

Elon Pras

List of Publications by Year in descending order

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Version: 2024-02-01

46
papers

1,999
citations

516710

16
h-index

276875

41
g-index

48
all docs

48
docs citations

48
times ranked

3622
citing authors

| # | ARTICLE | IF | CITATIONS |
|----|---|------|-----------|
| 1 | CD55-deficiency in Jews of Bukharan descent is caused by the Cromer blood type Dr(a ⁺) variant. <i>Human Genetics</i> , 2023, 142, 683-690. | 3.8 | 3 |
| 2 | Refining the Phenotypic Spectrum of KMT5B-Associated Developmental Delay. <i>Frontiers in Pediatrics</i> , 2022, 10, 844845. | 1.9 | 8 |
| 3 | The role of orotic acid measurement in routine newborn screening for urea cycle disorders. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 606-617. | 3.6 | 6 |
| 4 | Mild Phenotype of Wolfram Syndrome Associated With a Common Pathogenic Variant Is Predicted by a Structural Model of Wolframin. <i>Neurology: Genetics</i> , 2021, 7, e578. | 1.9 | 12 |
| 5 | Early and late manifestations of neuropathy due to <i>HSPB1</i> mutation in the Jewish Iranian population. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 1260-1268. | 3.7 | 3 |
| 6 | A single center experience with publicly funded clinical exome sequencing for neurodevelopmental disorders or multiple congenital anomalies. <i>Scientific Reports</i> , 2021, 11, 19099. | 3.3 | 13 |
| 7 | Identification of a homozygous <i>VRK1</i> mutation in two patients with adult-onset distal hereditary motor neuropathy. <i>Muscle and Nerve</i> , 2020, 61, 395-400. | 2.2 | 10 |
| 8 | Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene <i>ATOH1</i> . <i>Clinical Genetics</i> , 2020, 98, 353-364. | 2.0 | 15 |
| 9 | Familial Mediterranean fever: Penetrance of the p.[Met694Val];[Glu148Gln] and p.[Met694Val];[-] genotypes. <i>Human Mutation</i> , 2020, 41, 1866-1870. | 2.5 | 10 |
| 10 | Reduction in Filamin C transcript is associated with arrhythmogenic cardiomyopathy in Ashkenazi Jews. <i>International Journal of Cardiology</i> , 2020, 317, 133-138. | 1.7 | 11 |
| 11 | Clues and challenges in the diagnosis of intermittent maple syrup urine disease. <i>European Journal of Medical Genetics</i> , 2020, 63, 103901. | 1.3 | 26 |
| 12 | Chromosomal Microarray Evaluation of Fetal Ventriculomegaly. <i>Israel Medical Association Journal</i> , 2020, 22, 639-644. | 0.1 | 3 |
| 13 | Evaluation of Diagnostic Yield in Fetal Whole-Exome Sequencing: A Report on 45 Consecutive Families. <i>Frontiers in Genetics</i> , 2019, 10, 425. | 2.3 | 33 |
| 14 | Prenatal diagnosis for <i>de novo</i> mutations: Experience from a tertiary center over a 10-year period. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00573. | 1.2 | 10 |
| 15 | Noncoding deletions reveal a gene that is critical for intestinal function. <i>Nature</i> , 2019, 571, 107-111. | 27.8 | 24 |
| 16 | <i>BRPF1</i> -associated intellectual disability, ptosis, and facial dysmorphism in a multiplex family. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e665. | 1.2 | 21 |
| 17 | A false-carrier state for the c.579G>A mutation in the <i>NCF1</i> gene in Ashkenazi Jews. <i>Journal of Medical Genetics</i> , 2018, 55, 166-172. | 3.2 | 5 |
| 18 | Absence of AGG Interruptions Is a Risk Factor for Full Mutation Expansion Among Israeli FMR1 Premutation Carriers. <i>Frontiers in Genetics</i> , 2018, 9, 606. | 2.3 | 14 |

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|----|--|-----|-----------|
| 19 | <i>SMYD1</i> is the underlying gene for the AnWj ⁻ negative blood group phenotype. <i>European Journal of Haematology</i> , 2018, 101, 496-501. | 2.2 | 7 |
| 20 | Novelties in the field of autoimmunity – 1st Saint Petersburg congress of autoimmunity, the bridge between east and west. <i>Autoimmunity Reviews</i> , 2017, 16, 1175-1184. | 5.8 | 17 |
| 21 | Anakinra for Colchicine-Resistant Familial Mediterranean Fever: A Randomized, Double-Blind, Placebo-Controlled Trial. <i>Arthritis and Rheumatology</i> , 2017, 69, 854-862. | 5.6 | 147 |
| 22 | Colorectal and Endometrial Cancer Risk and Age at Diagnosis in BLMASH Mutation Carriers. <i>Israel Medical Association Journal</i> , 2017, 19, 365-367. | 0.1 | 8 |
| 23 | A high and equal prevalence of the Q703K variant in NLRP3 patients with autoinflammatory symptoms and ethnically matched controls. <i>Clinical and Experimental Rheumatology</i> , 2017, 35 Suppl 108, 82-85. | 0.8 | 1 |
| 24 | Mutations in <i>STN1</i> cause Coats plus syndrome and are associated with genomic and telomere defects. <i>Journal of Experimental Medicine</i> , 2016, 213, 1429-1440. | 8.5 | 100 |
| 25 | Expanding preconception carrier screening for the Jewish population using high throughput microfluidics technology and next generation sequencing. <i>BMC Medical Genomics</i> , 2016, 9, 24. | 1.5 | 2 |
| 26 | Patients' Attitudes Towards Disclosure of Genetic Test Results to Family Members: The Impact of Patients' Sociodemographic Background and Counseling Experience. <i>Journal of Genetic Counseling</i> , 2016, 25, 314-324. | 1.6 | 8 |
| 27 | The risk for developing cancer in Israeli ATM, BLM, and FANCC heterozygous mutation carriers. <i>Cancer Genetics</i> , 2016, 209, 70-74. | 0.4 | 29 |
| 28 | TECPR2 mutations cause a new subtype of familial dysautonomia like hereditary sensory autonomic neuropathy with intellectual disability. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 69-79. | 1.6 | 45 |
| 29 | The limited effect of information on Israeli pregnant women at advanced maternal age who decide to undergo amniocentesis. <i>Israel Journal of Health Policy Research</i> , 2015, 4, 23. | 2.6 | 16 |
| 30 | Rare genetic variants in Tunisian Jewish patients suffering from age-related macular degeneration. <i>Journal of Medical Genetics</i> , 2015, 52, 484-492. | 3.2 | 19 |
| 31 | Factors that affect the decision to undergo amniocentesis in women with normal Down syndrome screening results: it is all about the age. <i>Health Expectations</i> , 2015, 18, 2306-2317. | 2.6 | 18 |
| 32 | Whole-exome sequencing in undiagnosed genetic diseases: interpreting 119 trios. <i>Genetics in Medicine</i> , 2015, 17, 774-781. | 2.4 | 284 |
| 33 | A novel mutation in the <i>C7orf11</i> gene causes nonphotosensitive trichothiodystrophy in a multiplex highly consanguineous kindred. <i>European Journal of Medical Genetics</i> , 2015, 58, 685-688. | 1.3 | 7 |
| 34 | <i>FMR1</i> CGG allele length in Israeli <i>BRCA1</i> / <i>BRCA2</i> mutation carriers and the general population display distinct distribution patterns. <i>Genetical Research</i> , 2014, 96, e11. | 0.9 | 5 |
| 35 | Mutations in <i>FYCO1</i> Cause Autosomal-Recessive Congenital Cataracts. <i>American Journal of Human Genetics</i> , 2011, 88, 827-838. | 6.2 | 132 |
| 36 | Familial Mediterranean Fever in the First Two Years of Life: A Unique Phenotype of Disease in Evolution. <i>Journal of Pediatrics</i> , 2010, 156, 985-989. | 1.8 | 66 |

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|----|---|------|-----------|
| 37 | A single mutated MEFV allele in Israeli patients suffering from familial Mediterranean fever and Behçet's disease (FMF-BD). <i>European Journal of Human Genetics</i> , 2001, 9, 191-196. | 2.8 | 74 |
| 38 | Non-type I cystinuria caused by mutations in SLC7A9, encoding a subunit (bo,+AT) of rBAT. <i>Nature Genetics</i> , 1999, 23, 52-57. | 21.4 | 280 |
| 39 | Late-onset familial Mediterranean fever (FMF): A subset with distinct clinical, demographic, and molecular genetic characteristics. <i>American Journal of Medical Genetics Part A</i> , 1999, 87, 30-35. | 2.4 | 59 |
| 40 | MEFV mutation analysis in patients suffering from amyloidosis of familial Mediterranean fever. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , 1999, 6, 1-6. | 3.0 | 182 |
| 41 | Analysis of Allelic Association Between D6S461 Marker and Multiple Sclerosis in Ashkenazi and Iraqi Jewish Patients. <i>Journal of Molecular Neuroscience</i> , 1998, 11, 265-270. | 2.3 | 7 |
| 42 | A splicing mutation (891+4Aâ†'G) in SLC3A1 leads to exon 4 skipping and causes cystinuria in a Moslem Arab family. <i>Human Mutation</i> , 1998, 11, S28-S30. | 2.5 | 12 |
| 43 | Clinical differences between North African and Iraqi Jews with familial Mediterranean fever. , 1998, 75, 216-219. | | 220 |
| 44 | Biochemical and clinical studies in Libyan Jewish cystinuria patients and their relatives. , 1998, 80, 173-176. | | 11 |
| 45 | Lack of evidence of mycobacteria in synovial tissue from patients with rheumatoid arthritis. <i>Arthritis and Rheumatism</i> , 1996, 39, 2080-2081. | 6.7 | 7 |
| 46 | Mapping of the Familial Mediterranean Fever Gene to Chromosome 16. <i>American Journal of Reproductive Immunology</i> , 1992, 28, 241-242. | 1.2 | 8 |