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79	Impact of EFEMP1 on the survival outcome of biliary atresia in Thai infants. 2022, 12,	0
78	Association of rs2072446 in the NGFR gene with the risk of Alzheimer's disease and amyloid-□ deposition in the brain.	1
77	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,	О
76	Pleiotropic modifiers of age-related diabetes and neonatal intestinal obstruction in cystic fibrosis. 2022 , 109, 1894-1908	1
75	Loss-of-function mutations in CEP78 cause male infertility in humans and mice. 2022, 8,	2
74	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,	O
73	E-SNPs&GO: embedding of protein sequence and function improves the annotation of human pathogenic variants.	0
72	Characterization of hotspot exonuclease domain mutations in the DNA polymerase? gene in endometrial cancer. 12,	O
71	The fly homolog of SUPT16H, a gene associated with neurodevelopmental disorders, is required in a cell-autonomous fashion for cell survival.	1
70	Dermokine mutations contribute to epithelial-mesenchymal transition and advanced melanoma through ERK/MAPK pathways.	O
69	Candidate genes and sequence variants for susceptibility to mycobacterial infection identified by whole-exome sequencing. 13,	0
68	Novel pathogenic variant (c.2947C > T) of the carbamoyl phosphate synthetase 1 gene in neonatal-onset deficiency. 16,	O
67	PhenoScore: Al-based phenomics to quantify rare disease and genetic variation.	1
66	Conformational Stability and Denaturation Processes of Proteins Investigated by Electrophoresis under Extreme Conditions. 2022 , 27, 6861	O
65	A Novel Homozygous Founder Variant of RTN4IP1 in Two Consanguineous Saudi Families. 2022 , 11, 3154	0
64	Exome-wide association study to identify rare variants influencing COVID-19 outcomes: Results from the Host Genetics Initiative. 2022 , 18, e1010367	О

63	Brain iron deposition and whole-exome sequencing of non-Wilson-disease hypoceruloplasminemia in a family. 2022 , 100027	O
62	In Silico Study of ULK1 Gene as a Susceptible Biomarker for Neurodegeneration.	O
61	A missense variant in NCF1 is associated with susceptibility to unexplained recurrent spontaneous abortion. 2022 , 17, 1443-1450	О
60	Bilateral Nonsyndromic Sensorineural Hearing Loss Caused by a NARS2 Mutation. 2022,	O
59	PRDM1 DNA-binding zinc finger domain is required for normal limb development and is disrupted in Split Hand/Foot Malformation.	O
58	Phenotypic screening models for rapid diagnosis of genetic variants and discovery of personalized therapeutics. 2022 , 101153	O
57	Isolation, identification and application of Aspergillus oryzae BL18 with high protease activity as starter culture in doubanjiang (broad bean paste) fermentation. 2023 , 51, 102225	0
56	Genetic aetiology of Down syndrome birth: novel variants of maternal DNMT3B and RFC1 genes increase risk of meiosis II nondisjunction in the oocyte.	O
55	Loss of WNT4 in the gubernaculum causes unilateral cryptorchidism and fertility defects. 2022, 149,	О
54	Clinical heterogeneity of hyperornithinemia-hyperammonemia-homocitrullinuria syndrome in thirteen palestinian patients and report of a novel variant in the SLC25A15 gene. 13,	O
53	Proteogenomic insights into the biology and treatment of pancreatic ductal adenocarcinoma. 2022 , 15,	0
52	Genome-wide association study reveals loci with sex-specific effects on plasma bile acids.	O
51	Diverse monogenic subforms of human spermatogenic failure. 2022 , 13,	1
50	Genetic analysis of the ATP11B gene in Chinese Han population with cerebral small vessel disease. 2022 , 23,	O
49	TranceptEVE: Combining Family-specific and Family-agnostic Models of Protein Sequences for Improved Fitness Prediction.	0
48	Rapid Targeted Sequencing Using Dried Blood Spot Samples for Patients With Suspected Actionable Genetic Diseases. 2023 , 43, 280-289	1
47	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.	O
46	A novel NONO variant that causes developmental delay and cardiac phenotypes. 2023 , 13,	O

45	Dermokine mutations contribute to epithelial-mesenchymal transition and advanced melanoma through ERK/MAPK pathways.	O
44	A biallelic variant in POLR2C is associated with congenital hearing loss and male infertility: Case report.	O
43	A novel frameshift mutation in TRPV6 is associated with hereditary pancreatitis. 13,	O
42	Primary failure of eruption: From molecular diagnosis to therapeutic management. 2023, 13, 169-176	O
41	Dendritic Spine in Autism Genetics: Whole-Exome Sequencing Identifying De Novo Variant of CTTNBP2 in a Quad Family Affected by Autism Spectrum Disorder. 2023 , 10, 80	О
40	Exome sequencing reveals aggregates of rare variants in glycosyltransferase and other genes influencing immunoglobulin G and transferrin glycosylation.	O
39	Proteogenomics of diffuse gliomas reveal molecular subtypes associated with specific therapeutic targets and immune-evasion mechanisms. 2023 , 14,	O
38	Haploinsufficiency as a Foreground Pathomechanism of Poirer-Bienvenu Syndrome and Novel Insights Underlying the Phenotypic Continuum of CSNK2B-Associated Disorders. 2023 , 14, 250	O
37	The PER3rs772027021 SNP induces pigmentation phenotypes of dyschromatosis universalis hereditaria. 2023 , 101, 279-294	O
36	A heterozygous missense variant in DLX3 leads to uterine leiomyomas and pregnancy losses in a consanguineous Iranian family. 2023 , 865, 147292	O
35	Whole-exome sequencing and electrophysiological study reveal a novel loss-of-function mutation of KCNA10 in epinephrine provoked long QT syndrome with familial history of sudden cardiac death. 2023 , 62, 102245	O
34	An inclusive study of deleterious missense PAX9 variants using user-friendly tools reveals structural, functional alterations, as well as potential therapeutic targets 2023 , 233, 123375	O
33	Deep genomic analysis of malignant peripheral nerve sheath tumor cell lines challenges current malignant peripheral nerve sheath tumor diagnosis. 2023 , 26, 106096	O
32	Common and rare variant associations with latent traits underlying depression, bipolar disorder, and schizophrenia. 2023 , 13,	O
31	The Usher syndrome 1C protein harmonin regulates canonical Wnt signaling. 11,	О
30	Model performance and interpretability of semi-supervised generative adversarial networks to predict oncogenic variants with unlabeled data. 2023 , 24,	O
29	Analysis of Elinical exomelpanel in Serbian patients with cognitive disorders. 2022, 54, 1351-1364	O
28	Whole-Exome Sequencing Analyses Support a Role of Vitamin D Metabolism in Ischemic Stroke. 2023 , 54, 800-809	O

27	A homozygous KASH5 frameshift mutation causes diminished ovarian reserve, recurrent miscarriage, and non-obstructive azoospermia in humans. 14,	0
26	Whole Exome Sequencing Reveals Novel Candidate Genes in Familial Forms of Glaucomatous Neurodegeneration. 2023 , 14, 495	О
25	N6-methyladenosine modification in 18S rRNA promotes tumorigenesis and chemoresistance via HSF4b/HSP90B1/mutant p53 axis. 2023 , 30, 144-158.e10	О
24	Genetic background of idiopathic neurodevelopmental delay patients with significant brain deviation volume. Publish Ahead of Print,	О
23	Evaluation of AlphaFold structure-based protein stability prediction on missense variations in cancer. 14,	О
22	Computational and mitochondrial functional studies of novel compound heterozygous variants in SPATA5 gene support a causal link with epileptogenic encephalopathy. 2023 , 17,	О
21	Evaluation of the Function of a Rare Variant in the 3'-Untranslated Region of the EGlobin Gene. 2022 , 46, 312-316	O
20	Whole exome sequencing in unexplained recurrent miscarriage families identified novel pathogenic genetic causes of euploid miscarriage.	O
19	Novel and recurrent genetic variants of VHL, SDHB, and RET genes in Chinese pheochromocytoma and paraganglioma patients. 14,	О
18	Identification of putative regulatory single-nucleotide variants in NTN1 gene associated with NSCL/P.	O
17	Novel mutations in the ABCD1 gene caused adrenomyeloneuropathy in the Chinese population. 14,	О
16	Identification of de novo Mutations in the Chinese Autism Spectrum Disorder Cohort via Whole-Exome Sequencing Unveils Brain Regions Implicated in Autism.	O
15	Genome Analysis Using Whole-Exome Sequencing of Non-Syndromic Cleft Lip and/or Palate from Malagasy Trios Identifies Variants Associated with Cilium-Related Pathways and Asian Genetic Ancestry. 2023 , 14, 665	O
14	Hypercholesterolemia in the Malaysian Cohort Participants: Genetic and Non-Genetic Risk Factors. 2023 , 14, 721	О
13	Mutational investigation of 17 causative genes in a cohort of 113 families with nonsyndromic early-onset high myopia in northwestern China.	О
12	PredDSMC: A predictor for driver synonymous mutations in human cancers. 14,	O
11	Whole exome sequencing reveals novel variants associated with diminished ovarian reserve in young women. 14,	О
10	A Novel Heterozygous Missense Variant in Parathyroid Hormone 1 is Related to the Occurrence of Developmental Dysplasia of the Hip. 2023 , 27, 74-80	О

9	Association of LHCGR gene variant rs2293275 with the polycystic ovary syndrome risk: an updated meta-analysis.	O
8	Clinical phenotyping and genetic diagnosis of a large cohort of Sudanese families with hereditary spinocerebellar degenerations.	O
7	Large-scale analysis of de novo mutations identifies risk genes for female infertility characterized by oocyte and early embryo defects. 2023 , 24,	0
6	Novel mutations reduce expression of meiotic regulators SYCE1 and BOLL in testis of azoospermic men from West Bengal, India.	O
5	GenomicEranscriptomic evolution in lung cancer and metastasis. 2023, 616, 543-552	0
4	The evolution of lung cancer and impact of subclonal selection in TRACERx. 2023 , 616, 525-533	O
3	PRDM1 DNA-binding zinc finger domain is required for normal limb development and is disrupted in split hand/foot malformation. 2023 , 16,	O
2	Whole-exome sequencing in moyamoya patients of Northern-European origin identifies gene variants involved in Nitric Oxide metabolism: A pilot study. 2023 , 101745	O
1	Genetic, clinical, and pathological study of patients with severe hypertension-associated renal microangiopathy.	O