Zsolt Urban

List of Publications by Year in descending order

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69 papers	4,723 citations	94269 37 h-index	110170 64 g-index
75 all docs	75 docs citations	75 times ranked	4783 citing authors

#	Article	IF	Citations
1	JAGGED1/NOTCH3 activation promotes aortic hypermuscularization and stenosis in elastin deficiency. Journal of Clinical Investigation, 2022, 132, .	3.9	20
2	LTBP4 in Health and Disease. Genes, 2021, 12, 795.	1.0	15
3	LTBP4 affects renal fibrosis by influencing angiogenesis and altering mitochondrial structure. Cell Death and Disease, 2021, 12, 943.	2.7	15
4	Novel defect in phosphatidylinositol 4â€kinase type 2â€alpha (<scp>PI4K2A</scp>) at the membraneâ€enzyme interface is associated with metabolic cutis laxa. Journal of Inherited Metabolic Disease, 2020, 43, 1382-1391.	1.7	7
5	Exploring the Paradoxical Relationship of a Creb 3 Regulatory Factor Missense Variant With Body Mass Index and Diabetes Among Samoans: Protocol for the Soifua Manuia (Good Health) Observational Cohort Study. JMIR Research Protocols, 2020, 9, e17329.	0.5	13
6	Loss of skin elasticity is associated with pulmonary emphysema, biomarkers of inflammation, and matrix metalloproteinase activity in smokers. Respiratory Research, 2019, 20, 128.	1.4	11
7	Bi-allelic Mutations in the Mitochondrial Ribosomal Protein MRPS2 Cause Sensorineural Hearing Loss, Hypoglycemia, and Multiple OXPHOS Complex Deficiencies. American Journal of Human Genetics, 2018, 102, 685-695.	2.6	61
8	Acromeliaâ€oligodontia syndrome. Clinical Case Reports (discontinued), 2017, 5, 968-974.	0.2	1
9	Novel ELN mutation in a family with supravalvular aortic stenosis and intracranial aneurysm. European Journal of Medical Genetics, 2017, 60, 110-113.	0.7	21
10	A thrifty variant in CREBRF strongly influences body mass index in Samoans. Nature Genetics, 2016, 48, 1049-1054.	9.4	201
11	Integrin Î²3 inhibition is a therapeutic strategy for supravalvular aortic stenosis. Journal of Experimental Medicine, 2016, 213, 451-463.	4.2	46
12	Integrin Î²3 inhibition is a therapeutic strategy for supravalvular aortic stenosis. Journal of General Physiology, 2016, 147, 1473OIA18.	0.9	0
13	Latent transforming growth factor binding protein 4 regulates transforming growth factor beta receptor stability. Human Molecular Genetics, 2015, 24, 4024-4036.	1.4	32
14	Biomechanical Properties of the Skin in Cutis Laxa. Journal of Investigative Dermatology, 2014, 134, 2836-2838.	0.3	11
15	Cutis laxa: Intersection of elastic fiber biogenesis, TGFÎ² signaling, the secretory pathway and metabolism. Matrix Biology, 2014, 33, 16-22.	1.5	40
16	A Novel Elastin Gene Mutation in a Vietnamese Patient with Cutis Laxa. Pediatric Dermatology, 2014, 31, 347-349.	0.5	4
17	Comprehensive Clinical and Molecular Analysis of 12 Families with Type 1 Recessive Cutis Laxa. Human Mutation, 2013, 34, 111-121.	1.1	67
18	The complexity of elastic fibre biogenesis in the skin â€“ a perspective to the clinical heterogeneity of cutis laxa. Experimental Dermatology, 2013, 22, 88-92.	1.4	47

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19	The Complexity of Elastic Fiber Biogenesis: The Paradigm of Cutis Laxa. Journal of Investigative Dermatology, 2012, 132, E12-E14.	0.3	21
20	Alternative Splicing and Tissue-specific Elastin Misassembly Act as Biological Modifiers of Human Elastin Gene Frameshift Mutations Associated with Dominant Cutis Laxa. Journal of Biological Chemistry, 2012, 287, 22055-22067.	1.6	28
21	GLUT10 is required for the development of the cardiovascular system and the notochord and connects mitochondrial function to TGFÎ² signaling. Human Molecular Genetics, 2012, 21, 1248-1259.	1.4	52
22	Cutis laxa: A review. Journal of the American Academy of Dermatology, 2012, 66, 842.e1-842.e17.	0.6	163
23	New insights into the pathogenesis of autosomalâ€dominant cutis laxa with report of fiveELNmutations. Human Mutation, 2011, 32, 445-455.	1.1	116
24	Cutis laxa and fatal pulmonary hypertension. Clinical Dysmorphology, 2011, 20, 77-81.	0.1	2
25	Jagged1 (JAG1) mutations in patients with tetralogy of fallot or pulmonic stenosis. Human Mutation, 2010, 31, 594-601.	1.1	113
26	Mechanisms of emphysema in autosomal dominant cutis laxa. Matrix Biology, 2010, 29, 621-628.	1.5	27
27	Loss-of-function mutations in ATP6V0A2 impair vesicular trafficking, tropoelastin secretion and cell survival. Human Molecular Genetics, 2009, 18, 2149-2165.	1.4	115
28	Decreased bone density and treatment in patients with autosomal recessive cutis laxa. Acta Paediatrica, International Journal of Paediatrics, 2009, 98, 490-494.	0.7	13
29	DANCE/fibulin-5 promotes elastic fiber formation in a tropoelastin isoform-dependent manner. Clinical Biochemistry, 2009, 42, 713-721.	0.8	20
30	Chediak-Steinbrinck-Higashi Syndrome. , 2009, , 314-314.		0
31	Mutations in LTBP4 Cause a Syndrome of Impaired Pulmonary, Gastrointestinal, Genitourinary, Musculoskeletal, and Dermal Development. American Journal of Human Genetics, 2009, 85, 593-605.	2.6	131
32	Impaired glycosylation and cutis laxa caused by mutations in the vesicular H+-ATPase subunit ATP6V0A2. Nature Genetics, 2008, 40, 32-34.	9.4	330
33	Defining the phenotype in an autosomal recessive cutis laxa syndrome with a combined congenital defect of glycosylation. European Journal of Human Genetics, 2008, 16, 28-35.	1.4	67
34	Characterization of the Molecular Interaction between Tropoelastin and DANCE/Fibulin-5. Journal of Biochemistry, 2008, 143, 633-639.	0.9	29
35	Mechanisms and treatment of cardiovascular disease in Williams-Beuren syndrome. Journal of Clinical Investigation, 2008, 118, 1606-1615.	3.9	175
36	Domains 16 and 17 of tropoelastin in elastic fibre formation. Biochemical Journal, 2007, 402, 63-70.	1.7	24

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37	Distinct Steps of Cross-linking, Self-association, and Maturation of Tropoelastin Are Necessary for Elastic Fiber Formation. Journal of Molecular Biology, 2007, 369, 841-851.	2.0	67
38	Molecular genetics of congenital diaphragmatic defects. Annals of Medicine, 2007, 39, 261-274.	1.5	54
39	Population Differences in Elastin Maturation in Optic Nerve Head Tissue and Astrocytes. , 2007, 48, 3209.		39
40	Fibulin-4: A Novel Gene for an Autosomal Recessive Cutis Laxa Syndrome. American Journal of Human Genetics, 2006, 78, 1075-1080.	2.6	266
41	A transgenic mouse model of autosomal cutis laxa. Matrix Biology, 2006, 25, S65-S65.	1.5	0
42	Inflammatory Destruction of Elastic Fibers in Acquired Cutis Laxa Is Associated with Missense Alleles in the Elastin and Fibulin-5 Genes. Journal of Investigative Dermatology, 2006, 126, 283-290.	0.3	67
43	Echocardiographic findings in patients with Williams-Beuren syndrome. Wiener Klinische Wochenschrift, 2006, 118, 538-542.	1.0	19
44	Congenital Heart Disease: Molecular Diagnostics of Supravalvular Aortic Stenosis. , 2006, 126, 129-156.		10
45	Fibulin-5 mutations: mechanisms of impaired elastic fiber formation in recessive cutis laxa. Human Molecular Genetics, 2006, 15, 3379-3386.	1.4	84
46	Autosomal Dominant Cutis Laxa with Severe Lung Disease: Synthesis and Matrix Deposition of Mutant Tropoelastin. Journal of Investigative Dermatology, 2005, 124, 1193-1199.	0.3	87
47	Sensorineural hearing loss in children and adults with Williams syndrome. American Journal of Medical Genetics, Part A, 2005, 138A, 318-327.	0.7	52
48	Aortic aneurysmal disease and cutis laxa caused by defects in the elastin gene. Journal of Medical Genetics, 2005, 43, 255-258.	1.5	125
49	Association of Polymorphisms and Haplotypes in the Elastin Gene in Dutch Patients With Sporadic Aneurysmal Subarachnoid Hemorrhage. Stroke, 2004, 35, 2064-2068.	1.0	55
50	Frontonasal dysplasia in 3H1Br/Br mice. The Anatomical Record, 2003, 271A, 291-302.	2.3	24
51	Loss of ATP-dependent Transport Activity in Pseudoxanthoma Elasticum-associated Mutants of Human ABCC6 (MRP6). Journal of Biological Chemistry, 2002, 277, 16860-16867.	1.6	216
52	Connection between Elastin Haploinsufficiency and Increased Cell Proliferation in Patients with Supravalvular Aortic Stenosis and Williams-Beuren Syndrome. American Journal of Human Genetics, 2002, 71, 30-44.	2.6	166
53	A Serial Analysis of Gene Expression in Sun-Damaged Human Skin. Journal of Investigative Dermatology, 2002, 119, 3-13.	0.3	20
54	Differences by sex in cardiovascular disease in Williams syndrome. Journal of Pediatrics, 2001, 139, 849-853.	0.9	49

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55	Supravalvular aortic stenosis: genetic and molecular dissection of a complex mutation in the elastin gene. Human Genetics, 2001, 109, 512-520.	1.8	55
56	Elastin Gene Deletions in Williams Syndrome Patients Result in Altered Deposition of Elastic Fibers in Skin and a Subclinical Dermalâ€ƒPhenotype. Pediatric Dermatology, 2000, 17, 12-20.	0.5	37
57	Mutations in a gene encoding an ABC transporter cause pseudoxanthoma elasticum. Nature Genetics, 2000, 25, 223-227.	9.4	512
58	Isolated supravalvular aortic stenosis: functional haploinsufficiency of the elastin gene as a result of nonsense-mediated decay. Human Genetics, 2000, 106, 577-588.	1.8	19
59	A Quantitative Evaluation of SAGE. Genome Research, 2000, 10, 1241-1248.	2.4	82
60	Elastic-Fiber Pathologies: Primary Defects in Assemblyâ€”and Secondary Disorders in Transport and Delivery. American Journal of Human Genetics, 2000, 67, 4-7.	2.6	33
61	Genetic disorders of the elastic fiber system. Matrix Biology, 2000, 19, 471-480.	1.5	165
62	Isolated supravalvular aortic stenosis: functional haploinsufficiency of the elastin gene as a result of nonsense-mediated decay. Human Genetics, 2000, 106, 577-588.	1.8	67
63	Supravalvular aortic stenosis: a splice site mutation within the elastin gene results in reduced expression of two aberrantly spliced transcripts. Human Genetics, 1999, 104, 135-142.	1.8	54
64	Altered bladder function in transgenic mice expressing rat elastin. Neurourology and Urodynamics, 1999, 18, 55-68.	0.8	27
65	Pseudoxanthoma Elasticum Maps to an 820-kb Region of the p13.1 Region of Chromosome 16. Genomics, 1999, 62, 1-10.	1.3	61
66	A geneâ€dosage PCR method for the detection of elastin gene deletions in patients with Williams syndrome. Clinical Genetics, 1998, 54, 129-135.	1.0	4
67	A tetranucleotide repeat polymorphism within the human elastin gene (ELNi1). Clinical Genetics, 1997, 51, 133-134.	1.0	15
68	Structure and the Promoter Region of the Mouse Gene Encoding the 67-kD Form of Glutamic Acid Decarboxylase. DNA and Cell Biology, 1996, 15, 1081-1091.	0.9	54
69	A 30 kb deletion within the elastin gene results in familial supravalvular aortic stenosis. Human Molecular Genetics, 1995, 4, 1677-1679.	1.4	100