Jean-Philippe F Gourdine

List of Publications by Year
in descending order

25	2,478	394421 19	580821 25
papers	citations	h-index	g-index

29	29	29	5541
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. Nucleic Acids Research, 2019, 47, D1018-D1027.	14.5	539
2	The Monarch Initiative: an integrative data and analytic platform connecting phenotypes to genotypes across species. Nucleic Acids Research, 2017, 45, D712-D722.	14.5	306
3	Innate immune lectins kill bacteria expressing blood group antigen. Nature Medicine, 2010, 16, 295-301.	30.7	267
4	Microbial glycan microarrays define key features of host-microbial interactions. Nature Chemical Biology, 2014, 10, 470-476.	8.0	191
5	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. American Journal of Human Genetics, 2017, 100, 843-853.	6.2	181
6	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. Genetics, 2017, 207, 9-27.	2.9	165
7	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	6.2	142
8	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	6.2	96
9	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
10	Navigating the Phenotype Frontier: The Monarch Initiative. Genetics, 2016, 203, 1491-1495.	2.9	65
11	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	2.4	60
12	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	6.2	59
13	Galectins are human milk glycan receptors. Glycobiology, 2016, 26, 655-669.	2.5	44
14	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	6.2	43
15	Looking beyond the exome: a phenotype-first approach to molecular diagnostic resolution in rare and undiagnosed diseases. Genetics in Medicine, 2018, 20, 464-469.	2.4	42
16	Forming Consensus To Advance Urobiome Research. MSystems, 2021, 6, e0137120.	3.8	42
17	Analysis of a cDNA-derived sequence of a novel mannose-binding lectin, codakine, from the tropical clam Codakia orbicularis. Fish and Shellfish Immunology, 2007, 22, 498-509.	3.6	35
18	High Affinity Interaction between a Bivalve C-type Lectin and a Biantennary Complex-type N-Glycan Revealed by Crystallography and Microcalorimetry. Journal of Biological Chemistry, 2008, 283, 30112-30120.	3.4	35

#	Article	IF	Citations
19	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	6.2	35
20	Significantly different clinical phenotypes associated with mutations in synthesis and transamidase+remodeling glycosylphosphatidylinositol (GPI)-anchor biosynthesis genes. Orphanet Journal of Rare Diseases, 2020, 15, 40.	2.7	21
21	The three-dimensional structure of codakine and related marine C-type lectins. Fish and Shellfish Immunology, 2007, 23, 831-839.	3.6	16
22	ELECTROPHORETIC SEPARATION OF GILL PROTEINS OF THE CLAMCODAKIA ORBICULARIS. Preparative Biochemistry and Biotechnology, 2002, 32, 341-353.	1.9	8
23	Representing glycophenotypes: semantic unification of glycobiology resources for disease discovery. Database: the Journal of Biological Databases and Curation, 2019, 2019, .	3.0	5
24	Application of Recombinant Phage Display Antibody System in Study of <I>Codakia orbicularis</I> Gill Proteins. Applied Biochemistry and Biotechnology, 2005, 125, 041-052.	2.9	4
25	Degradation of extracellular chondroitin sulfate delays recovery of network activity after perturbation. Journal of Neurophysiology, 2015, 114, 1346-1352.	1.8	4