A method and server for predicting damaging missense mutations

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441	ENPP1 Deficiency: A Clinical Update on the Relevance of Individual Variants Using a Locus-Specific Patient Database.	0
440	Germline Variants in 32 Cancer-Related Genes among 700 Chinese Breast Cancer Patients by Next-Generation Sequencing: A Clinic-Based, Observational Study. **2022**, 23, 11266	0

439	Identification of a pleiotropic effect of ADIPOQ on cardiac dysfunction and Alzheimer disease based on genetic evidence and health care records. **2022**, 12,	0
438	An SNN retrocopy insertion upstream of GPR22 is associated with dark red coat color in Poodles.	0
437	Hb Alessandria [7(C3)Trp->Leu; HBB: c.113G>T]: a Novel Variant on the Globin Chain with Slightly Increased Affinity for Oxygen Detected by Capillary Electrophoresis. 1-5	0
436	Tracing Bai-Yue ancestry in aboriginal Li people on Hainan Island.	0
435	Genotyping and In Silico Analysis of Delmarva (DMV/1639) Infectious Bronchitis Virus (IBV) Spike 1 (S1) Glycoprotein. **2022**, 13, 1617	0
434	A novel loss-of-function mutation of the voltage-gated potassium channel Kv10.2 involved in epilepsy and autism. **2022**, 17,	0
433	Comprehensive analysis predicting effects of deleterious SNPs of human progesterone receptor gene on its structure and functions: a computational approach. 1-16	0
432	Lymphoma as an Exclusion Criteria for CVID Diagnosis Revisited.	0
431	Two Loci Contribute to Age-Related Hearing Loss Resistance in the Japanese Wild-Derived Inbred MSM/Ms Mice. **2022**, 10, 2221	0
430	Investigation of TLR2 and TLR4 Polymorphisms and Sepsis Susceptibility: Computational and Experimental Approaches. **2022**, 23, 10982	1
429	Vasor: Accurate prediction of variant effects for amino acid substitutions in multidrug resistance protein 3.	0
428	Genetic drivers of Cushing disease: Frequency and associated phenotypes. **2022**,	0
427	Functional and Structural Impact of Deleterious Missense Single Nucleotide Polymorphisms in the NR3C1, CYP3A5, and TNF- Genes: An In Silico Analysis. **2022**, 12, 1307	1
426	Genetic analyses of the electrocardiographic QT interval and its components identify additional loci and pathways. **2022**, 13,	1
425	A Missense Variant in PLP2 in Holstein Cattle with X-Linked Congenital Mast Cell Tumor. **2022**, 12, 2329	0
424	Cardiomyocyte ploidy is dynamic during postnatal development and varies across genetic backgrounds.	0
423	Novel clinical, molecular and bioinformatics insights into the genetic background of autism. **2022**, 16,	0
422	A novel germline mutation of the <i>PALB</i> gene in a young Yakut breast cancer woman. **2022**, 21, 72-79	0

421	RMDisease V2.0: an updated database of genetic variants that affect RNA modifications with disease and trait implication.	0
420	Mutation screening of the CYP1B1 gene reveals thirteen novel disease-causing variants in consanguineous Pakistani families causing primary congenital glaucoma. **2022**, 17, e0274335	0
419	Rapid genome sequencing for pediatrics.	0
418	Predicting clinical phenotypes of metachromatic leukodystrophy based on the arylsulfatase A activity and the ARSA genotype? Chances and challenges. **2022**,	0
417	Computational approaches for predicting variant impact: An overview from resources, principles to applications. 13,	0
416	Novel variants and phenotypes in NEUROG3 associated syndrome.	0
415	Identification of a novel ESR1 mutation in a Chinese PCOS woman with estrogen insensitivity in IVF treatment.	0
414	Prevalence and Phenotypic Burden of Monogenic Arrhythmias Using Integration of Electronic Health Records With Genetics.	0
413	ortho_seqs: A Python tool for sequence analysis and higher order sequence–phenotype mapping.	0
412	Towards a Better Understanding of Genotype–Phenotype Correlations and Therapeutic Targets for Cardiocutaneous Genes: The Importance of Functional Studies above Prediction. **2022**, 23, 10765	0
411	The Clinical and Genetic Characteristics in Children with Idiopathic Hypogonadotropin Hypogonadism. **2022**, 2022, 1-12	0
410	Roles of neuroligins in central nervous system development: focus on glial neuroligins and neuron neuroligins. **2022**, 20,	0
409	Diagnosis of a Single-Nucleotide Variant in Whole-Exome Sequencing Data for Patients With Inherited Diseases: Machine Learning Study Using Artificial Intelligence Variant Prioritization. **2022**, 3, e37701	0
408	Pedigree-based study to identify GOLGB1 as a risk gene for bipolar disorder. **2022**, 12,	0
407	Never-homozygous genetic variants in healthy populations are potential recessive disease candidates. **2022**, 7,	0
406	Long-Surviving Adult Siblings With Joubert Syndrome Harboring a Novel Compound Heterozygous CPLANE1 Variant. **2022**, 8, e200031	0
405	High detection rate from genetic testing in BRCA-negative women with familial epithelial ovarian cancer. **2022**,	2
404	Common variants in GNL3 gene contributed the susceptibility of hand osteoarthritis in Han Chinese population. **2022**, 12,	1

403	Identification of variants in genes associated with autoinflammatory disorders in a cohort of patients with psoriatic arthritis. **2022**, 8, e002561	0
402	The role of excitatory amino acid transporter 2 (EAAT2) in epilepsy and other neurological disorders.	0
401	Repeated turnovers keep sex chromosomes young in willows. **2022**, 23,	1
400	DPP9 deficiency: An inflammasomopathy that can be rescued by lowering NLRP1/IL-1 signaling. **2022**, 7,	1
399	Structure-Based Pathogenicity Relationship Identifier (SPRI): A Novel Structure-Guided Method to Evaluate Pathological Effect of Missense Mutations.	0
398	In silico analyses of Wnt1 nsSNPs reveal structurally destabilizing variants, altered interactions with Frizzled receptors and its deregulation in tumorigenesis. **2022**, 12,	0
397	P. Ala278Val mutation might cause a pathogenic defect in HEXB folding leading to the Sandhoff disease.	0
396	Whole-genome sequencing of multiple related individuals with type 2 diabetes reveals an atypical likely pathogenic mutation in the PAX6 gene.	0
395	Exome sequencing in a Swedish family with PMS2 mutation with varying penetrance of colorectal cancer: investigating the presence of genetic risk modifiers in colorectal cancer risk. Publish Ahead of Print,	0
394	Rare surfactant-related variants in familial and sporadic pulmonary fibrosis.	0
393	Pleiotropic modifiers of age-related diabetes and neonatal intestinal obstruction in cystic fibrosis. **2022**, 109, 1894-1908	1
392	A Novel Missense PRKAR1A Variant Causes Carney Complex.	0
391	Screening of candidate genes at GLC3B and GLC3C loci in Chinese primary congenital glaucoma patients with targeted next generation sequencing. 1-6	0
390	Unsupervised co-optimization of a graph neural network and a knowledge graph embedding model to prioritize causal genes for Alzheimers Disease.	0
389	A whole genome sequencing approach to anterior cruciate ligament rupture□ twin study in two unrelated families. **2022**, 17, e0274354	0
388	Molecular Dynamic Simulation of Neurexin1□Mutations Associated with Mental Disorder.	0
387	Protein Language Model Predicts Mutation Pathogenicity and Clinical Prognosis.	0
386	Genomics technologies and bioinformatics in allergy and immunology. **2022**, 221-260	0

385	Novel Titin Gene Mutation Causing Autosomal Dominant Limb-Girdle Muscular Dystrophy. **2022**,	0
384	Description of the molecular and phenotypic spectrum in Chinese patients with aggrecan deficiency: Novel ACAN heterozygous variants in eight Chinese children and a review of the literature. 13,	0
383	Cumulative evidence for associations between genetic variants in interleukin 17 family gene and risk of human diseases. 13,	0
382	ImprovedKCNQ2gene missense variant interpretation with artificial intelligence.	0
381	Pathogenic variants of ornithine transcarbamylase deficiency: Nation-wide study in Japan and literature review. 13,	0
380	Autosomal dominant STAT6 gain of function causes severe atopy associated with lymphoma.	0
379	E-SNPs&GO: embedding of protein sequence and function improves the annotation of human pathogenic variants.	0
378	The PINK1 p.Asn521Thr Variant Is Associated with Earlier Disease Onset in GRN/C9orf72 Frontotemporal Lobar Degeneration. **2022**, 23, 12847	0
377	Leveraging global multi-ancestry meta-analysis in the study of idiopathic pulmonary fibrosis genetics. **2022**, 2, 100181	0
376	Channel HCN4 mutation R666Q associated with sporadic arrhythmia decreases channel electrophysiological function and increases protein degradation. **2022**, 102599	0
375	Frontotemporal degeneration genetic risk loci and transcription regulation as a possible mechanistic link to disease risk. **2022**, 101, e31078	0
374	Expanding the genetic spectrum for Chinese familial hypercholesterolemia population with six genetic mutations identified using a next-generation sequencing-based laboratory-developed screening test.	0
373	The Combination of Salmonella Phage ST-3 and Antibiotics to Prevent Salmonella Typhimurium In Vitro. **2022**, 79,	0
372	Integrative genomic analysis of childhood acute lymphoblastic leukaemia lacking a genetic biomarker in the UKALL2003 clinical trial..	0
371	POU6F2mutation identified in humans with pubertal failure shifts isoform formation and alters GnRH transcript expression.	0
370	Rare Germline ATM Variants Influence the Development of Chronic Lymphocytic Leukemia.	0
369	The fly homolog of SUPT16H, a gene associated with neurodevelopmental disorders, is required in a cell-autonomous fashion for cell survival.	1
368	A missense, loss-of-function YARS1 variant in a patient with proximal-predominant motor neuropathy. mcs.a006246	0

367	PCNA thermosensitivity underlies an Ataxia Telangiectasia-like disorder.	0
366	The determination of the effect(s) of solute carrier family 22-member 2 (SLC22A2) haplotype variants on drug binding via molecular dynamic simulation systems. **2022**, 12,	0
365	MetaRNN: differentiating rare pathogenic and rare benign missense SNVs and InDels using deep learning. **2022**, 14,	0
364	Evolutionary coupling analysis guides identification of mistrafficking-sensitive variants in cardiac K+ channels: Validation with hERG. 13,	0
363	A congenital CSF3R mutation in chronic neutropenia reveals a vital role for a cytokine receptor extracellular hinge motif in the response to granulocyte colony-stimulating factor.	0
362	HINT1 neuropathy in Lithuania: clinical, genetic, and functional profiling. **2022**, 17,	0
361	Candidate genes and sequence variants for susceptibility to mycobacterial infection identified by whole-exome sequencing. 13,	0
360	Identification of potential driver mutations in glioblastoma using machine learning.	0
359	The role of contactin-associated protein-like 2 in neurodevelopmental disease and human cerebral cortex evolution. 15,	0
358	Eosinophilic Infiltration of the Sino-Atrial Node in Sudden Cardiac Death Caused by Long QT Syndrome. **2022**, 23, 11666	0
357	Matrix metalloproteinase gene mutations and bioinformatics of telocytes in hepatocellular carcinoma.	0
356	Genomic characteristics of two breast malignant phyllodes tumors during pregnancy and lactation identified through whole-exome sequencing. **2022**, 17,	0
355	Anterior Umbilication of Lens in a Family with Congenital Cataracts Associated with a Missense Mutation of MIP Gene. **2022**, 13, 1987	0
354	Mild phenotypes of phosphoglycerate dehydrogenase deficiency by a novel mutation of PHGDH gene: case report and literature review.	0
353	Genetic and Clinical Spectrum of GNE Myopathy in Russia. **2022**, 13, 1991	0
352	Gene Sequencing Identifies Perturbation in Nitric Oxide Signaling as a Nonlipid Molecular Subtype of Coronary Artery Disease.	0
351	Predictive functional, statistical and structural analysis of CSNK2A1 and CSNK2B variants linked to neurodevelopmental diseases. 9,	0
350	Novel pathogenic variant (c.2947C > T) of the carbamoyl phosphate synthetase 1 gene in neonatal-onset deficiency. 16,	0

349	Disease-associated mutations in WDR34 lead to diverse impacts on the assembly and function of dynein-2.	0
348	PhenoScore: AI-based phenomics to quantify rare disease and genetic variation.	1
347	Intragenic compensation through the lens of deep mutational scanning. **2022**, 14, 1161-1182	0
346	Comprehensive In Silico Functional Prediction Analysis of CDKL5 by Single Amino Acid Substitution in the Catalytic Domain. **2022**, 23, 12281	0
345	Conformational Stability and Denaturation Processes of Proteins Investigated by Electrophoresis under Extreme Conditions. **2022**, 27, 6861	0
344	A Novel Homozygous Founder Variant of RTN4IP1 in Two Consanguineous Saudi Families. **2022**, 11, 3154	0
343	Visual inspection reveals a novel pathogenic mutation in PKD1 missed by the variant caller in whole-exome sequencing. **2022**, 26,	0
342	Exome sequencing identified five novel USH2A variants in Korean patients with retinitis pigmentosa. 1-8	0
341	Unusual Dependence between Gene Expression and Negative Selection in Euplotes.	0
340	Analysis of von Willebrand Disease in the Heart of Europe**2022**, 06, e335-e346	0
339	Global population genetics and diversity in the TAS2R bitter taste receptor family. 13,	1
338	Bowhead NEIL1: molecular cloning, characterization, and enzymatic properties. **2022**,	0
337	Biallelic variants in PIGN cause Fryns syndrome, multiple congenital anomalies-hypotonia-seizures syndrome, and neurologic phenotypes: A genotypephenotype correlation study. **2022**,	0
336	Parallel functional annotation of cancer-associated missense mutations in histone methyltransferases. **2022**, 12,	0
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334	Ceramide analysis in combination with genetic testing may provide a precise diagnosis for self-healing collodion babies. **2022**, 100308	0
333	Delineation of a KDM2B-related neurodevelopmental disorder and its associated DNA methylation signature. **2022**,	0
332	Expanding SPTAN1 monoallelic variant associated disorders: From epileptic encephalopathy to pure spastic paraplegia and ataxia. **2022**,	1

331	Concurrent Pathogenic Variants of BRCA1, MUTYH and CHEK2 in a Hereditary Cancer Family. **2022**,	0
330	Evaluation of in silico pathogenicity prediction tools for the classification of small in-frame indels.	0
329	Human Hereditary Cardiomyopathy Shares a Genetic Substrate With Bicuspid Aortic Valve.	0
328	Characterization of Arabian Peninsula whole exomes: Contributing to the catalogue of human diversity. **2022**, 25, 105336	0
327	Deep Molecular and In Silico Protein Analysis of p53 Alteration in Myelodysplastic Neoplasia and Acute Myeloid Leukemia. **2022**, 11, 3475	0
326	A rare PRSS1 p.S127C mutation is associated with chronic pancreatitis and causes misfolding-induced ER-stress. **2022**,	0
325	Comprehensive in silico prioritization of pathogenic nsSNPs in human □adducin gene towards finding its relation with cancer. **2022**, 34, 201119	0
324	Novel mutations in TUBB8 and ZP3 cause human oocyte maturation arrest and female infertility. **2022**, 279, 132-139	0
323	In silico assessment of missense point mutations on human cathelicidin LL-37. **2023**, 118, 108368	0
322	Identification of prognostic factors in classic Hodgkin lymphoma by integrating whole slide imaging and next generation sequencing.	0
321	A missense variant in NCF1 is associated with susceptibility to unexplained recurrent spontaneous abortion. **2022**, 17, 1443-1450	0
320	Diagnostic reasoning in neurogenetics: a general approach. **2022**, 80, 944-952	0
319	An in vitro and computational validation of a novel loss-of-functional mutation in PAX9 associated with non-syndromic tooth agenesis.	0
318	Counterintuitive structural and functional effects due to naturally-occurring mutations targeting the active site of the disease-associated NQO1 enzyme.	1
317	Theme 02 - Genetics and Genomics. **2022**, 23, 40-56	0
316	Spectrum of DDC variants causing aromatic l-amino acid decarboxylase (AADC) deficiency and pathogenicity interpretation using ACMG-AMP/ACGS recommendations. **2022**,	0
315	Genomic integration and expression of Felis catus papillomavirus type 2 oncogenes in feline Merkel cell carcinoma. 030098582211391	0
314	A Comprehensive Study of De Novo Mutations on the Protein-Protein Interaction Interfaces Provides New Insights into Developmental Delay. **2022**, 12, 1643	0

313	Reconstructing mutational lineages in breast cancer by multi-patient-targeted single-cell DNA sequencing. **2022**, 100215	0
312	FAVOR: functional annotation of variants online resource and annotator for variation across the human genome.	0
311	Mutations in the tail domain of the neurofilament heavy chain gene increase the risk of amyotrophic lateral sclerosis.	0
310	A bioinformatics approach to the identification of novel deleterious mutations of human TPMT through validated screening and molecular dynamics. **2022**, 12,	1
309	Analysis of 1276 Haplotype-Resolved Genomes Allows Characterization of Cis- and Trans-Abundant Genes. **2023**, 237-272	0
308	Population Genomics for Insect Conservation. **2023**, 11,	1
307	Identification and Characterization of Novel Mutations in Chronic Kidney Disease (CKD) and Autosomal Dominant Polycystic Kidney Disease (ADPKD) in Saudi Subjects by Whole-Exome Sequencing. **2022**, 58, 1657	0
306	Clinical and Genetic Characterization of Patients with Artemis Deficiency in Japan.	0
305	ASCARIS: Positional Feature Annotation and Protein Structure-Based Representation of Single Amino Acid Variations.	0
304	In Silico Evaluation of Nonsynonymous SNPs in Human ADAM33: The Most Common Form of Genetic Association to Asthma Susceptibility. **2022**, 2022, 1-29	0
303	Bilateral Nonsyndromic Sensorineural Hearing Loss Caused by a NARS2 Mutation. **2022**,	0
302	Genetic and clinical characteristics of ALS patients with NEK1 gene variants. **2022**,	0
301	Germline selection of PTPN11 (HGNC:9644) variants make a major contribution to both Noonan syndrome's high birth rate and the transmission of sporadic cancer variants resulting in fetal abnormality.	0
300	Single-cell genome-wide association reveals that a nonsynonymous variant in ERAP1 confers increased susceptibility to influenza virus. **2022**, 2, 100207	0
299	Potential CRISPR Base Editing Therapeutic Options in a Sorsby Fundus Dystrophy Patient. **2022**, 13, 2103	0
298	Diagnostic Efficacy of Genetic Studies in a Series of Hereditary Cerebellar Ataxias in Eastern Spain. **2022**, 8, e200038	0
297	Episignature Mapping of TRIP12 Provides Functional Insight into Clark Baraitser Syndrome. **2022**, 23, 13664	0
296	TREML2 Gene Expression and Its Missense Variant rs3747742 Associate with White Matter Hyperintensity Volume and Alzheimer's Disease-Related Brain Atrophy in the General Population. **2022**, 23, 13764	0

295	Mutation analysis performed on tumor biopsies from patients with newly-diagnosed germinal center aggressive B cell lymphomas. **2022**, 13, 1237-1244	0
294	Carrier frequency and incidence estimation of familial hemophagocytic lymphohistiocytosis in East Asian populations by genome aggregation database (gnomAD) based analysis. 10,	0
293	Next-generation sequencing of postmortem molecular markers to support for medicolegal autopsy. **2022**, 6, 100300	0
292	Identification of a novel ESR1 mutation in a Chinese PCOS woman with estrogen insensitivity in IVF treatment. **2022**, 20,	0
291	Cross-protein transfer learning substantially improves zero-shot prediction of disease variant effects.	0
290	Pharmacogenomics of GLP-1 receptor agonists: a genome-wide analysis of observational data and large randomised controlled trials. **2023**, 11, 33-41	1
289	Novel variants in GABAA receptor subunits: A possible association with benzodiazepine resistance in patients with drug-resistant epilepsy. **2023**, 189, 107056	0
288	Mutant MESD links cellular stress to type I collagen aggregation in osteogenesis imperfecta type XX. **2023**, 115, 81-106	0
287	Molecular mechanisms underlying the defects of two novel mutations in the HSD17B3 gene found in the Tunisian population. **2023**, 227, 106235	0
286	OAF: a new member of the BRICHOS family. **2022**, 2,	0
285	Reporting Two Novel Mutations in Two Iranian Families with Cystic Fibrosis, Molecular and Bioinformatic Analysis. **2022**, 26, 398-405	0
284	Structural characterization and conformational dynamics of alpha-1 antitrypsin pathogenic variants causing alpha-1-antitrypsin deficiency. 9,	0
283	Somatic Variants in SVIL in Cerebral Aneurysms. **2022**, 8, e200040	0
282	Inferring the effects of protein variants on protein–protein interactions with an interpretable deep-learning model.	0
281	Insilico prediction and functional analysis of nonsynonymous SNPs in human CTLA4 gene. **2022**, 12,	0
280	Congenital Stationary Night Blindness: Clinical and Genetic Features. **2022**, 23, 14965	0
279	Mendelian Disorders in an Interstitial Cystitis/Bladder Pain Syndrome Cohort. 2200013	0
278	Screening of high-risk deleterious missense variations in the CYP1B1 gene implicated in the pathogenesis of primary congenital glaucoma: A comprehensive in silico approach. 10, e14132	1

277	Genetic analysis of a novel SUMF1 variation associated with a late infantile form of multiple sulfatase deficiency. **2022**, 36,	0
276	Molecular epidemiology analysis of symptomatic and asymptomatic norovirus infections in Chinese infants.	0
275	Neuron-specific protein network mapping of autism risk genes identifies shared biological mechanisms and disease-relevant pathologies. **2022**, 41, 111678	1
274	Novel and Founder Pathogenic Variants in X-Linked Alport Syndrome Families in Greece. **2022**, 13, 2203	0
273	Arrhythmogenic cardiomyopathy is under-recognized in end-stage pediatric heart failure: A 36-year single-center experience.	0
272	Genetic analysis and clinical features of three Chinese patients with Oguchi disease.	0
271	Loss of WNT4 in the gubernaculum causes unilateral cryptorchidism and fertility defects. **2022**, 149,	0
270	Somatic mutations in tumor and plasma of locoregional recurrent and/or metastatic head and neck cancer using a next-generation sequencing panel: A preliminary study.	0
269	BiallelicSTAB1pathogenic variants cause hereditary hyperferritinemia.	0
268	Genetic Variants in ARHGEF6 Cause Congenital Anomalies of the Kidneys and Urinary Tract in Humans, Mice, and Frogs. ASN.2022010050	0
267	Efficacy of low-dose hCG on FET cycle in patients with recurrent implantation failure. 13,	0
266	Case report: Identification of three novel compound heterozygous SGLT2 variants in three Chinese pediatric patients with familial renal glucosuria. 10,	0
265	Analysis of 363 variants in the coagulation Factor 5 gene via an interactive web database reveals new insights into FV deficiency and FV Leiden.	0
264	Computational Analysis Reveals the Temporal Acquisition of Pathway Alterations during the Evolution of Cancer. **2022**, 14, 5817	0
263	A likely HOXC4 predisposition variant for Chiari malformations. **2022**, 1-9	1
262	A High-resolution Haplotype-resolved Reference Panel Constructed from the China Kadoorie Biobank Study.	0
261	Analysis of ProP1 Gene in a Cohort of Tunisian Patients with Congenital Combined Pituitary Hormone Deficiency. **2022**, 11, 7525	1
260	Evaluation of in silico predictors on short nucleotide variants in HBA1, HBA2, and HBB associated with haemoglobinopathies. 11,	0

259	Genome-wide association study reveals loci with sex-specific effects on plasma bile acids.	0
258	Genetic Interaction Between GABRA1 and ERBB4 Variants in the Pathogenesis of Genetic Generalized Epilepsy. **2022**, 107070	0
257	De NovoVariants inMRTFBhave gain of function activity inDrosophilaand are associated with a novel neurodevelopmental phenotype with dysmorphic features.	0
256	Analysis of SLC26A4, FOXI1, and KCNJ10 Gene Variants in Patients with Incomplete Partition of the Cochlea and Enlarged Vestibular Aqueduct (EVA) Anomalies. **2022**, 23, 15372	1
255	Analysis of single-nucleotide polymorphisms in genes associated with triple-negative breast cancer. 13,	0
254	HBD-2 variants and SARS-CoV-2: New insights into inter-individual susceptibility. 13,	0
253	An ELF4 hypomorphic variant results in NK cell deficiency. **2022**, 7,	0
252	DDI2 promotes tumor metastasis and resists antineoplastic drugs-induced apoptosis in colorectal cancer.	0
251	The Genetic Spectrum of Maturity-Onset Diabetes of the Young (MODY) in Qatar, a Population-Based Study. **2023**, 24, 130	0
250	Diverse monogenic subforms of human spermatogenic failure. **2022**, 13,	1
249	Pitfalls of whole exome sequencing in undefined clinical conditions with a suspected genetic etiology.	0
248	Genetic analysis of the ATP11B gene in Chinese Han population with cerebral small vessel disease. **2022**, 23,	0
247	Unraveling the Structural Changes in the DNA-Binding Region of Tumor Protein p53 (TP53) upon Hotspot Mutation p53 Arg248 by Comparative Computational Approach. **2022**, 23, 15499	0
246	Genetic Factors Causing Thyroid Dyshormonogenesis as the Major Etiologies for Primary Congenital Hypothyroidism: Clinical and Genetic Characterization of 33 Patients. **2022**, 11, 7313	0
245	A Translational Approach to Spinal Neurofibromatosis: Clinical and Molecular Insights from a Wide Italian Cohort. **2023**, 15, 59	0
244	Optical genome mapping and revisiting short-read genome sequencing data reveal previously overlooked structural variants disrupting retinal disease□ssociated genes. **2022**, 100345	1
243	ACE2 polymorphisms impact COVID-19 severity in obese patients. **2022**, 12,	2
242	Genetic variants in African-American and Hispanic patients with breast cancer. **2022**, 25,	0

241	A novel frameshift variant of LMX1A that leads to autosomal dominant nonsyndromic sensorineural hearing loss: functional characterization of the C-terminal domain in LMX1A.	0
240	MUG: a mutation overview of GPCR subfamily A17 receptors. **2022**,	0
239	Defensin 19/119 mediates sperm chemotaxis and is associated with idiopathic infertility. **2022**, 3, 100825	0
238	Cylicins are a structural component of the sperm calyx being indispensable for male fertility in mice and human.	0
237	Structural Evaluation and Conformational Dynamics of ZNF141T474I Mutation Provoking Postaxial Polydactyly Type A. **2022**, 9, 749	1
236	Whole genome sequencing identifies rare variants inANK1, LRRN1, HAS1, and other genes and gene regulatory regions for stroke in type 1 diabetes.	0
235	TogoVar: A comprehensive Japanese genetic variation database. **2022**, 9,	0
234	A multiple sclerosis protective coding variant reveals an essential role for HDAC7 in regulatory T cells. **2022**, 14,	1
233	In Silico Profiling of Non-Synonymous SNPs of Fat Mass and Obesity- Associated Gene: Possible impacts on the treatment of Non-Alcoholic Fatty Liver Disease.	0
232	Targeted deep sequencing analyses of long QT syndrome in a Japanese population. **2022**, 17, e0277242	0
231	Identification and drug metabolic characterization of four new CYP2C9 variants CYP2C9*72-*75 in the Chinese Han population. 13,	0
230	Rare Variants in Genes of the Cholesterol Pathway Are Present in 60% of Patients with Acute Myocardial Infarction. **2022**, 23, 16127	0
229	Genetic architecture and evolution of color variation in American black bears. **2022**,	0
228	Integrative genomic analysis of childhood acute lymphoblastic leukaemia lacking a genetic biomarker in the UKALL2003 clinical trial.	0
227	An allelic series rare variant association test for candidate gene discovery.	0
226	TranceptEVE: Combining Family-specific and Family-agnostic Models of Protein Sequences for Improved Fitness Prediction.	0
225	The complex etiology of Epilepsy: Genetic analysis and HLA association in patients in the Middle East.	0
224	A Whole-Genome Sequencing Study Implicates GRAMD1B in Multiple Sclerosis Susceptibility. **2022**, 13, 2392	0

223	De novo mutation hotspots in homologous protein domains identify function-altering mutations in neurodevelopmental disorders. **2022**,	0
222	Rubbing Salt in the Wound: Molecular Evolutionary Analysis of Pain-Related Genes Reveals the Pain Adaptation of Cetaceans in Seawater. **2022**, 12, 3571	0
221	Effects of the pathological E200K mutation on human prion protein: A computational screening and molecular dynamics approach.	0
220	Calibration of computational tools for missense variant pathogenicity classification and ClinGen recommendations for PP3/BP4 criteria. **2022**, 109, 2163-2177	2
219	Rapid Targeted Sequencing Using Dried Blood Spot Samples for Patients With Suspected Actionable Genetic Diseases. **2023**, 43, 280-289	1
218	Computational analysis of missense variant CYP4F2*3 (V433M) in association with human CYP4F2 dysfunction: A functional and structural impact.	0
217	Heterozygous mutations in SOX2 may cause idiopathic hypogonadotropic hypogonadism via dominant-negative mechanisms.	0
216	Whole-exome sequencing identifies FANC heterozygous germline mutation as an adverse factor for immunosuppressive therapy in Chinese aplastic anemia patients aged 40 or younger: a single-center retrospective study.	0
215	SwissGenVar: A platform for clinical grade interpretation of genetic variants to foster personalized health care in Switzerland.	0
214	The mutation spectrum of SLC25A13 gene in citrin deficiency: identification of novel mutations in Vietnamese pediatric cohort with neonatal intrahepatic cholestasis.	0
213	Case report: KPTN gene-related syndrome associated with a spectrum of neurodevelopmental anomalies including severe epilepsy. 13,	0
212	Quantitative differentiation of benign and misfolded glaucoma-causing myocilin variants on the basis of protein thermal stability. **2023**, 16,	0
211	Comprehensive in vitro and in silico assessments of metabolic capabilities of 24 genomic variants of CYP2C19 using two different substrates. 14,	0
210	Identification and characterization of novel compound heterozygous variants in FSHR causing primary ovarian insufficiency with resistant ovary syndrome. 13,	0
209	Successful Treatment of Large B-Cell Lymphoma in a Child with Compound Heterozygous Mutation in the ATM Gene. **2023**, 24, 1099	0
208	Identification of a Novel Non-Canonical Splice-Site Variant in ABCD1. **2023**, 12, 473	0
207	Systemic structural analysis of alterations reveals a common structural basis of driver mutations in cancer. **2023**, 5,	0
206	A novel NONO variant that causes developmental delay and cardiac phenotypes. **2023**, 13,	0

205	LRP6 High Bone Mass Characterized In Two Generations Harboring A Unique Mutation Of Low-Density Lipoprotein Receptor-Related Protein 6.	0
204	Poor prognostic implications of myelodysplasia-related mutations in both older and younger patients with de novo AML. **2023**, 13,	0
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