CURRENT POSITIONS:

CEO and Medical Director, Connective Tissue Gene Tests, LLC, Allentown, PA

EDUCATION:

M.D.	June 1993	Jefferson Medical College
Ph.D.	May 1985	Biology, Temple University
M.A.	January 1982	Biology, Temple University
B.A.	August 1974	Biology, Temple University

BOARD CERTIFICATIONS:

October 1999	Anatomic/Clinical Pathology
September 2000	Hematology

PROFESSIONAL MEMBERSHIPS:

American College of Pathologists

American Society of Clinical Pathologists

American Society of Human Genetics

Association for Molecular Pathology

Pennsylvania Medical Society

PROFESSIONAL EXPERIENCE:

August 1, 2004 – Present	Medical Director, Connective Tissue Gene Tests, LLC, Allentown, PA
Sept 1,2004 – Mar 31, 2006	Staff Hematopathologists, Lehigh Valley Hospital, Allentown, PA.
July 1, 2000 – June 15, 2004	Assistant Professor of Clinical Pathology, Department of Pathology and Laboratory Medicine and Director of Molecular Diagnostics, Martix DNA Diagnostics, Tulane University Medical Center, New Orleans, LA

	Hematopatholgy coverage: Medical Center of Louisiana, New Orleans, Charity Hospital, New Orleans, LA.
July 1,1998-June 30, 2000	Hematopathology Fellow, Thomas Jefferson University Hospital, Jefferson Health System, Philadelphia, PA.
July 1, 1994-June 30, 1998	Resident, AP/CP Program, Thomas Jefferson University Hospital, Philadelphia, PA
July 1, 1993-June 30, 1994	Transition Resident, Chestnut Hill Hospital, Philadelphia, Penna. 19118
March 1, 1993- June 1, 1993	Department of Dermatology, Jefferson Medical College, Thomas Jefferson University, Philadelphia, PA.
September 1991- August 1992	Visiting Scientist, Department of Medical Biochemistry, University of Oulu, Oulu, Finland.
January 1989- September 1989	Consultant, Department of Biochemistry and Molecular Biology, Jefferson Institute of Molecular Medicine, Thomas Jefferson University, Philadelphia, PA.
January 1986- January 1989	Research Associate, Department of Biochemistry and Molecular Biology, Jefferson Institute of Medicine, Thomas Jefferson University and Post-doctoral Fellow, Department of Biochemistry, Robert Wood Johnson Medical School Piscataway, NJ.
March 1985- December 1985	Research Specialist, Cancer Center, Department of Medicine University of Pennsylvania, Philadelphia. PA.
September 1979-March 1985	Graduate Student, Department of Biology, Temple University, Philadelphia PA.
January 1979- April 1979	Instructor in Biology, Temple University, Philadelphia, PA.

GRANTS AND AWARDS

First Prize 1998, Residents Forum, AFIP- American Registry of Pathology. Best submitted publication.

Fogarty International Fellowship, September 1991-August 1992.

Public Health Service Training Grant, 27 months at Jefferson Medical College. Various Research and Teaching Assistantships at Temple University.

RECENT CONFERENCES/ PRESENTATIONS

6th International Symposium on the Marfan Syndrome.
Seattle, WA, 2001

ASCP/CAP Joint Annual Meeting Philadelphia, PA. 2001

Tutorial on Neoplastic Hematopathology Miami, FL 2003

Collagen Gordon Research Conference. New London NH. Poster Presentation. 2003

6th International Skeletel Dysplasia Society Conference.Warrenton, VA. Poster Presentation. 2003

Annual Meeting of the American Society of Human Genetics, Salt Lake, UT, 2005

Annual Meeting of the American Society of Human Genetics, New Orleans, LA, 2006

Annual Meeting of the Association for Molecular Pathology, Los Angeles, CA, 2007

Annual Meeting of the Canadian Collage of Medical Genetics, St. John's, NL, Canada - 2008

The American Society of Human Genetics, Philadelphia PA - 2008

The American College of Medical Genetics, Tampa, FL, Poster – 2009

9th International Skeletel Dysplasia Society Conference, Boston, MA - 2009

The American Society of Human Genetics, Honolulu, HI - 2009

The American College of Medical Genetics, Albuquerque, NM – 2010

The American College of Medical Genetics, Vancouver, Canada – 2011

The American Society of Human Genetics, Montreal, Canada - 2011

The American College of Medical Genetics, Charlotte, NC – 2012

The American Society of Human Genetics, Boston, MA - 2013

The American Society of Human Genetics, San Diego, CA - 2014

ADDITIONAL TRAINING Two Months at AFIP-Studied Molecular Biology of Testicular Lymphoma.

PUBLICATIONS:

1 Rovera G, Hyland J, Ming P-M (1977) . Temperature-Sensitive Variants for Saturation Density and Anchorage Dependency of a Simian Virus 40- Transformed Human x Mouse Hybrid Cell Line. *J Natl Cancer Inst* 58:711.

2 Hyland J, Conner C, Putzrath R, Brownstein B (1980). Alterations in the Cell Surface of a Cytochalazin B-Resistant Rat Liver Epithelial Cell Line. *Eur J Cell Biol* 22:264.

3 Hyland J, (1984). Differentiated and Non-Differentiated Mouse 3T3-F442A Cells Exhibit the Same Heat Shock Response. *Federation Proc* 43:2052.

4 Hyland J, The Expression of Heat Shock Genes in Differentiated and Non-Differentiated 3T3-F442A Cells Exhibit the Same Heat Shock Response. Dissertation May 19th, 1985.

5 Ala-Kokko L, Kontusaari S, Olsen A, Hyland J, Jimenez SA, Prockop DJ (1989) Expression of Human Type II Procollagen Genes in Mouse Fibroblasts Transfected with a Chimeric Gene Construct Containing the Promoter of the Type I Pro-collagen Gene. *Arthritis Rheum* 32:384.

6 Prockop DJ, Olsen A, Kontusaari S, Hyland J, Ala-Kokko L, Vasan NS, Barton E, Buck S, Harrison K, Brent RL (1990) Mutations in Procollagen Genes. Consequences of the Mutations in Man and in Transgenic Mice. Ann NY Acad Sci Feb. 19:330

7 Ala-Kokko L, Hyland J, Jimenez SA, Prockop DJ, (1990) Expression of the Human Type II Procollagen Gene in Mouse NIH 3T3 Cells by the Use of a Vector Containing the Promoter Region, The First Exon and The First Intron of the Pro a1 Chain for the Human Type I Procollagen. Matrix *Coll Rel Res* 10

8 Geddis AL, Hyland J, Ala-Kokko L, Fertala A, Prockop DJ, (1991) Expression of a Human a1 Collagen in Stably Transfected mouse 3T3 Fibroblasts. The Effect of Amino Acid Substitution on Thermal Stability. East Coast Connective Tissue Society. Eleventh Annual Meeting. Piscataway, N.J. USA.

9 Ala-Kokko L, Hyland J, Smith C, Kivirikko KI, Jimenez SA, Prockop DJ (1991)Expression of a Human Cartilage Procollagen Gene (COL2A1) in Mouse 3T3 Cells. *J Biol Chem* 266:14175.

10 Hyland J, Ala-Kokko L, Royce P, Steinman B, Kivirikko KI, Myllyla R (1992) A Homozygous Mutation Creating a Stop Codon in the Lysyl Hydroxylase Gene in a Family with Ehlers-Danlos Syndrome Type VI. Thirteenth FECTS Meeting, Davos- Platz, Switzerland.

11 Hyland J, Ala-Kokko L, Royce P, Steinman B, Kivirikko KI, Myllyla R (1992) A Homozygous Stop Codon in the Lysyl Hydroxylase Gene in Two Sibilings with Ehlers-Danlos Syndrome Type VI. *Nature Genetics* 2:228

12 Ritvaniemi P, Hyland J, Ignatius J, Kivirikko KI, Prockop DJ, Ala-Kokko L (1993) A Fourth Example Suggests Premature Termination Codons in the COL2A1 Gene Are a Common Cause of the Stickler Syndrome: Analysis of the COL2A Gene by Denaturing Gradient Gel Electrophoresis. *Genomics* 17:218

13 Hautala T, Hyland J, Heikkinen J, Krieg T, Kivirikko KI, Myllya R (1993) A Case of Type VI Ehlers-Danlos Syndrome Caused by a Homozygous Deletion of Two Exons in the mRNA for Lysyl Hydroxylase. Mutations in the Human Genome Orta San Giulio, Italy.

14 Ignatius J, Ritvaniemi P, Korkko J, Hyland J, Haataja L, Kivirikko KI, Prockop DJ, Ala-Kokko L (1993) Phenotype Variation in Stickler/Wager Syndrome Due to Different Mutations in the Type II Procollagen (COL2A1) Gene. Twenty-fifth Annual Meeting. European Society Human Genetics.

15 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 I, Punnett H, Shimoya K, Spotila L, Tate A, Williams CJ, (1994) Molecular Characterization of Blau Syndrome Genetic Linkage to Chromosome 16. American Society of Human Genetics, Montreal, Canada

16 Ritavaniemi P, Korkko J, Bonaventure J, Vikkula M, Hyland J, Passilta P, Kaitila I, Kaarianen H, Sokolov BP, Hakala M. et. al. (1995) Identification of COL2A1 Gene Mutation in Patients with Chondrodysplasias and Familial Osteoarthritis. *Arthritis Rheum* 38:999

17 Tromp G, Kuivaniemi H, Raphael S, Ala-Kokko L, Christiano A, Considine E, Dhulipala R, Hyland J, Jokinen A, Kivirikko S, et. al. (1996) Genetic Linkage of Familial Granulomatous Inflammatory Arthritis, Skin Rash and Uveitis to Chromosome 16. *Am J Hum Genet* 59:1097

18 Pousi B, Hautala T, Hyland J, Schroter J, Eckes B, Kivirikko K, Myllyla R (1998) A Coumpound Heterozygote Patient with Ehlers-Danlos Syndrome Type VI has a Deletion in One Allele and a Splicing Defect in the Other Allele of the Lysyl Hydroxylase Gene. *Human Mutation* 11: 55

19 Hyland J, Lasota J, Jasinski M, Petersen R.O, Nordling S, Miettinen M. (1998) Molecular Pathological Analysis of Primary Testicular Diffuse Large Cell Lymphomas Shows Differences From Nodal Large Cell Lymphomas. *Human Pathology* 29:1231

20 Murphy TJ, Mowad CM, Feig SA, Nussbaum SA, Hyland JC. (1998) Breast imaging case of the day. Dermatopathic lymphadenopathy. *Radiographics* 18:536

21 Schafer A, Stein J, Hyland J, Schwarze U, Byers P, Clark A. (2002) Single Nucleotide Substitution in the COL1A1 gene (IVS13-10T>A) Results in use of a New Splice Acceptor Site, mRNA Instability, and Mild Osteogenesis Imperfecta. *Am J Hum Genet* 71:4

22 Collod-Béroud G, Le Bourdelles S, Ades L, Ala-Kokko L, Booms P, Boxer M, Child A, Comeglio P, De Paepe A, Hyland JC, Holman K, Kaitila I, Loeys, B, Matyass G, Nuytinck, L, Peltonen L, Rantamaki T, Robinson P, Steinmann B, Junien C, Béroud C, Boileau C (2003) Update of the UMD-FBN1 Mutation Database and Creation of an FBN1 Polymorphism Database. *Hum Mutat* 22:199.

23 Schafer IA, Stein J, Hyland JC, Clark B. (2004) Gene symbol: COL1A1. Disease: Osteogenesis imperfecta type 1. *Hum Genet* 114:404.

24 Hartikka H, Kuurila K, Körkkö J, Kaitila I, Grénman R, Pynnönen S, Hyland JC, Ala-Kokko L. (2004) Lack of correlation between the type of *COL1A1* or *COL1A2* mutation and hearing loss in osteogenesis imperfecta patients. *Hum Mutat* 24, 147.

25 Hyland J. Ala-Kokko L. Prenatal Diagnosis of Connective Tissue Disorders. (2004) In: Milunsky A. editor. Genetic Disorders and the Fetus 5th edition. Baltimore. John Hopkins University Press, pp 700-718.

26 Sutton VR, Hyland JC, Phillips WA, Schlessinger AE, Brill PW. (2005) A dominantly inherited spondylometaphyseal dysplasia with "corner fractures" and congenital scoliosis. *Am J Med Genet* 133, 209.

27 Leung L, Hyland JC, Young A, Goldberg MF, Handa JT. (2006) A novel mutation in intron 11 of the COL2A1 gene in a patient with type 1 Stickler syndrome. Retina 26:106-109.

28 Hyland J. Skin and Connective Tissue Disorders. (2007) In: Leonard D. editor. Molecular Pathology in Clinical Practice. New York: Springer Science and Business Media, pp. 191-203.

29 Walter K, Tansek M, Tobias M T, Ikegawa S, Coucke P, Hyland J, Mortier G, Iwaya T, Nishimura G, Superti-Furga A, Unger S. (2007) COL2A1-related skeletal dysplasias with predominant metaphyseal involvement. Am J Med Genet 134A, 161.

30 Sutherell J, Zarate Y, Tinkle BT, Markham LW, Cripe LH, Hyland JC, Witte D, Hopkin RJ, Hinton RB. (2007) Novel fibrillin 1 mutation in a case of neonatal Marfan syndrome: the increasing importance of early recognition. Congenit Heart Dis 2342-346.

31 Marini JC, Forlino A, Cabral WA, Barnes AM, San Antonio JD, Milgrom S, Hyland JC, Körkkö J, Prockop DJ, De Paepe A, Coucke P, Symoens S, Glorieux FH, Roughley PJ, Lund AM, Kuurila-Svahn K, Hartikka H, Cohn DH, Krakow D, Mottes M, Schwarze U, Chen D, Yang K, Kuslich C, Troendle J, Dalgleish R, Byers PH. Consortium for osteogenesis imperfecta mutations in the helical domain of type I collagen: regions rich in lethal mutations align with collagen binding sites for integrins and proteoglycans. (2007) Hum Mutat 28: 209-221.

32 Schievink WI, Gordon OK, Hyland JC, Ala-Kokko L. (2008) Absence of TGFBR2 mutations in patients with spontaneous spinal CSF leaks and intracranial hypotension. J Headache Pain 9: 99-102.

33 Milewicz DM, Ostergaard JR, Ala-Kokko LM, Khan N, Grange DK, Mendoza-Londono R, Bradley TJ, Olney AH, Adès L, Maher JF, Guo D, Buja LM, Kim D, Hyland JC, Regalado ES. De novo ACTA2 mutation causes a novel syndrome of multisystemic smooth muscle dysfunction. *Am J Med Genet A* 152A:2437-2443, 2010.

34 Dwyer E, Hyland J, Modaff P, Pauli RM. Genotype – phenotype correlation in *DTDST* dysplasias: atelosteogenesis type II and diastrophic dysplasia variant in one family. *Am J Med Genet A* 152A:3043-3050, 2010.

35 Hyland J. Osteogenesis Imperfecta. In: Lang F. editor Encylopedic Reference of Molecular Mechanisms of Disease Heidelberg. Springer –Verlag. In Press.

36 Baker S, Booth C, Fillman C, Shapiro M, Blair MP, Hyland JC, Ala-Kokko L. 2011. A loss of function mutation in the COL9A2 gene causes autosomal recessive Stickler syndrome. *Am J Med Genet Part A* 155:1668–1672, 2011.

37 Kannu P, O'Rielly DD, Hyland JC, Ala Kokko L. 2011. Avascular necrosis of the femoral head due to a novel C propeptide mutation in COL2A1. *Am J Med Genet Part A* 155:1759-1762, 2011.

38 Zadeh N, Bernstein JA, Niemi AK, Dugan S, Kwan A, Liang D, Hyland JC, Hoyme HE, Hudgins L, Manning MA. Ectopia lentis as the presenting and primary feature in Marfan syndrome. *Am J Med Genet Part A* 155:2661-2668, 2011.

39 Khalifa O, Imtiaz F, Allam R, Al-Hassnan Z, Al-Hemidan A, Al-Mane K, Abuharb G, Balobaid A, Sakati N, Hyland J, Al-Owain M. A recessive form of Marshall syndrome is caused by a mutation in the COL11A1 gene. *J Med Genet* 49: 246-248, 2012.

40 Kochhar A, Kirmani S, Cetta F, Younge B, Hyland JC and Michels V. Similarity of geleophysic dysplasia and Weill–Marchesani syndrome. *Am J Med Genet Part A* 161:3130-3132, 2013.

41 Regalado E, Guo D-C, Prakash S, Bensend T, Flynn K, Estrera A, Safi H, Liang D, Hyland J, Child A, Arno G, Boileau C, Jondeau G, Moran R, Morisaki T, Morisaki H, Pyeritz R, Coselli J, LeMaire SA, Milewicz D. Aortic Disease Presentation and Outcome Associated with ACTA2 mutations. *Circulation*, in press.