Elon Pras

List of Publications by Year
in descending order

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Version: 2024-02-01

46	1,999	516710 16	276875 41
papers	citations	h-index	g-index

48	48	48	3622
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Whole-exome sequencing in undiagnosed genetic diseases: interpreting 119 trios. Genetics in Medicine, 2015, 17, 774-781.	2.4	284
2	Non-type I cystinuria caused by mutations in SLC7A9, encoding a subunit (bo,+AT) of rBAT. Nature Genetics, 1999, 23, 52-57.	21.4	280
3	Clinical differences between North African and Iraqi Jews with familial Mediterranean fever. , 1998, 75, 216-219.		220
4	MEFV mutation analysis in patients suffering from amyloidosis of familial Mediterranean fever. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 1999, 6, 1-6.	3.0	182
5	Anakinra for Colchicineâ€Resistant Familial Mediterranean Fever: A Randomized, Doubleâ€Blind, Placeboâ€Controlled Trial. Arthritis and Rheumatology, 2017, 69, 854-862.	5.6	147
6	Mutations in FYCO1 Cause Autosomal-Recessive Congenital Cataracts. American Journal of Human Genetics, 2011, 88, 827-838.	6.2	132
7	Mutations in STN1 cause Coats plus syndrome and are associated with genomic and telomere defects. Journal of Experimental Medicine, 2016, 213, 1429-1440.	8.5	100
8	A single mutated MEFV allele in Israeli patients suffering from familial Mediterranean fever and BehÃ§et's disease (FMF-BD). European Journal of Human Genetics, 2001, 9, 191-196.	2.8	74
9	Familial Mediterranean Fever in the First Two Years of Life: A Unique Phenotype of Disease in Evolution. Journal of Pediatrics, 2010, 156, 985-989.	1.8	66
10	Late-onset familial Mediterranean fever (FMF): A subset with distinct clinical, demographic, and molecular genetic characteristics. American Journal of Medical Genetics Part A, 1999, 87, 30-35.	2.4	59
11	TECPR2 mutations cause a new subtype of familial dysautonomia like hereditary sensory autonomic neuropathy with intellectual disability. European Journal of Paediatric Neurology, 2016, 20, 69-79.	1.6	45
12	Evaluation of Diagnostic Yield in Fetal Whole-Exome Sequencing: A Report on 45 Consecutive Families. Frontiers in Genetics, 2019, 10, 425.	2.3	33
13	The risk for developing cancer in Israeli ATM, BLM, and FANCC heterozygous mutation carriers. Cancer Genetics, 2016, 209, 70-74.	0.4	29
14	Clues and challenges in the diagnosis of intermittent maple syrup urine disease. European Journal of Medical Genetics, 2020, 63, 103901.	1.3	26
15	Noncoding deletions reveal a gene that is critical for intestinal function. Nature, 2019, 571, 107-111.	27.8	24
16	BRPF1â€associated intellectual disability, ptosis, and facial dysmorphism in a multiplex family. Molecular Genetics & Genomic Medicine, 2019, 7, e665.	1.2	21
17	Rare genetic variants in Tunisian Jewish patients suffering from age-related macular degeneration. Journal of Medical Genetics, 2015, 52, 484-492.	3.2	19
18	Factors that affect the decision to undergo amniocentesis in women with normal Down syndrome screening results: it is all about the age. Health Expectations, 2015, 18, 2306-2317.	2.6	18

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19	Novelties in the field of autoimmunity â€“ 1st Saint Petersburg congress of autoimmunity, the bridge between east and west. Autoimmunity Reviews, 2017, 16, 1175-1184.	5.8	17
20	The limited effect of information on Israeli pregnant women at advanced maternal age who decide to undergo amniocentesis. Israel Journal of Health Policy Research, 2015, 4, 23.	2.6	16
21	Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene <scp>ATOH1</scp>. Clinical Genetics, 2020, 98, 353-364.	2.0	15
22	Absence of AGG Interruptions Is a Risk Factor for Full Mutation Expansion Among Israeli FMR1 Premutation Carriers. Frontiers in Genetics, 2018, 9, 606.	2.3	14
23	A single center experience with publicly funded clinical exome sequencing for neurodevelopmental disorders or multiple congenital anomalies. Scientific Reports, 2021, 11, 19099.	3.3	13
24	A splicing mutation (891+4Aâ†'G) in SLC3A1 leads to exon 4 skipping and causes cystinuria in a Moslem Arab family. Human Mutation, 1998, 11, S28-S30.	2.5	12
25	Mild Phenotype of Wolfram Syndrome Associated With a Common Pathogenic Variant Is Predicted by a Structural Model of Wolframin. Neurology: Genetics, 2021, 7, e578.	1.9	12
26	Biochemical and clinical studies in Libyan Jewish cystinuria patients and their relatives. , 1998, 80, 173-176.		11
27	Reduction in Filamin C transcript is associated with arrhythmogenic cardiomyopathy in Ashkenazi Jews. International Journal of Cardiology, 2020, 317, 133-138.	1.7	11
28	Prenatal diagnosis for de novo mutations: Experience from a tertiary center over a 10â€year period. Molecular Genetics & Genomic Medicine, 2019, 7, e00573.	1.2	10
29	Identification of a homozygous VRK1 mutation in two patients with adultâ€onset distal hereditary motor neuropathy. Muscle and Nerve, 2020, 61, 395-400.	2.2	10
30	Familial Mediterranean fever: Penetrance of the p.[Met694Val];[Glu148Gln] and p.[Met694Val];[=] genotypes. Human Mutation, 2020, 41, 1866-1870.	2.5	10
31	Mapping of the Familial Mediterranean Fever Gene to Chromosome 16. American Journal of Reproductive Immunology, 1992, 28, 241-242.	1.2	8
32	Patientsâ€™ Attitudes Towards Disclosure of Genetic Test Results to Family Members: The Impact of Patientsâ€™ Sociodemographic Background and Counseling Experience. Journal of Genetic Counseling, 2016, 25, 314-324.	1.6	8
33	Colorectal and Endometrial Cancer Risk and Age at Diagnosis in BLMAsh Mutation Carriers. Israel Medical Association Journal, 2017, 19, 365-367.	0.1	8
34	Refining the Phenotypic Spectrum of KMT5B-Associated Developmental Delay. Frontiers in Pediatrics, 2022, 10, 844845.	1.9	8
35	Lack of evidence of mycobacteria in synovial tissue from patients with rheumatoid arthritis. Arthritis and Rheumatism, 1996, 39, 2080-2081.	6.7	7
36	Analysis of Allelic Association Between D6S461 Marker and Multiple Sclerosis in Ashkenazi and Iraqi Jewish Patients. Journal of Molecular Neuroscience, 1998, 11, 265-270.	2.3	7

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37	A novel mutation in the C7orf11 gene causes nonphotosensitive trichothiodystrophy in a multiplex highly consanguineous kindred. European Journal of Medical Genetics, 2015, 58, 685-688.	1.3	7
38	<scp>SMYD</scp>1 is the underlying gene for the AnWjâ€negative blood group phenotype. European Journal of Haematology, 2018, 101, 496-501.	2.2	7
39	The role of orotic acid measurement in routine newborn screening for urea cycle disorders. Journal of Inherited Metabolic Disease, 2021, 44, 606-617.	3.6	6
40	FMR1 CGG allele length in Israeli BRCA1/BRCA2 mutation carriers and the general population display distinct distribution patterns. Genetical Research, 2014, 96, e11.	0.9	5
41	A false-carrier state for the c.579G>A mutation in the NCF1 gene in Ashkenazi Jews. Journal of Medical Genetics, 2018, 55, 166-172.	3.2	5
42	Early and late manifestations of neuropathy due to HSPB1 mutation in the Jewish Iranian population. Annals of Clinical and Translational Neurology, 2021, 8, 1260-1268.	3.7	3
43	Chromosomal Microarray Evaluation of Fetal Ventriculomegaly. Israel Medical Association Journal, 2020, 22, 639-644.	0.1	3
44	CD55-deficiency in Jews of Bukharan descent is caused by the Cromer blood type Dr(aâˆ’) variant. Human Genetics, 2023, 142, 683-690.	3.8	3
45	Expanding preconception carrier screening for the Jewish population using high throughput microfluidics technology and next generation sequencing. BMC Medical Genomics, 2016, 9, 24.	1.5	2
46	A high and equal prevalence of the Q703K variant in NLRP3 patients with autoinflammatory symptoms and ethnically matched controls. Clinical and Experimental Rheumatology, 2017, 35 Suppl 108, 82-85.	0.8	1