Chris Ottolenghi

List of Publications by Year
in descending order

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95 papers	5,565 citations	61984 43 h-index	85541 71 g-index
97 all docs	97 docs citations	97 times ranked	8310 citing authors

#	Article	IF	Citations
1	SDH Mutations Establish a Hypermethylator Phenotype in Paraganglioma. Cancer Cell, 2013, 23, 739-752.	16.8	606
2	Foxl2 disruption causes mouse ovarian failure by pervasive blockage of follicle development. Human Molecular Genetics, 2004, 13, 1171-1181.	2.9	468
3	Foxl2 is required for commitment to ovary differentiation. Human Molecular Genetics, 2005, 14, 2053-2062.	2.9	298
4	Loss of Wnt4 and Foxl2 leads to female-to-male sex reversal extending to germ cells. Human Molecular Genetics, 2007, 16, 2795-2804.	2.9	293
5	Loss of succinate dehydrogenase activity results in dependency on pyruvate carboxylation for cellular anabolism. Nature Communications, 2015, 6, 8784.	12.8	169
6	Cysteamine therapy delays the progression of nephropathic cystinosis in late adolescents and adults. Kidney International, 2012, 81, 179-189.	5.2	162
7	Foxl2functions in sex determination and histogenesis throughout mouse ovary development. BMC Developmental Biology, 2009, 9, 36.	2.1	113
8	Serum 2-Hydroxyglutarate Production in IDH1- and IDH2-Mutated De Novo Acute Myeloid Leukemia: A Study by the Acute Leukemia French Association Group. Journal of Clinical Oncology, 2014, 32, 297-305.	1.6	109
9	Novel Paralogy Relations Among Human Chromosomes Support a Link between the Phylogeny of doublesex -Related Genes and the Evolution of Sex Determination. Genomics, 2002, 79, 333-343.	2.9	104
10	Successful Treatment of Severe Cardiomyopathy in Glycogen Storage Disease Type III With D,L-3-Hydroxybutyrate, Ketogenic and High-Protein Diet. Pediatric Research, 2011, 70, 638-641.	2.3	96
11	Germline Mutations in the Mitochondrial 2-Oxoglutarate/Malate Carrier SLC25A11 Gene Confer a Predisposition to Metastatic Paragangliomas. Cancer Research, 2018, 78, 1914-1922.	0.9	96
12	Constitutively active Foxo3 in oocytes preserves ovarian reserve in mice. Nature Communications, 2013, 4, 1843.	12.8	95
13	Natural history of Barth syndrome: a national cohort study of 22 patients. Orphanet Journal of Rare Diseases, 2013, 8, 70.	2.7	93
14	Alteration of ornithine metabolism leads to dominant and recessive hereditary spastic paraplegia. Brain, 2015, 138, 2191-2205.	7.6	88
15	The Region on 9p Associated with 46,XY Sex Reversal Contains Several Transcripts Expressed in the Urogenital System and a Novel Doublesex-Related Domain. Genomics, 2000, 64, 170-178.	2.9	87
16	Update on Lysinuric Protein Intolerance, a Multi-faceted Disease Retrospective cohort analysis from birth to adulthood. Orphanet Journal of Rare Diseases, 2017, 12, 3.	2.7	78
17	Aging of Oocyte, Ovary, and Human Reproduction. Annals of the New York Academy of Sciences, 2004, 1034, 117-131.	3.8	77
18	DelK32-lamin A/C has abnormal location and induces incomplete tissue maturation and severe metabolic defects leading to premature death. Human Molecular Genetics, 2012, 21, 1037-1048.	2.9	77

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19	Triheptanoin dramatically reduces paroxysmal motor disorder in patients with GLUT1 deficiency. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 550-553.	1.9	73
20	Long-term neurological outcome of a cohort of 80 patients with classical organic acidurias. Orphanet Journal of Rare Diseases, 2013, 8, 148.	2.7	72
21	Highly specific determination of IDH status using edited in vivo magnetic resonance spectroscopy. Neuro-Oncology, 2018, 20, 907-916.	1.2	72
22	C<scp>IC</scp> inactivating mutations identify aggressive subset of 1p19q codeleted gliomas. Annals of Neurology, 2015, 78, 355-374.	5.3	71
23	Impaired Transferrin Receptor Palmitoylation and Recycling in Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2018, 102, 266-277.	6.2	69
24	Long-term outcomes in Ornithine Transcarbamylase deficiency: a series of 90 patients. Orphanet Journal of Rare Diseases, 2015, 10, 58.	2.7	66
25	Mutations in human lipoyltransferase gene LIPT1cause a Leigh disease with secondary deficiency for pyruvate and alpha-ketoglutarate dehydrogenase. Orphanet Journal of Rare Diseases, 2013, 8, 192.	2.7	64
26	Early and Late Complications After Liver Transplantation for Propionic Acidemia in Children: A Two Centers Study. American Journal of Transplantation, 2015, 15, 786-791.	4.7	61
27	Inducible mEDA-A1 transgene mediates sebaceous gland hyperplasia and differential formation of two types of mouse hair follicles. Human Molecular Genetics, 2003, 12, 2931-2940.	2.9	60
28	Testis determination in mammals: more questions than answers. Molecular and Cellular Endocrinology, 2001, 179, 3-16.	3.2	58
29	Biallelic Mutations in LIPT2 Cause a Mitochondrial Lipoylation Defect Associated with Severe Neonatal Encephalopathy. American Journal of Human Genetics, 2017, 101, 283-290.	6.2	55
30	The environmental carcinogen benzo[a]pyrene induces a Warburg-like metabolic reprogramming dependent on NHE1 and associated with cell survival. Scientific Reports, 2016, 6, 30776.	3.3	54
31	In Vivo Detection of Succinate by Magnetic Resonance Spectroscopy as a Hallmark of SDHx Mutations in Paraganglioma. Clinical Cancer Research, 2016, 22, 1120-1129.	7.0	54
32	Mouse ovary developmental RNA and protein markers from gene expression profiling. Developmental Biology, 2005, 279, 271-290.	2.0	53
33	A driver role for GABA metabolism in controlling stem and proliferative cell state through GHB production in glioma. Acta Neuropathologica, 2017, 133, 645-660.	7.7	53
34	Controversies and research agenda in nephropathic cystinosis: conclusions from a â€œKidney Disease: Improving Global Outcomesâ€•(KDIGO) Controversies Conference. Kidney International, 2016, 89, 1192-1203.	5.2	52
35	Deletions of 9p and the Quest for a Conserved Mechanism of Sex Determination. Molecular Genetics and Metabolism, 2000, 71, 397-404.	1.1	51
36	Acute rhabdomyolysis and inflammation. Journal of Inherited Metabolic Disease, 2015, 38, 621-628.	3.6	51

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37	Cystathionine as a marker for 1p/19q codeleted gliomas by in vivo magnetic resonance spectroscopy. Neuro-Oncology, 2019, 21, 765-774.	1.2	51
38	Combination of lipid metabolism alterations and their sensitivity to inflammatory cytokines in human lipin-1-deficient myoblasts. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2013, 1832, 2103-2114.	3.8	50
39	Determination and stability of sex. BioEssays, 2007, 29, 15-25.	2.5	49
40	The Human Doublesex-Related Gene, DMRT2, Is Homologous to a Gene Involved in Somitogenesis and Encodes a Potential Bicistronic Transcript. Genomics, 2000, 64, 179-186.	2.9	48
41	Liver Engraftment and Repopulation by In Vitro Expanded Adult Derived Human Liver Stem Cells in a Child with Ornithine Carbamoyltransferase Deficiency. JIMD Reports, 2013, 13, 65-72.	1.5	46
42	Leukoencephalopathy with cysts and hyperglycinemia may result from NFU1 deficiency. Mitochondrion, 2014, 15, 59-64.	3.4	46
43	Renal transplantation in 4 patients with methylmalonic aciduria: A cell therapy for metabolic disease. Molecular Genetics and Metabolism, 2013, 110, 106-110.	1.1	44
44	Dihydrolipoamide dehydrogenase deficiency: A still overlooked cause of recurrent acute liver failure and Reye-like syndrome. Molecular Genetics and Metabolism, 2013, 109, 28-32.	1.1	42
45	A Metabolic Study of Huntingtonâ€™s Disease. PLoS ONE, 2016, 11, e0146480.	2.5	41
46	Sonographic biometrical range of external genitalia differentiation in the first trimester of pregnancy: analysis of 2593 cases. Prenatal Diagnosis, 2004, 24, 677-684.	2.3	40
47	Evolutionary Diversification of SPANX-N Sperm Protein Gene Structure and Expression. PLoS ONE, 2007, 2, e359.	2.5	37
48	FISH mapping of the sex-reversal region on human chromosome 9p in two XY females and in primates. European Journal of Human Genetics, 2000, 8, 167-173.	2.8	35
49	Absence of Mutations Involving the Lim Homeobox Domain GeneLHX9in 46,XY Gonadal Agenesis and Dysgenesis. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 2465-2469.	3.6	35
50	Two persistent organic pollutants which act through different xenosensors (alpha-endosulfan and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 human hepatocyte lipid and glucose metabolism. Biochimie, 2015, 116, 79-91.	2.6	35
51	Lung involvement in children with lysinuric protein intolerance. Journal of Inherited Metabolic Disease, 2015, 38, 257-263.	3.6	35
52	Two new cases of serine deficiency disorders treated with l-serine. European Journal of Paediatric Neurology, 2016, 20, 53-60.	1.6	35
53	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disordersâ€”A successful strategy for clinical research of rare diseases. Journal of Inherited Metabolic Disease, 2019, 42, 93-106.	3.6	35
54	From Nf1 to Sdhb knockout: Successes and failures in the quest for animal models of pheochromocytoma. Molecular and Cellular Endocrinology, 2016, 421, 40-48.	3.2	34

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55	NUP98â€“HMGB3: a novel oncogenic fusion. Leukemia, 2010, 24, 654-658.	7.2	33
56	Autism spectrum disorders in propionic acidemia patients. Journal of Inherited Metabolic Disease, 2018, 41, 623-629.	3.6	32
57	Clinical and biological characterization of 20 patients with TANGO2 deficiency indicates novel triggers of metabolic crises and no primary energetic defect. Journal of Inherited Metabolic Disease, 2021, 44, 415-425.	3.6	31
58	The transcription factor Srf regulates hematopoietic stem cell adhesion. Blood, 2010, 116, 4464-4473.	1.4	30
59	Clinical and biochemical heterogeneity associated with fumarase deficiency. Human Mutation, 2011, 32, 1046-1052.	2.5	28
60	Acute Psychosis in Propionic Acidemia. Journal of Child Neurology, 2014, 29, 274-279.	1.4	28
61	Multiple sources of metabolic disturbance in ETHE1â€related ethylmalonic encephalopathy. Journal of Inherited Metabolic Disease, 2010, 33, 443-453.	3.6	25
62	Longâ€term metabolic followâ€up and clinical outcome of 35 patients with maple syrup urine disease. Journal of Inherited Metabolic Disease, 2017, 40, 783-792.	3.6	25
63	Epileptic Phenotype of Two Siblings with Asparagine Synthesis Deficiency Mimics Neonatal Pyridoxine-Dependent Epilepsy. Neuropediatrics, 2016, 47, 399-403.	0.6	21
64	Long-term follow-up in an open-label trial of triheptanoin in GLUT1 deficiency syndrome: a sustained dramatic effect. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 1291-1293.	1.9	21
65	Comorbidity and metabolic context are crucial factors determining neurological sequelae of hypoglycaemia. Developmental Medicine and Child Neurology, 2012, 54, 1012-1017.	2.1	20
66	Longâ€term outcome of methylmalonic aciduria after kidney, liver, or combined liverâ€kidney transplantation: The French experience. Journal of Inherited Metabolic Disease, 2020, 43, 234-243.	3.6	20
67	Population pharmacokinetics and pharmacodynamics of cysteamine in nephropathic cystinosis patients. Orphanet Journal of Rare Diseases, 2011, 6, 86.	2.7	18
68	A Thermolabile Aldolase A Mutant Causes Fever-Induced Recurrent Rhabdomyolysis without Hemolytic Anemia. PLoS Genetics, 2014, 10, e1004711.	3.5	18
69	Isocitrate dehydrogenase (IDH)2 R140Q mutation induces myeloid and lymphoid neoplasms in mice. Leukemia, 2014, 28, 1343-1346.	7.2	18
70	Pyruvate carboxylase deficiency: An underestimated cause of lactic acidosis. Molecular Genetics and Metabolism Reports, 2015, 2, 25-31.	1.1	18
71	Peak hyperammonemia and atypical acute liver failure: The eruption of an urea cycle disorder during hyperemesis gravidarum. Journal of Hepatology, 2018, 68, 185-192.	3.7	18
72	Treatment of acute decompensation of maple syrup urine disease in adult patients with a new parenteral amino-acid mixture. Journal of Inherited Metabolic Disease, 2013, 36, 939-944.	3.6	17

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73	No effect of triheptanoin on exercise performance in McArdle disease. Annals of Clinical and Translational Neurology, 2019, 6, 1949-1960.	3.7	17
74	Functional analysis of the NUP98-CCDC28A fusion protein. Haematologica, 2012, 97, 379-387.	3.5	14
75	5-Fluorouracil rechallenge after 5-fluorouracil-induced hyperammonemic encephalopathy. Anti-Cancer Drugs, 2019, 30, 313-317.	1.4	14
76	Neurocognitive profiles in MSUD schoolâ€age patients. Journal of Inherited Metabolic Disease, 2017, 40, 377-383.	3.6	13
77	A randomized, controlled, double-blind, crossover trial of triheptanoin in alternating hemiplegia of childhood. Orphanet Journal of Rare Diseases, 2017, 12, 160.	2.7	11
78	Gestational ageâ€related reference values for amniotic fluid organic acids. Prenatal Diagnosis, 2010, 30, 43-48.	2.3	10
79	Early prenatal diagnosis of recurrent 46,XY partial gonadal dysgenesis. Prenatal Diagnosis, 2003, 23, 716-721.	2.3	9
80	Hyperprolinemia in Type 2 Glutaric Aciduria and MADD-Like Profiles. JIMD Reports, 2015, 27, 39-45.	1.5	9
81	<scp>OTC</scp> deficiency in females: Phenotypeâ€genotype correlation based on a 130â€family cohort. Journal of Inherited Metabolic Disease, 2021, 44, 1235-1247.	3.6	9
82	Absence of Mutations Involving the Lim Homeobox Domain Gene LHX9 in 46,XY Gonadal Agenesis and Dysgenesis. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 2465-2469.	3.6	9
83	Neonatal gene therapy achieves sustained disease rescue of maple syrup urine disease in mice. Nature Communications, 2022, 13, .	12.8	8
84	The genomic structure of C14orf1 is conserved across eukarya. Mammalian Genome, 2000, 11, 786-788.	2.2	7
85	Phosphoethanolamine normal range in pediatric urines for hypophosphatasia screening. Clinical Chemistry and Laboratory Medicine, 2012, 50, 2231-2233.	2.3	7
86	Transition from ketogenic diet to triheptanoin in patients with GLUT1 deficiency syndrome. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 444-445.	1.9	7
87	Enteral tube feeding in patients receiving dietary treatment for metabolic diseases: A retrospective analysis in a large French cohort. Molecular Genetics and Metabolism Reports, 2021, 26, 100655.	1.1	7
88	Long-term renal outcome in methylmalonic acidemia in adolescents and adults. Orphanet Journal of Rare Diseases, 2021, 16, 220.	2.7	7
89	Placing Parallel Stranded DNA in an Evolutionary Context. Journal of Theoretical Biology, 2000, 206, 317-322.	1.7	6
90	Transcriptional Control of Ovarian Development in Somatic Cells. Seminars in Reproductive Medicine, 2007, 25, 252-263.	1.1	5

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91	Realâ€world management of maple syrup urine disease (MSUD) metabolic decompensations with branched chain amino acid â€free formulas in France and Germany: A retrospective observational study. JIMD Reports, 2021, 59, 110-119.	1.5	5
92	A comparison of immediate release and delayed release cysteamine in 17 patients with nephropathic cystinosis. Orphanet Journal of Rare Diseases, 2021, 16, 387.	2.7	5
93	Reply:ALDH18A1gene mutations cause dominant spastic paraplegia SPG9: loss of function effect and plausibility of a dominant negative mechanism. Brain, 2016, 139, e4-e4.	7.6	4
94	Neonatal factors related to survival and intellectual and developmental outcome of patients with early-onset urea cycle disorders. Molecular Genetics and Metabolism, 2020, 130, 110-117.	1.1	4
95	Genetic divergence between mouse and humans: A useful direction for gene pathway analysis. Teratology, 1998, 58, 82-87.	1.6	0