Panel de gènes des maladies autoinflammatoires (260 gènes)

ABCG8	*ACP5*	*ADA2/CECR1*	*ADAM17*	*ADAR*	*AIM2*	*ALPI*	*ALPK1*	*ALPL*	*AP1S3*	*ARF1*	*ARPC1B*	*ARPC5*
ATAD3A	*B2M*	*BACH2*	*BNIP3*	*C1QA*	*C1QB*	*C1QC*	*C1R*	*C1S*	*C2*	*C2orf69*	*C3*	*CARD10*
CARD11	*CARD14*	*CARD9*	*CARMIL2*	*CASP1*	*CASP8*	*CCR2*	*CD27*	*CD3E*	*CD3G*	*CD40*	*CD40LG*	*CD70*
CDC42	*CEBPE*	*CFH*	*COPA*	*CORO1A*	*CTLA4*	*CYBA*	*CYBB*	*CYBC1*	*DEF6*	*DNASE1*	*DNASE1L3*	*DNASE2*
DOCK11	*DOCK2*	*DOCK8*	*DPP8*	*DPP9*	*EGFR*	*ELANE*	*ELF4*	*ELP1*	*ERBIN*	*F12*	*FAS*	*FASLG*
FBLIM1	*FBXW11*	*FDPS*	*FOXP3*	*G6PC3*	*GALNT3*	*GATA2*	*GBP5*	*GIMAP6*	*GSDMD*	*HAVCR2*	*HCK*	*HPS1*
HPS4	*HPS6*	*IFIH1*	*IKBKB*	*IKBKG*	*IKZF1*	*IKZF2*	*IL10*	*IL10RA*	*IL10RB*	*IL18*	*IL18BP*	*IL1R1*
IL1RN	*IL2RA*	*IL2RB*	*IL36RN*	*IL6R*	*IL6ST*	*IRAK1*	*IRAK4*	*ISG15*	*ITGB2*	*ITK*	*JAK1*	*LACC1*
LCK	*LCP2*	*LIG4*	*LPIN2*	*LRBA*	*LSM11*	*LYN*	*LYST*	*MAGT1*	*MALT1*	*MAN2B2*	*MCM10*	*MEFV*
MLKL	*MRTFA*	*MVD*	*MVK*	*MYD88*	*NCF1*	*NCF2*	*NCF4*	*NCKAP1L*	*NCSTN*	*NFAT5*	*NFIL3*	*NFKB1*
NFKB2	*NFKBIA*	*NLRC4*	*NLRP1*	*NLRP12*	*NLRP3*	*NLRP4*	*NLRP9*	*NOD2*	*NRAS*	*OAS1*	*ORAI1*	*OTUD7B*
OTULIN	*PDGFRB*	*PGM3*	*PIK3CG*	*PIK3R1*	*PLCG1*	*PLCG2*	*PMVK*	*PNPT1*	*POFUT1*	*POLA1*	*POMP*	*POU2F2*
PRF1	*PRKCD*	*PSEN1*	*PSENEN*	*PSMA3*	*PSMA5*	*PSMA6*	*PSMB10*	*PSMB4*	*PSMB8*	*PSMB9*	*PSMC3*	*PSMD12*
PSMG2	*PSTPIP1*	*PTPN22*	*PTPN6*	*PYCARD*	*RAB27A*	*RAC1*	*RAC2*	*RAG1*	*RAG2*	*RASGRP1*	*RBCK1*	*RC3H1*
RELA	*RELB*	*REXO2*	*RHBDF1*	*RHOG*	*RIGI/DDX58*	*RIPK1*	*RIPK3*	*RMND1*	*RNASEH2A*	*RNASEH2B*	*RNASEH2C*	*RNASET2*
RND1	*RNF213*	*RNF31*	*RNU7-1*	*SAA1*	*SAMD9*	*SAMD9L*	*SAMHD1*	*SBNO2*	*SH2D1A*	*SH3BP2*	*SHARPIN*	*SKIC2*
SLC12A2	*SLC29A3*	*SLC37A4*	*SLC39A4*	*SOCS1*	*SPINK5*	*STAT1*	*STAT2*	*STAT3*	*STAT4*	*STAT5B*	*STAT6*	*STIM1*
STING1/TMEM173	*STK4*	*STX11*	*STXBP2*	*STXBP3*	*SUPV3L1*	*SYK*	*TBK1*	*TET2*	*TGFB1*	*TLR4*	*TLR7*	*TLR8*
TNFAIP3	*TNFRSF11A*	*TNFRSF13B*	*TNFRSF13C*	*TNFRSF1A*	*TNFRSF9*	*TRAF3*	*TRAP1*	*TREX1*	*TRIM21*	*TRIM22*	*TRNT1*	*TTC37/SKIC3*
TTC7A	*UBA1*	*UNC13D*	*UNC93B1*	*USP18*	*USP43*	*WAS*	*WDR1*	*WIPF1*	*WNT6*	*XIAP*	*ZAP70*	*ZNF341*

Gène	OMIM	Orphanet	Transcrit GRCh37 (NCBI)*	Code couleur Nouveaux gènes Gènes candidats de MAI	Phénotype	Hérédité
ABCG8	605460		NM_022437.3		Sitosterolemia with arthritis and splenomegaly	Récessive
ACP5	171640	259358	NM_001111035.2		Spondyloenchondrodysplasia with immune dysregulation (SPENCDI)	Récessive
ADA2/ CECR1	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
AIM2	604578		NM_004833.2			Candidat
ALPI	171740		NM_001631.4		Inflammatory bowel disease, Behçet-like	Récessive
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

Gène	OMIM	Orphanet	Transcrit GRCh37 (NCBI)*	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
AP1S3	615781	402648	NM_001039569.1	AP1 deficiency-mediated pustular psoriasis	Dominante
ARF1	103180		NM_001658.4	Type I interferonopathy	Dominante
ARPC1B	604223		NM_005720.4	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease	Récessive
ARPC5	604227		NM_005717.4	ARPC5 deficiency	Récessive
ATAD3A	612316	469968	NM_001170535.2	Type I interferonopathy	Récessive/ 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
BNIP3	603293		NM_004052.4	EBV-driven lymphoproliferative disease	Récessive
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
C2orf69	619219		NM_153689.6	Combined oxidative phosphorylation deficiency with autoinflammation	Récessive
C3	120700	160064	NM_000064.3	C3 deficiency, Monogenic SLE	Récessive
CARD10	607209		NM_014550.4	CARD10 Deficiency with auto-immunity	Récessive
CARD11	607210	333084	NM_032415.5	Severe combined immunodeficiency with atopic dermatitis and increased IgE, B cell expansion with NFkB and T cell anergy (BENTA)	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 GRCh37 (NCBI)*	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
CASP8	601763	138851	NM_001228.4	Autoimmune lymphoproliferative syndrome with immunodeficiency	Récessive
CCR2	601267		NM_001123396.4	CCR2 deficiency	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
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
CYBB	300481	120935	NM_000397.3	Chronic granulomatous disease	Liée à l'X
CYBC1	618334		NM_001033046.3	Chronic granulomatous disease	Récessive
DEF6	610094		NM_022047.3	Immunodeficiency syndrome with systemic autoimmunity and aberrant CTLA-4 homeostasis, DEF6 Deficiency	Récessive

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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
DOCK11	300681		NM_144658.4	DOCK11 deficiency actinopathy and autoimmunity	Liée à l'X
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/ Dominante
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
ERBIN	606944	470292	NM_001253697.1	ERBIN defiency	Dominante
F12	610619	121663	NM_000505.3	Cold-induced Urticarial Autoinflammatory Syndrome Related to Factor XII Activation (FACAS)	Dominante
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
FBXW11	605651		NM_001378974.1	Recurrent periodic fever and severe headaches associated with *FBXW11* -LOF	Dominante
FDPS	134629	444574	NM_002004.3	Porokeratosis	Dominante
FOXP3	300292	121913	NM_014009.3	Immunodysregulation, polyendocrinopathy and enteropathy (IPEX)	Liée à l'X

Gène	OMIM	Orphanet	Transcrit GRCh37 (NCBI)*	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
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
GIMAP6	616960		NM_024711.6	GIMAP6 Deficiency	Récessive
GSDMD	617042		NM_024736.6		Candidat
HAVCR2	606652		NM_032782.4	T-cell lymphoma subcutaneous panniculitis-like	Récessive
HCK	142370		NM_002110.5	HCK-associated autoinflammatory disease	Dominante
HPS1	604982	122480	NM_000195.4	Hermansky-Pudlak syndrome	Récessive
HPS4	606682	122486	NM_022081.5	Hermansky-Pudlak syndrome	Récessive
HPS6	607522	122492	NM_024747.5	Hermansky-Pudlak syndrome	Récessive
IFIH1	606951	402570	NM_022168.4	Aicardi-Goutieres syndrome, Singleton-Merten syndrome	Dominante
IKBKB	300248	122614	NM_001556.3	Ectodermal dysplasia and immunodeficiency	Récessive
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
IKZF2	606234		NM_001387220.1	HELIOS deficiency	Récessive/ 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
IL18	600953	470496	NM_001562.3		Candidat

Gène	OMIM	Orphanet	Transcrit GRCh37 (NCBI)*	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
IL18BP	604113		NM_001039660.2	IL-18BP deficiency with human fulminant viral hepatitis	Récessive
IL1R1	147810		NM_000877.4	Monogenic CRMO, Loss of IL-1R1 Sensitivity to IL-1Ra (LIRSA)	Dominante
IL1RN	147679	212902	NM_173842.2	Deficit of IL-1 receptor agonist (DIRA)	Récessive
IL2RA	147730	160265	NM_000417.2	Immunodeficiency with lymphoproliferation and autoimmunity	Récessive
IL2RB	146710	355507	NM_000878.4	Immunodeficiency with lymphoproliferation and autoimmunity	*Récessive*
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
IL6ST	600694		NM_002184.4	Hyper-IgE recurrent infection syndrome (HIES)	Récessive/ Dominante
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
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/Somatique
LACC1	613409	454001	NM_001128303.2	LACC1 deficiency, Juvenile arthritis	Récessive
LCK	153390	280151	NM_001042771.2	Immunodeficiency	Récessive
LCP2	601603		NM_005565.5	SLP76 Deficiency	Récessive
LIG4	601837	123058	NM_002312.3	LIG4 Syndrome	Récessive
LPIN2	605519	123109	NM_014646.2	Majeed syndrome	Récessive
LRBA	606453	303168	NM_006726.4	Common variable immunodeficiency (CVID) with autoimmunity	Récessive

Gène	OMIM	Orphanet	Transcrit GRCh37 (NCBI)*	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
LSM11	617910		NM_173491.3	Aicardi-Goutieres syndrome	Récessive
LYN	165120		NM_002350.3	LYN associated autoinflammatory disease (LAID) LYN kinase associated vasculopathy and liver fibrosis (LAVLI) syndrome	Dominante
LYST	606897	123318	NM_000081.3	Chediak-Higashi syndrome	Récessive
MAGT1	300715	250175	NM_032121.5	Immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia	Liée à l'X
MALT1	604860	159756	NM_006785.4	Immunodeficiency	Récessive
MAN2B2	609458	123328	NM_015274.3	MAN2B2-CDG	Récessive
MCM10	609357		NM_018518.5	Immunodeficiency HLH-like	Récessive
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
MRTFA	606078	409876	NM_020831.4	Immunodeficiency	Récessive
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/ Somatique
NCF1	608512	123695	NM_000265.5	Chronic granulomatous disease	Récessive
NCF2	608515	123701	NM_000433.3	Chronic granulomatous disease	Récessive
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

Gène	OMIM	Orphanet	Transcrit GRCh37 (NCBI)*	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
NFIL3	605327		NM_005384.2	NFIL3 deficiency	Récessive
NFKB1	164011	438054	NM_003998.3	NFKB1 deficiency, activated PI3Kδ syndrome (APDS) 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
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
NLRP3	606636	123821	NM_001243133.1	NLRP3-associated autoinflammatory disease	Dominante
NLRP4	609648		NM_134444.4	PMID: 35467709	Candidat
NLRP9	609663		NM_176820.4	PMID: 35090055	Candidat
NOD2	605956	123845	NM_022162.2	Blau syndrome	Dominante
NRAS	164790	221346	NM_002524.4	RAS-associated autoimmune leukoproliferative disease	Somatique
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
PIK3CG	601232		NM_002649.2	Immunodeficiency with autoinflammation	Récessive

Gène	OMIM	Orphanet	Transcrit GRCh37 (NCBI)*	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
PIK3R1	171833	352217	NM_181523.2	Immunodeficiency	Récessive/ Dominante
PLCG1	172420		NM_002660.3	PLCG1-associated immune dysregulation disease	Dominante
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
POFUT1	607491		NM_015352.2	hidradenitis suppurativa-Dowling-Degos	Dominante
POLA1	312040	460832	NM_016937.3	X-linked pigmentary disorder, reticulate, with systemic manifestations	Liée à l'X
POMP	613386	281391	NM_015932.5	*POMP* -related autoinflammation and immune dysregulation disease (PRAID), *Proteasome-associated autoinflammatory syndrome (PRAAS)*	Dominante
POU2F2	164176		NM_001394376.1	B cell deficiencies and arthritis	Candidat (Dominant)
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
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
PSMA5	176844		NM_002790.4	Proteasome-associated autoinflammatory syndrome (PRAAS)	Récessive/ Dominante
PSMA6	602855		NM_002791.3	Proteasome-associated autoinflammatory syndrome (PRAAS)	Récessive
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

Gène	OMIM	Orphanet	Transcrit GRCh37 (NCBI)*	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
PSMC3	186852		NM_002804.5	Proteasome-associated autoinflammatory syndrome (PRAAS)	Récessive/ Dominante
PSMD12	604450		NM_002816.5	Type I interferonopathy	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
PTPN22	600716	317445	NM_015967.6		Candidat
PTPN6	176883		NM_002831.6		Candidat
PYCARD	606838		NM_013258.4		Candidat
RAB27A	603868	118188	NM_183235.2	Griscelli syndrome with HLH	Récessive
RAC1	602048	470948	NM_018890.3	Mental retardation with recurrent infections	Dominante
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
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
RELA	164014	395954	NM_021975.3	Chronic mucocutaneous ulceration	Dominante
RELB	604758		NM_006509.3	Immunodeficiency	Récessive
REXO2	607149		NM_015523.4	Type I interferonopathy	Dominante
RHBDF1	614403		NM_022450.5		Candidat
RHOG	179505		NM_001665.3	RhoG deficiency	*Récessive*

Gène	OMIM	Orphanet	Transcrit GRCh37 (NCBI)*	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
RIGI/DDX58	609631	422901	NM_014314.3	Singleton-Merten syndrome	Dominante
RIPK1	603453		NM_003804.5	Cleavage resistant RIPK1-associated fever (CRAF), Immunodeficiency with autoinflammation	Récessive/ Dominante
RIPK3	605817		NM_006871.3		Candidat
RMND1	614917		NM_017909.4	Type I interferonopathy	Récessive
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
RNASET2	612944		NM_003730.6	RNASET2-related leukodystrophy and Aicardi-Goutieres syndrome	Récessive
RND1	609038		NM_014470.3		Candidat
RNF213	613768		NM_001256071.3	RNF213-associated urticarial lesions with hypercytokinemia	Dominante
RNF31	612487	432144	NM_017999.4	HOIP deficiency (LUBAC deficiency)	Récessive
RNU7-1	617876		NR_023317.1	Aicardi-Goutieres syndrome-like	Récessive
SAA1	104750	330340	NM_001178006.2	Hereditary AA Amyloidosis	*Dominante*
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
SBNO2	615729		NM_014963.3	Juvenile arthritis and autoimmunity (in press)	Récessive
SH2D1A	300490	118679	NM_002351.4	SAP deficiency (XLP1) with lymphoproliferative syndrome	Liée à l'X
SH3BP2	602104	118689	NM_003023.4	Cherubism	Dominante
SHARPIN	611885	471052	NM_030974.3	LUBAC deficiency	Récessive

Gène	OMIM	Orphanet	Transcrit GRCh37 (NCBI)*	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
SKIC2	600478	299641	NM_006929.4	Trichohepatoenteric syndrome	Récessive
SLC12A2	600840		NM_001046.3	Gastrointestinal inflammation	Dominante
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
SLC39A4	607059		NM_130849.4	Acrodermatitis enteropathica	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
STAT1	600555	119879	NM_007315.3	Immunodeficiency	Récessive/ 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		NM_003151.4	Disabling pansclerotic morphea (DPM)	Dominante
STAT5B	604260	119882	NM_012448.3	Hypereosinophilic syndrome with urticarial rash	Somatique
STAT6	601512		NM_003153.5	Severe Atopy Associated with Lymphoma	Dominante
STIM1	605921	201300	NM_003156.3	Immunodeficiency	Récessive
STING1/ TMEM173	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
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*
SUPV3L1	605122		NM_003171.5	Type I interferonopathy	Récessive

Gène	OMIM	Orphanet	Transcrit GRCh37 (NCBI)*	Code couleur Nouveaux gènes Gènes candidats de MAI Phénotype	Hérédité
SYK	600085		NM_003177.6	SYK-associated autoinflammatory disease	*Dominante*
TBK1	604834	318865	NM_013254.3	TBK1 deficiency autoinflammation driven by TNF-induced cell death	Récessive/ Dominante
TET2	612839	268325	NM_001127208.2	Immunodeficiency	Récessive
TGFB1	190180	120047	NM_000660.6	Camurati-Engelman disease	Dominante
TLR4	603030		NM_138554.5	TLR4 deficiency	Récessive
TLR7	300365		NM_016562.4	Monogenic SLE	Lié à l'X
TLR8	300366		NM_138636.5	Immunodeficiency with autoinflammation	Dominante/ Somatique
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	Immunodeficiency and EBV-induced lymphoproliferation (4-1BB deficiency or CD137 deficiency)	Récessive
TRAF3	601896		NM_145725.3	Immunodeficiency, autoimmunity, and increased risk of B cell malignancy	Dominante
TRAP1	606219		NM_016292.2	TRAP1 chaperone protein-associated autoinflammation	Récessive
TREX1	606609	120255	NM_033629.5	Aicardi-Goutieres syndrome	Récessive/ Dominante
TRIM21	109092		NM_003141.3	TRIM21 Deficiency and Autoimmune B Cells Monogenic SLE	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/SKIC3	614589	238544	NM_014639.3	Trico hepato enteric syndrome	Récessive