Paola Torreri

List of Publications by Year in descending order

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713332 567144 21 735 15 21 citations h-index g-index papers 22 22 22 1689 docs citations all docs times ranked citing authors

#	Article	IF	CITATIONS
1	The RD-Connect Registry & Siobank 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
2	The Occurrence of 275 Rare Diseases and 47 Rare Disease Groups in Italy. Results from the National Registry of Rare Diseases. International Journal of Environmental Research and Public Health, 2018, 15, 1470.	1.2	15
3	Recommendations for Improving the Quality of Rare Disease Registries. International Journal of Environmental Research and Public Health, 2018, 15, 1644.	1.2	116
4	Gaucher disease due to saposin C deficiency is an inherited lysosomal disease caused by rapidly degraded mutant proteins. Human Molecular Genetics, 2014, 23, 5814-5826.	1.4	33
5	Specificity of Îμ and Non-Îμ Isoforms of Arabidopsis 14-3-3 Proteins Towards the H+-ATPase and Other Targets. PLoS ONE, 2014, 9, e90764.	1.1	49
6	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
7	Understanding the role of the Q338H MUTYH variant in oxidative damage repair. Nucleic Acids Research, 2013, 41, 4093-4103.	6.5	25
8	Counteracting Effects Operating on Src Homology 2 Domain-containing Protein-tyrosine Phosphatase 2 (SHP2) Function Drive Selection of the Recurrent Y62D and Y63C Substitutions in Noonan Syndrome*. Journal of Biological Chemistry, 2012, 287, 27066-27077.	1.6	35
9	Phosphorylation on threonineÂ11 of βâ€dystrobrevin alters its interaction with kinesin heavy chain. FEBS Journal, 2012, 279, 4131-4144.	2.2	5
10	Retinoblastoma-independent antiproliferative activity of novel intracellular antibodies against the E7 oncoprotein in HPV 16-positive cells. BMC Cancer, 2011, 11, 17.	1.1	15
11	Functional analysis of MUTYH mutated proteins associated with familial adenomatous polyposis. DNA Repair, 2010, 9, 700-707.	1.3	39
12	The Interaction with HMG20a/b Proteins Suggests a Potential Role for \hat{l}^2 -Dystrobrevin in Neuronal Differentiation. Journal of Biological Chemistry, 2010, 285, 24740-24750.	1.6	29
13	Identification of Key Regions and Genes Important in the Pathogenesis of Sézary Syndrome by Combining Genomic and Expression Microarrays. Cancer Research, 2009, 69, 8438-8446.	0.4	68
14	A global benchmark study using affinity-based biosensors. Analytical Biochemistry, 2009, 386, 194-216.	1.1	85
15	Prion Protein Paralog Doppel Protein Interacts with Alpha-2-Macroglobulin: A Plausible Mechanism for Doppel-Mediated Neurodegeneration. PLoS ONE, 2009, 4, e5968.	1.1	7
16	Diverse driving forces underlie the invariant occurrence of the T42A, E139D, I282V and T468M SHP2 amino acid substitutions causing Noonan and LEOPARD syndromes. Human Molecular Genetics, 2008, 17, 2018-2029.	1.4	79
17	Association of Dystrobrevin and Regulatory Subunit of Protein Kinase A: A New Role for Dystrobrevin as a Scaffold for Signaling Proteins. Journal of Molecular Biology, 2007, 371, 1174-1187.	2.0	18
18	Concerted mutation of Phe residues belonging to the ?-dystroglycan ectodomain strongly inhibits the interaction with ?-dystroglycan ini;½vitro. FEBS Journal, 2006, 273, 4929-4943.	2.2	12

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19	The role of loop ZA and Pro371 in the function of yeast Gcn5p bromodomain revealed through molecular dynamics and experiment. Journal of Molecular Recognition, 2006, 19, 1-9.	1.1	20
20	Molecular Basis of Dystrobrevin Interaction with Kinesin Heavy Chain: Structural Determinants of their Binding. Journal of Molecular Biology, 2005, 354, 872-882.	2.0	13
21	\hat{l}^2 -Dystrobrevin interacts directly with kinesin heavy chain in brain. Journal of Cell Science, 2003, 116, 4847-4856.	1.2	28