Name: Zsolt Urban, Ph.D.

Title: Associate Professor

Address (work): Department of Human Genetics
University of Pittsburgh
Graduate School of Public Health
130 DeSoto Street
Crabtree Hall A308
Pittsburgh, PA 15261
Phone: (412) 648-8269
Fax: (412) 624-3020
Email: urbanz@pitt.edu

Date prepared: June 10, 2015

EDUCATION

M.Sc. (Diploma) in Molecular Biology and Biotechnology, University of Szeged, Szeged, Hungary, 1987-1992.

Exchange student, University of Manchester, School of Biological Sciences, January 1992 - July 1992.

Ph.D student, Institute of Biochemistry, Biological Research Institute, Szeged, Hungary, 1992-1994

Ph.D. student in a joint program with UMDNJ-Robert Wood Johnson Medical School, New Brunswick, NJ, USA and the Department of Pediatrics, Semmelweis University of Medicine, Budapest, Hungary, 1994-1996.

Ph.D. June 5, 1997
Thesis title: Elastin gene mutations in patients with supravalvular aortic stenosis and Williams syndrome.

POST-DOCTORAL TRAINING:

1. Research Fellow, Department of Pediatrics, Semmelweis University of Medicine, Budapest, Hungary, 1996-1997.

2. Junior Researcher, Pacific Biomedical Research Center, University of Hawaii, 1997-2001.

ACADEMIC APPOINTMENTS:

1. Assistant Researcher, Pacific Biomedical Research Center, University of Hawaii, 2001-2004

2. Adjunct Assistant Professor, Department of Anatomy and Reproductive Biology, John A. Burns School of Medicine, University of Hawaii 2001-2004

3. Associate Member, Cancer Research Center of Hawaii 2001-2004

4. Assistant Professor, Department of Anatomy, Biochemistry, Physiology and Reproductive Biology, John A. Burns School of Medicine, University of Hawaii February 2004 – July 2004.

5. Assistant Professor, Department of Pediatrics and Department of Genetics, Washington University School of Medicine, 2004 – 2009.

6. Associate Member, Siteman Cancer Center, Washington University School of Medicine, 2004 – 2009.

7. Assistant Professor, Department of Internal Medicine, Washington University School of Medicine, 2008 – 2009.

8. Associate Professor, Department of Human Genetics, Graduate School of Public Health, University of Pittsburgh, 2010 – to date.

SOCIETY MEMBERSHIP

1. Hungarian Society for Biochemistry 1992- 2001

2. American Society of Human Genetics 1998- to date

3. American Heart Association,
   Council on Basic Cardiovascular Science 1998-2003, 2014- to date
   Council on Cardiovascular Disease in the Young 2003- 2005, 2014- to date
   Council on Arteriosclerosis, Thrombosis and Vascular Biology 2003- 2005.

4. Williams Syndrome Association, Professional Member 1998-2008

5. American Society for Matrix Biology, Charter Member 2000-to date

6. American Society for Investigative Dermatology 2003-2006

PEER REVIEWER - JOURNALS

1. American Journal of Human Genetics, 2000-2009
2. American Journal of Medical Genetics, 2007-2015
3. American Journal of Physiology - Lung Cellular and Molecular Physiology, 2008
4. American Journal of Pathology, 2008-2009
5. American Journal of Respiratory Cell and Molecular Biology, 2013
6. Arteriosclerosis, Thrombosis, and Vascular Biology, 2015
7. Biochemistry, 2003
8. Biochimica et Biophysica Acta, 2008
9. Biology of Reproduction, 2011-2012
10. BMC Pediatrics, 2013
11. Circulation, 2003-2014
12. Circulation: Cardiovascular Genetics 2009
13. Circulation Research 2010
14. Clinical Biochemistry, 2002-2014
15. Clinical Genetics, 2008
16. Disease Models & Mechanisms, 2014-2015
17. European Jounal of Human Genetics, 2013
18. European Journal of Pediatrics, 2002
19. Experimental Dermatology, 2007-2008
20. Expert Reviews in Molecular Medicine, 2006
21. FEBS Journal, 2005
22. Genetics Home Reference (NLM, web), 2012

23. Heart and Vessels, 2006

24. Human Molecular Genetics, 2007-2013

25. Human Mutation, 2006

26. Investigative Ophthalmology & Visual Science, 2009

27. Journal of Biological Chemistry, 2004

28. Journal of Clinical Investigation, 2003

29. Journal of Investigative Dermatology, 2005-2015

30. Journal of Medical Genetics, 2002-2004

31. Journal of Vascular Research, 2004-2005

32. Laboratory Investigation, 2007

33. Matrix Biology, 2002-2015

34. Molecular and Cellular Biology, 2006

35. Neuroscience Letters, 2007-2008

36. Orphanet Journal of Rare Diseases, 2012

37. Pediatric and Developmental Pathology, 2006

38. PLOS One, 2012-2013

39. Proceedings of the National Academy of Sciences USA, 2012

40. Stroke, 2004-2009

EDITORIAL RESPONSIBILITIES

1. Current Genetic Medicine Reports (Springer) , Reproductive and Developmental Genetics, Section Editor, 2011 - to date

ADVISORY AND REVIEW BOARDS

1. Hawaii Community Foundation, Medical Advisory Board, 2003- 2004.

2. American Institute of Biological Sciences, Grant Review Panels, 2003-2010.

3. Italian Telethon Foundation, External Reviewer, 2006, 2015.

4. Association Française contre les Myopathies, External Reviewer, 2006.

5. French National Research Agency (ANR), External Reviewer, 2006-2013.

6. Health Research Board, Ireland, External Reviewer, 2007.

7. National Science Foundation, Reviewer, 2007.

8. Wellcome Trust, UK, Reviewer, 2008

9. Medical Research Council, UK, Reviewer, 2008-2014

10. National Institutes of Health
    Special Emphasis Panel ZRG1 GGG F, 2009
    Special Emphasis Panel ZRG1 MOSS G, 2009
    Special Emphasis Panel ZRG1 SBIB-X, 2011

11. US Army Medical Research and Materiel Command, Reviewer, 2009

12. Fund for Scientific Research Flanders (FWO), Belgium, Reviewer, 2010-2012

13. Deusche Forschungsgemeinshaft (DFG), Germany, Reviewer, 2013

14. University of Pittsburgh, Clinical & Translational Science Institute, Reviewer, 2013-2014

15. Washington University, St. Louis, Nutrition and Obesity Research Center Pilot and Feasibility Studies Program, Reviewer, 2013

16. Qatar National Research Fund, Reviewer, 2014-2015

OTHER SERVICE

1. Faculty Promotion Dossier, External Reviewer, Cleveland Clinic, 2012-2014

2. Faculty Promotion Dossier, External Reviewer, Washington University School of Medicine, 2013

3. Chair, Faculty Search Committee, Department of Human Genetics, University of Pittsburgh, 2015

TEACHING

1. Graduate Program Membership
   - Cell and Molecular Biology (CMB) Graduate Program, University of Hawaii, 2000-2004
   - CMB Graduate Program Admissions Committee, University of Hawaii 2003-2004
   - Faculty Member, Division of Biology and Biomedical Sciences, Quantitative Human and Statistical Genetics Program, Molecular Cell Biology Program, Washington University, 2005-2009
   - Faculty Member, Human Genetics Graduate Program, University of Pittsburgh, 2010-to date
   - Faculty Member, Molecular Genetics and Developmental Biology Graduate Program, University of Pittsburgh, 2011- to date.
   - University of Pittsburgh MSTP Program, Human Genetics Program Director, 2014- to date

2. Courses taught:
   - CMB 621, Molecular Biology of the Cell, University of Hawaii, Lecturer, 1997-2004
   - BIOM 699, Directed Reading, University of Hawaii, 2001- 2004
   - ANAT 695, Directed Research, University of Hawaii, 2001-2002
   - Pediatric Fellows' Core Lecture: „Getting Published", Washington University, 2006-2008
   - BIO 5128, Extracellular Matrix and Cell Matrix Interactions Journal Club, Washington University, Coursemaster, 2007-2009
   - Summer Institute Program to Increase Diversity, Lecture: „Genetic Epidemiology of COPD", Washington University 2008-2009
   - BIO 5258, Fundamentals of Mammalian Genetics, Washington University, Lecturer, 2009
   - INTBP 2290, Scientific Ethics and the Responsible Conduct of Research, University of Pittsburgh, Breakout session moderator, 2010
   - HUGEN 2040, Molecular Basis of Human Inherited Disease, University of Pittsburgh, Co-Instructor, 2010- to date

- HUGEN 2051, Inborn Errors of Development, University of Pittsburgh, Primary Instructor, 2011- to date
- HUGEN 2036, Genetic Counseling Internship, University of Pittsburgh, Guest Lecturer, 2011- 2014
- HUGEN 2025, Human Genetics Noon Seminar, Co-Organizer, 2011- to date
- Human Genetics Summer Research in Progress Seminar, Organizer, 2012- to date
- MSCMP 3735, Extracellular Matrix in Tissue Biology and Bioengineering, Guest Lecturer, 2013-to date
- MS-1, Cellular and Pathologic Basis of Disease, Guest Lecturer, 2014-to date

3. Postdoctoral trainees:

Gabriella Skuta, MD, PhD	2001-2002
Feng Hao, MD, PhD	2003-2004
Zoltan Szabo, PhD	2003-2005
Vishwanathan Hucthagowder, PhD	2003-2009
Andrew Maxfield, PhD	2006-2008
Jyothirmai Talasila, DDS	2007-2009
Laura Miller-Smith, MD	2008-2009
Chi-Ting Su, MD, MPH, PhD	2014-to date

4. Ph.D. students supervised:

Claudia Lupp	1997
Gregg Maeda	1998-2000
Peter Nyerki	2001-2002
Qirui Hu	2002-2007
Bert Callewaert, MD	2008
Andy Willaert	2009
Sandeeep Khatri	2010-to date
Gulab Sher	2011-2012
Chi-Ting Su, MD, MPH	2011-2014
Sevinc Alkan	2012-to date
Michelle Zorrilla	2013-to date

5. Masters students supervised:

Thomas L. Seidl	2000-2002
Ulrich Seitz	2003
Susanne Bohn	2004-2005
Rachel Westman	2010-2011
Meghan MacNeal	2010-2012
Shazina Saeed	2010-2011
Chi-Ting Su, MD	2011-2012
Christine Weckenmann	2012
Christa Lorenchick	2012-2014
Kathrin Hammon	2013
Martin Requena	2015-to date

6. Rotation students supervised:

Sandra Dunn	2003
Jeffrey Squires	2003

7. Pre-medical graduate students:	Alok N. Sharma	1999-2000
	Jodi Katahira	2000- 2002
8. Undergraduate students:	Kerstin Wagner	2001-2002
	Silke Wolterink	2002-2003
	Melanie Roth	2003-2004
	Michael Angstmann	2005
	Stefanie Lerche	2005
	Nina Sausgruber	2005-2006
	Aleksander Keselman	2005-2006
	Carmen Fischer	2006
	Lena Schreiber	2006-2007
	Lenny Jonggadipo	2007
	Annika Aldinger	2007-2008
	Carla Sens	2008
	Nura Schürmann	2008-2009
	Mark Robertshaw	2009-2010
	Lena Collenburg	2009-2010
	Silvia Henger	2010
	Christine Pfeiffer	2010-2011
	Franziska Zuber	2011
	Maureen McGowan	2011-2012
	Bianca Schulz	2011-2012
	Amelie Bauer	2012-2013
	Kerstin Ehm	2013-2014
	Sheila Longo	2013-2014
	Daniel Thomas	2014

9. Examination committee membership:
   - Qirui Hu, Cell and Molecular Biology Program, University of Hawaii, Ph.D. 2002-2004
   - Chao-Tsung Yang, Developmental Biology Program, Washington University, Ph.D. 2006
   - Justin Weinberg, Molecular Cell Biology Program, Washington University, Ph.D. 2006-2007
   - John Gansner, Molecular Cell Biology Program, Washington University, Ph.D. 2006-2008
   - Sandeep Khatri, Genetics Program, University of Pittsburgh, Ph.D. 2010- to date, 'Fibulin-4 in cardiovascular development'
   - Rachel Westman, Human Genetics, Genetic Counseling Program, University of Pittsburgh, M.S.G.C. 2010-2011
   - Madhav Sankunny, Human Genetics Program, University of Pittsburgh, Ph.D. 2011-2013 'The Role of the ATR-CHEK1 Pathway in Therapeutic Resistance Resulting from Distal 11q Loss in Carcinoma Cells'
   - Heejung Kang, Human Genetics Program, University of Pittsburgh, Ph.D. 2011-2014. 'Medium Chain Acyl-CoA Dehydrogenase Deficiency'

- Kaitlyn Kormanik, Human Genetics Program, University of Pittsburgh, Ph.D. 2011-2014. 'Characterization of the Biochemical and Physiological Functions of Acyl-CoA Dehydrogenase 10'
- Prya Mittal (Qualifying, Comprehensive, Dissertation), Human Genetics Program, University of Pittsburgh, Ph.D. 2012-to date
- Brianna Heath (Qualifying, Comprehensive, Dissertation), Human Genetics Program, University of Pittsburgh, Ph.D. 2012-to date. 'Neuroprotective effects of melatonin in Huntington's disease'
- Sevinc Alkan (Qualifying, Comprehensive, Dissertation), Human Genetics Program, University of Pittsburgh, Ph.D. 2012-to date, 'Transforming growth factor beta signaling in ELN-related cutis laxa'
- Meghan Mac Neal, Human Genetics, Genetic Counseling Program, University of Pittsburgh, M.S.G.C. 2012
- Chi Ting Su (Qualifying, Comprehensive, Dissertation), Human Genetics Program, University of Pittsburgh, Ph.D. 2012-2014, 'Molecular mechanisms of LTBP4-related cutis laxa'. Winner of 'Best Human Genetics PhD Dissertation' award.
- Megan Beck (Qualifying, Comprehensive, Dissertation) Human Genetics Program, University of Pittsburgh, Ph.D. 2012-to date
- Lora McClain (Qualifying, Comprehensive, Dissertation) Human Genetics Program, University of Pittsburgh, Ph.D. 2013-to date, 'HSV-1 infection in human iPSC-derived neurons: cellular models of quiescence and drug discovery'.
- Hatem O. Kaseb (Qualifying, Comprehensive, Dissertation) Human Genetics Program, University of Pittsburgh, Ph.D. 2013-to date, 'Spheroid-enriched cancer stem-like cells as a model for targeted therapy in oral cancer with distal 11q loss'.
- Anatalia Labilloy (Dissertation) Human Genetics Program, University of Pittsburgh, Ph.D. 2013-2015, 'Signaling pathways in cell models of Fabry disease nephropathy'
- Michelle Zorrilla (Qualifying) Human Genetics Program, University of Pittsburgh, Ph.D. 2013-to date
- Ian Casci (Qualifying) Human Genetics Program, University of Pittsburgh, Ph.D. 2014-to date
- Megan Breski (Comprehensive, Thesis) Human Genetics Program, University of Pittsburgh, M.S. 2015, 'Allele specific approach to study histone demethylation using engineered KDM4A- (2) ketoglutarate pairs'
- Stephen McCalley (Qualifying, Dissertation) Human Genetics Program, University of Pittsburgh, Ph.D. 2015-to date
- Brandon Blobner (Qualifying, Dissertation) Human Genetics Program, University of Pittsburgh, Ph.D. 2015-to date

10. MPH essay reader
    - Shazina Saeed, Human Genetics Program, University of Pittsburgh, First Reader 2011
    - Chi Ting Su, Human Genetics Program, University of Pittsburgh, First Reader 2012

- Meghan Mac Neal, Human Genetics Program, University of Pittsburgh, First Reader 2012

11. External thesis reviewer/examiner:
- Caroline E. Ridley, External Examiner, Faculty of Medical and Human Sciences, University of Manchester, UK, Ph.D. 2009
- Laetitia Sabatier, External Examiner, Department of Anatomy and Cell Biology, McGill University, Montreal, Canada, Ph.D. 2011
- Sulman Basit, External Examiner, Department of Biochemistry, Quaid-i-Azam University, Islamabad, Pakistan, Ph.D. 2012 'Genetic mapping of genes involved in human hereditary alopecias and skeletal deformities'.
- Andreja Simpson, External Examiner, Faculty of Medical and Human Sciences, University of Manchester, UK, Ph.D. 2013 'Fibulin-4 mutations in cutis laxa'.
- Sofia Hussain, External Examiner, Department of Biochemistry, Quaid-i-Azam University, Islamabad, Pakistan, Ph.D. 2014 'Molecular characterization of inherited kidney diseases in the Pakistani population'.

12. Other committee membership:
- Department of Human Genetics, Member, Curriculum Committee, 2012-to date
- Graducate School of Public Health, Alternate Member, Educational Policies and Curriculum Committee, 2012-to date

GRANT SUPPORT

Completed

1. 1998-2000 American Heart Association, Hawaii Affiliate, Beginning Grant-in Aid (HIGB-13-98), PI, 'The role of elastin gene mutations and polymorphisms in the pathogenesis of supravalvular aortic stenosis', $28,820 for the first year, total award: $58,780.

2. 1999-2002 NIH, NIAMS R03 (AR46379), PI, 'Elastin gene mutations in skin and vascular diseases.', $49,850 direct costs for the first year, total direct costs: $149,550.

3. 2001-2002 Connective Tissue Imagineering LLC, PI, 'Hydrophilic elastin peptides: cellular responses' $126,000 direct costs.

4. 2001-2003 American Heart Association, National Grant-in-Aid (0150587N), PI, 'Genetic dissection of elastin and elastin receptor interactions in obstructive vascular disease', $71,500 for the first year, total award: $214,500.

5. 2001-2004, NIH, NCRR, P20 (RR16453)'A COBRE center for cardiovascular research' Project 5, PI, 'Elastin and elastin receptor in vascular diseases', $135,829 direct costs for the first year, total direct costs: $450,000.

6. 2002-2006 (2006-2007 no cost extension), NIH, NHLBI R01 (RFA) (HL73703), PI, 'Elastin gene mutations: mechanisms causing SVAS and ADCL' $150,000 direct costs for the first year, total direct costs: $650,000.

7. 2007-2008, Washington University, Center for Aging Pilot Research Grant, PI, 'Fibulin-2: a novel determinant of the longevity of elastic fibers', total direct costs $30,000.

8. 2006-2009. American Heart Association, Heartland Grant-in-Aid (0655626Z), 'Williams syndrome heart study' $65,000 direct costs for the first year, total direct costs $130,000.

9. 2006-2009, NIH, NHLBI SCCOR (P50 HL084922), PI: Holtzman, M., 'Alveolar and airway mechanisms for COPD' Project 2, Project leader: Mecham, M., Co-investigator: Urban, Z., 'Genetic determinants: elastin quality and quantity' funds for Z. U. $100,000 direct costs for the first year, total direct costs to Z.U. $300,000.

10. 2010, NIH, NHLBI SCCOR (P50 HL084922), PI: Holtzman, M., 'Alveolar and airway mechanisms for COPD' Project 2, Project leader: Mecham, M., Co-investigator, Subcontract with Washington University, PI, total costs : 50,000.

11. 2009-2012, March of Dimes, Research Grant (#1-FY09-556), PI, 'Fibulin-4 in cardiovascular and connective tissue development' $77,382 for the first year, total direct costs: $235,764.

12. 2011-2012, University of Pittsburgh, Clinical and Translational Science Institute Basic to Clinical Collaborative Research Pilot Program, PI, 'Genetics of emphysema: extracellular and growth factor signaling', total direct costs: $25,000

13. 2013 (NIH) (R13 HL120348), PI, '2013 Elastin, Elastic Fibers & Microfibrils Gordon Research Conference and Gordon Research Seminar' $10,000 direct costs.

14. 2013-2014, NIH (R01 HL090648, supplement), PI, 'Genetics of extracellular matrix in health and disease' $40,750 direct costs for the first year, total direct costs: $40,750.

15. 2010-2015, NIH (R01 HL090648), PI, 'Genetics of extracellular matrix in health and disease' $250,000 direct costs for the first year, total direct costs: $1,000,000.

Active

16. 2014-2015, NIH (R01 HL090648, supplement), PI, 'Genetics of extracellular matrix in health and disease' $38,950 direct costs for the first year, total direct costs: $38,950.

17. 2015-2019, NIH (R01 HL090648), PI, 'Genetics of extracellular matrix in health and disease' $395,333 direct costs for the first year, total direct costs: $1,558,920.

Pending

19. 2015-2017, NIH (R01 HL090648, supplement), PI, 'Genetics of extracellular matrix in health and disease' $32,130 direct costs for the first year, total direct costs: $74,971.

PATENTS

1. Methods and composition for diagnosing and treating pseudoxanthoma elasticum and realted conditions. Inventors: Boyd CD, Csiszar K, Le Saux O, **Urban Z,** Terry S. US Patent 6,780,587, South Africa Patent 2002/7641, European Patents 1258649, 1259649.

PEER REVIEWED RESEARCH PUBLICATIONS

Metrics (as of 1/20/2015):	Total citations:	2050
	Average citations:	44
	Median citations:	29
	G factor:	45
	H factor	27

1. Szabo G, Katarova Z, Körtvély E, Greenspan R, **Urban Z** (1996) Structure and the promoter region of the mouse gene encoding the 67-kD form of glutamic acid decarboxylase. **DNA Cell Biol** 15:1081-1091.

2. Körtvély E, **Urban Z**, Katarova Z, Szabo G (1997) Transcriptional regulation of the mouse gene encoding the 67 KDa form of glutamic acid decarboxylase. **Neurobiology** 5:172

3. Olson TM, Michels VV, **Urban Z**, Csiszar K, Christiano A, Driscoll DJ, Feldt RH, Boyd CD, Thibodeau SN (1995) A 30 kb deletion within the elastin gene results in familial supravalvular aortic stenosis. **Hum Mol Genet** 4:1677-1679

4. **Urban Z**, Helms C, Fekete G, Csiszar K, Bonnet D, Munnich A, Donis-Keller H, Boyd CD (1996) 7q11.23 deletions in Williams syndrome arise as a consequence of unequal meiotic crossover. **Am J Hum Genet** 59:958-962. PMCID: PMC1914803

5. **Urban Z**, Csiszar K, Fekete G, Boyd CD (1997) A tetranucleotide repeat polymorphism within the human elastin gene (ELNi1). **Clin Genet** 51:133-134.

6. **Urban Z**, Kiss E, Kadar K, Szabolcs J, Csiszar K, Boyd CD, Fekete G (1997) A Williams-szindróma genetikai diagnózisa. (Genetic diagnosis of Williams syndrome) **Orv Hetil** 138: 1749-1752.

7. delRio T, **Urban Z**, Csiszar K, Boyd CD (1998) A gene–dosage PCR method for the detection of elastin gene deletions in patients with Williams syndrome. **Clin Genet** 54: 129-135.

8. Lemack GE, Szabo Z, **Urban Z**, Boyd CD, Csiszar K, Vaughan ED Jr, Felsen D (1999) Altered bladder function in transgenic mice expressing rat elastin. **Neurourol Urodyn** 18: 55-68

9. **Urban Z**, Michels VV, Thibodeau SN, Donis-Keller H, Csiszar K, Boyd CD (1999) Supravalvular aortic stenosis: a splice site mutation within the elastin gene results in reduced expression of two aberrantly spliced transcripts. **Hum Genet** 104:135-142.

10. Le Saux O, **Urban Z**, Göring HHH, Csiszar K, Pope FM, Richards A, Pasquali-Ronchetti, Terry S, Bercovitch L, Lebwohl MG, Breuning M, van der Berg P, Kornet L, Doggett N, Ott J, de Jong PTVM, Bergen AAB, Boyd CD (1999) Pseudoxanthoma elsticum maps to an 820 kb region of the p13.1 region of chromosome 16. **Genomics** 62:1-10.

11. **Urban Z**, Peyrol S, Zabot M-T, Plauchu H, Lebwohl M, Schilling K, Green M, Boyd CD, Csiszar K, (2000) Elastin gene deletions in Williams syndrome patients result in altered deposition of elastic fibers in skin and a subclinical dermal phenotype. **Pediatr Dermatol** 17:12-20.

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 (2000) Mutations in a gene encoding an ABC transporter (ABCC6) cause pseudoxanthoma elasticum. **Nat Genet** 25:223-226.

13. **Urban Z**, Michels VV, Thibodeau SN, Davis EC, Bonnefont JP, Munnich A, Eyskens, Gewillig M, Devriendt K, Boyd CD (2000) Isolated supravalvular aortic stenosis: haploinsuffciency of the elastin gene due to nonsense mediated decay. **Hum Genet** 106:577-588.

14. Stollberg J, Urschitz J, **Urban Z,** Boyd CD (2000) A quantitative assessment of SAGE. **Genome Res** 10: 1241-1248. PMCID: PMC310928

15. Dedic J, Weiss AS, Katahira J, Yu B, **Urban Z**. (2001) A novel elastin gene mutation in a family with supravalvular aortic stenosis: a mutation cluster within exon 20. **Hum Mutation** 17: 81.

16. **Urban Z**, Zhang Y, Davis EC, Maeda G, Kumar A, Stalker H, Belmont J, Boyd CD, Wallace MR. (2001) Supravalvular aortic stenosis: genetic and molecular dissection of a complex mutation in the elastin gene **Hum Genet** 109: 512-520.

17. Sadler LS, Pober BR, Grandinetti A, Scheiber D, Fekete G, Sharma AN, **Urban Z.** (2001) Differences by sex in cardiovascular disease in Williams syndrome. **J Pediatr** 139:849-853.

18. Ilias A, **Urban Z**, Seidl TL, Le Saux O, Sinko E, Boyd CD, Sarkadi B, Varadi A. (2002) Loss of ATP-dependent transport activity in pseudoxanthoma associated mutants of human ABCC6 (MRP6) **J Biol Chem** 277: 16860-16867.

19. **Urban Z**, Riazi S, Seidl TL, Katahira J, Smoot LB, Chitayat D, Boyd CD, Hinek A. (2002) Connection between Elastin Haploinsufficiency and Increased Cell Proliferation in Patients with Supravalvular Aortic Stenosis and Williams-Beuren Syndrome. **Am J Hum Genet** 71:30-44. PMCID: PMC384991

20. Urschitz J, Iobst S, **Urban Z**, Granda C, Souza KM, Lupp C, Schilling K, Scott I, Csiszar K, Boyd CD (2002) A serial analysis of gene expression in sun-damaged human skin. **J Invest Dermatol** 119:3-13.

21. McBratney BM, Margaryan E, Ma W, **Urban Z,** Lozanoff S (2003) Frontonasal dysplasia in 3H1 Br/Br mice. **Anat Rec** 271A: 291-302.

22. Ruigrok Y, Seitz U, Wolterink S, Wijmenga C, Rinkel G, **Urban Z** (2004) Association of polymorphisms and haplotypes in the elastin gene in Dutch patients with sporadic aneurysmal subarachnoid hemorrhage. **Stroke** 35:2064-2068.

23. **Urban Z,** Gao J, Pope FM, Davis EC (2005) Autosomal dominant cutis laxa with severe lung disease: synthesis and matrix deposition of mutant tropoelastin. **J Invest Dermatol** 124:1193-1199.

24. Marler JA, Elfenbein JL, Ryals BM, **Urban Z,** Netzloff ML (2005) Sensorineural hearing loss in children and young adults with Williams syndrome **Am J Med Genet** 138:318-327.

25. Hu Q, Reymond JL. Pinel N, Zabot MT, **Urban Z** (2006) Inflammatory destruction of elastic fibers in acquired cutis laxa is associated with missense alleles in the elastin and fibulin-5 genes. **J Invest Dermatol** 126:283-290.

26. Szabo Z, Crepeau MW, Mitchell AL, Stephan MJ, Puntel RA, Loke KY, Kirk RC, **Urban Z** (2006) Aortic aneurysmal disease and cutis laxa caused by defects in the elastin gene. **J Med Genet** 43:255-258. PMCID: PMC2563239

27. Hucthagowder V, Sausgruber N, Kim KH, Angle B, Marmorstein LY, **Urban Z** (2006) Fibulin-4: A novel gene for an autosomal recessive cutis laxa syndrome. **Am J Hum Genet** 78:1075-1080. PMCID: PMC1474103

28. Scheiber D, Fekete G, **Urban Z**, Tarjan I, Balaton G, Kosa L, Nagy K, Vajo Z (2006) Echocardiographic findings in patients with Williams-Beuren syndrome. **Wien Klin Wochenschr** 118:17-18.

29. Hu Q, Loeys BL, Coucke PJ, De Paepe A, Mecham RP, Choi J, Davis EC, **Urban Z** (2006) Fibulin-5 mutations: mechanisms of impaired elastic fiber formation in recessive cutis laxa. **Hum Mol Genet** 15:3379-3386.

30. 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 (2007) Domains 16 and 17 of tropoelastin in elastic fiber formation. **Biochem J** 402:63-70. PMCID: PMC1783983

31. **Urban Z**, Agapova O, Hucthagowder V, Yang P, Starcher BC, Hernandez MR (2007) Population differences in elastin maturation in optic nerve head tissue and astrocytes. **Invest Ophthalmol Vis Sci** 48:3209-3215.

32. Sato F, Wachi H, Ishida M, Nonaka R, Onoue S, **Urban Z**, Starcher BC, Seyama Y (2007) Distinct steps of cross-linking, self-association, and maturation of tropoelastin are necessary for elastic fiber formation. **J Mol Biol** 369:841-851.

33. Watts CR, Marler JA, **Urban Z** (2007) The effects of supravalvular aortic stenosis mutation on voice production. **J Med Speech Lang Pathol** 15:395-406.

34. 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, J van Reeuwijk, Brunner HG, Wevers RA. (2008) Defining the phenotype of an autosomal recessive cutis laxa syndrome with a combined defect of glycosylation. **Eur J Hum Genet** 16:28-35.

35. Kornak U, Reynders E, Dimopoulou A, van Reeuwijk J, Fischer B, Rajab A, Budde B, Nuernberg P, Foulquier F, the ARCL Debré-type study group, Lefeber D, **Urban Z,** Gruenewald S, Annaert W, van Bokhoven H, Wevers R, Morava E, Matthijs G, Van Maldergem L, Mundlos S. (2008) Mutations in the a2 subunit of the v-type H^+-ATPase Impair Glycosylation in the Golgi-Apparatus and Cause Autosomal Recessive Cutis Laxa Type II**. Nat Genet** 40:32-34.

36. Wachi H, Nonaka R, Sato F, Shibata-Sato K, Ishida M, Iketani S, Maeda I, Okamoto K, **Urban Z**, Onoue S, Seyama Y (2008) Characterization of the molecular interaction between tropoelastin and DANCE/fibulin-5. **J Biochem** 143:633-639.

37. Noordam C, Funke S, Knoers NV, Jira P, Wevers RA, **Urban Z**, Morava E. (2009) Decreased bone density and treatment with patients with autosomal recessive cutis laxa. **Acta Paediatr** 98:490-494.

38. Nonaka R, Onoue S, Wachi H, Sato F, **Urban Z**, Starcher BC, Seyama Y. (2009) DANCE/Fibulin-5 promotes elastic fiber formation in a tropoelastin isoform-dependent manner. **Clin Biochem** 42:713-721.

39. Hucthagowder V, Morava E, Kornak U, Lefeber DJ, Dimopoulu A, Fischer B, Dimopoulu A, Aldinger A, Choi J, Davis EC, Abuelo DN, Adamowicz M, Al-Aama

Jumana, 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.** (2009) Loss-of-function mutations in *ATP6V0A2* impair vesicular trafficking, tropoelastin secretion, and cell survival. **Hum Mol Genet** 18:2149-2165. PMCID: PMC2685755

40. **Urban Z**, Hucthagowder V, Schürmann N, Todorovic V, Zilberberg L, Choi J, Sens C, Brown CW, Clark RD, Holand KE, Marble M, Sakai LY, Dabovic B, Rifkin DB, Davis EC. (2009) Mutations in LTBP4 cause a syndrome of impaired pulmonary, gastrointestinal, genitourinary, musculoskeletal and dermal development. **Am J Hum Genet** 85:593-605. PMCID: PMC2775835

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. (2010) Jagged1 (JAG1) mutations in patients with tetralogy of Fallot or pulmonic stenosis. **Hum Mutat** 31:594-601. PMCID: PMC2914103

42. Hu Q, Shifren A, Choi J, Szabo Z, Starcher BC, Knutsen RH, Shipley JM, Davis EC, Mecham RP, **Urban Z**. (2010) Mechanisms of emphysema in autosomal dominant cutis laxa. **Matrix Biol** 29:621-628. PMCID: PMC3606561

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**. (2011) New insights into the pathogenesis of autosomal dominant cutis laxa with report of five *ELN* mutations. **Hum Mutat** 32:445-455. PMCID: PMC3383654

44. Brunetti-Pierri N, Piccolo P, Morava E, Wevers RA, McGuirk M, Johnson YR, **Urban Z**, Dishop MK, Potocki L. (2011) Cutis laxa and fatal pulmonary hypertension: a newly recognized syndrome? **Clin Dysmorphol** 20:77-81.

45.Willaert A, Khatri SM, Callewaert BL, Coucke PJ, Crosby SD, Lee JG, Davis EC, Shiva S, Tsang M, De Paepe A, **Urban Z**. (2012) GLUT10 is required for the development of the cardiovascular system and the notochord and connects mitochondrial function to TGFβ signaling. **Hum Mol Genet** 21:1248-1259. PMCID: PMC3284116

46. Sugitani H, Hirano E, Knutsen RH, Shifren A, Wagenseil JE, Ciliberto C, Kozel B, **Urban Z**, Davis EC, Mecham RP. (2012) A humanized mouse model of autosomal dominant cutis laxa: insight into disease pathogenesis. **J Biol Chem** 287:22055-22067. PMCID: PMC3381164

47. Callewaert B, Su CT, Van Damme T, Vlummens P, Malfait F, Vanakker O, Schultz B, Mac Neal M, Davis EC, Lee JGH, Salhi A, Unger S, Heimdal K, De Almeida S, Kornak U, Gaspar 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. (2013) Comprehensive clinical and molecular analysis of 12 families with type 1 recessive cutis laxa. **Human Mutation** 34:111-121. PMCID:PMC4105850

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

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** doi: 10.1093/hmg/ddv139.

INVITED PAPERS, REVIEWS

1. **Urban Z,** Boyd CD (2000) Elastic-fiber pathologies: primary defects in assembly – and secondary disorders in transport and delivery. **Am J Hum Genet** 67:4-7. PMCID: PMC1287100

2. Milewicz DM, **Urban Z,** Boyd CD (2000) Genetic disorders of the elastic fiber system. **Matrix Biol** 19: 471-480.

3. Bielinska M, Jay PY, Erlich JM, Mannisto S, **Urban Z**, Heikinheimo M, Wilson DB (2007) Molecular genetics of diaphragmatic defects. **Ann Med** 39:261-274. PMCID: PMC2174621

4. Pober BR, Johnson MC, **Urban Z** (2008) Mechanisms and treatment of cardiovascular disease in Williams-Beuren syndrome. **J Clin Invest** 118:1606-1615. PMCID: PMC2358987

5. Berk DR, Bentley DD, Lind A, Bayliss SJ, **Urban Z.** (2012) Cutis laxa: a review. **J Am Acad Dermatol** 66:842.e1-842.e17.

6. **Urban Z**. (2012) The complexity of elastic fiber biogenesis: the paradigm of cutis laxa. **J Invest Dermatol** 132:E12-14.

7. Uitto J, Li Q, **Urban Z**. (2013) The complexity of elastic fiber biogenesis in the skin – a perspective to the clinical heterogeneity of cutis laxa. **Exp Dermatol** 22:88-92. PMCID: PMC3556375

8. **Urban Z**, Davis EC. (2014) Cutis laxa: Intersection of elastic fiber biogenesis, TGFβ signaling, the secretory pathway and metabolism. **Matrix Biol** 33:16-22.

BOOK CHAPTERS

1. Katahira J, Nyerki P, Crepeau M, Davis EC, Pope FM, Hinek A, **Urban Z.** (2002) Elastin gene mutations in supravalvular aortic stenosis, pulmonary artery stenosis and cutis laxa: an emerging genotype-phenotype correlation. In: Tamburro AM, Pepe A. (eds) **Elastin 2002**. EditricErmes, Potenza, Italy, pp 191-201.

2. Tassabehji M, **Urban Z** (2006) Molecular diagnostics of supravalvular aortic stenosis. In: Kearns-Jonker M (ed) **Congenital heart disease: molecular diagnostics.** Humana, Totowa, NJ pp 129-156.

3. **Urban Z** (2009) Cutis laxa. In: Lang F (ed) **Encyclopedia of Molecular Mechanisms of Disease.** Springer, Heidelberg, pp 477-479.

4. Loeys BL, De Paepe A, **Urban Z.** *EFEMP2*-related cutis laxa. In: Pagon RA, Bird TD, Dolan CR, Stephens K, (eds) **GeneReviews** [Internet]. Seattle (WA): University of Washington, Seattle; 1993-. 2011 May 12.

5. **Urban Z**. (2014) LTBP4 and Urban-Rifkin-Davis syndrome. In: Erickson RP, Wynshaw-Boris A, editors. **Epstein's Inborn Errors of Development, 3rd ed.** Oxford University Press, New York (NY), (in press).

6. Callewaert BL, **Urban Z**. LTBP4-related cutis laxa. In: Pagon RA, Bird TD, Dolan CR, Stephens K, (eds) **GeneReviews** [Internet]. Seattle (WA): University of Washington, Seattle; 1993-. (in press)

CONFERENCE ORGANIZATION

1. Session Co-Chair, Heritable Elastic Fiber Diseases, Gordon Conference on Elastin and Elastic Fibers July 31- August 5, 2005, Meriden, NH

2. Session Co-Chair, Genetic Disorders and Genomics, Member, Scientific Committee, 4th European Meeting on Elastin, July 9-12, 2006, Lyon, France.

3. Session Chair, Elastic Fibers in Skin Development, Diseases, and Aging, Gordon Conference on Elastin and Elastic Fibers, July 29- August 3, 2007, Biddeford, ME.

4. Organizer, 1st American Meeting on Cutis Laxa, June 20-21, 2008, St. Louis, MO.

5. Session Co-Chair, Disorders Associated with Elastic Fiber Genes, 5th European Meeting on Elastin, July 16-19, 2008, Acala de Henares, Spain.

6. Session Chair, Inherited Disorders of Elastic Fibers, Gordon Conference on Elastin and Elastic Fibers, July 26-31, 2009, Biddeford, ME.

7. Session Co-Chair, Inherited and Acquired Elastic Fibre Disorders, The Sixth European Elastin Meeting, June 28-July 2, 2010, Maratea, Italy.

8. Organizer, 2nd American Meeting on Cutis Laxa, April 2, 2011, Pittsburgh, PA

9. Session Chair, Genetic Basis and Treatment of Diseases of Elastic Fibers, Gordon Conference on Elastin and Elastic Fibers, July 24-29, 2011, Biddeford, ME.

10. Session Co-Chair, Microfibrils and elastic fibres: structure, assembly and function, 7th European Elastin Meeting, September 1-4, 2012, Ghent, Belgium.

11. Vice Chair, Session Chair, and Poster Judge, Gordon Conference on Elastin and Elastic Fibers 2013.

12. Chair, Gordon Conference on Elastin and Elastic Fibers 2015.

INVITED PRESENTATIONS

1. Speaker, East Coast Connective Tissue Society Meeting, March 22-23, 1996, New York, New York

2. Speaker, XVth Meeting of the Federation of the European Connective Tissue Societies, Elastin Workshop, August 4-9, 1996, Munich, Germany.

3. Invited Speaker, Annual Meeting of the Molecular Biology Section of the Hungarian Biochemical Society, May 13-16, 1997, Lillafured, Hungary

4. Speaker, International Centennial Meeting on Pseudoxanthoma Elasticum, November 6-7, 1997, Bethesda, Maryland.

5. Speaker, XVIth Meeting of the Federation of the European Connective Tissue Societies, Elastin Workshop, August 1-6, 1998, Uppsala, Sweden.

6. Invited Speaker, PXE International Colorado Regional Patient Support Meeting October 31, 1998, Denver, Colorado. 'Mapping of the PXE gene'

7. Speaker, Gordon Conference on Elastin and Elastic Fibers July 18-23, 1999, Meriden, New Hampshire.

8. Invited Speaker, PXE International California Regional Patient Support Group Meeting October 23, 1999, San Francisco, California. 'Mapping of the PXE gene'

9. Invited Speaker, 8th International Professional Conference on Williams Syndrome, July 21-23, 2000, Dearborn, Michigan.

10. Invited Seminar Speaker, Division of Cardiovascular Research, Hospital for Sick Children, October 18, 2000. 'Inherited disorders of the elastic fibers'

11. Invited Speaker, Gordon Conference on Elastin and Elastic Fibers, Meriden, NH, July 29-August 3, 2001.

12. Speaker, American Heart Association Scientific Sessions 2001, Anaheim, CA, November 11-14, 2001

13. Speaker, Elastin 2002, Acquafredda di Maratea, Italy, July 10-14, 2002

14. Speaker, American Society for Matrix Biology, First Meeting, Houston, Texas, November 6-9, 2002

15. Invited Speaker, Gordon Conference on Elastin and Elastic Fibers, Meriden, NH, August 2-8, 2003.

16. Platform Presenter, American Society of Human Genetics, 53rd Annual Meeting, Los Angeles, CA, November 4-8, 2003.

17. Speaker, American Heart Association, Scientific Sessions, Orlando, Florida, November 9-12, 2003.

18. Invited Seminar Speaker, Washington University School of Medicine, St Louis, MO, November 13, 2003. 'Elastin gene in rare and common diseases'.

19. Platform Presenter, Indian Society of Human Genetics, 29th Annual Meeting, Bangalore, India, January 8-11, 2004

20. Invited Seminar Speaker, National University of Singapore, Singapore, January 13, 2004. 'Pediatric diseases caused by elastin gene defects'

21. Invited Seminar Speaker, Washington University School of Medicine, St Louis, MO, March 10, 2004. 'Locus heterogeneity in cutis laxa'

22. Invited Seminar Speaker, Institute of Enzymology, Hungarian Academy of Sciences, Budapest, Hungary, June 25, 2004. 'Elastin gene defects in vascular and skin diseases'

23. Platform Speaker, Elastin 2004, 3rd European Symposium, Manchester, UK, June 30-July 3, 2004.

24. Invited Seminar Speaker, Center for Medical Genetics, University Medical Center, Ghent, Belgium, July 6, 2004. 'Locus heterogeneity in cutis laxa'

25. Invited Seminar Speaker, Johnson & Johnson Skin Research Center, Skilman, NJ, May 16, 2005. 'Elastin gene defects in vascular and skin diseases'

26. Invited Speaker and Session Co-Chair, Gordon Conference on Elastin and Elastic Fibers, Waterville Valley, NH, July 31- August 5, 2005

27. Invited Speaker and Session Co-Chair, 4th European Meeting on Elastin, July 9-12, 2006, Lyon, France.

28. Invited Speaker, 11th International Professional Conference on Williams Syndrome, Richmond, VA, July 23-24, 2006.

29. Invited Seminar Speaker, Institute of Enzymology, Hungarian Academy of Sciences, Budapest, Hungary, May 31, 2007. 'Fibulin gene mutations in developmental defects'

30. Invited Speaker and Session Chair, Gordon Conference on Elastin and Elastic Fibers, Biddeford, ME, July 29-August 3, 2007.

31. Invited Speaker and Session Co-Chair, 5th European Elastin Meeting, Alcalá De Henares, Spain, July 16-19, 2008.

32. Invited Speaker, American Society for Matrix Biology 2008 Meeting, San Diego, CA, December 7-10, 2008.

33. Invited Seminar Speaker, Department of Human Genetics, Graduate School of Public Health, University of Pittsburgh, May 29, 2009. 'Human genetics of the elastic fiber/TGF-beta network'

34. Invited Speaker and Session Chair, Gordon Conference on Elastin and Elastic Fibers, Biddeford, ME, July 26-31, 2009.

35. Invited Seminar Speaker, Wellcome Trust Centre for Cell Matrix Research, Manchester, UK, November 2-3, 2009. 'Elastic fibre synthesis and assembly: insights from human genetics'

36. Invited Speaker and Session Chair, The Sixth European Elastin Meeting, Maratea, Italy, June 28-July 2, 2010.

37. Invited Speaker, 8th International Symposium on Marfan Syndrome, Warrenton, VA, September 11-14, 2010

38. Invited Speaker, 33rd Annual Meeting of the Society of Craniofacial Genetics, Washington, DC, November 2, 2010

39. Invited Seminar Speaker, Department of Anatomy, Biochemistry and Physiology, John A. Burns School of Medicine, University of Hawaii, Honolulu, HI, January 10, 2011. 'The molecular basis of elastic fiber assembly and TGFβ signaling: insights from human genetics'

40. Invited Seminar Speaker, Department of Environmental and Occupational Health, Graduate School of Public Health, University of Pittsburgh, Pittsburgh, PA, April 28, 2011. 'An elastic view of genes, environment and disease'

41. Invited Speaker and Session Chair, Genetic Basis and Treatment of Diseases of Elastic Fibers, Gordon Conference on Elastin and Elastic Fibers July 24-29, 2011, Biddeford, ME.

42. Invited Speaker, 4th Days on Cutis Laxa, Cutis Laxa Internationale Meeting, Lyon, France, September 16-17, 2011. 'Animal models of cutis laxa'

43. Invited Seminar Speaker, Institute of Enzymology, Hungarian Academy of Sciences, Budapest, Hungary, September 20, 2011. 'Arterial tortuosity: new intracellular and extracellular pathways of TGFβ regulation'

44. Invited Seminar Speaker, Department of Anatomy and Cell Biology, McGill University, Montreal, Canada, October 12, 2011. 'Human genetic insights into the elastic fiber/TGFβ network'

45. Invited Speaker, Pittsburgh International Lung Conference, Pittsburgh, PA October 28-29, 2011. 'The elastic fiber/TGF-beta network in rare and common forms of COPD'

46. Invited Seminar Speaker, Department of Pediatrics, University of Pittsburgh, Pittsburgh, PA, March 20, 2012. 'Arterial tortuosity: new intracellular pathways of TGFβ regulation'

47. Invited Speaker, 7th European Elastin Meeting, September 1-4, 2012, Ghent, Belgium. 'Transforming growth factor-beta dysregulation in cutis laxa'

48. Invited speaker, Joint Meeting of the Society for Glycobiology & American Society for Matrix Biology San Diego, CA, November 11-14, 2012. 'Gene discovery in cutis laxa: insights into elastic fiber formation and TGF-beta signaling'

49. Invited Seminar Speaker, Wellcome Trust Centre for Cell Matrix Research, Manchester, UK, May 16, 2013. 'Arterial tortuosity: new intracellular and extracellular pathways of TGFβ regulation'

50. Invited Speaker, Gordon Research Conference on Elastin, Elastic Fibers & Microfibrils, Biddeford, ME, July 21-26, 2013. 'Gene discovery in cutis laxa'

MEETINGS AND WORKSHOPS

1. *Society for Neuroscience Annual Meeting, St. Louis, MO, October 28 - November 2, 1990.*

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. Soc Neurosci Abstr 16:1293.

2. *Society for Neuroscience Annual Meeting, New Orleans, LA, November 10-15, 1991.*

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. Soc Neurosci Abstr 17:1513.

3. *East Coast Connective Tissue Society, Fifteenth Annual Meeting, Somerset, New Jersey, U.S.A., March 19-20, 1995.*

Olson TM, Michels VV, **Urban Z**, Csiszar K, Christiano A, Driscoll DJ, Feldt RH, Boyd CD, Thibodeau SN. An intragenic deletion in the elastin gene in a family with supravalvular aortic stenosis (SVAS).

4. *Gordon Conference - Elastin and Elastic Fibers, Meriden, New Hampshire, U.S.A. July 23-28, 1995.*

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.

Olson TM, Michels VV, **Urban Z**, Csiszar K, Christiano A, Driscoll DJ, Feldt RH, Boyd CD, Thibodeau SN. A 30 kb deletion within the elastin gene results in familial supravalvular aortic stenosis.

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.

5. *East Coast Connective Tissue Society Meeting, New York, U.S.A., March 22-23, 1996.*

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.

Urban Z, Csiszar K, Thibodeau SN, Boyd CD. Isolated supravalvular aortic stenosis is characterized by a spectrum of mutations within the elastin gene.

del Rio T, **Urban Z**, Boyd CD. The detection of elastin gene deletions in Williams syndrome patients by gene dosage PCR.

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.

6. *Sixth International Conference on the Molecular Biology and Pathology of Matrix, Philadelphia, Pennsylvania, June 16-19, 1996*.

Urban Z, Csiszar K, Thibodeau SN, Boyd CD. Isolated supravalvular aortic stenosis is characterized by a spectrum of mutations within the elastin gene. Matrix Biol. 15:183 (1996).

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. Matrix Biol. 15:183 (1996).

7. *The Seventh International Professional Williams Syndrome Conference, Valley Forge, Pennsylvania, July 23-25, 1996*.

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.

8. *XVth Meeting of the Federation of the European Connnective Tissue Societies, Munich, Germany, August 4-9, 1996*.

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.

9. *46th Annual Meeting: The American Society of Human Genetics, San Francisco, California, October 29-November 2, 1996*.

Boyd CD., **Urban Z**, Helms C, Peyrol S, Fekete G, Bonnet D, Munnich A, Donis-Keller H, Csiszár K. Williams syndrome is characterized by 1 megabase deletions encompassing the elastin gene. Am J Hum Genet 59:A249, 1435 (1996).

10. *2nd Annual Meeting Hungarian Biochemical Society, Lillafured, Hungary, May 13-16, 1997*.

Urban Z, Csiszar K, Fekete G, Peyrol S, Helms C, Donis-Keller H, Thibodeau S, Munnich A, Boyd CD. Elastin gene mutations in supravalvular aortic stenosis and Williams syndrome.

11. *International Centennial Meeting on Pseudoxanthoma Elasticum, Bethesda, Maryland, November 6-7, 1997.*

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.

12. *Association of European Pediatric Cardiologists (AEPC) Annual Meeting, Dublin, Ireland, June 10-13, 1998.*

Urban Z, Kadar K, Kiss E, Csiszar K, Boyd CD, Bojeldein S, Szabolcs J, Fekete G. Williams syndrome: Cardiac and molecular genetic diagnosis.

13. *Pädiatrische Forschung: 7. Tagung mitteleuropäischer Länder. Vienna, Austria, June 19, 1998.*

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.

14. *XVIth Meeting of the Federation of the European Connective Tissue Societies, Uppsala, Sweden, August 1-6, 1998.*

Urban Z, Michels VV, Thibodeau SN, Zhang J, Wallace MR, Bonnefont JP, Munnich A, Donis-Keller H, Csiszár K, Boyd CD. A spectrum of point mutations within the elastin gene in patients with isolated SVAS.

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.

15. *48th Annual Meeting: The American Society of Human Genetics, Denver, Colorado, October 27-31, 1998.*

Urban Z, Michels VV, Thibodeau SN, Zhang J, Wallace MR, Devriendt K, Bonnefont JP, Munnich A, Donis-Keller H, Csiszár K, Boyd CD. Supravalvular aortic stenosis (SVAS): predominance of truncating point mutations within the elastin gene. Am J Hum Genet 63: A390

16. *Gordon Conference on Elastin and Elastic Fibers, Meriden, New Hampshire July 18-23, 1999.*

Urban Z, Michels VV, Thibodeau SN, Bonnefont JP, Munnich A, Eyskens, Gewillig M, Devriendt K, Boyd CD. Premature termination mutations in the elastin gene (ELN) are responsible for isolated supravalvular aortic stenosis (SVAS).

Le Saux O, **Urban Z**, Göring HHH, Csiszar K, Pope FM, Richards A, Pasquali-Ronchetti, Terry S, Bercovitch L, Lebwohl MG, Breuning M, van der Berg P, Kornet L, Doggett N, Ott J, de Jong PTVM, Bergen AAB, Boyd CD. Pseudoxanthoma elsticum maps to an 820 kb region of the p13.1 region of chromosome 16.

17. *49th Annual Meeting: The American Society of Human Genetics, San Francisco, California, October 19-23, 1999.*

Urban Z, Davis EC, Zhang J, Wallace MR, Michels VV, Thibodeau SN, Eyskens B, Devriendt K, and Boyd CD. Functional haploinsufficiency of the elastin gene in patients with isolated supravalvular aortic stenosis (SVAS). Am J Hum Genet 65: A113

Le Saux O, **Urban Z**, Göring HHH, Csiszar K, Pope FM, Richards A, Pasquali-Ronchetti, Terry S, Bercovitch L, Lebwohl MG, Breuning M, van der Berg P, Kornet L, Doggett N, Ott J, de Jong PTVM, Bergen AAB, Boyd CD. Pseudoxanthoma elsticum maps to an 820 kb region of the p13.1 region of chromosome 16. Am J Hum Genet 65: A475

18. *1st Symosium of the International Society for Matrix Biology, Philadelphia, Pennsylvania, June 14-17, 2000.*

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.

19. *8th International Professional Conference on Williams Syndrome, Dearborn, Michigan, July 21-23, 2000.*

Urban Z, Boyd CD. Molecular genetics of the elastin gene: contributing to the genetic dissection of Williams syndrome.

Urban Z, Sadler LS, Pober BR, Scheiber D, Fekete G, Sharma AN, Grandinetti A. Gender differences in cardiovascular disease in Williams syndrome (WS).

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.

20. *XXI David W. Smith Workshop on Malformation and Morphogenesis, La Jolla, California, August 2-5, 2000.*

Sadler L, **Urban Z**, Pober B, Scheiber D, Fekete G, Sharma AN, Grandinetti A. Gender differences in cardiovascular disease in Williams syndrome.

21. *International Conference on Biology and Pathology of the Extracellular Matrix, St. Louis, Missouri, October 12-15, 2000.*

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.

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.

22. *3rd FEBS Advanced Course, ATP-Binding Casette (ABC) Proteins: From Genetic Disease to Multidrug Resistance, Gosau, Austria, March 3-10, 2001*

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.

23. *Society for Investigative Dermatology, 62nd Annual Meeting, Washington DC, May 9-12, 2001*

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.

24. *1st European symposium: Elastin 2001, Reims, France July 10-14, 2001*

Urban Z, Le Saux O, Beck K, Urschitz J, Boyd CD. The pathogenetics of primary and secondary elastinopathies.

25. *Gordon Conference on Elastin and Elastic Fibers, Meriden, NH, July 29-August 3, 2001.*

Urban Z, Gao J, Pope FM, Davis EC. A tandem duplication within the elastin gene is associated with autosomal dominant cutis laxa.

26. *The American Society of Human Genetics, 51st Annual Meeting, San Diego, CA, October 12-16, 2001*

Urban Z, Gao J, Pope FM, Davis EC. A tandem duplication within the elastin gene is associated with autosomal dominant cutis laxa. Am J Hum Genet 69: A2543

27. *Canadian Cardiovascular Congress, Halifax, Nova Scotia, Canada, October 20-24, 2001*

Riazi S, Chitayat D, **Urban Z**, Hinek A. The role of elastin and elastin receptor subunit in the modulation of cellular proliferation.

28. *American Heart Association Scientific Sessions 2001, Anaheim, CA, November 11-14, 2001*

Urban Z, Riazi S, Katahira J, Seidl T, 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. Circulation 104: II-356.

29. *The Biochemical Society Joint Meeting with The Physiological Society, York, UK, December 17-19, 2001*

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.

30. *American Heart Association Asia Pacific Scientific Forum, Honolulu, HI, April 23-26, 2002*

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.

Wagner K, Seidl TL, Yanagisawa H, **Urban Z.** Mutational and expression studies on fibulin-5 in patients with aortic tortuosity and cutis laxa.

Urban Z, Riazi S, Nyerki P, Skuta G, Hinek A. Inverse relationship between cell proliferation and elastin deposition in dermal and vascular cells.

31. *Elastin 2002, Second European Symposium, Aquafredda di Maratea, Italy, July 10-14, 2002*

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.

Wagner K, Yanagisawa H, **Urban Z.** Mutational and expression sutudies on fibulin-5 in patients with aortic tortuosity and cutis laxa.

Davis EC, Gao J, Pope FM, **Urban Z**. A tandem duplication within the elastin gene is associated with autosomal dominant cutis laxa.

32. *The Western Neurosurgical Society, 48th Annual Meeting, Delte Ocean Pointe Resort, Victoria, BC, Canada, October 12-15, 2002*

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.

33. *American Society for Matrix Biology, First Meeting, Houston, TX, November 6-9, 2002*

Urban Z. Elastin-cell interactions: insights from genetic diseases.

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.

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.

34. *Gordon Conference on Elastin and Elastic Fibers, Meriden, NH, August 3-8, 2003*

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.

Hu Q, Wagner K, Crepeau MW, Reymond J-L, Zabot M-T, The CL Research Consortium, Yanagisawa H, **Urban, Z**. Digenic inheritance of cutis laxa.

35. *American Society of Human Genetics, 53rd Annual Meeting, Los Angeles, CA, November 4-8, 2003*

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. Am J Hum Genet 73: A245

36. *American Heart Association, Scientific Sessions, Orlando, FL, November 9-12, 2003*

Crepeau MW, Szabo Z, Stephan MJ, Mitchell A, **Urban Z**. Familial thoracic aneurysm with cutis laxa caused by a mutation in the elastin gene. Circulation 108: IV-2473

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. Circulation 108: IV-392

37. *Indian Society of Human Genetics, 29th Annual Meeting, Bangalore, India, January 8-11, 2004*

Urban Z, Crepeau MW, Szabo Z, Hu Q, Huchthagowder V, The Cutis Laxa Consortium. Locus heterogeneity in cutis laxa.

Huchthagowder V, Crepeau MW, The Cutis Laxa Consortium, **Urban Z.** DHPLC-based candidate gene scanning for cutis laxa.

38. *29th International Stroke Conference, San Diego, CA, February 5-7, 2004*

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.

39. *Society for Investigative Dermatology, 65th Annual Meeting, Providence, RI, April 28-May 1, 2004*

Hu Q, Crepeau MW, Hucthagowder V, Szabo Z, The Cutis Laxa Consortium, **Urban Z**. Locus heterogeneity in cutis laxa. J Invest Dermatol 122: A485.

Szabo Z, Crepeau MW, Stephan MJ, Puntel RA, Mitchell A, **Urban Z**. Cutis laxa caused by mutation in the tropoelastin gene. J Invest Dermatol 122: A486.

40. *Elastin 2004, 3rd European Symposium, Manchester, UK, June 30-July 3, 2004*

Crepeau MW, Szabo Z, Hu Q, Hucthagowder V, The Cutis Laxa Consortium, **Urban Z.** Elastin and fibulin-5 gene defects in cutis laxa.

41. *American Society of Human Genetics, 54th Annual Meeting, Toronto, Canada, October 26-30, 2004*

Urban Z, Ruigrok YM, Wolterink S, Böhn S, Wijmenga C, Rinkel GJE. Association of polymorphisms and haplotypes in the elastin gene in dutch patients with sporadic aneurysmal subarachnoid hemorrhage.

Hucthagowder V, Coucke P, The Cutis Laxa Consortium, Fong K, Csiszar K, **Urban Z**. Lysyl oxidase genes in cutis laxa.

42. *American Society for Matrix Biology, 2nd Meeting, San Diego, CA, November 10-13, 2004*

Hu Q, Wagner K, Reymond J-L, Zabot M-T, The CL Research Consortium, Yanagisawa H,, **Urban Z**. Digenic inheritance of cutis laxa.

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.

43. *SeattleSNPs PGA Variation Workshop, St. Louis, MO, April 25-26, 2005*

44. *Society for Investigative Dermatology, 66th Annual Meeting, St. Louis, MO, May 4-7, 2005*

Hucthagowder V, CL Consortium, Fong K, Csiszar K, **Urban Z**. Lysyl oxidase-like gene mutation in cutis laxa.

Szabo Z, Mitchell AL, Stephan MJ, Pope FM, Davis EC, **Urban Z**. Elastin gene mutations: a novel mechanism causing cutis laxa.

45. *International Evoked Response Audiometry Study Group Biennial Symposium, Havana, Cuba, June 12-16, 2005*

Marler JA, Elfenbein JL, Ryals B, **Urban Z**, Netzloff ML. Sensorineural hearing loss in children and young adults with Williams syndrome.

46. *Gordon Conference on Elastin and Elastic Fibers, Waterville Valley, NH, July 31-August 5, 2005*

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.

Hu Q, Reymond J-L, Pinel N, Zabot M-T, Coucke PJ, **Urban Z**. Inflammatory destruction of elastic fibers in acquired cutis laxa is associated with mutations in the elastin and fibulin-5 genes.

Marler JA, Elfenbein JL, Ryals BM, Netzloff ML, **Urban Z**. Sensorineural hearing loss in Williams syndrome and supravalvular aortic stenosis: a novel role for elastin in auditory function.

47. *The Annual American Speech-Language-Hearing Association Convention, San Diego, CA, November 18-20, 2005*

Marler JA, Elfenbein JL, Ryals BM, **Urban Z**, Netzloff ML. Auditory Acuity in Williams Syndrome & Supravalvular Aortic Stenosis.

48, *Association for Research in Otolaryngology 27th Mid-Winter Meeting, Baltimore, MD, February 5-9, 2006*

Marler JA, Ryals BM, Kujawa SG, **Urban Z**. DPOAE and ABR measures of auditory function in ELN-heterozygous knockout mice.

49. *38th European Human Genetics Conference, Amsterdam. Netherlands, May 6-9, 2006*

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.

50. *4th European Meeting on Elastin, Lyon, France, July 9-12, 2006*

Hucthagowder V, Sausgruber N, Kim KH, Angle B, Marmorstein LY, **Urban Z**. Fibulin-4 mutation in a novel recessive cutis laxa syndrome.

Wachi H, Sato F, Nonaka R, Szabo Z, **Urban Z**, Maeda I, Okamoto K, Starcher BC, Li DY, Mecham RP and Seyama Y. Involvement of exon 16 and 17 in tropoelastin molecule on the elastic fiber formation.

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.

51. *11th International Professional Conference on Williams Syndrome, Richmond, VA, July 23-24, 2006*

Hucthagowder V, Kaplan P, Pober BR, **Urban Z**. Elastin in Williams syndrome: cardiovascular disease and beyond.

Marler JA, **Urban Z**. Auditory function in Williams syndrome and supravalvular aortic stenosis.

52. *The Annual American Speech-Language-Hearing Association Convention, Miami Beach, FL, November 16-18, 2006*

Marler JA, Roy JL, **Urban Z**. Auditory function in connective tissue disorders.

53. *American Society of Human Genetics 56th Annual Meeting, New Orleans, LA, October 9-13, 2006*

Hucthagowder V, Susgruber 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.

54. *American Society of Matrix Biology Biennial National Meeting, Nashville, TN, November 1-4, 2006*

Maxfield AB, Hucthagowder V, Gitlin JD, Joseph EM, **Urban Z**. Fibulin-4 function in zebrafish.

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.

Hu Q, Coucke PJ, Sommer P, Davis EC, Reinhardt DP, Mecham RP, **Urban Z**. Fibulin-5 mutations in cutis laxa.

55. *Gordon Conference on Elastin and Elastic Fibers, Biddeford, ME, July 29-August 3, 2007*

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.

Maxfield AB, Hucthagowder V, Joseph EM, **Urban Z.** Fibulin-4 function in zebrafish.

56. *Midwest Pediatric Cardiology Society 31st Annual Scientific Meeting, St. Louis, MO, September 20-21, 2007*

Hucthagowder V, Johnson MC, Kaplan P, Singh GK, **Urban Z**. Genetic Analysis of Congenital Heart Disease in Williams Syndrome.

57. *American Thoracic Society International Conference, Toronto, Canada, May 16-21, 2008*

Miller-Smith L, **Urban Z**, Mecham RP, Shifren A. Pulmonary Circulatory Abnormalities in Human Elastinopathies.

58. *5th European Elastin Meeting, Alcalá De Henares, Spain, July 16-19, 2008*

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.

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.

Maxfield AB, Hucthagowder V, Joseph EM, **Urban Z**. Fibulin-4 function in cardiovasular development.

59. *Gordon Conference on Elastin and Elastic Fibers, Biddeford, ME, July 26-31, 2009*

Hucthagowder V, Morava E, Kornak U, Lefeber DJ, Dimopoulu A, Fischer B, 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.

60. *6th European Elastin Meeting, Maratea, Italy, June 28-July 2, 2010*

Urban Z, Hucthagowder V, Henger S, Westman R, Collenburg L, 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. LTBP4 mutations in Urban-Rifkin-Davis syndrome.

Maxfield AB, Khatri SM, Mecham RP, Joseph EM, **Urban Z.** Fibulin-4 is necessary for the development of the notochord and the cardiovascular system.

61. *8th International Symposium on Marfan Syndrome, Warrenton, VA, September 11-14, 2010*

Urban Z, Hucthagowder V, Henger S, Westman R, Collenburg L, 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. Disease phenotypes and mechanisms of LTBP4 mutations.

62. American Society of Matrix Biology, 2010 Biennial Meeting, Charleston, SC, October 24-27, 2010

Maxfield AB, Khatri SM, Mecham RP, Joseph EM, **Urban Z.** Fibulin-4 is necessary for the development of the notochord and the cardiovascular system.

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 TGFβ signaling.

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.

63. *33rd Annual Meeting of the Society of Craniofacial Genetics, Washington, DC, November 2, 2010*

Urban Z, Hucthagowder V, Henger S, Westman R, Collenburg L, 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 of LTBP4 result in defective craniofacial extracellular matrix formation in patients with Urban-Rifkin-Davis syndrome. In GH Sperber. 2011. The society of craniofacial genetics. Abstracts of the 2010 annual meeting. Am J Med Genet Part A 155:280–286.

64. *The American Society of Human Genetics, 60th Annual Meeting, Washington, DC, November 2-6, 2010*

Urban Z, Maxfield AB, Khatri SM, Mecham RP, Joseph EM. Fibulin-4 is required for the development of the notochord and the cardiovascular system.

Westman R, Henger S, Shifren A, Yusen RD, Mecham RP, Sciurba FC, **Urban Z**. Chronic lung disease in patients with cutis laxa.

65. *Gordon Conference on Elastin and Elastic Fibers, Biddeford, ME, July 24-29, 2011*

Willaert A, Khatri S, Callewaert BL, Coucke PJ, Crosby SD, De Paepe A, **Urban Z**. GLUT10 connects cellular metabolism to TGFβ signaling.

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.

66. *12th International Congress of Human Genetics, The American Society of Human Genetics, 61th Annual Meeting, Montreal, Canada, October 11-15, 2011*

Urban Z, Willaert A, Khatri SM, Callewaert BL, Coucke PJ, Crosby SD, Loeys BL, Tsang M, De Paepe A. GLUT10 connects TGFβ signaling to cellular metabolism in cardiovascular development.

67. *Pittsburgh International Lung Conference, Personalized Medicine of Lung Disease, Pittsburgh, PA, October 28-29, 2011*

Levine K, Mac Neal M, Sciurba F, **Urban Z**. Genotype-phenotype correlations for pulmonary manifestations of cutis laxa.

Urban, Z. The elastic fiber/TGF-beta network in rare and common forms of COPD.

68. *2012 MidWinter Meeting of the Association for Research in Otolaryngology, San Diego, CA February 25-29, 2012*

Marler JA, Goodman SS, **Urban Z**. Middle-ear anomalies and cochlear function in elastic-fiber disorders.

69. *European Human Genetics Conference 2012, Nürnberg, Germany, June 23-26, 2012*

Callewaert BL, Su CT, Van Damme T, Vlummens P, Malfait F, Vanakker O, Schulz B, Mac Neal M, Davis EC, Lee JGH, 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.

70. *7th European Elastin Meeting, Ghent, Belgium, September 1-4, 2012*

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.

Su CT, Khatri SM, McGowan M, Lawrence E, **Urban Z**. Transforming growth factor-beta dysregulation in cutis laxa.

71. *The American Society of Human Genetics, 62nd Annual Meeting, San Francisco, CA, November 6-10, 2012*

Lawrence EC, McGowan M, Su CT, Mac Neal M, Levine K, Zaenglein A, Bodzioch M, Kiss A, **Urban Z**. ELN mutations in autosomal dominant cutis laxa.

Callewaert B, Su CT, Van Damme T, Vlummens P, Malfait F, Vanakker O, Schulz B, Mac Neal M, Davis EC, Lee JGH, 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.

Urban Z, Khatri SM, Maxfield AB. Fibulin-4b is required for cardiovascular and musculoskeletal development as an antagonist of transforming growth factor-beta.

72. *Joint Meeting of the Society for Glycobiology & American Society for Matrix Biology, San Diego, CA, November 11-14, 2012*

Urban Z. Gene discovery in cutis laxa: insights into elastic fiber formation and TGF-beta signaling.

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.**

Su CT, Weckenmann C, Lawrence EC, **Urban Z**. LTBP4 regulates both the magnitude and the direction of TGFβ response.

73. *Gordon Research Conference on Elastin, Elastic Fibers & Microfibrils, Biddeford, ME, July 21-26, 2013*

Urban Z. Gene discovery in cutis laxa.

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.

74. National Society of Genetic Counselors 32nd Annual Education Conference, Anaheim, CA, October 9-12, 2013

Lorechick C, Weinberg SM, Levine K, Madan-Khetarpal S, Grubs R, **Urban Z**. Craniofacial phenotype in cutis laxa.

75. Society of Craniofacial Genetics and Developmental Biology, 36th Annual Meeting, Boston, MA, October 22, 2013

Urban Z, Parsons TE, Lorechick C, Levine K, Madan-Khetarpal S, Weinberg SM. Craniofacial phenotypes in cutis laxa.

76. The American Society of Human Genetics, 63rd Annual Meeting, Boston, MA, October 22-26, 2013

Lawrence EC, McGowan M, Levine K, 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.

Urban Z, Lorechick C, Parsons TE, Levine K, Madan-Khetarpal S, Weinberg SM. Craniofacial phenotypes in cutis laxa. *Am J Med Genet A*. 2014; 219:S136-S137

77. American Thoracic Society, 2014 International Conference, San Diego, CA, May 16-21, 2014

Wilson R, Chandra D, **Urban Z**, Bon J, 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.

78. *American Society for Matrix Biology, Biennial Meeting, Cleveland, OH, October 12-15, 2014*

Su CT*, Lawrence EC, Dabovic B, Rifkin DB, **Urban Z**. LTBP4 regulates TGFβ signaling by stabilizing TGFβ receptors. ***Winner of ASMB Travel Award.**

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.

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.

Zorrilla M, **Urban Z**. Structure and function of zebrafish elastins.

Alkan S, **Urban Z**. TGFβ signaling in patients with *ELN*-related cutis laxa.

79. *American Society of Nephrology, Kidney Week 2014, Philadelphia, PA, November 11-16, 2014*

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

80. *American Thoracic Society, 2015 International Conference, Denver, CO, May 15-20, 2015*

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.

COMMUNITY SERVICE, PROFESSIONAL DEVELOPMENT

1. Human Genome Project
   High School Education Mentor 2001-2009
   Keynote Speaker: Hawaii Science Teachers Association Workshop 2003
   „The Human Genome Project – Past, Present and Future"

2. High School Student Mentor NIDDK Drew Research Training Program:
   Science project mentor: Kiani Arkus, Kamehameha High School, 2002
   Natasha Peay, Campbell High School, 2003

3. Gifted Resource Council, St. Louis, MO, Member 2007-2008.

4. Academy of Science, St. Louis
   Speaker, Academy of Science Group, Ladue Middle School 2007

5. Young Scientist Program, Washington University,
   Summer Focus Mentor: Stephanie Carson, Sumner High School, 2008

6. Dean's Day Judge, Univeristy of Pittsburgh, Graduate School of Public Health, 2010-to date

7. Coordinator: 'Grants and Papers' discussion group, Department of Human Genetics University of Pittsburgh, 2011-2013.