612316	469968	NM_001170535.2	Type I interferonopathy	*Dominante*
B2M	109700	317771	NM_004048.3	MHC class I deficiency with cutaneous granulomas	*Récessive*
BACH2	605394		NM_021813.3	BACH2-related immunodeficiency and autoimmunity (BRIDA)	Dominante
BTK	300300	119094	NM_000061.2	Agammaglobulinemia Isolates growth hormone deficiency type III	Liée à l'X
C1QA	120550	221162	NM_015991.3	C1q deficiency, Monogenic SLE	Récessive
C1QB	120570	221166	NM_000491.4	C1q deficiency, Monogenic SLE	Récessive
C1QC	120575	221168	NM_001114101.2	C1q deficiency, Monogenic SLE	Récessive
C1R	613785	360362	NM_001733.4	Ehlers-Danlos syndrome, Monogenic SLE	Récessive
C1S	120580	221171	NM_201442.3	C1s deficiency, Ehlers-Danlos syndrome, Monogenic SLE	Récessive
C2	613927	119106	NM_000063.5	C2 deficiency, Monogenic SLE	Récessive
C3	120700	160064	NM_000064.3	C3 deficiency, Monogenic SLE	Récessive
CARD11	607210	333084	NM_032415.5	Severe combined immunodeficiency with atopic dermatitis and increased IgE, B cell expansion	Récessive/ Dominante
CARD14	607211	304694	NM_024110.4	CARD14-mediated psoriasis	Dominante
CARD9	607212	225286	NM_052813.4	Familial Candidiasis, Lyn deficiency-associated autoimmune and inflammatory diseases	Récessive

Gène	OMIM	Orphanet	Transcrit (RefSeq)	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
CARMIL2	610859	470980	NM_001013838.2	CARMIL2-Immunodeficiency with veo-IBD	Récessive
CASP1	147678	470036	NM_001257118.2		Candidat
CASP10	601762	119179	NM_032977.3	Autoimmune lymphoproliferative syndrome type II (ALPS)	Dominante
CASP8	601763	138851	NM_001228.4	Autoimmune lymphoproliferative syndrome with immunodeficiency	Récessive
CD27	186711	356847	NM_001242.4	Lymphoproliferative syndrome	Récessive
CD3E	186830	159200	NM_000733.3	Immunodeficiency, SCID-like	Récessive
CD3G	186740	201284	NM_000073.2	Immunodeficiency	Récessive
CD40	109535	119244	NM_001250.5	Immunodeficiency with hyper-IgM	Récessive
CD40LG	300386	119249	NM_000074.2	Immunodeficiency with hyper-IgM	Liée à l'X
CD70	602840	470052	NM_001252.4	Lymphoproliferative syndrome	Récessive
CDC42	116952	470056	NM_001791.3	Neonatal onset of pancytopenia, autoinflammation, rash, and episodes of hemophagocytic lymphohistiocytosis (NOCARH) / Takenouchi-Kosaki syndrome	Dominante
CEBPE	600749	183890	NM_001805.3	Noncanonical autoinflammatory inflammasomopathy with defective neutrophil function (CAIN)	Récessive
CFH	134370	119363	NM_000186.3	Complement factor H deficiency, Familial HLH	Récessive
COG6	606977	365113	NM_020751.2	Congenital disorder of glycosylation (CGD) type Iil	Récessive
COL1A1	120150	120704	NM_000088.3	Infantile cortical hyperostosis	Dominante
COL7A1	120120	120738	NM_000094.3	Dystrophic epidermolysis bullosa	Récessive/ Dominante
COPA	601924	448689	NM_001098398.1	Autoimmune interstitial lung, joint, and kidney disease	Dominante
CORO1A	605000	179462	NM_001193333.2	Immunodeficiency	Récessive
CTLA4	123890	120878	NM_005214.5	CTLA-4 haploinsufficiency with autoimmune infiltration (CHAI)	Dominante
CYBA	608508	138719	NM_000101.3	Chronic granulomatous disease	Récessive

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CYBB	300481	120935	NM_000397.3	Chronic granulomatous disease	Liée à l'X
CYBC1	618334		NM_001033046.3	Chronic granulomatous disease	Récessive
DCLRE1C	605988	121027	NM_001033855.2	Omenn Syndrome; Athabascan-type severe combined immunodeficiency	Récessive
DDX58	609631	422901	NM_014314.3	Singleton-Merten syndrome	Dominante
DEF6	610094		NM_022047.3	Immunodeficiency syndrome with systemic autoimmunity and aberrant CTLA-4 homeostasis	Récessive
DKC1	300126	121091	NM_001363.4	Dyskeratosis congenita	Liée à l'X
DNASE1	125505	353586	NM_005223.3	Monogenic systemic lupus erythematosus	Récessive/ Dominante
DNASE1L3	602244	291856	NM_004944.3	Monogenic systemic lupus erythematosus	Récessive/ Dominante
DNASE2	126350		NM_001375.2	Monogenic systemic lupus erythematosus	Récessive/ Dominante
DOCK2	603122	457458	NM_004946.3	Immunodeficiency	Récessive
DOCK8	611432	220922	NM_203447.3	Hyper-IgE recurrent infection syndrome (HIES)	Récessive
DPP8	606819		NM_130434.4		Candidat
DPP9	608258	353250	NM_139159.4	DPP9 deficiency (NLRP1-related inflammasomopathy)	Récessive
EGFR	131550	121311	NM_005228.4	Neonatal inflammatory skin and bowel disease 2	Récessive
ELANE	130130	121511	NM_001972.3	Cyclic neutropenia, severe congenital neutropenia	Dominante
ELF4	300775	306189	NM_001421.3	Deficiency in ELF4, X-linked (DEX)	*Liée à l'X*
ELP1	603722	122607	NM_003640.4	Dysautonomia, familial	Récessive
EPCAM	185535	226018	NM_002354.2	Congenital diarrhea with tufting enteropathy	Récessive
ERBIN	606944	470292	NM_001253697.1	ERBIN defiency	Dominante
ERCC4	133520	121595	NM_005236.2		Candidat

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ERCC6L2	615667	403281	NM_001010895.2	Bone marrow failure syndrome	Récessive
F12	610619	121663	NM_000505.3	Cold-induced Urticarial Autoinflammatory Syndrome Related to Factor XII Activation (FACAS)	Dominante
FAAP24	610884		NM_152266.4	EBV-driven lymphoproliferative disease	*Récessive*
FAS	134637	121733	NM_000043.5	Autoimmune lymphoproliferative syndrome (ALPS)	Récessive/ Dominante
FASLG	134638	121740	NM_000639.2	Autoimmune lymphoproliferative syndrome (ALPS)	Récessive/ Dominante
FBLIM1	607747		NM_017556.3	Monogenic CRMO	Récessive
FCN3	604973	332191	NM_003665.3	Immunodeficiency due to ficolin 3 deficiency	Récessive
FDPS	134629	444574	NM_002004.3	Porokeratosis	Dominante
FERMT1	607900	159172	NM_017671.4	Kindler syndrome	Récessive
FOXP3	300292	121913	NM_014009.3	Immunodysregulation, polyendocrinopathy and enteropathy (IPEX)	Liée à l'X
G3BP1	608431		NM_005754.2		Candidat
G6PC3	611045	171034	NM_138387.3	Severe Congenital neutropenia	Récessive
GALNT3	601756	122006	NM_004482.3	Hyperphosphatemic familial tumoral calcinosis with CRMO	Récessive
GATA2	137295	274222	NM_001145661.1	Immunodeficiency	Dominante
GBP5	611467		NM_052942.3		Candidat
GSDMD	617042		NM_024736.6		Candidat
GUCY2C	601330	317726	NM_004963.3	Familial Diarrhea	Dominante
HAVCR2	606652		NM_032782.4	T-cell lymphoma subcutaneous panniculitis-like	Récessive
HPS1	604982	122480	NM_000195.4	Hermansky-Pudlak syndrome	Récessive
HPS4	606682	122486	NM_022081.5	Hermansky-Pudlak syndrome	Récessive

Gène	OMIM	Orphanet	Transcrit (RefSeq)	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
HPS6	607522	122492	NM_024747.5	Hermansky-Pudlak syndrome	Récessive
IARS1	600709		NM_013417.3	Very early-onset IBD	Récessive
ICOS	604558	122565	NM_012092.3	Common variable immunodeficiency (CVID)	Récessive
IFIH1	606951	402570	NM_022168.4	Aicardi-Goutieres syndrome, Singleton-Merten syndrome	Dominante
IKBIP	609861		NM_153687.3		Candidat
IKBKB	300248	122614	NM_001556.3	Ectodermal dysplasia and immunodeficiency	Récessive/ Dominante
IKBKG	300248	122614	NM_003639.4	Ectodermal dysplasia and immunodeficiency NEMO deleted exon 5-autoinflammatory syndrome (NEMO-NDAS)	Liée à l'X
IKZF1	603023	318745	NM_006060.5	Common variable immunodeficiency (CVID), Monogenic SLE	Dominante
IL10	124092	122623	NM_000572.3	Early onset inflammatory bowel disease	Récessive
IL10RA	146933	244737	NM_001558.3	Early onset inflammatory bowel disease (IBD)	Récessive
IL10RB	123889	244742	NM_000628.4	Early onset inflammatory bowel disease (IBD)	Récessive
IL16	603035		NM_004513.5		Candidat
IL18	600953	470496	NM_001562.3		Candidat
IL1RAPL1	300206	159931	NM_014271.3		Candidat
IL1RN	147679	212902	NM_173842.2	Deficit of IL-1 receptor agonist (DIRA)	Récessive
IL21	605384	402379	NM_021803.4	IL-21 deficiency with early onset IBD	Récessive
IL21R	605383	357435	NM_021798.3	Immunodeficiency	Récessive
IL27RA	605350		NM_004843.3		Candidat
IL2RA	147730	160265	NM_000417.2	Immunodeficiency with lymphoproliferation and autoimmunity	Récessive
IL2RB	146710	355507	NM_000878.4	Immunodeficiency with lymphoproliferation and autoimmunity	*Récessive*

Gène	OMIM	Orphanet	Transcrit (RefSeq)	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
IL2RG	308380	122641	NM_000206.2	Severe Combined Immunodeficiency; Moderate Immunodeficiency	Liée à l'X
IL36RN	605507	281457	NM_173170.1	Deficit of IL-36 receptor agonist (DITRA)	Récessive
IL6R	147880		NM_000565.3	Hyper-IgE recurrent infection syndrome (HIES)	Récessive
IL7R	146661	159734	NM_002185.4	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type	Récessive
IRAK1	300283	369336	NM_001569.3		Candidat
IRAK4	606883	122674	NM_001114182.2	Immunodeficiency	Récessive
ISG15	147571	315470	NM_005101.4	Mendelian susceptibility to mycobacterial disease with basal ganglia calcification (MSMD)	Récessive
ITCH	606409	232245	NM_031483.7	Autoimmune disease, multisystem with facial dysmorphism	Récessive
ITGB2	600065	122698	NM_000211.4	Leukocyte adhesion deficiency (LAD)	Récessive
ITK	186973	244711	NM_005546.3	Lymphoproliferative syndrome	Récessive
JAK1	147795		NM_002227.3	Autoinflammation, immune dysregulation and eosinophilia	Dominante
JAK2	147796	122727	NM_004972.3		Candidat
KPNA1	600686	470578	NM_002264.3		Candidat
KPNA2	600685		NM_002266.3		Candidat
KRAS	19007	122879	NM_033360.3	RAS-associated autoimmune leukoproliferative disease Lymphoproliferative RAS-MAPK signaling interferonopathy	*Somatique*
LACC1	613409	454001	NM_001128303.2	LACC1 deficiency, Juvenile arthritis	Récessive
LCK	153390	280151	NM_001042771.2	Immunodeficiency	Récessive
LIG4	601837	123058	NM_002312.3	LIG4 Syndrome	Récessive
LIPA	613497	123063	NM_001127605.2	Wolman disease (HLH)	Récessive
LPIN2	605519	123109	NM_014646.2	Majeed syndrome	Récessive

Gène	OMIM	Orphanet	Transcrit (RefSeq)	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
LRBA	606453	303168	NM_006726.4	Common variable immunodeficiency (CVID) with autoimmunity	Récessive
LSM11	617910		NM_173491.3	Aicardi-Goutieres syndrome	Récessive
LYN	165120		NM_002350.3	LYN associated autoinflammatory disease (LAID)	Candidat (Dominante)
LYST	606897	123318	NM_000081.3	Chediak-Higashi syndrome	Récessive
MALT1	604860	159756	NM_006785.4	Immunodeficiency	Récessive
MAN2B1	609458	123328	NM_000528.3	Metabolic IFNpathy	Candidat (Récessive)
MASP2	605102	332185	NM_006610.3	MASP2 deficiency	Récessive
MCM3AP	603294		NM_003906.4		Candidat
MCM9	610098	448630	NM_017696.2		Candidat
MEFV	608107	123191	NM_000243.2	Pyrin-associated autoinflammatory diseases (PAAD) including Familial Mediterranean fever (FMF) and Pyrin-associated autoinflammation with neutrophilic dermatosis (PAAND)	Récessive/ Dominante
MLKL	615153		NM_152649.3	PMID: 32561755	Candidat
MPO	606989	123468	NM_000250.1	Neutrophilic pustular skin disease	Candidat (Récessive)
MRTFA	606078	409876	NM_020831.4	Immunodeficiency	Récessive
MSN	309845	470694	NM_002444.2	Immunodeficiency	Liée à l'X
MVD	603236	444568	NM_002461.2	Porokeratosis	Récessive/ Dominante
MVK	251170	123588	NM_000431.3	Mevalonate kinase deficiency (MKD), Porokeratosis	Récessive/ Dominante
MYD88	602170	201114	NM_001172567.1	Immunodeficiency	Récessive
NAE1	603385		NM_003905.3	Groupe des Ubiquitin like modifier activating enzymes (UBA)	Candidat
NCF1	608512	123695	NM_000265.5	Chronic granulomatous disease	Récessive
NCF2	608515	123701	NM_000433.3	Chronic granulomatous disease	Récessive

Gène	OMIM	Orphanet	Transcrit (RefSeq)	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
NCF4	601488	204415	NM_000631.4	Chronic granulomatous disease	Récessive
NCKAP1L	141180		NM_005337.4	Immunodeficiency with autoinflammation	Récessive
NCSTN	605254	268347	NM_015331.2	Hidradenitis suppurativa (HS)	Dominante
NFAT5	604708	470770	NM_138714.3	NFAT5 deficiency	Dominante
NFIL3	605327		NM_005384.2	NFIL3 deficiency	Récessive
NFKB1	164011	438054	NM_003998.3	Common variable immunodeficiency (CVID)	Dominante
NFKB2	164012	371170	NM_001077494.3	Common variable immunodeficiency (CVID)	Dominante
NFKBIA	164008	159788	NM_020529.2	Ectodermal dysplasia and immunodeficiency	Dominante
NGLY1	610661	406885	NM_018297.3	Congenital disorder of deglycosylation	Récessive
NLRC3	615648		NM_178844.3	PMID: 32447396	Candidat (Récessive)
NLRC4	605254	444736	NM_021209.4	Autoinflammation with infantile enterocolitis (AIFEC), Familial cold autoinflammatory syndrome (FCAS)	Dominante
NLRP1	606636	139184	NM_033004.3	NLRP1- associated autoinflammation with arthritis and dyskeratosis (NAIAD)	Récessive/ Dominante
NLRP12	609648	138361	NM_144687.3	Familial cold autoinflammatory syndrome (FCAS)	Dominante
NLRP13	609660		NM_176810.2		Candidat
NLRP3	606636	123821	NM_001243133.1	NLRP3-associated autoinflammatory disease	Dominante
NLRP4	609648		NM_134444.4	PMID: 35467709	Candidat
NLRP7	609661	123834	NM_001127255.1	Ulcerative colitis and hydatidiform mole	Dominante
NOD1	605980		NM_006092.3		Candidat
NOD2	605956	123845	NM_022162.2	Blau syndrome	Dominante
NOX1	300225		NM_007052.4	NOX1 deficiency	Dominante

Gène	OMIM	Orphanet	Transcrit (RefSeq)	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
NRAS	164790	221346	NM_002524.4	RAS-associated autoimmune leukoproliferative disease	Somatique
NSMCE3	608243		NM_138704.3	Lung disease, immunodeficiency, and chromosome breakage syndrome (LICS)	Récessive
OAS1	164350		NM_002534.3	OAS1-associated polymorphic autoinflammatory immunodeficiency (OPAID)	*Dominante*
ORAI1	610277	201295	NM_032790.3	Immunodeficiency due to ORAI1 deficiency	Récessive
OTUD7B	611748		NM_020205.3		Candidat
OTULIN	615712		NM_138348.5	OTULIN-related autoinflammatory syndrome (ORAS)	Récessive
PDGFRB	173410	138376	NM_002609.3	Penttinen syndrome	Dominante
PGM3	172100	446811	NM_001199917.1	PGM3-congenital disorders of glycosylation (CDG) with severe immunodeficiency	Récessive
PIK3CD	602839	400623	NM_005026.4	Immunodeficiency	Dominante
PIK3R1	171833	352217	NM_181523.2	Immunodeficiency	Récessive/ Dominante
PKN1	601032		NM_213560.1		Candidat
PKN2	602549		NM_006256.3		Candidat
PLCG2	600220	303819	NM_002661.4	Autoinflammation, antibody deficiency, and immune dysregulation syndrome (APLAID), Familial cold autoinflammatory syndrome (FCAS)	Dominante
PMVK	607622	444563	NM_006556.3	Porokeratosis	Dominante
PNPT1	610316	317560	NM_033109.4	Combined oxidative phosphorylation deficiency (Mitochondrial disease) / Deafness	Récessive
POLA1	312040	460832	NM_016937.3	X-linked pigmentary disorder, reticulate, with systemic manifestations	Liée à l'X
POLE	174762	356190	NM_006231.3	Facial dysmorphism-immunodeficiency-livedo-short stature syndrome (FILS)	Récessive
POMP	613386	281391	NM_015932.5	*POMP* -related autoinflammation and immune dysregulation disease (PRAID), *Proteasome-associated autoinflammatory syndrome (PRAAS)*	Dominante
PRF1	170280	117995	NM_005041.5	Familial hemophagocytic lymphohistiocytosis (HLH)	Récessive
PRKCD	176977	332077	NM_006254.3	Autoimmune lymphoproliferative syndrome (ALPS), type III / Monogenic SLE	Récessive

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PSEN1	104311	118099	NM_000021.3	Hidradenitis suppurativa (HS)	Dominante
PSENEN	607632	268351	NM_172341.3	Hidradenitis suppurativa (HS)	Dominante
PSMA3	176843	470916	NM_002788.3	Proteasome-associated autoinflammatory syndrome (PRAAS)	Récessive/ Dominante
PSMB10	176847		NM_002801.3	Proteasome-associated autoinflammatory syndrome (PRAAS)	Récessive/ Dominante
PSMB4	602177	470918	NM_002796.2	Proteasome-associated autoinflammatory syndrome (PRAAS)	Récessive/ Dominante
PSMB8	177046	258610	NM_148919.3	Proteasome-associated autoinflammatory syndrome (PRAAS)	Récessive/ Dominante
PSMB9	177045	470920	NM_002800.4	Proteasome-associated autoinflammatory syndrome (PRAAS)	Récessive/ Dominante
PSMG2	609702		NM_020232.4	Proteasome-associated autoinflammatory syndrome (PRAAS)	Récessive/ Dominante
PSTPIP1	606347	118114	NM_003978.4	*PSTPIP1* -associated autoinflammatory diseases (PAID)	Dominante
PSTPIP2	616046		NM_024430.3		Candidat
PTPN22	600716	317445	NM_015967.6		Candidat
PYCARD	606838		NM_013258.4		Candidat
RAB27A	603868	118188	NM_183235.2	Griscelli syndrome with HLH	Récessive
RAC2	602049	204411	NM_002872.4	Immunodeficiency with defective neutrophil chemotaxis	Récessive/ Dominante
RAG1	179615	118215	NM_000448.2	Combined cellular and humoral immune defects with granulomas	Récessive/ Dominante
RAG2	179616	118218	NM_000536.3	Combined cellular and humoral immune defects with granulomas	Récessive/ Dominante
RASGRP1	603962		NM_005739.3	Immunodeficiency	Récessive
RASGRP3	609531		NM_001139488.1		Candidat
RBCK1	610924	331658	NM_031229.3	Polyglucosan body myopathy with or without immunodeficiency (LUBAC deficiency)	Récessive
RC3H1	609424		NM_172071.4	Immune dysregulation and systemic hyperinflammation syndrome	Récessive

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RELA	164014	395954	NM_021975.3	Chronic mucocutaneous ulceration	Dominante
RELB	604758		NM_006509.3	Immunodeficiency	Récessive
RHOG	179505		NM_001665.3	RhoG deficiency	*Récessive*
RIPK1	603453		NM_003804.5	Cleavage resistant RIPK1-associated fever (CRAF), Immunodeficiency with autoinflammation	Récessive/ Dominante
RIPK3	605817		NM_006871.3		Candidat
RNASEH2A	606034	118329	NM_006397.2	Aicardi-Goutieres syndrome	Récessive
RNASEH2B	610326	118335	NM_024570.3	Aicardi-Goutieres syndrome	Récessive
RNASEH2C	610330	118340	NM_032193.3	Aicardi-Goutieres syndrome	Récessive
RND1	609038		NM_014470.3		Candidat
RNF31	612487	432144	NM_017999.4	HOIP deficiency (LUBAC deficiency)	Récessive
RTEL1	608833	330673	NM_016434.3	Pulmonary fibrosis and/or bone marrow failure	Dominante
SAA1	104750	330340	NM_001178006.2	Hereditary AA Amyloidosis	*Dominante*
SAE1	613294		NM_005500.2	Groupe des Ubiquitin like modifier activating enzymes (UBA)	Candidat
SAMD9	610456	118460	NM_017654.3	Myelodysplasia, infection, growth restriction, adrenal hypoplasia, genital phenotypes, and enteropathy (MIRAGE) syndrome, Inherited bone marrow failure	Dominante
SAMD9L	611170	461215	NM_152703.4	SAMD9L-associated autoinflammatory disease (SAMD9L-SAAD)	Dominante
SAMHD1	606754	201570	NM_015474.3	Aicardi-Goutieres syndrome	Récessive
SERPING1	606860	118618	NM_000062.2	Hereditary Angioedema	Récessive/ Dominante
SH2D1A	300490	118679	NM_002351.4	Lymphoproliferative syndrome	Liée à l'X
SH3BP2	602104	118689	NM_003023.4	Cherubism	Dominante
SHARPIN	611885	471052	NM_030974.3	LUBAC deficiency	Récessive

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SKIV2L	600478	299641	NM_006929.4	Trichohepatoenteric syndrome	Récessive
SLC11A1	600266	158401	NM_000578.3		Candidat
SLC27A1	600691		NM_198580.2	Merkelsson-Rosenthal syndrome	Dominante
SLC27A6	604196		NM_014031.4	PMID: 32447396	Candidat (Récessive)
SLC29A3	612373	173057	NM_018344.5	Pigmentary hypertrichosis and non-autoimmune insulin-dependent diabetes (H syndrome)	Récessive
SLC37A4	602671	119474	NM_001164277.1	Glycogen storage disease (GSD)	Récessive
SLC7A7	603593	119709	*NM_001126105.2*	Lysinuric protein intolerance with HLH	Récessive
SLC9A3	182307	456093	NM_004174.3	Congenital diarrhea with secretory sodium	Récessive
SLCO2A1	601460	291830	NM_005630.2	Primary hypertrophic osteoarthropathy	Récessive
SOCS1	603597		NM_003745.1	Early-onset autoimmunity associated with SOCS1 haploinsufficiency / Monogenic systemic lupus erythematosus	Dominante
SPINK5	605010	119840	NM_001127698.1	Netherton syndrome	Récessive
SPINT2	605124	178801	NM_021102.3	Congenital diarrhea with secretory sodium	Récessive
STAT1	600555	119879	NM_007315.3	Immunodeficiency	Dominante
STAT2	600556	434238	NM_005419.3	Immunodeficiency with HLH	Récessive
STAT3	102582	138523	NM_139276.2	Autoimmune disease, multisystem, infantile-onset	Dominante
STAT4	600558	320815	NM_003151.3		Candidat
STAT5B	604260	119882	NM_012448.3	Hypereosinophilic syndrome with urticarial rash	*Somatique*
STIM1	605921	201300	NM_003156.3	Immunodeficiency	Récessive
STING1	612374	438066	NM_198282.3	STING-associated vasculopathy, infantile-onset (SAVI)	Dominante
STK4	604965	317798	NM_006282.4	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations (TIIAC)	Récessive

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STX11	605014	119892	NM_003764.3	Familial hemophagocytic lymphohistiocytosis (HLH)	Récessive
STXBP2	601717	227102	NM_006949.3	Familial hemophagocytic lymphohistiocytosis (HLH)	Récessive
STXBP3	608339		NM_007269.3	Very Early Onset Inflammatory Bowel Disease, Bilateral Sensorineural Hearing Loss and Immune Dysregulation	*Récessive/ Dominante*
SYK	600085		NM_003177.6	SYK-associated autoinflammatory disease	*Dominante*
TAP1	170260	123369	NM_000593.5	MHC class I deficiency with vasculitis or pyoderma gangrenosum	*Récessive*
TAP2	170261	119922	NM_000544.3	MHC class I deficiency with vasculitis or pyoderma gangrenosum	*Récessive*
TAPBP	601962	123371	NM_003190.4	MHC class I deficiency with vasculitis or pyoderma gangrenosum	*Récessive*
TBK1	604834	318865	NM_013254.3	TBK1 deficiency autoinflammation driven by TNF-induced cell death	Récessive
TERT	187270	138451	NM_198253.2	pulmonary fibrosis and Høyeraal-Hreidarsson syndrome	Dominante
TET2	612839	268325	NM_001127208.2	Immunodeficiency	Récessive
TGFB1	190180	120047	NM_000660.6	Camurati-Engelman disease	Dominante
TGFBR1	190181	120065	NM_004612.3	Loeys-Dietz syndrome, type 1	Dominante
TGFBR2	190182	120069	NM_001024847.2	Loeys-Dietz syndrome, type 2	Dominante
TNFAIP3	191163	369146	NM_006290.3	Haploinsufficiency in A20 or Familial autoinflammatory syndrome (HA20), Behcet-like	Dominante
TNFRSF11A	603499	120160	NM_003839.3	TRAPS-like disease	Dominante
TNFRSF13B	604907	120169	NM_012452.2	TACI deficiency	Récessive/ Dominante
TNFRSF13C	606269	231494	NM_052945.3	Immunodeficiency	Récessive
TNFRSF1A	191190	120173	NM_001065.3	TNF-receptor associated periodic fever syndrome (TRAPS)	Dominante
TNFRSF9	602250	471244	NM_001561.5		Candidat
TNFSF15	604052	317398	NM_005118.3		Candidat

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TRAF3IP2	607043	370364	NM_147686.3	ACT1 deficiency, chronic mucocutaneous candidiasis	Récessive
TRAP1	606219		NM_016292.2	TRAP1 chaperone protein-associated autoinflammation	Récessive
TREM2	605086	120249	NM_018965.3	Familial hemophagocytic lymphohistiocytosis	Candidat
TREX1	606609	120255	NM_033629.5	Aicardi-Goutieres syndrome	Récessive/ Dominante
TRIM21	109092		NM_003141.3		Candidat
TRIM22	606559	471254	NM_006074.4	Granulomatous colitis and severe perianal disease	Récessive
TRNT1	612907	413615	NM_182916.2	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay (SIFD)	Récessive
TTC37	614589	238544	NM_014639.3	Trico hepato enteric syndrome	Récessive
TTC7A	609332	353460	NM_020458.3	Gastrointestinal defects and immunodeficiency syndrome	Récessive
UBA1	314370	140036	NM_003334.3	VEXAS syndrome	Somatique
UBA2	613295		NM_005499.2	Groupe des Ubiquitin like modifier activating enzymes (UBA)	Candidat
UBA3	603172		NM_003968.3	Groupe des Ubiquitin like modifier activating enzymes (UBA)	Candidat
UBA5	610552	469050	NM_024818.4	Groupe des Ubiquitin like modifier activating enzymes (UBA)	Candidat
UBA6	611361		NM_018227.5	Groupe des Ubiquitin like modifier activating enzymes (UBA)	Candidat
UBA7	191325		NM_003335.2	Groupe des Ubiquitin like modifier activating enzymes (UBA)	Candidat
UBAC1	608129		NM_016172.2	Groupe des Ubiquitin like modifier activating enzymes (UBA)	Candidat
UBAC2		285499	NM_001144072.1	Groupe des Ubiquitin like modifier activating enzymes (UBA)	Candidat
UNC13D	608897	120416	NM_199242.2	Familial hemophagocytic lymphohistiocytosis (HLH)	Récessive
USP18	607057		NM_017414.3	Pseudo-TORCH syndrome	Récessive
USP43			NM_153210.4	USP43-associated autoinflammatory disease (USP43-AID)	Récessive

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WAS	300392	120490	NM_000377.2	Wiskott-Aldrich syndrome	Liée à l'X
WASF2	605875		NM_006990.4		Candidat
WDR1	604734		NM_017491.4	Periodic fever, immunodeficiency, and thrombocytopenia syndrome (PFIT)	Récessive
WIPF1	602357	292448	NM_001077269.1	Wiskott-Aldrich syndrome	Récessive
WNT2B	601968		NM_024494.2	Diarrhea	Récessive
WNT6	604663		NM_006522.3		Candidat
XIAP	300079	138732	NM_001167.3	XIAP deficiency-associated lymphoproliferative syndrome	Liée à l'X
ZAP70	176947	120579	NM_001079.3	Immunodeficiency	Récessive
ZBTB24	614064	268103	NM_014797.2	Immunodeficiency-centromeric instability-facial anomalies syndrome	Récessive
ZNF341	618269		NM_032819.4	Hyper-IgE recurrent infection syndrome (HIES)	Récessive