CURRICULUM VITAE

Zsolt Urban, Ph.D.
University of Pittsburgh

Business Address:	PUBHL 3130 130 Desoto Street Pittsburgh, PA 15213	**Business Phone:**	412-648-8269
Business Fax:	412-624-3020	**Email Address:**	urbanz@pitt.edu
Citizenship:	Naturalized Citizen (USA)		

EDUCATION and TRAINING

Undergraduate

1987 - 1992	University of Szeged Szeged, Hungary	MS Molecular Biology and Biotechnology
1/1992 - 7/1992	University of Manchester Manchester, England	Exchange Student / School of Biological Sciences

Graduate

1992 - 1994	Institute of Biochemistry, Biological Research Institute Szeged, Hungary	PhD Student Dr. Gábor Szabó
1994 - 1996	Joint program with UMDNJ-Robert Wood Johnson Medical School, New Brunswick, NJ, USA and the Department of Pediatrics, Semmelweis University of Medicine Budapest, Hungary	PhD Biology Dr. Charles D Boyd, Dr. György Fekete

APPOINTMENTS and POSITIONS

Academic

2010 - Present	Graduate School of Public Health University of Pittsburgh	Associate Professor Department of Human Genetics
2008 - 2009	Washington University School of Medicine	Assistant Professor Department of Internal Medicine
2/2004 - 7/2004	John A. Burns School of Medicine University of Hawaii	Assistant Professor Department of Anatomy, Biochemistry, Physiology and Reproductive Biology
2004 - 2009	Washington University School of Medicine	Assistant Professor Department of Pediatrics and Department of Genetics
2004 - 2009	Washington University School of Medicine	Associate Member Siteman Cancer Center
2001 - 2004	University of Hawaii	Assistant Researcher Pacific Biomedical Research Center
2001 - 2004	John A. Burns School of Medicine University of Hawaii	Adjunct Assistant Professor Department of Anatomy and Reproductive Biology
2001 - 2004	Cancer Research Center of Hawaii	Associate Member

Non-Academic

1996 - 1997	Semmelweis University of Medicine Budapest, Hungary	Research Fellow/Postdoc Department of Pediatrics
1997 - 2001	University of Hawaii	Junior Researcher/Postdoc Pacific Biomedical Research Center

MEMBERSHIP in PROFESSIONAL and SCIENTIFIC SOCIETIES

1992 - 2001	Member, Hungarian Society for Biochemistry
1998 - 2009	Professional Member, Williams Syndrome Association
1998 - Present	Member, American Society of Human Genetics
1998 - Present	Member, American Heart Association, Council on Basic Cardiovascular Science
2000 - Present	Charter Member, American Society for Matrix Biology
2003 - 2005	Member, American Heart Association, Council on Arteriosclerosis, Thrombosis and Vascular Biology
2003 - 2006	Member, American Society for Investigative Dermatology
2003 - 2015	Member, American Heart Association, Council on Cardiovascular Disease in the Young

PUBLICATIONS

Peer-reviewed Publications

1.		Olson TM, Michels VV, Urban Z, Csiszar K, Christiano AM, Driscoll DJ, Feldt RH, Boyd CD, Thibodeau SN. A 30 kb deletion within the elastin gene results in familial supravalvular aortic stenosis. Human molecular genetics. 1995 Sep; 4 (9):1677-9. PMID: 8541862.
2.		Szabo G, Katarova Z, Kortvely E, Greenspan R, Urban Z. Structure and the promoter region of the mouse gene encoding the 67-kD form of glutamic acid decarboxylase. DNA Cell Biol. 1996; 15:1081-1091.
3.		Urban Z, Helms C, Fekete G, Csiszar K, Bonnet D, Munnich A, Donis-Keller H, Boyd C. 7q11.23 deletions in Williams syndrome arise as a consequence of unequal meiotic crossover. Am J Hum Genet. 1996; 958-962.
4.		Kortvely E, Urban Z, Katarova Z, Szabo G. Transcriptional regulation of the mouse gene encoding the 67 KDa form of glutamic acid decarboxylase. Neurobiology. 1997; 5:172.
5.		Urbán Z, Kiss E, Kádár K, Szabolcs J, Csiszár K, Boyd DC, Fekete G. [Genetic diagnosis of Williams syndrome]. Orvosi hetilap. 1997 Jul 6; 138 (27):1749-52. PMID: 9273487.
6.		Urban Z, Csiszar K, Fekete G, Boyd C. A tetranucleotide repeat polymorphism within the human elastin gene (ELNi1). Clin Genet. 1997; 133-134.
7.		del Rio T, Urbán Z, Csiszár K, Boyd CD. A gene-dosage PCR method for the detection of elastin gene deletions in patients with Williams syndrome. Clin Genet Aug 1998; 54(2): 129-35.
8.		Lemack GE, Szabo Z, Urban Z, Boyd CD, Csiszar K, Vaughan ED Jr, Felsen D. Altered bladder function in transgenic mice expressing rat elastin. Neurourology and urodynamics. 1999; 18 (1):55-68. PMID: 10090127.
9.		Urbán Z, Michels VV, Thibodeau SN, Donis-Keller H, Csiszár K, Boyd CD. Supravalvular aortic stenosis: a splice site mutation within the elastin gene results in reduced expression of two aberrantly spliced transcripts. Human genetics. 1999 Feb; 104 (2):135-42. PMID: 10190324.

10.		Le Saux O, Urban Z, Göring HH, Csiszar K, Pope FM, Richards A, Pasquali-Ronchetti I, Terry S, Bercovitch L, Lebwohl MG, Breuning M, van den Berg P, Kornet L, Doggett N, Ott J, de Jong PT, Bergen AA, Boyd CD. Pseudoxanthoma elasticum maps to an 820-kb region of the p13.1 region of chromosome 16. Genomics. 1999 Nov 15; 62 (1):1-10. PMID: 10585762.
11.		Urban Z, Peyrol S, Plauchu H, Zabot MT, Lebwohl M, Schilling K, Green M, Boyd CD, Csiszar K. Elastin gene deletions in Williams syndrome patients result in altered deposition of elastic fibers in skin and a subclinical dermal phenotype. Pediatric dermatology. 2000; 17 (1):12-20. PMID: 10720981.
12.		Le Saux O, Urban Z, Tschuch C, Csiszar K, Bacchelli B, Quaglino D, Pasquali-Ronchetti I, Pope FM, Richards A, Terry S, Bercovitch L, de Paepe A, Boyd CD. Mutations in a gene encoding an ABC transporter cause pseudoxanthoma elasticum. Nature genetics. 2000 Jun; 25 (2):223-7. PMID: 10835642.
13.		Urbán Z, Michels VV, Thibodeau SN, Davis EC, Bonnefont JP, Munnich A, Eyskens B, Gewillig M, Devriendt K, Boyd CD. Isolated supravalvular aortic stenosis: functional haploinsufficiency of the elastin gene as a result of nonsense-mediated decay. Human genetics. 2000 Jun; 106 (6):577-88. PMID: 10942104.
14.		Stollberg J, Urschitz J, Urban Z, Boyd CD. A quantitative evaluation of SAGE. Genome research. 2000 Aug; 10 (8):1241-8. PMCID: PMC310928. PMID: 10958642.
15.		Dedic J, Weiss AS, Katahira J, Yu B, Trent RJ, Urbán Z. A novel elastin gene mutation (1281delC) in a family with supravalvular aortic stenosis: a mutation cluster within exon 20. Human mutation. 2001; 17 (1):81. PMID: 11139266.
16.		Urbán Z, Zhang J, Davis EC, Maeda GK, Kumar A, Stalker H, Belmont JW, Boyd CD, Wallace MR. Supravalvular aortic stenosis: genetic and molecular dissection of a complex mutation in the elastin gene. Human genetics. 2001 Nov; 109 (5):512-20. PMID: 11735026.
17.		Sadler LS, Pober BR, Grandinetti A, Scheiber D, Fekete G, Sharma AN, Urbán Z. Differences by sex in cardiovascular disease in Williams syndrome. The Journal of pediatrics. 2001 Dec; 139 (6):849-53. PMID: 11743512.
18.		Iliás A, Urbán Z, Seidl TL, Le Saux O, Sinkó E, Boyd CD, Sarkadi B, Váradi A. Loss of ATP-dependent transport activity in pseudoxanthoma elasticum-associated mutants of human ABCC6 (MRP6). The Journal of biological chemistry. 2002 May 10; 277 (19):16860-7. PMID: 11880368.
19.		Urschitz J, Iobst S, Urban Z, Granda C, Souza KA, Lupp C, Schilling K, Scott I, Csiszar K, Boyd CD. A serial analysis of gene expression in sun-damaged human skin. The Journal of investigative dermatology. 2002 Jul; 119 (1):3-13. PMID: 12164917.
20.		Urbán Z, Riazi S, Seidl TL, Katahira J, Smoot LB, Chitayat D, Boyd CD, Hinek A. Connection between elastin haploinsufficiency and increased cell proliferation in patients with supravalvular aortic stenosis and Williams-Beuren syndrome. American journal of human genetics. 2002 Jul; 71 (1):30-44. PMCID: PMC384991. PMID: 12016585.
21.		McBratney BM, Margaryan E, Ma W, Urban Z, Lozanoff S. Frontonasal dysplasia in 3H1 Br/Br mice. The anatomical record. Part A, Discoveries in molecular,

		cellular, and evolutionary biology. 2003 Apr; 271 (2):291-302. PMID: 12629672.
22.		Ruigrok YM, Seitz U, Wolterink S, Rinkel GJ, Wijmenga C, Urbán Z. Association of polymorphisms and haplotypes in the elastin gene in Dutch patients with sporadic aneurysmal subarachnoid hemorrhage. Stroke; a journal of cerebral circulation. 2004 Sep; 35 (9):2064-8. PMID: 15297630.
23.		Urban Z, Gao J, Pope FM, Davis EC. Autosomal dominant cutis laxa with severe lung disease: synthesis and matrix deposition of mutant tropoelastin. J Invest Dermatol Jun 2005; 124(6): 1193-9.
24.		Marler JA, Elfenbein JL, Ryals BM, Urban Z, Netzloff ML. Sensorineural hearing loss in children and adults with Williams syndrome. American journal of medical genetics. Part A. 2005 Nov 1; 138 (4):318-27. PMID: 16222677.
25.		Hu Q, Reymond JL, Pinel N, Zabot MT, Urban Z. Inflammatory destruction of elastic fibers in acquired cutis laxa is associated with missense alleles in the elastin and fibulin-5 genes. J Invest Dermatol Feb 2006; 126(2): 283-90.
26.		Szabo Z, Crepeau MW, Mitchell AL, Stephan MJ, Puntel RA, Yin Loke K, Kirk RC, Urban Z. Aortic aneurysmal disease and cutis laxa caused by defects in the elastin gene. J Med Genet Mar 2006; 43(3): 255-8.
27.		Hucthagowder V, Sausgruber N, Kim KH, Angle B, Marmorstein LY, Urban Z. Fibulin-4: a novel gene for an autosomal recessive cutis laxa syndrome. American journal of human genetics. 2006 Jun; 78 (6):1075-80. PMCID: PMC1474103. PMID: 16685658.
28.		Scheiber D, Fekete G, Urban Z, Tarjan I, Balaton G, Kosa L, Nagy K, Vajo Z. Echocardiographic findings in patients with Williams-Beuren syndrome. Wiener klinische Wochenschrift. 2006 Sep; 118:538-42. PMID: 17009066.
29.		Hu Q, Loeys BL, Coucke PJ, De Paepe A, Mecham RP, Choi J, Davis EC, Urban Z. Fibulin-5 mutations: mechanisms of impaired elastic fiber formation in recessive cutis laxa. Human molecular genetics. 2006 Dec 1; 15 (23):3379-86. PMID: 17035250.
30.		Watts CR, Marler JA, Urban Z. The effects of supravalvular aortic stenosis mutation on voice production. J Med Speech Lang Pathol. 2007; 15 (4):395-406.
31.		Wachi H, Sato F, Nakazawa J, Nonaka R, Szabo Z, Urban Z, Yasunaga T, Maeda I, Okamoto K, Starcher BC, Li DY, Mecham RP, Seyama Y. Domains 16 and 17 of tropoelastin in elastic fibre formation. The Biochemical journal. 2007 Feb 15; 402 (1):63-70. PMCID: PMC1783983. PMID: 17037986.
32.		Sato F, Wachi H, Ishida M, Nonaka R, Onoue S, Urban Z, Starcher BC, Seyama Y. Distinct steps of cross-linking, self-association, and maturation of tropoelastin are necessary for elastic fiber formation. Journal of molecular biology. 2007 Jun 8; 369 (3):841-51. PMID: 17459412.
33.		Urban Z, Agapova O, Hucthagowder V, Yang P, Starcher BC, Hernandez MR. Population differences in elastin maturation in optic nerve head tissue and astrocytes. Investigative ophthalmology & visual science. 2007 Jul; 48 (7):3209-15. PMID: 17591890.
34.		Kornak U, Reynders E, Dimopoulou A, van Reeuwijk J, Fischer B, Rajab A, Budde B, Nürnberg P, Foulquier F, ARCL Debré-type Study Group, Lefeber D, Urban Z, Gruenewald S, Annaert W, Brunner HG, van Bokhoven H, Wevers R, Morava E, Matthijs G, Van Maldergem L, Mundlos S. Impaired glycosylation and

		cutis laxa caused by mutations in the vesicular H+-ATPase subunit ATP6V0A2. Nature genetics. 2008 Jan; 40 (1):32-4. PMID: 18157129.
35.		Morava E, Lefeber DJ, Urban Z, de Meirleir L, Meinecke P, Gillessen Kaesbach G, Sykut-Cegielska J, Adamowicz M, Salafsky I, Ranells J, Lemyre E, van Reeuwijk J, Brunner HG, Wevers RA. Defining the phenotype in an autosomal recessive cutis laxa syndrome with a combined congenital defect of glycosylation. Eur J Hum Genet Jan 2008; 16(1): 28-35.
36.		Wachi H, Nonaka R, Sato F, Shibata-Sato K, Ishida M, Iketani S, Maeda I, Okamoto K, Urban Z, Onoue S, Seyama Y. Characterization of the molecular interaction between tropoelastin and DANCE/fibulin-5. Journal of biochemistry. 2008 May; 143 (5):633-9. PMID: 18267938.
37.		Noordam C, Funke S, Knoers NV, Jira P, Wevers RA, Urban Z, Morava E. Decreased bone density and treatment in patients with autosomal recessive cutis laxa. Acta Paediatr Mar 2009; 98(3): 490-4.
38.		Nonaka R, Onoue S, Wachi H, Sato F, Urban Z, Starcher BC, Seyama Y. DANCE/fibulin-5 promotes elastic fiber formation in a tropoelastin isoform-dependent manner. Clinical biochemistry. 2009 May; 42:713-21. PMID: 19167375.
39.		Hucthagowder V, Morava E, Kornak U, Lefeber DJ, Fischer B, Dimopoulou A, Aldinger A, Choi J, Davis EC, Abuelo DN, Adamowicz M, Al-Aama J, Basel-Vanagaite L, Fernandez B, Greally MT, Gillessen-Kaesbach G, Kayserili H, Lemyre E, Tekin M, Türkmen S, Tuysuz B, Yüksel-Konuk B, Mundlos S, Van Maldergem L, Wevers RA, Urban Z. Loss-of-function mutations in ATP6V0A2 impair vesicular trafficking, tropoelastin secretion and cell survival. Human molecular genetics. 2009 Jun 15; 18 (12):2149-65. PMCID: PMC2685755. PMID: 19321599.
40.		Urban Z, Hucthagowder V, Schürmann N, Todorovic V, Zilberberg L, Choi J, Sens C, Brown CW, Clark RD, Holland KE, Marble M, Sakai LY, Dabovic B, Rifkin DB, Davis EC. Mutations in LTBP4 cause a syndrome of impaired pulmonary, gastrointestinal, genitourinary, musculoskeletal, and dermal development. Am J Hum Genet Nov 2009; 85(5): 593-605.
41.		Bauer RC, Laney AO, Smith R, Gerfen J, Morrissette JJ, Woyciechowski S, Garbarini J, Loomes KM, Krantz ID, Urban Z, Gelb BD, Goldmuntz E, Spinner NB. Jagged1 (JAG1) mutations in patients with tetralogy of Fallot or pulmonic stenosis. Human mutation. 2010 May; 31 (5):594-601. PMCID: PMC2914103. PMID: 20437614.
42.		Hu Q, Shifren A, Sens C, Choi J, Szabo Z, Starcher BC, Knutsen RH, Shipley JM, Davis EC, Mecham RP, Urban Z. Mechanisms of emphysema in autosomal dominant cutis laxa. Matrix biology : journal of the International Society for Matrix Biology. 2010 Sep; 29 (7):621-8. PMCID: PMC3606561. PMID: 20600892.
43.		Callewaert B, Renard M, Hucthagowder V, Albrecht B, Hausser I, Blair E, Dias C, Albino A, Wachi H, Sato F, Mecham RP, Loeys B, Coucke PJ, De Paepe A, Urban Z. New insights into the pathogenesis of autosomal-dominant cutis laxa with report of five ELN mutations. Hum Mutat Apr 2011; 32(4): 445-55.
44.		Brunetti-Pierri N, Piccolo P, Morava E, Wevers RA, McGuirk M, Johnson YR, Urban Z, Dishop MK, Potocki L. Cutis laxa and fatal pulmonary hypertension: a

		newly recognized syndrome? Clin Dysmorphol Apr 2011; 20(2): 77-81.
45.		Willaert A, Khatri S, Callewaert BL, Coucke PJ, Crosby SD, Lee JG, Davis EC, Shiva S, Tsang M, De Paepe A, Urban Z. GLUT10 is required for the development of the cardiovascular system and the notochord and connects mitochondrial function to TGFß signaling. Human molecular genetics. 2012 Mar 15; 21 (6):1248-59. PMCID: PMC3284116. PMID: 22116938.
46.		Sugitani H, Hirano E, Knutsen RH, Shifren A, Wagenseil JE, Ciliberto C, Kozel BA, Urban Z, Davis EC, Broekelmann TJ, Mecham RP. Alternative splicing and tissue-specific elastin misassembly act as biological modifiers of human elastin gene frameshift mutations associated with dominant cutis laxa. The Journal of biological chemistry. 2012 Jun 22; 287 (26):22055-67. PMCID: PMC3381164. PMID: 22573328.
47.		Callewaert B, Su CT, Van Damme T, Vlummens P, Malfait F, Vanakker O, Schulz B, Mac Neal M, Davis EC, Lee JG, Salhi A, Unger S, Heimdal K, De Almeida S, Kornak U, Gaspar H, Bresson JL, Prescott K, Gosendi ME, Mansour S, Piérard GE, Madan-Khetarpal S, Sciurba FC, Symoens S, Coucke PJ, Van Maldergem L, Urban Z, De Paepe A. Comprehensive clinical and molecular analysis of 12 families with type 1 recessive cutis laxa. Human mutation. 2013 Jan; 34 (1):111-21. PMID: 22829427. PMID: 24758204.
48.		Siefring ML, Lawrence EC, Nguyen TC, Lu D, Pham G, Lorenchick C, Levine KL, Urban Z. (2014) A novel elastin gene mutation in a Vietnamese patient with cutis laxa. Pediatr Dermatol 31:347-349. PMCID: PMC4108164
49.		Kozel, BA, Su CT, Danback JR, Minster RL, Madan-Khetarpal S, McConnell J, Mac Neal MK, Levine KL, Wilson RC, Sciurba, FC, Urban Z. (2014) Biomechanical properties of the skin in cutis laxa. J Invest Dermatol 134:2836-2838. PMCID: PMC4199921. PMID: 24844858
50.		Su CT, Huang JW, Chiang CK, Lawrence EC, Levine KL, Dabovic B, Jung C, Davis EC, Madan-Khetarpal S, Urban Z. (2015) Latent transforming growth factor binding protein 4 regulates transforming growth factor beta receptor stability. Hum Mol Genet 24:4024-4036. PMCID: PMC4476448
51.		Misra A, Sheikh AQ, Kumar A, Luo J, Zhang J, Hinton RB, Smoot L, Kaplan P, Urban Z, Qyang Y, Tellides G, Greif DM. (2016) Integrin beta3 inhibition as a therapeutic strategy for supravalvular aortic stenosis. J Exp Med 213:451-453 PMCID: PMC4813675
52.		Minster RL, Hawley NL, Su CT, Sun G, Kershaw EE. Cheng H, Buhule OD, Lin J, Reupena MS, Viali S, Tuitele J, Naseri T, Urban Z*, Deka R*, Weeks DE*, McGarvey ST*. (2016) A thrifty variant in CREBRF strongly influences body mass index in Samoans. Nat Genet. 48:1049-1054 PMCID:PMC5069069 *Joint last authorship.
53.		Jelsig AM, Urban Z, Hucthagowder V, Nissen H, Ousager LB. Novel ELN mutation in a family with supravalvular aortic stenosis and intracranial aneurysm. Eur J Med Genet 60:110-113.
54.		Talasila J, Pachigolla R, Yarlagadda KVSN, Vuppala R, Grzeschik KH, Kiran SKVS, Rose CM, Gottesman GS, Urban Z. (2017) Acromelia-oligodontia syndrome. Clin Case Rep 5(6):968-974. PMCID:PMC5458011
55.		Gardeitchik T, Mohamed M, Ruzzenente B, Karall D, Guerrero-Castillo S,

		Dalloyaux D, van den Brand M, van Kraaij S, van Asbeck E, Assouline Z, Rio M, de Lonlay P, Scholl-Buergi S, Wolthuis DFGJ, Hoischen A, Rodenburg RJ, Sperl W, Urban Z, Brandt U, Mayr JA, Wong S, de Brouwer APM, Nijtmans L, Munnich A, Rötig A, Wevers RA, Metodiev MD, Morava E. (2018) Biallelic mutations in the mitochondrial ribosomal protein MRPS2 cause sensorineural hearing loss, hypoglycemia and multiple OXPHOS complex deficiencies. Am J Hum Genet 102:1-11. PMCID:PMC5985281
56.		O'Brien ME, Chandra D, Wilson RC, Karoleski CM, Fuhrman CR, Leader JK, Pu J, Zhang Y, Morris A, Nouraie S, Bon J, Urban Z, Sciurba FC. (2019) Loss of skin elasticity is associated with pulmonary emphysema, biomarkers of inflammation, and matrix metalloproteinase activity in smokers. Respir Res 20:128
57.		Mohamed M, Gardeitchik T, Balasubramaniam S, Guerrero-Castillo S, Dalloyaux D, van Kraaij S, Venselaar H, Hoischen A, Urban Z, Brandt U, Al-Shawi R, Simons JP, Frison M, Ngu LH, Callewaert B, Spelbrink H, Kallemeijn WW, Aerts JMFG, Waugh MG, Morava E, Wevers RA. (2020) Novel defect in phosphatidylinositol 4-kinase type 2-alpha (PI4K2A) at the membrane-enzyme interface is associated with metabolic cutis laxa. J Inherit Metab Dis doi: 10.1002/jimd.12255. Online ahead of print.
58.		Hawley NL, Pomer A, Rivara AC, Rosenthal SL, Duckham RL, Carlson JC, Naseri T, Reupena MS, Selu M, Lupematisila V, Unasa F, Vesi L, Fatu T, Unasa S, Faasalele-Savusa K, Wetzel AI, Soti-Ulberg C, Prescott AT, Siufaga G, Penaia C, To S, LaMonica LC, Lameko V, Choy CC, Crouter SE, Redline S, Deka R, Kershaw EE*, Urban Z*, Minster RL*, Weeks DE*, and McGarvey ST*. Exploring the paradoxical relationship of a CREBRF missense variant with body mass index and diabetes among Samoans: study protocol for the Soifua Manuia ('Good Health') observational cohort study. JMIR Research Protocols (in press).

Reviews, Proceedings of Conferences and Symposia (not peer-reviewed), Editorials

1.		Urbán Z, Boyd CD. Elastic-fiber pathologies: primary defects in assembly-and secondary disorders in transport and delivery. Am J Hum Genet Jul 2000; 67(1): 4-7.
2.		Milewicz DM, Urbán Z, Boyd C. Genetic disorders of the elastic fiber system. Matrix biology : journal of the International Society for Matrix Biology. 2000 Nov; 19 (6):471-80. PMID: 11068201.
3.		Bielinska M, Jay PY, Erlich JM, Mannisto S, Urban Z, Heikinheimo M, Wilson DB. Molecular genetics of congenital diaphragmatic defects. Annals of medicine. 2007; 39 (4):261-74. PMCID: PMC2174621. PMID: 17558598.
4.		Pober BR, Johnson M, Urban Z. Mechanisms and treatment of cardiovascular disease in Williams-Beuren syndrome. The Journal of clinical investigation. 2008 May; 118 (5):1606-15. PMCID: PMC2358987. PMID: 18452001.
5.		Urban Z. The complexity of elastic fiber biogenesis: the paradigm of cutis laxa. The Journal of investigative dermatology. 2012; 132:E12-4. PMID: 23154624.
6.		Berk DR, Bentley DD, Bayliss SJ, Lind A, Urban Z. Cutis laxa: a review. Journal of the American Academy of Dermatology. 2012 May; 66 (5):842.e1-17. PMID: 22387031.

7.		Uitto J, Li Q, Urban Z. The complexity of elastic fibre biogenesis in the skin--a perspective to the clinical heterogeneity of cutis laxa. Experimental dermatology. 2013 Feb; 22 (2):88-92. PMCID: PMC3556375. PMID: 23088642.
8.		Urban Z, Davis EC. Cutis laxa: intersection of elastic fiber biogenesis, TGFß signaling, the secretory pathway and metabolism. Matrix Biology : Journal of The International Society For Matrix Biology. 2014 Jan; 33:16-22. PMID: 23954411.

Books, Book Chapters, Monographs

1.		Katahira J, Nyerki P, Crepeau M, Davis EC, Pope FM, Hinek A, Urban Z. Elastin gene mutations in supravalvular aortic stenosis, pulmonary artery stenosis and cutis laxa: an emerging genotype-phenotype correlation. In: Tamburro AM, Pepe A editors. Elastin 2002. Potenza, Italy: EditricErmes, 2002. 191-201.
2.		Tassabehji M, Urban Z. Congenital heart disease: Molecular diagnostics of supravalvular aortic stenosis. Methods in molecular medicine. 2006; 126:129-56. PMID: 16930010.
3.		Urban Z. Cutis laxa. In: Lang F, editor. Encyclopedia of molecular mechanisms of disease. Heidelberg: Springer, 2009. 477-479.
4.		Loeys BL, De Paepe A, Urban Z. EFEMP2-related cutis laxa. In: Pagon RA, Bird TD, Dolan CR, Stephens K, editors. GeneReviews. Seattle, WA: University of Washington, 2011.
5.		Callewaert BL, Urban Z. LTBP4-related cutis laxa. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Fong CT, Mefford HC, Smith RJH, Stephens K, (eds) GeneReviews [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2016
6.		Urban Z. LTBP4 and autosomal recessive cutis laxa type IC. In: Erickson RP, Wynshaw-Boris A, editors. Epstein's Inborn Errors of Development, 3rd ed. New York, NY: Oxford University Press, 2016.

Published Abstracts

1.		Szabo G, Katarova Z, Urban Z, Gorcs TJ, Greenspan R. Regulation of glutamic acid decarboxylase (GAD) expression in developing mouse brain: embryonic transcript codes for a truncated protein. [abstract]. In: Society for Neuroscience Annual Meeting; Soc Neurosci Abstr. 1990 Oct 28-Nov 2; St. Louis, MO 1990. 16:1513.
2.		Szabo G, Katarova Z, Urban Z, Mann J, Mugniani E, Greenspan R. Regulation of mouse glutamic acid decarboxylase gene (Gad1(2)) expression in adult brain and in developing mouse embryo. [abstract]. In: Society for Neuroscience Annual Meeting; Soc Neurosci Abstr. 1991 Nov 10-15 ; New Orleans, LA 1991. 17:1513.
3.		Urban Z, Helms C, Peyrol S, Fekete G, Csiszar K, Bonnet D, Munnich A, Donis-Keller H, Boyd CD. Williams syndrome is characterized by 1 megabase 7q deletions encompassing the elastin gene. [abstract]. Matrix Biol. 1996; 15:183.
4.		Urban Z, Csiszar K, Thibodeau SN, Boyd CD. Isolated supravalvular aortic stenosis is characterized by a spectrum of mutations within the elastin gene. [abstract]. Matrix Biol. 1996; 15:183.
5.		Urban Z, Michels VV, Thibodeau SN, Zhang J, Wallace MR, Devriendt K,

		Bonnefont JP, Munnich A, Donis-Keller H, Csiszar K, Boyd CD. Supravalvular aortic stenosis (SVAS): predominance of truncating point mutations within the elastin gene. [abstract]. Am J Hum Genet. 1998; 63:A390.
6.		Boyd CD, Urban Z, Helms C, Peyrol S, Fekete G, Bonnet D, Munnich A, Donis-Keller H, Csiszar K. Williams syndrome is characterized by 1 megabase deletions encompassing the elastin gene. [abstract]. Am J Hum Genet. 1996; 59:A249.
7.		Le Saux O, Urban Z, Goring HH, Csiszar K, Pope FM, Richards A, Pasquali-Ronchetti I, Terry S, Bercovitch L, Lebwohl M, Breuning M, van der Berg P, Kornet L, Doggett N, Ott J, de Jong PT, Bergen AA, Boyd CD. Pseudoxanthoma elsticum maps to an 820 kb region of the p13.1 region of chromosome 16. [abstract]. Am J Hum Genet. 1999; 65:A475.
8.		Urban Z, Davis EC, Zhang J, Wallace MR, Michels VV, Thibodeau SN, Eyskens B, Devriendt K, Boyd CD. Functional haploinsufficiency of the elastin gene in patients with isolated supravalvular aortic stenosis (SVAS). [abstract]. Am J Hum Genet. 1999; 65:A113.
9.		Urban Z, Gao J, Pope FM, Davis EC. A tandem duplication within the elastin gene is associated with autosomal dominant cutis laxa. [abstract]. Am J Hum Genet. 2001; 69:A2543.
10.		Urban Z, Riazi S, Katahira J, Seidl TJ, Smoot L, Boyd CD, Hinek A. Elastin gene mutations in supravlavular aortic stenosis result in reduced elastin synthesis and increased proliferation in skin fibroblasts and aortic smooth muscle cells. [abstract]. Circulation. 2001; 104:II-356.
11.		Urban Z, Crepeau MW, Szabo Z, Pope FM, Stephan MJ, Mitchell A, Yin LK, Davis EC. Elastin gene mutations in patients with cutis laxa: genotype-specific pulmonary and vascular lesions. [abstract]. Am J Hum Genet. 2003; 73:A245.
12.		Crepeau MW, Szabo Z, Stephan MJ, Mitchell A, Urban Z. Familial thoracic aneurysm with cutis laxa caused by a mutation in the elastin gene. [abstract]. Circulation. 2003; 108:IV-2473.
13.		Ruigrok YM, Wolterink S, Seitz U, Rinkel GJ, Wijmenga C, Urban Z. Association of single nucleotide polymorphisms and pairwise haplotypes in the elastin gene with subarachnoid hemorrhage: a case-control study. [abstract]. Circulation. 2003; 108:IV-392.
14.		Hu Q, Crepeau MW, Hucthagowder V, Szabo Z, Urban Z. Locus heterogeneity in cutis laxa. [abstract]. J Invest Dermatol. 2004; 122:A458.
15.		Szabo Z, Crepeau MW, Stephan MJ, Puntel RA, Mitchell A, Urban Z. Cutis laxa caused by mutation in the tropoelastin gene. [abstract]. J Invest Dermatol. 2004; 122:A486.
16.		Urban Z, Hucthagowder V, Henger S, Westman R, Collenburg L, Schurmann N, Todorovic V, Zilberberg L, Choi J, Sens C, Brown CW, Clark RD, Holland KE, Marble M, Sakai LY, Dabovic B, Rifkin DB, Davis EC. Mutations of LTBP4 result in defective craniofacial extracellular matrix formation in patients with Urban-Rifkin-Davis syndrome. [abstract]. Am J Med Genet Part A. 2011; 155:280-286.
17.		Urban Z, Parsons TE, Lorenchick C, Levine KL, Madan-Khetarpal S, Weinberg SM. Craniofacial phenotypes in cutis laxa. [abstract] Am J Med Genet A. 2014 219:S136-S137.

18.		Su CT, Huang JW, Chiang CK, Dabovic B, Rifkin DB, Urban Z. Latent transforming growth factor beta binding protein 4 (Ltbp4) enhances renal fibrosis in obstructive nephropathy. J Am Soc Nephrol. 2014; 25:732A
19.		Su CT, Fang IH, Liu Y, Jao TM, Urban Z. Latent transforming growth factor-beta-binding protein 4 deficiency attenuated renal fibrosis after myocardial infarction. Circulation. 2019; 140:A14169
20.		Su CT, Fang IH, Liu Y, Jao TM, Urban Z. Latent transforming growth factor beta binding protein 4 (LTBP4) attenuates tubulointerstitial fibrosis and ameliorates inflammation and mitochondrial dysfunction in CKD. J Am Soc Nephrol. 2019; 30:538A

RESEARCH

Current research support

Funding Agency:	NIH, NIA
Grant Number:	AG057726
Title of Grant:	Training in molecular epidemiology: linking genes to physical function in older adults
Principal Investigator:	Adam Santanasto
Urban Role on Grant:	Co-Mentor
Years Inclusive:	2018-2023
Percent Effort:	NA
Total Direct Costs:	$566,501
Total Amount Awarded:	$611,821
Funding Agency:	NIH, NHLBI
Grant Number:	HL093093
Title of Grant:	Integrated cellular, mouse and human research on a novel missense variant influencing adiposity in Samoans
Principal Investigator:	Zsolt Urban
Urban Role on Grant:	Co-PI
Years Inclusive:	2016 – 2021 (no cost extension)
Percent Effort:	20%
Total Direct Costs:	$2,903,304
Total Amount Awarded:	$3,035,442

Pending research support

None

Past research support

Funding Agency:	Clinical and Translational Science Institute
Grant Number:	Research Initiative for Special Populations Pilot Program
Title of Grant:	Obesity Gene Regulation in Samoans
Principal Investigator:	Zsolt Urban
Urban Role on Grant:	PI (sole)
Years Inclusive:	2018-2019
Percent Effort:	5% (cost shared)
Total Direct Costs:	$25,000
Total Amount Awarded:	$25,000
Grant Number:	HL090648 (supplement)
Title of Grant:	Genetics of extracellular matrix in health and disease (supplement)
Principal Investigator:	Zsolt Urban
Urban Role on Grant:	PI (sole)
Years Inclusive:	2014 - 2015
Total Direct Costs:	$38,750
Total Amount Awarded:	$59,094
Funding Agency:	NIH
Grant Number:	R01 HL090648
Title of Grant:	'Genetics of Extracellular Matrix in Health and Disease'
Principal Investigator:	Zsolt Urban
Urban Role on Grant:	PI (sole)
Years Inclusive:	2010 - 2014
Percent Effort:	40.0 %
Total Direct Costs:	$1,000,000
Funding Agency:	NIH NHLBI
Grant Number:	R13 HL120348
Title of Grant:	2013 Elastin, Elastic Fibers & Microfibrils Gordon Research Conference and Gordon Research Seminar
Principal Investigator:	Zsolt Urban
Urban Role on Grant:	PI (sole)
Years Inclusive:	2013 - 2014
Total Direct Costs:	$10,000
Total Amount Awarded:	$10,000
Funding Agency:	NIH, NHLBI
Grant Number:	R01 HL090648 (supplement)
Title of Grant:	Genetics of extracellular matrix in health and disease (supplement)

Principal Investigator:	Zsolt Urban
Urban Role on Grant:	PI (sole)
Years Inclusive:	2013 - 2014
Total Direct Costs:	$40,750
Total Amount Awarded:	$55,428
Funding Agency:	University of Pittsburgh, Clinical and Translational Science Institute
Grant Number:	Basic to Clinical Collaborative Research Pilot Program
Title of Grant:	Genetics of Emphysema: Extracellular and Growth Factor Signaling
Principal Investigator:	Zsolt Urban
Urban Role on Grant:	PI (multiple)
Years Inclusive:	2011 - 2012
Percent Effort:	5.0 % (donated)
Total Direct Costs:	$25,000
Funding Agency:	March of Dimes
Grant Number:	Research Grant (#1-FY09-402)
Title of Grant:	’Fibulin-4 in Cardiovascular and Connective Tissue Development
Principal Investigator:	Zsolt Urban
Urban Role on Grant:	PI (sole)
Years Inclusive:	2009 - 2012
Total Direct Costs:	$235,764
Funding Agency:	NIH
Grant Number:	NHLBI SCCOR (P50 HL084922),
Title of Grant:	’Alveolar and airway mechanisms for COPD’
Principal Investigator:	M. Holtzman
Urban Role on Grant:	Co-Investigator
Years Inclusive:	2010 - 2010
Total Amount Awarded:	$50,000
Funding Agency:	American Heart Association
Grant Number:	Heartland Grant-in-Aid (0655626Z)
Title of Grant:	’Williams syndrome heart study’
Years Inclusive:	2006 - 2009
Total Direct Costs:	$130,000
Funding Agency:	NIH
Grant Number:	NHLBI SCCOR (P50 HL084922),
Title of Grant:	’Alveolar and airway mechanisms for COPD’
Principal Investigator:	M. Holtzman

Urban Role on Grant:	Co-Investigator
Years Inclusive:	2006 - 2009
Total Direct Costs:	$3,000,000
Funding Agency:	Washington University, Center for Aging Pilot Research Grant
Title of Grant:	Fibulin-2: a Novel Determinant of the Longevity of Elastic Fibers’
Principal Investigator:	Zsolt Urban
Urban Role on Grant:	PI (sole)
Years Inclusive:	2007 - 2008
Total Amount Awarded:	$30,000
Funding Agency:	NIH
Grant Number:	NHLBI R01 (RFA) (HL73703)
Title of Grant:	’Elastin gene mutations: mechanisms causing SVAS and ADCL’
Principal Investigator:	Zsolt Urban
Urban Role on Grant:	PI (sole)
Years Inclusive:	2002 - 2006
Total Amount Awarded:	$650,000
Funding Agency:	NIH
Grant Number:	NCRR, P20 (RR16453)
Title of Grant:	’A COBRE center for cardiovascular research’ Project 5, PI, ‘Elastin and elastin receptor in vascular diseases’
Principal Investigator:	Zsolt Urban
Urban Role on Grant:	PI (sole)
Years Inclusive:	2001 - 2004
Total Direct Costs:	$450,000
Funding Agency:	American Heart Association
Grant Number:	Grant-in-Aid (0150587N),
Title of Grant:	’Genetic dissection of elastin and elastin receptor interactions in obstructive vascular disease’
Principal Investigator:	Zsolt Urban
Urban Role on Grant:	PI (sole)
Years Inclusive:	2001 - 2003
Total Amount Awarded:	$214,500
Funding Agency:	Connective Tissue Imagineering LLC
Title of Grant:	’Hydrophilic elastin peptides: cellular responses
Principal Investigator:	Zsolt Urban
Urban Role on Grant:	PI (sole)

Years Inclusive:	2001 - 2002
Total Direct Costs:	$126,000
Funding Agency:	NIH
Grant Number:	NIAMS R03 (AR46379)
Title of Grant:	'Elastin gene mutations in skin and vascular diseases.'
Principal Investigator:	Zsolt Urban
Urban Role on Grant:	PI (sole)
Years Inclusive:	1999 - 2002
Total Direct Costs:	$149,550
Funding Agency:	American Heart Association, Hawaii Affiliate
Grant Number:	Beginning Grant-in Aid (HIGB-13-98)
Title of Grant:	'The role of elastin gene mutations and polymorphisms in the pathogenesis of supravalvular aortic stenosis'
Principal Investigator:	Zsolt Urban
Urban Role on Grant:	PI (sole)
Years Inclusive:	1998 - 2000
Total Amount Awarded:	$58,780

OTHER SCHOLARLY ACTIVITIES

Editorial Board(s)

2011 - 2016	Section Editor, Current Genetic Medicine Reports (Springer)
2020 – Present	Editorial Board Member, Biomolecules

Manuscript Reviewer

2000 - 2009	American Journal of Human Genetics
2002	European Journal of Pediatrics
2002 - 2004	Journal of Medical Genetics
2002 - 2019	Matrix Biology
2002 - 2014	Clinical Biochemistry
2003	Biochemistry
2003 - 2015	Journal of Clinical Investigation
2003 – 2020	Circulation
2004	Journal of Biological Chemistry
2004 - 2005	Journal of Vascular Research
2004 - 2020	Stroke
2005	FEBS Journal
2005 - 2016	Journal of Investigative Dermatology
2006	Expert Reviews in Molecular Medicine
2006	Heart and Vessels

2006	Human Mutation
2006	Molecular and Cellular Biology
2006	Pediatric and Developmental Pathology
2007	Laboratory Investigation
2007 - 2020	Experimental Dermatology
2007 - 2008	Neuroscience Letters
2007 – 2020	Human Molecular Genetics
2007 - 2015	American Journal of Medical Genetics
2008	American Journal of Physiology - Lung Cellular and Molecular Physiology
2008	Biochimica et Biophysica Acta
2008	Clinical Genetics
2008 - 2009	American Journal of Pathology
2009 - 2016	Circulation: Cardiovascular Genetics
2009	Investigative Ophthalmology and Visual Science
2010 - 2018	Circulation Research
2011 - 2012	Biology of Reproduction
2012	Genetics Home Reference (NLM, web)
2012	Orphanet Journal of Rare Diseases
2012	Proceedings of the National Academy of Sciences USA
2012 - 2013	PLOS One
2013	American Journal of Respiratory Cell and Molecular Biology
2013	BMC Pediatrics
2013	European Journal of Human Genetics
2014 - 2015	Disease Models & Mechanisms
2015	Archives of Dermatological Research
2015	JAMA Dermatology
2015 - 2019	Arteriosclerosis, Thrombosis, and Vascular Biology
2016	Annals of the American Thoracic Society
2016	Biopolymers
2016	British Journal of Dermatology
2016	Expert Opinion on Therapeutic Targets
2016	Scientific Reports
2017-2018	Molecular Syndromology
2018	BMC Dermatology
2019	JCI Insight
2020	Pediatric Allergy, Immunology, and Pulmonology

Grant Reviewer

2003 - 2004	Medical Advisory Board Member, Hawaii Community Foundation
2003 - 2010	Grant Review Panel Member, American Institute of Biological Sciences
2006 - 2015	External Reviewer, Italian Telethon Foundation

2006 - 2006	External Reviewer, Association Française contre les Myopathies
2006 - 2013	External Reviewer, French National Research Agency (ANR)
2007 - 2007	External Reviewer, Health Research Board, Ireland
2007 - 2007	Reviewer, National Science Foundation
2008 - 2008	Reviewer, Wellcome Trust, UK
2008 - 2014	Reviewer, Medical Research Council, UK
2009 - 2009	Member, NIH, Special Emphasis Panel ZRG1 GGG F
2009 - 2009	Member, NIH, Special Emphasis Panel ZRG1 MOSS G
2009 - 2009	Reviewer, US Army Medical Research and Materiel Command
2010 - 2012	Reviewer, Fund for Scientific Research, Flanders, Belgium (FWO)
2011 - 2011	Member, NIH, Special Emphasis Panel ZRG1 SBIB-X
2013 - 2013	Reviewer, Deutsche Forschungsgemeinshaft (DFG), Germany
2013 - 2013	Reviewer, Washington University, St. Louis, Nutrition and Obesity Research Center Pilot and Feasibility Studies Program
2013 - 2014	Reviewer, University of Pittsburgh, Clinical and Translational Science Institute
2014 - 2016	Reviewer, Qatar National Research Fund
2017	Member, NIH, Genetics of Health and Disease GHD
2018	Reviewer, University of Pittsburgh, Central Research and Development Fund
2018-2020	Member, NIH, Special Emphasis Panel ZHL1 CSR-S

PATENTS

1.		Urban Z, Boyd C, Csiszar K, Le Saux O, Terry M Methods and composition for diagnosing and treating pseudoxanthoma elasticum and related conditions. US Patent 6,780,587, South Africa Patent 2002/7641, European Patents 1258649, 1259649.

INVITED PRESENTATIONS

1.		Urban Z, Helms C, Fekete G, Csiszar K, Donis-Keller H, Boyd CD. 0.9cM deletions distal to the elastin gene locus in patients with Williams syndrome. Presented at: East Coast Connective Tissue Society Meeting; 1996 Mar 22-23; New York, NY.
2.		Urban Z, Helms C, Peyrol S, Fekete G, Csiszar K, Bonnet D, Munnich A, Donis-Keller H, Boyd CD. 0.9 cM deletions distal to the elastin gene locus in patients with Williams syndrome. Presented at: XVth Meeting of the Federation of the European Connnective Tissue Societies; 1996 Aug 4; Munich, Germany.
3.		Urban Z, Csiszar K, Fekete G, Peyrol S, Helms C, Donis-Keller H, Thibodeau SN, Munnich A, Boyd CD. Elastin gene mutations in supravalvular aortic stenosis and Williams syndrome. Presented at: 2nd Annual Meeting Hungarian Biochemical Society; 1997 May 13-16; Lillafured, Hungary.

4.		Le Saux O, Urban Z, Csiszar K, Struk B, Terry S, Bercovitch L, Magro C, Pope M, Lebwohl M, Uitto J, Lindpaintner K, Boyd CD. Pseudoxanthoma elasticum: sib-pair and haplotype analysis reveals a largely recessive disorder with variable penetrance in unrelated families from the United Kingdom and North America. Presented at: International Centennial Meeting on Pseudoxanthoma Elasticum; 1997 Nov 6-7; Bethesda, MD.
5.		Urban Z, Helms C, Peyrol S, Fekete G, Csiszar K, Bonnet D, Munnich A, Donis-Keller H, Boyd CD. 0.9 cM deletions distal to the elastin gene locus in patients with Williams syndrome. Presented at: Pädiatrische Forschung: 7. Tagung Mitteleuropäischer Länder; 1998 Jun 19; Vienna, Austria.
6.		Urban Z, Michels VV, Thibodeau SN, Zhang J, Wallace MR, Bonnefont JP, Munnich A, Donis-Keller H, Csiszar K, Boyd CD. A spectrum of point mutations within the elastin gene in patients with isolated SVAS. Presented at: XVIth Meeting of the Federation of the European Connective Tissue Societies; 1998 Aug 1-6; Uppsala, Sweden.
7.		Urban Z. Mapping of the PXE gene. Presented at: PXE International Colorado Regional Patient Support Meeting; 1998 Oct 31; Denver, CO.
8.		Urban Z. Premature termination mutations in the elastin gene (ELN) are responsible for isolated supravalvular aortic stenosis (SVAS). Presented at: Gordon Conference on Elastin and Elastic Fibers; 1999 Jul 18-23; Meriden, NH.
9.		Le Saux O, Urban Z, Goring HH, Csiszar K, Pope FM, Richards A, Pasquali-Ronchetti I, Terry S, Bercovitch L, Lebwohl MG, Breuning M, van der Berg P, Kornet L, Doggett N, Ott J, de Jong PT, Bergen AA, Boyd CD. Pseudoxanthoma elsticum maps to an 820 kb region of the p13.1 region of chromosome 16. Presented at: Gordon Conference on Elastin and Elastic Fibers; 1999 Jul 18-23; Meriden, NH.
10.		Urban Z. Mapping of the PXE gene. Presented at: PXE International California Regional Patient Support Group Meeting; 1999 Oct 23; San Francisco, CA.
11.		Le Saux O, Urban Z, Tschuch C, Csiszar K, Bacchelli B, Quaglino D, Pasquali-Rochetti I, Pope FM, Richards A, Terry S, Bercovitch L, de Paepe A, Boyd CD. Mutations in a gene encoding an ABC transporter (ABCC6) cause pseudoxanthoma elasticum. Presented at: 1st Symosium of the International Society for Matrix Biology; 2000 Jun 14-17; Philadelphia, PA.
12.		Urban Z, Boyd CD. Molecular genetics of the elastin gene: contributing to the genetic dissection of Williams syndrome. Presented at: 8th International Professional Conference on Williams Syndrome; 2000 Jul 21-23; Dearborn, MI.
13.		Urban Z. Inherited disorders of the elastic fibers. Presented at: Cardiovascular Research Seminar; 2000 Oct 18; Hospital for Sick Children, Toronto, Canada.
14.		Urban Z, Le Saux O, Beck K, Urschitz J, Boyd CD. The pathogenetics of primary and secondary elastinopathies. Presented at: 1st European symposium: Elastin 2001; 2001 Jul 10-14; Rheims, France.
15.		Urban Z, Gao J, Pope FM, Davis EC. A tandem duplication within the elastin gene is associated with autosomal dominant cutis laxa. Presented at: Gordon Conference on Elastin and Elastic Fibers; 2001 Jul 29-Aug 3; Meriden, NH.
16.		Urschitz J, Iobst S, Le Saux O, Urban Z, Csiszar K, Boyd CD. Functional genomic approaches to understanding heritable and acquired disorders of

		connective tissue. Presented at: The Biochemical Society Joint Meeting with The Physiological Society; 2001 Dec 17-19; York, UK.
17.		Katahira J, Nyerki P, Crepeau M, Davis EC, Pope FM, Hinek A, Urban Z. Elastin gene mutations in supravalvular aortic stenosis, pulmonary artery stenosis and cutis laxa: an emerging genotype-phenotype correlation. Presented at: Elastin 2002, Second European Symposium; 2002 Jul 10-14; Aquafredda di Maratea, Italy.
18.		Urban Z. Elastin-cell interactions: insights from genetic diseases. Presented at: American Society for Matrix Biology, First Meeting; 2002 Nov 6-9; Houston, TX.
19.		Crepeau MW, Szabo Z, Pope FM, Stephan MJ, Mitchell A, Yin LK, Davis EC, Urban Z. Novel mutations in the elastin gene in patients with cutis laxa. Presented at: Gordon Conference on Elastin and Elastic Fibers; 2003 Aug 3-8; Meriden, NH.
20.		Urban Z. Elastin gene in rare and common diseases. Presented at: Cell Biology Research Seminar; 2003 Nov 13; Washington University School of Medicine, St. Louis, MO.
21.		Urban Z, Crepeau MW, Szabo Z, Hu Q, Hucthagowder V. Locus heterogeneity in cutis laxa. Presented at: Indian Society of Human Genetics, 29th Annual Meeting; 2004 Jan 8-11; Bangalore, India.
22.		Urban Z. Pediatric diseases caused by elastin gene defects. Presented at: Pediatrics Seminar; 2004 Jan 13; National University of Singapore, Singapore.
23.		Urban Z. Locus heterogeneity in cutis laxa. Presented at: Pediatrics Research Seminar; 2004 Mar 10; Washington University School of Medicine, St. Louis, MO.
24.		Urban Z. Elastin gene defects in vascular and skin diseases. Presented at: Enzymology Research Seminar; 2004 Jun 25; Institute of Enzymology, Hungarian Academy of Sciences, Budapest, Hungary.
25.		Crepeau MW, Szabo Z, Hu Q, Hucthagowder V, Urban Z. Elastin and fibulin-5 gene defects in cutis laxa. Presented at: Elastin 2004, 3rd European Symposium; 2004 Jun 30-Jul 3; Manchester, UK.
26.		Urban Z. Locus heterogeneity in cutis laxa. Presented at: Center for Medical Genetics Seminar; 2004 Jul 6; University Medical Center, Ghent, Belgium.
27.		Urban Z. Elastin gene defects in vascular and skin diseases. Presented at: Skin Research Seminar; 2005 May 16; Johnson & Johnson Skin Research Center, Skilman, NJ.
28.		Szabo Z, Pope FM, Stephan MJ, Mitchell AL, Loke KY, Coucke PJ, Davis EC, Urban Z. Elastin gene mutations in patients with inherited cutis laxa. Presented at: Gordon Conference on Elastin and Elastic Fibers; 2005 Jul 31-Aug 5; Waterville Valley, NH.
29.		Hucthagowder V, Sausgruber N, Kim KK, Angle B, Marmorstein LY, Urban Z. Fibulin-4 mutation in a novel recessive cutis laxa syndrome. Presented at: 4th European Meeting on Elastin; 2006 Jul 9-12; Lyon, France.
30.		Hucthagowder V, Kaplan P, Pober BR, Urban Z. Elastin in Williams syndrome: cardiovascular disease and beyond. Presented at: 11th International Professional Conference on Williams Syndrome; 2006 Jul 23-24; Richmond, VA.
31.		Urban Z. Fibulin gene mutations in developmental defects. Presented at: Enzymology Research Seminar; 2007 May 31; Institute of Enzymology,

		Hungarian Academy of Sciences, Budapest, Hungary.
32.		Maxfield AB, Hucthagowder V, Joseph EM, Urban Z. Fibulin-4 function in zebrafish. Presented at: Gordon Conference on Elastin and Elastic Fibers; 2007 Jul 29-Aug 3; Biddeford, ME.
33.		Hucthagowder V, Johnson MC, Kaplan P, Singh GK, Urban Z. Genetic Analysis of Congenital Heart Disease in Williams Syndrome. Presented at: Midwest Pediatric Cardiology Society 31st Annual Scientific Meeting; 2007 Sep 20-21; St. Louis, MO.
34.		Hucthagowder V, Morava E, Kornak U, Lefeber DJ, Dimopoulu A, Fischer B, Aldinger A, Choi J, Davis EC, Abuelo DN, Basel-Vanagaite L, Bruner HG, Gillessen-Kaesbach G, Kayserili H, Lemyre E, Mundlos S, Van Maldergem L, Wevers RA, Urban Z. Loss-of-function mutations in ATP6V0A2 impair elastin deposition and cell survival. Presented at: 5th European Elastin Meeting; 2008 Jul 16-19; Alcalá De Henares, Spain.
35.		Urban Z. Elastin mutations in cutis laxa. Presented at: American Society for Matrix Biology, 2008 Meeting; 2008 Dec 7-10; San Diego, CA.
36.		Urban Z. Human genetics of the elastic fiber/TGF-beta network. Presented at: Human Genetics Seminar; 2009 May 29; Graduate School of Public Health, University of Pittsburgh.
37.		Hucthagowder V, Morava E, Kornak U, Lefeber DJ, Dimopoulu A, Fischer B, Aldinger A, Choi, Davis EC, Abuelo DN, Adamowicz M, Al-Aama J, Basel-Vanageite L, Fernandez B, Greally MT, Gillessen-Kaesbach G, Kayserili H, Lemyre E, Tekin M, Turkmen S, Tuysuz B, Yuksel-Konuk B, Mundlos S, Van Maldergem L, Wevers RA, Urban Z. Loss-of-function mutations in ATP6V0A2 impair vesicular trafficking, tropoelastin secretion, and cell survival. Presented at: Gordon Conference on Elastin and Elastic Fibers; 2009 Jul 26-31; Biddeford, ME.
38.		Urban Z. 'Elastic fibre synthesis and assembly: insights from human genetics. Presented at: Cell Matrix Seminar; 2009 Nov 3; Wellcome Trust Centre for Cell Matrix Research, Manchester, UK.
39.		Urban Z, Hucthagowder V, Henger S, Westman R, Collenburg L, Schurmann N, Todorovic V, Zilberberg L, Choi J, Sens C, Brown CW, Clark RD, Holland KE, Marble M, Sakai LY, Dabovic B, Rifkin DB, Davis EC. LTBP4 mutations in Urban-Rifkin-Davis syndrome. Presented at: 6th European Elastin Meeting; 2010 Jun 28-Jul 2; Maratea, Italy.
40.		Urban Z, Hucthagowder V, Henger S, Westman R, Collenburg L, Schurmann N, Todorovic V, Zilberberg L, Choi J, Sens C, Brown CW, Clark RD, Holland KE, Marble M, Sakai LY, Dabovic B, Rifkin DB, Davis EC. Disease phenotypes and mechanisms of LTBP4 mutations. Presented at: 8th International Symposium on Marfan Syndrome; 2010 Sep 11-14; Warrenton, VA.
41.		Urban Z. The molecular basis of elastic fiber assembly and TGFß signaling: insights from human genetics. Presented at: Anatomy Research Seminar; 2011 Jan 10; John A. Burns School of Medicine, University of Hawaii, Honolulu, HI.
42.		An elastic view of genes, environment and disease. Presented at: Environmental and Occupational Health Seminar; 2011 Apr 28; Graduate School of Public Health, University of Pittsburgh, Pittsburgh, PA.

43.		Willaert A, Khatri SM, Callewaert BL, Coucke PJ, Crosby SD, De Paepe A, Urban Z. GLUT10 connects cellular metabolism to TGFß signaling. Presented at: Gordon Conference on Elastin and Elastic Fibers; 2011 Jul 24-29; Biddeford, ME.
44.		Urban Z. Animal models of cutis laxa. Presented at: 4th Days on Cutis Laxa, Cutis Laxa Internationale Meeting; 2011 Sep 16-17; Lyon, France.
45.		Urban Z. Arterial tortuosity: new intracellular and extracellular pathways of TGFß regulation. Presented at: Enzymology Seminar; 2011 Sep 20; Institute of Enzymology, Hungarian Academy of Sciences, Budapest, Hungary.
46.		Urban Z. Human genetic insights into the elastic fiber/TGFß network. Presented at: Anatomy and Cell Biology Seminar; 2011 Oct 12; McGill University, Montreal, Canada.
47.		Urban Z. The elastic fiber/TGF-beta network in rare and common forms of COPD. Presented at: Pittsburgh International Lung Conference, Personalized Medicine of Lung Disease; 2011 Oct 28-29; Pittsburgh, PA.
48.		Urban Z. Arterial tortuosity: new intracellular pathways of TGFß regulation. Presented at: Molecular Medicine Research Seminar; 2012 Mar 20; Department of Pediatrics, University of Pittsburgh, Pittsburgh, PA.
49.		Callewaert BL, Su CT, Van Damme T, Vlummens P, Malfait F, Vanakker O, Schulz B, Mac Neal M, Davis EC, Lee JG, Salhi A, Unger S, Heimdal K, De Almeida S, Kornak U, Gaspar H, Bresson J, Prescott K, Gosendi ME, Mansour S, Pierard GE, Madan-Khetarpal S, Sciurba FC, Coucke PJ, Van Maldergem L, Urban Z, De Paepe A. Comprehensive clinical and molecular analysis of 12 families with type I recessive cutis laxa. Presented at: European Human Genetics Conference 2012; 2012 Jun 23-26; Nurnberg, Germany.
50.		Callewaert BL, Su CT, Van Damme T, Malfait F, Vanakker O, Davis EC, Coucke PJ, Van Maldergem L, Urban Z, De Paepe A. Comprehensive clinical and molecular analysis of 12 families with type 1 recessive cutis laxa. Presented at: 7th European Elastin Meeting; 2012 Sep 1-4; Ghent, Belgium.
51.		Su CT, Khatri SM, McGowan ME, Lawrence E, Urban Z. Transforming growth factor-beta dysregulation in cutis laxa. Presented at: 7th European Elastin Meeting; 2012 Sep 1-4; Ghent, Belgium.
52.		Urban Z. Gene discovery in cutis laxa: insights into elastic fiber formation and TGF-beta signaling. Presented at: Joint Meeting of the Society for Glycobiology & American Society for Matrix Biology; 2012 Nov 11-14; San Diego, CA.
53.		Urban Z. Arterial tortuosity: new intracellular and extracellular pathways of TGFß regulation. Presented at: Cell Matrix Seminar; 2013 May 16; Wellcome Trust Centre for Cell Matrix Research, Manchester, UK.
54.		Urban Z. Gene discovery in cutis laxa. Presented at: Gordon Research Conference on Elastin, Elastic Fibers & Microfibrils; 2013 Jul 21-26; Biddeford, ME.
55.		Urban Z. Unusual elastin mutations in cutis laxa. Presented at: 9th European Elastin Meeting, June 17-19 2016; Stuttgart, Germany.
56.		Khatri SM, Urban Z. Fibulin-4a enhances vascular and inhibits cardiac progenitor fate in the lateral plate mesoderm by inhibiting transforming growth factor beta signaling. Presented at: Gordon Research Conference on Elastin, Elastic Fibers & Microfibrils, July 30-August 4, 2017; Biddeford, ME.

57.		Zorrilla M, Givi JP, Watkins SC, Roman BL, Urban Z. Zebrafish elastin a and elastin b are required for cardiac valve development and juvenile survival. Presented at: 10th European Elastin Meeting, Nijmegen, Netherlands, June 14-16, 2018
58.		Zorrilla M, Givi JP, Waqas K, Watkins SC, Kim K, Roman BL, Urban Z. Zebrafish elastin a is required for cardiac valve development. NIH Workshop, Heart and Soul, Brain Behavior, and cardiovascular gene dosage effects in 7q11.23 and 22q11.2 CNVs, Bethesda, MD, April 2-3, 2019
59		Urban Z. Zebrafish models of elastic fiber diseases Presented at: Gordon Research Conference on Elastin, Elastic Fibers & Microfibrils, July 21-26, 2019; Manchester, NH.

OTHER PRESENTATIONS

1.		Olson TM, Michels VV, Urban Z, Csiszar K, Christiano AM, Driscoll DJ, Feldt RH, Boyd CD, Thibodeau SN. An intragenic deletion in the elastin gene in a family with supravalvular aortic stenosis (SVAS). Poster presented at: East Coast Connective Tissue Society, Fifteenth Annual Meeting; 1995 Mar 19-20; Somerset, NJ.
2.		Urban Z, Kiss E, Read C, Fekete G, Donis-Keller H, Csiszar K, Thibodeau SN, Boyd CD. Detection of mutational heterogeneity in SVAS patients using a tetranucleotide repeat polymorphism within the first intron of the elastin gene. Poster presented at: Gordon Conference - Elastin and Elastic Fibers; 1995 Jul 23-28; Meriden, NH.
3.		Olson TM, Michels VV, Urban Z, Csiszar K, Christiano AM, Driscoll DJ, Feldt RH, Boyd CD, Thibodeau SN. A 30 kb deletion within the elastin gene results in familial supravalvular aortic stenosis. Poster presented at: Gordon Conference - Elastin and Elastic Fibers; 1995 Jul 23-28; Meriden, NH.
4.		Felsen D, Lemack GE, Szabo Z, McConnell F, Urban Z, Csiszar K, Vaughan D, Boyd CD. Expression of rat topoelastin in transgenic mouse bladder: physiologic and histologic effects. Poster presented at: Gordon Conference - Elastin and Elastic Fibers; 1995 Jul 23-28; Meriden, NH.
5.		Urban Z, Csiszar K, Thibodeau SN, Boyd CD. Isolated supravalvular aortic stenosis is characterized by a spectrum of mutations within the elastin gene. Poster presented at: East Coast Connective Tissue Society Meeting; 1996 Mar 22-23; New York, NY.
6.		del Rio T, Urban Z, Boyd CD. The detection of elastin gene deletions in Williams syndrome patients by gene dosage PCR. Poster presented at: East Coast Connective Tissue Society Meeting; 1996 Mar 22-23; New York, NY.
7.		Mohl M, Urban Z, Csiszar K, Schilling K, Green M, Boyd CD. A highly informative PCR-based assay for the quantitation of low-abundance levels of tropoelastin mRNA. Presented at: East Coast Connective Tissue Society Meeting; 1996 Mar 22-23; New York, NY.
8.		Urban Z, Helms C, Peyrol S, Fekete G, Csiszar K, Bonnet D, Munnich A, Donis-Keller H, Boyd CD. Williams syndrome is characterized by 1 megabase 7q deletions encompassing the elastin gene. Presented at: The Seventh

		International Professional Williams Syndrome Conference; 1996 Jul 23-25; Valley Forge, PA.
9.		Urban Z, Kadar K, Kiss E, Csiszar K, Boyd CD, Bojeldein S, Szabolcs J, Fekete G. Williams syndrome: Cardiac and molecular genetic diagnosis. Presented at: Association of European Pediatric Cardiologists (AEPC) Annual Meeting; 1998 Jun 10-13; Dublin, Ireland.
10.		Le Saux O, Urban Z, Csiszar K, Terry S, Pope M, Lebwohl M, Pasquali-Ronchetti I, Boyd CD. Pseudoxanthoma elasticum is a recessive disorder linked to a gene on chromosome 16p13.1. Poster presented at: XVIth Meeting of the Federation of the European Connective Tissue Societies; 1998 Aug 1-6; Uppsala, Sweden.
11.		Urban Z, Sadler LS, Pober BR, Scheiber D, Fekete G, Sharma AN, Grandinetti A. Gender differences in cardiovascular disease in Williams syndrome (WS). Poster presented at: 8th International Professional Conference on Williams Syndrome; 2000 Jul 21-23; Dearborn, MI.
12.		Scheiber D, Szabolcs J, Urban Z, Sharma AN, Kiss E, Csoszanszky N, Fekete G. The natural history of cardiovascular disease in Hungarian patients with Williams-Beuren syndrome. Poster presented at: 8th International Professional Conference on Williams Syndrome, Dearborn, Michigan; 2000 Jul 21-23; Dearborn, MI.
13.		Sadler LS, Urban Z, Pober BR, Scheiber D, Fekete G, Sharma AN, Grandinetti A. Gender differences in cardiovascular disease in Williams syndrome. Presented at: XXI David W. Smith Workshop on Malformation and Morphogenesis; 2000 Aug 2-5; La Jolla, CA.
14.		Urban Z, Katahira J, Davis EC, Wallace MR, Michels VV, Thibodeau SN, Devriendt K, Boyd CD. Molecular genetic and biochemical analysis of supravalvular aortic stenosis: insights into the development of obstructive vascular disease. Poster presented at: Conference on Biology and Pathology of the Extracellular Matrix; 2000 Oct 12-15; St. Louis, MO.
15.		Le Saux O, Urban Z, Tschuch C, Csiszar K, Bacchelli B, Quaglino D, Pasquali-Ronchetti I, Pope FM, Richards A, Terry S, Bercovitch L, de Paepe A, Boyd CD. Mutations in a gene encoding an ABC transporter (ABCC6) cause pseudoxanthoma elasticum. Poster presented at: International Conference on Biology and Pathology of the Extracellular Matrix; 2000 Oct 12-15; St. Louis, MO.
16.		Ilias A, Urban Z, Seidl TL, Szakacs G, Le Saux O, Boyd CD, Varadi A. Expression, transport and ATPase activity of human MRP6/ABCC6, a membrane transporter mutated in pseudoxanthoma elasticum. Poster presented at: 3rd FEBS Advanced Course, ATP-Binding Casette (ABC) Proteins: From Genetic Disease to Multidrug Resistance; 2001 Mar 3-10; Gosau, Austria.
17.		Le Saux O, Urban Z, Beck K, Sachsinger C, Sylvestri C, de Paepe A, Pasquali-Ronchetti I, Pope FM, Terry S, Boyd CD. A spectrum of ABCC6 (MRP6) mutations are responsible for pseudoxanthoma elasticum. Presented at: Society for Investigative Dermatology, 62nd Annual Meeting; 2001 Mar 9-12; Washington, DC.
18.		Riazi S, Chitayat D, Urban Z, Hinek A. The role of elastin and elastin receptor subunit in the modulation of cellular proliferation. Presented at: Canadian Cardiovascular Congress; 2001 Oct 20-24; Halifax, Nova Scotia, Canada.

19.		Seidl TL, Katahira J, Skuta G, Smoot LB, Michels VV, Babovic-Vuksanovic D, Boyd CD, Urban Z. Functional haploinsufficiency of the elastin gene: a unifying pathomechanism for supravalvular aortic stenosis. Poster presented at: American Heart Association Asia Pacific Scientific Forum; 2002 Apr 23-26; Honolulu, HI.
20.		Wagner K, Seidl TL, Yanagisawa H, Urban Z. Mutational and expression studies on fibulin-5 in patients with aortic tortuosity and cutis laxa. Poster presented at: American Heart Association Asia Pacific Scientific Forum; 2002 Apr 23-26; Honolulu, HI.
21.		Urban Z, Riazi S, Nyerki P, Skuta G, Hinek A. Inverse relationship between cell proliferation and elastin deposition in dermal and vascular cells. Poster presented at: American Heart Association Asia Pacific Scientific Forum; 2002 Apr 23-26; Honolulu, HI.
22.		Davis EC, Gao J, Pope FM, Urban Z. A tandem duplication within the elastin gene is associated with autosomal dominant cutis laxa. Poster presented at: Elastin 2002, Second European Symposium; 2002 Jul 10-14; Aquafredda di Maratea, Italy.
23.		Wagner K, Yanagisawa H, Urban Z. Mutational and expression sutudies on fibulin-5 in patients with aortic tortuosity and cutis laxa. Poster presented at: Elastin 2002, Second European Symposium; 2002 Jul 10-14; Aquafredda di Maratea, Italy.
24.		Urban Z, Katahira J, Wagner K, Boyd CD, Kirsch WM, Weller S, Lennart A, Dickson C, Gundry SR. A candidate gene approach to investigate the molecular basis of a complex aneurysm case. Presented at: The Western Neurosurgical Society, 48th Annual Meeting; 2002 Oct 12-15; Victoria, BC, Canada.
25.		Crepeau M, Nyerki P, Davis EC, Pope FM, Hinek A, Urban Z. Elastin gene mutations in supravalvular aortic stenosis, pulmonary artery stenosis and cutis laxa: distinct disease mechanisms. Poster presented at: American Society for Matrix Biology, First Meeting; 2002 Nov 6-9; Houston, TX.
26.		Urschitz J, Iobst S, Urban Z, Granda C, Souza KM, Lupp C, Schilling K, Scott I, Csiszar K, Boyd CD. A serial analysis of gene expression in sun-damaged human skin. Poster presented at: American Society for Matrix Biology, First Meeting; 2002 Nov 6-9; Houston, TX.
27.		Hu Q, Wagner K, Crepeau MW, Reymond JL, Zabot MT, Yanagisawa H, Urban Z. Digenic inheritance of cutis laxa. Poster presented at: Gordon Conference on Elastin and Elastic Fibers; 2003 Aug 3-8; Meriden, NH.
28.		Hucthagowder V, Crepeau MW, Urban Z. DHPLC-based candidate gene scanning for cutis laxa. Poster presented at: Indian Society of Human Genetics, 29th Annual Meeting; 2004 Jan 8-11; Bangalore, India.
29.		Ruigrok YM, Wolterink S, Seitz U, Rinkel GJ, Wijmenga C, Urban Z. Association of polymorphisms and pairwise haplotypes in the elastin gene with aneurysmal subarachnoid hemorrhage. Presented at: 29th International Stroke Conference; 2004 Apr 28-May 1; San Diego, CA.
30.		Urban Z, Ruigrok YM, Wolterink S, Bohn S, Wijmenga C, Rinkel GJ. Association of polymorphisms and haplotypes in the elastin gene in dutch patients with sporadic aneurysmal subarachnoid hemorrhage. Poster presented at: American Society of Human Genetics, 54th Annual Meeting; 2004 Oct 26-30; Toronto,

		Canada.
31.		Hucthagowder V, Coucke P, Fong K, Csiszar K, Urban Z. Lysyl oxidase genes in cutis laxa. Poster presented at: American Society of Human Genetics, 54th Annual Meeting; 2004 Oct 26-30; Toronto, Canada.
32.		Hu Q, Wagner K, Reymond JL, Zabot MT, Yanagisawa H, Urban Z. Digenic inheritance of cutis laxa. Poster presented at: American Society for Matrix Biology, 2nd Meeting; 2004 Nov 10-13; San Diego, CA.
33.		Szabo Z, Crepeau MW, Stephan MJ, Puntel RA, Mitchell AL, Loke KY, Kirk RC, Urban Z. Elastin gene defects in cutis laxa associated with thoracic aortic aneurysms. Poster presented at: American Society for Matrix Biology, 2nd Meeting; 2004 Nov 10-13; San Diego, CA.
34.		Hucthagowder V, Fong K, Csiszar K, Urban Z. Lysyl oxidase-like gene mutation in cutis laxa. Poster presented at: Society for Investigative Dermatology, 66th Annual Meeting; 2005 May 4-7; St. Louis, MO.
35.		Szabo Z, Mitchell AL, Stephan MJ, Pope FM, Davis EC, Urban Z. Elastin gene mutations: a novel mechanism causing cutis laxa. Poster presented at: Society for Investigative Dermatology, 66th Annual Meeting; 2005 May 4-7; St. Louis, MO.
36.		Marler JA, Elfenbein JL, Ryals B, Urban Z, Netzloff ML. Sensorineural hearing loss in children and young adults with Williams syndrome. Poster presented at: International Evoked Response Audiometry Study Group Biennial Symposium; 2005 Jun 12-16; Havana, Cuba.
37.		Hu Q, Reymond JL, Pinel N, Zabot MT, Coucke PJ, Urban Z. Inflammatory destruction of elastic fibers in acquired cutis laxa is associated with mutations in the elastin and fibulin-5 genes. Poster presented at: Gordon Conference on Elastin and Elastic Fibers; 2005 Jul 31-Aug 5; Waterville Valley, NH.
38.		Marler JA, Elfenbein JL, Ryals B, Netzloff ML, Urban Z. Sensorineural hearing loss in Williams syndrome and supravalvular aortic stenosis: a novel role for elastin in auditory function. Poster presented at: Gordon Conference on Elastin and Elastic Fibers; 2005 Jul 31-Aug 5; Waterville Valley, NH.
39.		Marler JA, Elfenbein JL, Ryals BM, Urban Z, Netzloff ML. Auditory Acuity in Williams Syndrome & Supravalvular Aortic Stenosis. Poster presented at: The Annual American Speech-Language-Hearing Association Convention; 2005 Nov 18-20; San Diego, CA.
40.		Marler JA, Ryals BM, Kujawa SG, Urban Z. DPOAE and ABR measures of auditory function in ELN-heterozygous knockout mice. Presented at: Association for Research in Otolaryngology 27th Mid-Winter Meeting; 2006 Feb 5-9; Baltimore, MD.
41.		Callewaert B, Albrecht B, Loeys B, Gillessen-Kaesbach G, Hausser I, Vanakker O, Coucke PJ, Urban Z, De Paepe A. Two novel mutations in the ELN gene in patients with autosomal dominant cutis laxa and systemic manifestations. Poster presented at: 38th European Human Genetics Conference; 2006 May 6-9; Amsterdam, Netherlands.
42.		Wachi H, Sato F, Nonaka R, Szabo Z, Urban Z, Maeda I, Okamoto K, Starcher BC, Li DY, Mecham RP, Seyama Y. Involvement of exon 16 and 17 in tropoelastin molecule on the elastic fiber formation. Poster presented at: 4th European

		Meeting on Elastin; 2006 Jul 9-12; Lyon, France.
43.		Callewaert B, Loeys B, Albrecht B, Gillessen-Kaesbach G, Hausser I, Vanakker O, Coucke PJ, Urban Z, De Paepe A. Two novel mutations in the ELN gene in patients with autosomal dominant cutis laxa and systemic manifestations. Poster presented at: 4th European Meeting on Elastin; 2006 Jul 9-12; Lyon, France.
44.		Marler JA, Urban Z. Auditory function in Williams syndrome and supravalvular aortic stenosis. Poster presented at: 11th International Professional Conference on Williams Syndrome; 2006 Jul 23-24; Richmond, VA.
45.		Hucthagowder V, Sausgruber N, Kim KH, Angle B, Marmorstein LY, Urban Z. A loss of function mutation in the fibulin-4 gene causes a severe form of recessive cutis laxa. Poster presented at: American Society of Human Genetics 56th Annual Meeting; 2006 Oct 9-13; New Orleans, LA.
46.		Maxfield AB, Hucthagowder V, Gitlin JD, Joseph EM, Urban Z. Fibulin-4 function in zebrafish. Poster presented at: American Society of Matrix Biology Biennial National Meeting; 2006 Nov 1-4; Nashville, TN.
47.		Hu Q, Choi J, Davis EC, Szabo Z, Shifren A, Knutsen RH, Pierce RA, Shipley JM, Mecham RP, Urban Z. A transgenic mouse model of autosomal dominant cutis laxa. Poster presented at: American Society of Matrix Biology Biennial National Meeting; 2006 Nov 1-4; Nashville, TN.
48.		Hu Q, Coucke PJ, Sommer P, Davis EC, Reinhardt DP, Mecham RP, Urban Z. Fibulin-5 mutations in cutis laxa. Poster presented at: American Society of Matrix Biology Biennial National Meeting; 2006 Nov 1-4; Nashville, TN.
49.		Marler JA, Roy JL, Urban Z. Auditory function in connective tissue disorders. Poster presented at: The Annual American Speech-Language-Hearing Association Convention; 2006 Nov 16-18; Miami Beach, FL.
50.		Hu Q, Shifren A, Choi J, Szabo Z, Starcher BC, Knutsen RH, Shipley JM, Davis EC, Mecham RP, Urban Z. Emphysematous lung disease in a transgenic mouse model of autosomal dominant cutis laxa. Poster presented at: Gordon Conference on Elastin and Elastic Fibers; 2007 Jul 29-Aug 3; Biddeford, ME.
51.		Miller-Smith L, Urban Z, Mecham RP, Shifren A. Pulmonary Circulatory Abnormalities in Human Elastinopathies. Poster presented at: American Thoracic Society International Conference; 2008 May 16-21; Toronto, Canada.
52.		Hu Q, Shifren AS, Choi J, Szabo Z, Starcher BC, Knutsen RH, Shipley JM, Davis EC, Mecham RP, Urban Z. Mechanisms of emphysema in autosomal dominant cutis laxa. Poster presented at: 5th European Elastin Meeting; 2008 Jul 16-19; Alcalá De Henares, Spain.
53.		Maxfield AB, Hucthagowder V, Joseph EM, Urban Z. Fibulin-4 function in cardiovasular development. Poster presented at: 5th European Elastin Meeting; 2008 Jul 16-19; Alcalá De Henares, Spain.
54.		Maxfield AB, Khatri SM, Mecham RP, Joseph EM, Urban Z. Fibulin-4 is necessary for the development of the notochord and the cardiovascular system. Poster presented at: 6th European Elastin Meeting; 2010 Jun 28-Jul 2; Maratea, Italy.
55.		Maxfield AB, Khatri SM, Mecham RP, Joseph EM, Urban Z. Fibulin-4 is necessary for the development of the notochord and the cardiovascular system. Presented at: American Society of Matrix Biology, 2010 Biennial Meeting; 2010

		Oct 24-27; Charleston, SC.
56.		Willaert A, Callewaert BL, Coucke PJ, Crosby SD, Loeys BL, De Paepe A, Urban Z. GLUT10 is required for the development of the cardiovascular system and the notochord and connects cellular metabolism to TGFß signaling. Presented at: American Society of Matrix Biology, 2010 Biennial Meeting; 2010 Oct 24-27; Charleston, SC.
57.		Opoka A, Osinska H, Juraszek AL, Doyle J, Eghtesady P, Urban Z, Dietz HC, Mecham RP, Aronow BJ, Bove KE, Hinton RB. Elastic Fiber Dysregulation in Syndromic and Nonsyndromic Aortic Valve Disease. Poster presented at: American Society of Matrix Biology, 2010 Biennial Meeting; 2010 Oct 24-27; Charleston, SC.
58.		Urban Z, Maxfield AB, Khatri SM, Mecham RP, Joseph EM. Fibulin-4 is required for the development of the notochord and the cardiovascular system. Poster presented at: The American Society of Human Genetics, 60th Annual Meeting; 2010 Nov 2-6; Washington, DC.
59.		Westman R, Henger S, Shifren A, Yusen RD, Mecham RP, Sciurba FC, Urban Z. Chronic lung disease in patients with cutis laxa. Poster presented at: The American Society of Human Genetics, 60th Annual Meeting; 2010 Nov 2-6; Washington, DC.
60.		Khatri SM, Maxfield AB, Mecham RP, Joseph EM, Urban Z. Fibulin-4b is required for cardiovascular system development and as an antagonist of transforming growth factors tgfb2 and tgfb3. Poster presented at: Gordon Conference on Elastin and Elastic Fibers; 2011 Jul 24-29; Biddeford, ME.
61.		Urban Z, Willaert A, Khatri SM, Callewaert BL, Coucke P, Crosby SD, Loeys BL, Tsang M, De Paepe A. GLUT10 connects TGFß signaling to cellular metabolism in cardiovascular development. Poster presented at: 12th International Congress of Human Genetics, The American Society of Human Genetics, 61th Annual Meeting; 2011 Oct 11-15; Montreal, Canada.
62.		Levine K, Mac Neal M, Sciurba FC, Urban Z. Genotype-phenotype correlations for pulmonary manifestations of cutis laxa. Poster presented at: Pittsburgh International Lung Conference, Personalized Medicine of Lung Disease; 2011 Oct 28-29; Pittsburgh, PA.
63.		Marler JA, Goodman SS, Urban Z. Middle-ear anomalies and cochlear function in elastic-fiber disorders. Presented at: 2012 MidWinter Meeting of the Association for Research in Otolaryngology; 2012 Feb 25-29; San Diego, CA.
64.		Lawrence EC, McGowan ME, Su CT, Mac Neal M, Levine K, Zaenglein A, Bodzioch M, Kiss A, Urban Z. ELN mutations in autosomal dominant cutis laxa. Poster presented at: The American Society of Human Genetics, 62nd Annual Meeting; 2012 Nov 6-10; San Francisco.
65.		Callewaert B, Su CT, Van DT, Vlummens P, Malfait F, Vanakker O, Schulz B, Mac Neal M, Davis EC, Lee JG, Salhi A, Unger S, Heimdal K, De Almeida S, Kornak U, Gaspard H, Bresson JL, Prescott K, Gosendi ME, Mansour S, Pierard GE, Madan-Khetarpal S, Sciurba FC, Symoens S, Coucke PJ, Van Maldergem L, Urban Z, De Paepe A. Comprehensive clinical and molecular analysis of 12 families with type I recessive cutis laxa. Presented at: The American Society of Human Genetics, 62nd Annual Meeting; 2012 Nov 6-10; San Francisco, CA.

66.		Urban Z, Khatri SM, Maxfield AB. Fibulin-4b is required for cardiovascular and musculoskeletal development as an antagonist of transforming growth factor-beta. Poster presented at: The American Society of Human Genetics, 62nd Annual Meeting; 2012 Nov 6-10; San Francisco, CA.
67.		Khatri SM, Maxfield AB, Joseph EM, Urban Z. Antagonistic regulation of transforming growth factor-beta by fibulin-4a and fibulin-4b is required for cardiovascular and musculoskeletal development in zebrafish. *Winner of ASMB travel award. Poster presented at: Joint Meeting of the Society for Glycobiology & American Society for Matrix Biology; 2012 Nov 11-14; San Diego, CA.
68.		Su CT, Weckenmann C, Lawrence EC, Urban Z. LTBP4 regulates both the magnitude and the direction of TGFß response. Poster presented at: Joint Meeting of the Society for Glycobiology & American Society for Matrix Biology; 2012 Nov 11-14; San Diego, CA.
69.		Khatri SM, Maxfield AB, Joseph EM, Urban Z. Antagonistic regulation of transforming growth factor-beta by fibulin-4a and fibulin-4b is required for cardiovascular and musculoskeletal development in zebrafish. Poster presented at: Gordon Research Conference on Elastin, Elastic Fibers & Microfibrils; 2013 Jul 21-26; Biddeford, ME.
70.		Lorenchick C, Weinberg S, Levine K, Madan-Khetarpal S, Grubs RE, Urban Z. Craniofacial phenotype in cutis laxa. Poster presented at: 32nd Annual Education Conference of the National Society of Genetic Counselors; 2013 Oct 9-12; Anaheim, CA.
71.		Urban Z, Parsons TE, Lorenchick C, Levine KL, Madan-Khetarpal S, Weinberg SM. Craniofacial phenotypes in cutis laxa. Poster presented at: Society of Craniofacial Genetics and Developmental Biology, 36th Annual Meeting; 2013 Oct 22; Boston, MA.
72.		Lawrence EC, McGowan M, Levine KL, Lorenchick C, Alkan S, Salvaggio H, Zaenglein A, Bodzioch M, Kiss A, Siefring M, Urban Z. Novel ELN mutations and vascular phenotype in autosomal dominant cutis laxa. Presented at: The American Society of Human Genetics, 63rd Annual Meeting; 2013 Oct 22-26; Boston, MA.
73.		Urban Z, Lorenchick C, Parsons TE, Levine KL, Madan-Khetarpal S, Weinberg SM. Craniofacial phenotypes in cutis laxa. Poster presented at: The American Society of Human Genetics, 63rd Annual Meeting; 2013 Oct 22-26; Boston, MA.
74.		Wilson RC, Chandra D, Urban Z, Bon JM, Isenberg JS, Karoleski CM, Champion HC, Morris A, Sciurba FC. Association of skin elasticity with pulmonary diffusing capacity, markers of peripheral endothelial function, and arterial stiffness in smokers. Poster presented at: American Thoracic Society, 2014 International Conference; 2014 May 16-21; San Diego, CA.
75.		Su CT*, Lawrence EC, Dabovic B, Rifkin DB, Urban Z. LTBP4 regulates TGFβ signaling by stabilizing TGFβ receptors. American Society for Matrix Biology, Biennial Meeting, 2014 Oct 12-15, Cleveland, OH. *Winner of ASMB Travel Award.
76.		Khatri SM, Maxfield AB, Joseph EM, Urban Z. Antagonistic regulation of transforming growth factor-beta by fibulin-4a and fibulin-4b is required for cardiovascular and musculoskeletal development in zebrafish. American Society

		for Matrix Biology, Biennial Meeting, 2014 Oct 12-15, Cleveland, OH.
77.		Urban Z, Kozel BA, Su CT, Sciurba FC, Weinberg SM, Madan-Khetarpal S, Lorenchick C, Lawrence EC, Levine KL. Multi-system phenotyping in cutis laxa. American Society for Matrix Biology, Biennial Meeting, 2014 Oct 12-15, Cleveland, OH.
78.		Alkan S, Urban Z. TGFβ signaling in patients with ELN-related cutis laxa. American Society for Matrix Biology, Biennial Meeting, 2014 Oct 12-15, Cleveland, OH.
79.		Su CT, Huang JW, Chiang CK, Dabovic B, Rifkin DB, Urban Z. Latent transforming growth factor beta binding protein 4 (Ltbp4) enhances renal fibrosis in obstructive nephropathy. American Society of Nephrology, Kidney Week 2014, 2014 Nov 11-16, Philadelphia, PA.
80.		Wilson R, Chandra D, Bon J, Karoleski CM, Morris AM, Fuhrman CR, Leader JK, Urban Z, Sciurba FC. Skin elasticity is associated with emphysema in smokers: the Pittsburgh SCCOR cohort. American Thoracic Society, 2015 International Conference, 2015 May 15-20, Denver, CO.
81.		Alkan S, Urban Z. Mutation-specific TGFβ singaling changes in patients with ELN-related cutis laxa. Gordon Research Conference on Elastin, Elastic Fibers & Microfibrils, 2015 Jul 26-31, Biddeford, ME. Winner of the 2016 Human Genetics travel award
82.		Khatri S, Maxfield AB, Joseph EM, Urban Z. Antagonistic regulation of transforming growth factor-beta by fibulin-4a and fibulin-4b is required for cardiovascular development. Gordon Research Conference on Elastin, Elastic Fibers & Microfibrils, 2015 Jul 26-31, Biddeford, ME.
83.		Lawrence E, McGowan M, Levine K, Lorenchick C, Alkan S, Madan-Khetarpal S, Urban Z. Novel ELN mutations and vascular phenotype in autosomal dominant cutis laxa. Gordon Research Conference on Elastin, Elastic Fibers & Microfibrils, 2015 Jul 26-31, Biddeford, ME.
84.		Su CT, Urban Z. LTBP4 regulates TGFβ receptor stability. Gordon Research Conference on Elastin, Elastic Fibers & Microfibrils, 2015 Jul 26-31, Biddeford, ME.
85.		Zorrilla M, Urban Z. Elastin function in age-related vascular diseases. Gordon Research Conference on Elastin, Elastic Fibers & Microfibrils, 2015 Jul 26-31, Biddeford, ME.
86.		Alkan S, Urban Z. Genotype-dependent alterations in TGFβ signaling in ELN-related cutis laxa. The American Society of Human Genetics, 65th Annual Meeting, 2015 Oct 6-10, Baltimore, MD.
87.		Urban Z, Huang JW, Chiang CK, Lawrence EC, Dabovic B, Su CT. Transforming growth factor beta receptor stability and signaling in LTBP4-related cutis laxa. The American Society of Human Genetics, 65th Annual Meeting, 2015 Oct 6-10, Baltimore, MD.
88.		Wilson R, Chandra D, Bon J, Karoleski CM, Morris AM, Fuhrman CR, Leader JK, Zhang Y, Morris Z, Urban Z, Sciurba FC. Facial wrinkling is associated with emphysema independent of airflow obstruction: the Pittsburgh SCCOR cohort. American Thoracic Society, 2016 International Conference, San Francisco, CA, May 13-18, 2016

89.		Zhao Y, Davidson LA, Watkins SC, Lewis DW, Urban Z, Adibi JJ. A light shines through the trees: fluid-filled bulb formation from placental villous explants in long-term culture. International Federation of Placenta Associations, 2016 Meeting, Portland, OR, September 13-16, 2016
90.		Akcay S, Lawrence EC, Urban Z. Increased expression of mutant elastin alleles in autosomal dominant cutis laxa is normalized by transforming growth factor beta treatment. The American Society of Human Genetics, 66th Annual Meeting, Vancouver, BC, Canada October 18-22, 2016
91.		Su CT, Minster RL, Hawley NL, Kershaw EE, Deka R, Weeks DE, McGarvey ST, Urban Z. Thrifty functional characteristics of the major variant in CREBRF associated with body mass index in Samoans. The American Society of Human Genetics, 66th Annual Meeting, Vancouver, BC, Canada October 18-22, 2016
92.		Minster RL, Hawley NL, Su CT, Sun G, Kershaw EE, Cheng H, Buhule OD, Lin J, Reupena MS, Viali S, Tuitele J, Naseri T, Urban Z, Deka R, Weeks DE, McGarvey ST (2016) A thrifty variant in CREBRF strongly influences body mass index in Samoans. The American Society of Human Genetics, 66th Annual Meeting, Vancouver, BC, Canada October 18-22, 2016
93.		Khatri SM, Maxfield AB, Joseph EM, Urban Z. Fbln4a is a maternal gene regulating sonic hedgehog signaling and the fate of cardiovascular progenitors. American Society for Matrix Biology, Biennial Meeting, St. Petersburg, FL, November 13-16, 2016
94.		Zorrilla M, Urban Z. Contribution of elastin to cardiovascular development in zebrafish. American Society for Matrix Biology, Biennial Meeting, St. Petersburg, FL, November 13-16, 2016
95.		Watkin LB, Morrow M, Vargas-Hernandez A, Mace EM, Urban Z, Orange JS. Mutations in the H^+-ATPase subunit ATP6V0A2 associated with cutis laxa also cause NK cell deficiency. Clinical Immunology Society, 2017 Annual Meeting, Immune Deficiency and Dysregulation North American Conference, Seattle, WA, March 23-26, 2017
96.		Lawrence EC, Rosenthal SL, Schusterman MA, Kokai LE, Rubin P, Kershaw EE, Urban Z. Simultaneous extraction of DNA, RNA and protein for genomic and proteomic applications from fat tissue. European Human Genetics Conference, Copenhagen, Denmark, May 27-30, 2017
97.		Dreikorn E, Urban Z. Cutis laxa mutations decrease the secretion of fibulin-5, leading to impaired elastic fiber formation. Gordon Research Conference on Elastin, Elastic Fibers & Microfibrils, Biddeford, ME, July 30-August 4, 2017
98.		Givi JP, Peck J, Wallace C, Watkins SC, Urban Z. Automated histochemical image capture and analysis to quantify dermal elastin in cutis laxa. Gordon Research Conference on Elastin, Elastic Fibers & Microfibrils, Biddeford, ME, July 30-August 4, 2017
99.		Zorrilla M, Urban Z. Contribution of elastin to cardiovascular development in zebrafish. Gordon Research Conference on Elastin, Elastic Fibers & Microfibrils, Biddeford, ME, July 30-August 4, 2017
100.		Khatri SM, Urban Z. Fibulin-4a inhibits vascular and enhances cardiac cell fate by inhibiting transforming growth factor beta signaling. American Society of Human Genetics, 2017 Annual Meeting, Orlando, FL, October 17-21, 2017

111.		Lawrence EC, Rosenthal SL, Schusterman MA, Kokai LE, Rubin P, Kershaw EE, Urban Z. Optimizing simultaneous isolation of biomolecules and cells from cryopreserved adipose tissue for omics applications. American Society of Human Genetics, 2017 Annual Meeting, Orlando, FL, October 17-21, 2017
112.		Watkin LB, Morrow M, Vargas-Hernandes A, Forbes LR, Stray-Pedersen A, Issekutz T, Issekutz AC, Lupski JR, Mace EM, Urban Z, Orange JS. ATP6V0A2 is required for NK cell expansion and function. NK2018 – The 17th meeting of the Society for Natural Immunity, San Antonio, TX, May 28-June 1, 2018
113.		Lawrence EC, Morava E, Mohamed M, Waugh MG, Wevers RA, Urban Z. Reduced elastin deposition in PI4K2A-related cutis laxa. 10th European Elastin Meeting, Nijmegen, Netherlands, June 14-16, 2018
114.		Dreikorn EN, Urban Z. Custis laxa mutations in fibulin-5 decrease elastin deposition, GPCR and TGF-β signaling. American Society for Matrix Biology Biennial Meeting, Las Vegas, NV, October 14-17, 2018
115.		Zorrilla M, Givi JP, Watkins SC, Roman BL, Kim K, Urban Z. Zebrafish elastin is required for cardiac valve development and function. American Society for Matrix Biology Biennial Meeting, Las Vegas, NV, October 14-17, 2018
116.		Lawrence EC, Morava E, Liu F, Mohamed M, Waugh MG Wevers RA, Urban Z. Reduced elastin deposition in a PI4K2A-related cutis laxa patient. American Society of Human Genetics, 68th Annual Meeting, San Diego, CA, October 16-20, 2018
117.		Rosenthal SL, Liu F, Lawrence EC, Su CT, Hawley NL, McGarvey ST, Urban Z. Transcript profile of a CRISPR derived CREBRF exon deletion in 3T3-L1 preadipocytes. American Society of Human Genetics, 68th Annual Meeting, San Diego, CA, October 16-20, 2018
118.		Beyens A, De Rycke R, Syryn H, Fischer-Zirnsak B, Van Damme T, Hausser I, De Bruyne M, Morroni M, Nampoothiri S, Mahesh K, Kornak U, Urban Z, Hadj-Rabia S, Bodemer C, De Schepper S, Davis EC, Callewaert B. Ultrastructural elastic fiber morphology in cutis laxa reflects the underlying pathogenesis and supports a novel clinical classification. European Human Genetics Conference, Gothenburg, Sweden June 15-18, 2019

TEACHING

Graduate Courses

Year(s)	Course Number & Title	Role
1997 - 2004	CMB 621, Molecular Biology of the Cell	Guest Lecturer
2000 - 2004	Cell and Molecular Biology (CMB)	Guest Lecturer
2001 - 2002	ANAT 695, Directed Research	Primary Instructor
2001 - 2004	BIOM 699, Directed Reading	Primary Instructor
2006 - 2008	Pediatric Fellows' Core Lecture: "Getting Published",	Guest Lecturer

2007 - 2009	BIO 5128, Extracellular Matrix and Cell Matrix Interactions Journal Club	Coursemaster
2008 - 2009	Summer Institute Program to Increase Diversity, Lecture: “Genetic Epidemiology of COPD”	Guest Lecturer
2009 - 2009	BIO 5258, Fundamentals of Mammalian Genetics	Guest Lecturer
2010 - 2010	INTBP 2290, Scientific Ethics and the Responsible Conduct of Research	Breakout session moderator
2010 - Present	HUGEN 2040, Molecular Basis of Human Inherited Disease	Co-Instructor
2011 - 2014	HUGEN 2036 Genetic Counseling Internship	Guest Lecturer
2011 - 2019	HUGEN 2051, Inborn Errors of Development	Co-Instructor
2011 - 2017	HUGEN 2025 Human Genetics Seminar Spring	Primary Instructor
2012 - 2016	Human Genetics Summer Research in Progress Seminar	Primary Instructor
2017, 2020	Human Genetics Summer Research in Progress Seminar	Co-Instructor
2013 - Present	MSCMP 3735 Extracellular Matrix in Tissue Biology and Bioengineering	Guest Lecturer
2014	HUGEN 2027 Human Genetics Fall Journal Club	Co-Instructor
2014 - Present	MS 1, Tissues in Health and Disease	Guest Lecturer
2020 - Present	HUGEN 2028 Human Genetics Spring Journal Club	Primary Instructor

MENTORING AND ADVISING

Undergraduate Students

Year(s)	Student’s Name & Degree/Discipline	Advisor’s Role
2001 - 2002	Kerstin Wagner Fibulin mutations in cutis laxa	Research Advisor
2002 - 2003	Silke Wolterink Elastin gene variants in intracranial aneurysms	Research Advisor
2003 - 2004	Melanie Roth Elastin gene variants in lymphoblasts	Research Advisor
2005 - 2005	Michael Angstmann	Research Advisor

	Elastin gene variants in intracranial aneurysms	
2005 - 2005	Stefanie Lerche Transgenic mouse model of autosomal dominant cutis laxa	Research Advisor
2005 - 2006	Nina Sausgruber Fibulin mutations in cutis laxa	Research Advisor
2005 - 2006	Aleksander Keselman Elastin mutations in cutis laxa	Research Advisor
2006 - 2006	Carmen Fischer Elastin mutations in supravalvular aortic stenosis	Research Advisor
2006 - 2007	Lena Schreiber Lysyl oxidases in cutis laxa	Research Advisor
2007 - 2007	Lenny Jonggadipo Lysyl oxidases in cutis laxa	Research Advisor
2007 - 2008	Annika Aldinger ATP6V0A2 mutations in cutis laxa	Research Advisor
2008 - 2008	Carla Sens Transgenic mouse model of autosomal dominant cutis laxa	Research Advisor
2008 - 2009	Nura Schürmann Mutations and functions of LTBP4	Research Advisor
2009 - 2010	Mark Robertshaw COL8A1 mutations in cutis laxa	Research Advisor
2009 - 2010	Lena Collenburg Mutations and Functions of LTBP4	Research Advisor
2010 - 2010	Silvia Henger LTBP4 mutations in cutis laxa	Research Advisor
2010 - 2011	Christine Pfeiffer LTBP4 expression in skin fibroblasts	Research Advisor

2011 - 2011	Franziska Zuber Mutations and functions of LTBP4	Research Advisor
2011 - 2012	Maureen McGowan Elastin mutations in cutis laxa	Research Advisor
2011 - 2012	Bianca Schulz LTBP4 mutations in cutis laxa	Research Advisor
2012 - 2013	Amelie Bauer LTBP4 mutations in cutis laxa	Research Advisor
2013 - 2014	Kerstin Ehm Suppression of nonsense mediated mRNA decay in connective tissue diseases	Research Advisor
2013 - 2014	Sheila Longo GORAB mutations in cutis laxa	Research Advisor
2014	Daniel Thomas Suppression of nonsense mediated mRNA decay in connective tissue diseases	Research Advisor
2016 - 2017	Jerome Givi Automated histochemical image capture and analysis of elastin content Recipient: Brackenridge Fellowship 2017	Research Advisor

Master Students

Year(s)	**Student's Name & Degree/Discipline**	**Advisor's Role**
2000 - 2002	Thomas L. Seidl MS (2002) Elastin mutations in supravalvular aortic stenosis	Research Advisor
2003 - 2003	Ulrich Seitz MS (2011) Elastin gene variants in intracranial aneurysms	Research Advisor
2004 - 2005	Susanne Bohn MS (2005)	Research Advisor

	Elastin gene variants in intracranial aneurysms	
2010 - 2011	Shazina Saeed MPH (2011) Aortic aneurysm and its genetic causes	Research and Essay Advisor
2010 - 2011	Rachel Westman MSGC (2011) Chronic lung disease in cutis laxa	Research and Thesis Advisor
2010 - 2012	Meghan Mc Neal MSGC, MPH (2012) Pulmonary disease and disability from lung disease in cutis laxa	Research and Thesis Advisor
2011 - 2012	Chi-Ting Su MPH (2012) Circulating TGFß1 as a biomarker of COPD	Practicum and Essay Advisor
2012 - 2012	Christine Weckenmann MS (2013) LTBP4 mutations in cutis laxa	Research Advisor
2012 - 2014	Christa Lorenchick MSGC (2014) Craniofacial anomalies in cutis laxa	Research Advisor
2013 - 2013	Kathrin Hammon MS (2014) LTBP4 mutations in cutis laxa	Research Advisor
2015	Megan Breski MS (2015) Allele specific approach to study histone demethylation using engineered KDM4A-(2) ketoglutarate pairs	Thesis Committee Member
2015-2016	Amy Biery MSGC (2016) Expression of Alk1 is regulated by a positive feedback mechanism involving blood flow and circulating ligand	Thesis Committee Member

2015 - 2017	Martin Requena MS (2017) A cell culture model of MRPS2-related cutis laxa	Research Advisor, Thesis Committee Chair
2015 - 2017	Michelle Morrow MSGC (2017) Immune function in ATP6V0A2-related cutis laxa	Research Advisor, Thesis Committee Chair
2016 - 2017	Teresa Capasso MS	Thesis Committee Member
2016 - 2017	Jennifer Peck MSGC Skin thickness and elasticity in cutis laxa	Research Advisor
2017	Annie Arockiaraj MS	Comprehensive Committee Member
2017 - 2018	Emily Spoth MSGC Biomechanical properties of the skin in arterial tortuosity syndrome	Research Advisor, Thesis Committee Chair
2017 - 2019	Bijun Li MS (2019) Zebrafish blm10 mutatns as a model for HHT-associated high output heart failure	Comprehensive and Thesis Committee Member
2018 - 2019	Jerome Givi MPH (2019) Elastin b in cardiovascular development	Research Advisor
2018 - 2019	Zeynep Erdogan Yildirim MS (2019) Functional investigation of ESR1 fusions identified in endocrine therapy refractory estrogen receptor positive breast cancer	Comprehensive and Thesis Committee Member
2019 – 2020	Pooja Solanki MSCG, MPH (2020)	Research Advisor MPH Essay Reader
2020	Anushe Munir MS (2020)	Comprehensive and Thesis Committee

		Member

Doctoral Students

Year(s)	Student's Name & Degree/Discipline	Advisor's Role
1997 - 1997	Claudia Lupp PhD (2003) University of Hawaii Elastin mutations in supravalvular aortic stenosis	Research Advisor
1998 - 2000	Gregg Maeda PhD (2004) University of Hawaii Elastin mutations in supravalvular aortic stenosis	Research Advisor
2001 - 2002	Peter Nyerki, University of Hawaii Hydrophilic elastin peptides	Research Advisor
2002 - 2006	Qirui Hu PhD (2006) University of Hawaii Fibulin-5 in cutis laxa	Research Advisor
2003 - 2003	Sandra Dunn, University of Hawaii Elastin minigenes in transgenic mice	Research advisor
2006 - 2006	Chao-Tsung Yang PhD (2006) Washington University, St. Louis	Thesis Committee Member
2006 - 2008	Justin Weinberg PhD (2008) Washington University, St. Louis	Thesis Committee Member
2006 - 2008	John Gansner PhD (2008) Washington University, St. Louis	Thesis Committee Member
2008 - 2009	Bert Callewaert PhD (2010) Washington University, St. Louis Inherited Elastinopathies: Novel Clinical and Etiopathogenetic Insights	Research Advisor
2009 - 2010	Andy Willaert PhD (2010)	Research Advisor

	Washington University, St. Louis Molecular characterization of heritable connective tissue disorders affecting bone and arteries,	
2009 - 2009	Caroline E. Ridley PhD (2009) University of Manchester, UK	Thesis External Examiner
2010 - 2017	Sandeeep Khatri PhD (2017) Fibulin-4a in zebrafish development	Research Advisor Qualifying, Comprehensive and Dissertation Committee Member
2011 - 2011	Laetitia Sabatier PhD (2011) McGill University, Canada	Thesis External Examiner
2011 - 2012	Gulab Sher PhD LTBP4 mutations in cutis laxa	Research Advisor
2011 - 2013	Madhav Sankunny PhD (2013) The role of the ATR-CHEK1 pathway in therapeutic resistance resulting from distal 11q loss in carcinoma cells	Dissertation Committee Member
2011 - 2014	Heejung Kang PhD (2014) Medium chain acyl-CoA dehydrogenase deficiency	Dissertation Committee Member
2011 - 2014	Kaitlyn Kormanik PhD (2014) Characterization of the biochemical and physiological functions of acyl-CoA dehydrogenase 10	Dissertation Committee Member
2012 - 2012	Sulman Basit PhD (2012) Genetic mapping of genes involved in human hereditary alopecias and skeletal deformities	Thesis External Examiner

2012 - 2014	Chi-Ting Su PhD (2014) Molecular mechanisms of LTBP4-related cutis laxa 2014 winner of the 'Best Human Genetics PhD Dissertation' award	Research Advisor, Qualifying, Comprehensive and Dissertation Committee Member
2012 - 2015	Prya Mittal PhD (2015) The role of mediator complex subunit 12 (MED12) in the murine reproductive tract	Qualifying, Comprehensive and Dissertation Committee Member
2012 - 2016	Brianna Heath PhD (2016) MT1 receptor mediated neuroprotection in R6/2 mouse model of Huntington's disease	Qualifying, Comprehensive and Dissertation Committee Member
2012 - 2016	Sevinc Alkan PhD (2016) Molecular consequences of elastin gene mutations in autosomal dominant cutis laxa and supravalvular aortic stenosis	Research Advisor, Qualifying, Comprehensive and Dissertation Committee Member
2012	Meghan Beck PhD	Qualifying Committee Member
2013 - 2013	Andreja Simpson PhD (2013) Fibulin-4 mutations in cutis laxa	Thesis External Examiner
2013 - 2016	Lora McClain PhD (2013) HSV-1 infection in human iPSC-derived neurons: cellular models of quiescence and drug discovery	Qualifying, Comprehensive and Dissertation Committee Member
2013 - 2015	Hatem O. Kaseb PhD (2015) Spheroid-enriched cancer stem-like cells as a model for targeted therapy in oral cancer with distal 11q loss	Qualifying, Comprehensive and Dissertation Committee Member
2013 - 2015	Anatalia Labilloy PhD (2015)	Dissertation Committee Member

	Signaling pathways in cell models of Fabry disease nephropathy	
2013 - 2018	Michelle Zorrilla PhD (2018) Contribution of elastin to cardiovascular development in zebrafish	Research Advisor, Qualifying, Comprehensive and Dissertation Committee Chair
2014	Sofia Hussain PhD (2014) Molecular characterization of inherited kidney diseases in the Pakistani population	Thesis External Reviewer
2014 - 2017	Ian Casci PhD (2017) Identifying novel modifiers of Fus-associated toxicity in a Drosophila model of amyotrphic lateral sclerosis	Qualifying, Comprehensive and Dissertation Committee Member
2015	Brandon Blobner PhD	Qualifying Committee Member
2015 - 2018	Stephen McCalley PhD (2018) Elucidating the mitochondrial architecture of branched-chain amino acid metabolism enzymes: implications for treatment	Qualifying, Comprehensive and Dissertation Committee Member
2015 - 2017	Megan Breski PhD (2017) Engineering lysine demethylases to orthogonally probe cellular functions	Qualifying, Comprehensive and Dissertation Committee Member
2016 - Present	Katya Orlova PhD	Qualifying, Comprehensive and Dissertation Committee Member
2016 - 2019	Aneesh Ramaswamy PhD (2019) in Bioengineering, Swanson School of Engineering Adipose stromal cell-based elastogenesis therapy for adult and pediatric aortic defects	Dissertation Committee Member

2016 - 2020	Nandini Ramesh PhD	Qualifying, Comprehensive and Dissertation Committee Member
2016 - 2019	Bruce Nmezi PhD (2019) The role of lamin B1 in the organization of the nuclear envelope and myelin regulation in development and disease	Qualifying, Comprehensive and Dissertation Committee Member
2017 - 2019	Erika Dreikorn PhD Functional analysis of fibulin-5 mutations in cutis laxa	Research Advisor, Qualifying Committee Chair
2017 - 2019	Megan Eshbach PhD (2019) Tubular proteinuria and vitamin D deficiency in sickle cell disease	Comprehensive and Dissertation Committee Member
2019 - Present	Teresa Capasso PhD	Dissertation Committee Member
2019	Melissa Bulik PhD	Qualifying Committee Member
2019	Olivia D'Annibale PhD	Qualifying Committee Chair
2019	Laura Savariau PhD	Qualifying Committee Chair
2019 - Present	Alexandra Schmidt	Qualifying Committee Chair, Comprehensive and Dissertation Committee Member
2020 - Present	Annie Arockiaraj CRISPR/dCas9-mediated upregulation of *LAMA1* as a potential therapeutic approach for merosin deficient congenital muscular dystrophy (MDC1A)	Comprehensive and Dissertation Committee Member

2020 – Present	Tyler Fortuna Integrative Systems Biology Program Functional studies on GEMIN5-related spinal muscular atrophy	Comprehensive and Dissertation Committee Member
2020	Rachel Forman-Rubinsky Molecular Genetics and Developmental Biology Program Investigating the role of chromatin modification and Ras/MAPK singaling in hypoplastic left heart syndrome using zebrafish	Comprehensive Committee Member
2020	Talia Oranburg	Qualifying Committee Member
2020	Erika Dreikorn PhD	Dissertation Committee Member

Other Students

Year(s)	**Student's Name & Degree/Discipline**	**Advisor's Role**
1999 - 2000	Alok N. Sharma Williams syndrome	Research Advisor
2000 - 2002	Jodi Katahira Elastin mutations in supravalvular aortic stenosis	Research Advisor

Postdoc or Fellow

Year(s)	**Student's Name & Degree/Discipline**
2001 - 2002	Gabriella Skuta, MD, PhD Hydrophilic elastin peptides
2003 - 2003	Jeffrey Squires
2003 - 2004	Feng Hao Hydrophilic elastin peptides

2003 - 2005	Zoltan Szabo Transgenic mouse model of autosomal dominant cutis laxa
2003 - 2009	Vishwanathan Hucthagowder Genetics of cutis laxa
2006 - 2008	Andrew Maxfield Fibulin-4 in zebrafish development
2007 - 2009	Jyothirmai Talasila Williams syndrome heart study
2008 - 2009	Laura Miller-Smith, MD Williams syndrome heart study
2014 - 2015	Chi-Ting Su Functional studies on CREBRF in obesity
2017 - 2020	Samantha Rosenthal Cellular and genomic studies on CREBRF in obesity Recipient: T32 Fellowship – Interdisciplinary training in skin biology and cancer Recipient: T32 Fellowship - Endocrinology
2017 - 2018	Sandeep Khatri Fibulin-4a in zebrafish development
2018	Michelle Zorrilla Contribution of elastin to cardiovascular development in zebrafish

SERVICE

Service to Internal Organizations

Department Committees

2012 - Present	Member, Curriculum Committee
2015	Chair, Faculty Search Committee
2018 - 2019	Member, Genetic Counseling Program Curriculum Evaluation Committee
2018-2019	Member, Faculty Search Committee, Department of Human

	Genetics
2018- Present	Member, Strategic Planning Committee
2018- Present	Member, Laboratory Committee
2019	Member, Faculty Search Committee, Environmental and Occupational Health Department
2020	Member, Faculty Search Committee, Department of Human Genetics
2020-present	Member, Faculty Search Committee, Department of Epidemiology

School/University Committees

2003-2004	Member, Admissions Committee, Cell and Molecular Biology Graduate Program, University of Hawaii
2012 - Present	Alternate Member, Educational Policies and Curriculum Committee, Graduate School of Public Health
2014 - Present	University of Pittsburgh MSTP Program, Human Genetics Program Director
2016 - 2019	Member, Genomics Research Core Advisory Committee, University of Pittsburgh
2019 - Present	Member, Faculty Appointment, Promotion and Tenure Committee, Graduate School of public Health
2019	Member, Fulbright Interview Committee, University of Pittsburgh Honors College

Service to External Organizations

2012 - 2013	Reviewer, Faculty Promotions Dossier, Cleveland Clinic
2013 - 2013	Reviewer, Faculty Promotions Dossier, Washington University School of Medicine
2016 – Present	Medical/Scientific Advisory Board Member, A Twist of Fate-ATS, a patient support group for arterial tortuosity syndrome
2019	Member, Faculty Promotion Assessment Committee, University of Copenhagen, Denmark

Other Service

2001 - 2003	Keynote Speaker, Human Genome Project High School Education Mentor Hawaii Science Teachers Association Workshop 2003 “The Human Genome Project - Past, Present and Future”
2002 - 2003	High School Student Mentor NIDDK Drew Research Training Program: Kiani Arkus, Kamehameha High School, 2002 Natasha Peay, Campbell High School, 2003
July 31- August 5 2005	Session Co-Chair, Heritable Elastic Fiber Diseases, Gordon Conference on Elastin and Elastic Fibers, 2005, Meriden, NH

July 9-12 2006	Session Co-Chair, Genetic Disorders and Genomics, Member, Scientific Committee, 4th European Meeting on Elastin, Lyon, France.
2007 - 2008	Speaker, Gifted Resource Council, St. Louis, MO, Member 2007-2008.
2008	Summer Focus Mentor, Young Scientist Program, Washington University Stephanie Carson, Sumner High School
2010 - Present	Dean's Day Judge, Graduate School of Public Health
2011 - 2013	Organizer, Grants and Papers Discussion Group, Department of Human Genetics
2013	Vice Chair, Session Chair, and Poster Judge, Gordon Conference on Elastin and Elastic Fibers
2015	Chair, Gordon Conference on Elastin and Elastic Fibers & Microfibrils
April 2-3, 2019	Session Moderator, NIH Workshop, Heart and Soul, Brain behavior, and cardiovascular gene dosage effects in 7q11.23 and 22q11.2 CNVs, Bethesda, MD.