Resource Summary Report

Generated by NIF on Apr 29, 2025

INFEVERS

RRID:SCR_007738
Type: Tool

Proper Citation

INFEVERS (RRID:SCR_007738)

Resource Information

URL: http://fmf.igh.cnrs.fr/ISSAID/infevers

Proper Citation: INFEVERS (RRID:SCR_007738)

Description: Registry for Familial Mediterranean Fever (FMF) and hereditary inflammatory disorders mutations. As of 2014, it includes twenty genes including: MEFV, MVK, TNFRSF1A, NLRP3, NOD2, PSTPIP1, LPIN2 and NLRP7, and contains over 1338 sequence variants. Confidential data, simple and complex alleles are accepted. For each gene, a menu offers: 1) a tabular list of the variants that can be sorted by several parameters; 2) a gene graph providing a schematic representation of the variants along the gene; 3) statistical analysis of the data according to the phenotype, alteration type, and location of the mutation in the gene; 4) the cDNA and gDNA sequences of each gene, showing the nucleotide changes along the sequence, with a color-based code highlighting the gene domains, the first ATG, and the termination codon; and 5) a download menu making all tables and figures available for the users, which, except for the gene graphs, are all automatically generated and updated upon submission of the variants. The entire database was curated to comply with the HUGO Gene Nomenclature Committee (HGNC) and HGVS nomenclature guidelines, and wherever necessary, an informative note was provided.

Abbreviations: Infevers

Synonyms: Internet Fevers

Resource Type: service resource, data or information resource, storage service resource, data set, data repository

Defining Citation: PMID:18409191, PMID:15300846, PMID:12520003

Keywords: sequence variant, mutation, allele, genetics, dna, rna, protein, disease, heredity, inflammation, gene, function, phenotype, complex allele, simple allele, exon, intron, cdna sequence, genomic sequence, gdna, FASEB list

Related Condition: Familial Mediterranean Fever, Auto-inflammatory Disorder, Hereditary Auto-inflammatory Disorder

Funding: European Union

Availability: Acknowledgement required, Free, Public

Resource Name: INFEVERS

Resource ID: SCR_007738

Alternate IDs: nif-0000-03022

Alternate URLs: http://fmf.igh.cnrs.fr/infevers

Record Creation Time: 20220129T080243+0000

Record Last Update: 20250429T055157+0000

Ratings and Alerts

No rating or validation information has been found for INFEVERS.

No alerts have been found for INFEVERS.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 37 mentions in open access literature.

Listed below are recent publications. The full list is available at NIF.

Karamanakos A, et al. (2024) The expanding clinical spectrum of autoinflammatory diseases with NOD2 variants: a case series and literature review. Frontiers in immunology, 15, 1342668.

Qian M, et al. (2024) A rare missense p.C125Y mutation in the TNFRSF1A gene identified in a Chinese family with tumor necrosis factor receptor-associated periodic fever syndrome. Frontiers in genetics, 15, 1413641.

Li Z, et al. (2023) Tumor necrosis factor receptor-associated cycle syndrome: a case report and literature review. Frontiers in pediatrics, 11, 1296487.

Bustaffa M, et al. (2022) The impact of the Eurofever criteria and the new InFevers MEFV classification in real life: Results from a large international FMF cohort. Seminars in arthritis and rheumatism, 52, 151957.

Guzel F, et al. (2021) Next Generation Sequencing Based Multiplex Long-Range PCR for Routine Genotyping of Autoinflammatory Disorders. Frontiers in immunology, 12, 666273.

Wang Y, et al. (2021) Case Report: Pyogenic Arthritis, Pyoderma Gangrenosum, and Acne: A Single-Center Experience and Literature Review. Frontiers in immunology, 12, 735851.

Fernandes FP, et al. (2020) Inflammasome genetics and complex diseases: a comprehensive review. European journal of human genetics : EJHG, 28(10), 1307.

Fujimoto K, et al. (2020) Clinical and Genetic Analysis of 22 Japanese Patients with Familial Mediterranean Fever: An Examination of MEFV and 10 Other Genes Related to Autoinflammatory Syndromes. Internal medicine (Tokyo, Japan), 59(11), 1373.

Hua Y, et al. (2019) Phenotypes and genotypes of Chinese adult patients with systemic autoinflammatory diseases. Seminars in arthritis and rheumatism, 49(3), 446.

Malireddi RKS, et al. (2018) TAK1 restricts spontaneous NLRP3 activation and cell death to control myeloid proliferation. The Journal of experimental medicine, 215(4), 1023.

Cordero MD, et al. (2018) Gain of function mutation and inflammasome driven diseases in human and mouse models. Journal of autoimmunity, 91, 13.

Rigante D, et al. (2018) New mosaic tiles in childhood hereditary autoinflammatory disorders. Immunology letters, 193, 67.

Grandemange S, et al. (2017) Clinical dose effect and functional consequences of R92Q in two families presenting with a TRAPS/PFAPA-like phenotype. Molecular genetics & genomic medicine, 5(2), 110.

Ruiz-Ortiz E, et al. (2017) Disease Phenotype and Outcome Depending on the Age at Disease Onset in Patients Carrying the R92Q Low-Penetrance Variant in TNFRSF1A Gene. Frontiers in immunology, 8, 299.

Radhakrishna SM, et al. (2017) Novel mutation identified in severe early-onset tumor necrosis factor receptor-associated periodic syndrome: a case report. BMC pediatrics, 17(1), 108.

Papa R, et al. (2017) A web-based collection of genotype-phenotype associations in hereditary recurrent fevers from the Eurofever registry. Orphanet journal of rare diseases, 12(1), 167.

Peciuliene S, et al. (2016) Perinatal manifestation of mevalonate kinase deficiency and efficacy of anakinra. Pediatric rheumatology online journal, 14(1), 19.

Finetti M, et al. (2016) Chronic Infantile Neurological Cutaneous and Articular (CINCA) syndrome: a review. Orphanet journal of rare diseases, 11(1), 167.

Sönmez HE, et al. (2016) Familial Mediterranean fever: current perspectives. Journal of inflammation research, 9, 13.

Park YH, et al. (2016) Pyrin inflammasome activation and RhoA signaling in the autoinflammatory diseases FMF and HIDS. Nature immunology, 17(8), 914.