Claudio Carta

List of Publications by Year in descending order

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33 papers	3,612 citations	236925 25 h-index	377865 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. Neurological Sciences, 2019, 40, 447-456.	1.9	20
2	The RD-Connect Registry & Biobank Finder: a tool for sharing aggregated data and metadata among rare disease researchers. European Journal of Human Genetics, 2018, 26, 631-643.	2.8	33
3	Recommendations for Improving the Quality of Rare Disease Registries. International Journal of Environmental Research and Public Health, 2018, 15, 1644.	2.6	116
4	Linked Registries: Connecting Rare Diseases Patient Registries through a Semantic Web Layer. BioMed Research International, 2017, 2017, 1-13.	1.9	28
5	The Italian pilot external quality assessment program for cystic fibrosis sweat test. Clinical Biochemistry, 2016, 49, 601-605.	1.9	8
6	Protracted late infantile ceroid lipofuscinosis due to TPP1 mutations: Clinical, molecular and biochemical characterization in three sibs. Journal of the Neurological Sciences, 2015, 356, 65-71.	0.6	27
7	The Italian National Centre for Rare Diseases: where research and public health translate into action. Blood Transfusion, 2014, 12 Suppl 3, s591-605.	0.4	4
8	Complete Sequence of the IncT-Type Plasmid pT-OXA-181 Carrying the bla _{OXA-181} Carbapenemase Gene from Citrobacter freundii. Antimicrobial Agents and Chemotherapy, 2013, 57, 1965-1967.	3.2	46
9	Draft Genome Sequence of Stenotrophomonas maltophilia Strain EPM1, Found in Association with a Culture of the Human Parasite Giardia duodenalis. Genome Announcements, 2013, 1, e0018213.	0.8	8
10	A boy with Burkitt lymphoma associated with Noonan syndrome due to a mutation in RAF1. American Journal of Medical Genetics, Part A, 2013, 161, 1401-1404.	1.2	5
11	Klebsiella pneumoniae ST258 Producing KPC-3 Identified in Italy Carries Novel Plasmids and OmpK36/OmpK35 Porin Variants. Antimicrobial Agents and Chemotherapy, 2012, 56, 2143-2145.	3.2	169
12	Complete sequencing of an IncH plasmid carrying the blaNDM-1, blaCTX-M-15 and qnrB1 genes. Journal of Antimicrobial Chemotherapy, 2012, 67, 1645-1650.	3.0	114
13	A Restricted Spectrum of Mutations in the SMAD4 Tumor-Suppressor Gene Underlies Myhre Syndrome. American Journal of Human Genetics, 2012, 90, 161-169.	6.2	77
14	A restricted spectrum of NRAS mutations causes Noonan syndrome. Nature Genetics, 2010, 42, 27-29.	21.4	271
15	Genomic duplication of PTPN11 is an uncommon cause of Noonan syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 2122-2128.	1.2	28
16	GermlineBRAFmutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: Molecular diversity and associated phenotypic spectrum. Human Mutation, 2009, 30, 695-702.	2.5	251
17	Spectrum of MEK1 and MEK2 gene mutations in cardio-facio-cutaneous syndrome and genotypeâ€“phenotype correlations. European Journal of Human Genetics, 2009, 17, 733-740.	2.8	74
18	Clinical and molecular characterization of 40 patients with Noonan syndrome. European Journal of Medical Genetics, 2008, 51, 566-572.	1.3	45

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19	Diversity, parental germline origin, and phenotypic spectrum of de novoHRASmissense changes in Costello syndrome. Human Mutation, 2007, 28, 265-272.	2.5	123
20	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. Nature Genetics, 2007, 39, 75-79.	21.4	523
21	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. Nature Genetics, 2007, 39, 1007-1012.	21.4	624
22	Germline Missense Mutations Affecting KRAS Isoform B Are Associated with a Severe Noonan Syndrome Phenotype. American Journal of Human Genetics, 2006, 79, 129-135.	6.2	205
23	Structural and functional effects of disease-causing amino acid substitutions affecting residues Ala72 and Glu76 of the protein tyrosine phosphatase SHP-2. Proteins: Structure, Function and Bioinformatics, 2006, 66, 963-974.	2.6	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). Clinical Endocrinology, 2006, 64, 105-109.	2.4	77
25	Biochemical and molecular characterization of the novel BRAFV599Ins mutation detected in a classic papillary thyroid carcinoma. Oncogene, 2006, 25, 4235-4240.	5.9	56
26	Activating PTPN11 mutations play a minor role in pediatric and adult solid tumors. Cancer Genetics and Cytogenetics, 2006, 166, 124-129.	1.0	48
27	Somatic PTPN11 mutations in childhood acute myeloid leukaemia. British Journal of Haematology, 2005, 129, 333-339.	2.5	78
28	Differences in the prevalence of PTPN11 mutations in FAB M5 paediatric acute myeloid leukaemia. British Journal of Haematology, 2005, 130, 801-803.	2.5	23
29	NF1 Gene Mutations Represent the Major Molecular Event Underlying Neurofibromatosis-Noonan Syndrome. American Journal of Human Genetics, 2005, 77, 1092-1101.	6.2	139
30	Genetic evidence for lineage-related and differentiation stage-related contribution of somatic PTPN11 mutations to leukemogenesis in childhood acute leukemia. Blood, 2004, 104, 307-313.	1.4	265
31	Accentuated response to phenylhydrazine and erythropoietin in mice genetically impaired for their GATA-1 expression (GATA-1low mice). Blood, 2001, 97, 3040-3050.	1.4	62
32	Erythropoietin-Dependent Suppression of the Expression of the Î² Subunits of the Interleukin-3 Receptor during Erythroid Differentiation. Blood Cells, Molecules, and Diseases, 2000, 26, 467-478.	1.4	1
33	In vivo expansion of purified hematopoietic stem cells transplanted in nonablated W/Wv mice. Experimental Hematology, 1999, 27, 1655-1666.	0.4	25