

HEREDITARY AUTO-INFLAMMATORY DISEASES

IMPORTANT: CHECK-LIST BEFORE SHIPPING

- ☐ Medical prescription
- ☐ Attestation and consent forms signed by the patient AND the prescriber (http://www.labcerba.com/images/espace_medecin/CONSSG.pdf)
- ☐ Clinical form (see other side)
- ☐ PREREQUISITES (according to the national network of AID genetic diagnosis laboratories):
- ☐ At least 3 unexplained inflammatory episodes
 - ☐ High C-Reactive Protein (CRP) during an episode (attach test results)
 - ☐ High mevalonaturia during episodes (Mevalonate Kinase Deficiency (MKD))

COLLECTION LABORATORY

BLOOD SAMPLE (5mL EDTA whole blood at room temperature)

Customer: C /

Stamp

Sampling date:

Time of collection: :

PATIENT

PRESCRIBER

SURNAME

FIRST NAME

Birth name

Date of birth

Address

ZIP code Town

Phone

Geographical origin

Parental consanguinity ☐ Yes ☐ No ☐ Unknown

Family history of auto-inflammatory syndrome (Crohn's, ulcerative colitis, Behçet's...):

☐ Yes ☐ No ☐ Unknown

SURNAME

FIRST NAME

Address

ZIP code Town

Phone

Fax

E-mail address

Signature:

INDICATION

REQUIRED TEST (CODE NATURE: SGE)

☐ Clinical suspicion

- ☐ Familial Mediterranean Fever (FMF)
- ☐ TNF receptor 1-associated periodic syndrome (TRAPS)
- ☐ Cryopyrin-associated periodic syndrome (CAPS)
- ☐ NLRP12-associated hereditary periodic fever syndrome (NAPS12)
- ☐ Periodic fever syndrome with hyper-Ig D (MKD)

☐ Family study (attach index case results)

☐ MEFV gene study (exons 2, 3, 5, 9 and 10) (code OPL : MEFV)

☐ TNFRSF1A gene study (exons 2, 3 and 4) (code OPL : TRAPS)

☐ NLRP3 gene study (exon 3) (code OPL : CAPS)

☐ NLRP12 gene study (exon 3) (code OPL : NAPS)

☐ MVK gene study (exons 2 to 11) (code OPL : MVK)

☐ Targeted study (MEFV, TNFRSF1A, NLRP3, NLRP12, MVK gene)

☐ Known mutation(s):

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CLINICAL FORM

- ☐ **Triggering factor:** ☐ Hot ☐ Cold ☐ Vaccine ☐ Stress ☐ Fatigue
- ☐ **Frequency of episodes:** ☐ <1/month ☐ 1-2/months ☐ >2/months ☐ Chronic ☐ Unknown
- ☐ **Duration of episodes:** ☐ **Age of first symptoms :**

☐ **Inflammation**

- ☐ Fever ☐ Yes ☐ No ☐ Unknown
- ☐ High CRP ☐ Yes ☐ No ☐ Unknown

☐ **Thoracic**

- ☐ Pain ☐ Yes ☐ No ☐ Unknown
- ☐ Pleuritis-Pericarditis ☐ Yes ☐ No ☐ Unknown
- ☐ Pneumopathy ☐ Yes ☐ No ☐ Unknown

☐ **Neurosensory**

- ☐ VCA/TIA ☐ Yes ☐ No ☐ Unknown
- ☐ Calcification ☐ Yes ☐ No ☐ Unknown
- ☐ Cephalaea ☐ Yes ☐ No ☐ Unknown
- ☐ Conjunctivitis ☐ Yes ☐ No ☐ Unknown
- ☐ Encephalitis ☐ Yes ☐ No ☐ Unknown
- ☐ Epilepsy ☐ Yes ☐ No ☐ Unknown
- ☐ Meningitis ☐ Yes ☐ No ☐ Unknown
- ☐ Papillitis ☐ Yes ☐ No ☐ Unknown
- ☐ Intellectual disability ☐ Yes ☐ No ☐ Unknown
- ☐ Hearing loss ☐ Yes ☐ No ☐ Unknown
- ☐ Uveitis ☐ Yes ☐ No ☐ Unknown

☐ **Locomotor**

- ☐ Arthralgia ☐ Yes ☐ No ☐ Unknown
- ☐ Arthritis ☐ Yes ☐ No ☐ Unknown
- ☐ Myalgia ☐ Yes ☐ No ☐ Unknown
- ☐ Deforming arthropathy ☐ Yes ☐ No ☐ Unknown
- ☐ Osteitis ☐ Yes ☐ No ☐ Unknown

☐ **Renal**

- ☐ Amyloidosis ☐ Yes ☐ No ☐ Unknown
- ☐ Kidney failure ☐ Yes ☐ No ☐ Unknown
- ☐ Proteinuria ☐ Yes ☐ No ☐ Unknown

☐ **Gastrointestinal**

- ☐ Abscess ☐ Yes ☐ No ☐ Unknown
- ☐ Hepatomegaly ☐ Yes ☐ No ☐ Unknown
- ☐ Cytolytic hepatitis ☐ Yes ☐ No ☐ Unknown
- ☐ Splenomegaly ☐ Yes ☐ No ☐ Unknown
- ☐ Diarrhea ☐ Yes ☐ No ☐ Unknown
- ☐ Abdominal pain ☐ Yes ☐ No ☐ Unknown
- ☐ Hemorrhage ☐ Yes ☐ No ☐ Unknown
- ☐ Vomiting ☐ Yes ☐ No ☐ Unknown

☐ **Mucocutaneous**

- ☐ Oral ulcer ☐ Yes ☐ No ☐ Unknown
- ☐ Genital ulcer ☐ Yes ☐ No ☐ Unknown
- ☐ Erythema nodosum ☐ Yes ☐ No ☐ Unknown
- ☐ Folliculitis/Acne ☐ Yes ☐ No ☐ Unknown
- ☐ Lipodystrophy ☐ Yes ☐ No ☐ Unknown
- ☐ Maculopapules ☐ Yes ☐ No ☐ Unknown
- ☐ Necrosis ☐ Yes ☐ No ☐ Unknown
- ☐ Edema ☐ Yes ☐ No ☐ Unknown
- ☐ Pseudoerysipelas ☐ Yes ☐ No ☐ Unknown
- ☐ Psoriasis ☐ Yes ☐ No ☐ Unknown
- ☐ Pyoderma ☐ Yes ☐ No ☐ Unknown
- ☐ Tenosynovitis ☐ Yes ☐ No ☐ Unknown
- ☐ Hives ☐ Yes ☐ No ☐ Unknown
- ☐ Vasculitis ☐ Yes ☐ No ☐ Unknown

☐ **Other**

- ☐ Failure to thrive ☐ Yes ☐ No ☐ Unknown
- ☐ Adenopathy ☐ Yes ☐ No ☐ Unknown
- ☐ Allergy ☐ Yes ☐ No ☐ Unknown
- ☐ Recurrent infections ☐ Yes ☐ No ☐ Unknown
- ☐ Pharyngitis ☐ Yes ☐ No ☐ Unknown

TREATMENT

- ☐ **Colchicine:** ☐ No ☐ Yes ► Response ☐ No ☐ Complete ☐ Partial
- ☐ **Corticoids:** ☐ No ☐ Yes ► Response ☐ No ☐ Complete ☐ Partial
- ☐ **NSAID:** ☐ No ☐ Yes ► Response ☐ No ☐ Complete ☐ Partial
- ☐ **IL-1 inhibitor:** ☐ No ☐ Yes ► Response ☐ No ☐ Complete ☐ Partial
- ☐ **TNF inhibitor:** ☐ No ☐ Yes ► Response ☐ No ☐ Complete ☐ Partial