

Claudio Carta

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/8878083/publications.pdf>

Version: 2024-02-01

33
papers

3,612
citations

257101

24
h-index

377514

34
g-index

35
all docs

35
docs citations

35
times ranked

4842
citing authors

#	ARTICLE	IF	CITATIONS
1	Red Flags for early referral of people with symptoms suggestive of narcolepsy: a report from a national multidisciplinary panel. <i>Neurological Sciences</i> , 2019, 40, 447-456.	0.9	20
2	The RD-Connect Registry & Biobank Finder: a tool for sharing aggregated data and metadata among rare disease researchers. <i>European Journal of Human Genetics</i> , 2018, 26, 631-643.	1.4	33
3	Recommendations for Improving the Quality of Rare Disease Registries. <i>International Journal of Environmental Research and Public Health</i> , 2018, 15, 1644.	1.2	116
4	Linked Registries: Connecting Rare Diseases Patient Registries through a Semantic Web Layer. <i>BioMed Research International</i> , 2017, 2017, 1-13.	0.9	28
5	The Italian pilot external quality assessment program for cystic fibrosis sweat test. <i>Clinical Biochemistry</i> , 2016, 49, 601-605.	0.8	8
6	Protracted late infantile ceroid lipofuscinosis due to TPP1 mutations: Clinical, molecular and biochemical characterization in three sibs. <i>Journal of the Neurological Sciences</i> , 2015, 356, 65-71.	0.3	27
7	The Italian National Centre for Rare Diseases: where research and public health translate into action. <i>Blood Transfusion</i> , 2014, 12 Suppl 3, s591-605.	0.3	4
8	Complete Sequence of the IncT-Type Plasmid pT-OXA-181 Carrying the <i>bla</i> _{OXA-181} Carbapenemase Gene from <i>Citrobacter freundii</i> . <i>Antimicrobial Agents and Chemotherapy</i> , 2013, 57, 1965-1967.	1.4	46
9	Draft Genome Sequence of <i>Stenotrophomonas maltophilia</i> Strain EPM1, Found in Association with a Culture of the Human Parasite <i>Giardia duodenalis</i> . <i>Genome Announcements</i> , 2013, 1, e0018213.	0.8	8
10	A boy with Burkitt lymphoma associated with Noonan syndrome due to a mutation in <i>RAF1</i> . <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1401-1404.	0.7	5
11	<i>Klebsiella pneumoniae</i> ST258 Producing KPC-3 Identified in Italy Carries Novel Plasmids and <i>OmpK36/OmpK35</i> Porin Variants. <i>Antimicrobial Agents and Chemotherapy</i> , 2012, 56, 2143-2145.	1.4	169
12	Complete sequencing of an IncH plasmid carrying the <i>bla</i> _{NDM-1} , <i>bla</i> _{CTX-M-15} and <i>qnrB1</i> genes. <i>Journal of Antimicrobial Chemotherapy</i> , 2012, 67, 1645-1650.	1.3	114
13	A Restricted Spectrum of Mutations in the SMAD4 Tumor-Suppressor Gene Underlies Myhre Syndrome. <i>American Journal of Human Genetics</i> , 2012, 90, 161-169.	2.6	77
14	A restricted spectrum of NRAS mutations causes Noonan syndrome. <i>Nature Genetics</i> , 2010, 42, 27-29.	9.4	271
15	Genomic duplication of <i>PTPN11</i> is an uncommon cause of Noonan syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 2122-2128.	0.7	28
16	Germline <i>BRAF</i> mutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: Molecular diversity and associated phenotypic spectrum. <i>Human Mutation</i> , 2009, 30, 695-702.	1.1	251
17	Spectrum of MEK1 and MEK2 gene mutations in cardio-facio-cutaneous syndrome and genotype-phenotype correlations. <i>European Journal of Human Genetics</i> , 2009, 17, 733-740.	1.4	74
18	Clinical and molecular characterization of 40 patients with Noonan syndrome. <i>European Journal of Medical Genetics</i> , 2008, 51, 566-572.	0.7	45

#	ARTICLE	IF	CITATIONS
19	Diversity, parental germline origin, and phenotypic spectrum of de novo HRAS missense changes in Costello syndrome. <i>Human Mutation</i> , 2007, 28, 265-272.	1.1	123
20	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. <i>Nature Genetics</i> , 2007, 39, 75-79.	9.4	523
21	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. <i>Nature Genetics</i> , 2007, 39, 1007-1012.	9.4	624
22	Germline Missense Mutations Affecting KRAS Isoform B Are Associated with a Severe Noonan Syndrome Phenotype. <i>American Journal of Human Genetics</i> , 2006, 79, 129-135.	2.6	205
23	Structural and functional effects of disease-causing amino acid substitutions affecting residues Ala72 and Glu76 of the protein tyrosine phosphatase SHP-2. <i>Proteins: Structure, Function and Bioinformatics</i> , 2006, 66, 963-974.	1.5	31
24	Genotyping of an Italian papillary thyroid carcinoma cohort revealed high prevalence of BRAF mutations, absence of RAS mutations and allowed the detection of a new mutation of BRAF oncoprotein (BRAFV599Ins). <i>Clinical Endocrinology</i> , 2006, 64, 105-109.	1.2	77
25	Biochemical and molecular characterization of the novel BRAFV599Ins mutation detected in a classic papillary thyroid carcinoma. <i>Oncogene</i> , 2006, 25, 4235-4240.	2.6	56
26	Activating PTPN11 mutations play a minor role in pediatric and adult solid tumors. <i>Cancer Genetics and Cytogenetics</i> , 2006, 166, 124-129.	1.0	48
27	Somatic PTPN11 mutations in childhood acute myeloid leukaemia. <i>British Journal of Haematology</i> , 2005, 129, 333-339.	1.2	78
28	Differences in the prevalence of PTPN11 mutations in FAB M5 paediatric acute myeloid leukaemia. <i>British Journal of Haematology</i> , 2005, 130, 801-803.	1.2	23
29	NF1 Gene Mutations Represent the Major Molecular Event Underlying Neurofibromatosis-Noonan Syndrome. <i>American Journal of Human Genetics</i> , 2005, 77, 1092-1101.	2.6	139
30	Genetic evidence for lineage-related and differentiation stage-related contribution of somatic PTPN11 mutations to leukemogenesis in childhood acute leukemia. <i>Blood</i> , 2004, 104, 307-313.	0.6	265
31	Accentuated response to phenylhydrazine and erythropoietin in mice genetically impaired for their GATA-1 expression (GATA-1 ^{low} mice). <i>Blood</i> , 2001, 97, 3040-3050.	0.6	62
32	Erythropoietin-Dependent Suppression of the Expression of the β^2 Subunits of the Interleukin-3 Receptor during Erythroid Differentiation. <i>Blood Cells, Molecules, and Diseases</i> , 2000, 26, 467-478.	0.6	1
33	In vivo expansion of purified hematopoietic stem cells transplanted in nonablated W/W ^v mice. <i>Experimental Hematology</i> , 1999, 27, 1655-1666.	0.2	25