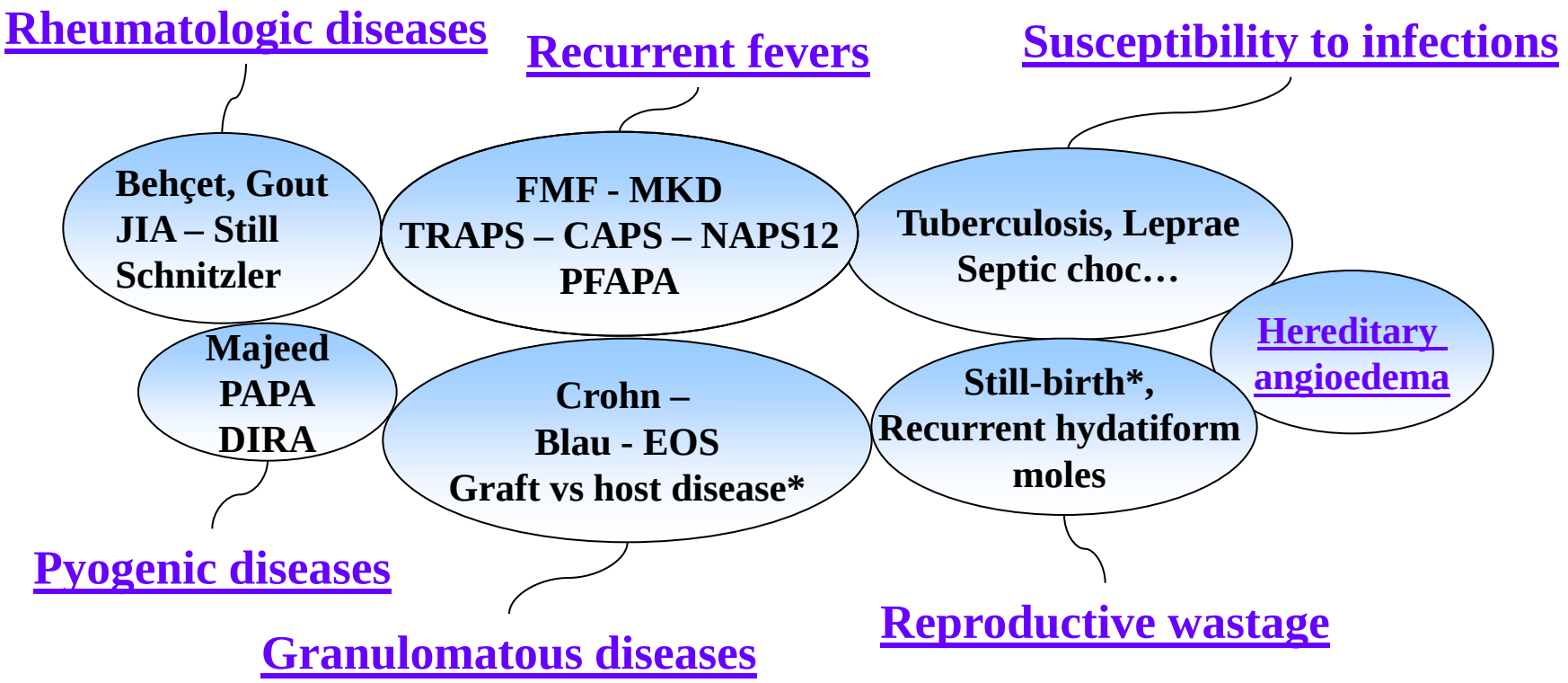
Auto-inflammatory diseases
A non-exhaustive classification



*: Certain forms of

DISEASE					THE GENE AND ITS PRODUCT				
Acronyme	**Extended**	**Synonyme(s)**	**OMIM**	**Transmission**	**Locus Map**	**Acronyme**	**Extended**	**OMIM**	**Protein and synonyme(s)**
FMF	Familial Mediterranean fever	Periodic fever, recurrent	249100	Recessive	16p13	*MEFV*	MEditerranean FeVer	608107	Pyrin, Marenostrin
MKD	Mevalonate Kinase Deficiency			Recessive	12q24	*MVK*	Mevalonate Kinase	251170	
MA	Mevalonic Aciduria		610377						
HIDS	Hyper IgD Syndrome		260920						
TRAPS	TNF-Receptor Associated Periodic Syndrome	Familial Hibernian Fever; FHF	142680	Dominant	12p13.2	*TNFRSF1A*	Tumor Necrosis Factor Receptor Super Family 1A	191190	
CAPS	Cryopyrin Associated Periodic Syndrome			Dominant	1q44	*NLRP3*	NLR pyrin domain containing protein 3	606416	Cryopyrin, Cold Induced Autoinflammatory Syndrome 1, CIAS1, Nacht Domain-, Leucine-Rich Repeat and Pyd-Containing Protein 3; NALP3 Pyrin Domain-Containing Apaf1-Like Protein 1; PYPAF1, Caterpiller 1.1; CLR 1.1
FCAS	Familial Cold Autoinflammatory Syndrome	Familial Cold Urticaria; FCU	120100						
MWS	Muckle-Wells Syndrome		191900						
CINCA	Chronic Neurologic Cutaneous and Articular Syndrome	Neonatal-Onset Multisystem Inflammatory Disease; NOMID	607115						
NAPS12	NLRP12 Associated Periodic Syndrome	Familial Cold Urticaria 2; FCAS2	611762	Dominant	19q13.3	*NLRP12*	NLR pyrin domain containing protein 12	609648	
PFAPA	Periodic Fever Aphtosis Pharyngitis Adenitis	Marshall syndrome		Unknown					

RHEUMATOLOGIC DISEASES					MAJOR GENE AND ITS PRODUCT				
Acronyme	**Extended**	**Synonyme(s)**	**OMIM**	**Transmission**	**Locus Map**	**Acronyme**	**Extended**	**OMIM**	**Protein and synonyme(s)**
(None)	Schnitzler syndrome			Unknown					
JIA	Juvenile Idiopathic Arthritis	Still disease (child)	604302	Multifactorial					
(None)	Gout		300323	Multifactorial	Xq26-q27.2	*HPRT*	Hypoxanthine Guanine Phosphoribosyltransferase 1	308000	Hypoxanthine Guanine Phosphoribosyltransferase 1
(None)	Still disease (adult)	Wissler-Fanconi		Unknown					
BD	Behcet disease	Adamantiades syndrome	109650	Multifactorial					

PYOGENIC DISEASES					THE GENE AND ITS PRODUCT				
Acronyme	**Extended**	**Synonyme(s)**	**OMIM**	**Transmission**	**Locus Map**	**Acronyme**	**Extended**	**OMIM**	**Protein and**
CRMO and CDA	Chronic Recurrent Multifocal Osteomyelitis, Congenital Dyserythropoietic Anemia, And Neutrophilic Dermatosis	Majeed Syndrome	609628	Recessive	18p11.31	*LPIN2*		605519	
PAPA	Pyogenic Sterile		604416	Dominant	15q24-q25.1	*PSTPIP1*	Proline-Serine-	606347	CD2 Binding
DIRA	Deficiency of Interleukin 1 Receptor Antagonist		612852	Recessive	2q14.2	*IL1RN*	Interleukin 1 receptor antagonist	147679	IL1RN

GRANULOMATOUS DISEASES					THE GENE AND ITS PRODUCT				
Acronyme	**Extended**	**Synonyme(s)**	**OMIM**	**Transmission**	**Locus Map**	**Acronyme**	**Extended**	**OMIM**	**Protein and synonyme(s)**
BS	Blau syndrome	Joubs syndrome	186580	Dominant	16q12	*NOD2*	Nucleotide-binding oligomerization domain 2	605956	Caspase recruitment domain-containing protein 15, CARD15, Caterpiller 16.3; CLR 16,3
EOS	Early Onset Sarcoidosis		609464	Dominant					
CD	Crohn's disease		266600	Multifactorial					

REPRODUCTIVE WASTAGE				THE GENE AND ITS PRODUCT				
Acronyme	**Extended**	**OMIM**	**Transmission**	**Locus Map**	**Acronyme**	**Extended**	**OMIM**	**Protein and synonyme(s)**
RHM	Recurrent hydatiform moles	231090	Recessive	19q13.4	*NLRP7*	NLR pyrin domain containing protein 7	609661	Nacht Domain-, Leucine-Rich Repeat-, and Pyd-Containing Protein 7, NALP7; Pyrin-containing APAF-1-like protein 3, PYPAF3; Nucleotide-binding oligomerization domain 12; NOD12, PAAD and NACHT containing protein 7; PAN7, Caterpiller 19.4; CLR 19,4

ANGIOEDEMA					THE GENE AND ITS PRODUCT				
Acronyme	**Extended**	**Synonyme(s)**	**OMIM**	**Transmission**	**Locus Map**	**Acronyme**	**Extended**	**OMIM**	**Protein and synonyme(s)**
HAE	Hereditary angioedema	Quincke oedema	106100	Dominant	11q11-q13.1	*C1NH*	Complement Component 1 Inhibitor	606860	SERPING1

SUSCEPTIBILITY TO BACTERIAL INFECTIONS					THE GENE AND ITS PRODUCT				
Acronyme	**Extended**	**Synonyme(s)**	**OMIM**	**Transmission**	**Locus Map**	**Acronyme**	**Extended**	**OMIM**	**Protein and synonyme(s)**
(None)	Lyme disease	Borreliosis		Unknown	4p14	*TLR1*		601194	
	Leprosy		246300	Unknown	4q32	*TLR2*		603028	Till4
	Endoxin hyporesponsiveness, tuberculosis			Unknown	9q32-q33	*TLR4*	Toll Receptor 4	603030	

What are auto-inflammatory diseases?

Autoinflammatory diseases are defined as illnesses caused by primary dysfunction of the innate immune system. This new concept includes a broad number of disorders, but the spotlight has been focused for the past two years on periodic fevers (FMF, Familial Mediterranean fever; MVK, Mevalonate kinase deficiency; TRAPS, TNF Receptor Associated Periodic Syndrome; CAPS, cryopyrin associated periodic syndrome), Crohn's disease and Blau syndrome, thanks to the recent understanding of their molecular basis. Indeed, until recently, these conditions were defined only by phenotypical features, the main ones being recurrent attacks of fever, abdominal pain, arthritis and cutaneous signs, which sometimes overlap, obscuring diagnosis. Search for distinguishing signs such as periorbital oedema in TRAPS, and use of specific functional tests where available, are of valuable help.

Needless to say, molecular screening of the causative genes has dramatically improved patient quality of life, by providing early and accurate diagnosis, allowing subsequent appropriate treatment. Some patients, however, remain hard to manage despite the advent of new genetic tests, and/or due to lack of effective treatment. The original clinical link between these diseases can now be confirmed by a molecular one, following the exciting discovery that most of the altered proteins are involved in inflammation and apoptosis. These protein mediate regulation of NFκB activation, cell apoptosis and IL1β secretion through cross-regulated and sometimes common signalling pathways. Knowledge of the defective steps in autoinflammatory has already led to elucidation of the mechanisms of action of existing drugs and may allow development of new therapies.