# Mauno Vihinen

## List of Publications by Citations

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|---|---|---|---|
| 274 papers | 11,337 citations | 55 h-index | 97 g-index |
| 297 ext. papers | 12,717 ext. citations | 6.3 avg, IF | 6.47 L-index |

| # | Paper | IF | Citations |
|---|---|---|---|
| 274 | The EUROclass trial: defining subgroups in common variable immunodeficiency. *Blood*, **2008**, 111, 77-85 | 2.2 | 556 |
| 273 | Copy number analysis indicates monoclonal origin of lethal metastatic prostate cancer. *Nature Medicine*, **2009**, 15, 559-65 | 50.5 | 513 |
| 272 | Performance of mutation pathogenicity prediction methods on missense variants. *Human Mutation*, **2011**, 32, 358-68 | 4.7 | 399 |
| 271 | Bruton's tyrosine kinase (Btk): function, regulation, and transformation with special emphasis on the PH domain. *Immunological Reviews*, **2009**, 228, 58-73 | 11.3 | 344 |
| 270 | Microbial amylolytic enzymes. *Critical Reviews in Biochemistry and Molecular Biology*, **1989**, 24, 329-418 | 8.7 | 306 |
| 269 | V(D)J recombination defects in lymphocytes due to RAG mutations: severe immunodeficiency with a spectrum of clinical presentations. *Blood*, **2001**, 97, 81-8 | 2.2 | 278 |
| 268 | Accuracy of protein flexibility predictions. *Proteins: Structure, Function and Bioinformatics*, **1994**, 19, 141-9 | 4.2 | 241 |
| 267 | Relationship of protein flexibility to thermostability. *Protein Engineering, Design and Selection*, **1987**, 1, 477-80 | 1.9 | 234 |
| 266 | Performance of protein stability predictors. *Human Mutation*, **2010**, 31, 675-84 | 4.7 | 228 |
| 265 | The Tec family of cytoplasmic tyrosine kinases: mammalian Btk, Bmx, Itk, Tec, Txk and homologs in other species. *BioEssays*, **2001**, 23, 436-46 | 4.1 | 227 |
| 264 | Isolation and initial characterization of a novel zinc finger gene, DNMT3L, on 21q22.3, related to the cytosine-5-methyltransferase 3 gene family. *Genomics*, **2000**, 65, 293-8 | 4.3 | 221 |
| 263 | Characterization of CA XIII, a novel member of the carbonic anhydrase isozyme family. *Journal of Biological Chemistry*, **2004**, 279, 2719-27 | 5.4 | 187 |
| 262 | Pathogenic or not? And if so, then how? Studying the effects of missense mutations using bioinformatics methods. *Human Mutation*, **2009**, 30, 703-14 | 4.7 | 184 |
| 261 | Autoinhibition of Jak2 tyrosine kinase is dependent on specific regions in its pseudokinase domain. *Molecular Biology of the Cell*, **2003**, 14, 1448-59 | 3.5 | 170 |
| 260 | The autoimmune regulator protein has transcriptional transactivating properties and interacts with the common coactivator CREB-binding protein. *Journal of Biological Chemistry*, **2000**, 275, 16802-9 | 5.4 | 168 |
| 259 | Bruton's tyrosine kinase: cell biology, sequence conservation, mutation spectrum, siRNA modifications, and expression profiling. *Immunological Reviews*, **2005**, 203, 200-15 | 11.3 | 159 |
| 258 | How to evaluate performance of prediction methods? Measures and their interpretation in variation effect analysis. *BMC Genomics*, **2012**, 13 Suppl 4, S2 | 4.5 | 152 |

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|---|---|---|---|
| 257 | BTKbase: the mutation database for X-linked agammaglobulinemia. *Human Mutation*, **2006**, 27, 1209-17 | 4.7 | 147 |
| 256 | Finding our way through phenotypes. *PLoS Biology*, **2015**, 13, e1002033 | 9.7 | 144 |
| 255 | Characterization of CA XV, a new GPI-anchored form of carbonic anhydrase. *Biochemical Journal*, **2005**, 392, 83-92 | 3.8 | 140 |
| 254 | Human Proteinpedia enables sharing of human protein data. *Nature Biotechnology*, **2008**, 26, 164-7 | 44.5 | 138 |
| 253 | PON-P2: prediction method for fast and reliable identification of harmful variants. *PLoS ONE*, **2015**, 10, e0117380 | 3.7 | 130 |
| 252 | Immunodeficiency mutation databases (IDbases). *Human Mutation*, **2006**, 27, 1200-8 | 4.7 | 101 |
| 251 | A systematic analysis of recombination activity and genotype-phenotype correlation in human recombination-activating gene 1 deficiency. *Journal of Allergy and Clinical Immunology*, **2014**, 133, 1099-108 | 11.5 | 100 |
| 250 | Mutation screening of the BTK gene in 56 families with X-linked agammaglobulinemia (XLA): 47 unique mutations without correlation to clinical course. *Pediatrics*, **1998**, 101, 276-84 | 7.4 | 98 |
| 249 | Aromatic interactions define the binding of the alphavirus spike to its nucleocapsid. *Structure*, **1996**, 4, 519-29 | 5.2 | 95 |
| 248 | SH3-Domain binding function of HIV-1 Nef is required for association with a PAK-related kinase. *Virology*, **1998**, 250, 273-82 | 3.6 | 93 |
| 247 | APECED-causing mutations in AIRE reveal the functional domains of the protein. *Human Mutation*, **2004**, 23, 245-57 | 4.7 | 90 |
| 246 | VariBench: a benchmark database for variations. *Human Mutation*, **2013**, 34, 42-9 | 4.7 | 87 |
| 245 | Screening for BRCA1, BRCA2, CHEK2, PALB2, BRIP1, RAD50, and CDH1 mutations in high-risk Finnish BRCA1/2-founder mutation-negative breast and/or ovarian cancer individuals. *Breast Cancer Research*, **2011**, 13, R20 | 8.3 | 85 |
| 244 | Tec homology (TH) adjacent to the PH domain. *FEBS Letters*, **1994**, 350, 263-5 | 3.8 | 85 |
| 243 | Of genes and phenotypes: the immunological and molecular spectrum of combined immune deficiency. Defects of the gamma(c)-JAK3 signaling pathway as a model. *Immunological Reviews*, **2000**, 178, 39-48 | 11.3 | 84 |
| 242 | CD40Lbase: a database of CD40L gene mutations causing X-linked hyper-IgM syndrome. *Trends in Immunology*, **1996**, 17, 511-516 | | 84 |
| 241 | Structure-function analysis of PrsA reveals roles for the parvulin-like and flanking N- and C-terminal domains in protein folding and secretion in Bacillus subtilis. *Journal of Biological Chemistry*, **2004**, 279, 19302-14 | 5.4 | 83 |
| 240 | Mutations in severe combined immune deficiency (SCID) due to JAK3 deficiency. *Human Mutation*, **2001**, 18, 255-63 | 4.7 | 83 |

| | | | |
|---|---|---|---|
| 239 | PON-P: integrated predictor for pathogenicity of missense variants. *Human Mutation*, **2012**, 33, 1166-74 | 4.7 | 79 |
| 238 | Deletion within the Src homology domain 3 of Bruton's tyrosine kinase resulting in X-linked agammaglobulinemia (XLA). *Journal of Experimental Medicine*, **1994**, 180, 461-70 | 16.6 | 78 |
| 237 | Identification of candidate disease genes by integrating Gene Ontologies and protein-interaction networks: case study of primary immunodeficiencies. *Nucleic Acids Research*, **2009**, 37, 622-8 | 20.1 | 76 |
| 236 | Mutations of the human BTK gene coding for bruton tyrosine kinase in X-linked agammaglobulinemia. *Human Mutation*, **1999**, 13, 280-5 | 4.7 | 76 |
| 235 | BTKbase: a database of XLA-causing mutations. International Study Group. *Trends in Immunology*, **1995**, 16, 460-5 | | 76 |
| 234 | Variation Interpretation Predictors: Principles, Types, Performance, and Choice. *Human Mutation*, **2016**, 37, 579-97 | 4.7 | 75 |
| 233 | PhenCode: connecting ENCODE data with mutations and phenotype. *Human Mutation*, **2007**, 28, 554-62 | 4.7 | 72 |
| 232 | Site-directed mutagenesis of a thermostable alpha-amylase from Bacillus stearothermophilus: putative role of three conserved residues. *Journal of Biochemistry*, **1990**, 107, 267-72 | 3.1 | 69 |
| 231 | Solution structure of the SH3 domain from Bruton's tyrosine kinase. *Biochemistry*, **1998**, 37, 2912-24 | 3.2 | 68 |
| 230 | Structural basis for chromosome X-linked agammaglobulinemia: a tyrosine kinase disease. *Proceedings of the National Academy of Sciences of the United States of America*, **1994**, 91, 12803-7 | 11.5 | 68 |
| 229 | Guidelines for reporting and using prediction tools for genetic variation analysis. *Human Mutation*, **2013**, 34, 275-82 | 4.7 | 64 |
| 228 | Spectrum of disease-causing mutations in protein secondary structures. *BMC Structural Biology*, **2007**, 7, 56 | 2.7 | 62 |
| 227 | BTKbase, mutation database for X-linked agammaglobulinemia (XLA). *Nucleic Acids Research*, **1998**, 26, 242-7 | 20.1 | 60 |
| 226 | Primary immunodeficiency mutation databases. *Advances in Genetics*, **2001**, 43, 103-88 | 3.3 | 58 |
| 225 | Structural basis for pleckstrin homology domain mutations in X-linked agammaglobulinemia. *Biochemistry*, **1995**, 34, 1475-81 | 3.2 | 57 |
| 224 | A novel family of DNA-polymerase-associated B subunits. *Trends in Biochemical Sciences*, **1999**, 24, 14-6 | 10.3 | 56 |
| 223 | Sequence specificity in CpG mutation hotspots. *FEBS Letters*, **1996**, 396, 119-22 | 3.8 | 56 |
| 222 | X-linked agammaglobulinemia (XLA): a genetic tyrosine kinase (Btk) disease. *BioEssays*, **1996**, 18, 825-34 | 4.1 | 56 |

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|---|---|---|---|
| 221 | CD40lbase: a database of CD40L gene mutations causing X-linked hyper-IgM syndrome. *Trends in Immunology*, **1996**, 17, 511-6 | | 56 |
| 220 | Pattern of somatic androgen receptor gene mutations in patients with hormone-refractory prostate cancer. *Laboratory Investigation*, **2002**, 82, 1591-8 | 5.9 | 55 |
| 219 | Mapping and cloning of FAD2 gene to develop allele-specific PCR for oleic acid in spring turnip rape (Brassica rapa ssp. oleifera). *Molecular Breeding*, **1998**, 4, 543-550 | 3.4 | 54 |
| 218 | Identification of Btk mutations in 20 unrelated patients with X-linked agammaglobulinaemia (XLA). *Human Molecular Genetics*, **1995**, 4, 693-700 | 5.6 | 54 |
| 217 | KinMutBase: a registry of disease-causing mutations in protein kinase domains. *Human Mutation*, **2005**, 25, 435-42 | 4.7 | 53 |
| 216 | Recommendations for locus-specific databases and their curation. *Human Mutation*, **2008**, 29, 2-5 | 4.7 | 52 |
| 215 | BTKbase, mutation database for X-linked agammaglobulinemia (XLA). *Nucleic Acids Research*, **1996**, 24, 160-5 | 20.1 | 51 |
| 214 | Determination and analysis of antigenic epitopes of prostate specific antigen (PSA) and human glandular kallikrein 2 (hK2) using synthetic peptides and computer modeling. *Protein Science*, **1998**, 7, 259-69 | 6.3 | 51 |
| 213 | Profiling genetic variation along the androgen biosynthesis and metabolism pathways implicates several single nucleotide polymorphisms and their combinations as prostate cancer risk factors. *Cancer Research*, **2006**, 66, 743-7 | 10.1 | 51 |
| 212 | ETS1 mediates MEK1/2-dependent overexpression of cancerous inhibitor of protein phosphatase 2A (CIP2A) in human cancer cells. *PLoS ONE*, **2011**, 6, e17979 | 3.7 | 49 |
| 211 | Thermal unfolding of small proteins with SH3 domain folding pattern. *Proteins: Structure, Function and Bioinformatics*, **1998**, 31, 309-19 | 4.2 | 49 |
| 210 | Selection of a representative set of structures from Brookhaven Protein Data Bank. *Proteins: Structure, Function and Bioinformatics*, **1992**, 14, 265-76 | 4.2 | 47 |
| 209 | Delta3,5-delta2,4-dienoyl-CoA isomerase from rat liver. Molecular characterization. *Journal of Biological Chemistry*, **1998**, 273, 349-55 | 5.4 | 46 |
| 208 | Immunome: a reference set of genes and proteins for systems biology of the human immune system. *Cellular Immunology*, **2006**, 244, 87-9 | 4.4 | 44 |
| 207 | Genome wide analysis of pathogenic SH2 domain mutations. *Proteins: Structure, Function and Bioinformatics*, **2008**, 72, 779-92 | 4.2 | 43 |
| 206 | Guidelines for establishing locus specific databases. *Human Mutation*, **2012**, 33, 298-305 | 4.7 | 42 |
| 205 | Bioinformatic analysis of protein structure-function relationships: case study of leukocyte elastase (ELA2) missense mutations. *Human Mutation*, **2006**, 27, 1230-43 | 4.7 | 42 |
| 204 | Probing the alpha-complementing domain of E. coli beta-galactosidase with use of an insertional pentapeptide mutagenesis strategy based on Mu in vitro DNA transposition. *Proteins: Structure, Function and Bioinformatics*, **2004**, 54, 681-92 | 4.2 | 42 |

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|---|---|---|---|
| 203 | Variation Ontology for annotation of variation effects and mechanisms. *Genome Research*, **2014**, 24, 356-64 | 9.7 | 41 |
| 202 | Changes in apoptosis-related pathways in acute myelocytic leukemia. *Cancer Genetics and Cytogenetics*, **2003**, 146, 89-101 | | 41 |
| 201 | Prediction of disease-related mutations affecting protein localization. *BMC Genomics*, **2009**, 10, 122 | 4.5 | 40 |
| 200 | Curating gene variant databases (LSDBs): toward a universal standard. *Human Mutation*, **2012**, 33, 291-7 | 4.7 | 39 |
| 199 | BTKbase, mutation database for X-linked agammaglobulinemia (XLA). *Nucleic Acids Research*, **1997**, 25, 166-71 | 20.1 | 39 |
| 198 | Capturing all disease-causing mutations for clinical and research use: toward an effortless system for the Human Variome Project. *Genetics in Medicine*, **2009**, 11, 843-9 | 8.1 | 37 |
| 197 | BTKbase: XLA-mutation registry. *Trends in Immunology*, **1996**, 17, 502-506 | | 37 |
| 196 | Conservation of functional residues between yeast and E. coli inorganic pyrophosphatases. *BBA - Proteins and Proteomics*, **1990**, 1038, 338-45 | | 37 |
| 195 | Conservation and covariance in PH domain sequences: physicochemical profile and information theoretical analysis of XLA-causing mutations in the Btk PH domain. *Protein Engineering, Design and Selection*, **2004**, 17, 267-76 | 1.9 | 36 |
| 194 | VariSNP, a benchmark database for variations from dbSNP. *Human Mutation*, **2015**, 36, 161-6 | 4.7 | 35 |
| 193 | The structural basis of hyper IgM deficiency - CD40L mutations. *Protein Engineering, Design and Selection*, **2007**, 20, 133-41 | 1.9 | 35 |
| 192 | B cells. *International Journal of Biochemistry and Cell Biology*, **2005**, 37, 518-23 | 5.6 | 35 |
| 191 | A statistical score for assessing the quality of multiple sequence alignments. *BMC Bioinformatics*, **2006**, 7, 484 | 3.6 | 35 |
| 190 | Identification of novel transcription factor-like gene from human intestinal cells. *Biochemical and Biophysical Research Communications*, **2000**, 276, 660-6 | 3.4 | 35 |
| 189 | Bioinformatics in proteomics. *New Biotechnology*, **2001**, 18, 241-8 | | 34 |
| 188 | Structural Basis of Bloom Syndrome (BS) Causing Mutations in the BLM Helicase Domain. *Molecular Medicine*, **2000**, 6, 155-164 | 6.2 | 33 |
| 187 | How good are pathogenicity predictors in detecting benign variants?. *PLoS Computational Biology*, **2019**, 15, e1006481 | 5 | 32 |
| 186 | Abnormal cerebellar development and ataxia in CARP VIII morphant zebrafish. *Human Molecular Genetics*, **2013**, 22, 417-32 | 5.6 | 32 |

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|---|---|---|---|
| 185 | PON-tstab: Protein Variant Stability Predictor. Importance of Training Data Quality. *International Journal of Molecular Sciences*, **2018**, 19, | 6.3 | 31 |
| 184 | Physicochemical feature-based classification of amino acid mutations. *Protein Engineering, Design and Selection*, **2008**, 21, 37-44 | 1.9 | 31 |
| 183 | Intermolecular interactions between the SH3 domain and the proline-rich TH region of Bruton's tyrosine kinase. *FEBS Letters*, **2001**, 489, 67-70 | 3.8 | 31 |
| 182 | Pleckstrin homology domains of tec family protein kinases. *Biochemical and Biophysical Research Communications*, **1999**, 265, 151-7 | 3.4 | 31 |
| 181 | Characterization of all possible single-nucleotide change caused amino acid substitutions in the kinase domain of Bruton tyrosine kinase. *Human Mutation*, **2015**, 36, 638-47 | 4.7 | 30 |
| 180 | Different gene expression in immunoglobulin-mutated and immunoglobulin-unmutated forms of chronic lymphocytic leukemia. *Cancer Genetics and Cytogenetics*, **2004**, 153, 69-72 | | 30 |
| 179 | cDNA microarray analysis of gene expression in coeliac disease jejunal biopsy samples. *Journal of Autoimmunity*, **2004**, 22, 249-65 | 15.5 | 30 |
| 178 | Structure-based prediction of the effects of a missense variant on protein stability. *Amino Acids*, **2013**, 44, 847-55 | 3.5 | 29 |
| 177 | Structural basis of Wiskott-Aldrich syndrome causing mutations in the WH1 domain. *Journal of Molecular Medicine*, **2000**, 78, 530-7 | 5.5 | 29 |
| 176 | Working toward precision medicine: Predicting phenotypes from exomes in the Critical Assessment of Genome Interpretation (CAGI) challenges. *Human Mutation*, **2017**, 38, 1182-1192 | 4.7 | 28 |
| 175 | PON-mt-tRNA: a multifactorial probability-based method for classification of mitochondrial tRNA variations. *Nucleic Acids Research*, **2016**, 44, 2020-7 | 20.1 | 28 |
| 174 | PON-Sol: prediction of effects of amino acid substitutions on protein solubility. *Bioinformatics*, **2016**, 32, 2032-4 | 7.2 | 28 |
| 173 | Immunome knowledge base (IKB): an integrated service for immunome research. *BMC Immunology*, **2009**, 10, 3 | 3.7 | 28 |
| 172 | Missense mutations affecting a conserved cysteine pair in the TH domain of Btk. *FEBS Letters*, **1997**, 413, 205-10 | 3.8 | 28 |
| 171 | Six X-linked agammaglobulinemia-causing missense mutations in the Src homology 2 domain of Bruton's tyrosine kinase: phosphotyrosine-binding and circular dichroism analysis. *Journal of Immunology*, **2000**, 164, 4170-7 | 5.3 | 28 |
| 170 | C-terminal truncations of a thermostable Bacillus stearothermophilus alpha-amylase. *Protein Engineering, Design and Selection*, **1994**, 7, 1255-9 | 1.9 | 28 |
| 169 | Bruton tyrosine kinase (BTK) in X-linked agammaglobulinemia (XLA). *Frontiers in Bioscience - Landmark*, **2000**, 5, D917-28 | 2.8 | 28 |
| 168 | PROlocalizer: integrated web service for protein subcellular localization prediction. *Amino Acids*, **2011**, 40, 975-80 | 3.5 | 27 |

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|---|---|---|---|
| 167 | Molecular characterization of the immune system: emergence of proteins, processes, and domains. *Immunogenetics*, **2007**, 59, 333-48 | 3.2 | 27 |
| 166 | Modeling of prostate specific antigen and human glandular kallikrein structures. *Biochemical and Biophysical Research Communications*, **1994**, 204, 1251-6 | 3.4 | 27 |
| 165 | Novel immunodeficiency data servers. *Immunological Reviews*, **2000**, 178, 177-85 | 11.3 | 26 |
| 164 | Structural basis for SH2D1A mutations in X-linked lymphoproliferative disease. *Biochemical and Biophysical Research Communications*, **2000**, 269, 124-30 | 3.4 | 26 |
| 163 | copy number variation analysis in familial BRCA1/2-negative Finnish breast and ovarian cancer. *PLoS ONE*, **2013**, 8, e71802 | 3.7 | 25 |
| 162 | Specific autoantibody profiles and disease subgroups correlate with circulating micro-RNA in systemic sclerosis. *Rheumatology*, **2015**, 54, 2100-7 | 3.9 | 24 |
| 161 | Classification of mismatch repair gene missense variants with PON-MMR. *Human Mutation*, **2012**, 33, 642-50 | 4.7 | 24 |
| 160 | Different molecular behavior of CD40 mutants causing hyper-IgM syndrome. *Blood*, **2010**, 116, 5867-74 | 2.2 | 24 |
| 159 | Predicting Severity of Disease-Causing Variants. *Human Mutation*, **2017**, 38, 357-364 | 4.7 | 23 |
| 158 | Phylogeny of Tec family kinases identification of a premetazoan origin of Btk, Bmx, Itk, Tec, Txk, and the Btk regulator SH3BP5. *Advances in Genetics*, **2008**, 64, 51-80 | 3.3 | 23 |
| 157 | RankViaContact: ranking and visualization of amino acid contacts. *Bioinformatics*, **2003**, 19, 2161-2 | 7.2 | 23 |
| 156 | Types and effects of protein variations. *Human Genetics*, **2015**, 134, 405-21 | 6.3 | 22 |
| 155 | Novel association of neurofibromatosis type 1-causing mutations in families with neurofibromatosis-Noonan syndrome. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 579-87 | 2.5 | 21 |
| 154 | Systematic classification of primary immunodeficiencies based on clinical, pathological, and laboratory parameters. *Journal of Immunology*, **2009**, 183, 7569-75 | 5.3 | 21 |
| 153 | Identification of differentially expressed genes after PPM1D silencing in breast cancer. *Cancer Letters*, **2008**, 259, 61-70 | 9.9 | 21 |
| 152 | IDR knowledge base for primary immunodeficiencies. *Immunome Research*, **2007**, 3, 6 | | 21 |
| 151 | Molecular dynamics simulation of the effects of cytosine methylation on structure of oligonucleotides. *Computational and Theoretical Chemistry*, **2001**, 546, 51-62 | | 21 |
| 150 | Molecular modeling of the Jak3 kinase domains and structural basis for severe combined immunodeficiency. *Clinical Immunology*, **2000**, 96, 108-18 | 9 | 21 |

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|---|---|---|---|
| 149 | Structural analysis of an anti-estradiol antibody. *Molecular Immunology*, **1997**, 34, 1215-26 | 4.3 | 20 |
| 148 | Structural aspects of signal transduction in B-cells. *Critical Reviews in Immunology*, **1996**, 16, 251-75 | 1.8 | 20 |
| 147 | BTKbase: XLA-mutation registry. *Trends in Immunology*, **1996**, 17, 502-6 | | 20 |
| 146 | Performance of protein disorder prediction programs on amino acid substitutions. *Human Mutation* , **2014**, 35, 794-804 | 4.7 | 19 |
| 145 | National research contributions: A case study on Finnish biomedical research. *Scientometrics*, **2008**, 77, 207-222 | 3 | 19 |
| 144 | Molecular and Structural Characterization of Five Novel Mutations in the Bruton Tyrosine Kinase Gene from Patients with X-Linked Agammaglobulinemia. *Molecular Medicine*, **1997**, 3, 477-485 | 6.2 | 18 |
| 143 | X-linked agammaglobulinemia: lack of mature B lineage cells caused by mutations in the Btk kinase. *Seminars in Immunopathology*, **1998**, 19, 369-81 | | 18 |
| 142 | FGF family members differentially regulate maturation and proliferation of stem cell-derived astrocytes. *Scientific Reports*, **2019**, 9, 9610 | 4.9 | 17 |
| 141 | Sharing data between LSDBs and central repositories. *Human Mutation*, **2009**, 30, 493-5 | 4.7 | 17 |
| 140 | Stimulation of B and T cells activates expression of transcription and differentiation factors. *Biochemical and Biophysical Research Communications*, **1998**, 249, 475-80 | 3.4 | 17 |
| 139 | No more hidden solutions in bioinformatics. *Nature*, **2015**, 521, 261 | 50.4 | 17 |
| 138 | How to Define Pathogenicity, Health, and Disease?. *Human Mutation*, **2017**, 38, 129-136 | 4.7 | 16 |
| 137 | ImmTree: database of evolutionary relationships of genes and proteins in the human immune system. *Immunome Research*, **2007**, 3, 4 | | 16 |
| 136 | KinMutBase, a database of human disease-causing protein kinase mutations. *Nucleic Acids Research*, **1999**, 27, 362-4 | 20.1 | 16 |
| 135 | Immunodeficiency mutation databases--a new research tool. *Trends in Immunology*, **1996**, 17, 495-6 | | 16 |
| 134 | Model-based prediction of sequence alignment quality. *Bioinformatics*, **2008**, 24, 2165-71 | 7.2 | 15 |
| 133 | Online registry of genetic and clinical immunodeficiency diagnostic laboratories, IDdiagnostics. *Journal of Clinical Immunology*, **2004**, 24, 53-61 | 5.7 | 15 |
| 132 | Large differences in proportions of harmful and benign amino acid substitutions between proteins and diseases. *Human Mutation*, **2017**, 38, 839-848 | 4.7 | 14 |

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|---|---|---|---|
| 131 | Expanded national database collection and data coverage in the FINDbase worldwide database for clinically relevant genomic variation allele frequencies. *Nucleic Acids Research*, **2017**, 45, D846-D853 | 20.1 | 14 |
| 130 | Classification of Amino Acid Substitutions in Mismatch Repair Proteins Using PON-MMR2. *Human Mutation*, **2015**, 36, 1128-34 | 4.7 | 14 |
| 129 | Majority vote and other problems when using computational tools. *Human Mutation*, **2014**, 35, 912-4 | 4.7 | 14 |
| 128 | VarioML framework for comprehensive variation data representation and exchange. *BMC Bioinformatics*, **2012**, 13, 254 | 3.6 | 14 |
| 127 | Androgen-sensitive human prostate cancer cells, LNCaP, produce both N-terminally mature and truncated prostate-specific antigen isoforms. *FEBS Journal*, **1998**, 255, 329-35 | | 14 |
| 126 | High-precision mapping of protein protein interfaces: an integrated genetic strategy combining en masse mutagenesis and DNA-level parallel analysis on a yeast two-hybrid platform. *Nucleic Acids Research*, **2007**, 35, e103 | 20.1 | 14 |
| 125 | Interaction between Btk TH and SH3 domain. *Biopolymers*, **2002**, 63, 325-34 | 2.2 | 14 |
| 124 | Coevolution of the domains of cytoplasmic tyrosine kinases. *Molecular Biology and Evolution*, **2001**, 18, 312-21 | 8.3 | 14 |
| 123 | TNF- and -synuclein fibrils differently regulate human astrocyte immune reactivity and impair mitochondrial respiration. *Cell Reports*, **2021**, 34, 108895 | 10.6 | 14 |
| 122 | Variation benchmark datasets: update, criteria, quality and applications. *Database: the Journal of Biological Databases and Curation*, **2020**, 2020, | 5 | 13 |
| 121 | PseudoGeneQuest - service for identification of different pseudogene types in the human genome. *BMC Bioinformatics*, **2008**, 9, 299 | 3.6 | 13 |
| 120 | Proteome analysis of B-cell maturation. *Proteomics*, **2006**, 6, 5152-68 | 4.8 | 13 |
| 119 | Analysis of Btk mutations in patients with X-linked agammaglobulinaemia (XLA) and determination of carrier status in normal female relatives: a nationwide study of Btk deficiency in Greece. *Scandinavian Journal of Immunology*, **2001**, 54, 321-7 | 3.4 | 13 |
| 118 | IDR: the ImmunoDeficiency Resource. *Nucleic Acids Research*, **2002**, 30, 232-4 | 20.1 | 13 |
| 117 | Structural similarity of the binding sites of cyclophilin A-cyclosporin A and FKBP-FK506 systems. *Biochemical and Biophysical Research Communications*, **1993**, 192, 912-7 | 3.4 | 13 |
| 116 | Human Variome Project Quality Assessment Criteria for Variation Databases. *Human Mutation*, **2016**, 37, 549-58 | 4.7 | 13 |
| 115 | Representativeness of variation benchmark datasets. *BMC Bioinformatics*, **2018**, 19, 461 | 3.6 | 13 |
| 114 | Harmful somatic amino acid substitutions affect key pathways in cancers. *BMC Medical Genomics*, **2015**, 8, 53 | 3.7 | 12 |

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|---|---|---|---|
| 113 | Pan-cancer analysis of neoepitopes. *Scientific Reports*, **2018**, 8, 12735 | 4.9 | 12 |
| 112 | Clustering of gene ontology terms in genomes. *Gene*, **2014**, 550, 155-64 | 3.8 | 12 |
| 111 | Gene expression profiles in human and mouse primary cells provide new insights into the differential actions of vitamin D3 metabolites. *PLoS ONE*, **2013**, 8, e75338 | 3.7 | 12 |
| 110 | Somatic mutation databases as tools for molecular epidemiology and molecular pathology of cancer: proposed guidelines for improving data collection, distribution, and integration. *Human Mutation*, **2009**, 30, 275-82 | 4.7 | 12 |
| 109 | Crystal structure of a 1.6-hexanediol bound tetrameric form of Escherichia coli lac-repressor refined to 2.1 A resolution. *Proteins: Structure, Function and Bioinformatics*, **2009**, 75, 748-59 | 4.2 | 12 |
| 108 | Identification of csk tyrosine phosphorylation sites and a tyrosine residue important for kinase domain structure. *Biochemical Journal*, **1997**, 322 ( Pt 3), 927-35 | 3.8 | 12 |
| 107 | Medical Expert Systems. *Current Bioinformatics*, **2008**, 3, 56-65 | 4.7 | 12 |
| 106 | Efficiency of the immunome protein interaction network increases during evolution. *Immunome Research*, **2008**, 4, 4 | | 12 |
| 105 | Immunity genes and their orthologs: a multi-species database. *International Immunology*, **2007**, 19, 1361-70 | 4.9 | 12 |
| 104 | Virtual bioinformatics distance learning suite*. *Biochemistry and Molecular Biology Education*, **2004**, 32, 156-60 | 1.3 | 12 |
| 103 | Stimulation-induced gene expression in Ramos B-cells. *Genes and Immunity*, **2003**, 4, 343-50 | 4.4 | 12 |
| 102 | Microarray analysis of B-cell stimulation. *Vitamins and Hormones*, **2002**, 64, 77-99 | 2.5 | 12 |
| 101 | Nucleolar proteins with altered expression in leukemic cell lines. *Leukemia Research*, **2012**, 36, 232-6 | 2.7 | 11 |
| 100 | Mutation (variation) databases and registries: a rationale for coordination of efforts. *Nature Reviews Genetics*, **2011**, 12, 881; discussion 881 | 30.1 | 11 |
| 99 | On exact string matching of unique oligonucleotides. *Computers in Biology and Medicine*, **2005**, 35, 173-81 | 1.7 | 11 |
| 98 | Structural basis of ICF-causing mutations in the methyltransferase domain of DNMT3B. *Protein Engineering, Design and Selection*, **2002**, 15, 1005-14 | 1.9 | 11 |
| 97 | Conserved residues of liquefying alpha-amylases are concentrated in the vicinity of active site. *Biochemical and Biophysical Research Communications*, **1990**, 166, 61-5 | 3.4 | 11 |
| 96 | Muddled genetic terms miss and mess the message. *Trends in Genetics*, **2015**, 31, 423-5 | 8.5 | 10 |

| | | | |
|---|---|---|---|
| 95 | Benchmarking subcellular localization and variant tolerance predictors on membrane proteins. *BMC Genomics*, **2019**, 20, 547 | 4.5 | 10 |
| 94 | Human Variome Project country nodes: documenting genetic information within a country. *Human Mutation*, **2012**, 33, 1513-9 | 4.7 | 10 |
| 93 | Representative selection of proteins based on nuclear families. *Protein Engineering, Design and Selection*, **1995**, 8, 501-3 | 1.9 | 10 |
| 92 | Accurate prediction of protein secondary structural class with fuzzy structural vectors. *Protein Engineering, Design and Selection*, **1995**, 8, 505-12 | 1.9 | 10 |
| 91 | Simultaneous comparison of several sequences. *Methods in Enzymology*, **1990**, 183, 447-56 | 1.7 | 10 |
| 90 | Performance of in silico tools for the evaluation of p16INK4a (CDKN2A) variants in CAGI. *Human Mutation*, **2017**, 38, 1042-1050 | 4.7 | 9 |
| 89 | Expression of sterol regulatory element-binding transcription factor (SREBF) 2 and SREBF cleavage-activating protein (SCAP) in human atheroma and the association of their allelic variants with sudden cardiac death. *Thrombosis Journal*, **2008**, 6, 17 | 5.6 | 9 |
| 88 | Dynamic covariation between gene expression and proteome characteristics. *BMC Bioinformatics*, **2005**, 6, 215 | 3.6 | 9 |
| 87 | Mobile access to biological databases on the Internet. *IEEE Transactions on Biomedical Engineering*, **2002**, 49, 1477-9 | 5 | 9 |
| 86 | Structure-function effects in primary immunodeficiencies. *Scandinavian Journal of Immunology*, **2000**, 52, 226-32 | 3.4 | 9 |
| 85 | BTK gatekeeper residue variation combined with cysteine 481 substitution causes super-resistance to irreversible inhibitors acalabrutinib, ibrutinib and zanubrutinib. *Leukemia*, **2021**, 35, 1317-1329 | 10.7 | 9 |
| 84 | Structure-Function Relationships of Covalent and Non-Covalent BTK Inhibitors. *Frontiers in Immunology*, **2021**, 12, 694853 | 8.4 | 9 |
| 83 | Standard development at the Human Variome Project. *Database: the Journal of Biological Databases and Curation*, **2015**, 2015, | 5 | 8 |
| 82 | Bioinformatics services related to diagnosis of primary immunodeficiencies. *Current Opinion in Allergy and Clinical Immunology*, **2009**, 9, 531-6 | 3.3 | 8 |
| 81 | Registries of immunodeficiency patients and mutations. *Human Mutation*, **1997**, 10, 261-7 | 4.7 | 8 |
| 80 | A structured simple form for ordering genetic tests is needed to ensure coupling of clinical detail (phenotype) with DNA variants (genotype) to ensure utility in publication and databases. *Human Mutation*, **2007**, 28, 931-2 | 4.7 | 8 |
| 79 | On preprocessing of protein sequences for neural network prediction of polyproline type II secondary structures. *Computers in Biology and Medicine*, **2001**, 31, 385-98 | 7 | 8 |
| 78 | Site-directed mutagenesis of putative active-site residues of Bacillus stearothermophilus □amylase. *Molecular Engineering*, **1991**, 1, 267-273 | | 8 |

| | | | |
|---|---|---|---|
| 77 | Proper reporting of predictor performance. *Nature Methods*, **2014**, 11, 781 | 21.6 | 7 |
| 76 | NMD and microRNA expression profiling of the HPCX1 locus reveal MAGEC1 as a candidate prostate cancer predisposition gene. *BMC Cancer*, **2011**, 11, 327 | 4.8 | 7 |
| 75 | Proteomic changes during B cell maturation: 2D-DIGE approach. *PLoS ONE*, **2013**, 8, e77894 | 3.7 | 7 |
| 74 | ProTstab - predictor for cellular protein stability. *BMC Genomics*, **2019**, 20, 804 | 4.5 | 7 |
| 73 | Strategy for Disease Diagnosis, Progression Prediction, Risk Group Stratification and Treatment-Case of COVID-19. *Frontiers in Medicine*, **2020**, 7, 294 | 4.9 | 6 |
| 72 | Statistical methods for identifying conserved residues in multiple sequence alignment. *Statistical Applications in Genetics and Molecular Biology*, **2004**, 3, Article28 | 1.2 | 6 |
| 71 | Novel insertions of Bruton tyrosine kinase in patients with X-linked agammaglobulinemia. *Human Mutation*, **2002**, 20, 480-1 | 4.7 | 6 |
| 70 | Efficient estimation of emission probabilities in profile hidden Markov models. *Bioinformatics*, **2003**, 19, 2359-68 | 7.2 | 6 |
| 69 | Solubility of proteins.. *ADMET and DMPK*, **2020**, 8, 391-399 | 1.3 | 6 |
| 68 | Fast iterative gene clustering based on information theoretic criteria for selecting the cluster structure. *Journal of Computational Biology*, **2004**, 11, 660-82 | 1.7 | 6 |
| 67 | Variation ontology: annotator guide. *Journal of Biomedical Semantics*, **2014**, 5, 9 | 2.2 | 5 |
| 66 | A novel mutation in CD40 and its functional characterization. *Human Mutation*, **2009**, 30, 985-94 | 4.7 | 5 |
| 65 | Genome-wide selection of unique and valid oligonucleotides. *Nucleic Acids Research*, **2005**, 33, e115 | 20.1 | 5 |
| 64 | Interactions between SH2 and SH3 domains. *Biochemical and Biophysical Research Communications*, **1998**, 242, 351-6 | 3.4 | 5 |
| 63 | Computer modeling of two inorganic pyrophosphatases. *Biochemical and Biophysical Research Communications*, **1992**, 186, 122-8 | 3.4 | 5 |
| 62 | Systematics for types and effects of DNA variations. *BMC Genomics*, **2018**, 19, 974 | 4.5 | 5 |
| 61 | PON-P and PON-P2 predictor performance in CAGI challenges: Lessons learned. *Human Mutation*, **2017**, 38, 1085-1091 | 4.7 | 4 |
| 60 | Problems in variation interpretation guidelines and in their implementation in computational tools. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1206 | 2.3 | 4 |

| | | | |
|---|---|---|---|
| 59 | Guidelines for systematic reporting of sequence alignments. *Biology Methods and Protocols*, **2020**, 5, bpaa001 | 2.4 | 4 |
| 58 | PON-SC - program for identifying steric clashes caused by amino acid substitutions. *BMC Bioinformatics*, **2017**, 18, 531 | 3.6 | 4 |
| 57 | Identification of core T cell network based on immunome interactome. *BMC Systems Biology*, **2014**, 8, 17 | 3.5 | 4 |
| 56 | On Postprocessing of Neural Network Prediction of Polyproline Type II Secondary Structures: Network Spectrum, Response Analysis, and Scattering. *Neural Computing and Applications*, **2003**, 11, 238-243 | 4.8 | 4 |
| 55 | Distribution of immunodeficiency fact files with XML--from Web to WAP. *BMC Medical Informatics and Decision Making*, **2005**, 5, 21 | 3.6 | 4 |
| 54 | Rational design and purification of human Bruton's tyrosine kinase SH3-SH2 protein for structure-function studies. *Protein Expression and Purification*, **2000**, 20, 365-71 | 2 | 4 |
| 53 | MULTICOMP: a program package for multiple sequence comparison. *Bioinformatics*, **1992**, 8, 35-8 | 7.2 | 4 |
| 52 | Flexibility in MuA transposase family protein structures: functional mapping with scanning mutagenesis and sequence alignment of protein homologues. *PLoS ONE*, **2012**, 7, e37922 | 3.7 | 4 |
| 51 | Functional effects of protein variants. *Biochimie*, **2021**, 180, 104-120 | 4.6 | 4 |
| 50 | Mu transpososome activity-profiling yields hyperactive MuA variants for highly efficient genetic and genome engineering. *Nucleic Acids Research*, **2018**, 46, 4649-4661 | 20.1 | 3 |
| 49 | VariOtator, a Software Tool for Variation Annotation with the Variation Ontology. *Human Mutation*, **2016**, 37, 344-9 | 4.7 | 3 |
| 48 | NDDVD: an integrated and manually curated Neurodegenerative Diseases Variation Database. *Database: the Journal of Biological Databases and Curation*, **2018**, 2018, | 5 | 3 |
| 47 | Assessing computational predictions of the phenotypic effect of cystathionine-beta-synthase variants. *Human Mutation*, **2019**, 40, 1530-1545 | 4.7 | 3 |
| 46 | Genetic tests need the Human Variome Project. *Genetic Testing and Molecular Biomarkers*, **2011**, 15, 3 | 1.6 | 3 |
| 45 | Accuracy of protein hydropathy predictions. *International Journal of Data Mining and Bioinformatics*, **2010**, 4, 735-54 | 0.5 | 3 |
| 44 | Immunological systems biology: gene expression analysis of B-cell development in Ramos B-cells. *Molecular Immunology*, **2007**, 44, 3537-51 | 4.3 | 3 |
| 43 | Fast Iterative Gene Clustering Based on Information Theoretic Criteria for Selecting the Cluster Structure. *Journal of Computational Biology*, **2004**, 11, 660-682 | 1.7 | 3 |
| 42 | Poikilosis - pervasive biological variation. *F1000Research*, **2020**, 9, 602 | 3.6 | 3 |

| | | | |
|---|---|---|---|
| 41 | Contribution of pseudogenes to sequence diversity. *Methods in Molecular Biology*, **2014**, 1167, 15-24 | 1.4 | 3 |
| 40 | Genetic Variation in Bruton Tyrosine Kinase. *Rare Diseases of the Immune System*, **2015**, 75-85 | 0.2 | 3 |
| 39 | Hematologically important mutations: The autosomal forms of chronic granulomatous disease (third update). *Blood Cells, Molecules, and Diseases*, **2021**, 92, 102596 | 2.1 | 3 |
| 38 | Establishment of an international database for genetic variants in esophageal cancer. *Annals of the New York Academy of Sciences*, **2016**, 1381, 45-49 | 6.5 | 2 |
| 37 | Conserved and quickly evolving immunome genes have different evolutionary paths. *Human Mutation*, **2012**, 33, 1456-63 | 4.7 | 2 |
| 36 | Primary Immunodeficiencies: Genotype Phenotype Correlations **2006**, 443-460 | | 2 |
| 35 | Bruton tyrosine kinase BTK in X-linked agammaglobulinemia XLA. *Frontiers in Bioscience - Landmark*, **2000**, 5, d917-928 | 2.8 | 2 |
| 34 | Structural consequences of neopullulanase mutations. *BBA - Proteins and Proteomics*, **1996**, 1295, 195-200 | | 2 |
| 33 | HYDRO: a program for protein hydropathy predictions. *Computer Methods and Programs in Biomedicine*, **1993**, 41, 121-9 | 6.9 | 2 |
| 32 | An algorithm for simultaneous comparison of several sequences. *Bioinformatics*, **1988**, 4, 89-92 | 7.2 | 2 |
| 31 | Simulation of the dynamics of primary immunodeficiencies in CD4+ T-cells. *PLoS ONE*, **2017**, 12, e0176509 | 3.7 | 2 |
| 30 | Strategy for Disease Diagnosis, Progression Prediction, Risk Group Stratification and Treatment – Case of COVID-19 | | 2 |
| 29 | Variation Benchmark Datasets: Update, Criteria, Quality and Applications | | 2 |
| 28 | Circulating Plasma microRNAs In Systemic Sclerosis-Associated Pulmonary Arterial Hypertension. *Rheumatology*, **2021**, | 3.9 | 2 |
| 27 | Systematics for types and effects of RNA variations. *RNA Biology*, **2021**, 18, 481-498 | 4.8 | 2 |
| 26 | The Importance of Proper Testing of Predictor Performance. *Human Mutation*, **2015**, 36, iii-iv | 4.7 | 1 |
| 25 | Evaluation of accuracy and applicability of protein models: retrospective analysis of biological and biomedical predictions. *In Silico Biology*, **2009**, 9, 307-31 | 2 | 1 |
| 24 | Problems with anti-plagiarism database. *Nature*, **2009**, 457, 26 | 50.4 | 1 |

| | | | |
|---|---|---|---|
| 23 | Detection of Pathogenic Mutation Prone Locations from Protein Sequences using Solvent Accessibility Measurements **2007**, | | 1 |
| 22 | Immunome, Immtree and Immunity: Databases for Systems Biology of Immune System **2007**, | | 1 |
| 21 | Combination of Gatekeeper Mutations and Cysteine 481 Replacement Causes Super Resistance to the Irreversible BTK Inhibitors Ibrutinib, Acalabrutinib and Zanubrutinib. *Blood*, **2019**, 134, 5759-5759 | 2.2 | 1 |
| 20 | How good are pathogenicity predictors in detecting benign variants? | | 1 |
| 19 | Measuring and interpreting pervasive heterogeneity, poikilosis. *FASEB BioAdvances*, **2021**, 3, 611-625 | 2.8 | 1 |
| 18 | PON-Sol2: Prediction of Effects of Variants on Protein Solubility. *International Journal of Molecular Sciences*, **2021**, 22, | 6.3 | 1 |
| 17 | Immunodeficiency, Primary: Affecting the Adaptive Immune System1-6 | | 1 |
| 16 | Parkinson's disease and multiple system atrophy patient iPSC-derived oligodendrocytes exhibit alpha-synuclein-induced changes in maturation and immune reactive properties.. *Proceedings of the National Academy of Sciences of the United States of America*, **2022**, 119, e2111405119 | 11.5 | 1 |
| 15 | Simulation of the Dynamics of Primary Immunodeficiencies in B Cells. *Frontiers in Immunology*, **2018** , 9, 1785 | 8.4 | 0 |
| 14 | Poikilosis □pervasive biological variation. *F1000Research*,9, 602 | 3.6 | 0 |
| 13 | Prognostic implications of troponin T variations in inherited cardiomyopathies using systems biology. *Npj Genomic Medicine*, **2021**, 6, 47 | 6.2 | 0 |
| 12 | Generic model for biological regulation. *F1000Research*,11, 419 | 3.6 | 0 |
| 11 | One Gene, Several Diseases: The Characteristics of Pleiotropic Proteins. *Human Mutation*, **2017**, 38, 241 | 4.7 | |
| 10 | Checklist for gene/disease-specific variation database curators to enable ethical data management. *Human Mutation*, **2019**, 40, 1634-1640 | 4.7 | |
| 9 | Clarity and claims in variation/mutation databasing. *Nature Biotechnology*, **2011**, 29, 790-2; author reply 792-4 | 44.5 | |
| 8 | Dynamic covariation between gene expression and genome characteristics. *Gene*, **2008**, 410, 53-66 | 3.8 | |
| 7 | Signal transduction-related bioinformatics services. *Briefings in Bioinformatics*, **2003**, 4, 325-31 | 13.4 | |
| 6 | The diverse genetic basis of immunodeficiencies **2003**, 45-76 | | |

5 Immunodeficiency mutation databases a new research tool. *Trends in Immunology*, **1996**, 17, 495-496

4 Bioinformatics in Proteomics **2003**, 419-428

3 Modelling of the 3D structure of Bacillus circulant var. alkalophilus cyclomaltodextrin glucanotransferase and comparison with amylase **1993**, 433-434

2 Mixed T- and B-Lymphocyte Deficiency Disorders1-7

1 Immunodeficiencies and Immunome: Diseases and Information Services **2009**, 71-85