Luigi Boccuto

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8146978/publications.pdf

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57	2,165	331538	243529
papers	citations	21	44
		h-index	g-index

58	58	58	3254
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Gut microbiota and non-alcoholic fatty liver disease. Minerva Gastroenterology, 2022, 67, .	0.3	10
2	Genetic and metabolic profiling of individuals with <scp>Phelanâ€McDermid</scp> syndrome presenting with seizures. Clinical Genetics, 2022, 101, 87-100.	1.0	9
3	Current Status and Future Therapeutic Options for Fecal Microbiota Transplantation. Medicina (Lithuania), 2022, 58, 84.	0.8	21
4	Phelan-McDermid syndrome: a classification system after 30Âyears of experience. Orphanet Journal of Rare Diseases, 2022, 17, 27.	1.2	32
5	Phenotypic Variability in Phelanâ€“McDermid Syndrome and Its Putative Link to Environmental Factors. Genes, 2022, 13, 528.	1.0	5
6	Clinical findings from the landmark MEF2C<scp>â€related</scp> disorders natural history study. Molecular Genetics & Genomic Medicine, 2022, 10, e1919.	0.6	3
7	State of the Science for Kidney Disorders in Phelan-McDermid Syndrome: UPK3A, FBLN1, WNT7B, and CELSR1 as Candidate Genes. Genes, 2022, 13, 1042.	1.0	4
8	Sudden olfactory loss as an early marker of COVID-19: a nationwide Italian survey. European Archives of Oto-Rhino-Laryngology, 2021, 278, 247-255.	0.8	15
9	Nutrition and COVID-19 pandemic: the case of Mediterranean diet. Gazzetta Medica Italiana Archivio Per Le Scienze Mediche, 2021, 179, .	0.0	1
10	Dietary Polyphenols and Non-Alcoholic Fatty Liver Disease. Nutrients, 2021, 13, 494.	1.7	70
11	Medulloblastoma Associated with Down Syndrome: From a Rare Event Leading to a Pathogenic Hypothesis. Diagnostics, 2021, 11, 254.	1.3	3
12	Anxiety and Gastrointestinal Symptoms Related to COVID-19 during Italian Lockdown. Journal of Clinical Medicine, 2021, 10, 1221.	1.0	13
13	Editorial: Recent Advances in Pediatric Cancer Predisposition Syndromes. Frontiers in Pediatrics, 2021, 9, 661894.	0.9	1
14	Individuals with SATB2-associated syndrome with and without autism have a recognizable metabolic profile and distinctive cellular energy metabolism alterations. Metabolic Brain Disease, 2021, 36, 1049-1056.	1.4	4
15	Molecular Characterization of Medulloblastoma in a Patient with Neurofibromatosis Type 1: Case Report and Literature Review. Diagnostics, 2021, 11, 647.	1.3	4
16	Expansion of the clinical and molecular spectrum of an <scp>XPD</scp><scp>â€related disorder linked to biallelic mutations in <scp>ERCC2</scp> gene. Clinical Genetics, 2021, 99, 842-848.	1.0	4
17	Tremors: A concept analysis. Nursing Open, 2021, 8, 2419-2428.	1.1	1
18	Overgrowth in myth and art. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 176-181.	0.7	1

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19	New Strategies for Clinical Trials in Autism Spectrum Disorder. Reviews on Recent Clinical Trials, 2021, 16, 131-137.	0.4	1
20	Genetic and environmental factors influencing the interaction between the gut microbiota and the human host: implications for gastrointestinal disorders and treatment approaches. Minerva Gastroenterology, 2021, , .	0.3	1
21	Comprehensive investigation of the phenotype of <scp>MEF2Câ€related</scp> disorders in human patients: A systematic review. American Journal of Medical Genetics, Part A, 2021, 185, 3884-3894.	0.7	9
22	Are probiotics effective in reversing non-alcoholic steatohepatitis?. Hepatobiliary Surgery and Nutrition, 2021, 10, 407-409.	0.7	0
23	Genetic Findings as the Potential Basis of Personalized Pharmacotherapy in Phelan-McDermid Syndrome. Genes, 2021, 12, 1192.	1.0	9
24	Development of a cell-based metabolic test for the identification of individuals with autism spectrum disorder. Research in Autism Spectrum Disorders, 2021, 85, 101790.	0.8	0
25	Position effects of 22q13 rearrangements on candidate genes in Phelan-McDermid syndrome. PLoS ONE, 2021, 16, e0253859.	1.1	8
26	Upfront treatment with <scp>mTOR</scp> inhibitor everolimus in pediatric lowâ€grade gliomas: A singleâ€center experience. International Journal of Cancer, 2021, 148, 2522-2534.	2.3	19
27	Crohn disease. Nurse Practitioner, 2021, 46, 22-30.	0.2	3
28	A new test for autism spectrum disorder: Metabolic data from different cell types. Data in Brief, 2021, 39, 107598.	0.5	0
29	Abnormalities in the genes that encode Large Amino Acid Transporters increase the risk of Autism Spectrum Disorder. Molecular Genetics & Genomic Medicine, 2020, 8, e1036.	0.6	40
30	Neurofibromatosis type 2 in Phelan-McDermid syndrome: Institutional experience and review of the literature. European Journal of Medical Genetics, 2020, 63, 104042.	0.7	12
31	The Reality of Patient-Reported Outcomes of Health-Related Quality of Life in an Italian Cohort of Patients with Inflammatory Bowel Disease: Results from a Cross-Sectional Study. Journal of Clinical Medicine, 2020, 9, 2416.	1.0	16
32	Cancer Predisposition Syndromes and Medulloblastoma in the Molecular Era. Frontiers in Oncology, 2020, 10, 566822.	1.3	17
33	COVID-19 and Inflammatory Bowel Disease: Patient Knowledge and Perceptions in a Single Center Survey. Medicina (Lithuania), 2020, 56, 407.	0.8	14
34	Low-Grade Gliomas in Patients with Noonan Syndrome: Case-Based Review of the Literature. Diagnostics, 2020, 10, 582.	1.3	21
35	Epidemiology of Coronavirus Disease Outbreak: The Italian Trends. Reviews on Recent Clinical Trials, 2020, 15, 87-92.	0.4	24
36	Gut Microbiota and Obesity: A Role for Probiotics. Nutrients, 2019, 11, 2690.	1.7	335

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37	Diet and Non-Alcoholic Fatty Liver Disease: The Mediterranean Way. International Journal of Environmental Research and Public Health, 2019, 16, 3011.	1.2	86
38	The Skin in Celiac Disease Patients: The Other Side of the Coin. Medicina (Lithuania), 2019, 55, 578.	0.8	28
39	A Multi-strain Probiotic Reduces the Fatty Liver Index, Cytokines and Aminotransferase levels in NAFLD Patients: Evidence from a Randomized Clinical Trial. Journal of Gastrointestinal and Liver Diseases, 2019, 27, 41-49.	0.5	159
40	Identification of 22q13 genes most likely to contribute to Phelan McDermid syndrome. European Journal of Human Genetics, 2018, 26, 293-302.	1.4	54
41	Variability in Phelanâ€McDermid syndrome: The impact of the PNPLA3 p.I148M polymorphism. Clinical Genetics, 2018, 94, 590-591.	1.0	11
42	Obeticholic Acid: A New Era in the Treatment of Nonalcoholic Fatty Liver Disease. Pharmaceuticals, 2018, 11, 104.	1.7	60
43	Beneficial effects of probiotic combination with omega-3 fatty acids in NAFLD: a randomized clinical study. Minerva Medica, 2018, 109, 418-428.	0.3	82
44	The impact of genetic polymorphisms on liver diseases. European Journal of Gastroenterology and Hepatology, 2017, 29, 1102-1103.	0.8	5
45	Polyphenols treatment in patients with nonalcoholic fatty liver disease. Journal of Translational Internal Medicine, 2017, 5, 144-147.	1.0	35
46	Spermine synthase deficiency causes lysosomal dysfunction and oxidative stress in models of Snyder-Robinson syndrome. Nature Communications, 2017, 8, 1257.	5.8	64
47	Genetic and Epigenetic Profile of Patients With Alcoholic Liver Disease. Annals of Hepatology, 2017, 16, 490-500.	0.6	32
48	Phelanâ€“McDermid Syndrome. , 2016, , 347-364.		3
49	Autism spectrum disorder in Phelan-McDermid syndrome: initial characterization and genotype-phenotype correlations. Orphanet Journal of Rare Diseases, 2015, 10, 105.	1.2	53
50	Controlling false discoveries in high-dimensional situations: boosting with stability selection. BMC Bioinformatics, 2015, 16, 144.	1.2	95
51	Letter to the editor regarding Disciglio et al.: Interstitial 22q13 deletions not involving SHANK3 gene: A new contiguous gene syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1679-1680.	0.7	7
52	22q13.2q13.32 genomic regions associated with severity of speech delay, developmental delay, and physical features in Phelanâ€“McDermid syndrome. Genetics in Medicine, 2014, 16, 318-328.	1.1	71
53	Clinical and genomic evaluation of 201 patients with Phelanâ€“McDermid syndrome. Human Genetics, 2014, 133, 847-859.	1.8	142
54	Decreased tryptophan metabolism in patients with autism spectrum disorders. Molecular Autism, 2013, 4, 16.	2.6	124

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55	Prevalence of SHANK3 variants in patients with different subtypes of autism spectrum disorders. European Journal of Human Genetics, 2013, 21, 310-316.	1.4	210
56	Association between deletion size and important phenotypes expands the genomic region of interest in Phelan-McDermid syndrome (22q13 deletion syndrome). Journal of Medical Genetics, 2011, 48, 761-766.	1.5	112
57	Posterior fossa ependymoma in neurodevelopmental syndrome caused by a de novo germline pathogenic Polr2a variant. American Journal of Medical Genetics, Part A, 0, , .	0.7	2