Infevers Data Entry Form

SIMPLE ALLELE

I) Name of the mutation: **Usual Name:** Please refer to the HUGO-HVGS mutation nomenclature guideline
example: S108R (exonic variants) or c.1260+18C>G (non exonic variants)

Sequence variant:
example: c.195C>T or c.76_84del

Protein variant [nomenclature] :
Example: p.Ser108Arg or p.Cys28_Met30del

II) Functional tests? Yes ○ No ○ Unknown ○

III) Location in the gene:

Position **The position is defined according to the c.DNA numbering, including for intronic variants.**
Examples:
- 5' UT: c.-25C>T, the position is -25.
- Intronic mutations: c.2321+85A>G, the position is 2321 and not 85.
- 3' UT: c.*58A>G, the position is 58.

5' flanking ○ **The mutation is located upstream to the first exon (named with a negative number)**

5'UT ○ **The mutation is exonic but located upstream to the initiation codon (named with a negative number)**

Exon ○ **Exon number** **The mutation is between the initiation and the termination codons**

Intron ○ **Intron number**

3'UT	○	**The mutation is exonic but located downstream to the termination codon (named with an asterix)**
3' flanking	○	**The mutation is located downstream to the last exon (named with an asterix)**

IV) Alteration:

	Select	Number of base(s) mutated		Sequence mutated
Substitution	○			[ ] > [ ] (ex : A>T)
Deletion	○		[ ]	[ ]
Insertion	○		[ ]	[ ]
Duplication	○		[ ]	[ ]
Deletion/ Insertion	○	Del	[ ]	[ ]
		Ins	[ ]	[ ]

V) Technique(s) used:

Please choose one or more technique(s) below:

ARMS
ASO
Chips
DGGE
dHPLC

If other, please describe the technique(s) below:

[ ]

VI) Does the sequence change or define a Restriction Fragment Length Polymorphism ?

Yes ○ No ○ Unknown ○ Information about restriction enzymes (e.g.isoschizomers)

If yes, please choose one enzyme

AarI cacctgc
AatI agg/cct
AatII gacgt/c
AauI t/gtaca

choose one actic the restriction site.

VII) Consequence

Select a consequence

If complex effect or other, please describe the consequences below:

VIII) Number of control chromosomes tested

Known ○ Unknown ○

If known, please fill this field in:

IX) Was this variant observed in a symptomatic individual?

Yes ○ No ○ Unknown ○

X) Associated phenotype? (if the individual is symptomatic)

Yes ○ No ○ Unknown ○

If Yes (Ctrl+click):

- Behçet's disease
- Blau syndrome
- CINCA/NOMID
- Crohn's disease
- Early Arthritis

Other: please define this or these disease(s) in the text field:

XI) Origin of the patient where the mutation was INITIALLY found

○ Country of origin ----------Please choose a value--------

○ Other (please define)

○ Mixed (please define)

○ Unknown

○ Ancestry ----------Please choose a value--------

○ Other (please define)

○ Mixed (please define)

○ Unknown

XII) Note (write here any relevant information)

If you have a personal site about this disease, please note your URL:

XIII) References

Name of author(s):

Ex: Smith,JS; Gates,BG; (...)

Type of reference:

- ○ Personal communication
- ○ Congress abstract
  (Name, Town, Country, Date)
- ○ Publication
  - ○ PubMed:
    (http://...)
  - ○ In press:
    (Title, Journal,Year)