

Raul Tonda

List of Publications by Year in descending order

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Version: 2024-02-01

40
papers

1,036
citations

471509

17
h-index

454955

30
g-index

42
all docs

42
docs citations

42
times ranked

2128
citing authors

#	ARTICLE	IF	CITATIONS
1	The RDConnect GenomePhenome Analysis Platform: Accelerating diagnosis, research, and gene discovery for rare diseases. <i>Human Mutation</i> , 2022, , .	2.5	18
2	PrecisionFDA Truth Challenge V2: Calling variants from short and long reads in difficult-to-map regions. <i>Cell Genomics</i> , 2022, 2, 100129.	6.5	72
3	The Genetic Landscape of Inherited Retinal Diseases in a Mexican Cohort: Genes, Mutations and Phenotypes. <i>Genes</i> , 2021, 12, 1824.	2.4	15
4	Diagnostic value of bone marrow core biopsy patterns in lymphoplasmacytic lymphoma/Waldenström macroglobulinaemia and description of its mutational profiles by targeted NGS. <i>Journal of Clinical Pathology</i> , 2020, 73, 571-577.	2.0	7
5	A de novo heterozygous missense BSCL2 variant in 2 siblings with intractable developmental and epileptic encephalopathy. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2019, 71, 161-165.	2.0	8
6	Chromosome 12p Amplification in Triple-Negative/BRCA1-Mutated Breast Cancer Associates with Emergence of Docetaxel Resistance and Carboplatin Sensitivity. <i>Cancer Research</i> , 2019, 79, 4258-4270.	0.9	17
7	A De Novo FOXP1 Truncating Mutation in a Patient Originally Diagnosed as C Syndrome. <i>Scientific Reports</i> , 2018, 8, 694.	3.3	11
8	Novel mutation in the choroideremia gene and multi-Mendelian phenotypes in Spanish families. <i>British Journal of Ophthalmology</i> , 2018, 102, 1378-1386.	3.9	7
9	Heterozygous STUB1 mutation causes familial ataxia with cognitive affective syndrome (SCA48). <i>Neurology</i> , 2018, 91, e1988-e1998.	1.1	81
10	Deep analysis of acquired resistance to FGFR1 inhibitor identifies MET and AKT activation and an expansion of AKT1 mutant cells. <i>Oncotarget</i> , 2018, 9, 31549-31558.	1.8	8
11	Whole-Exome Sequencing of Congenital Glaucoma Patients Reveals Hypermorphic Variants in GPATCH3, a New Gene Involved in Ocular and Craniofacial Development. <i>Scientific Reports</i> , 2017, 7, 46175.	3.3	22
12	Benchmarking of Whole Exome Sequencing and Ad Hoc Designed Panels for Genetic Testing of Hereditary Cancer. <i>Scientific Reports</i> , 2017, 7, 37984.	3.3	35
13	ATRX driver mutation in a composite malignant pheochromocytoma. <i>Cancer Genetics</i> , 2016, 209, 272-277.	0.4	24
14	Novel Candidate Genes and a Wide Spectrum of Structural and Point Mutations Responsible for Inherited Retinal Dystrophies Revealed by Exome Sequencing. <i>PLoS ONE</i> , 2016, 11, e0168966.	2.5	40
15	Runs of homozygosity reveal signatures of positive selection for reproduction traits in breed and non-breed horses. <i>BMC Genomics</i> , 2015, 16, 764.	2.8	125
16	Combined Genetic and High-Throughput Strategies for Molecular Diagnosis of Inherited Retinal Dystrophies. <i>PLoS ONE</i> , 2014, 9, e88410.	2.5	32
17	Next generation sequencing gives an insight into the characteristics of highly selected breeds versus non-breed horses in the course of domestication. <i>BMC Genomics</i> , 2014, 15, 562.	2.8	24
18	An In-Depth Characterization of the Major Psoriasis Susceptibility Locus Identifies Candidate Susceptibility Alleles within an HLA-C Enhancer Element. <i>PLoS ONE</i> , 2013, 8, e71690.	2.5	45

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19	Relative contributions of collagen and tissue factor to thrombus formation on damaged vascular vessels: in-vitro studies with circulating blood. <i>Coronary Artery Disease</i> , 2009, 20, 392-399.	0.7	5
20	Haemodialysis through a cellulose membrane induces dephosphorylation of CD11b and promotes leukocyte adhesion to endothelial cells. <i>Clinical and Investigative Medicine</i> , 2009, 32, 48.	0.6	2
21	Platelets interact with tissue factor immobilized on surfaces: effects of shear rate. <i>European Journal of Clinical Investigation</i> , 2008, 38, 34-42.	3.4	6
22	Effects on primary haemostasis of an anti-inflammatory agent with 5-lipoxygenase and cyclooxygenase inhibitory activity. <i>Journal of Cardiovascular Medicine</i> , 2006, 7, 859-865.	1.5	3
23	Role of Tissue Factor in Platelet Adhesion: Real-Time Perfusion Studies.. <i>Blood</i> , 2006, 108, 1794-1794.	1.4	0
24	Tissue Factor Firmly Immobilized on Surfaces Promotes Platelet Adhesion and Fibrin Formation in Studies with Circulating Human Blood: Importance of Shear Rate Conditions and FVIIa.. <i>Blood</i> , 2006, 108, 3925-3925.	1.4	0
25	Effects of a new pathogen-reduction technology (Mirasol PRT) on functional aspects of platelet concentrates. <i>Transfusion</i> , 2005, 45, 911-919.	1.6	108
26	Antithrombotic effect of a new nitric oxide donor (LA419) on experimental thrombogenesis. <i>European Journal of Clinical Investigation</i> , 2005, 35, 337-342.	3.4	8
27	In vitro evaluation of platelet reactivity toward annuloplasty devices treated with heparin coating: Studies under flow conditions. <i>Journal of Biomedical Materials Research - Part A</i> , 2005, 75A, 192-198.	4.0	4
28	Efficient tyrosine phosphorylation of proteins after activation of platelets with thrombin depends on intact glycoprotein Ib. <i>Platelets</i> , 2005, 16, 453-461.	2.3	5
29	Effect of P-Selectin on Phosphatidylserine Exposure and Surface-Dependent Thrombin Generation on Monocytes. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2005, 25, 1065-1070.	2.4	77
30	Recombinant Activated FVII (rFVIIa) Corrects Platelet Dysfunction in Hemophilic Patients: Study on Collagen and Tissue Factor Surfaces at High Shear Rates.. <i>Blood</i> , 2005, 106, 4034-4034.	1.4	18
31	Biocompatibility of Cellulosic and Synthetic Membranes Assessed by Leukocyte Activation. <i>American Journal of Nephrology</i> , 2004, 24, 235-241.	3.1	24
32	Evaluation of effects of rofecoxib on platelet function in an in vitro model of thrombosis with circulating human blood. <i>European Journal of Clinical Investigation</i> , 2004, 34, 297-302.	3.4	20
33	Hemostatic effect of activated recombinant factor VIIa in Bernard-Soulier syndrome: studies in an in vitro model. <i>Transfusion</i> , 2004, 44, 1790-1791.	1.6	6
34	Platelet membrane fragments enhance the procoagulant effect of recombinant factor VIIa in studies with circulating human blood under conditions of experimental thrombocytopenia. <i>Seminars in Hematology</i> , 2004, 41, 157-162.	3.4	12
35	Direct Activation of Platelets by Tissue Factor Is Amplified by FVIIa and Potentiated by Serotonin: Influence of Shear Rate.. <i>Blood</i> , 2004, 104, 3906-3906.	1.4	0
36	Increased local procoagulant action: a mechanism contributing to the favorable hemostatic effect of recombinant FVIIa in PLT disorders. <i>Transfusion</i> , 2003, 43, 885-892.	1.6	47

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37	Atomic Force Microscopy of Liposomes Bearing Fibrinogen. <i>Bioconjugate Chemistry</i> , 2003, 14, 593-600.	3.6	20
38	Hemostatic effect of activated recombinant factor VII (rFVIIa) in liver disease: studies in an in vitro model. <i>Journal of Hepatology</i> , 2003, 39, 954-959.	3.7	22
39	Recombinant factor VIIa (NovoSeven [®]) restores deficient coagulation: Experience from an ex vivo model. <i>Seminars in Hematology</i> , 2001, 38, 10-14.	3.4	16
40	Recombinant factor VIIa (Novoseven[reg]) restores deficient coagulation: Experience from an ex vivo model. <i>Seminars in Hematology</i> , 2001, 38, 10-14.	3.4	5