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List of Publications by Year in descending order

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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	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. Nature Genetics, 2007, 39, 1007-1012.	9.4	624
2	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. Nature Genetics, 2007, 39, 75-79.	9.4	523
3	A restricted spectrum of NRAS mutations causes Noonan syndrome. Nature Genetics, 2010, 42, 27-29.	9.4	271
4	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.	0.6	265
5	GermlineBRAFmutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: Molecular diversity and associated phenotypic spectrum. Human Mutation, 2009, 30, 695-702.	1.1	251
6	Germline Missense Mutations Affecting KRAS Isoform B Are Associated with a Severe Noonan Syndrome Phenotype. American Journal of Human Genetics, 2006, 79, 129-135.	2.6	205
7	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.	1.4	169
8	NF1 Gene Mutations Represent the Major Molecular Event Underlying Neurofibromatosis-Noonan Syndrome. American Journal of Human Genetics, 2005, 77, 1092-1101.	2.6	139
9	Diversity, parental germline origin, and phenotypic spectrum of de novoHRASmissense changes in Costello syndrome. Human Mutation, 2007, 28, 265-272.	1.1	123
10	Recommendations for Improving the Quality of Rare Disease Registries. International Journal of Environmental Research and Public Health, 2018, 15, 1644.	1.2	116
11	Complete sequencing of an IncH plasmid carrying the blaNDM-1, blaCTX-M-15 and qnrB1 genes. Journal of Antimicrobial Chemotherapy, 2012, 67, 1645-1650.	1.3	114
12	Somatic PTPN11 mutations in childhood acute myeloid leukaemia. British Journal of Haematology, 2005, 129, 333-339.	1.2	78
13	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.	1.2	77
14	A Restricted Spectrum of Mutations in the SMAD4 Tumor-Suppressor Gene Underlies Myhre Syndrome. American Journal of Human Genetics, 2012, 90, 161-169.	2.6	77
15	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.	1.4	74
16	Accentuated response to phenylhydrazine and erythropoietin in mice genetically impaired for their GATA-1 expression (GATA-1low mice). Blood, 2001, 97, 3040-3050.	0.6	62
17	Biochemical and molecular characterization of the novel BRAFV599Ins mutation detected in a classic papillary thyroid carcinoma. Oncogene, 2006, 25, 4235-4240.	2.6	56
18	Activating PTPN11 mutations play a minor role in pediatric and adult solid tumors. Cancer Genetics and Cytogenetics, 2006, 166, 124-129.	1.0	48

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19	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.	1.4	46
20	Clinical and molecular characterization of 40 patients with Noonan syndrome. European Journal of Medical Genetics, 2008, 51, 566-572.	0.7	45
21	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.	1.4	33
22	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.	1.5	31
23	Genomic duplication of PTPN11 is an uncommon cause of Noonan syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 2122-2128.	0.7	28
24	Linked Registries: Connecting Rare Diseases Patient Registries through a Semantic Web Layer. BioMed Research International, 2017, 2017, 1-13.	0.9	28
25	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.	0.3	27
26	In vivo expansion of purified hematopoietic stem cells transplanted in nonablated W/Wv mice. Experimental Hematology, 1999, 27, 1655-1666.	0.2	25
27	Differences in the prevalence of PTPN11 mutations in FAB M5 paediatric acute myeloid leukaemia. British Journal of Haematology, 2005, 130, 801-803.	1.2	23
28	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.	0.9	20
29	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
30	The Italian pilot external quality assessment program for cystic fibrosis sweat test. Clinical Biochemistry, 2016, 49, 601-605.	0.8	8
31	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.	0.7	5
32	The Italian National Centre for Rare Diseases: where research and public health translate into action. Blood Transfusion, 2014, 12 Suppl 3, s591-605.	0.3	4
33	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.	0.6	1