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# Annual report, 2004-2005

Jouni Uitto  
*Thomas Jefferson University*

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DEPARTMENT OF DERMATOLOGY AND CUTANEOUS BIOLOGY  
JEFFERSON MEDICAL COLLEGE

STATEMENT OF THE CHAIR

Academic Year 2005

Jouni Uitto, M.D., Ph.D.

August 15, 2005

PREFACE

The year 2005 marks my 19<sup>th</sup> year as the Chair of the Department of Dermatology and Cutaneous Biology at Jefferson Medical College. Looking back almost two decades, I have observed the expansion of the Department and the University, and I have been a witness to the extraordinary growth in academic enterprise. When I assumed the Chair in November 1986, the full-time faculty of the Department consisted of one lone full-time academician, Dr. Herbert A. Luscombe, my predecessor as the Chair for 28 years. In contrast, at the end of the Academic Year 2005, we currently have a full-time faculty of 24, comprising of 8 clinicians and 16 research faculty; the total number of trainees currently stands at 16, including 12 residents and 4 research fellows, and the total personnel of the department is about 78.

Our clinical enterprise continues to flourish noted by the increasing demand for our clinical services. However, the inability to offer equitable financial incentives to academic dermatologists

looms over us and poses as a substantial threat to the solvency and overall financial health of the department.

Like many other departments at Jefferson, Dermatology continues to face the challenge of increasing costs and expenses. Cutbacks in the guise of reduced reimbursements, soaring of malpractice costs, reductions in the institutional support for growth and maintenance of the departmental infrastructure and a shrinking faculty, all pose serious threats to our continued ability to fulfill the Departmental mission in education, clinical care and research. Despite daunting circumstances, our department remains proud of its standing as one of the leading institutions nationwide in providing quality dermatologic care and performing state-of-the-art cutaneous biology research.

I am pleased to present this report on the clinical, teaching, and research activities of the Department of Dermatology and Cutaneous Biology at Jefferson Medical College during the Academic year 2005.

### **CLINICAL ACTIVITIES**

Our clinical enterprise, under the leadership and guidance of Young C. Kauh, MD, Professor and Senior Vice Chair of Departmental Development, and Guy F. Webster, MD, Ph.D., Professor and Clinical Vice Chair, has experienced continued growth. Patient demand for dermatology services continues while we seek to expand our clinical faculty so that we may accommodate this growing population. The remaining clinical faculty, comprised of Dr. Tanya Humphreys, Director of Cutaneous Surgery, Dr. Jason Lee, Director of Dermatopathology, Dr. Patrice Hyde, Chief of Pediatric Dermatology, Dr. Stacy Katchman, Clinical Assistant Professor of Dermatology; Dr.

Franziska Ringpfeil, Assistant Professor of Dermatology; and Dr. Barbara Vail, Instructor of Dermatology, have contributed much to our overall clinical practice success. During this academic year, our clinical faculty has valiantly tried to keep up with the workplace demands of an increasing patient load, despite our inability to recruit faculty to replace the attendings who left our full-time employment in 2004. We want to thank our present faculty for their long-term contributions and commitment to the Department's clinical activities.

Mr. Alan Strigga, the Departmental Business Manager, with the assistance of Ms. Karen Griffin, Director of Practice Operations, has been instrumental in maintaining the high quality of standards of our clinical operations. We thank them and the entire clinical office staff for their superb help and a job well done.

### **Specialty Services**

In addition to general dermatology, a number of specialty services are available to our patients at our Center City offices.

**The Cutaneous Surgery Unit**, headed by Dr. Tanya Humphreys, offers a state-of-the-art array of surgical services, including Mohs micrographic surgery, laser surgery, and a number of cosmetic procedures.

**The Division of Pediatric Dermatology**, is directed by Dr. Patrice Hyde who, in addition to our center city office, practices at A.I. DuPont Hospital for Children. Over the past several years, we have also expanded our pediatric dermatology services to Temple Children's Hospital under the direction of Dr. Franziska Ringpfeil, Assistant Professor of Dermatology.

### **Dermatopathology:**

The dermatopathology service of the Department of Dermatology and Cutaneous Biology is directed by Dr. Jason Lee, Clinical Associate Professor of Dermatology and Cutaneous Biology. Since joining us in 2002, his service has grown and provides an excellent source of teaching material for our residency training program. We have realigned our dermatopathology teaching with complementary services within the Department of Pathology that is responsible for hospital-based diagnostics. We expect our dermatopathology services to grow, and we look forward to a potential recruitment of an additional dermatopathologist in the near future.

### **Molecular Diagnostics**

The clinical services are augmented by Molecular Diagnostics of Heritable Skin Diseases Clinic attended by myself and Dr. Gabriele Richard, Associate Professor of Dermatology and Cutaneous Biology, a dermatologist, genetic counselor, and molecular geneticist, with extensive expertise and experience in clinical and molecular genetic studies of ichthyosis and other disorders of cornification. Dr. Richard's research has elucidated the molecular basis of numerous ichthyoses, palmar plantar keratodermas and erythrokeradermias, and she is a recognized expert in research on connexins disorders. Dr. Richard also serves with me as the co-director of the Molecular Diagnostics Laboratory located in our Department.

In summary, the clinical enterprise of the Department of Dermatology and Cutaneous Biology continues to offer broad and diversified specialty services to our patients, and we look forward to continued expansion of these services. The clinical faculty deserves my congratulations for their outstanding work.

## TEACHING

### Residency Training Program

It is with great pleasure that I announce that Dr. Jason Lee has been appointed as the Residency Training Program Director, effective July 1, 2005. A graduate of our own training program, Jason brings a unique perspective to this post. Our Dermatology Residency Training Program continues to be fully accredited towards the eligibility for certification by the American Board of Dermatology. The program currently has twelve residents in good standing. In June 2005, three of our residents completed their training: The Chief Resident, Dr Christine Ambro entered a Cutaneous Surgery fellowship in Seattle, WA; Dr. Carmen Campanelli joined a private practice in Delaware as has Dr. John Wildemore who has remained in the Philadelphia area. I would also like to extend our thanks to our current chief resident, Dr. Gina Ang for doing an outstanding job.

### Medical Student Teaching

The Department participates in the medical student training by giving lectures in dermatology for the second year students. These lectures have been extremely well received by the students as judged from the evaluation feedback. In addition, the Department offers dermatology electives to the medical students, primarily from Jefferson Medical College, but space permitting, also from other universities. Dr. Barbara Vail has assumed a leadership role by serving as a departmental liaison in coordinating the medical students' educational activities. All the clinical work by the students is under direct supervision by the full-time clinical faculty.

In addition to medical students, our clinics have a number of rotating students, including House Staff members from other Jefferson Departments, as well as from other U.S. medical schools and

from abroad. During this academic year, we have had several international clinical fellows from Korea and Brazil join us for a four to eight week rotation.

## RESEARCH

### Research Faculty

Our research faculty has remained stable and has continued to be a successful in biomedical research. During the past year, Dr. Sonsoles Piera-Velasquez was appointed to the Faculty at the rank of Assistant Professor.

I would like to take this opportunity to discuss the recent cuts in NIH funding which has resulted in several of our research faculty resigning and returning to private sector. Dr. Gabriele Richard, now Adjunct Associate Professor of Dermatology and Cutaneous Biology and Dr. Ellen Pfendner, Assistant Professor of Dermatology and Cutaneous Biology have left our faculty and are now employed at GeneDx situated in Bethesda, MD. NIH funding, which over the years has supported dermatology research here at Jefferson, has remained steady, but there has been no increases as in years during the doubling of NIH budgets in 1999-2003. While the extramural support to our research has remained stable and the Department of Dermatology and Cutaneous Biology has continued to successfully compete for grant support from the National Institutes of Health and other sources, the challenge to hold our ground is ever present. The Department has remained among the top five dermatology departments in the country in NIH ranking, having been no. 1 in 1998. In addition to NIH, Dr. Andrzej Fertala continues to receive grant support from NASA while Drs. Mÿ Mahoney, John Klement and Uli Rodeck have funding from the Department of Defense. Drs. Vitali Alexeev, Reza Ghohestani, Mÿ Mahoney, Fatima Rouan, Gabriele Richard and Franziska Ringpfeil have been recipients of Career Development Awards from a variety of sources, including the National Institutes of Health, Dermatology Foundation, and the American Skin Association. While

these Career Development Awards are reaching their end, the challenge is to obtain steady funding that will guarantee continued support to the research of these young investigators. I would like to express our thanks to Dr. Mon-Li Chu, Professor and Vice Chair of Research, for her contributions in support of further development of our departmental research enterprise.

### **Jefferson Institute of Molecular Medicine**

The Institute continues to serve as an umbrella for broad inter-departmental and inter-disciplinary research at Jefferson with emphasis on matrix biology and pathology, cancer biology and human molecular genetics. The Institute provides core facilities that serve not only the Departmental researchers but also the entire Jefferson research community at large. The current cores include: Tissue culture core (Director: Dr. Ellen Pfendner); Morphology core (Director: Dr. David Birk; shared with the Department of Pathology); Transgenic animal facilities (Director: Dr. John F. Klement); and DNA syntheses and sequencing core (Director: Dr. Mon-Li Chu; shared with the Kimmel Cancer Center). Other functions of the Institute include organization of on-going seminar and science lecture series in coordination with Orthopedic Research and Rheumatology/Medicine as well as departmental research in progress series. I am pleased to announce that Dr. Ulrich Rodeck, Professor of Dermatology and Cutaneous Biology, has accepted the position as the Deputy Director of the Jefferson Institute of Molecular Medicine, primarily in charge of the long-term research planning.

### **DebRA Molecular Diagnostics Laboratory**

This laboratory was established in 1996 as a global referral center for molecular diagnostics of epidermolysis bullosa (EB), a group of heritable blistering disorders. This laboratory, a partnership between the lay organization, the Dystrophic Epidermolysis Bullosa Research Association of



America, and the Department of Dermatology and Cutaneous Biology at Jefferson Medical College, has continued to provide DNA diagnostics to the global network of EB patients. In fact, as of today, the Center has analyzed DNA from over a thousand families with different forms of EB. Dr. Ellen Pfindner, Assistant Professor of Dermatology, has served as the Manager of this increasingly busy laboratory. The laboratory has expanded its services to other heritable skin diseases, including various keratinization disorders; this effort has been spearheaded by Dr. Gabriele Richard, Co-Director of the laboratory. Over the past year, the laboratory has also partnered with PXE International, Inc., a patient advocacy organization supporting research on the causes and treatment of pseudoxanthoma elasticum, a group of disorders characterized by calcification of elastic structures in the skin, eyes, and cardiovascular system. As a result of this partnership, the laboratory has identified disease-causing mutations in a cohort of ~200 families with PXE and has participated in development of a diagnostic kit for these mutations.

### **JEFFERSON CENTER FOR INTERNATIONAL DERMATOLOGY**

Over the past two decades, Jefferson Dermatology has become increasingly recognized as a leader in international dermatology. These activities are concentrated under the umbrella of Jefferson Center for International Dermatology which was established in 1987 to promote the exchange of information and dermatological knowledge within the global network of international dermatology. Within this framework, Jefferson Dermatology has continued to host visiting residents, scholars, and scientists from a variety of countries.

### **Summary**

The Department of Dermatology and Cutaneous Biology at Jefferson Medical College continues to excel in academic enterprise, including clinical activities, research endeavors and teaching

programs. The quality of our research is attested to by our high ranking as one of the leaders in research in cutaneous biology and pathology. Our international relations continue to flourish and bring recognition not only to the Department but also to Jefferson Medical College at large. The Department is fully committed to further development of activities in the areas of research, education and patient care, to the maximum extent allowed by fiscal realities.

On behalf of the entire Department of Dermatology and Cutaneous Biology at Jefferson, I would like to thank the President and the Board of Trustees of Thomas Jefferson University, the Dean of Jefferson Medical College, and the President of Thomas Jefferson University Hospital, for their continued support to the programs of the Department of Dermatology and Cutaneous Biology.

Jouni Uitto, MD, PhD

Professor and Chair

Department of Dermatology and Cutaneous Biology

Director, Jefferson Institute of Molecular Medicine

## **MON-LI CHU, PhD**

*Professor and Vice-Chair of Research*

*Department of Dermatology and Cutaneous Biology*

### **PUBLICATIONS:**

Lampe, A.K., Dunn, D.M., von Niederhausern, A.C., Hamil, C., Aoyagi, A., Laval, S.H., Chu, M.-L., Swoboda, K., Muntoni, F., Bönnemann, C.G., Flanigan, K.M., Bushby, K.M.D.; and Weiss, R.B. (2005) Automated genomic sequence analysis of the three collagen VI genes: Applications to Ullrich congenital muscular dystrophy and Bethlem myopathy. *J. Med. Genet.* 42:108-120.

Bardin, A., Moll, F., Margueron, R., Delfour, C., Chu, M.-L., Maudelonde, T., Cavailles, V., and Pujol, P. (2005) Transcriptional and posttranscriptional regulation of fibulin-1 by estrogens leads to differential induction of messenger ribonucleic Acid variants in ovarian and breast cancer cells. *Endocrinology* 146:760-768.

Lucioli, S., Giusti, B., Mercuri, E., Camacho Vanegas, O., Lucarini, L., Pietroni, V., Urtizberea, A., Yaou, R.B., De Visser, M., Bönnemann, C., Iannaccone, S.T., Merlini, L., Bushby, K., Muntoni, F., Bertini, E., Chu, M.-L., and Pepe, G. (2005) Detection of common and private mutations in the COL6A1 gene of Bethlem myopathy patients. *Neurology*, 64:1931-1937.

Lucarini, L., Giusti, B., Zhang, R.Z., Pan, T.C., Jimenez-Mallebrera, C., Mercuri, E., Muntoni, F., Pepe, G. and Chu, M.-L. (2005) A homozygous COL6A2 intron mutation causes in-frame triple-helical deletion and nonsense-mediated mRNA decay in a patient with Ullrich congenital muscular dystrophy. *Hum Genet*, 117:460-466.

Tsuda, T. Chu, M.-L. Biological role of fibulin-2 in cardiovascular development. (2005) In Artman, M, Woodrow Benson, D, Srivastava, D and Nakazawa M (eds) "Cardiovascular Development and Congenital Malformations: Molecular & Genetic Mechanisms." Blackwell Futura. pp. 20-23.

Pepe, G., Lucarini, L., Zhang, R.-Z., Pan, T.-C., Giusti, B., Quijano-Roy, S., Gartioux, C., Bushby, K.M.D., Guicheney, P., and Chu, M.-L. COL6A1 Genomic Deletions in Bethlem Myopathy and Ullrich Muscular Dystrophy. Submitted.

### **PARTICIPATION IN SCIENTIFIC MEETINGS:**

Second National Meeting of the American Society for Matrix Biology, November 10-13, 2004, San Diego, CA.

Invited speaker, "Fibulin-2 in Elastic Fiber Organization and Extracellular Matrix Remodeling"

Italian Telethon Scientific Convention, March 6-8, 2005. Salsomaggiore Terme, Italy.

**MON-LI CHU, PH.D.**

*Professor and Vice-Chair of Research*

*Department of Dermatology and Cutaneous Biology*

**PARTICIPATION IN SCIENTIFIC MEETINGS:**

*(continued)*

Invited speaker, "Molecular Genetics of Collagen VI Muscular Dystrophies"

Co-Chair, "Extracellular Matrix, Intracellular Signaling and Mitochondria: Novel Insights into Muscle Disorders"

12<sup>th</sup> International Symposium on Basement Membranes, June 15-17, 2005, Philadelphia, PA.

Vice President, Organizing Committee at Thomas Jefferson University

Matrix Biology Symposium, June 17-18, 2005, Philadelphia, PA.

Invited speaker, "Lessons from Darwin: Heritable Disorders of Collagen Type VI"

**DEPARTMENTAL COMMITTEES:**

Coordinator of "Science Lecture Series" and "Research in Progress"

**PROFESSIONAL ORGANIZATIONS:**

American Society for Biochemistry and Molecular Biology

American Association for the Advancement of Science

International Society for Matrix Biology

American Society for Matrix Biology

Alexander von Humboldt Association of America

**ANDRZEJ FERTALA, PhD**

*Associate Professor of Dermatology and Cutaneous Biology*

**PUBLICATIONS**

A. Steplewski, H. Ito, E. Rucker, R. J. Brittingham, T. Alabyeva, M. Gandhi, F. K. Ko, D. E. Birk, S. A. Jimenez, A. Fertala Position of Single Amino Acid Substitutions in the Collagen Triple Helix Determines Their Effect on Structure of Collagen Fibrils. *J Struct Biol*, 2004 148(3):326-337.

Leitinger B, Steplewski A, Fertala A.J The D2 period of collagen II contains a specific binding site for the human discoidin domain receptor, DDR2. *Mol Biol*. 2005 Feb 18;346(2):661.

Sun YL, Luo ZP, Fertala A, An KN. Stretching type II collagen with optical tweezers. *J Biomech*. 2004 37(11):1665-9.

Brittingham R, Colombo M, Ito H, Steplewski A, Birk DE, Uitto J, Fertala A. "Single Amino Acid Substitutions in Procollagen VII Affect Early Stages of Assembly of Anchoring Fibrils." *J Biol Chem*. 2005 Jan 7;280(1):191-198.

Brennan D, Hu Y, Kljuic A, Choi Y, Joubeh S, Bashkin M, Wahl J, Fertala A, Pulkkinen L, Uitto J, Christiano AM, Panteleyev A, Mahoney MG. Differential structural properties and expression patterns suggest functional significance for multiple mouse desmoglein 1 isoforms. *Differentiation*. 2004 Oct;72(8):434-49.

Kisiel DG, Calvete JJ, Katzhendler J, Fertala A, Lazarovici P, Marcinkiewicz C. Structural determinants of the selectivity of KTS-disintegrins for the alpha1beta1 integrin. *FEBS Lett*. 2004 Nov 19;577(3):478-82.

**PARTICIPATION IN SCIENTIFIC/CLINICAL MEETINGS AND PRESENTATIONS.**

2<sup>nd</sup> ASMB Conference, 2004, San Diego, CA. "Rational Engineering of Collagen-Like Proteins for Tissue Repair"

**PROFESSIONAL ORGANIZATIONS.**

Member:

American Society for Matrix Biology

Protein Society

**REZA F GHOHESTANI, M.D., PhD**

*Resident, Department of Dermatology and Cutaneous Biology*

**PUBLICATION:**

Metz BJ, Ruggeri SY, Hsu S, Reed JA, Ghohestani AS, Uitto J, Ghohestani RF. Linear IgA dermatosis with IgA and IgG autoantibodies to the 180 kDa bullous pemphigoid antigen (BP180): evidence for a distinct subtype.

Int J Dermatol. 2004 Jun;43(6):443-6.

**PRESENTATIONS:**

12<sup>th</sup> International Symposium on Matrix Biology Philadelphia, June 2005

A. M Barria, AS Ghohestani, G. Zhang,, J Uitto, RF Ghohestani

DNA IMMUNIZATION WITH DESMOGLEIN-3, A CADHERIN TYPE CELL-CELL ADHESION MOLECULE, RECAPITULATES PEMPHIGUS VULGARIS

B. T. Roudner\*, A.S. Ghahestani, M. Matsui\*\*, J. Uitto, R.F. Ghohestani

CLINICAL FEATURES OF PHOTODAMAGED HUMAN SKIN ARE ASSOCIATED WITH REDUCTION IN TYPE XVII COLLAGEN AND LAMININ-5, THE MAIN COMPONENTS OF HEMIDESMOSOME/ANCHORING FILAMENT COMPLEXES

C. M. Barria, A. Claudy\*, B. Hudson\*\* , J. Uitto, R.F. Ghohestani

THE  $\alpha$ 5 AND  $\alpha$ 6 CHAINS OF TYPE IV COLLAGEN ARE THE TARGET OF IgG AND IgA AUTOANTIBODIES IN A NOVEL AUTOIMMUNE DISEASE CHARACTERIZED BY CRESCENTIC GLOMERULONEPHRITIS AND SUB-EPIDERMAL BLISTERS.

**HONORS:**

Johnson Award in Basic Sciences Dermatology Research, College of Physicians of Philadelphia

**PROFESSIONAL ACTIVITIES:**

Editor, European Journal of Dermatology

Special Emphasis Panel Member and Grant Reviewer, NIH-NIDCR Rooster, Bethesda

Resident Delegate, Governing Council, Pennsylvania Medical Society, Hershey

Resident Delegate (PA) to the American Medical Association, Chicago June 2005

Council Member on Membership and Member Services Committee, Pennsylvania Medical Society, Hershey

**DAVID J. HERRICK, PhD**

*Research Associate*

*Department of Dermatology and Cutaneous Biology*

**PUBLICATIONS:**

Liao, B., Patel, M., Hu, Y., Charles, S., Herrick, D. J., and Brewer, G. Targeted knockdown of the RNA-binding protein CRD-BP promotes cell proliferation via an insulin-like growth factor II-dependent pathway in human K562 leukemia cells. *J. Biol. Chem.* 279: 48716-48724, 2004.

Zhou, D., Herrick, D. J., Rosenbloom, J., and Chaqour, B. Cyr61 mediates the expression of VEGF,  $\alpha$ -integrin and  $\alpha$ -actin genes through cytoskeletally based mechanotransduction mechanisms in bladder smooth muscle cells. *J Appl Physiol.* 98: 2344-2354, 2005.

Liao, B., Hu, Y., Herrick, D. J., and Brewer, G. The RNA-binding protein IMP-3 is a translational activator of IGF-II leader-3 mRNA during proliferation of human K562 leukemia cells. *J. Biol. Chem.* 280: 18517-18524, 2005.

**PARTICIPATION IN SCIENTIFIC/CLINICAL MEETINGS AND PRESENTATIONS:**

Herrick, D.J., Wang, D., Uitto, J.: Functional analysis of the  $\alpha_1(I)$  procollagen 5'-stem loop and 3'-UTR: Role of the  $\alpha$ CP, AUF1 and PABP RNA-binding proteins. (Presented at the Translational Control Meeting, Cold Spring Harbor Laboratories, 2004.)

Castorino, J.J., Brewer, G.A., Herrick, D.J: Functional dissection of the *c-myc* coding region determinant. (Presented at the Translational Control Meeting, Cold Spring Harbor Laboratories, 2004.)

**OLGA IGOUCHEVA, PhD**

*Assistant Professor of Dermatology and Cutaneous Biology*

**PUBLICATIONS**

Igoucheva, O., Alexeev, V. and Yoon, K. Oligonucleotide-directed mutagenesis and targeted gene correction: a mechanistic point of view. *Current Molecular Medicine*, 4, 445-463, 2004.

Igoucheva, O. and Yoon, K. "Gene targeting by oligonucleotides in keratinocytes" in press, *Epidermal Cell Protocols, Methods in Molecular Biology*, ed, Turksen, K. Humana Press, pp. 287-302, 2004.

Igoucheva, O., Alexeev, V. and Yoon, K. Mechanism of gene repair open for discussion. *Oligonucleotides*, 14, 311-321, 2004.

Igoucheva, O., Alexeev, V. and Yoon, K. Differential cellular responses to exogenous DNA in mammalian cells and its effect on oligonucleotide-directed gene modification. *Gene Therapy*, 2005 (in press).

Igoucheva, O., Alexeev, V., Scharer, O. and Yoon, K. Involvement of ERCC1/XPF and XPG in Oligodeoxynucleotide-Directed Gene Modification *Oligonucleotides*, 2005 (submitted)

**PARTICIPATION IN SCIENTIFIC/CLINICAL MEETINGS AND PRESENTATIONS.**

Matrix Biology Days, 2005



**QIUJIE JIANG, MD, PhD**

*Post-Doctoral Fellow*

*Department of Dermatology and Cutaneous Biology*

**PUBLICATIONS:**

1. Muhle C, **Jiang QJ**, Charlesworth A, Bruckner-Tuderman L, Meneguzzi G, Schneider H. Novel and recurrent mutations in the laminin-5 genes causing lethal junctional epidermolysis bullosa: molecular basis and clinical course of Herlitz disease. *Hum Genet* 2005;116(1-2):33-42.
2. **Jiang QJ**, Uitto J. Animal models of epidermolysis bullosa--targets for gene therapy. *J Invest Dermatol.* 2005;124(3):xi-xiii.
3. Klement JF, Matsuzaki Y, **Jiang QJ**, Terlizzi J, Choi HY, Li K, Pulkkinen L, Birk DE, Sundberg JP, Uitto J. A mouse model for pseudoxanthoma elasticum: target ablation of ABCC6 results in aberrant calcification of connective tissues. *Mol. Cell. Biol* (in press)
4. Matsuzaki Y, Nakano A, **Jiang QJ**, Pulkkinen L, Uitto J. Tissue-specific expression of the ABCC6 gene. *J Invest Dermatol.* (in press)
5. **Jiang QJ**, Matsuzaki Y, Li K and Uitto J. Transcriptional regulation and characterization of the promoter region of the human ABCC6 gene.(submitted JID)

**PRESENTATIONS:**

1. Klement JF, Matsuzaki Y, **Jiang QJ**, Terlizzi J, Choi HY, Li K, Pulkkinen L, Birk DE, Sundberg JP, Uitto J. A mouse model for pseudoxanthoma elasticum: target ablation of ABCC6 results in aberrant calcification of connective tissues. *12<sup>th</sup> international symposium on basement membranes*, June 15-18, 2005, Philadelphia, USA
2. **Jiang QJ**, Matsuzaki Y, Pulkkinen L, Uitto J. Tissue-specific expression of the ABCC6 gene. *The 66th Annual Meeting of the Society for Investigative Dermatology*, May 4-7, 2005, St.Louis, Missouri, USA

**JOHN F. KLEMENT, PhD**

*Research Assistant Professor of Dermatology and Cutaneous Biology*

**PUBLICATIONS:**

**Klement, J.F., Y. Matsuzaki, Q-J. Jiang, J. Terlizzi, H.Y. Choi, N. Nujimoto, K. Li, L. Pulkkinen, D.E. Birk, J. P. Sundberg, and J. Uitto.** 2005. Targeted ablation of the *Abcc6* gene results in ectopic mineralization of connective tissues. *Mol Cell Biol* **18**: (In Press; Expected publication in Sept. 15 issue)

**UNIVERSITY COMMITTEES**

Institutional Animal Care and Use Committee  
Extracellular Matrix Visioning Committee

**PROFESSIONAL ORGANIZATIONS**

American Society for Microbiology  
American Association for the Advancement of Science

**TORELLO M. LOTTI, MD**

*Professor of Dermatology*

*Director of the Operative Unit of Physiotherapeutic Dermatology*

*Department of Dermatological Sciences*

*University of Florence Medical School*

*Florence, Italy*

**PUBLICATIONS 2004-2005:**

**JOURNALS:**

1. E. Tsourelis-Nikita, G. Menchini, TM Lotti. Il prurito: eziologia, meccanismi fisiopatologici, inquadramento e gestione del paziente. *Il Dermatologo* 2004;4:4-9
2. I. Ghersetich, G. Buggiani, B. Brazzini, T. Lotti. La vasculite cutanea dei piccoli vasi. *G Ital Dermatol Venereol* 2004;139:389-413
3. G. Buggiani, T. Lotti. La terapia della vitiligine: novità. *New pages of therapy* 2004;vol3,n2:13-22
4. Formigli L, Zecchi S, Protopapa C, Caporale D, Cammarota N, Lotti TM. Bio-Alcamid: an electron microscopy study after skin implantation. *Plast Reconstr Surg* 2004;113(3):1104-06
5. Ghersetich I, Buggiani G, Brazzini B, Lotti T. La vitiligine: cosa c'è di nuovo? *G Ital Dermatol Venereol* (registrato n° 3494, IN PRESS)
6. Holikova Z, Massi D, Lotti T, Hercogova J. Insight into the pathogenesis of sporadic basal cell carcinoma. *Int J Dermatol* 2004;43:865-869
7. Lotti T, Greco A, Hercogova J. Vitiligo: Problems and Solutions. *Acta Dermatovenereologica Albanica* 2004;1:47-50

**BOOK CHAPTERS:**

1. T Lotti, G Hautmann, J Hercogova. Vitiligo: disease or symptom? From the confusion of the past to current doubts. In: *Vitiligo: problems and solutions*. Marcel Dekker Inc. 2004:1-14
2. E Del Bianco, G Muscarella, T Lotti. Vitiligo: a disorder of the microvessels? In: *Vitiligo: problems and solutions*. Marcel Dekker Inc. 2004:93-98
3. G Hautmann, S Moretti, T Lotti, J Hercogova. Pathogenesis of vitiligo: evidence of a possible ongoing disorder of the cutaneous microenvironment. In: *Vitiligo: problems and solutions*. Marcel Dekker Inc. 2004:99-122

**TORELLO M. LOTTI, MD**

*Professor of Dermatology*

*University of Florence Medical School*

*Florence, Italy*

4. G Hautmann, T Lotti, J Hercogova. Vitiligo: emotional aspects and personality. In: Vitiligo: problems and solutions. Marcel Dekker Inc. 2004:225-234
5. E Tsourelis-Nikita, C Comacchi, G Menchini, T Lotti. Vitamins and vitiligo. In: Vitiligo: problems and solutions. Marcel Dekker Inc. 2004:281-284
6. I Ghersetich, B Brazzini, T Lotti, G Menchini. Alternative treatments for vitiligo. In: Vitiligo: problems and solutions. Marcel Dekker Inc. 2004:285-292
7. G Menchini, T Lotti, E Tsourelis-Nikita, J Hercogova. UV-B narrow band microphototherapy: a new treatment for vitiligo. In: Vitiligo: problems and solutions. Marcel Dekker Inc. 2004:323-334
8. G Menchini, T Lotti, E Tsourelis-Nikita, J Hercogova. Vitiligo: problems and nonsurgical solutions. In: Vitiligo: problems and solutions. Marcel Dekker Inc. 2004:335-340
9. G Menchini, T Lotti, E Tsourelis-Nikita, J Hercogova. Vitiligo and the internet. In: Vitiligo: problems and solutions. Marcel Dekker Inc. 2004:365-368
10. B Bianchi, T Lotti, J Hercogova. Nevus depigmentosus. In: Vitiligo: problems and solutions. Marcel Dekker Inc. 2004:413-418
11. E Tsourelis-Nikita, G Menchini, T Lotti, H Grossman. Albinism. In: Vitiligo: problems and solutions. Marcel Dekker Inc. 2004:461-472
12. T Lotti. Introduzione. In: Ulcerazione e cicatrizzazione cutanea. UTET Periodici 2005:2-11
13. B Bianchi, M Stante, T Lotti. Procollagene, collagene e fibronectina: la riparazione tessutale avviene così. In: Ulcerazione e cicatrizzazione cutanea. UTET Periodici 2005:42-45
14. M Benci, T Lotti. Gli attivatori del plasminogeno e la riparazione tessutale. In: Ulcerazione e cicatrizzazione cutanea. UTET Periodici 2005:56-62
15. G Buggiani, T Lotti. Collagene e fattori di crescita: ruolo nella riparazione tessutale. In: Ulcerazione e cicatrizzazione cutanea. UTET Periodici 2005:74-80
16. M Benci, T Lotti. Ruolo dei fattori genetici nell'induzione delle ulcere degli arti inferiori. In: Ulcerazione e cicatrizzazione cutanea. UTET Periodici 2005:114-121

**TORELLO M. LOTTI, MD**

*Professor of Dermatology*

*University of Florence Medical School*

*Florence, Italy*

**ABSTRACTS:**

1. G. Buggiani, T. Lotti. La vitiligine: nuovi concetti e nuove terapie. Atti XLI Congresso Nazionale ADOI, Capri, 1-4 Oct 2004
2. B. Brazzini, I. Ghersetich, T. Lotti. UVB microphototherapy: why, where and how. 13<sup>th</sup> EADV Congress, Florence, Italy, 17-21 Nov 2004
3. S. Giorgini, A. Greco, M. Stante, T. Lotti. Tattoos: from vanity to body art an increasing challenge for dermatologists. 13<sup>th</sup> EADV Congress, Florence, Italy, 17-21 Nov 2004
4. A. Greco, S. Giorgini, S. Francalanci, T. Lotti. Clinical relevance and contact allergy to gold sodium thiosulfate in Florence. 13<sup>th</sup> EADV Congress, Florence, Italy, 17-21 Nov 2004
5. S. Giorgini, A. Greco, T. Lotti. Hair coloring allergies. 13<sup>th</sup> EADV Congress, Florence, Italy, 17-21 Nov 2004
6. T. Lotti. Quality of life: a strategic point in Herpes Zoster treatment. 13<sup>th</sup> EADV Congress, Florence, Italy, 17-21 Nov 2004
7. L.M. Lova, P. Campolmi, P. Bonan, G. Cannarozzo, B. Brazzini, T. Lotti. Pathophysiology of lower limb teleangectasias bridging the gap between haemodynamics and clinical practice. 13<sup>th</sup> EADV Congress, Florence, Italy, 17-21 Nov 2004
8. G. Cannarozzo, P. Campolmi, P. Bonan, R.M. Lova, B. Brazzini, P. Cappugi, T. Lotti. Laser treatment of lower limb teleangectasias by using the 1064 Nd:Yag laser: techniques and results. 13<sup>th</sup> EADV Congress, Florence, Italy, 17-21 Nov 2004
9. Lotti T, Buggiani G. Terapia termale. Convegno Regionale Monotematico "Dermatite Seborroica". Comelico Superiore (BL), 4-5 Mar 2005
10. Lotti T, Urpe M, Buggiani G. Psycho-neuro-endocrine-immunology: what's new? 1<sup>st</sup>/17<sup>th</sup> Congress of the Association of Dermatovenereologists of Serbia and Montenegro, Belgrade, 2-4 Jun 2005 (Atti)

## **TORELLO M. LOTTI, MD**

*Professor of Dermatology*

*University of Florence Medical School*

*Florence, Italy*

### **VISITING PROFESSORSHIPS AND INVITED LECTURESHIPS:**

Visiting Professor & Director, International Centre for Balneology, Cell Biology, and Physiotherapy Thomas Jefferson University, Philadelphia (PA, USA)

Visiting Professor of Dermatology, Charles University, Prague, republica Ceca

Visiting Professor of Dermatology, Bowman Gray University, Winston Salem (NC, USA)

Visiting Professor of Dermatology, University of Louisville, Louisville (KY, USA)

Visiting Professor of Dermatology, Medical University of South Carolina, Charleston (SC,USA)

Visiting Professor of Dermatology, Charles University, Prague, Czech Republic

University Medal, Charles University, Czech Republic

Key Note Lecturer, Japanese Society of Cosmetic Dermatology

Chairman Centro Universitario di Dermatologia Biologica e Psicosomatica, Università di Firenze, Siena e Milano, Italy.

Full Professor of Dermatology, Dept of Dermatological Sciences, University of Florence, Italy.

### **PARTICIPATION TO SCIENTIFIC/CLINICAL MEETINGS AND PRESENTATIONS**

**2004 Chair**, 62<sup>nd</sup> Annual Meeting American Academy of Dermatology.

Symposium: Itch. Washington D.C., USA, 6-11 September

**2004 Chair and Invited Speaker**, First International Congress of Cosmetic Dermatology and Laser. Vitiligo: what is new for the Dermatocosmetologists? Cairo, Egypt. 26-30 Sept.

**2004 Chair**, Dolore e Medicina Psicosomatica. Tavola Rotonda: Dolore e Psicosomatica: dall'epistemologia alla clinica. Firenze, Italy. 2 Oct.

**2004 Chair and Invited Speaker** 6° Congresso Internazionale di Medicina Estetica. Sessione 3: la cura della pelle e delle irregolarità cutanee La Vitiligine: Nuovi Concetti e Nuove Terapie. Milano, Italy. 8-10 Oct.

**2004 Chair and Invited Speaker** XLII Congresso Nazionale A.D.O.I. Vitiligine: Cosa non si deve fare. Rimini, Italy. 13-16 Ott.

**TORELLO M. LOTTI, MD**

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**2004 Invited Speaker** Patologo e Clinico a Confronto: Vulva e Corpo Uterino – Corso teorico pratico per il ginecologo, il dermatologo ed il patologo. Discromie Cutanee: Vitiligine. Firenze, Italy. 22-23 Ott.

**2004 President, Chair and Invited Speaker** 13th Congress of the European Academy of Dermatology and Venereology – Florence, Italy, 17-21 November Symposium – Managing histamine mediated skin disease: antihistamines in adults and children Symposium – New treatment strategy for Herpes Zoster treatment: “Quality of life: a strategic point in Herpes Zoster treatment” Workshop – UVB microphototherapy: why, when and how

**2005 Chair and Invited Speaker** 21° Congresso Nazionale SIMG Cute e sistema endocrino Firenze, Italy, 25-27 November

**2005 Chair and Invited Lecturer** 8° Congresso Internazionale SIES “Novità e aggiornamenti in tema di Medicina e Chirurgia Estetica” Bologna, Italia, 11-13 Febbraio

**2005 Chair and Invited Speaker** V Corso di Aggiornamento “Dermatologia Estetica” Ipomelanosi: eziopatogenesi Ipomelanosi: possibili trattamenti Montecatini Terme, Italy, 7-11 March

**2005 Chair and Invited Speaker** Associazione Dermatologica Ionica International Congress Aspetti Immunologici della vitiligine Giardini Naxos Taormina, Italy, 21-24 April

**2005 Invited Speaker** 15th World Congress of Aesthetic Medicine Lecture – Hypomelanotic skin disorders: problems and aesthetic solutions. Rome, Italy, 5-8 May

**2005 Chair and Invited Speaker** 3<sup>rd</sup> EADV Spring Symposium – Sofia, Bulgaria, 19-22 May

**2005 Honorary Membership** Association of Dermatovenereologists of Serbia and Montenegro

**2005 Invited Lecturer** 1st/17th Congress of the Association of Dermatovenereologists of Serbia and Montenegro. Psycho-neuro-endocrine-immunology: what’s new? Belgrade, Serbia and Montenegro, 2-4 June

**2005 Invited Speaker** 2° Congresso Nazionale Unificato di Dermatologia e Venereologia. Le macchie bianche: quali terapie? La sindrome di Raynaud La vitiligine: nuove acquisizioni. Genova, Italy, 8-11 June

**MEMBERSHIP IN SCIENTIFIC SOCIETIES:**

European Academy of Dermatology and Venereology (EADV)

Societa' Italiana di Dermatologia e Venereologia (SIDEV)

Societa' Italiana per lo Studio della Emostasi e della Trombosi (ISD)

European Society for Dermatologic Research (ESDR)

**TORELLO M. LOTTI, MD**

*Professor of Dermatology*

*University of Florence Medical School*

*Florence, Italy*

**MEMBERSHIP IN SCIENTIFIC SOCIETIES:**

International Society of Dermatology (ISD)

American Academy of Dermatology (AAD)

Society for Investigative Dermatology (SID)

Society for Cutaneous Ultrastructure Research (SCUR)

Società Italiana per la Riparazione Tissutale (SIRT)

European Wound Management Association (EWMA)

College of Physicians of Philadelphia

**HONORS/SPECIAL RECOGNITIONS:**

Honorary member : Czech Society of Dermatology

Honorary member: Polish Society of Dermatology

Honorary member: American Dermatological Association

Honorary member: Bulgarian Society of Dermatology

Honorary member: French Society of Dermatology

Honorary member Rumanian Society of Dermatology

Honorary member: Argentine Society of Dermatology

Honorary member: Association of Dermatovenereologists of Serbia and Montenegro

**EDITORIAL ACTIVITIES 2004-2005:**

1. **Editor:** International Journal of Immunopathology and Pharmacology, Section of Dermatology, Biomedical Research Press & Pacini Editore, Pisa, Italia.
2. **Editorial Board:** Journal of the American Academy of Dermatology, 1993-.
3. **Editor:** Il Dermatologo. Periodico Mensile. CIC Edizioni Internazionali, Italy. since 2003
4. **Editor:** European Handbook of Dermatological Treatments. Springer 2004
5. **Editor:** Vitiligo: Problems and Solutions. Marcel Dekker 2004
6. **Editor:** Manuale Europeo di Clinica e Terapia Dermatologica. CIC Edizioni 2004.
7. **Editor:** Ulcerazione e Cicatrizzazione Cutanea: nuovi concetti e nuove terapie. UTET 2005
8. **Editor:** Dermatologia Pratica per Immagini 2° Edizione. UTET 2005

**REVIEWER FOR:**

Annals of Medical Science

Acta Dermato Venereologica

British Journal of Dermatology

Journal of Investigative Dermatology

Journal of the American Academy of Dermatology

International Angiology

International Journal of Dermatology



Italian General Review of Dermatology  
Giornale Italiano di Dermatologia e Venereologia

**PARTICIPATION TO CONGRESS ORGANIZATION 2004-2005:**

2004 First International Congress of Cosmetic Dermatology and Laser. Cairo, Egypt.  
26-30 Sept.

2004 Congress President – 6<sup>th</sup> International Congress of Aesthetic Medicine. Milan,  
Italy. 8-10 October

2004 Congress President – 13th Congress of the European Academy of Dermatology  
and Venereology – Florence, Italy, 17-21 November

2005 Coordinamento e Segreteria Scientifica – V Corso di Aggiornamento  
“Dermatologia Estetica”. Montecatini Terme, 7-11 Marzo

**LAWRENCE CHARLES PARISH, MD**

*Clinical Professor*

*Department of Dermatology and Cutaneous Biology*

**PUBLICATIONS:**

Routh HB, Parish LC, La Sen S, Bhowmik B. Skin torture. *Clin Dermatol.* 2005 May-Jun;23(3):307-9.

Wolf R, Parish JL, Marcus BS, Parish LC. Continued use of nonsteroidal topical treatments: comment on the FDA talk paper. *Skinmed.* 2005 May-Jun;4(3):133-4.

Wolf R, Parish LC. Life-threatening skin diseases: part I. *Clin Dermatol.* 2005 Mar-Apr;23(2):131-3.

Wolf R, Parish JL, Parish LC. Propecia: the goose that lays the golden eggs. *Skinmed.* 2005 Mar-Apr;4(2):67-8.

Parish LC, Lowthian PT, Witkowski JA. The Infected Decubitus Ulcer: Superman's Kryptonite. *Skinmed.* 2005 Jan-Feb; 4(1): 7-8.

Oumeish OY, Parish LC. Sexually transmitted diseases in clinical practice. *Clin Dermatol.* 2004 Nov-Dec;22(6):451

Witkowski JA, Parish LC. The assessment of acne: an evaluation of grading and lesion counting in the measurement of acne. *Clin Dermatol.* 2004 Sep-Oct;22(5):394-7.

Witkowski JA, Parish LC. On being blue: argyria still exists. *Skinmed.* 2004 Nov-Dec;3(6):304-5.]

Parish LC. Being thoroughly modern dermatologically in the nineteenth century: L. Duncan Bulkley (1845-1928) revisited. *Skinmed.* 2004 Sep-Oct;3(5):249-50.

Parish LC, Witkowski JA. Quackery and the dermatologist. *Skinmed.* 2004 Jul Aug;3(4):185-6.

Witkowski JA, Parish JL, Parish LC. Traumatic purpuric penile ulcer. *Acta Dermatovenerol Croat.* 2004;12(2):96-8.

**BOOKS AND CHAPTERS**

Parish, LC, Crissey, JT. Historical aspects of nail disease. In Scher, RK, Daniel, CR. *Nails: Therapy, Diagnosis, Surgery*, 3<sup>rd</sup> Ed, Philadelphia, Elsevier Saunders. 2005:7-12

**LAWRENCE CHARLES PARISH, MD**

*Clinical Professor*

*Department of Dermatology and Cutaneous Biology*

**BOOK REVIEWS:**

The Book of Skin. J Hist Med All Sci. 2005; 60:2.

Locating Medical History: The stories and their meanings. Ann Int Med. 2004;7:580.

**LAY PAPERS:**

Clinical trials in children Pediatrics for Parents 2005; 21(8): 4.

Molluscum contagiosum. Pediatrics for Parents. 2005: 21(6), 4.

Atopic dermatitis. Pediatrics for Parents. 2005: 21(5), 4.

Myths about acne. Pediatrics for Parents. 2005: 21(4), 4.

**SPECIAL ISSUES – EDITOR:**

Wolf R, Parish LC. Life-Threatening Skin Diseases (Part I), Clin Dermatol. 2005; 23 (2): 131-18.

Wolf R, Parish LC. Life-Threatening Skin Diseases (Part II), Clin Dermatol. 2005; 23 (3): 223-310.

Oumeish, OY, Parish, LC. Sexually Transmitted Skin Diseases in Clinical Practice. Clin Dermatol. 2004; 22 (6): 451-547.

**EXHIBITS:**

Parish, LC, Parish, JL, Routh, HB: The psoriatic patient. American Academy of Dermatology, New Orleans, Louisiana, February 18-22, 2005

Parish, LC, Parish, JL, Routh, HB: Doxycycline 20 mg tablets administered twice daily as maintenance therapy for the treatment of acne. American Academy of Dermatology, New Orleans, Louisiana, February 18-22, 2005

**LAWRENCE CHARLES PARISH, MD**

*Clinical Professor*

*Department of Dermatology and Cutaneous Biology*

**PRESENTATIONS:**

The terminology of balneology, mineral water, and spas. European Academy of Dermatology and Venereology, Sofia, Bulgaria, May 19, 2005

Integrative dermatology and the clinician. American Academy of Dermatology, New Orleans, Louisiana, February 18, 2005

Integrative medicine and cosmetic dermatology. European Academy of Dermatology and Venereology, Florence, Italy, November 20, 2004

The basics of balneology, mineral water, and spas. European Academy of Dermatology and Venereology, Florence, Italy, November 19, 2004

**SEMINARS ORGANIZED AND CHAIRED:**

History of Dermatology. History of Dermatology Society, Sofia, Bulgaria, May 19, 2005

History of Dermatology. History of Dermatology Society, New Orleans, Louisiana, February 17, 2005

**MEETINGS ATTENDED:**

3rd Spring Symposium. European Academy of Dermatology and Venereology, Sofia, Bulgaria, May 19-22, 2005

65th Annual Meeting of the American Academy of Dermatology, New Orleans, Louisiana, February 18-22, 2005

13th European Academy of Dermatology and Venereology, Florence, Italy, November 17-20, 2004

**HONORS/SPECIAL RECOGNITION:**

Appointed Visiting Professor of Dermatology and Venereology, Sofia Medical University, Sofia, Bulgaria

**LAWRENCE CHARLES PARISH, MD**

*Clinical Professor*

*Department of Dermatology and Cutaneous Biology*

**PROFESSIONAL ORGANIZATIONS:**

**MEMBER OR FELLOW:**

American Association for the History of Medicine, American College of Physicians, American Dermatological Association, American Academy of Dermatology, Canadian Dermatology Association, Canadian Society for the History of Medicine, College of Physicians of Philadelphia (Section on the History of Medicine of the College of Physicians of Philadelphia, Section on Dermatology of the College of Physicians of Philadelphia), European Academy of Dermatology and Venereology, European Society for the History of Dermatology and Venereology, French Society for the History of Dermatology and Venereology, History of Dermatology Society (president), International Academy of Cosmetic Dermatology (president), Pennsylvania Academy of Dermatology, Philadelphia Dermatological Society

**HONORARY MEMBER OR FELLOW:**

American College of Veterinary Dermatology, Bulgarian Society of Dermatology, Florida Society of Dermatology, Italian Society of Dermatology and Venereology, Polish Academy of Dermatology, Venezuelan Society of Dermatology,

**CORRESPONDING MEMBER OR FELLOW:**

British Association of Dermatologists, French Society of Dermatology and Syphilology, Israeli Society of Dermatology

**ELLEN G. PFENDNER, PhD**

*Assistant Professor of Dermatology and Cutaneous Biology*

**PUBLICATIONS**

Schara U, Tucke J, Mortier W, Nusslein T, Rouan F, Pfindner E, Zillikens D, Bruckner-Tuderman L, Uitto J, Wiche G, Schroder R. 2004 Severe mucous membrane involvement in epidermolysis bullosa simplex with muscular dystrophy due to a novel plectin gene mutation. *European Journal of Pediatrics* 163(4-5):218-22.

Pfindner, E and J Uitto 2005 Plectin mutations can cause epidermolysis bullosa with pyloric atresia *Journal of Investigative Dermatology* 124 (1):111-115.

Pfindner, E, F Rouan, J Uitto. 2005 Progress in epidermolysis bullosa: the phenotypic spectrum of plectin mutations. *Experimental Dermatology* 14 (4):241-249.

Pfindner, E, S Sadowski and J Uitto. 2005 Epidermolysis Bullosa Simplex: Recurrent and De Novo Mutations in the KRT5 and KRT14 Genes, Phenotype/Genotype Correlations, and Implications for Genetic Counseling and Prenatal Diagnosis. *Journal of Investigative Dermatology* 125(2):239-243.

**SCIENTIFIC MEETINGS:**

Sa'd, J. Abu, M. Indelman, E. Pfindner, G. Richard, J. Uitto, R. Bergman, M. Kanaan, E. Sprecher, 2005 Mutation analysis in a cohort of Israeli patients with dystrophic epidermolysis bullosa. *Journal of Investigative Dermatology* 124(4) suppl:A74.

Pfindner ,E., F Rouan, and J Uitto. 2005 The phenotypic spectrum of plectin mutations in epidermolysis bullosa. *Journal of Investigative Dermatology* 124(4) suppl:A85.

Varki, R., E. Pfindner, and J Uitto 2005 The spectrum of COL7A1 mutations in dystrophic epidermolysis bullosa. *Journal of Investigative Dermatology* 124(4) suppl:A85.

**MEMBERSHIP:**

Society of Investigative Dermatology

## **SONSOLES PIERA-VELAZQUEZ, PhD**

*Assistant Professor*

*Department of Dermatology and Cutaneous Biology*

### **PUBLICATIONS:**

Colter D.C., **Piera-Velázquez S.**, Hawkins D.F., Whitecavage M., Jimenez S.A., Stokes D.G. Regulation of the human Sox9 by the CCAAT-binding factor. *Matrix Biology*. 24(3):185-97, 2005.

Mendoza F.A., Arlett C.M. Sandorfi N., Latinis K, **Piera-Velázquez S.**, Jimenez S.A. 2005. Description of twelve cases of nephrogenic fibrosing dermopathy and review of the literature. *Seminars in Arthritis and Rheumatism* (in press).

**Piera-Velázquez S.**, Hawkins D.F., Whitecavage M., Colter D.C., Jimenez S.A., Stokes D.G. 2005. Regulation of the human Sox9 by the SP1 and CREB-binding factors. *Matrix Biology* (Submitted).

**S. Piera-Velázquez;** C. Derk; S.A. Jiménez. Patogénesis de la Esclerosis Sistémica. Revista Peruana de Reumatología. Sociedad Peruana de Reumatología. Vol. 10 (3), 2004.

### **ABSTRACTS:**

**Piera-Velázquez S.**, Hawkins D.F., Whitecavage MK, Jimenez S.A. and Stokes D.G. Characterization of the human Sox9 proximal promoter region. OARSI. Chicago, USA, 2004.

### **MEETING PARTICIPATION:**

ACR/ARHP Annual Scientific Meeting, San Antonio, TX. October 16-21, 2004.

9<sup>th</sup> World Congress of the OARSI. Chicago, IL. December 2-5, 2004.

12<sup>th</sup> International Symposium on Basement Membranes. Matrix Biology Symposium. Philadelphia, PA. June 15-18, 2005.

**SONSOLES PIERA-VELAZQUEZ, PhD**

*Assistant Professor*

*Department of Dermatology and Cutaneous Biology*

**PRESENTATIONS:**

Constitutive expression of the two novel cyclooxygenase-1 (prostaglandin h2 synthase) isoforms cox-3 and cox-1a by normal and osteoarthritic (OA) human articular cartilage chondrocytes. **S. Piera-Velazquez** and S. A. Jimenez. OARSI, Chicago, IL. December 2-5, 2004.

**PROFESSIONAL ORGANIZATIONS:**

Membership, OARSI (Osteoarthritis Research Society International).



## **GABRIELE RICHARD, MD**

*Associate Professor*

*Department of Dermatology and Cutaneous Biology*

### **PUBLICATIONS:**

1. Richard, N. Brown, A. Ishida-Yamamoto and A. Krol. Expanding the phenotypic spectrum of Cx26 disorders: Bart-Pumphrey Syndrome is caused by a novel missense mutation in *GJB2*. J Invest Dermatol, 123:856-63, 2004
2. Sprecher, E, Tesfaye-Kedjela, A, Ratajczak, P, Bergman, R, Richard-G: Deleterious mutations in SPINK5 in a patient with congenital ichthyosiform erythroderma: molecular testing as a helpful diagnostic tool for Netherton syndrome. Clin Exp Dermat, 29:513-517, 2004
3. Langschuetzer CM, Klaussegger A, Pohla-Gubo G, Hametner R, Richard G, Uitto J, Hintner H, Bauer JW. Gene symbol: KRT14. Disease: epidermolysis bullosa simplex, Koebner. Hum Genet. 115:171, 2004
4. Ishida-Yamamoto, C. Deraison, C. Bonnart, E. Bitoun, T.J. O'Brien, K. Wakamatsu, S. Ootsubo, H. Takahashi, Y. Hashimoto, P. JC Dopping-Hepenstal, J. A. McGrath, H. Iizuka, G. Richard, A. Hovnanian: vTransport of LEKTI via lamellar granules: relevance to desquamation in normal human skin and the pathogenesis of Netherton syndrome. J Invest Dermatol, 124:360-366, 2005
5. A.S. Geyer, P. Ratajczak, M. Pol-Rodriguez, M. Garzon, G. Richard. Netherton syndrome with extensive skin peeling and failure to thrive due to a homozygous frameshift mutation in *SPINK*. Dermatology, 210: (4): 308-314, 2005
6. S.C. Chao, G. Richard, J.Y. Lee. Netherton syndrome: report of two Taiwanese siblings with staphylococcal scalded skin syndrome and mutation of SPINK5. Br J Dermatol. 152(1):159-165, 2005
7. R. Kellermayer, M. Keller, P. Ratajczak, E. Richardson, F. Harangi, E. Mérei, B. Meleg, G. Kosztolányi, G. Richard. Bigenic connexin mutations in a patient with hidrotic ectodermal dysplasia. Eur J Dermatol, 15(2):75-79, 2005
8. O. Topaz, R. Bergman, G. Richard, E. Sprecher. Absence of intraepidermal glycosyltransferase ppGalNAc-T3 expression in familial tumoral calcinosis. Am J Pathol, 27: 211-215, 2005
9. S.M. Morley, M.I. White, M. Rogers, D. Wasserman, P. Ratajczak, W.H.I. McLean, and G. Richard. A New, Recurrent Mutation of *GJB3* (Cx31) in Erythrokeratoderma Variabilis. Br J Dermatol, 152: 1143-48, 2005
10. D.L. Shurman, L. Glazewski, A. Gumpert, J.D. Zieske and G. Richard. Connexin expression in the human corneal epithelium in vivo and in vitro. Invest Ophthalmol Vis Sci, in press

## **GABRIELE RICHARD, MD**

*Associate Professor*

*Department of Dermatology and Cutaneous Biology*

11. Y. Frishberg, O. Topaz, R. Bergman, D. Behar, D. Fisher, D. Gordon, G. Richard, E. Sprecher. Hyperostosis-hyperphosphatemia syndrome is caused by mutations in *GALNT3* and is allelic to familial tumoral calcinosis. J Molec Medicine, in press
12. F Erdem, D Yucelten, N Elcioglu, T Keck, P Ratajczak, and G Richard. KID syndrome: rapid response to itraconazole in a child with fungating Candida infection of the scalp. J Eur Acad Dermatol Venerol, under review
13. CK Abrams, VK Verselis, TA Bargiello, MVL Bennett, DP Kelsell, G Richard and FF Bukauskas: Properties of connexin 31, a gap junction protein, associated with diseases of the skin and auditory system. PNAS, under review
14. I Chefetz, R Heller, A Galli-Tsinopoulou, G Richard, B Wollnik, M Indelman, F Korber, O Topaz, R Bergman, E Sprecher, E Schönau. A novel homozygous missense mutation in *FGF23* causes familial tumoral calcinosis associated with disseminated visceral calcification. Hum Genet, in press
15. SC Kelly, P Ratajczak, M Keller, SM Purcell, TD Griffin, G Richard. A novel *GJA1* mutation in oculo-dento-digital dysplasia with curly hair and hyperkeratosis. J Am Acad Dermatol, submitted
16. Richardson E, Lee J, Hyde P, Richard G. A novel mutation and large size polymorphism affecting the V2 domain of keratin 1 in an African-American family with severe, diffuse PPK of the Ichthyosis Hystrix Curth-Macklin type. J Invest Dermatol, under review

### **Invited Reviews**

17. G. Richard. Molecular genetics of the ichthyoses. In: Genetic Disorders of the Skin. Der Kaloustian and Happle (eds), Am J Med Genet (Semin Med Genet) 131C:32-44, 2004
18. J. Uitto and G. Richard. Progress in epidermolysis bullosa: Genetic classification and clinical implications. In: Genetic Disorders of the Skin. Der Kaloustian and Happle (eds), Am J Med Genet (Semin Med Genet) 131C:61-74, 2004
19. G. Richard. Connexin Disorders of the Skin. In: 'Molecular Genetics in Dermatology'. G. Richard (ed). Clinics in Dermatology, 23:23-32, 2005
20. G. Richard. Erythrokeratoderma Variabilis: An Update on Clinics and Genetics. G Ital Dermatol Venereol, in press

### **Other Publications and Editorials**

21. G. Richard and J. Uitto. The complexity of genetics in heritable skin diseases. Editorial (In this issue). J Invest Dermatol 2005

## **GABRIELE RICHARD, MD**

*Associate Professor*

*Department of Dermatology and Cutaneous Biology*

### **Book Chapters**

22. G. Richard and J. Uitto. Inherited Skin disease. In: *Encyclopedic Reference of Genomics and Proteomics in Molecular Medicine*. D. Ganten & K. Ruckpaul (eds) Springer International, in press.
23. J. Uitto, G. Richard and A. Christiano. Epidermolysis Bullosa. In: *Principles of Molecular Medicine*, in press
24. G. Richard. Disorders of Cornification. In: 'Inherited hair disorders'. E. Sprecher (ed), Nova Science Publishers, Inc., in press
25. G. Richard. Erythrokeratodermias. In: *Treatment of Skin Diseases. Comprehensive therapeutic strategies*. 2<sup>nd</sup> edition. M. Lebwohl et al (eds), Mosby, Philadelphia, in review
26. G. Richard. Human connexins in skin development and skin disorders. In: *Gap Junctions in Development and Disease*. 1<sup>st</sup> edition. Elke Winterhager et al. (ed), Springer, Berlin, in review

### **Participation in Scientific/Clinical Meetings and Presentations.**

#### **Oral presentations**

1. Summer Meeting of the American Academy of Dermatology. July 2004. Invited speaker 'Connexins in Pediatric Dermatoses'. G. Richard
2. Invited lecture. Grand Rounds, Department of Dermatology, Northwestern University, Chicago. May 26, 2004. Human Connexin Disorders: Mind the Gap Junction. G. Richard
3. Invited lecture. Cell Biology Interest Group Meeting, Northwestern University, Chicago. May 26, 2004. Connexin mutations and their functional consequences in ectodermal epithelia. G. Richard
4. Invited speaker for the Stanley lecture. National Institutes of Health/NCI. Epithelial Connexin Disorders-Mind the Gap. G. Richard
5. Philadelphia Genetics Group, January 18, 2005. Skin calcification disorders-The genetics of FTC. D. Shurman, O. Topaz, E. Sprecher, G. Richard
6. Guest speaker. ARO symposium "Recent advances in understanding the function of gap junctions", New Orleans, Feb 21, 2005. Connexin Disorders of the Skin – Clinic, Genetics, and Pathogenesis. G. Richard

## **GABRIELE RICHARD, MD**

*Associate Professor*

*Department of Dermatology and Cutaneous Biology*

7. Annual Meeting of the American Academy of Dermatology. February 2005. Co-organizer and Faculty, Forum 'Impact of Molecular Genetics on Clinical Dermatology and Dermatopathology'. Molecular Genetics in Dermatology. G. Richard.

### **Abstracts and poster presentations**

1. 54th Annual Meeting of The American Society of Human Genetics, October 26-30, 2004. Familial tumoral calcinosis is an O-glycosylation disorder due to loss of function mutations of GALNT3 and is genetically heterogeneous. G. Richard, O. Topaz, R. Bergman, D.L. Shurman, M. Indelman, R. Ratajczak, M. Mizrachi, Z. Khamaysi, D. Behar, D. Petronius, V. Friedman, I. Zelikovic, S. Raimer, A. Metzker, E. Sprecher.
2. 5th Molecular Biology of Hearing and Deafness meeting, Bethesda, MD, September 30-October 3, 2004. In vitro functional analysis of a connexin 26 protein bearing an F142L amino acid substitution. HY Tang, CW Brown, G Richard, RL Alford.
3. 66th Annual Meeting of The Society for Investigative Dermatology, 2005. Mutation analysis in a cohort of Israeli patients with dystrophic epidermolysis bullosa. J. Abu Sa'd, M. Indelman, E. Pfindner, G. Richard, J. Uitto, R. Bergman, M. Kanaan, E. Sprecher.
4. 66th Annual Meeting of The Society for Investigative Dermatology, 2005. A common, recurrent mutation in *GALNT3* demonstrates that hyperphosphatemic familial tumoral calcinosis and hyperostosis-hyperphosphatemia syndrome are allelic disorders. Topaz O, Frishberg Y, Bergman R, Behar D, Fisher D, Derek Gordon D, Richard G, Sprecher E.
5. 66th Annual Meeting of The Society for Investigative Dermatology, 2005. Erythrodermic peeling skin syndrome is a variant of Netherton syndrome due to detrimental mutations in *SPINK5*. P. Ratajczak, A. S. Geyer, R. Silverman, M. Pol-Rodriguez, W. S. Millar, M. Garzon, J. Uitto<sup>1</sup>, G. Richard.
6. 66th Annual Meeting of The Society for Investigative Dermatology, 2005. Aberrant expression of gap junction proteins in a SCC tumor of a patient with Keratitis-Ichthyosis-Deafness syndrome. DL Shurman, P Benedetto, CP Hybarger, IFrieden, JJ Uitto, G Richard.
7. 66th Annual Meeting of The Society for Investigative Dermatology, 2005. Molecular basis of epidermolysis bullosa simplex with mottled pigmentation in a large autosomal dominant pedigree. JM Losi-Sasaki, D Shurman, RE Grimwood, EH Tichy, J Uitto, and G Richard.

**GABRIELE RICHARD, MD**

*Associate Professor*

*Department of Dermatology and Cutaneous Biology*

**Departmental Committees**

Co-Director, Molecular Diagnostic Laboratory

Member of the Organizing Committee for the 12th International Symposium on Basement Membranes, Philadelphia, June 15 - 18, 2005

**Professional organizations.**

American Society of Human Genetics

Society of Investigative Dermatology

Association for Research in Vision and Ophthalmology

German Society of Dermatology

Society of Pediatric Dermatology

Philadelphia Dermatological Society

Medical Advisory Board, FIRST

## FRANZISKA RINGPFEIL, MD

*Assistant Professor of Dermatology and Cutaneous Biology*

### Publications

1. Ringpfeil F. Selected Disorders of Connective Tissue: Pseudoxanthoma Elasticum, Cutis Laxa and Lipoid Proteinosis. *Clin Dermatol* 23: 41-46, 2005.
2. Ringpfeil F, McGuigan K, Kozic H, Lebwohl M, Larralde M, Uitto J. Pseudoxanthoma Elasticum is a Recessive Disease characterized by Compound Heterozygosity. (submitted)

### Book Chapters

1. Ringpfeil F, Uitto J. Pseudoxanthoma Elasticum. In: *Encyclopedia of Diagnostic Genomics and Proteomics*. Fuchs J, Podda M, Marcel Dekker, pp.1095-98, New York, NY, 2004
2. Uitto J, Ringpfeil F. Heritable Disorders of Connective Tissue. In: *Principles of Molecular Medicine* (2<sup>nd</sup> Edition). Diaz LA, Goldsmith LA, The Humana Press, Totowa, NJ (in press)
3. Uitto J, Ringpfeil F. Heritable diseases affecting the elastic tissues: cutis laxa, pseudoxanthoma elasticum and related disorders. In: *Emery and Rimoin's principles and practice of medical genetics*. Fifth edition. Elsevier, London (in press)

### Abstracts

1. Ang G, Webster G, Ringpfeil F, Campanelli C, Lee JB. "Numerous sebaceous neoplasms in a patient without visceral malignancies." American Society of Dermatopathology 41st meeting. Boston, MA. October 2004
2. LaRusso J, Fuchsel L, Uitto J, Ringpfeil F. Improved mutation screening in pseudoxanthoma elasticum based on ethnicity. Society for Investigative Dermatology, St Louis, MO, May 4-7, 2005
3. Pramick M, Fuchsel L, Uitto J, Ringpfeil F. Role of coding polymorphisms in the ABCC6 gene for cardiovascular symptoms in pseudoxanthoma elasticum. Society for Investigative Dermatology, St Louis, MO, May 4-7, 2005

### Grant Support

NIH, R01: Biochemistry and Morphology of Connective Tissue; Co-Investigator

### Participation in Scientific/ Clinical Meetings and Presentations

1. Ringpfeil F. Molecular Basis of Pseudoxanthoma Elasticum. Grand Rounds of the Department of Dermatology, Ludwig Maximillians University, Munich, Germany, June 2<sup>nd</sup>, 2004 (invited speaker)
2. Society for Pediatric Dermatology, 17<sup>th</sup> Annual Pre-AAD Meeting, New Orleans, LA, February 17, 2005
3. American Academy of Dermatology, 63<sup>rd</sup> Annual Meeting, New Orleans, LA, February 18-22, 2005

**FRANZISKA RINGPFEIL, MD**

*Assistant Professor of Dermatology and Cutaneous Biology*

**University Committees**

JUP Advisory Council

**Membership in Professional Organizations**

- American Academy of Dermatology
- Dermatology Foundation, Vice Chair, Pennsylvania
- Society for Investigative Dermatology
- Society for Pediatric Dermatology
- Philadelphia Dermatological Society
- Medical Advisory Board, NAPE (National Association for Pseudoxanthoma elasticum)
- Airline Ambassadors International

## **ULRICH RODECK, MD, PhD**

*Professor of Dermatology and Cutaneous Biology*

### **PUBLICATIONS**

Dicker, A.P, and Rodeck, U. 2004. Targeting the EGFR in neoplasia – More questions than answers. *Int. J. Rad. Onc. Biol. Phys.* 58: 899-902.

Rodeck, U. 2004. Extracellular matrix composition modulates cell cycle checkpoint control – A new twist to radiation sensitivity. *Cancer Biology and Therapy* 3:54.

Jost, M., and Rodeck, U. 2004. Keratinocyte culture in the absence of substrate attachment. In: *Methods in Molecular Biology* Vol. 289:23-28. (K. Turksen, ed.). Humana Press,

Dicker A.P., and Rodeck, U. 2005. Predicting the future from trials of the past: epidermal growth factor receptor expression and outcome of fractionated radiation therapy trials. *Journal of Clinical Oncology.* 23:5437-9.

Quadros, M.R., Peruzzi, F., Kari, C., and Rodeck, U. 2004. Complex regulation of signal transducer and activator of transcription 3 activation in normal and malignant keratinocytes. 2004. *Cancer Res.* 64:3934-3939.

McAleer, M.F., Rodeck, U., Dicker, A. 2004. Novel use of zebrafish as a vertebrate model to screen radiation protectors and sensitizers. 2004 *International Journal of Radiation Oncology, Biology, Physics* 61:10-13, 2005.

Carbone, A., Mauri, F.A., Sozzi, M., Gaspari, F., Smirne, C., Prati, A., Addeo, A., Novarino, A., Robecchi, A., Berretto, O., Emanuelli, G., Rodeck, U., and Bellone, G. 2005. Human pancreatic carcinoma cells secrete bioactive Interleukin-18 after treatment with 5-Fluorouracil: Implications for antitumor immune response. *Cancer Biology & Therapy* 4:231-241.

Duffy, K., McAleer M., Kari, L., Kari, C, Liu, C-G., Farber, S.A., Wickstrom, E., Dicker, A., and Rodeck, U. 2005. Coordinate regulation of cell cycle regulatory genes in zebrafish embryos – functional consequences of cyclinD knockdown. *Nucleic Acids Res.* 33:4914-21.

Quadros, M.R., Connelly, S., Kari, C., Abrams, M.T., Wickstrom, E, and Rodeck, U. 2005. EGFR-dependent downregulation of Bim in epithelial cells requires MAPK and PKC-.

Quadros, M.R., Peruzzi, F., Kari, C., and Rodeck, U.: Complex regulation of signal transducer and activator of transcription 3 activation in normal and malignant keratinocytes. 2004. *Cancer Res.* 64:3934-3939, 2004. (submitted).



## **ULRICH RODECK, MD, PhD**

*Professor of Dermatology and Cutaneous Biology*

Ren, Q., Kari, C., Quadros, M.R., Dicker, A.P., and U. 2005. Integration of pathways to cell survival in epidermal keratinocytes – an emergent role for TNF- $\alpha$ /NF $\kappa$ B signaling. (submitted)

Bellone, G., Anna Carbone, A., Novarino, A., Smirne, C., Stacchini, A., Bertetto, O., Robecchi, A., Emanuelli, G., and Rodeck U. Effects of tumor-derived cytokines on the polarization of dendritic cells in pancreatic carcinoma (submitted).

Davidson, W., Kari, G., Kashi, O., Daroczi, B., McAleer, M.-F., Duffy, K., Wickstrom, E., Dicker, A.P. and Rodeck, U., Divergent effects of p53 and p73 on radiosensitivity of zebrafish embryos. (submitted).

Kari, G., Davidson, W., McAleer, M.-F., Rodeck, U. and Dicker, A.P. Off-target effects of Pifithrin-alpha related to inhibition of p73 function (submitted).

### **Scientific meetings**

Inaugural meeting “RTK systems Biology”, Yokohama, Japan  
Presentation: ‘Integration of signaling pathways in complex environments’ U. Rodeck

### **Honors/Special Recognitions**

Founding member “RTK Systems Biology”. February 2005

### **Departmental Committees**

Departmental Review Committee, Dept. of Dermatology & Cutaneous Biology

### **University Committees**

Chairman, Library Committee, Thomas Jefferson University (2004-05)

Member, Ad Hoc Cancer Center Review Committee, Thomas Jefferson University (Summer 2004)

Member, Task force ‘Bone and Extracellular Matrix Biology’ (2005)

Member, Task Force ‘Cancer Biology and Research’ (2005)

Member, Committee on Research (2005)

Ad hoc member, Strategic Research Oversight Committee (2005)

**ULRICH RODECK, MD, PhD**

*Professor of Dermatology and Cutaneous Biology*

**Professional Organizations**

Memberships in Professional and Scientific Societies:

American Association of Cancer Research (1988-)  
Tellers Committee of the AACR (1994-95)  
American Association for the Advancement of Science (1989-)  
German Society of Dermatology (1990-)  
German Society of Immunology (1991-)  
German Society of Hematology and Oncology (1991-)  
Philadelphia Cancer Research Association (1993-)

**ARADHNA SAXENA, M.D.**

*Resident, Department of Dermatology and Cutaneous Biology*

**PUBLICATIONS:**

Saxena A, Lee JB, Humphreys TR. Mohs Micrographic Surgery for Squamous Cell Carcinoma associated with Epidermolysis Bullosa. Manuscript accepted for publication in *Dermatologic Surgery*.

Kasper DA, Saxena A, Katz KA. Pyoderma gangrenosum. In: Baer AB, Holstege CP, Pines JM, Brady WJ, editors. Visual Diagnosis for the Emergency Physician. British Medical Journal Press. In press 2006.

Saxena A, Lee JB et al. Lichen Sclerosis et Atrophicus – like Graft versus Host Disease. Manuscript submitted to *Cutis* for publication, status pending

Saxena A, Campanelli CD, Kasper DA, Lee JB, Humphreys TR, Webster GF. Pigmented Squamous Cell Carcinoma Mimicking Melanoma of the Nail Matrix. Manuscript submitted to *The Journal of the American Academy of Dermatology* for publication, status pending.

**PARTICIPATION IN SCIENTIFIC/CLINICAL MEETINGS AND PRESENTATIONS**

Saxena A. Lichen Sclerosis et Atrophicus – like Graft versus Host Disease. Oral Presentation at the Gross & Microscopic Symposium, American Academy of Dermatology Annual Meeting 2/19/05

Saxena A. Mohs Micrographic Surgery for Squamous Cell Carcinoma associated with Epidermolysis Bullosa. Oral Presentation for the College of Physicians Section on Dermatology Meeting, 4/13/05.

**HONORS/SPECIAL RECOGNITIONS**

Stelwagon Award Winner, 3/05: achievement in clinically-oriented research  
The College of Physicians of Philadelphia, Section on Dermatology

ASDS Dermatologic Surgery Preceptorship Program Recipient –2005

ASDS Resident Scholarship Program award

**ARADHNA SAXENA, M.D.**

*Resident, Department of Dermatology and Cutaneous Biology*

**PROFESSIONAL ORGANIZATIONS**

Women's Dermatologic Society (WDS): 2004-present

Group/Table Facilitator at WDS Networking Reception 2/05

American Society for Dermatologic Surgery (ASDS): 2003-present

American Society for Mohs Surgery: 2005-present

American Academy of Dermatology: 2003-present

## **JOUNI UITTO, MD, PhD**

*Professor of Dermatology and Cutaneous Biology  
Professor of Biochemistry and Molecular Biology*

### **PUBLICATIONS:**

#### **Peer Reviewed Publications**

##### **Year 2004**

Odanagi, M., Kikuchi, Y., Yamazaki, T., Kaneko, T., Nakano, H., Tamai, K., **Uitto, J.**, and Hanada, K.: Transcriptional Regulation of the 230-kDa Bullous Pemphigoid Antigen Gene Expression by Interferon Regulatory Factor 1 and Interferon Regulatory Factor 2 in Normal Human Epidermal Keratinocytes. *Exp. Dermatol.* 13:773-779, 2004.

Brennan, D., Hu, Y., Kljuic, A., Choi, Y.W., Joubeh, S., Bashkin, M., Wahl, J., Fertala, A., Pulkkinen, L., **Uitto, J.**, Christiano, A.M., Panteleyev, A., and Mahoney, M.G.: Differential Structural Properties and Expression Patterns Suggest Functional Significance for Multiple Mouse Desmoglein 1 Isoforms. *Differentiation* 72:434-449. 2004.

El-Domyati, M.M., Attia, S.K., Saleh, F.Y., Ahmad, H.M., and **Uitto, J.**: Effect of Topical Tretinoin on Photoaged Facial Skin: A Histometric, Immunohistochemical and Ultrastructural Study. *J. Cosmetic Dermatol* 3:191-201, 2004.

Aho, S., Li, K., McGee, C., Ryoo, Y., Ishida-Yamamoto, A., and **Uitto, J.**, Klement, J.F.: Periplakin Gene Targeting Reveals a Constituent of the Cornified Cell Envelope Dispensable for Normal Mouse Development. *Mol. Cell. Biol.* 24:6410-6418, 2004.

Schara, U., Tücke, J., Mortier, W., Nüßlein, T., Rouan, F., Pfendner, E., Zillikens, D., Bruckner-Tuderman, L., **Uitto, J.**, Wiche, G., and Schröder, R.: Severe Mucous Membrane Involvement in Epidermolysis Bullosa Simplex with Muscular Dystrophy Due to a Novel Plectin Gene Mutation. *Eur. J. Ped.* 163:218-222, 2004.

El-Domyati, M.B.M., Attia, S.K., Saleh, F.Y., Ahmad, H.M., and **Uitto, J.**: Trichloroacetic Acid Peeling Versus Dermabrasion: A Histometric, Immunohistochemical and Ultrastructural Comparison. *Dermatol. Surgery* 30:179-188, 2004.

**JOUNI UITTO, MD, PhD**

*Professor of Dermatology and Cutaneous Biology*

*Professor of Biochemistry and Molecular Biology*

**PUBLICATIONS:**

**Peer Reviewed Publications**

**Year 2004** (continued)

Metz, B.J., Ruggeri, S.Y., Hsu, S., Reed, J.A., Ghohestani, A.S., **Uitto, J.**, and Ghohestani, R.F.: Linear IgA Dermatitis with IgA and IgG Autoantibodies to the 180 kDa Bullous Pemphigoid Antigen (BP180): Evidence for a Distinct Subtype. *Int. J. Derm.* 43:443-446, 2004.

**Year 2005**

Matsuzaki, Y., Nakano, A., Jiang, Q.-J., Pulkkinen, L., and **Uitto, J.**: Tissue-Specific Expression of the ABCC6 Gene. *J. Invest. Dermatol.* (in press).

Fujimoto, N., Terlizzi, J., Brittingham, R., Fertala, A., McGrath, J.A., and **Uitto, J.**: Extracellular Matrix Protein 1 Interacts with the Domain III of Fibulin-1C and 1D Variants Through its Central Tandem Repeat 2. *Biochem. Biophys. Res. Commun.* 333:1327-1333, 2005.

Klement, J.F., Matsuzaki, Jiang, Q.-J., Y., Terlizzi, J., Choi, H.Y., Fujimoto, N., Li, K., Pulkkinen, L., Birk, D.E., Sundberg, J.P., and **Uitto, J.**: Targeted Ablation of the ABCC6 Gene Results in Ectopic Mineralization of Connective Tissues. *Mol. Cell Biol.* (in press).

Hata, D., Miyazaki, M., Seto, S., Furusho, K., Kadota, E., Nakano, A., Tamai, K., **Uitto, J.**, Moriyama, K., and Miyazaki, K.: Nephrotic Syndrome and Aberrant Expression of Laminin Isoforms in Glomerular Basement Membranes in an Infant with Herlitz Junctional Epidermolysis Bullosa. *Pediatrics* (in press).

Pfendner, E.G., Sadowski, S.G., and **Uitto, J.**: Epidermolysis Bullosa Simplex: Recurrent and *De Novo* Mutations in the KRT5 and KRT14 Genes, Phenotype/Genotype Correlations, and Implications for Genetic Counseling and Prenatal Diagnosis. *J. Invest. Dermatol.* (in press).

## **JOUNI UITTO, MD, PhD**

*Professor of Dermatology and Cutaneous Biology*

*Professor of Biochemistry and Molecular Biology*

### **PUBLICATIONS:**

#### **Peer Reviewed Publications**

##### **Year 2005** (continued)

Aumailley, M., Bruckner-Tuderman, L., Carter, W.G., Deutzmann, R., Edgar, D., Ekblom, P., Engel, J., Engvall, E., Hohenester, E., Jones, J.C.R., Kleinman, H.K., Marinkovich, M.P., Martin, G.R., Mayer, U., Meneguzzi, G., Miner, J.H., Miyazaki, K., Patarroyo, M., Paulsson, M., Quaranta, V., Sanes, J., Sasaki, T., Sekiguchi, K., Sorokin, L.M., Talts, J.F., Tryggvason, K., **Uitto, J.**, Virtanen, I., von der Mark, K., Wewer, U.M., Yamada, Y., and Yurchenco, P.: A Simplified Laminin Nomenclature. *Matrix Biol.* (in press).

Pfendner, E., and **Uitto, J.**: Plectin Gene Mutations Can Cause Epidermolysis Bullosa with Pyloric Atresia. *J. Invest. Dermatol.* 124:111-115, 2005.

Kalinin, A.E., Kalinin, A.E., Aho, M., **Uitto, J.**, and Aho, S.: Breaking the Connection: Caspase 6 Cleaves Periplakin Releasing the Intermediate Filament Binding Tail Region from the Actin Binding N-Terminal Domain. *J. Invest. Dermatol.* 124:48-55, 2005.

Brittingham, R., Colombo, M., Ito, H., Steplewski, A., Birk, D.E., **Uitto, J.**, Fertala, A.: Single Amino Acid Substitutions in Procollagen VII Affect Early Stages of Assembly of Anchoring Fibrils. *J. Biol. Chem.* 280:191-198, 2005.

Takahashi, Y., Rouan, F., **Uitto, J.**, Ishida-Yamamoto, A., Iizuka, H., Owaribe Tanigawa, M., Ishii, N., Yasumoto, S., Hashimoto, T.: Plectin Deficient Epidermolysis Bullosa Simplex with 27-Year-History of Muscular Dystrophy. *J. Dermatol. Sci.* 37:87-93, 2005.

### **SUBMITTED PUBLICATIONS:**

Nakano, H., Mauviel, A., Iozzo, R., and **Uitto, J.**: Interferon- $\gamma$  Counteracts the Transcriptional Upregulation of Type VII Collagen Gene Expression Elicited by Transforming Growth Factor- $\beta$  Evidence for Involvement of Stat-1 but not the Smad Pathway in Human Epidermal Keratinocytes.

## **JOUNI UITTO, MD, PhD**

*Professor of Dermatology and Cutaneous Biology*

*Professor of Biochemistry and Molecular Biology*

### **SUBMITTED PUBLICATIONS:**

*(continued)*

Kaneko, T., Tamai, K., Matsuzaki, Y., Yamazaki, T., Nakano, H., Kon, A., Hashimoto, I., Hanada, K., Kaneda, Y., and **Uitto, J.**: Interferon- $\gamma$  Downregulates Expression of the 230-kD Bullous Pemphigoid Antigen Gene (BPAG1) via Novel Chimeric Sequences of ISRE and GAS in Epidermal Keratinocytes.

Jiang, Q-J., Mtsuzaki, Y., Li, K., and **Uitto, J.**: Transcriptional Regulation and Characterization of the Promoter Region of the Human ABCC6 Gene.

Fujimoto, N., Terlizzi, J., Aho, S., Brittingham, R., Fertala, A., Oyama, N., McGrath, J.A., and **Uitto, J.**: Extracellular Matrix Protein 1 Inhibits the Activity of Matrix Metalloproteinase 9 Through High-Affinity Protein/Protein Interactions.

Ringpfeil, F., McGuigan, K., Fuchsel, L., Kozic, H., Larralde, M., Lebwohl, M., and **Uitto, J.**: Pseudoxanthoma Elasticum is a Recessive Disease Characterized by Compound Heterozygosity.

### **CHAPTERS IN BOOKS, REVIEW ARTICLES AND EDITORIALS**

#### **Year 2004**

**Uitto, J.**, and Richard, G.: Progress in Epidermolysis Bullosa: Genetic Classification and Clinical Implications. In: Genetic Disorders of the Skin. *Am. J. Med. Genet. Seminar.* 131C:61-74, 2004.

Lanschuetzer, C.M., Klausegger, A., Pohla-Gubo, G., Hametner, R., Richard, G., **Uitto, J.**, Hintner, H., Bauer, J.W.: Gene Symbol: KRT14 Disease: Epidermolysis Bullosa Simplex, Koebner. *Hum Genet.* 115:171.

**Uitto, J.**: Clinical Implications of Basic Research on Heritable Skin Diseases. Abstracts, The IX International Congress of Dermatology, Beijing, China, May 19-22, 2004, Abstract 04.



**JOUNI UITTO, MD, PhD**

*Professor of Dermatology and Cutaneous Biology*

*Professor of Biochemistry and Molecular Biology*

**CHAPTERS IN BOOKS, REVIEW ARTICLES AND EDITORIALS**

**Year 2004**

*(continued)*

Murrell, D.F., Turner, A., Moran, K., Pfenner, E., **Uitto, J.**, Lloyd, L., Klingberg, S.: Prenatal Diagnosis of Epidermolysis Bullosa in Australia. Abstracts of the Australasian College of Dermatologists 57<sup>th</sup> Annual Meeting, Sydney, New South Wales, Australia, May 16-19, 2004.

**Uitto, J.:** Epidermolysis Bullosa: The Expanding Mutation Database. (Commentary) *J. Invest. Dermatol.* 123:xii-xii, 2004.

**Uitto, J.:** Structure & Function of the Skin. (CRS 203). The 62<sup>nd</sup> Annual Meeting of the American Academy of Dermatology, Washington, DC, February 7, 2004.

**Uitto, J.:** Pseudoxanthoma Elasticum – A Connective Tissue Disorder or a Metabolic Disease at the Genome/Environmental Interface? (Commentary). *J. Invest. Dermatol.* 122:ix-x, 2004.

Ringfeil, F., and **Uitto, J.:** Pseudoxanthoma Elasticum. Encyclopedic Reference of Genomics and Proteomics (in press).

**Year 2005**

**Uitto, J.:** The Ehlers-Danlos Syndrome – Phenotypic Spectrum and Molecular Genetics. *Eur. J. Derm.* (in press).

**Uitto, J.:** The Gene Family of ABC Transporters – Novel Mutations, New Phenotypes. *Trends Mol. Med.* (in press).

Alexeev, V. and **Uitto, J.:** DNA Pharmaceuticals for Skin Diseases. In: DNA Pharmaceuticals (Schleef, M.), Wiley-VCH, Weinheim, Germany (in press).

**Uitto, J.,** Richard, G.: Progress in Epidermolysis Bullosa: From Eponyms to Molecular Genetic Classification. *Clin. Dermatol.* 23:33-40, 2005.

## **JOUNI UITTO, MD, PhD**

*Professor of Dermatology and Cutaneous Biology*

*Professor of Biochemistry and Molecular Biology*

### **CHAPTERS IN BOOKS, REVIEW ARTICLES AND EDITORIALS**

#### **Year 2005**

*(continued)*

Lanschuetzer, C.M., Klausegger, A., Pohla-Gubo, G., Hametner, R., Richard, G., **Uitto, J.**, Hintner, H., and Bauer, J.W.: Gene Symbol: KRT14 Disease: Epidermolysis Bullosa Simplex, Koebner. *Hum. Genet.* 115:171, 2004.

Richard, G., and **Uitto, J.**: The Complexity of Genetics in Heritable Skin Diseases. *J. Invest. Dermatol.* 124:vii-viii, 2005.

Pfendner, E., Rouan, F., and **Uitto, J.**: Progress in Epidermolysis Bullosa: The Phenotypic Spectrum of Plectin Mutations. *Exp. Derm.* 14:241-249, 2005.

Jiang, Q-J. and **Uitto, J.**: Animal Models of Epidermolysis Bullosa – Targets for Gene Therapy. *J. Invest. Dermatol.* 124:xi-xiii, 2005.

### **ABSTRACTS**

#### **Year 2004**

El-Domyati, M., Attia, S., Saleh, F., Ahmad, and **Uitto, J.**: Effect of Topical Tretinoin on Photoaged Skin. Abstract, The 13<sup>th</sup> Congress of the European Academy of Dermatology and Venereology. *J. Eur. Acad. Derm. Venereol.* 18 (suppl 2), p 140, 2004.

**Uitto, J.**: Progress in Heritable Skin Diseases. The 13<sup>th</sup> Congress of the European Academy of Dermatology and Venereology. *J. Eur. Acad. Derm. Venereol.* 18 (suppl 2), p 37, 2004.

Ghahestani, A.S., Nikbakht, N., Fry, L., Reunala, T., **Uitto, J.**, and Ghohestani, R.: Circulating IgA Autoantibodies to Epidermal Transglutaminase 3 Can Not Distinguish Dermatitis Herpetiformis from Celiac Disease. *J. Invest. Dermatol.* 122: Abstract 081, 2004.

## **JOUNI UITTO, MD, PhD**

*Professor of Dermatology and Cutaneous Biology*

*Professor of Biochemistry and Molecular Biology*

### **ABSTRACTS**

#### **Year 2004**

*(continued)*

Nguyen, P., Bazzi, H., Kljuic, A., Vargas, J.H., Dmochowski, M., Barria, M., Ghohestani, R.F., Chan, L.S., **Uitto, J.**, Diaz, L.A., Christiano, A.M., Mahoney, M.G., and Nguyen, V.T.: Identification of a Novel Population of Pathogenic Autoantibodies Against Desmoglein 4 in Different Major Variants of Pemphigus. *J. Invest. Dermatol.* 122: Abstract 085, 2004.

Fujimoto, N., Terlizzi, J., Li, K., Aho, S., and **Uitto, J.**: Assembly of Elastic Fibers: Elastin and MFAP3 Interactions Determined by Yeast Two-Hybrid Genetic System: *J. Invest. Dermatol.* 122: Abstract 215, 2004.

Brennan, D.M., Hu, Y., Kljuic, A., **Uitto, J.**, Panteleyev, A., Christiano, A.M., Mahoney, M.G.: Complexity of Dsg1- $\alpha$ , - $\beta$ , and - $\gamma$  Expression in Diverse Mouse Tissues and During Hair Follicle Formation. *J. Invest. Dermatol.* 122: Abstract 222, 2004.

Terlizzi, J., Li, K., Aho, S., Fujimoto, N., Oyama, N., Hamada, T., Chan, I., McGrath, J.A., and **Uitto, J.**: Characterization of ECM-1 Protein Interactions by Yeast Two-Hybrid System. *J. Invest. Dermatol.* 122: Abstract 225, 2004.

Kalinin, A.E., Kalinin, A.E., Aho, M., **Uitto, J.**, and Aho, S.: Breaking the Connection: Caspase 6 Cleaves Periplakin Releasing the Intermediate Filament Binding Tail Region from the Actin Binding N-terminal Domain. *J. Invest. Dermatol.* 122: Abstract 437, 2004.

Richard, G., Ratajczak, P.A., Amin, S., Ilyas, H., Tesfaye Kedjela, A., Siegfried, E.C., and **Uitto, J.**: Netherton Syndrome: Novel and Recurrent Mutations in SPINK5 and Implications for Screening and Diagnosis. *J. Invest. Dermatol.* 122: Abstract 483, 2004.

Sprecher, E., Abu Sa'd, J., Pfindner, E., Indelman, M., Ciubutaro, D., Mizrachi, M., Lestringant, G., Pulkkinen, L., Richard, G., Kanaan, M., **Uitto, J.**, and Bergman, R.: Molecular Epidemiology of Epidermolysis Bullosa in Middle East Populations. *J. Invest. Dermatol.* 122: Abstract 493, 2004.

**JOUNI UITTO, MD, PhD**

*Professor of Dermatology and Cutaneous Biology*

*Professor of Biochemistry and Molecular Biology*

**ABSTRACTS**

**Year 2004**

*(continued)*

**Uitto, J.** and Pfindner, E.: Compound Heterozygosity for Unique In-Frame Insertion and Deletion Mutations in the Plectin Gene in a Mild Case of Epidermolysis Bullosa with Very Late Onset Muscular Dystrophy. *J. Invest. Dermatol.* 122: Abstract 513, 2004.

Pfindner, E. and **Uitto, J.:** Novel and Recurrent Mutations in the Keratin 5 and 14 Genes in Epidermolysis Bullosa Simplex: Implications for Genetic Counseling. *J. Invest. Dermatol.* 122: Abstract 516, 2004.

Fratta, S., Ringpfeil, F., Terry, S., Terry, P., **Uitto, J.** and Pfindner, E.: dHPLC Screening Detects Novel and Recurrent Mutations in Pseudoxanthoma Elasticum. *J. Invest. Dermatol.* 122: Abstract 517, 2004.

Jiang, Q., Matsuzaki, Y., Li, K. and **Uitto, J.:** Transcriptional Regulation of ABCC6 Gene Expression: Identification of Novel *Cis*-Elements, Role of Sp1, and Cytokine Modulation of the Promoter Activity. *J. Invest. Dermatol.* 122: Abstract 522, 2004.

Colombo, M., Brittingham, R.J., Klement, J., Birk, D.E., **Uitto, J.**, and Fertala, A.: Procollagen VII Self-Assembly and its Perturbations in Dystrophic Epidermolysis bullosa. *J. Invest. Dermatol.* 122: Abstract 525, 2004.

Ringpfeil, F., McGuigan, K., Fuchsel, L., Kozic, H., Lebwohl, M., and **Uitto, J.:** Autosomal Dominant Inheritance in Pseudoxanthoma Elasticum Revisited. *J. Invest. Dermatol.* 122: Abstract 551, 2004.

Fuchsel, L., Kozic, H., McGuigan, K., Skvarka, C., Jacobson, M., **Uitto, J.** and Ringpfeil, F.: Genotype-Phenotype Correlation in 62 Patients with Pseudoxanthoma Elasticum. *J. Invest. Dermatol.* 122: Abstract 554, 2004.

Ringpfeil, F., McGuigan, K., Fuchsel, L., Kozic, H., Lebwohl, M., Larralde, M., and **Uitto, J.:** Autosomal Dominant Inheritance in Pseudoxanthoma Elasticum – Debunking the Myth. Abstract, International Research Meeting on Pseudoxanthoma Elasticum, October 14 & 15, 2004, Bethesda, MD, p. 5.

## **JOUNI UITTO, MD, PhD**

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### **ABSTRACTS**

#### **Year 2004**

*(continued)*

Pfendner, El, Fratta, S., and **Uitto, J.**: Standardization of a Mutation Detection Strategy for ABCC6. Abstract, International Research Meeting on Pseudoxanthoma Elasticum, October 14 & 15, 2004, Bethesda, MD, p. 6.

Klement, J.F., Matsuzaki, Y., Terlizzi, J., Jiang, Q-J., Choic H.Y., Li, K., Pulkkinen, L., Birk, D.E., Sundberg, J.P., and **Uitto, J.**: A Mouse Model for Pseudoxanthoma Elasticum: Targeted Ablation for ABCC6 Results in Aberrant Calcification of Connective Tissues. Abstract, International Research Meeting on Pseudoxanthoma Elasticum, October 14 & 15, 2004, Bethesda, MD, p. 11.

Terry, S., Hariri, S., Fernandez, J., Fratta, S., Pfendner, E., Ramsey, M., Couke, P., McCloskey, J., Viljoen, D., DePaepe, A., **Uitto, J.**, Bercovitch, L., and Terry, P.: Genotype/Phenotype Correlations. Abstract, International Research Meeting on Pseudoxanthoma Elasticum, October 14 & 15, 2004, Bethesda, MD, p. 13.

#### **Year 2005**

**Uitto, J.**: Collagen and Elastin and their Alterations in Acquired and Inherited Skin Diseases. Abstracts, Course 203: Structure and Function of the Skin, The 63<sup>rd</sup> Annual Meeting of the American Academy of Dermatology, New Orleans, Louisiana, February 18-22, 2005.

Katsuto, Y., Ogura, S., Amano, S., Klement, J., and **Uitto, J.**: Effect of tropoelastin overexpression on elastic fiber accumulation *in vitro*. *J. Invest. Dermatol.* 124: Abstract 195, 2005.

Aho, S., Kalinin, A., Aho, M., Casciola-Rosen, L., Rosen, A., and **Uitto, J.**: Specific cleavage of periplakin by granzyme B. *J. Invest. Dermatol.* 124: Abstract 409, 2005

Brennan, D., Hu, Y., Joubeh, S., Choi, Y., Whitaker-Menezes, D., **Uitto, J.**, and Mahoney, M.: Suprabasal desmoglein 2 expression enhances epidermal proliferation and alters differentiation. *J. Invest. Dermatol.* 124: Abstract 426.

Abu Saed, J., Indelman, M., Pfendner, E., Richard, G., **Uitto, J.**, Bergman, R., Kanaan, M., and Sprecher, E.: Mutation analysis and molecular epidemiology of dystrophic epidermolysis bullosa in an Israeli cohort. *J. Invest. Dermatol.* 124: Abstract 443, 2005.

## **JOUNI UITTO, MD, PhD**

*Professor of Dermatology and Cutaneous Biology*

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### **ABSTRACTS**

#### **Year 2005**

*(continued)*

Ratajczak, P.A., Geyer, A.S., Silverman, R., Pol-Rodriguez, M., Millar, W.S., Garzon, M., **Uitto, J.**, and Richard, G.: Erythrodermic peeling skin syndrome is a variant of Netherton syndrome due to detrimental mutations in *SPINK5*. *J. Invest. Dermatol.* 124: Abstract 446, 2005.

Losi-Sasaki, J.M., Shurman, D.L., Grimwood, R.E., Tichy, E.H., **Uitto, J.**, and Richard, G.: Molecular basis of epidermolysis bullosa simplex with mottled pigmentation in a large autosomal dominant pedigree. *J. Invest. Dermatol.* 124: Abstract 468, 2005.

Pramick, M., Fuchsel, L., **Uitto, J.**, and Ringpfeil, F.: Role of coding polymorphisms in the *ABCC6* gene for cardiovascular symptoms in pseudoxanthoma elasticum. *J. Invest. Dermatol.* 124: Abstract 481, 2005.

Shurman, D., Benedetto, P., Hybarger, C.P., Frieden, I., **Uitto, J.**, and Richard, G.: Aberrant expression of gap junction proteins in a SCC tumor of a patient with keratitis-ichthyosis-deafness syndrome. *J. Invest. Dermatol.* 124: Abstract 482, 2005.

Jiang, Q., Matsuzaki, Y., Pulkkinen, L., and **Uitto, J.**: Tissue-specific expression of the *ABCC6* gene. *J. Invest. Dermatol.* 124: Abstract 494, 2005.

**Uitto, J.**, Matsuzaki, Y., Terlizzi, J., Jiang, Q., Choi, H., Li, K., Pulkkinen, L., Birk, D., Sundberg, A., and Klement, J.: A mouse model for pseudoxanthoma elasticum: targeted ablation of *ABCC6* results in aberrant calcification of connective tissues. *J. Invest. Dermatol.* 124: Abstract 499, 2005.

Fujimoto, N., Terlizzi, J., Aho, S., Oyama, N., Chan, I., McGrath, J., and **Uitto, J.**: Extracellular matrix protein 1 (ECM1) inhibits the activity of matrix metalloproteinase 9 (MMP9) through protein/protein interactions: relevance to pathogenesis of lipoid proteinosis. *J. Invest. Dermatol.* 124: Abstract 500, 2006.

LaRusso, J., Fuchsel, L., **Uitto, J.**, and Ringpfeil, F.: Improved mutation screening in pseudoxanthoma elasticum based on ethnicity. *J. Invest. Dermatol.* 124: Abstract 501, 2005.

Pfendner, E., Rouan, F., and **Uitto, J.**: The phenotypic spectrum of plectin mutations in epidermolysis bullosa. *J. Invest. Dermatol.* 124: Abstract 509, 2005.

**JOUNI UITTO, MD, PhD**

*Professor of Dermatology and Cutaneous Biology*

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**ABSTRACTS**

**Year 2005**

*(continued)*

Varki, R., Pfindner, E., and **Uitto, J.**: The spectrum of COL7A1 gene mutations in dystrophic epidermolysis bullosa. *J. Invest. Dermatol.* 124: Abstract 510, 2005.

**VISITING PROFESSORSHIPS AND INVITED LECTURESHIPS:**

**Year 2004**

Grand Round Speaker, Department of Dermatology, Johns Hopkins University School of Medicine, Baltimore, Maryland, June 9, 2004.

Invited Speaker, Third European Symposium, Elastin-2004, University of Manchester, Manchester, United Kingdom, June 30 – July 3, 2004.

Invited Speaker, the Child Health Institute of New Jersey, Robert Wood Johnson Medical School – University of Medicine and Dentistry of New Jersey, New Brunswick, New Jersey, October 27, 2004.

Invited Speaker, Symposium Organizer and Plenary Session Chair, The 13<sup>th</sup> Congress of the European Academy of Dermatology and Venereology – The Renaissance of Dermatology – Florence, Italy, November 17-21, 2004

**Year 2005**

Invited Speaker, Medicis Winter Skin Seminar, The University of Colorado, Snowmass, Colorado, January 29-February 3, 2005.

Procter & Gamble Scientific Lecturer and Grand Round Speaker, University of Cincinnati, Cincinnati, Ohio, February 9-10, 2005.

Invited Speaker, Structure and Function of the Skin (Course 202), The 63<sup>rd</sup> Meeting of the American Academy of Dermatology, New Orleans, Louisiana, February 18-22, 2005.

**JOUNI UITTO, MD, PhD**

*Professor of Dermatology and Cutaneous Biology*

*Professor of Biochemistry and Molecular Biology*

**VISITING PROFESSORSHIPS AND INVITED LECTURESHIPS:**

**Year 2005**

*(continued)*

Visiting Professor, Department of Medicine, Division of Dermatology, University of Texas – San Antonio, Texas, March 22 & 23, 2005.

Visiting Professor, Department of Dermatology, The University of Texas Southwestern School of Medicine, Dallas, Texas, April 25-27, 2005.

Invited Speaker, Department of Medicine, University of Southern California School of Medicine, Los Angeles, California, December 15, 2004.

Visiting Professor and Arnold W. Gurevitch Memorial Grand Round Speaker, Harbor-UCLA Medical Center, Torrance, California, December 16-17, 2004.

**AWARDS AND HONORS:**

**2005**

President and Chair of the Organizing Committee, The 12<sup>th</sup> International Symposium on Basement Membranes, to be held at Jefferson Medical College, Philadelphia, Pennsylvania, June 16-19, 2005 (appointed in the 11<sup>th</sup> Symposium, Japan, March 6-9, 2003).

The 19<sup>th</sup> James N. Gilliam Lecturer, Department of Dermatology, The University of Texas Southwestern Medical Center at Dallas, and Dallas Dermatological Society, and Fort Worth Dermatological Society, Dallas, Texas, April 26, 2005.

**UNIVERSITY COMMITTEES:**

Jefferson Medical College Admissions Committee



**GUY WEBSTER, MD, PhD**

*Professor*

*Department of Dermatology and Cutaneous Biology*

**PUBLICATIONS:**

GF Webster, Mechanism-Based Treatment of Acne Vulgaris: The Value of Combination Therapy, *Journal of Drugs in Dermatology*, 2005 in press

GF Webster, G Zimmerman, M Corvest, U Mrowietz, Under-served patients with moderate or severe psoriasis: insights and management considerations. In MS

Torok HM, Webster G, Dunlap FE, Egan N, Jarratt M, Stewart D. Combination sodium sulfacetamide 10% and sulfur 5% cream with sunscreens versus metronidazole 0.75% cream for rosacea. *Cutis* 2005, 75:357-363.

GF Webster, Acne and Rosacea, *Conn's Current Therapy* 2006. in press

GF Webster, Rosacea, in *Dermatology*, J. Bologna, J. Jorizzo, R. Rapini eds, 2007, in manuscript

**INVITED TALKS:**

Visiting Professor, University of Oklahoma School of Medicine, 2004

Acne and Rosacea, Pennsylvania Academy of Family Practice, Hershey, PA 2004

Psoriasis Therapy, Update on Biologicals, University of Pennsylvania, Philadelphia, PA 2004

Update on Acne, Treatment of Rosacea, Winter Clinical Dermatology, Kona, HI, 2005

Visiting Professor, University of San Diego, San Diego, CA, 2005

Visiting Professor, Loma Linda University, Loma Linda CA, 2005

Visiting Professor, Brown University, Providence, RI, 2005

Visiting Professor, University of South Florida/West Coast Dermatological Soc, Tampa, FL, 2005

Acne Poster Session Moderator, AAD Annual Meeting, New Orleans, LA, 2005

**GUY WEBSTER, MD, PhD**

*Professor*

*Department of Dermatology and Cutaneous Biology*

**INVITED TALKS:**

*(continued)*

What's Coming in Acne and Rosacea, AAD Annual Meeting, New Orleans, LA, 2005

Update on Acne, Sacramento Valley Derm. Soc., Squaw Valley, CA, 2005

Topical Anti-inflammatory Therapy, Sacramento Valley Derm. Soc., Squaw Valley, CA,  
2005

Visiting Professor, University of Missouri Dept. Dermatology, Columbia, MO, 2005

Retinoid Modulation of Toll-like Receptors, Hawaii Dermatology Symposium, Maui,  
HI. 2005.

Visiting Professor, Baylor School of Medicine Dept Dermatology, Houston, TX, 2005

Overview of Acne Therapy; HIV related skin diseases, Valley of the Sun Symposium,  
Phoenix, AZ 2005

Acne and its Treatment, Plenary Session, AANP Annual Meeting, Ft. Lauderdale, FL,  
2005

Pathogenesis of acne, AAD '05, Chicago, IL, 2005

What's New in Acne, North Carolina Dermatology Assc, Summer Beach , NC, 2005

**KYONGGEUN YOON, PH.D**

*Professor of Dermatology and Cutaneous Biology*

**PUBLICATIONS:**

1. Igoucheva, O. and Alexeev, V. and **Yoon, K.** "Site-directed mutagenesis and targeted gene correction", *Current Molecular Medicine*, 4, 445-463, 2004.
2. **Yoon, K.** "Oligonucleotide-directed gene correction in epidermis", *J. Invest. Derm. Symp. Proc.* 9, 276-283, 2004
3. Igoucheva, O. and Alexeev, V. and **Yoon, K.** "Mechanism of gene repair open for discussion", *Oligonucleotides*. 14, 311-321, 2004.
4. Igoucheva, O. and **Yoon, K.** "Gene targeting by oligonucleotides in keratinocytes", *Epidermal Cell Protocols*, ed, Turksen, K. *Methods Mol Biol.*, Humana Press, pp287-302, 2004.
5. Abrams, M. T., Robertson, N., **Yoon, K** and Wickstrom, E., "Inhibition of glucocorticoid-induced apoptosis by targeting the major splice variants of BIM mRNA with siRNA and shRNA", *J. Biol. Chem.* 279, 55809-55817, 2004
6. Igoucheva, O., Alexeev, V. and **Yoon, K.,** "Differential cellular responses to exogenous DNA in mammalian cells and its effect on oligonucleotide-directed gene modification", accepted in *Gene Therapy*.
7. Igoucheva, O., Alexeev, V. and **Yoon, K.,** "Involvement of nucleotide excision repair in oligonucleotide-directed gene modification", submitted to *Gene Therapy*.
8. Alexeev, V. and **Yoon, K.,** "Distinctive role of the c-kit receptor tyrosine kinase signaling in mammalian melanocytes", submitted to *J. Invest Derm.*
9. Cesarone, G., Garofalo C., Abrams M., Igoucheva O., Alexeev, V., **Yoon, K.**, Surmacz, E., and Wickstrom, E. "RNAi-mediated Knockdown of Insulin Receptor Substrate 1 (IRS-1) Enhances Tamoxifen-Induced Cell Death in MCF-7 Breast Cancer Cells" submitted to *J. Biol. Chem.*
10. Dejneka, D.S., Bennicelli, J. L., Igoucheva, O., Alexeev, V., Fu, L., Dahl, J. P., Chung, D. C., Maguire, A.M., **Yoon, K.** and Bennett, J. "Gene Targeting in the Murine Retina with Adeno-Associated Virus", submitted to *Nat Biotech*

KYONGGEUN YOON, PH.D

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**PROFESSIONAL ORGANIZATIONS.**

American Society of Biological Chemistry and Molecular Biology

American Association of Advanced Science

Society of Investigative Dermatology

NIH study section, ad hoc reviewer of the Gene Therapy and Inborn Errors Study section (2005)

NIH study section, ad hoc reviewer of the Innovative Technologies for the molecular analysis of cancer (2005)