CURRICULUM VITAE

PERSONAL DATA

Name: **Ala-Kokko (nee Hämäläinen), Leena Maria**
Place of Birth: Oulu, Finland

CURRENT POSITIONS

Professor, Department of Medical Biochemistry and Molecular Biology, University of Oulu, Oulu, Finland
President, Connective Tissue Gene Tests, LLC. Allentown, PA.

EDUCATION AND BOARD CERTIFICATIONS

1980 Graduated from Oulu's Lyseo High School
1982 Bachelor of Medicine, University of Oulu, Finland
1986 M.D. Degree, University of Oulu, Finland
1987 Doctor of Medical Sciences (equivalent to Ph.D.), University of Oulu, Finland

PROFESSIONAL EXPERIENCE

1982-1987	Research Assistant, Medical Biochemistry, University of Oulu
1987-1989	Research Associate, Dept. of Biochemistry and Molecular Biology, Jefferson Institute of Molecular Medicine, Thomas Jefferson University, Philadelphia, PA
1989-1991	Instructor, Dept. of Biochemistry and Molecular Biology, Jefferson Institute of Molecular Medicine, Thomas Jefferson University, Philadelphia, PA
1990-	Docent in Medical Biochemistry, University of Oulu, Finland
1991-1992	Instructor, Medical Biochemistry, University of Oulu, Finland
1991-1996	Research Assistant Professor, Dept. of Biochemistry and Molecular Biology, Jefferson Institute of Molecular Medicine, Thomas Jefferson University, Philadelphia, PA
1996-1997	Assistant Professor/Senior Lecturer, Dept. of Medical Biochemistry, University of Oulu, Finland
1996-1999	Adjunct Associate Professor, Center for Gene Therapy, MCP Hahnemann University, Philadelphia, PA
1997-2002	Senior Research Fellow/Associate Professor (The Academy of Finland), Dept. of Medical Biochemistry, University of Oulu, Finland
1997-2000	Consultant, Fibrogen Europe, Oulu, Finland
1998-2000	Associate Professor, Center for Gene Therapy, MCP Hahnemann University, Philadelphia, PA
2000-2001	Associate Professor, Department of Medicine and Center for Gene Therapy, Tulane University Health Sciences Center, New Orleans, LA
2001-2004	Professor with tenure, Department of Medicine and Center for Gene Therapy, Tulane University Health Sciences Center, New Orleans, LA
2003-	Professor with tenure, Department of Medical Biochemistry and Molecular Biology, University of Oulu, Oulu, Finland
2004-	President, Connective Tissue Gene Tests, LLC., Allentown, PA.

SUPERVISOR OF THE DOCTORAL THESIS STUDIES

Ph.D. Thesis: Pertti Ritvaniemi (University of Oulu, Finland - 1994); Constance Yuan (Thomas Jefferson University, Philadelphia, PA, USA - 1997); Jarmo Körkkö (University of Oulu, Finland - 1998); Susanna Annunen and Petteri Paassilta (University of Oulu, Finland - 1999); Tero Pihlajamaa (University of Oulu, Finland – 2000); Jaro Karppinen co-supervisor with Professor Heikki Vanharanta (University of Oulu, Finland, 2001); Jussi Vuoristo (University of Oulu, Finland – 2002); Mirka Vuoristo (University of Oulu, Finland – 2003), Heini Hartikka (University of Oulu, Finland – 2005), Eveliina Jakkula (University of Oulu, Finland – 2005), Miia Melkoniemi (University of Oulu, Finland – 2005), Noora Noponen-Hietala (University of Oulu, Finland – 2005), Iita Daavittila (University of Oulu, Finland – 2007), Marja Majava

(University of Oulu, Finland – 2007), Juha Jäälinoja (University of Oulu, Finland – 2008), Olli-Pekka Kämäräinen (University of Oulu, Finland – 2009).

HONORS AND AWARDS

1999 The Finnish Science Award (together with Drs. Kari Kivirikko and Taina Pihlajaniemi)
1999 Young Scientist Award from the Finnish Life Insurance Companies
2001 Anders Jahre prize for young scientist

MAIN SCIENTIFIC ADVISORY FUNCTIONS

1997- Project Leader, Collagen Research Unit, University of Oulu
1998- Member of the Grants Committee, the Academy of Finland
1997- Project Leader, Biocenter Oulu
2001 Evaluator of Finnish Sports Medicine, Appointed by the Ministry of Education, Finland
Other evaluators: Dr. Bruce Beynnon (Univ. of Vermont), Dr. Hilkka Riihimäki (Finnish Institute of Occupational Health) and Dr. Bengt Saltin (University of Copenhagen).
2002- Member of the Grants Committee, the National Institute of Health

GRANTS (FINLAND)

1987-1988 The Finnish College of Physicians
1991-1992 The Finnish Cultural Fund
1992-1993 The University of Oulu, Finland
1992-1993 The Yrjö Jahnsson Foundation
1993-1995 EU, BIOMED1, Concerted Action
1997-2000 The Academy of Finland
1998-2004 Kuopio University Hospital (EVO) (PI: Heikki Helminen, M.D., Ph.D.)
1999-2000 The Finnish Work Environment Fund. (PI: Hilkka Riihimäki, M.D., Ph.D.)
1999-2007 Biocenter Oulu, Finland. (PI: Leena Ala-Kokko, M.D., Ph.D.)
2000-2005 The Academy of Finland (Independent group leader in the Collagen Research Unit that was nominated by the Academy of Finland a National Center of Excellence for 2000-2005).
2002-2005 EU, European Skeletal Dysplasia Network (ESDN).
2003-2005 EU, Cornea Engineering.

GRANTS (USA)

1999-2000 The National Marfan Foundation. PI: Leena Ala-Kokko, M.D., Ph.D.
1999-2004 NIH: 1RO1 AR45982-01A1 (Mutations causing disc disease and sciatica). PI: Leena Ala-Kokko, M.D., Ph.D.
2001-2004 Arthritis Foundation (Mutations causing osteoarthritis). PI: Leena Ala-Kokko, M.D., Ph.D.

OTHER ACADEMIC AND PROFESSIONAL MERITS AND ACTIVITIES

Found Qualified and Competent for the Professorship of Medical Biochemistry, University of Kuopio, Finland, 1997

Found Qualified and Competent for the Associate Professorship of Molecular Biochemistry, University of Oulu, Finland (2nd place of preference after Dr. R. Wierenga), 1997

Membership in the Graduate Faculty of Thomas Jefferson University, Philadelphia, PA, 1994-1996

Ad Hoc Referee for Scientific Journals, 1993- : Am. J. Hum. Genet., Am. J. Med. Genet., Arthritis and Rheumatism, Comp. Biochem. Physiol., Eur. J. Hum. Genet., Hum. Genet., Hum. Mol. Genet., Hum. Mutat., J. Biol. Chem., J. Invest. Dermatol., J. Med. Genet., J. Psychiat. Res., Matrix Biol., Proc. Natl. Acad. Sci. USA

Referee for the Doctoral Dissertation
University of Oulu, Finland, 1992, 1993, 1996, 1998, 1999, 2004
University of Kuopio, Finland, 1993
University of Turku, Finland, 1997, 2000
University of Helsinki, Finland, 1998

Official Opponent at Dissertations
University of Turku, Finland, 1997, 1998

University of Helsinki, Finland, 1998, 2000
Referee for a Docentship, University of Oulu, Finland, 1994; University of Turku, Finland, 2001

MEMBERSHIPS IN SCIENTIFIC SOCIETIES

1983- The Finnish Connective Tissue Society: Vice Chairperson 1998-2000, Chairperson 2000-2002
1983- Societas Biochemica, Biophysica et Microbiologica Fenniae
1983- The Biochemical Society of Oulu
1985- Finnish College of Physicians (Duodecim)
1996- International Society for Matrix Biology
2002- The American Society for Human Genetics

CONFERENCES

The Federation of European Connective Tissue Society, IXth Meeting, Budapest, Hungary, Poster - 1984
The Finnish Connective Tissue Society Meeting, Talk - 1985, 1986, 1999
The Federation of European Connective Tissue Societies, Xth Meeting, Manchester, England, Poster - 1986
Annual Meeting of the Society for Investigative Dermatology, Inc., Washington, DC, Talk - 1988
Second International Conference on Molecular Biology and Pathology of Matrix, Philadelphia, PA, Poster - 1988
Conference on Structure, Molecular Biology and Pathology of Collagen, Bethesda, MD, Poster - 1989
American College of Rheumatology, 53rd Annual Scientific Meeting, Cincinnati, OH, Talk - 1989
Orthopaedic Research Society, 36th Annual Meeting, New Orleans, LA, Talk - 1990
The Annual Meeting of the American Society of Clinical Investigation, Washington, DC, Talk - 1990
Third International Conference on the Molecular Biology and Pathology of Matrix, Philadelphia, PA, Poster - 1990
The American Society of Human Genetics, Cincinnati, OH, Poster - 1990
The Annual Meeting of Finnish Physical Medicine, Helsinki, Finland, Invited Talk -1992
The Federation of European Connective Tissue Societies, XIIIth Meeting, Davos, Switzerland, Talk - 1992
The American Society of Human Genetics, New Orleans, LA, Poster-1993
The Yutaka Nagai Symposium on Matrix Biology, Tokyo, Japan, Invited Talk - 1994
The Lumbar Spine. A Basic Science Approach. First International Symposium, Brussels, Belgium, Poster - 1994
Fifth International Conference on the Molecular Biology and Pathology of Matrix, Philadelphia, PA, Talk - 1994
The Finnish Arthritis Club Meeting, Helsinki, Finland, Invited Talk - 1994
Chromosome 18 DNA Markers and Manic-Depressive Illness. American College of Neuropsychopharmacology Annual Meeting Satellite, San Juan, Puerto Rico, Invited Talk - 1994
The 8th Annual Meeting of Japanese Society of Cartilage Metabolism, Tokyo, Japan, Invited Talk - 1995
Third International Chromosome 18 Workshop, Philadelphia, PA, Talk - 1995
Second Meeting of the Bone Dysplasia Society, Versailles, France, Talk - 1995
Skeletal Development of Chondrodysplasias. International Scientific Meeting of the EC-Concerted Action on Chondrodysplasias. Children's Hospital, University of Mainz, Mainz, Germany, Invited Talk - 1996
Sixth International Conference on the Molecular Biology and Pathology of Matrix, Philadelphia, PA, Invited Talk - 1996
The Federation of European Connective Tissue Societies, XVth Meeting, Munich, Germany, Invited Talk - 1996
Structure, Function and Genetic Analysis of Extracellular Matrix, Schloss Ringberg, Germany, Invited Talk - 1997
The American Society of Human Genetics, Denver, Colorado, Talk - 1998
The Clinical and Biological Basis of the Ehlers-Danlos Syndrome, Cold Spring Harbor Laboratory, New York, Invited Talk – 1999
Gordon Research Conference on Collagen, New London, NH, USA, Invited Talk – 1999
Annual Meeting of the Finnish Medical Association, Helsinki, Finland, Invited Talk –2000

The Finnish Internal Medicine Association Meeting, Levi, Finland, Invited Talk – 2000
28th Scandinavian Congress of Rheumatology, Turku, Finland, Invited Talk – 2000
3rd Workshop on Heritable Disorders of Connective Tissue: Pathogenesis of Connective Tissue Disorders, NIH, Bethesda, MD, USA, Invited Talk – 2000
6th International Symposium on the Marfan Syndrome, Seattle, USA, Invited Talk – 2001
American Academy of Orthopaedic Surgeons/National Institute of Health: Molecular Biology in Orthopaedics 2001 Workshop, Scottsdale, AZ, Invited Talk – 2001
Orthopaedic Research Society, 48th Annual Meeting, Dallas, TX, Invited Talk – 2002.
34th European Metabolic Group Meeting, Zurich, Switzerland, Invited Talk – 2002.
Segal Osteoarthritis Symposium, Chicago, Invited Talk – 2003
Gordon Research Conference on Collagen, New London, NH, Session Chair – 2003
The 6th International Society for Skeletal Dysplasia Conference, Warrenton, VA, Session chair - 2003
AOSPINE, Intervertebral Lumbar Disc Symposium. Davos, Switzerland, Key note speaker, 2005
The American Society of Human Genetics, Salt Lake City, Utah, Poster - 2005
Gordon Research Conference on Collagen, New London, NH, Session Chair – 2005
The American Society of Human Genetics, New Orleans, LA, Poster-2006
The American Society of Human Genetics, Philadelphia PA, Poster-2008
The American College of Medical Genetics, Tampa, FL, Poster - 2009
The American College of Medical Genetics, Albuquerque, NM, Poster – 2010
The American College of Medical Genetics, Vancouver, Canada, Invited Talk – 2011

INVITED SEMINARS

M.D. Anderson Hospital and Tumor Institute, Genetics Department, Houston, TX - 1990
University of Medicine and Dentistry of New Jersey, Dept. of Anatomy and Biochemistry, Newark, NJ - 1990
University of Oulu, Dept. of Physical Medicine and Rehabilitation, Oulu, Finland - 1991
University of Turku, Dept. of Medical Biochemistry, Turku, Finland - 1991
National Public Health Institute, Laboratory of Mol. Genetics, Helsinki, Finland - 1992
University of Oulu, Dept. of Dermatology, Oulu, Finland - 1992
University of Kuopio, Dept. of Obstetrics and Gynecology, Kuopio, Finland - 1992
University of Helsinki, Dept. of Medical Genetics, Helsinki, Finland – 1992
Osaka University Medical School, Dept. of Orthopaedic Surgery, Osaka, Japan - 1995
University of Kuopio, Dept. of Anatomy, Kuopio, Finland - 1997
University of Turku, Dept. of Medical Biochemistry, Turku, Finland - 1997
University of Kuopio, TULES Graduate School, Kuopio, Finland - 1997
Finnish Institute of Occupational Health, Helsinki, Finland - 1997
University of Oulu, Dept. of Physical Medicine and Rehabilitation, Oulu, Finland - 1997
University of Oulu, Dept. of Internal Medicine, Oulu, Finland - 1999
University of Helsinki. Dept .of Medical Genetics, Helsinki, Finland - 1999
Biocenter of Oulu, Oulu, Finland - 1999
University of Pennsylvania School Dental Medicine, Dept. of Biochemistry, Philadelphia, PA - 1999
Shriners Hospital for Children, Portland, OR - 1999
University of Hong Kong, Department of Biochemistry and Surgery, Hong Kong – 2000
Tulane University Health Sciences Center, Department of Biochemistry, New Orleans, LA – 2001
Tulane University Health Sciences Center, Department of Pharmacology, New Orleans, LA – 2001
University of Alabama at Birmingham, Cell Biology, Birmingham, AL – 2001
University of Helsinki, Institute of Biotechnology, Helsinki, Finland – 2001
Tulane University Health Sciences Center, Department of Genetics, New Orleans, LA – 2001
University of Kuopio, Department of Medicine, Kuopio, Finland – 2002
Karolinska Institute, Dept. of Medical Biochemistry and Biophysics, Stockholm, Sweden – 2002
Tulane University Health Sciences Center, Women's Center, New Orleans, LA - 2002
Tulane University Health Sciences Center, Department of Structural and Cellular Biology, New Orleans, LA – 2002
Tulane University Health Sciences Center, Department of Genetics, New Orleans, LA – 2003
Tulane University Health Sciences Center, Department of Clinical Immunology, New Orleans, LA – 2003

LIST OF PUBLICATIONS

Leena Ala-Kokko (Hämäläinen)

Original Papers

1. Oikarinen J, Pihlajaniemi T, Hämäläinen L, Kivirikko KI. Cortisol decreases the cellular concentration of translatable procollagen mRNA species in cultured human skin fibroblasts. ***Biochim Biophys Acta*** 741, 297-302, 1983.
2. Oikarinen J, Hämäläinen L, Oikarinen A. Modulation of glucocorticoid receptor activity by cyclic nucleotides and its implications on the regulation of human skin fibroblast growth and protein synthesis. ***Biochim Biophys Acta*** 799, 158-165, 1984.
3. Savolainen E-R, Hämäläinen L, Kivirikko KI. Hepatic type I procollagen messenger RNA levels in experimental liver fibrosis in rats. In: Hirayama C, Kivirikko KI. eds. ***Pathobiology of hepatic fibrosis***. Excerpta Medica. Amsterdam: Elsevier, 67-74, 1985.
4. Hämäläinen L, Oikarinen J, Kivirikko KI. Synthesis and degradation of type I procollagen mRNAs in cultured human skin fibroblasts and the effect of cortisol. ***J Biol Chem*** 260 720-725, l985.
5. Kuivaniemi H, Ala-Kokko L, Kivirikko KI. Secretion of lysyl oxidase by cultured human skin fibroblasts and effects of monensin, nigericin, tunicamycin and colchicine. ***Biochim Biophys Acta*** 883, 326-334, 1986.
6. Ala-Kokko L. Collagen gene expression in fibrotic human skin diseases and in rats with experimental liver fibrosis: effects of cortisol and malotilate. ***Acta Univ Oul A*** 185, 1-53, 1987.
7. Oikarinen A, Ala-Kokko L, Palatsi R, Peltonen L, Uitto J. Scleredema and paraproteinemia. Enhanced collagen production and elevated type I procollagen messenger RNA level in fibroblasts grown from cultures from the fibrotic skin of a patient. ***Arch Dermatol*** 123, 226-229, 1987.
8. Pääkkö P, Ala-Kokko L, Ryhänen L. A light microscopic and biochemical study of carbon tetrachloride-induced pulmonary fibrosis in rats: the preventive effect of malotilate. ***Eur J Clin Invest*** 17, 340-346, 1987.
9. Ala-Kokko L, Rintala A, Savolainen E-R. Collagen gene expression in keloids: Analysis of collagen metabolism and type I, III and V procollagen mRNAs in keloid tissue and keloid fibroblast cultures. ***J Invest Dermatol*** 89, 238-244, 1987.
10. Ala-Kokko L, Pihlajaniemi T, Myers JC, Kivirikko KI, Savolainen E-R. Gene expression of type I, III and IV collagens in hepatic fibrosis induced by dimethylnitrosamine in the rat. ***Biochem J*** 244, 75-79, 1987.
11. Ala-Kokko L, Stenbäck F, Ryhänen L. Preventive effect of malotilate on carbon tetrachloride-induced liver damage and collagen accumulation in the rat. ***Biochem J*** 246, 503-509, 1987.
12. Oikarinen A, Salo T, Ala-Kokko L, Tryggvason, K. Dexamethasone modulates the metabolism of type IV collagen and fibronectin in human basement-membrane-forming fibrosarcoma (HT-1080) cells. ***Biochem J*** 245, 235-241, 1987.
13. Savolainen E-R, Brocks D, Ala-Kokko L, Kivirikko KI. Serum concentrations of the N-propeptide of type III procollagen and two type IV collagen fragments and gene expression of the respective collagen types in liver in rats with dimethylnitrosamine-induced hepatic fibrosis. ***Biochem J*** 249, 753-757, 1988.
14. Stenbäck F, Ala-Kokko L, Ryhänen L. Morphological, immunohistochemical and ultrastructural changes in dimethylnitrosamine induced liver injury. Effect of malotilate. ***Histol Histopath*** 4, 95-104, 1989.
15. Ala-Kokko L, Stenbäck F, Ryhänen L. Preventive effect of malotilate on dimethylnitrosamine-induced liver fibrosis in the rat. ***J Lab Clin Med*** 113, 177-183, 1989.
16. Pääkkö P, Sormunen R, Risteli L, Risteli J, Ala-Kokko L, Ryhänen L. Malotilate prevents accumulation of type III pN-collagen, type IV collagen and laminin in carbon tetrachloride-induced pulmonary fibrosis in rats. ***Am Rev Respir Dis*** 139, 1105-1111, 1989.

17. Ala-Kokko L, Kontusaari S, Baldwin CT, Kuivaniemi H, Prockop DJ. Structure of cDNA clones coding for the entire preproα1(III) chain of human type III procollagen. Differences in protein structure from type I procollagen and conservation of codon preferences. ***Biochem J*** 260, 509-516, 1989.
18. Oikarinen A, Ala-Kokko L, Tamminen M, Karvonen J, Reunala T, Kallioinen M, Hannuksela M. Effect of long-term PUVA treatment of psoriasis on the collagen and elastin gene expression and growth of skin fibroblasts in vitro. ***British J Dermatol*** 123, 621-630, 1990.
19. Prockop DJ, Olsen A, Kontusaari S, Hyland J, Ala-Kokko L, Vasan NS, Barton E, Buck S, Harrison K, Brent RL. Mutations in human procollagen genes. Consequences of the mutations in man and in transgenic mice. ***Ann NY Acad Sci.*** 580, 330-339, 1990.
20. Zhao M, Kontusaari S, Kuivaniemi H, Tromp G, Sabol C, Ala-Kokko L, Klein SA, Ladda RL, Kousseff BG, Prockop DJ. Three single base mutations in type III procollagen that prevent correct RNA splicing in mild and severe variants of Ehlers-Danlos Syndrome IV. ***Ann NY Acad Sci*** 580, 554-555, 1990.
21. Karvonen K, Ala-Kokko L, Pihlajaniemi T, Helaakoski T, Henke S, Günzler V, Kivirikko KI, Savolainen E-R. Specific inactivation of prolyl 4-hydroxylase and inhibition of collagen synthesis by oxaproline-containing peptides in cultured human skin fibroblasts. ***J Biol Chem*** 265, 8415-8419, 1990.
22. Ala-Kokko L, Prockop DJ. Completion of the intron-exon structure of the gene for human type II procollagen (COL2A1). Variations in the nucleotide sequences of the alleles from three chromosomes. ***Genomics*** 8, 454-460, 1990.
23. Ala-Kokko L, Prockop DJ. Efficient procedure for preparing cosmid libraries from microgram quantities of genomic DNA fragments size fractionated by gel electrophoresis. ***Matrix. Coll Rel Res*** 10, 279-284, 1990.
24. Ala-Kokko L, Baldwin CT, Moskowitz RW, Prockop DJ. Single-base mutation in the type II procollagen gene (COL2A1) as a cause of primary osteoarthritis associated with a mild chondrodysplasia. ***Proc Natl Acad Sci USA*** 87, 6565-6568, 1990.
25. Ahmad NN, Ala-Kokko L, Knowlton RG, Jimenez SA, Weaver EJ, Maguire JT, Tasman W, Prockop DJ. Stop codon in the procollagen II gene as a cause of retinal detachment and arthro-ophthalmopathy (Stickler syndrome). ***Proc Natl Acad Sci USA*** 88, 6624-6627, 1991.
26. Vandenberg P, Khillan JS, Prockop DJ, Helminen H, Kontusaari S, Ala-Kokko L. Expression of a partially deleted gene of human type II procollagen (COL2A1) in transgenic mice produces a phenotype of a chondrodysplasia. ***Proc Natl Acad Sci USA*** 88, 7640-7644, 1991.
27. Ala-Kokko L, Hyland J, Smith C, Kivirikko K, Jimenez SA, Prockop DJ. Expression of a human cartilage procollagen gene (COL2A1) in mouse 3T3 cells. ***J Biol Chem*** 266, 14175-14178, 1991.
28. Jimenez S, Ala-Kokko L, Ahmad N, Baldwin C, Dharmavaram R, Reginato A, Knowlton R, Prockop DJ. Type II collagen gene mutations in familial osteoarthritis. In: Kuettner K et al, eds. ***Articular Cartilage and Osteoarthritis***. Raven Press, Ltd, New York, 167-178, 1992.
29. Vikkula M, Metsäranta M, Syvänen A-C, Ala-Kokko L, Vuorio E, Peltonen L. Structural analysis of the regulatory elements of the type II collagen gene: Conservation of promoter and first intron sequences between human and mouse. ***Biochem J*** 285, 287-294, 1992.
30. Ala-Kokko L, Günzler V, Hoek JB, Rubin E, Prockop DJ. Hepatic fibrosis in rats produced by carbon tetrachloride and dimethylnitrosamine. Observations suggesting immunoassays of serum for the 7S fragment of type IV collagen are a more sensitive index of liver damage than immunoassays for the NH_2-terminal propeptide of type III procollagen. ***Hepatology*** 16, 167-172, 1992.
31. Hyland J, Ala-Kokko L, Royce P, Steinmann B, Kivirikko KI, Myllylä R. A homozygous stop codon in the lysyl hydroxylase gene in two siblings with Ehlers-Danlos syndrome type VI. ***Nature Genet*** 2, 228-231, 1992.
32. Williams CJ, Harrison DA, Hopkinson I, Baldwin CT, Ahmad NN, Ala-Kokko L, Korn RM, Buxton PG, Dimascio J, Considine EL, Prockop DJ. Detection of sequence variants in the gene for human type II procollagen (COL2A1) by direct sequencing of polymerase chain reaction-amplified genomic DNA. ***Human Mutation*** 1, 403-416, 1992.

33. Vikkula M, Palotie A, Ritvaniemi P, Ott J, Ala-Kokko L, Sievers U, Aho K, Peltonen L. Early-onset osteoarthritis linked to type II procollagen gene: detailed clinical phenotype and further analyses of the gene. ***Arthritis Rheum*** 36, 401-409, 1993.
34. Vikkula M, Ritvaniemi P, Vuorio AF, Kaitila I, Ala-Kokko L, Peltonen L. A mutation in the amino-terminal end of the triple helix of type II collagen causing severe osteochondrodysplasia. ***Genomics*** 16, 282-285, 1993.
35. Körkkö J, Ritvaniemi P, Haataja L, Kääriäinen H, Kivirikko KI, Prockop DJ, Ala-Kokko L. Mutation in type II procollagen gene (COL2A1) that substitutes aspartate for glycine α1-67 and that causes cataracts and retinal detachment. Evidence for molecular heterogeneity in the Wagner syndrome and the Stickler syndrome (arthro-ophthalmopathy). ***Am J Hum Genet*** 53, 55-61, 1993.
36. Ritvaniemi P, Hyland J, Ignatius J, Kivirikko KI, Prockop DJ, Ala-Kokko L. A fourth example suggests premature termination codons in the COL2A1 gene are a common cause of the Stickler syndrome. Analysis of the COL2A1 gene by denaturing gradient gel electrophoresis. ***Genomics*** 17, 218-221, 1993.
37. Helminen HJ, Kiraly K, Pelttari A, Tammi MI, Vandenberg P, Pereira R, Dhulipala R, Khillan JS, Ala-Kokko L, Hume EL, Prockop DJ. An inbred line of transgenic mice expressing an internally deleted gene for type II procollagen (COL2A1). Young mice have a variable phenotype of a chondrodysplasia and older mice have osteoarthritic changes in joints. ***J Clin Invest*** 92, 582-595, 1993.
38. Sieron AL, Fertala A, Ala-Kokko L, Prockop DJ. Deletion of a large domain in recombinant human procollagen II does not alter the thermal stability of the triple helix. ***J Biol Chem*** 268, 21232-21237, 1993.
39. Fertala A, Sieron AL, Ganguly A, Li S-W, Ala-Kokko L, Anumula KR, Prockop DJ. Synthesis of recombinant human procollagen II in stably transfected tumor cell line (HT-1080). ***Biochem J*** 298, 31-37, 1994.
40. Ritvaniemi P, Sokolov BP, Williams CJ, Considine E, Yurgenev L, Meerson EM, Ala-Kokko L, Prockop, D.J. A single base mutation in the type II procollagen gene (COL2A1 that converts glycine α1-247 to serine in a family with spondylo-epiphyseal dysplasia. ***Hum Mutat*** 3, 261-267, 1994.
41. Freisinger P, Ala-Kokko L, LeGuellec D, Franc S, Bouvier R, Ritvaniemi P, Prockop DJ, Bonaventure J. A mutation in the COL2A1 gene in a patient with hypochondrogenesis. The mutation causes expression of the genes for type I procollagen in chondrocytes. ***J Biol Chem*** 269, 13663-13669, 1994.
42. Vikkula M, Metsäranta M, Ala-Kokko L. Type II collagen mutations in rare and common cartilage diseases. ***Ann Med*** 26, 107-114, 1994.
43. Bonaventure J, Cohen-Solal L, Ritvaniemi P, van Maldergem L, Khadom N, Delezoide AL, Maroteaux P, Prockop DJ, Ala-Kokko L. A substitution of aspartic acid for glycine at position 310 in type II collagen produces achondrogenesis II, and a substitution of serine at position 805 produces hypochondrogenesis. Analysis of genotype-phenotype relationships. ***Biochem J*** 307, 823-830, 1995.
44. Sokolov BP, Ala-Kokko L, Dhulipala R, Arita M, Khillan JS, Prockop DJ. Tissue-specific expression of the gene for type I procollagen (COL1A1) in transgenic mice. Only 476 bp of the promoter are required if collagen genes are used as reporters. ***J Biol Chem*** 270, 9622-9629, 1995.
45. Ala-Kokko L, Kvist A-P, Metsäranta M, Kivirikko KI, de Crombrugghe B, Prockop DJ, Vuorio E. Conservation of the sizes of 53 introns and over one hundred intronic sequences for the binding of common transcription factors in the human and mouse genes for type II procollagen (COL2A1). ***Biochem J*** 308, 923-929, 1995.
46. Ritvaniemi P, Körkkö J, Bonaventure J, Vikkula M, Hyland J, Paassilta P, Kaitila I, Kääriäinen H, Sokolov BP, Hakala M, Mannismäki P, Meerson EM, Klemola T, Williams C, Peltonen L, Kivirikko KI, Prockop, DJ, Ala-Kokko L. Identification of COL2A1 gene mutations in patients with chondrodysplasias and osteoarthritis. ***Arthritis Rheum*** 38, 999-1004, 1995.
47. Vuoristo MM, Pihlajamaa T, Vandenberg P, Prockop DJ, Ala-Kokko L. The human COL11A2 gene. The structure indicates that the gene has not evolved with the genes for the major fibrillar collagens. ***J Biol Chem*** 270, 22873-22881, 1995.

48. Thakker-Varia S, Anderson D, Kuivaniemi H, Tromp G, Shin H.-G, van der Rest M, Glorieux FH, Ala-Kokko L, Stolle CA Aberrant splicing of the type III procollagen mRNA leads to intracellular degradation of the protein in a patient with Ehlers-Danlos type IV. ***Hum Mutat*** 6, 116-125, 1995.
49. Kaitila L, Körkkö J, Marttinen E, Ala-Kokko L. Phenotypic expression of a Gly154Arg mutation in the type II in two unrelated patients with spondyloepimetaphyseal dysplasia (SEMD). ***Am J Med Genet*** 63, 111-122, 1996.
50. Pääkkö P, Anttila S, Sormunen R, Ala-Kokko L., Peura R, Ferrans VJ, Ryhänen L. Biochemical and morphological characterization of carbon tetrachloride-induced lung fibrosis in rats. ***Arch Toxicol*** 70, 540-552, 1996.
51. Ryhänen L, Stenbäck F, Ala-Kokko L and Savolainen ER, The Effect of malotilate on type III and type IV collagen, laminin and fibronectin metabolism in dimethyinitrosamine-induced liver fibrosis in the rat. ***J Hepatol*** 24, 238-245, 1996.
52. Tromp G, Kuivaniemi H, Raphael S, Ala-Kokko L, Christiano A, Considine E, Dhulipala R, Hyland J, Jokinen A, Kivirikko S, Korn R, Madhatheri S, McCarron S, Pulkkinen L, Punnett, H, Shimoya K, Spotila L, Tate A, Williams CJ Genetic linkage of familial granulomatous inflammatory arthritis, skin rash and uveitis to chromosome 16. ***Am J Hum Genet*** 59, 1097-1107, 1996.
53. Vandenberg P, Vuoristo MM, Ala-Kokko L, Prockop DJ. The mouse col11a2 gene. Some transcripts from the adjacent rxr-β gene extend into the col11a2 gene. ***Matrix Biol*** 15, 359-367, 1996.
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