

Curriculum Vitae

August 25, 2008

UTA FRANCKE

Professor of Genetics and Pediatrics
 Stanford University School of Medicine
 Stanford, California 94305-5323
 Telephone: (650) 725-8089
 FAX: (650) 725-8112
 EMail: ufrancke@stanford.edu

Citizenship U.S.A.**Married to** Heinz Furthmayr M.D., Professor of Pathology at Stanford, Emeritus**License** Physicians and Surgeons, State of California A25346**Education**

General Aufbaugymnasium Idstein, Germany, Abitur (Baccalaureat), 1961
 Medical University Medical School at Frankfurt (1961-62), Marburg (1962-63),
 Munich (1963-66)
 Final State Examination and Dissertation: Munich, 1967 (Dr.med)

Postgraduate Training

1967-1969 Intern, two-year rotating program, Klinikum rechts der Isar, Munich, Germany
 1969-1970 Resident in Pediatrics, Children's Hospital of Los Angeles, Los Angeles, CA
 1970-1971 Postdoctoral Fellow in Medical Genetics, University of California, Los Angeles, CA
 1971-1973 Postdoctoral Fellow in Medical Genetics, University of California, San Diego, CA
 1991 Cold Spring Harbor Course on Computational Genomics

Medical Specialty Board Certifications

1981 American Board of Pediatrics
 1982 American Board of Medical Genetics: Clinical Genetics and Clinical Cytogenetics
 1993, 2003 American Board of Medical Genetics: Clinical Molecular Genetics

Academic, Research and Hospital Staff Appointments

1973-1978 Assistant Professor of Pediatrics (in Residence), University of California San Diego
 1975-1978 Director of Medical Genetics, San Diego Children's Hospital and Health Center
 1975-1978 Director, Cytogenetics and Cell Genetics Laboratory, Department of Pediatrics,
 University of California San Diego
 1978-1985 Associate Professor of Human Genetics and Pediatrics, Department of Human
 Genetics, Yale University School of Medicine, New Haven, CT
 1985-1988 Professor of Human Genetics and Pediatrics, Director of Postdoctoral Training
 Program in Medical Genetics, Yale University School of Medicine, New Haven, CT
 1978-1988 Attending Physician, Clinical Genetics Service
 Yale-New Haven Hospital, New Haven, CT
 1984-1985 Visiting Scientist, European Molecular Biology Laboratory (with Dr. Hans Lehrach)
 Heidelberg, Germany
 1989-1996 Professor of Pediatrics, Stanford University School of Medicine
 1989-2000 Professor of Genetics and Investigator, Howard Hughes Medical Institute,
 Stanford University School of Medicine, Stanford, CA

Academic, Research and Hospital Staff Appointments (continued)

- 1989- Medical Staff, Stanford University Hospital, Stanford, CA
 1989- Medical Staff, Lucile Salter Packard Children's Hospital, Stanford, CA
 1989-2005 Director, Interdepartmental Medical Genetics Training Program,
 Stanford University School of Medicine, Stanford, CA
 1997-1998 Co-Director, UCSF/Stanford Medical Genetics Residency Program
 2001- Professor of Genetics and Pediatrics, Stanford University School of Medicine

Honors and Awards

- 1990 Elected Member, Institute of Medicine
 1990 Stanley Wright Memorial Lectureship Award, WSPR, Carmel, CA
 1995 Elected Fellow, American Association for the Advancement of Science
 1996 Antoine Marfan Award, National Marfan Foundation
 1997 Elected Member, American Academy of Arts and Sciences
 1999 President, American Society of Human Genetics
 2000 President, International Federation of Human Genetics Societies
 2001 March of Dimes/Colonel Harland Sanders Lifetime Achievement Award in Genetics
 2002 Original Member, Highly Cited Researchers database (ISI)
 2005 Irene Uchida Lectureship Award, University of Manitoba, Winnipeg Canada
 2005 Kurt Benirschke Lectureship Award, UCSD, San Diego CA

Editorial Activities

- 1979-1999 Associate Editor: *Cytogenetics and Cell Genetics*
 1977-1986 Board of Advisory Editors: *American Journal of Medical Genetics*
 2000- Board of Advisory Editors: *American Journal of Medical Genetics*
 1987-1991 Section Editor: *Genomics*
 1992-1999 Contributing Editor: *Human Mutation*
 2000- Editorial Advisory Board *Gene Function & Disease*

Editorial Boards:

- 1977-1979 *Cytogenetics and Cell Genetics*
 1983-1986 *American Journal of Human Genetics*
 1983-1988 *Genetic Epidemiology*
 1987-1990 *Molecular Biology and Medicine*
 1978-1998 *Human Genetics*
 1979-1999 *Somatic Cell and Molecular Genetics*
 1987-1990 *Oncogene Research*
 1989-1996 *Genes, Chromosomes & Cancer*
 1990-1997 *Mammalian Genome*
 1993-1998 *The Journal of Clinical Investigation*
 1995-2000 *American Journal of Medical Genetics*
 1999-2002 *Expert Reviews in Molecular Medicine*
 2000- *Current Opinion in Genetics & Development*
 2000- *BMC Genetics*
 2000- *BMC Medical Genetics*

Scientific Review Committees

- 1976-1980 Genetic Basis of Disease Review Committee (NIGMS/NIH)
 1984-1992 Basil O'Connor Starter Scholar Research Award Advisory Committee,
 March of Dimes Birth Defects Foundation
 1985-1989 Maternal and Child Health Research Committee (NICHD/NIH)
 1986-1988 Scientific Review Board, Genetics, Howard Hughes Medical Institute
 1990-1994 Member, Mammalian Genetics Study Section (DRG/NIH)

- 1992-1994 Chair, Mammalian Genetics Study Section (DRG/NIH)
 1994, 1997 Lawrence Berkeley Laboratories, Director's Review of Life Sciences Division
 1995 Panel to Assess NIH Investment in Research on Gene Therapy
 1996-1997 Chair, Committee on Breast Cancer Research, Institute of Medicine, NAS
 1997-2004 Scientific Advisory Board, Charles E. Culpeper Awards, Rockefeller Brothers Foundation
 1998-2001 Advisory Committee for the Career Awards in the Biomedical Sciences Program, Burroughs Wellcome Fund
 1998-2002 National Advisory Neurological Disorders & Stroke Council (NIH/NINDS)
 2000-2002 PubMed Central National Advisory Committee (NLM)
 2004, 2006 Scientific Advisory Board, Genome Canada
 2004-2008 Scientific Advisory Committee, VA CSP DNA bank
 2005-2008 Scientific Advisory Board, Austrian Genome Project (GEN-AU)
 2005-2006 Fachkommission und Gemeinsame Kommission, Exzellenzinitiative, German Government
 2008- CSP CSSMRB Genetic Epidemiology Scientific Review Subcommittee

Professional Society Memberships/ Directorships/ Advisory Boards

- 1976 Society for Pediatric Research
 1976-1986 Curator, Human Genetic Mutant Cell Repository, IMR, Camden, NJ
 1981-1984 Board of Directors, American Society of Human Genetics
 1981-1984 Program Committee, American Society of Human Genetics
 1983 Chair, Program Committee, American Society of Human Genetics
 1982-1986 Board of Directors, American Board of Medical Genetics
 1989 The Human Genome Organization
 1990-1995 Advisory Committee, March of Dimes Clinical Genetics Conferences
 1990 Society for Inherited Metabolic Disorders
 1990 Western Society for Pediatric Research
 1990 Pluto Club, Association of University Pathologists
 1990 International Mammalian Genome Society
 1991- Professional Advisory Board, National Marfan Foundation
 1993 Founding Member, American College of Medical Genetics
 1993 European Society of Human Genetics
 1994-1996 Awards Committee, American Society of Human Genetics
 1996 Fellow, Molecular Medicine Society
 1996-1999 Councilor, Association of Professors of Human and Medical Genetics
 1996-2007 Professional Advisory Board, International Rett Syndrome Association
 2001- Selection Committee, March of Dimes/Colonel Harland Sanders Lifetime Achievement Award in Genetics
 2001-2008 Genetics Prize Advisory Board, The Peter Gruber Foundation

International Scientific Activities

- 1982-1991 Member, International Committee for Human Cytogenetic Nomenclature
 1987-1991 Chair, International Committee for Human Cytogenetic Nomenclature
 1988-1991 Executive Program Committee, 8th International Congress of Human Genetics
 1995-1997 Scientific Advisory Committee on Human Genome Research, Federal Ministry of Education, Science, Research and Technology, Bonn, Germany
 1995-2001 Advisory Board, Center for Molecular Biology in Medicine, Univ. Koeln, Germany
 1996-2001 Member, International Standing Committee on Human Cytogenetic Nomenclature
 2000- Scientific Advisory Board, International Postgraduate Organization for Knowledge-Transfer Research and Teaching Excellent Students
 2002- International Advisory Board, IRSA RETT Phenotype Database

Meeting Organization

- 1991 Co-Director, Banbury Conference on Molecular Genetics and Cell Biology of Marfan Syndrome, Cold Spring Harbor, NY
- 1992 Co-Director, Second International Symposium on Marfan Syndrome
- 1992 Conference Director, 24th Annual March of Dimes Clinical Genetics Conference
- 1993 Chair, Conference on Fetal Research and Applications, Institute of Medicine, National Academy of Sciences, Irvine CA
- 1997 Co-Host, 13th Annual National Marfan Foundation Meeting, Stanford, CA
- 1997 Vice-Chair, Gordon Research Conference, Human Molecular Genetics, Newport, RI
- 1999 Chair, Gordon Research Conference, Human Molecular Genetics, Newport, RI
- 2001 Chair, Satellite Symposium on Rett Syndrome, 10th International Congress of Human Genetics, Vienna, Austria
- 2002 Chair, Keystone Conference "Genotype to Phenotype: Focus on Disease"

Consulting

- 1996-2001 Scientific Advisory Board, Genomica Corporation, Boulder, CO
- 2000-2001 Scientific Advisor, Genetic Health Inc., San Mateo, CA
- 2003-2004 Scientific Advisory Board, Naxcor, Mountain View, CA
- 2007 - Consultant, 23andMe Inc. Mountain View, CA

Invited Lectures (since 1989)

- 1989 Banbury Conference on Dystrophin, Cold Spring Harbor, NY
Molecular Neurogenetics FASEB Conference, Saxtons River, VT
Molecular Cytogenetics Symposium, Lake Tahoe, CA
Banbury Conference on Molecular Cytogenetics, Cold Spring Harbor, NY
Medical Genetics Training Program, University of California, Los Angeles, CA
- 1990 Stanley Wright Memorial Lecture, Western Society for Pediatric Research, Carmel, CA
Rett Syndrome Foundation Workshop, Washington, DC
4th International Workshop on Mouse Genome, Annapolis, MD
Howard Hughes Medical Institute, Lecture to Medical Students, Cloisters NIH, MD
- 1991 Banbury Conference on Molecular Genetics and Cell Biology of Marfan Syndrome, Cold Spring Harbor, NY
Short Course in Medical and Experimental Mammalian Genetics, Bar Harbor, ME
Summer School for Pediatric Endocrinology, Foer Islands, Germany
American Heart Association Convention, Postgraduate Seminar, Anaheim, CA
- 1992 Keystone Symposium, Molecular Biology of Human Genetic Disease, Copper Mountain, CO
CSH Genome Mapping and Sequencing Meeting, Cold Spring Harbor, NY
Ares Serono Symposium on Laron Syndrome, Lisbon, Portugal
First International Workshop on Chromosome 18 Mapping, Chicago, IL
Second International Symposium on the Marfan Syndrome, San Francisco, CA
Human Genetics Training Program, University of California, San Francisco, CA
- 1993 15th International Kabi Symposium on Growth and Growth Disorders, Florence, Italy
NIH Conference on Epigenetic Factors in Inheritance, Bethesda, MD
Joint LWPES/ESPE International Pediatric Endocrinology Meeting, San Francisco, CA
National Marfan Foundation Meeting and Research Symposium, Portland, OR

- Gordon Research Conference on Elastin, Kimball School, Meriden, New Hampshire
 IPOKRATES Postgraduate Course in Molecular Genetics, Krems, Austria
 International Workshop on Growth Hormone Insensitivity, Estoril, Portugal
- 1994 Miami 1994 Bio/Technology Winter Symposia, Advances in Gene Technology:
 Molecular Biology of Human Genetic Disease, Fort Lauderdale, FL
 Keystone Symposium, Molecular Biology of Human Genetic Disease, Copper
 Mountain, CO
 American Association for Cancer, Annual Convention, San Francisco, CA
 Symposium on Imprinting and Epigenetics, University of Oregon, Eugene, OR
 Life Sciences Division Seminar, Lawrence Berkeley Laboratory, UCB, CA
 Pediatrics Update, Continuing Medical Education, Travis Airforce Base, CA
 Carl Friedrich v. Siemens Stiftung, Nymphenburg, Munich, Germany
 Nobel Conference, Parental Imprinting: Causes and Consequences, Stockholm, Sweden
 Asia-Pacific Conference on Medical Genetics, Bangkok, Thailand
 Miami Bio/Technology European Symposium, Advances in Gene Technology:
 Molecular Biology of Human Genetic Disease, Monte Carlo, Monaco
- 1995 Western Society for Pediatric Research, Carmel, CA
 La Jolla Cancer Research Foundation, La Jolla, CA
 MSTP Program, Columbia University, New York, NY
 Genetics Department, Albert Einstein College of Medicine, Bronx, NY
 Biology Department, University of California, San Diego, CA
 Genetics Graduate Program, University of Chicago, Chicago, IL
 International Symposium on Clinical Immunology, San Francisco, CA
 Department of Cell Biology, Lawrence Berkeley Laboratory, Berkeley, CA
 Gordon Research Conference, Elastin and Elastic Fibers, Meriden, NH
 Gordon Research Conference, Human Molecular Genetics, Newport, RI
 American Society of Human Genetics Annual Meeting, Minneapolis, MN
 Medical Genetics and Mental Retardation Program, Baylor College of Medicine,
 Houston, TX
- 1996 Second International Growth Forum, Washington, D.C.
 Max Planck Institute for Molecular Genetics, Berlin-Dahlem, Germany
 Division of Immunology, Childrens Hospital of Los Angeles, Los Angeles, CA
 Interinstitutional Medical Genetics Training Program, UCLA, Los Angeles, CA
 12th Annual National Marfan Foundation Meeting, Houston, TX
 Childrens Hospital of Philadelphia and University of Pennsylvania, Philadelphia PA
 7th International Williams Syndrome Symposium, Philadelphia, PA
 9th International Congress of Human Genetics, Plenary Lecturer, Rio de Janeiro, Brazil
 Euroconference in Immunodeficiency Syndromes, Pasteur Institute, Paris, France
 Markey Distinguished Lectureship, University of Southern California, Los Angeles, CA
 6th International Congress of Cell Biology, Plenary Lecturer, San Francisco, CA
- 1997 Ciba Foundation Symposium: Epigenetics, London UK
 Public Symposium on Epigenetics, The Wellcome Centre for Medical Science, London UK
 13th Annual National Marfan Foundation Meeting, Stanford, CA
 Honors Lecture, New York University Medical Center, NYC
 Graduate Program in Cell Biology, Mt. Sinai School of Medicine, New York City
 Professional Women in Genetics, ASHG meeting, Baltimore MD
- 1998 Keystone Symposium: T Lymphocyte Activation, Differentiation and Death, Keystone CO
 Whitehead Policy Symposium: The Human Genome Project: Science, Law, and Social

- Change in the 21st Century, Plenary Lecturer, Cambridge, MA
 NIH Workshop: Genomic Alterations in Genetic Disease: Mechanisms of Structural Rearrangements, Bethesda, MD
 International Titisee Conference: Molecular Mechanisms in Human Malformation Syndromes, Titisee, Germany
 ASHG Symposium: Region Specific Repeats and Chromosomal Rearrangements, Denver, CO
 Pediatric Grand Rounds, Department of Pediatrics, UCSF, San Francisco, CA
- 1999 Medical Scientist Program Lecture, University of California at Irvine, CA
 Combined Endocrinology Conference, University of California at Los Angeles, CA
 Department of Molecular Genetics, University of Antwerp, Belgium
 Williams Syndrome Association Meeting, Palo Alto, CA
 American Society of Human Genetics, Presidential Address, San Francisco, CA
- 2000 National Advisory Neurological Disorders and Stroke Council February Meeting, Washington DC
 Department of Genetics Seminar, University of Koeln, Germany
 Workshop on Molecular Basis of Mental Handicap, Chamonix, France
 Annual Distinguished Scientist Lecture, Department of Molecular and Medical Genetics, University of Oregon Health Sciences Center, Portland OR
 Symposium Speaker at Society of Biological Psychiatry Annual Meeting, Chicago IL
 Symposium on Molecular Control of Organogenesis, University of Michigan, Ann Arbor MI
 Ihsan Dogramaci Lecture, Molecular Biology and Genetics, Bilkent University, Ankara, Turkey
 World Congress on Rett Syndrome, Karuizawa, Japan
 Northwest Rett Syndrome Foundation, Portland, OR
 The Olfactory Model System and Rett and Kallmann Syndromes, NIH, Bethesda MD
 Pediatrics Grand Rounds, Stanford University School of Medicine, Stanford CA
 Human Medical Genetics Graduate Program, University of Colorado, Denver CO
 National Marfan Foundation, Northern California Chapter, Palo Alto CA
 International Symposium of the Princess Takamatsu Cancer Research Fund, Tokyo, Japan
- 2001 Integrating Genomics Technologies in Health Care: Practice and Policy Challenges. Banbury Center. Cold Spring Harbor Laboratories, N.Y.
 International Symposium on GH and Growth Factors in Endocrinology and Metabolism. Plenary Lecture on the Human Genome Project. Valletta, Malta
 Weissenburg Symposium on Medicine and Molecular Biology. Lecture on Epigenetics and Human Disease. Weissenburg, Germany
 10th International Congress of Human Genetics. Public Lecture on the Implications of the Human Genome Project (in German), Vienna, Austria
 Satellite Symposium on Rett Syndrome. Vienna, Austria
 Rett Syndrome Research Foundation Symposium, Washington DC
 6th International Symposium on Marfan Syndrome, Seattle, WA
- 2002 State-of-the-Art Lecture. Western Society for Pediatric Research. Carmel, CA
 Advanced Human Genetics Graduate Course, UCSF, San Francisco, CA
 Kolloquium, Institut fuer Humangenetik, Universitaet Erlangen-Nuernberg, Germany
 Rett Syndrome Research Foundation Symposium, Baltimore MD
 Australian Society of Cytogenetics, Annual Meeting, Adelaide, Australia
 Human Genetics Society of Australasia Meeting, Plenary Lecture, Adelaide, Australia
 Royal Childrens Hospital and Murdoch Institute, Melbourne, Australia

- 2003 Society for Perinatology, Special Lecture, Taipei, Taiwan
 Introduction to Molecular Genetics, IpoKrates Postgraduate Education Course, Rust, Austria
 David W. Smith Workshop on Malformations and Morphogenesis, Vancouver, BC, Canada
 Institute of Genetic Medicine, 8th Annual Symposium, USC, Los Angeles CA
- 2004 Cardiovascular Medicine Seminar Series, Stanford, CA
 “Pharmacogenetics” Medical Genetics Grand Rounds, Stanford CA
 “Mouse models” Lecture in Medical Genomics Course, Stanford CA
 Plenary Lecture, Second Weissenburg Symposium on DNA Methylation, Germany
 David W. Smith Workshop on Morphogenesis and Malformations, Snowbird UT
 Plenary Lecture, Neurogenetics Conference, Genetics Society of Germany, Weimar
- 2005 Irene Uchida Lecture, University of Manitoba, Winnipeg, Canada
 Human Molecular Genetics research seminar, Univ. Manitoba, Winnipeg, Canada
 Kurt Benirschke Lecture, UCSD, San Diego CA
 Northern California Genetics Conference, Oakland CA
 Grand Rounds, Oakland Childrens Hospital CA
- 2006 Departmental Seminar, Univ. of Michigan, Department of Human Genetics, Ann Arbor MI
 50 Years of 46 Human Chromosomes: Progress in Cytogenetics, NIH, Bethesda MD
 11th International Congress of Human Genetics, Brisbane QLD, Australia
 Xth Oxford Conference on Modeling and Control of Breathing, Lake Louise, Banff, Canada
- 2007 UT Southwestern School of Medicine, Grand Rounds in Psychiatry, Dallas TX
 University of Iowa, Epigenetics Seminar Series, Iowa City IA
 RSRF Annual Symposium on Rett Syndrome research
- 2008 40th Biannual American Cytogenetics Conference, Monterey CA

Stanford University Service

- Director: Interdepartmental Postdoctoral Training Program in Medical Genetics
 (Principal Investigator, T32 GM08404, 1991-1997)
 (Principal Investigator, T32 GM08748, 2000-2005)
- Director: American Board of Medical Genetics accredited Training Programs in
 Clinical Genetics (1990-1997)
 Clinical Cytogenetics (1990-2004)
 Clinical Molecular Genetics (1993- 2004)
- Co-Director: UCSF/Stanford Joint Medical Genetics Residency Program (1997-1998)
- Clinical: Attending Physician, Medical Genetics Clinic and Consultation Service
 Attending Physician, Center for Marfan Syndrome and Related Connective
 Tissue Disorders
- Teaching: Genetics 201 (1989, 1990, 1991, 1992, 1993, 1994, 1995, 1997)
 Pathophysiology (1992)
 Advanced Human Genetics (Genetics 214) (1990, 1991)
 Mammalian Developmental Genetics (DB/Genetics 217) (1998)
 Sophomore Dialogue (Genetics Q102) (1998, 1999)

Human Genetics Journal Club (Organizer, 1989-)
 Human Biology 114 :Genomes, Diseases and Medicines (2002)
 Medical Genomics Course (2004)
 BioSci 109B: The Human Genome and Disease: Genetic Diversity and
 Personalized Medicine (2007, 2008)
 Human Biology Core 2A: Molecular Genetics Section(2007)

Medical School
 Committees:

Medical Scientist Training Program Committee (1989-1992)
 Program in Molecular and Genetic Medicine Advisory Committee (1989-1994)
 Pediatrics Department Chair Search Committee (1991-1992)
 General Clinical Research Center Advisory Committee (1992-1995)
 McCormick Lectureship Committee (1992-1999)
 Medical School Academic Senate, Member-at-large (1993-1999)
 Medical School Academic Senate, Executive Committee of Five (1996-1997)
 Appointments and Promotions Committee (1994-1998)
 Dean's Postdoctoral Fellowship Committee (1994-)
 Neurology Department Chair Search Committee (1995)
 Pediatric Genetics Division Chief Search Committee (1996-1997)
 Child Health Research Fund Advisory Committee (1996-98)
 Co-Chair, Stanford University School of Medicine/UCSF, Senate Committee
 on Academic Priorities and Strategies for Cooperation (1996-97)
 Chair, Advisory Committee, Center for Advanced Human Genetics (1996/97)
 Reproductive Endocrinology Faculty Search Committee (1997-99)
 Child and Adolescent Psychiatry and Child Development Faculty Search
 Committee (1998)
 Advisory Board, Women's Reproductive Health Research Career
 Development Center (1998-)
 Biochemical Genetics Faculty Search Committee (1998)
 Postdoctoral Affairs Committee (2001-2002)
 Chair, Faculty Search Committee, Dept. Genetics (2004-2005)
 Faculty Search Committee, Dept. Genetics (2005-2006)
 Pediatrics Department Chair Search Committee (2005)
 Genetic Counseling Faculty Search Committee (2006)
 Medical School Academic Senate, Member-at-large (2006-)
 Chair, Dean's Postdoctoral Fellowship Committee (2006-)
 Steering Committee of the Academic Senate (2007-2008)
 Advisory Committee; M.S. in Human Genetics and Genetic Counseling Program,
 Dept. Genetics (2007-)
 Advisory Committee: Stanford Career Development Program in the Genetics and
 Genomics of Lung Diseases (2008-)

University
 Committees:

Elected Member, Academic Senate (1990-1992)
 Academic Council, Committee on Research (1995-1998)
 Academic Council, Committee on Environmental Health and Safety (2005-2008)

PATENT:

U.S. Patent No US 6,709,817 B1;"METHOD OF SCREENING RETT
 SYNDROME BY DETECTING A MUTATION IN *MECP2*"

*Bibliography***UTA FRANCKE**

<u>Subject Area</u>	<u>Page</u>
I. Human Cytogenetics	10
II. Mouse Cytogenetics	13
III. Human Gene Mapping	14
IV. Human/Mouse Comparative Gene Mapping	27
V. Mouse Gene Mapping	31
VI. Human Genetic Disorders	34 - 46
A. New Syndromes Delineated	34
B. Lesch-Nyhan Syndrome	35
C. Duchenne/Becker Muscular Dystrophy (with microdeletions including Chronic Granulomatous Disease, Adrenal Hypoplasia and Glycerol Kinase Deficiency)	36
D. Growth Hormone Insensitivity Syndrome (Laron Syndrome)	38
E. Roberts/SC Phocomelia Syndrome	39
F. Prader Willi Syndrome and Genomic Imprinting	39
G. Marfan Syndrome and Fibrillinopathies	41
H. Rett Syndrome and MeCP2 Deficiency Disorders	44
I. Williams Beuren Syndrome	45
J. Wiskott Aldrich Syndrome	46
VII. Human Molecular Genetics	48 - 50
(Articles considered most important are in boldface.)	

I. HUMAN CYTOGENETICS

Francke, U.: Quinacrine mustard fluorescence of human chromosomes: Characterization of unusual translocations. *Am. J. Hum. Genet.* 24:189-213 (1972).

Crandall, B.F., Francke, U., Campbell, M.A., Sparkes, R.S.: Inherited t(13q14q) in two retarded sisters. *Am. J. Hum. Genet.* 24:416-424 (1972).

Spence, M.A., Francke, U., Forsythe, A.B.: Evidence against the peripheral location of the Y chromosome in human metaphase cells. *Cytogenet. Cell Genet.* 12:49-52 (1973).

Francke, U., Hammond, D.S., Schneider, J.A.: The band patterns of twelve D98/AH-2 marker chromosomes and their use for identification of intra-specific cell hybrids. *Chromosoma* 41:111-121 (1973).

Schneider, J.A., Francke, U., Hammond, D.S., Pellett, O.L., Becker, F.L.A.: Properties of cystinotic fibroblast-D98 cell hybrids studied by somatic cell hybridization. *Nature* 244:289-292 (1973).

Schrott, H.G., Sakaguchi, S., Francke, U., Luzzatti, L., Fialkow, P.J.: Translocation, t(4q-;13q+), in three generations resulting in partial trisomy of the long arm of chromosome 4 in the fourth generation. *J. Med. Genet.* 11:201-205 (1974).

Francke, U., Mahan, G.M., Dixon, B.K., Jones, O.W.: 10p-: A new autosomal deletion syndrome? In *Birth Defects: Original Article Series XI* (No. 5), pp. 207-212, The National Foundation, New York (1975).

Fawcett, W.A., McCord, W.K., Francke, U.: Trisomy 14q-. In *Birth Defects: Original Article Series XI* (No. 5), pp. 223-228, The National Foundation, New York (1975).

Francke, U., Jones, O.W., Moran, M.J.: Sex-chromosome abnormalities in husbands and wives. *Lancet* i:333-334 (1975).

Francke, U., Benirschke, K., Jones, O.W.: Prenatal diagnosis of trisomy 9. *Humangenetik* 29:243-250 (1975).

Spence, M.A., Forsythe, A.B., Nesbitt, M., Francke, U.: Methods for detecting non-random association of metaphase chromosomes. Technical Report No. 16. Health Sciences Computing Facility. University of California, Los Angeles (1975).

Francke, U., Kung, F.: Sporadic bilateral retinoblastoma and 13q- chromosomal deletion. *Med. Pediat. Oncol.* 2:379-385 (1976).

Francke, U.: Cytogenetics and somatic cell genetics: The impact of chromosome banding. In *Avenues of Clinical Genetic Research, Birth Defects: Original Article Series, XIII* (No. 6), pp. 79-103, The National Foundation, New York (1977).

Centerwall, W., Francke, U.: Familial trisomy 20p: Five cases and two carriers in three generations. A review. *Ann. Genet.* 20:77-83 (1977).

Sparkes, R.S., Francke, U., Muller, H., Toomey, K.: Partial 4q duplication due to inherited der(20),t(4;20)(q25;q13)mat. *Ann. Genet.* 20:31-35 (1977).

Francke, U.: Abnormalities of chromosomes 11 and 20. In *New Chromosomal Syndromes* (J. Yunis, ed.), pp. 245-272, Academic Press, New York (1977).

Taylor, K.M., Francke, U., Brown, M.G., George, D.L., Kaufhold, M.: Inverted tandem ("mirror") duplications in human chromosomes: inv dup 8p, 4q, 22q. *Am. J. Med. Genet.* **1**:3-19 (1977).

Riccardi, V.M., Sujansky, E., Smith, A.C., Francke, U.: Chromosomal imbalance in the aniridia-Wilms tumor association: 11p interstitial deletion. *Pediatrics* **61**:604-610 (1978).

Francke, U.: Clinical syndromes associated with partial duplications of chromosome 2 and 3: Dup(2)p, dup(2)q, dup(3)p, dup(3)q. In *Birth Defects Original Article Series XIV* (No. 6C), pp. 191-217, The National Foundation, New York (1978).

Oliver, N., Francke, U., Taylor, K.M.: Silver staining studies on the short arm variant of human chromosome 17. *Hum. Genet.* **42**:79-82 (1978).

Oliver, N., Francke, U.: Ideograms of high-resolution trypsin-Giemsa banded human chromosomes. *Cytogenet. Cell Genet.* **22**:668-671 (1978).

Francke, U., Oliver, N.: Quantitative analysis of high-resolution trypsin-Giemsa bands on human prometaphase chromosomes. *Hum. Genet.* **45:137-165 (1978).**

Francke, U.: Hageman (Factor XII) locus on 7q? Report of a second case with del(7)q35 and normal factor XII level. *Hum. Genet.* **45**:363-367 (1978).

Francke, U.: Chromosome 11q partial trisomy syndrome. In *Birth Defects Compendium* (D. Bergsma, ed.), pp. 203-204, The National Foundation, Alan Liss, Inc. (1979).

Francke, U.: Chromosome 11q- syndrome. In *Birth Defects Compendium* (D. Bergsma, ed.), pp. 204-205, The National Foundation, Alan Liss, Inc. (1979).

Begovich, A., Francke, U.: Karyotype evolution of the simian virus 40-transformed human cell line LNSV. *Cytogenet. Cell Genet.* **23**:3-11 (1979).

Francke, U., Brown, M.G., Jones, K.L.: Immediate chromosome diagnosis on bone marrow cells: An aid to management of the malformed newborn infant. *J. Pediat.* **94**:289-292 (1979).

Hittner, H.M., Riccardi, V.M., Francke, U.: Aniridia caused by a heritable chromosome 11 deletion. *Ophthalmology* **86**:1173-1183 (1979).

Francke, U., Holmes, L.B., Atkins, L., Riccardi, V.M.: Aniridia-Wilms' tumor association: Evidence for specific deletion of 11p13. *Cytogenet. Cell Genet.* **24:185-192 (1979).**

Riccardi, V.M., Hittner, H.M., Francke, U., Pippin, S., Holmquist, G.P., Kretzer, F.L., Ferrell, R.: Partial triplication and deletion of 13q: Study of a family presenting with bilateral retinoblastoma. *Clin. Genet.* **15**:332-345 (1979).

Monheit, A., Francke, U., Saunders, B., Jones, K.L.: The penta-X syndrome. *J. Med. Genet.* **17**:392-396 (1980).

Francke, U.: Role of high-resolution chromosome banding in the evaluation of mentally retarded individuals. In *Prevention of Mental Retardation and other Developmental Disabilities*, (M.K. McCormack, ed.), pp. 113-133, Marcel Dekker, Inc., New York (1980).

Riccardi, V.M., Hittner, H.M., Francke, U., Yunis, J.J., Ledbetter, D., Borges, W.: The aniridia-Wilms tumor association: The critical role of chromosome band 11p13. *Cancer Genet. Cytogenet.* **2**:131-137 (1980).

Francke, U.: Gene dosage studies in Down Syndrome: A review. In *Trisomy 21 (Down Syndrome) Research Perspectives* (F.F. de la Cruz, P.S. Gerald, eds.), pp. 237-251, University Park Press, Baltimore (1981).

Francke, U.: High resolution ideograms of trypsin-Giemsa banded human chromosomes. *Cytogenet. Cell Genet.* **31:24-32 (1981).**

O'Connor, R.D., Brown, M.G., Francke, U.: Immunologic and karyotypic studies in ataxia-telangiectasia: Specificity of break points on chromosomes 7 and 14 in lymphocytes from patients and relatives. In *Ataxia Telangiectasia--A Cellular and Molecular Link Between Cancer, Neuropathology and Immune Deficiency* (B.A. Bridges, D.G. Harnden, eds.) John Wiley and Sons, London (1982).

Haseltine, F.P., Lynch, V.A., van Dyke, D.L., Breg, W.R., Francke, U.: H-Y antigen expression in patients with X-autosomal translocations and gonadal dysgenesis. *Am. J. Med. Genet.* **13:115-123 (1982).**

Francke, U.: Specific chromosome changes in the human heritable tumors retinoblastoma and nephroblastoma. In *Chromosomes and Cancer* (J. Rowley, J. Ulmann, eds.), pp. 99-115, Academic Press, New York (1983).

Francke, U.: Random X inactivation resulting in mosaic nullisomy of region Xp21.1Æp21.3 associated with heterozygosity for ornithine transcarbamylase deficiency and for chronic granulomatous disease. *Cytogenet. Cell Genet.* **38:298-307 (1984).**

Yang-Feng, T.L., Finley S.C., Finley, W.H., Francke, U.: High resolution cytogenetic evaluation of couples with recurring fetal wastage. *Hum. Genet.* **69**:246-249 (1985).

Münke, M., de Martinville, B., Lieber, E., Francke, U.: Minute chromosomes replacing the Y chromosome carry Y-specific sequences by restriction fragment analysis and *in situ* hybridization. *Am. J. Med. Genet.* **22**:361-374 (1985).

Cowan, J.M., Halaban, R., Lane, A.T., Francke, U.: The involvement of 6p in melanoma. *Cancer Genet. Cytogenet.* **20:255-261 (1986).**

Francke, U.: Elusive chromosome anomalies. *Hospital Practice* **21**:175-193 (1986).

Francke, U., Harper, J.F., Darras, B.T., Cowan J.M.: Detection of microdeletions with high-resolution cytogenetics and DNA probes. Banbury Report "DNA Probes" pp 49-55 (1986).

Warburton, D., Anyane-Yeboa, K., Francke, U.: Mosaic tetrasomy 12p: Four new cases, and confirmation of the chromosomal origin of the supernumerary chromosome in one of the original Pallister-Mosaic syndrome cases. *Am. J. Med. Genet.* **27**:275-283 (1987).

Chenevix-Trench, G., Cowan, J.M., Behm, F.G., Goorha, R., Brown, J.A., Westin, E.H., Francke, U.: Cytogenetic and molecular genetic studies of a patient with atypical lymphoid hyperplasia. *Cancer Genet. Cytogenet.* **27**:251-259 (1987).

Francke, U.: Microdeletions and mendelian phenotypes. *7th Int. Congr. Human Genet.*, Berlin 1986. Springer-Verlag pp. 201-210 (1987).

Cowan, J.M., Halaban, R., Francke, U.: Cytogenetic analysis of melanocytes from premalignant nevi and melanomas. *J. Natl. Cancer Inst.* **80:1159-1164 (1988).**

Cowan, J.M., Francke, U.: Cytogenetic analysis in melanoma and nevi. In *Melanoma Research: Genetics Growth Factors, Metastases, and Antigens* (L. Nathanson, ed.), in Cancer Treatment and Research Series (W. McGuire, series ed.), Kluwer Academic Publishers, Boston, MA (1991).

Lindgren, V., Bryke, C.R., Özçelik, T., Yang-Feng, T.L., Francke, U.: Phenotypic, cytogenetic, and molecular studies of three patients with constitutional deletions of chromosome 5 in the region of the gene for familial adenomatous polyposis *Am. J. Hum. Genet.* **50**:988-997 (1992).

Francke, U.: Digitized and differentially shaded human chromosome ideograms for genomic applications. *Cytogenet. Cell Genet.* **65:206-218 (1994).**

Peoples, R., Milatovich, A., Francke, U.: Hemizygoty at the insulin-like growth factor 1 receptor (IGF1R) locus and growth failure in the ring chromosome 15 syndrome. *Cytogenet. Cell Genet.* **70:228-234 (1995).**

Laureys, G., Speleman, F., Versteeg, R., van der Drift, P., Chan, A., Leroy, J., Francke, U., Opdenakker, G., Van Roy, N.: Constitutional translocation t(1;17)(p36.31-p36.13;q11.2-q12.1) in a neuroblastoma patient. Establishment of somatic cell hybrids and identification of PND/A12M2 on chromosome 1 and NF1/SCYA7 on chromosome 17 as breakpoint flanking single copy markers. *Oncogene* **10**:1087-1093 (1995).

Maresco, D.L., Chang, E., Theil, K.S., Francke, U., Anderson, C.L.: The three genes of the human FCGR1 gene family encoding FcγRI flank the centromere of chromosome 1 at 1p12 and 1q21. *Cytogenet. Cell Genet.* **73**:157-163 (1996).

II. MOUSE CYTOGENETICS

Francke, U., Nesbitt, M.: Identification of the mouse chromosomes by quinacrine mustard staining. *Cytogenetics* **10:356-366 (1971).**

Nesbitt, M., Francke, U.: Linkage groups II and XII of the mouse: Cytological localization by fluorochrome staining. *Science* **174**:60-61 (1971).

Francke, U., Nesbitt, M.: Cattanach's translocation: Cytological characterization by quinacrine mustard staining. *Proc. Natl. Acad. Sci. USA* **68**:2918-2920 (1971).

Nesbitt, M., Francke, U.: Analysis of the T(3;?)6Ca and T(14;17)264Ca translocations in the mouse by quinacrine mustard staining. *Genetics* **69**:517-522 (1971).

Eicher, E.M., Nesbitt, M.N., Francke, U.: Cytological identification of the chromosomes involved in Searle's translocation and the location of the centromere in the X chromosome of the mouse. *Genetics* **71**:643-648 (1972).

Lerner, R.A., Jensen, F., Kennel, S.J., Dixon, F.J., Desroches, G., Francke, U.: Karyotypic, virologic and immunologic analyses of two continuous lymphocyte lines established from New Zealand Black mice: Possible relationship of lymphocyte mosaicism to autoimmunity. *Proc. Natl. Acad. Sci. USA* **69**:2965-2969 (1972).

Nesbitt, M.N., Francke, U.: A system of nomenclature for band patterns of mouse chromosomes. *Chromosoma* **41:145-158 (1973).**

Russell, S.W., Francke, U., Buettner, L., Cochrane, C.G.: Modes of growth and spread of a transplantable, virus-producing murine (Moloney) sarcoma: Karyotypic analyses. *J. Nat. Canc. Inst.* **53**:801-806 (1974).

Gazdar, A.F., Oie, H., Lalley, P., Moss, W.W., Minna, J.D., Francke, U.: Identification of mouse chromosomes required for murine leukemia virus replication. *Cell* **11**:949-956 (1977).

Halaban, R., Nordlund, J., Francke, U., Moellmann, G., Eisenstadt, J.M.: Supermelanotic hybrids derived from mouse melanomas and normal mouse cells. *Somat. Cell Genet.* **6**:29-44 (1980).

George, D.L., Francke, U.: Homogeneously staining chromosome regions and double minutes in a mouse adrenocortical tumor cell line. *Cytogenet. Cell Genet.* **28:217-226 (1980).**

Trowsdale, J., Hoch, J.A., Francke, U.: A methotrexate-resistant sub-line of mouse L1210 leukemia cells containing high levels of dihydrofolate reductase and with a homogeneously staining region on chromosome 4. *Oncodevelop. Biol. Med.* **1**:369-374 (1980).

Berenson, R.J., Francke, U., Dolnick, B.J., Bertino, J.R.: Karyotypic analysis of methotrexate-resistant and sensitive L5178Y cells. *Cytogenet. Cell Genet.* **29**:143-152 (1981).

Francke, U., Hsieh, C.-L., Kelly, D., Lai, E., Popko, B.: Induced reciprocal translocation in transgenic mice near sites of transgene integration. *Mamm. Genome* **3:209-216 (1992).**

III. HUMAN GENE MAPPING

Francke, U., Bakay, B., Conner, J.D., Coldwell, J.G., Nyhan, W.L.: Linkage relationships of X-linked enzymes glucose-6-phosphate dehydrogenase and hypoxanthine guanine phosphoribosyltransferase: Recombination in female offspring of compound heterozygotes. *Am. J. Hum. Genet.* **26:512-522 (1974).**

Francke, U.: Regional localization of the human genes for malate dehydrogenase-1 and isocitrate dehydrogenase-1 on chromosome 2 by interspecific hybridization using human cells with the balanced reciprocal translocation t(1;2)(q32;q13). *Cytogenet. Cell Genet.* **14**:308-312 (1975).

Francke, U., Busby, N.: Assignments of the human genes for lactate dehydrogenase-A and thymidine kinase to specific chromosomal regions. *Cytogenet. Cell Genet.* **14**:313-319 (1975).

Francke, U., Busby, N., Shaw, D., Hansen, S., Brown, M.G.: Intra-chromosomal gene mapping in man: Assignment of nucleoside phosphorylase to region 14cen→14q21 by interspecific hybridization of cells with a t(X;14)(p22;q21) translocation. *Somat. Cell Genet.* 2:27-40 (1976).

Buck, D.W., Bodmer, W.F., Bobrow, M. Francke, U.: The gene for the species antigen on human chromosome 11 is on the short arm. *Cytogenet. Cell Genet.* 16:97-98 (1976).

Busby, N., Courval, J., Francke, U.: Regional assignments of the genes for fumerate hydratase and guanylate kinase on chromosome 1 and for lysosomal acid phosphatase and esterase A4 on chromosome 11. *Cytogenet. Cell Genet.* 16:105-107 (1976).

Francke, U.: Retinoblastoma and chromosome 13. *Cytogenet. Cell Genet.* 16:131-134 (1976).

Francke, U., Busby, N., Shaw, D., Hansen, S., Brown, M.G.: Assignment of the *nucleoside phosphorylase (NP)* gene locus to region 14pter→14q21. *Cytogenet. Cell Genet.* 16:135-137 (1976).

Francke, U.: The human gene for β glucuronidase is on chromosome 7. *Am. J. Hum. Genet.* 28:357-362 (1976).

George, D.L., Francke, U.: Gene dose effect: Regional mapping of human nucleoside phosphorylase on chromosome 14. *Science* 194:851-852 (1976).

George, D.L., Francke, U.: Gene dose effect: Regional mapping of human glutathione reductase on chromosome 8. *Cytogenet. Cell Genet.* 17:282-286 (1976).

Francke, U., Pellegrino, M.A.: Assignment of the major histocompatibility complex to a region of the short arm of human chromosome 6. *Proc. Natl. Acad. Sci. USA* 74:1147-1151 and 5776 (correction) (1977).

Francke, U., Denney, R.M., Ruddle, F.H.: Intrachromosomal gene mapping in man: The gene for tryptophanyl-tRNA synthetase maps in region q21→qter of chromosome 14. *Somat. Cell Genet.* 3:381-389 (1977).

Francke, U., George, D.L., Brown, M.G., Riccardi, V.M.: Gene dose effect: Intraband mapping of the *LDH A* locus using cells from four individuals with different interstitial deletions of 11p. *Cytogenet. Cell Genet.* 19:197-207 (1977).

Francke, U., George, D.L., Pellegrino, M.A.: Regional mapping of gene loci on human chromosomes 1 and 6 by interspecific hybridization of cells with a t(1;6)(p3200;p2100) translocation and by correlation with linkage data. In *Human Cytogenetics: ICN-UCLA Symposia on Molecular and Cellular Biology*, Vol. VII, pp. 201-216, Academic Press, New York (1977).

George, D.L., Francke, U.: Regional mapping of human genes for hexosaminidase B and diphtheria toxin sensitivity on chromosome 5 using mouse x human hybrid cells. *Somat. Cell Genet.* 3:629-638 (1977).

George, D.L., Francke, U.: Regional mapping of human genes for phosphoglucomutase-1 on chromosome 1 and β -glucuronidase on chromosome 7 using mouse x human hybrids. *Hum. Hered.* 28:161-170 (1978).

Brown, S., Oie, H.K., Francke, U., Gazdar, A.F., Minna, J.D.: Assignment of a gene required for infection with endogenous baboon virus to human chromosome 19. *Cytogenet. Cell Genet.* **22**:239-242 (1978).

Weitkamp, L.R., Francke, U.: Report of the committee on the genetic constitution of chromosome 6. Winnipeg Conference (1977). *Cytogenet. Cell Genet.* **22**:92-105 (1978).

Francke, U., George, D.L.: Precise mapping of genes for phosphoglucomutase-1 and uridine monophosphate kinase on the short arm of human chromosome 1. *Cytogenet. Cell Genet.* **22**:384-388 (1978).

Francke, U., Brown, S.: Regional assignment of genes for phosphoglucomutase-2 and peptidase S to 4pter→4q21 in man. *Cytogenet. Cell Genet.* **22**:401-405 (1978).

George, D.L., Francke, U.: Evidence for localization of the gene for hexosaminidase B to the cen→q13 region of human chromosome 5 using mouse x human hybrid cells. *Cytogenet. Cell Genet.* **22**:408-411 (1978).

George, D.L., Francke, U.: Regional mapping of β -glucuronidase (β GUS) on human chromosome 7. *Cytogenet. Cell Genet.* **22**:437-440 (1978).

Oliver, N., Francke, U., Pellegrino, M.A.: Regional assignment of genes for mannose phosphate isomerase, pyruvate kinase-3 and β 2-microglobulin expression on human chromosome 15 by hybridization of cells from a t(15;22) (q14;q13.3) translocation carrier. *Cytogenet. Cell Genet.* **22**:506-510 (1978).

Brown, S., Oie, H.K., Gazdar, A.F., Minna, J.D., Francke, U.: Requirement of human chromosomes 19, 6 and possibly 3 for infection of hamster x human hybrid cells with Baboon M7 type C virus. *Cell* **18**:135-143 (1979).

Jeffreys, A.J., Craig, I.W., Francke, U.: Localization of the $G\gamma$ -, $A\gamma$ -, δ - and β -globin genes on the short arm of human chromosome 11. *Nature* **281:606-608 (1979).**

Francke, U., Weitkamp, L.R.: Report of the committee on the genetic constitution of chromosome 6. *Cytogenet. Cell Genet.* **25**:32-38 (1979).

Sanders-Haigh, L., Anderson, W.F., Francke, U.: The β -globin gene is on the short arm of human chromosome 11. *Nature* **283:683-686 (1980).**

Cox, D.R., Francke, U., Epstein, C.J.: Assignment of genes to the human X chromosome by the two-dimensional electrophoretic analysis of total cell proteins from rodent-human somatic cell hybrids. *Am. J. Hum. Genet.* **33**:495-512 (1981).

Pintar, J.E., Barbosa, J., Francke, U., Castiglione, C.M., Hawkins, Jr., M., Breakefield, X.O.: Gene for monoamine oxidase type A assigned to the human X chromosome. *J. Neuroscience* **1**:166-175 (1981).

Francke, U., Francke, B.: Requirement of the human chromosome 11 long arm for replication of herpes simplex virus type 1 in non-permissive Chinese hamster x human diploid fibroblast hybrids. *Somat. Cell Genet.* **7:171-191 (1981).**

George, D.L., Phillips III, J.A., Francke, U., Seeburg, P.H.: The genes for growth hormone and chorionic somatomammotropin are on the long arm of human chromosome 17 in region q21→qter. *Hum. Genet.* **57**:138-141 (1981).

Vora, S., Francke, U.: Assignment of the human gene for liver-type 6-phosphofructokinase isozyme (*PFKL*) to chromosome 21 by using somatic cell hybrids and monoclonal anti-L antibody. *Proc. Natl. Acad. Sci. USA* **78**:3738-3742 (1981).

Gehring, U., Francke, U.: Glucocorticoid sensitivity, specific receptors, and chromosome segregation in hybrid lymphoma cells. In *Hormones and Cell Regulation*, Vol. 5 (J.E. Dumont, J. Nunez, eds.), pp. 241-258, Elsevier/North Holland Biomed. Press (1981).

de Martinville, B., Wyman, A.R., White, R., Francke, U.: Assignment of the first random restriction fragment length polymorphism (RFLP) locus (*DI4SI*) to a region of human chromosome 14. *Am. J. Hum. Genet.* **34:216-226 (1982).**

Taggart, R.T., Francke, U.: Mapping of polypeptide genes by two-dimensional gel electrophoresis of hybrid cell extracts. *Cytogenet. Cell Genet.* **32:99-110 (1982).**

Vora, S., Durham, S., de Martinville, B., George, D.L., Francke, U.: Assignment of the human gene for muscle-type phosphofructokinase (*PFKM*) to chromosome 1 (region cen→q32) using somatic cell hybrids and monoclonal anti-M antibody. *Somat. Cell Genet.* **8**:95-104 (1982).

Skolnick, M.H., Francke, U.: Report of the committee on human gene mapping by recombinant DNA techniques. *Cytogenet. Cell Genet.* **32**:194-204 (1982).

Hershfield, M.S., Francke, U.: The human genes for S-adenosylhomocysteine hydrolase and adenosine deaminase are syntenic on chromosome 20. *Science* **216**:739-742 (1982).

Francke, U.: Mapping the X chromosome. In *Genetic Analysis of the X Chromosome* (H.F. Epstein, S. Wolf, eds.), pp. 79-94, Plenum Press, New York (1982).

Page, D., de Martinville, B., Barker, D., Wyman, A., White, R., Francke, U., Botstein, D.: Single-copy sequence hybridizes to polymorphic and homologous loci on the human X and Y chromosomes. *Proc. Natl. Acad. Sci. USA* **79**:5352-5356 (1982).

Francke, U., de Martinville, B.: Mapping of DNA sequences to chromosome regions in somatic cell hybrids. In *Banbury Report: Recombinant DNA Applications to Human Disease* (R. White, T. Caskey, eds.), pp. 175-187, Cold Spring Harbor Laboratory (1983).

de Martinville, B., Giacalone, J., Shih, C., Weinberg, R.A., Francke, U.: Oncogene from human EJ bladder carcinoma is located on the short arm of chromosome 11. *Science* **219:498-501 (1983).**

Francke, U., Foellmer, B.E., Haynes, B.F.: Chromosome mapping of human cell surface molecules: Monoclonal anti-human lymphocyte antibodies 4F2, A3D8 and A1G3 define antigens controlled by different regions of chromosome 11. *Somat. Cell Genet.* **9**:333-344 (1983).

Vora, S., Miranda, A.F., Hernandez, E., Francke, U.: Regional assignment of the human gene for platelet-type phosphofructokinase (*PFKP*) to chromosome 10p: Novel use of polyspecific rodent antisera to localize human enzyme genes. *Hum. Genet.* **63**:374-379 (1983).

Haynes, B.F., Telen, M.J., Harden, E.A., Francke, U., Palker, T.J.: Acquisition of a novel T cell surface protein during intrathymic T cell maturation encoded for by the short arm of chromosome 11. In *Human Leukocyte Markers Detected By Monoclonal Antibodies* (A. Bernard, L. Bounsell, eds.), Springer Verlag, New York (1983).

Francke, U., de Martinville, B.: Mapping of restriction fragment length polymorphisms (RFLP loci) to human chromosomes in somatic cell hybrids. In *Recombinant DNA and Medical Genetics* (A. Messer, I.A. Porter, eds.), pp. 21-34, Academic Press (1983).

Pearson, S.J., Tetri, P., George, D.L., Francke, U.: Activation of human α 1-antitrypsin gene in rat hepatoma x human fetal liver cell hybrids depends on presence of human chromosome 14. *Somat. Cell Genet.* 9:567-592 (1983).

de Martinville, B., Cunningham, J.M., Murray, M.J., Francke, U.: The N-*ras* oncogene assigned to the short arm of human chromosome 1. *Nucleic Acid Res.* 11:5267-5275 (1983).

de Martinville, B., Francke, U.: The c-Ha-*ras* 1, insulin and β -globin loci map outside the deletion associated with aniridia-Wilms tumour. *Nature* 305:641-643 (1983).

Francke, U., de Martinville, B., Coussens, L., Ullrich, A.: The human gene for the β -subunit of nerve growth factor is located on the proximal short arm of chromosome 1. *Science* 222:1248-1251 (1983).

de Martinville, B., Schafer, M., White, R., Francke, U.: Chromosomal assignments of three random RFLP loci defined by basepair changes in *Msp*I sites. *Molec. Biol. Med.* 1:415-424 (1983).

Skovby, F., Krassikoff, N., Francke, U.: Assignment of the gene for cystathionine β -synthase to human chromosome 21 in somatic cell hybrids. *Hum. Genet.* 65:291-294 (1984).

Francke, U., Brown, M.S., Goldstein, J.L.: Assignment of the human gene for the low density lipoprotein receptor to chromosome 19: Synteny of a receptor, a ligand, and a genetic disease. *Proc. Natl. Acad. Sci. USA* 81:2826-2830 (1984).

Brissenden, J.E., Ullrich, A., Francke, U.: Human chromosomal mapping of genes for insulin-like growth factors I and II and epidermal growth factor. *Nature* 310:781-784 (1984).

Lindgren, V., de Martinville, B., Horwich, A.L., Rosenberg, L.E., Francke, U.: Human ornithine transcarbamylase locus mapped to band Xp21.1 near the Duchenne muscular dystrophy locus. *Science* 226:698-700 (1984).

Aula, P., Astrin, K.H., Francke, U., Desnick, R.J.: Assignment of the structural gene encoding human aspartylglucosaminidase to the long arm of chromosome 4 (4q21-4qter). *Am. J. Hum. Genet.* 36:1215-1224 (1984).

Münke, M., Lindgren, V., de Martinville, B., Francke, U.: Comparative analysis of mouse-human hybrids with rearranged chromosome 1 by *in situ* hybridization and Southern blotting: High-resolution mapping of *NRAS*, *NGFB*, and *AMY* on human chromosome 1. *Somat. Cell Molec. Genet.* 10:589-599 (1984).

Lindgren, V., Ares Jr., M., Weiner, A.M., Francke, U: Human genes for U2 small nuclear RNA map to a major adenovirus 12 modification site on chromosome 17. *Nature* **314**:115-116 (1985).

Gehring, U., Segnitz, B., Foellmer, B., Francke, U: Assignment of the human gene for the glucocorticoid receptor to chromosome 5. *Proc. Natl. Acad. Sci. USA* **82**:3751-3755 (1985).

Engström, Y., Francke, U.: Assignment of the structural gene for subunit M1 of human ribonucleotide reductase to the short arm of chromosome 11. *Exptl. Cell Res.* **158**:477-483 (1985).

Cox, D.W., Francke, U.: Direct assignment of orosomucoid to human chromosome 9 and α 2HS-glycoprotein to chromosome 3 using human fetal liver x rat hepatoma hybrids. *Hum. Genet.* **70**:109-115 (1985).

Lindgren, V., Bernstein, L.B., Weiner A.M., Francke, U.: Human U1 small nuclear RNA pseudogenes do not map to the site of the U1 genes in 1p36 but are clustered in 1q12-q22. *Molec. Cell. Biol.* **5**:2172-2180 (1985).

Brissenden, J.E., Derynck, R., Francke, U.: Mapping of transforming growth factor α gene on human chromosome 2 close to the breakpoint of Burkitt's lymphoma t(2;8) variant translocation. *Cancer Res.* **45**:5593-5597 (1985).

Yang-Feng, T.L., Francke, U., Ullrich, A.: Gene for human insulin receptor: Localization to site on chromosome 19 involved in pre-B-cell leukemia. *Science* **228:728-731 (1985).**

Schechter A.L., Hung M-C., Vaidyanathan, L., Weinberg R., Yang-Feng, T.L., Francke, U., Ullrich A., Coussens, L.: The *neu* gene: An *erb-B* homologous gene distinct from and unlinked to the gene encoding the EGF receptor. *Science* **229**:976-978 (1985).

Yang-Feng, T.L., Floyd-Smith, G., Nemer, M., Drouin, J., Francke, U.: The pronatriodilatin gene is located on the distal short arm of human chromosome 1 and on mouse chromosome 4. *Am. J. Hum. Genet.* **37**:1117-1128 (1985).

Lindgren, V., Luskey, K.L., Russell, D.W., Francke, U.: Human genes involved in cholesterol metabolism: Chromosomal mapping of the loci for the low density lipoprotein receptor and 3-hydroxy-3-methylglutaryl-coenzyme A reductase with cDNA probes. *Proc. Natl. Acad. Sci. USA* **82:8567-8571 (1985).**

Coussens, L., Yang-Feng, T.L., Liao, Y.-C., Chen, E., Gray, A., McGrath, J., Seeburg, P.H., Libermann, T.A., Schlessinger, J., Francke, U., Levinson, A., and Ullrich, A.: Tyrosine kinase receptor with extensive homology to EGF receptor shares chromosomal location with *neu* oncogene. *Science* **230**:1132-1139 (1985).

Kraus, J.P., Williamson, C.L., Firgaira, F.A., Yang-Feng, T.L., Münke, M., Francke, U., Rosenberg, L.E.: Cloning and screening with nanogram amounts of immunopurified messenger RNAs: cDNA cloning and chromosomal mapping of cystathionine β -synthase and the β subunit of propionyl-CoA carboxylase. *Proc. Natl. Acad. Sci. USA* **83**:2047-2051 (1986).

Floyd-Smith, G., de Martinville, B., Francke, U.: An expressed β -tubulin gene, *TUBB*, is located on the short arm of human chromosome 6 and two related sequences are dispersed on chromosomes 8 and 13. *Exptl. Cell Res.* **163**:539-548 (1986).

Yang-Feng, T.L., Seeburg, P.H., Francke, U.: The human luteinizing hormone-releasing hormone gene (*LHRH*) is located on the short arm of chromosome 8 (region 8p11.2→p21). *Somat. Cell Mol. Genet.* **12**:95-100 (1986).

Barton, D.E., Yang-Feng, T.L., Francke, U.: The human tyrosine aminotransferase gene mapped to the long arm of chromosome 16 (region 16q22→q24) by somatic cell hybrid analysis and *in situ* hybridization. *Hum. Genet.* **72**:221-224 (1986).

Fujii, D., Brissenden, J.E., Derynck, R., Francke, U.: Transforming growth factor β gene maps to human chromosome 19 long arm and to mouse chromosome 7. *Somat. Cell Molec. Genet.* **12**:281-288 (1986).

Yang-Feng T.L., Opendakker G., Volckaert G., Francke U.: Human tissue-type plasminogen activator gene located near chromosomal breakpoint in myeloproliferative disorder. *Am. J. Hum. Genet.* **39**:79-87 (1986).

Brissenden, J.E., Page, D.C., de Martinville, B., Trowsdale, J., Botstein, D., Francke, U.: Regional assignments of three polymorphic DNA segments on human chromosome 15. *Genet. Epidemiol.* **3**:231-239 (1986).

Yang-Feng, T.L., Landau, N.R., Baltimore, D., Francke, U.: The terminal deoxynucleotidyltransferase gene is located on human chromosome 10 (10q23→10q24) and on mouse chromosome 19. *Cytogenet. Cell Genet.* **43**:121-126 (1986).

Floyd-Smith, G., Whitehead, A.S., Colten, H.R., Francke, U.: The human C-reactive protein gene (*CRP*) and serum amyloid P component gene (*ACPS*) are located on the proximal long arm of chromosome 1. *Immunogenetics* **24**:171-176 (1986).

Yang-Feng, T.L., Bruns, G.A.P., Carroll, A.J., Simola, K.O.J., Francke, U.: Localization of the LDHA gene to 11p14→11p15 by *in situ* hybridization of an LDHA cDNA probe to two translocations with breakpoints in 11p13. *Hum. Genet.* **74**:331-334 (1986).

Colb, M., Yang-Feng, T., Francke U., Mermer B., Parkinson, D.R., Krontiris, T.G.: A variable tandem repeat locus mapped to chromosome band 10q26 is amplified and rearranged in leukocyte DNAs of two cancer patients. *Nucleic Acids Res.* **14**:7929-7937 (1986).

Matsubara Y., Kraus, J.P., Yang-Feng, T.L., Francke U., Rosenberg, L.E., Tanaka, K.: Molecular cloning of cDNAs encoding rat and human medium chain acyl-CoA dehydrogenase and assignment of the gene to human chromosome 1. *Proc. Natl. Acad. Sci. USA.* **83**:6543-6547 (1986).

Yarden, Y., Escobedo, J.A., Kuang, W.-J., Yang-Feng, T.L., Daniel, T.O., Tremble, P.M., Chen, E.Y., Ando, M.E., Harkins, R.N., Francke, U., Fried, V.A., Ullrich, A., Williams, L.T.: Structure of the receptor for platelet-derived growth factor helps define a family of closely related growth factor receptors. *Nature* **323:226-232 (1986).**

Francke, U., Yang-Feng, T.L., Brissenden, J.E., Ullrich, A.: Chromosomal mapping of genes involved in growth control. *Cold Spring Harbor Symp. Quant. Biol.* **51:855-866 (1986).**

Coussens, L., Parker, P.J., Rhee, L., Yang-Feng, T.L., Chen, E., Waterfield, M.D., Francke, U., Ullrich, A.: Multiple, distinct forms of bovine and human protein kinase C suggest diversity in cellular signaling pathways. *Science* **233:859-866 (1986).**

Ullrich, A., Gray, A., Tam, A.W., Yang-Feng, T., Tsubokawa, M., Collins, C., Henzel, W., Le Bon, T., Kathuria, S., Chen, E., Jacobs, S., Francke, U., Ramachandran, J., Fujita-Yamaguchi, Y.: Insulin-like growth factor I receptor primary structure: Comparison with insulin receptor suggests structural determinants that define functional specificity. *EMBO J.* 5:2503-2512 (1986).

Chen, E., Coussens, L. Liao, Y.-C., Smith, D., Yang-Feng, T.L., McGrath, J., van Beveren, C., Verma, I.M., Libermann, T.A., Schlessinger, J., Francke, U., Levinson, A., Ullrich, A.: Structural features of growth factor receptors with oncogenic potential. *Biochem. Soc. Symp.* 52:65-82 (1986).

Kobilka, B.K., Dixon, R.A.F., Frielle, T., Dohlman, H.G., Bolanowski, M.A., Sigal, I.S., Yang-Feng, T.L., Francke, U., Caron, M.G., Lefkowitz, R.J.: cDNA for the human β_2 -adrenergic receptor: A protein with multiple membrane-spanning domains and encoded by a gene whose chromosomal location shared is with that of the receptor for platelet-derived growth factor. *Proc. Natl. Acad. Sci. USA* 84:46-50 (1987).

Kobilka, B.K., Freille, T., Collins, S., Yang-Feng, T., Kobilka, T.S., Francke, U., Lefkowitz, R.J., Caron, M.G.: An intronless gene encoding a potential member of the family of receptors coupled to guanine nucleotide regulatory proteins. *Nature* 329:75-79 (1987).

Yarden, Y., Kuang, W.-J., Yang-Feng, T., Coussens, L., Munemitsu, S., Dull, T.J., Chen, E., Schlessinger, J., Francke, U., Ullrich, A.: Human proto-oncogene *c-kit*: A new cell surface receptor tyrosine kinase for an unidentified ligand. *EMBO J.* 6:3341-3351 (1987).

Kobilka, B.K., Matsui, H., Kobilka, T.S., Yang-Feng, T.L., Francke, U., Caron, M.G., Lefkowitz, R.J., Regan, J.W.: Cloning, sequencing, and expression of the gene coding for the human platelet α_2 -adrenergic receptor. *Science* 238:650-656 (1987).

Radna, R.L., Foellmer, B., Feldman, L.A., Francke, U., Ozer, H.L.: Restriction of human adenovirus replication in Chinese hamster cell lines and their hybrids with human cells. *Virus Research* 8:277-299 (1987).

Spritz, R.A., Strunk, K., Surowy, C.S., Hoch, S.O., Barton, D.E., Francke, U.: The human U1 70k snRNP protein: cDNA cloning, chromosomal localization, expression, and alternative splicing and RNA-binding. *Nucleic Acids Res.* 15:10373-10391 (1987).

Kraus, J.P., Matsubara, Y., Barton, D., Yang-Feng, T.L., Glassberg, R., Ito, M., Ikeda, Y., Mole, J., Francke, U., Tanaka, K.: Isolation of cDNA clones coding for rat isovaleryl CoA dehydrogenase and assignment of the gene to human chromosome 15. *Genomics* 1:264-269 (1987).

Yang-Feng, T.L., Schneider, J.W., Lindgren, V., Shull, M.M., Benz, Jr., E.J., Lingrel, J.B., Francke, U.: Chromosomal localization of human Na⁺, K⁺-ATPase α - and β subunit genes. *Genomics* 2:128-138 (1988).

Alonso, M.A., Barton, D.E., Francke, U.: Assignment of the T-cell differentiation gene *MAL* to human chromosome 2, region cen \rightarrow q13. *Immunogenetics* 27:91-95 (1988).

Mangin, M., Webb, A.C., Dreyer, B.E., Posillico, J.T., Ikeda, K., Weir, E.C., Stewart, A.F., Bander, N.H., Milstone, L., Barton, D.E., Francke, U., Broadus, A.E.: Identification of a cDNA

encoding a parathyroid hormone-like peptide from a human tumor associated with humoral hypercalcemia of malignancy. *Proc. Natl. Acad. Sci. USA* **85**:597-601 (1988).

Leto, T.L., Fortugno-Erikson, D., Barton, D., Yang-Feng, T.L., Francke, U., Harris, A.S., Morrow, J.S., Marchesi, V.T., Benz, Jr., E.J.: Comparison of nonerythroid α -spectrin genes reveals strict homology among diverse species. *Molec. Cell Biol.* **8**:1-9 (1988).

Münke, M., Foellmer, B., Watkins, P.C., Cowan, J.M., Carroll, A.J., Gusella, J.F., Francke, U.: Regional assignment of six polymorphic DNA sequences on chromosome 21 by *in situ* hybridization to normal and rearranged chromosomes. *Am. J. Hum. Genet.* **42**:542-549 (1988).

Richmond, A., Balentien, E., Thomas, H.G., Flaggs, G., Barton, D.E., Spiess, J., Bordoni, R., Francke, U., Derynck, R.: Molecular characterization and chromosomal mapping of melanoma growth stimulatory activity, a growth factor structurally related to β -thromboglobulin. *EMBO J.* **7**:2025-2033 (1988).

Zeviani, M., Darras, B.T., Rizzuto, R., Salvati, G., Betto, R., Bonilla, E., Miranda, A.F., Du, J., Samitt, C., Dickson, G., Walsh, F.S., DiMauro, S., Francke, U., Schon E.A.: Cloning and expression of human nebulin cDNAs and assignment of the gene to chromosome 2q31-q32. *Genomics* **2**:249-256 (1988).

Barton, D.E., Kwon, B.S., Francke, U.: Human tyrosinase gene, mapped to chromosome 11 (q14→q21), defines second region of homology with mouse chromosome 7. *Genomics* **3**:17-24 (1988).

Ruta, M., Howk, R., Ricca, G., Drohan, W., Zabelshansky, M., Laureys, G., Barton, D.E., Francke, U., Schlessinger, J., Givol, D.: A novel protein kinase gene whose expression is modulated during endothelial cell differentiation. *Oncogene* **3**:9-15 (1988).

Brack-Werner, R., Barton, D.E., Werner, T., Foellmer, B.E., Leib-Mösch, C., Francke, U., Erfle, V., Hehlmann, R.: Human SSAV-related endogenous retroviral element: LTR-like sequence and chromosomal localization to 18q21. *Genomics* **4**:68-75 (1989).

Fodor, W.L., Darras, B., Seharaseyon, J., Falkenthal, S., Francke, U., Vanin, E.F.: Human ventricular/slow twitch myosin alkali light chain gene characterization, sequence and chromosomal location. *J. Biol. Chem.* **264**:2143-2149 (1989).

Staunton, D.E., Fisher, R.C., LeBeau, M.M., Lawrence, J.B., Barton, D.E., Francke, U., Dustin, M., Thorley-Lawson, D.A.: Blast-1 possesses a glycosyl-phosphatidylinositol (GPI) membrane anchor, is related to LFA-3 and OX-45, and maps to chromosome 1q21-q23. *J. Exp. Med.* **169**:1087-1099 (1989).

Leib-Mösch, C., Barton, D., Geigl, E.-M., Brack-Werner, R., Erfle, V., Hehlmann, R., Francke, U.: Two RFLPs associated with the human endogenous retroviral element S71 on chromosome 18q21. *Nucleic Acids Res.* **17**:2367 (1989).

Rozen, R., Barton, D., Du, J., Hum, D.W., MacKenzie, R.E., Francke, U.: Chromosomal localization of the gene for the human trifunctional enzyme, methylenetetrahydrofolate dehydrogenase-methenyltetrahydrofolate cyclohydrolase-formyltetrahydrofolate synthetase. *Am. J. Hum. Genet.* **44**:781-786 (1989).

Francke, U., Foellmer, B.E.: The glucocorticoid receptor gene is in 5q31-q32. *Genomics* **4**:610-612 (1989).

- Rizzuto, R., Nakase, H., Darras, B.T., Francke, U., Fabrizi, G.M., Mengel, T., Walsh, F., Kadenbach, B., DiMauro, S., Schon, E.A.: A gene specifying subunit VIII of human cytochrome *c* oxidase is localized to chromosome 11 and is expressed in both muscle and non-muscle tissues. *J. Biol. Chem.* **264**:10595-10600 (1989).
- Battat, L., Francke, U.: A common SacI polymorphism in the gene for the M1 subunit of ribonucleotide reductase (RRM1). *Nucleic Acids Res.* **17**:4005 (1989).
- Francke, U., Darras, B.T., Zander, N.F., Kilimann, M.W.: Assignment of human genes for phosphorylase kinase subunits α (PHKA) to Xq12-q13 and β (PHKB) to 16q12-q13. *Am. J. Hum. Genet.* **45**:276-282 (1989).**
- Yang-Feng, T.L., Xue, F., Zhong, W., Cotecchia, S., Frielle, T., Caron, M.G., Lefkowitz, R.J., Francke, U.: Chromosomal organization of adrenergic receptor genes. *Proc. Natl. Acad. Sci. USA* **87**:1516-1520 (1990).**
- Lomax, M.I., Welch, M.D., Darras, B.T., Francke, U., and Grossman, L.I. Novel use of a chimpanzee pseudogene for chromosomal mapping of human cytochrome *c* oxidase subunit IV. *Gene* **86**:209-216 (1990).
- Labeit, S., Barlow, D.P., Gautel, M., Gibson, T., Holt, J., Hsieh, C.-L., Francke, U., Leonard, K., Wardale, J., Whiting, A., Trinick, J.: A regular pattern of two types of 100-residue motif in the sequence of titin. *Nature* **345**:273-276 (1990).
- Seharaseyon, J., Bober, E., Hsieh, C.-L., Fodor, W.L., Francke, U., Arnold, H.-H., Vanin, E.F.: Human embryonic/atrial myosin alkali light chain gene: Characterization, sequence, and chromosomal location. *Genomics* **7**:289-293 (1990).
- Ferrero, E., Hsieh, C.-L., Francke, U., Goyert, S.M.: CD14 is a member of the family of leucine rich proteins and is encoded by a gene syntenic with multiple receptor genes. *J. Immunol.* **145**:311-336 (1990).
- Hsieh, C.-L., Swaroop, A., Francke, U.: Chromosomal localization and cDNA sequence of human *ralB*, a GTP binding protein. *Somat Cell Molec. Genet.* **16**:407-410 (1990).
- Cancela, L., Hsieh, C.-L., Francke, U., Price, P.A.: Molecular structure, chromosome assignment, and promoter organization of the human matrix Gla protein gene. *J. Biol. Chem.* **265**:15040-15048 (1990).
- Francke, U., Hsieh, C.-L., Foellmer, B.E., Lomax, K.J., Malech, H.L., Leto, T.L.: Genes for two autosomal recessive forms of chronic granulomatous disease assigned to 1q25 (NCF2) and 7q11.23 (NCF1). *Am. J. Hum. Genet.* **47**:483-492 (1990).**
- Archer III, B.T., Özçelik, T., Jahn, R., Francke, U., Südhof, T.C.: Structures and chromosomal localizations of two human genes encoding synaptobrevins 1 and 2. *J. Biol. Chem.* **265**:17267-17273 (1990).
- Perin, M.S., Johnston, P.A., Özçelik, T., Jahn, R., Francke, U., Südhof, T.C.: Structural and functional conservation of synaptotagmin (p65) in *Drosophila* and humans. *J. Biol. Chem.* **266**:615-622 (1991).

Keats, B.J.B., Sherman, S.L., Morton, N.E., Robson, E.B., Buetow, K.H., Cartwright, P.E., Chakravarti, A., Francke, U., Green, P.P., Ott, J.: Guidelines for Human Linkage Maps: An international system for human linkage maps (ISLM, 1990). *Genomics* 9:557-560 (1991).

Lomax, M.I., Hsieh, C.-L., Darras, B.T., Francke, U.: Structure of the human cytochrome c oxidase subunit Vb gene and chromosomal mapping of the coding gene and of seven pseudogenes. *Genomics* 10:1-9 (1991).

Agarwal, N., Hsieh, C.-L., Sills, D., Swaroop, M., Desai, B., Francke, U., Swaroop, A.: Sequence analysis, expression and chromosomal localization of a gene, isolated from a subtracted human retina cDNA library, that encodes an insulin-like growth factor binding protein (IGFBP2). *Exp. Eye Res.* 52:549-561 (1991).

Tse, W.T., Menninger, J.C., Yang-Feng, T.L., Francke, U., Sahr, K.E., Lux, S.E., Ward, D.C., Forget, B.G.: Isolation and chromosomal localization of a novel non-erythroid ankyrin gene. *Genomics* 10:858-866 (1991).

Milatovich, A., Song, K., Heller, R.A., Francke, U.: Tumor necrosis factor receptor genes, *TNFR1* and *TNFR2*, on human chromosomes 12 and 1. *Somat. Cell Molec. Genet.* 17: 519-523 (1991).

von Hoegen, I., Hsieh, C.-L., Scharting, R., Francke, U., Parnes, J.R.: Identity of human Lyb-2 and CD72 and localization of the gene to chromosome 9. *European J. Immunol.* 21:1425-1431 (1991).

Andria, M.L., Hsieh, C.-L., Oren, R., Francke, U., Levy, S.: Genomic organization and chromosomal localization of the TAPA-1 gene. *J. Immunol.* 147:1030-1036 (1991).

Davidson, J.J., Özçelik, T., Hamacher, C., Willems, P.J., Francke, U., Kilimann, M.W.: cDNA cloning of a liver isoform of the phosphorylase kinase a subunit and mapping of the gene to Xp22.2-p22.1, the region of human X-linked liver glycogenosis. *Proc. Natl. Acad. Sci. USA.* 89:2096-2100 (1992).

Murphy, P.M., Özçelik, T., Kenney, R.T., Tiffany, H.L., McDermott, D., Francke, U.: A structural homologue of the N-formyl peptide receptor: Characterization and chromosome mapping of a peptide chemoattractant receptor family. *J. Biol. Chem.* 267:7637-7643 (1992).

Welch, S.K., Francke, U.: Assignment of the human α 2-plasmin inhibitor gene (PLI) to chromosome 17, region pter-p12, by PCR analysis of somatic cell hybrids. *Genomics* 13:213-214 (1992).

Berkemeier, L.R., Özçelik, T., Francke, U., Rosenthal, A.: Human chromosome 19 contains the neurotrophin-5 gene locus and three related genes that may encode novel acidic neurotrophins. *Somat. Cell Molec. Genet.* 18:233-245 (1992).

Milatovich, A., Francke, U.: Human cyclin B1 gene (*CCNB1*) assigned to chromosome 5 (q13 qter). *Somat. Cell Molec. Genet.* 18:303-307 (1992).

Thigpen, A.E., Davis, D.L., Milatovich, A., Mendonca, B.B., Imperato-McGinley, J., Griffin, J.E., Francke, U., Wilson, J.D., Russell, D.W.: The molecular genetics of steroid 5 α -reductase 2 deficiency. *J. Clin. Invest.* 90:799-809 (1992).

Arnaudo, E., Hirano, M., Seelan, R.S., Milatovich, A., Hsieh, C.-L., Fabrizi, G.M., Grossman, L.I., Francke, U., Schon, E.A.: Tissue-specific expression and chromosome assignment of genes specifying two isoforms of subunit VIIa of human cytochrome *c* oxidase. *Gene* **119**:299-305 (1992).

Morrissey, J., Tkachuk, D.C., Milatovich, A., Francke, U., Link, M., Cleary, M.L.: A serine/proline-rich protein is fused to HRX in t(4;11) acute leukemias. *Blood* **81**:1124-1131 (1993).

Wilgenbus, K.K., Milatovich, A., Francke, U., Furthmayr, H.: Molecular cloning, cDNA sequence and chromosomal assignment of the human radixin gene and two dispersed pseudogenes. *Genomics* 16:199-206 (1993).

Le Beau, M.M., Overhauser, J., Straub, R.E., Silverman, G., Gilliam, T.C., Ott, J., O'Connell, P., Francke, U., Geurts van Kessel, A.: Report of the first international workshop on human chromosome 18 mapping. *Cytogenet. Cell Genet.* **63**:78-96 (1993).

Gao, J.-L., Kuhns, D.B., Tiffany, H.L., McDermott, D., Li, X., Francke, U., Murphy, P.M.: Structure and functional expression of the human macrophage inflammatory protein-1 α (MIP 1a)/RANTES receptor. *J. Exp. Med.* **177**:1421-1427 (1993).

Andres, D.A., Milatovich, A., Özçelik, T., Wenzlau, J.M., Brown, M.S., Goldstein, J.L., Francke, U.: cDNA cloning of the two subunits of human CAAX farnesyltransferase and chromosomal mapping of FNTA and FNTB loci and related sequences. *Genomics* 18:105-112 (1993).

Hanna, Z., Jankowski, M., Tremblay, P., Jiang, X., Milatovich, A., Francke, U., Jolicoeur, P.: The *Vin-1* gene, identified by provirus insertional mutagenesis, is the cyclin D2. *Oncogene* **8**:1661-1666 (1993).

Ibraghimov-Beskrovnaya, O., Milatovich, A., Özçelik, T., Yang, B., Koepnick, K., Francke, U., Campbell, K.P.: Human dystroglycan: Skeletal muscle cDNA, genomic structure, origin of tissue specific isoforms and chromosomal localization. *Hum. Molec. Genet.* **2**:1651-1657 (1993).

Milatovich, A., Kitamura, T., Miyajima, A., Francke, U.: Gene for the α -subunit of human interleukin-3 receptor (IL3RA) localized to the X-Y pseudoautosomal region. *Am. J. Hum. Genet.* 53:1146-1153 (1993).

Leib-Mösch, C., Haltmeier, M., Werner, T., Geigl, E.-M., Brack-Werner, R., Francke, U., Erfle, V., Hehlmann, R.: Genomic distribution and transcription of solitary HERV-K LTRs in human tissues. *Genomics* **18**:261-269 (1993).

Karasawa, M., Zwacka, R.M., Reuter, A., Fink, T., Hsieh, C.L., Lichter, P., Francke, U., Weiher, H.: The human homolog of the glomerulosclerosis gene *Mpv17*: Structure and genomic organization. *Hum. Molec. Genet.* **2**:1829-1834 (1993).

Tachibana, M., Pérez-Jurado, L., Nakayama, A., Hodgkinson, C.A., Li, X., Schneider, M., Miki, T., Fex, J., Francke, U., Arnheiter, H.: Cloning of MITF, the human homolog of the mouse *microphthalmia* gene and assignment to human chromosome 3p14.1-p12.3. *Hum. Mol. Genet.* **3**:553-557 (1994).

Garcia, C.K., Li, X., Luna, J., Francke, U.: cDNA cloning of the human monocarboxylate transporter 1 and chromosomal localization of the SLC16A1 locus to 1p13.2-p12. *Genomics* **23**:500-503 (1994).

Francke, U.: Clinical and molecular cytogenetics and gene mapping: principles and techniques. Asia Pacific Conference on Medical Genetics, Bangkok, *Southeastern Asian Journal of Tropical Medicine and Public Health* **26**:34-43 (1995).

Moore, K.J., Testa, J.R., Francke, U., Milatovich, A., Copeland, N.G., Jenkins, N.A.: Cloning and regional assignment of the human myosin heavy chain 12 (*MYH12*) gene to chromosome 15q21. *Cytogenet. Cell Genet.* **69**:53-58 (1995).

Yamamoto, R., Li, X., Winter, S., Francke, U., Kilimann, M.: Primary structure of human amphiphysin, the dominant autoantigen of paraneoplastic Stiff-Man syndrome, and mapping of its gene (*AMPH*) to chromosome 7p14-p13. *Hum. Molec. Genet.* **4**:265-268 (1995).

Bowen, M.A., Patel, D.D., Li, X., Modrell, B., Malacko, A.R., Wang, W.-C. Marquardt, H., Neubauer, M., Pesando, J.M., Francke, U., Haynes, B.F., Aruffo, A.: Cloning, mapping and characterization of activated leukocyte-cell adhesion molecule (ALCAM), a CD6 ligand. *J. Exp. Med.* **181**:2213-2220 (1995).

Li, X., Francke, U.: Assignment of the gene SLC1A2 coding for the human glutamate transporter EAAT2 to human chromosome 11 bands p13-p12. *Cytogenet. Cell Genet.* **71**:212-213 (1995).

Yamamoto, R., Li, X., Francke, U., Kilimann, M.W.: Genetic mapping of the human amphiphysin gene () at 7p14-p13 excludes its involvement in retinitis pigmentosa 9 or dominant cystoid macular dystrophy. *Am. J. Hum. Genet.* **57**:970-972 (1995).

Schanen, N.C., Scherer, S.W., Tsui, L.-C., Francke, U.: Assignment of the 5-hydroxytryptamine (serotonin) receptor 5A gene (*HTR5A*) to human chromosome band 7q36.1. *Cytogenet. Cell Genet.* **72**:187-188 (1996).

Gratas, C., Li, X., Wang, Y.-K., Francke, U., Becker, D.: Chromosomal assignment of three human melanocyte-specific genes. *Int. J. Oncol.* **9**:481-485 (1996).

Giacalone, J., Li, X., Lehrach, H., Francke, U.: High-density radiation hybrid map of human chromosome 18 and contig of 18p. *Genomics* 37:9-18 (1996).

Larrick, J.W., Lee, J., Ma, S., Li, X., Francke, U., Wright, S.C., Balint, R.F.: Structural, functional analysis and localization of the human CAP18 gene. *FEBS Letters* **398**:74-80 (1996).

Li, L., Li, X., Francke, U., Cohen, S.N.: The *TSG101* tumor susceptibility gene is located in chromosome 11, band p15 and is mutated in human breast cancer. *Cell* 88:143-154 (1997).

Gebe, J., Kiener, P.A., Ring, H.Z., Li, X., Francke, U., Aruffo, A.: Molecular cloning, mapping to human chromosome 1 q21-q23, and cell binding characteristics of Sp α , a new member of the Scavenger Receptor Cysteine-rich (SRCR) family of proteins. *J Biol. Chem.* **272**:6151-6158 (1997).

Carmeci, C., Thompson, D.A., Ring, H.Z., Francke, U., Weigel, R.J.: Identification of a gene (GPR30) with homology to the G-protein-coupled receptor superfamily associated with estrogen receptor expression in breast cancer. *Genomics* 45:607-617 (1997).

Carmeci, C., Thompson, D.A., Ring, H.Z., Francke, U., Weigel, R.J.: A novel G protein-coupled receptor isolated from an estrogen receptor-positive breast carcinoma cDNA library. *Surgical Forum XLVIII*:841-843 (1997).

Hitoshi, Y., Lorens, J., Kitada, S.I., Fisher, J., LaBarge, M., Ring, H.Z., Francke, U., Reed, J.C., Kinoshita, S., Nolan, G.P.: Toso, a cell-surface, specific regulator of Fas-induced apoptosis in T cells. *Immunity* 8:461-471 (1998).

Ring, H.Z., Vameghi-Meyers, V., Wang, W., Crabtree, G.R., Francke, U.: Five SWI/SNF-related, matrix-associated, actin-dependent regulator of chromatin (SMARC) genes are dispersed in the human genome. *Genomics* 51:140-143 (1998).

Burwinkel, B., Miglierini, G., Jenne, D.E., Gilbert, D.J., Copeland, N.G., Jenkins, N.A., Ring, H.Z., Francke, U., Kilimann, M.W.: Structure of the human paralemmin gene (*PALM*), mapping to human chromosome 19p13.3 and mouse chromosome 10, and exclusion of coding mutations in *grizzled*, *mocha jittery* and *hesitant* mice. *Genomics* 49:462-66 (1998).

IV. HUMAN/MOUSE COMPARATIVE GENE MAPPING

Minna, J.D., Lalley, P.A., Francke, U.: Comparative mapping using somatic cell hybrids. *In Vitro* 12:726-733 (1976).

Lalley, P.A., Minna, J.D., Francke, U.: Conservation of autosomal gene synteny groups in mouse and man. *Nature* 274:160-162 (1978).

Lalley, P.A., Francke, U., Minna, J.D.: Comparative gene mapping: The linkage relationships of the homologous genes for phosphoglucomutase and peptidase S are conserved in man and mouse. *Cytogenet. Cell Genet.* 22:573-576 (1978).

Lalley, P.A., Francke, U., Minna, J.D.: Comparative gene mapping in man and mouse: Assignment of the genes for lactate dehydrogenase-A, peptidase D, and isocitrate dehydrogenase-2 to mouse chromosome 7. *Cytogenet. Cell Genet.* 22:577-580 (1978).

Lalley, P.A., Francke, U., Minna, J.D.: The genes coding for pyruvate kinase (M2) and mannosephosphate isomerase are linked in man and mouse. *Cytogenet. Cell Genet.* 22:581-584 (1978).

Francke, U., Taggart, R.T.: Comparative gene mapping: Order of loci on the X chromosome is different in mice and humans. *Proc. Natl. Acad. Sci. USA* 77:3595-3599 (1980).

Francke, U.: Cytogenetic approaches to mouse models of human genetic diseases. *Am. J. Pathol.* 101:S41-S51 (1980).

Francke, U., Tetri, P., Taggart, R.T., Oliver, N.: Conserved autosomal syntenic group on mouse (MMU) chromosome 15 and human (HSA) chromosome 22: Assignment of a gene for

arylsulfatase A to MMU 15 and regional mapping of *DIAL*, *ARSA* and *ACO2* on HSA 22. *Cytogenet. Cell Genet.* **31**:58-69 (1981).

Francke, U: Gene mapping on human and mouse chromosomes. *Genetics: New Frontiers*. Proceedings of the XV International Congress of Genetics. pp 443-454, Oxford & IBH Publishing Co., New Delhi, (1984).

Yang-Feng, T.L., DeGennaro, L.J., Francke, U.: Genes for synapsin I, a neuronal phosphoprotein, map to conserved regions of human and murine X chromosomes. *Proc. Natl. Acad. Sci. USA* **83:8679-8683 (1986).**

Bucan, M. Yang-Feng, T., Colberg-Poley, A.M., Wolgemuth, D.J., Guenet, J.-L., Francke, U., Lehrach, H.: Genetic and cytogenetic localisation of the homeo box containing genes on mouse chromosome 6 and human chromosome 7. *EMBO J.* **5**:2899-2905 (1986).

Yang-Feng, T.L., Barton, D.E., Thelander, L., Lewis, W.H., Srinivasan, P.R., Francke, U.: Ribonucleotide reductase M2 subunit sequences mapped to four different chromosomal sites in humans and mice: Functional locus identified by its amplification in hydroxyurea-resistant cell lines. *Genomics* **1:77-86 (1987).**

Münke, M., Francke, U.: The physical map of *Mus musculus* chromosome 11 reveals evolutionary relationships with different syntenic groups of genes in *Homo sapiens*. *J. Mol. Evol.* **25:134-140 (1987).**

Barton, D.E., Arquint, M., Roder, J., Dunn, R., Francke, U.: The myelin-associated glycoprotein gene: Mapping to human chromosome 19 and mouse chromosome 7 and expression in quivering mice. *Genomics* **1**:107-112 (1987).

Brissenden, J.E., Caras, I., Thelander, L., Francke, U.: The structural gene for the M1 subunit of ribonucleotide reductase maps to chromosome 11, band p15, in human and to chromosome 7 in mouse. *Exptl. Cell Res.* **174**:302-308 (1988).

Laureys, G., Barton, D.E., Ullrich, A., Francke, U.: Chromosomal mapping of the gene for the type II insulin-like growth factor receptor/cation-independent mannose 6-phosphate receptor in man and mouse. *Genomics* **3**:224-229 (1988).

Barton, D.E., Foellmer, B.E., Du, J., Tamm, J., Derynck, R., Francke, U.: Chromosomal mapping of genes for transforming growth factors β_2 and β_3 in man and mouse: Dispersion of TGF- β gene family. *Oncogene Research* **3**:323-331 (1988).

Münke, M., Kraus, J.P., Ohura, T., Francke, U.: The gene for cystathionine β -synthase (*CBS*) maps to the subtelomeric region on human chromosome 21q and to the proximal mouse chromosome 17. *Am. J. Hum. Genet.* **42**:550-559 (1988).

Barton, D.E., Yang-Feng, T.L., Mason, A.J., Seeburg, P.H., Francke, U.: Mapping of genes for inhibin subunits α , β_A and β_B on human and mouse chromosomes and studies of *jsd* mice. *Genomics* **5:91-99 (1989).**

Barton, D.E., Foellmer, B.E., Wood, W.I., Francke, U.: Chromosome mapping of the growth hormone receptor gene in man and mouse. *Cytogenet. Cell Genet.* **50:137-141 (1989).**

Hsieh, C.-L., Vogel, U.S., Dixon, R.A.F., Francke, U.: Chromosome localization and cDNA sequence of murine and human genes for *ras* p21 GTPase activating protein (GAP). *Somat. Cell Molec. Genet.* **15**:579-590 (1989).

Hsieh, C.-L., Donlon, T.A., Darras, B.T., Chang, D.D., Topper, J.N., Clayton, D.A., Francke, U.: The gene for the RNA component of the mitochondrial RNA-processing endoribonuclease is located on human chromosome 9p and on mouse chromosome 4. *Genomics* **6**:540-544 (1990).

Hsieh, C.-L., Sturm, R., Herr, W., and Francke, U.: The gene for the ubiquitous octamer-binding protein Oct-1 is on human chromosome 1, region cen-q32, and near *Ly* 22 and *Ltw-4* on mouse chromosome 1. *Genomics* **6:666-672 (1990).**

Levanon, D., Hsieh, C.-L., Francke, U., Dawson, P.A., Ridgway, N.D., Brown, M.S., Goldstein, J.L.: cDNA cloning of human oxysterol-binding protein and localization of the gene to human chromosome 11 and mouse chromosome 19. *Genomics* **7:65-74 (1990).**

Hsieh, C.-L., Cheng-Deutsch, A., Gloor, S., Schachner, M., Francke, U.: Assignment of the AMOG (adhesion molecule on glia) gene to mouse chromosome 11 near *Zfp-3* and *Asgr-1,2* and to human chromosome 17. *Somat. Cell Molec. Genet.* **16**:401-405 (1990).

Özçelik, T., Lafreniere, R.G., Archer III, B.T., Johnston, P.A., Willard, H.F., Francke, U., Südhof, T.C.: Synaptophysin: Structure of the human gene and assignment to the X chromosome in man and mouse. *Am. J. Hum. Genet.* **47:551-561 (1990).**

Affholter, J.A., Hsieh, C.-L., Francke, U., Roth, R.A.: Insulin-degrading enzyme: Stable expression of the human complementary DNA, characterization of its protein product, and chromosomal mapping of the human and mouse genes. *Mol. Endo.* **4**:1125-1135 (1990).

Kuo, J., Conley, P.B., Hsieh, C.-L., Francke, U., Crabtree, G.R.: Molecular cloning, functional expression and chromosomal localization of murine hepatocyte nuclear factor-1. *Proc. Natl. Acad. Sci. USA* **87**:9838-9842 (1990).

Hsieh, C.-L., Bowcock, A.M., Farrer, L.A., Hebert, J.M., Huang, K.N., Cavalli-Sforza, L.L., Julius, D., Francke, U.: The serotonin receptor subtype 2 locus *HTR2* is on human chromosome 13 near genes for esterase D and retinoblastoma-1 and on mouse chromosome 14. *Somat. Cell Molec. Genet.* **16**:567-574 (1990).

Hsieh, C.-L., Kumar, N.M., Gilula, N.B., Francke, U.: Distribution of genes for Gap junction membrane channel proteins on human and mouse chromosomes. *Somat. Cell Molec. Genet.* **17**:191-200 (1991).

Hofmann, S.L., Topham, M., Hsieh, C.-L., Francke, U.: cDNA and genomic cloning of HRC, a human sarcoplasmic reticulum protein, and localization of the gene to human chromosome 19 and mouse chromosome 7. *Genomics* **9**:656-669 (1991).

Özçelik, T., Rosenthal, A., Francke, U.: Chromosomal mapping of brain-derived neurotrophic factor and neurotrophin-3 genes in man and mouse. *Genomics* **10:569-575 (1991).**

Hsieh, C.-L., Navankasattusas, S., Escobedo, J.A., Williams, L.T., Francke, U.: Chromosomal localization of the gene for AA-type platelet-derived growth factor receptor (PDGFRA) in humans and mice. *Cytogenet. Cell Genet.* **56**:160-163 (1991).

Milatovich, A., Travis, A., Grosschedl, R., Francke, U.: Gene for lymphoid enhancer binding factor (LEF1) mapped to human chromosome 4 (q23-q25) and mouse chromosome 3 near *Egf*. *Genomics* 11:1040-1048 (1991).

Jenkins, E.P., Hsieh, C.-L., Milatovich, A., Normington, K., Berman, D.M., Francke, U., Russell, D.W.: Characterization and chromosomal mapping of human steroid 5 α -reductase gene and pseudogene and mapping of the mouse homologue. *Genomics* 11:1102-1112 (1991).

Özçelik, T., Porteus, M.H., Rubenstein, J.L.R., Francke, U.: *DLX2 (Tes-1)*, a homeobox gene of the *distal-less* family, assigned to conserved regions on human and mouse chromosomes 2. *Genomics* 13:1157-1162 (1992).

Rupp, F., Özçelik, T., Linial, M., Peterson, K., Francke, U., Scheller, R.: Structure and chromosomal localization of the mammalian agrin gene. *J. Neurosci.* 12:3535-3544 (1992).

Bascom, R.A., García-Heras, J., Hsieh, C.-L., Gerhard, D.S., Jones, C., Francke, U., Willard, H.F., Ledbetter, D.H., McInnes, R.R.: Localization of the photoreceptor gene *ROM1* to human chromosome 11 and mouse chromosome 19. Sublocalization to human 11q13 between *PGA* and *PYGM*. *Am. J. Hum. Genet.* 51:1028-1035 (1992).

Milatovich, A., Hsieh, C.-L., Bonaminio, G., Tecott, L., Julius, D., Francke, U.: Serotonin receptor 1c gene assigned to X chromosome in human (band q24) and mouse (bands D-F4). *Hum. Molec. Genet.* 1:681-684 (1992).

Milatovich, A., Mendel, D.B., Crabtree, G.R., Francke, U.: Genes for the dimerization cofactor of hepatocyte nuclear factor-1 α (DCOH) are on human and murine chromosomes 10. *Genomics* 16:292-295 (1993).

McKenzie, A.N.J., Li, X., Largaespada, D.A., Sato, A., Kaneda, A., Zurawski, S., Doyle, E., Milatovich, A., Francke, U., Copeland, N.G., Jenkins, N.A., Zurawski, G.: Structural comparison and chromosomal localization of the human and mouse interleukin-13 genes. *J. Immunol.* 150:5436-5444 (1993).

Wilgenbus, K.K., Hsieh, C.-L., Lankes, W.T., Milatovich, A., Francke, U., Furthmayr, H.: Structure and localization of the X chromosome of the gene coding for the human filopodial protein moesin. *Genomics* 19:326-333 (1994).

Li, X., Nghiem, P., Schulman, H., Francke, U.: Localization of the *CAMKG* gene encoding γ isoforms of multifunctional calcium/calmodulin-dependent protein kinase (CaM kinase) to human chromosome 10 band q22 and mouse chromosome 14. *Cytogenet. Cell Genet* 66:113-116 (1994).

Milatovich, A., Qiu, R.-G., Grosschedl, R., Francke, U.: Gene for a tissue-specific transcriptional activator (EBF or Olf-1), expressed in early B lymphocytes, adipocytes and olfactory neurons, is located on human chromosome 5, band q34, and proximal mouse chromosome 11. *Mamm. Genome* 5:211-215 (1994).

Milatovich, A., Bolger, G., Michaeli, T., Francke, U.: Chromosome localizations of genes for five cAMP-specific phosphodiesterases in man and mouse. *Somat. Cell Mol. Genet.* 20:75-86 (1994).

Li, X., Yin, W., Pérez-Jurado, L., Bonadio, J., Francke, U.: Mapping of human and murine genes for latent TGF- β binding protein-2 (LTBP2). *Mamm. Genome* **6**:42-45 (1995).

Li, X., Ho, S.N., Luna J., Giacalone, J., Thomas, D.J., Timmerman, L.A., Crabtree, G.R., Francke, U.: Cloning and chromosomal localization of the human and murine genes for the T-cell transcription factors NFATc and NFATp. *Cytogenet. Cell Genet.* **68:185-191 (1995).**

Li, X., Rosahl, T.W., Südhof, T.C., Francke, U.: Mapping of synapsin II (SYN2) genes to human chromosome 3p and mouse chromosome 6 band F. *Cytogenet. Cell Genet.* **71**:301-305 (1995).

Li, X., Luna, J., Lombroso, P.J., Francke, U.: Molecular cloning of the human homolog of a striatum-enriched phosphatase (STEP) gene and chromosomal mapping of the human and murine loci. *Genomics* **28:442-449 (1995).**

Ellison, J.W., Li, X., Francke, U., Shapiro, L.J.: Rapid evolution of human pseudoautosomal genes and their mouse homologs. *Mamm. Genome* **7:25-30 (1996).**

Shang, J., Li, X., Ring, H.Z., Clayton, D.A., Francke, U.: Backfoot, a novel homeobox gene, maps to human chromosome 5 (*BFT*) and mouse chromosome 13 (*Bft*). *Genomics* **40**:108-113 (1997).

Wan, M., Cravatt, B.F., Ring, H.Z., Zhang, X., Francke, U.: Conserved chromosomal location and genomic structure of human and mouse fatty-acid amide hydrolase genes and evaluation of *clasper* as a candidate neurological mutation. *Genomics* **54**:408-414 (1998).

Ring, H.Z., Vameghi-Meyers, V., Nikolic, J.M., Min, H., Black, D.L., Francke, U.: Mapping of the *KHSRP* gene to a region of conserved synteny on human chromosome 19p13.3 and mouse chromosome 17. *Genomics* **56**:350-352 (1999).

Graef, I.A., Gastier, J.M., Francke, U., Crabtree, G.R. Evolutionary relationships among Rel domains indicate functional diversification by recombination. *Proc. Natl. Acad. Sci. U S A.* **98**:5740-5745 (2001).

V. MOUSE GENE MAPPING

Francke, U., Lalley, P.A., Moss, W., Ivy, J., Minna, J.D.: Gene mapping in *Mus musculus* by interspecific cell hybridization: Assignment of the genes for tripeptidase-1 to chromosome 10, depeptidase-2 to chromosome 18, acid phosphatase-1 to chromosome 12 and adenylate kinase-1 to chromosome 2. *Cytogenet. Cell Genet.* **19:57-84 (1977).**

Minna, J.D., Bruns, G.A.P., Krinsky, A.H., Lalley, P.A., Francke, U., Gerald, P.S.: Assignment of a *Mus musculus* gene for triosephosphate isomerase to chromosome 6 and for glyoxalase-I to chromosome 17 using somatic cell hybrids. *Somat. Cell Genet.* **4**:241-252 (1978).

Lalley, P.A., Francke, U., Minna, J.D.: Homologous genes for enolase, phosphogluconate dehydrogenase, phosphoglucomutase and adenylate kinase are syntenic on mouse chromosome 4 and human chromosome 1p. *Proc. Natl. Acad. Sci. USA* **75**:2382-2386 (1978).

Oie, H.K., Gazdar, A.F., Lalley, P.A., Russell, E.K., Minna, J.D., DeLarco, J., Todaro, G.J., Francke, U.: Mouse chromosome 5 codes for ecotropic murine leukaemia virus cell-surface receptor. *Nature* **274**:60-62 (1978).

Lalley, P.A., Francke, U., Minna, J.D.: Assignments of the genes coding for pyrophosphatase and hexokinase-1 to mouse chromosome 10: Implications for comparative gene mapping in man and mouse. *Cytogenet. Cell Genet.* **22**:570-572 (1978).

Park, S.S., Gazdar, A.F., Lalley, P.A., Minna, J.D., Francke, U.: Spontaneous expression of murine endogenous viruses in hamster x mouse hybrid cells. *Int. J. Cancer* **23**:52-61 (1979).

Francke, U., Taggart, R.T.: Assignment of the gene for cytoplasmic superoxide dismutase (*Sod-1*) to a region of chromosome 16 and of *Hprt* to a region of the X chromosome in the mouse. *Proc. Natl. Acad. Sci. USA* **76:5230-5233 (1979).**

Taggart, R.T., Tetri, P., Francke, U.: Assignment of the gene for NADH diaphorase *Dia-1* to mouse chromosome 15. *Somat. Cell Genet.* **6**:769-776 (1980).

Francke, U., Gehring, U.: Chromosome assignment of a murine glucocorticoid receptor gene (*Grl-1*) using intraspecies somatic cell hybrids. *Cell* **22:657-664 (1980).**

Francke, U., de Martinville, B., d'Eustachio, P., Ruddle, F.H.: Comparative gene mapping: Murine lambda light chain genes are located in region cen→B5 of mouse chromosome 16 not homologous to human chromosome 21. *Cytogenet. Cell Genet.* **33**:267-271 (1982).

Bishop, J.O., Clark, A. J., Clissold, P.M., Hainey, S., Francke, U.: Two main groups of mouse major urinary protein genes, both largely located on chromosome 4. *EMBO J.* **1**:615-620 (1982).

Cory, S., Adams, J.M., Gerondakis, S.D., Miller, J.F.A.P., Gamble, J., Wiener, F., Spira, J., Francke, U.: Fusion of DNA region to murine immunoglobulin heavy chain locus corresponds to plasmacytoma-associated chromosome translocation. *EMBO J.* **2**:213-216 (1983).

George, D.L., Scott, A.F., de Martinville, B., Francke, U.: Amplified DNA in Y1 mouse adrenal tumor cells: Isolation of cDNAs complementary to an amplified c-Ki-ras gene and localization of homologous sequences to mouse chromosome 6. *Nucleic Acids Res.* **12:2731-2743 (1984).**

Yamamoto, K., Floyd-Smith, G., Francke, U., Koch, N., Lauer, W., Dobberstein, B., Schäfer, R., Hammerling, G.J.: The gene encoding the Ia-associated invariant chain is located on chromosome 18 in the mouse. *Immunogenetics* **21**:83-90 (1985).

Pear, W.S., Ingversson, S., Steffen, D., Münke, M., Francke, U., Bazin, H., Klein, G., Sümegi, J.: Multiple chromosomal rearrangements in a spontaneously arising t(6;7) rat immunocytoma juxtapose *c-myc* and immunoglobulin heavy chain sequences. *Proc. Natl. Acad. Sci. USA* **83**:7376-7380 (1986).

Breier G., Bucan, M., Francke, U., Colberg-Poley, A.M., Gruss, P.: Sequential expression of murine homeo box genes during F9 EC cell differentiation. *EMBO J.* **5**:2209-2215 (1986).

Mason, I.J., Murphy, D., Münke, M., Francke, U., Elliott R.W., Hogan, B.L.M.: Developmental and transformation-sensitive expression of the mouse *Sparc* gene on chromosome 11. *EMBO J.* **5**:1831-1837 (1986).

- Münke, M., Harbers, K., Jaenisch R., Francke, U.: Chromosomal mapping of four different integration sites of Moloney murine leukemia virus including the locus for $\alpha 1(I)$ collagen in mouse. *Cytogenet. Cell Genet.* **43:140-149** (1986)**
- Münke, M., Cox, D.R., Jackson, I.J., Hogan, B.L.M., Francke, U.: The murine *Hox-2* cluster of homeo box containing genes maps distal on chromosome 11 near the tail-short (*Ts*) locus. *Cytogenet. Cell Genet.* **42:236-240** (1986).
- Cahilly-Snyder, L., Yang-Feng, T., Francke, U., George, D.L.: Molecular analysis and chromosomal mapping of amplified genes isolated from a transformed mouse 3T3 cell line. *Somat. Cell Molec. Genet.* **13:235-244** (1987).
- Ingvarsson, S., Sundaresan, S., Jin, P., Francke, U., Asker, C., Sümegi, J., Klein, G., Sejersen, T.: Chromosome localization and expression pattern of *Lmyc* and *Bmyc* in murine embryonal carcinoma cells. *Oncogene* **3:679-685** (1988).
- Spiegelman, B.M., Lowell, B., Napolitano, A., Dubuc, P., Barton, D., Francke, U., Groves, D.L., Cook, K.S., Flier, J.S.: Adrenal glucocorticoids regulate adiponin gene expression in genetically obese mice. *J. Biol. Chem.* **264:1811-1815** (1989).
- Sundaresan, S., Francke, U.: Genes for $\beta 2$ -adrenergic receptor and the platelet-derived growth factor receptor map to mouse chromosome 18. *Somat. Cell Molec. Genet.* **15:367-371** (1989).
- Sundaresan, S., Francke, U.: Insulin-like growth factor I receptor gene is concordant with the *C-Fes* protooncogene and mouse chromosome 7 in somatic cell hybrids. *Somat. Cell Molec. Genet.* **15:373-377** (1989).
- Sundaresan, S., Yang-Feng, T.L., Francke, U.: Genes for HMG-CoA reductase and serotonin 1a receptor are on mouse chromosome 13. *Somat. Cell Molec. Genet.* **15:465-469** (1989).
- Sundaresan, S., Francke, U.: Gene for $\alpha 2(I)$ collagen is on mouse chromosome 6 not 16. *Somat. Cell Molec. Genet.* **15:471-473** (1989).
- Friedman, J.M., Schneider, B.S., Barton, D.E., Francke, U.: Level of expression and chromosome mapping of the mouse cholecystokinin gene: Implications for murine models of genetic obesity. *Genomics* **5:463-469** (1989).
- Kwon, B.S., Haq, A.K., Wakulchik, M., Kestler, D., Barton, D.E., Francke, U., Lamoreux, M.L., Whitney, III, J.B., Halaban, R.: Isolation, chromosomal mapping, and expression of the mouse tyrosinase gene. *J. Investigative Dermatology* **93:589-594** (1989).
- Gridley, T., Gray, D.A., Orr-Weaver, T., Soriano, P., Barton, D.E., Francke, U., Jaenisch, R.: Molecular analysis of the *Mov 34* mutation: Transcript disrupted by proviral integration in mice is conserved in *Drosophila*. *Development* **109:235-242** (1990).
- Harbers, K., Francke, U., Soriano, P., Jaenisch, R., Müller, U.: Structure and chromosomal mapping of a highly polymorphic repetitive DNA sequence from the pseudoautosomal region of the mouse sex chromosomes. *Cytogenet. Cell Genet.* **53:129-133** (1990).**

You, K.-H., Hsieh, C.-L., Hayes, C., Stahl, N., Francke, U., Popko, B.: DNA sequence, genomic organization, and chromosomal localization of the mouse peripheral myelin protein zero gene: Identification of polymorphic alleles. *Genomics* **9**:751-757 (1991).

Kwon, B.S., Chintamaneni, C., Kozak, C.A., Copeland, N.G., Gilbert, D.J., Jenkins, N., Barton, D., Francke, U., Kobayashi, Y., Kim, K.K.: A melanocyte-specific gene, Pmel 17, maps near the silver coat color locus on mouse chromosome 10 and is in a syntenic region on human chromosome 12. *Proc. Natl. Acad. Sci. USA*. **88**:9228-9232 (1991).

Suter, U., Welcher, A.A., Özçelik, T., Snipes, G.J., Kosaras, B., Francke, U., Billings Gagliardi, S., Sidman, R.L., Shooter, E.M.: Trembler mouse carries a point mutation in a myelin gene. *Nature* **356:241-244 (1992).**

Wu, H., Fässler, R., Schnieke, A., Barker, D., Lee, K.H., Chapman, V., Francke, U., Jaenisch, R.: An X-linked human collagen transgene escapes X inactivation in a subset of cells. *Development* **116**:687-695 (1992).

Li, X., Pereira, L., Zhang, H., Sanguineti, C., Ramirez, F., Bonadio, J., Francke, U.: Fibrillin genes map to regions of conserved mouse/human synteny on mouse chromosomes 2 and 18. *Genomics* **18:667-672 (1993).**

Chen, Y., Faraco, J., Yin, W., Germiller, J., Francke, U., Bonadio, J.: Structure, chromosomal localization, and expression pattern of the murine *Magp* gene. *J. Biol. Chem.* **268**:27381-27389 (1993).

Kelly, D., Chancellor, K., Milatovich, A., Francke, U., Suzuki, K., Popko, B.: Autosomal recessive neuromuscular disorder in a transgenic line of mice. *J. Neuroscience* **14**:198-207 (1994).

Ho, S.N., Thomas, D.J., Timmerman, L.A., Li, X., Francke, U., Crabtree, G.R.: NFATc3 a lymphoid-specific NFATc family member that is calcium-regulated and exhibits distinct DNA binding specificity. *J. Biol. Chem.* **270**:19898-19907 (1995).

Coetzee, T., Li, X., Fujita, N., Marcus, J., Suzuki, K., Francke, U., Popko, B.: Molecular cloning, chromosomal mapping, and characterization of the mouse UDP-galactose:ceramide galactosyltransferase gene. *Genomics* **35**:215-222 (1996).

Fang, J., Li, X., Smiley, E., Francke, U., Mecham, R.P., Bonadio, J.: Mouse latent TGF- β binding protein-2: molecular cloning and developmental expression. *Biochim. Biophys. Acta* **1354**:219-230 (1997).

Ring, H.Z., Vameghi-Meyers, V., Min, H., Nikolic, J.M., Black, D.L., Francke, U.: The mouse *Fubp* gene maps near the distal end of chromosome 3. *Genomics* **56**:357-358 (1999).

VI. HUMAN GENETIC DISORDERS

A. New Syndromes Delineated

Francke, U., Kernahan, C., Bradshaw, C.: Del(10)p autosomal deletion syndrome: Clinical, cytogenetic and gene marker studies. *Humangenetik* **26**:343-351 (1975).

Francke, U., Jones, K.L.: The 2p partial trisomy syndrome. *Am. J. Dis. Child* **130:1244-1249 (1976).**

Taylor, K.M., Wolfinger, H.L., Brown, M.G., Chadwick, D.L., Francke, U.: Trisomy 20p derived from a t(18;20) translocation. *Human. Genet.* **34**:155-162 (1976).

Keller, M.A., Jones, K.L., Nyhan, W.L., Francke, U., Dixon, B.: A new syndrome of mental deficiency with craniofacial, limb, and anal abnormalities. *J. Pediat.* **88**:589-591 (1976).

Francke, U., Weber, F., Sparkes, R.S., Mattson, P.D., Mann, J.: Duplication 11q (q21 to 23→qter) syndrome. In *Birth Defects: Original Article Series XIII*, No. 3B, pp. 167-186, The National Foundation, New York (1977).

Francke, U., Arias, D.E., Nyhan, W.L.: Proximal 4p- deletion: Phenotype differs from classical 4p-syndrome. *J. Pediat.* **90**:250-252 (1977).

Francke, U., Gonzales y Rivera, E.L., Delgado, C.G., Ramos, M.G.: Saethre-Chotzen Syndrome (SCS) with additional abnormalities in a Mexican family. In *Birth Defects: Original Article Series XIII* (No. 3B), p. 241, The National Foundation, New York (1977).

Pallister, P.D., Meisner, L.F., Elejalde, B.R., Francke, U., Herrmann, J., Spranger, J., Tiddy, W., Inhorn, S.L., Opitz, J.M.: The Pallister mosaic syndrome. In *Birth Defects: Original Article Series XIII* (No. 3B), pp. 103-110, The National Foundation, New York (1977).

McNeal, R.M., Skoglund, R.R., Francke, U.: Congenital anomalies including the VATER association in a patient with a del(6)q deletion. *J. Pediat.* **91**:957-960 (1977).

Francke, U., Opitz, J.M.: Chromosome 3q duplication and the Brachmann-de Lange Syndrome (BDLS). *J. Pediat.* **95**:161-162 (1979).

Curry, C.J.R., Loughman, W.D., Francke, U., Hall, B.D., Golbus, M.S., Derstine, J., Epstein, C.J.: Partial trisomy for the distal long arm of chromosome 5 (region q34→qter). A new clinically recognizable syndrome. *Clin. Genet.* **15**:454-461 (1979).

Harrod, M.J.E., Byrne, J.B., Dev, V.G., Francke, U.: Duplication 12q mosaicism in two unrelated patients with a similar syndrome. *Am. J. Med. Genet.* **7:123-128 (1980).**

Puck, S.M., Haseltine, F.P., Francke, U.: Absence of H-Y antigen in an XY female with campomelic dysplasia. *Hum. Genet.* **57:23-27 (1981).**

Horwich, A., Riccardi, V.M., Francke, U.: Aqueductal stenosis leading to hydrocephalus - an unusual manifestation of neurofibromatosis. *Am. J. Med. Genet.* **14**:577-581 (1983).

Burke, B.A., Wick, M.R., King, R., Thompson, T., Hansen, J., Darras, B.T., Francke, U., Seltzer, W.K., McCabe E.R.B., Scheithauer, B.W.: Congenital adrenal hypoplasia and selective

absence of pituitary luteinizing hormone: A new autosomal recessive syndrome. *Am. J. Med. Genet.* **31**:75-97 (1988).

B. Lesch-Nyhan Syndrome

Francke, U., Bakay, B., Nyhan, W.L.: Detection of heterozygotes for the Lesch-Nyhan syndrome by electrophoresis of hair root lysates. *J. Pediat.* **82**:472-478 (1973).

Gartler, S.M., Francke, U.: Half chromatid mutations: Transmission in humans? *Am. J. Hum. Genet.* **27**:218-223 (1975).

Francke, U., Felsenstein, J., Gartler, S.M., Migeon, B.R., Dancis, J., Seegmiller, J.E., Bakay, B., Nyhan, W.L.: The occurrence of new mutants in the X-linked recessive Lesch-Nyhan disease. *Am. J. Hum. Genet.* **28**:123-137 (1976).

Bakay, B., Francke, U., Nyhan, W.L., Seegmiller, J.E.: Experience with detection of heterozygous carriers and prenatal diagnosis of Lesch-Nyhan disease. In *Purine Metabolism in Man - II: Regulation of pathways and enzyme defects.* (M.M. Müller, E. Kaiser, J.E. Seegmiller, eds.), pp. 351-358, Plenum Publishing Corp., New York (1977).

Francke, U., Felsenstein, J., Gartler, S.M., Nyhan, W.L., Seegmiller, J.E.: Genetic epidemiology of Lesch-Nyhan disease. Answer to criticism of Morton and Lalouel. *Am. J. Hum. Genet.* **29**:307-310 (1977).

Bakay, B., Nissinen, E., Sweetman, L., Francke, U., Nyhan, W.L.: Utilization of purines by an HPRT variant in an intelligent, nonmutilative patient with features of the Lesch-Nyhan syndrome. *Pediat. Res.* **13**:1365-1370 (1979).

Francke, U., Winter, R.M., Lin, D., Bakay, B., Seegmiller, J.E., Nyhan, W.L.: Use of carrier detection tests to estimate male to female ratio of mutation rates in Lesch-Nyhan disease. In *Population and Biological Aspects of Human Mutation* (E.B. Hook, I.H. Porter, eds.), pp. 117-127, Academic Press, New York (1981).

Marcus, S., Steen, A.-M., Andersson, B., Lambert, B., Kristoffersson, U., Francke, U.: Mutation analysis and prenatal diagnosis in a Lesch-Nyhan family showing non random X-inactivation interfering with carrier detection tests. *Hum. Genet.* **89**:395-400 (1992).

C. Duchenne Muscular Dystrophy

(with microdeletions including Chronic Granulomatous Disease, Adrenal Hypoplasia and Glycerol Kinase Deficiency)

De Martinville, B., Kunkel, L.M., Bruns, G., Morlé, F., Koenig, M., Mandel, J.L., Horwich A., Latt, S.A., Gusella, J.F., Housman, D., Francke, U.: Localization of DNA sequences in region Xp21 of the human X chromosome: Search for molecular markers close to the Duchenne muscular dystrophy locus. *Am. J. Hum. Genet.* **37**:235-249 (1985).

Francke, U., Ochs, H.D., de Martinville, B., Giacalone, J., Lindgren, V., Distèche C., Pagon, R.A., Hofker, M.H., van Ommen, G.-J.B., Pearson, P.L., Wedgwood, R.J.: Minor Xp21 chromosome deletion in a male associated with expression of Duchenne muscular

dystrophy, chronic granulomatous disease, retinitis pigmentosa and McLeod syndrome. *Am. J. Hum. Genet.* 37:250-267 (1985).

Francke, U. : Developing genetic markers based on restriction fragment length polymorphisms near the locus for X-linked muscular dystrophy. In "*Molecular Biology of Muscle Development*" (C. Emerson et al., eds.), pp 875-886, Alan Liss Inc., New York (1986).

Hofker, M.H., Bergen, A.A.B., Skraastad, M.I., Bakker, E., Francke, U., Wieringa, B., Bartley, J., van Ommen, G.J.B., Pearson, P.L.: Isolation of a random cosmid cline, cX5, which defines a new polymorphic locus DXS148 near the locus for Duchenne muscular dystrophy. *Hum. Genet.* 74:275-279 (1986).

Kunkel, L.M., 71 co-authors, Francke, U.: Analysis of deletions in DNA from patients with Becker and Duchenne muscular dystrophy. *Nature* 322:73-77 (1986).

Francke, U., Harper, J.F., Darras, B.T., Cowan, J.M., McCabe, E.R.B., Kohlschütter, A., Seltzer, W.K., Saito, F., Goto, J., Harpey, J.-P., Wise J.E.: Congenital adrenal hypoplasia, myopathy, and glycerol kinase deficiency: Molecular genetic evidence for deletions. *Am. J. Hum. Genet.* 40:212-227 (1987).

Darras, B.T., Harper, J.F., Francke, U.: Prenatal diagnosis and detection of carriers with DNA probes in Duchenne's muscular dystrophy. *N. Engl. J. Med.* 316:985-992 (1987).

Darras, B.T., Francke, U.: A partial deletion of the muscular dystrophy gene transmitted twice by an unaffected male. *Nature* 329:556-558 (1987).

Darras, B.T., Koenig, M., Kunkel, L.M., Francke, U.: Direct method for prenatal diagnosis and carrier detection in Duchenne/Becker muscular dystrophy using the entire dystrophin cDNA. *Am. J. Med. Genet.* 29:713-726 (1988).

Darras, B.T., Francke, U.: Myopathy in complex glycerol kinase deficiency patients is due to 3' deletions of the dystrophin gene. *Am. J. Hum. Genet.* 43:126-130 (1988).

Darras, B.T., Francke, U.: Normal human genomic restriction-fragment patterns and polymorphisms revealed by hybridization with the entire dystrophin cDNA. *Am. J. Hum. Genet.* 43:612-619 (1988).

Darras, B.T., Blattner, P., Harper, J.F., Spiro, A.J., Alter, S., Francke, U.: Intragenic deletions in 21 Duchenne Muscular Dystrophy (DMD)/Becker Muscular Dystrophy (BMD) families studied with the dystrophin cDNA: Location of breakpoints on *HindIII* and *BglIII* exon-containing fragment maps, meiotic and mitotic origin of the mutations. *Am. J. Hum. Genet.* 43:620-629 (1988).

Miranda, A.F., Francke, U., Bonilla, E., Martucci, G., Schmidt, B., Salviati, G., Rubin, M.: Dystrophin immunocytochemistry in muscle culture: Detection of a carrier of Duchenne muscular dystrophy. *Am J. Med. Genet.* 32:268-273 (1989).

Francke, U., Darras, B.T., Hersh, J.H., Berg, B.O., Miller, R.G.: Brother/sister pairs affected with early-onset, progressive muscular dystrophy: Molecular studies reveal etiologic heterogeneity. *Am. J. Hum. Genet.* 45:63-72 (1989).

Battat, L., Francke, U.: Nsi I RFLP at the X-linked chronic granulomatous disease locus (CYBB). *Nucleic Acids Res.* 17:3619 (1989).

Koenig, M., Beggs, A.H., Moyer, M., Scherpf, S., Heindrich, K., Bettecken, T., Meng, G., Müller, C.R., Lindlöf, Kaarianinen, H., de la Chapelle, A., Kiuru, A., Savontaus, M.-L., Gilgenkrantz, H., Récan, D., Chelly, J., Kaplan, J.-C., Covone, A.E., Archidiacono, N., Romeo, G., Liechti-Gallati, S., Schneider, V., Braga, S., Moser, H., Darras, B.T., Murphy, P., Francke, U., Chen, J.D., Morgan, G., Denton, M., Greenberg, C.R., Wrogemann, K., Blondin, L.A.J., van Paassen, H.M.B., van Ommen, G.J.B., Kunkel, L.M.: The molecular basis for Duchenne versus Becker muscular dystrophy: Correlation of severity with type of deletion. *Am. J. Hum. Genet.* **45**:498-506 (1989).

Francke, U., Ochs, H.D., Darras, B.T., Swaroop, A.: Origin of mutations in two families with X-linked chronic granulomatous disease. *Blood* 76:602-606 (1990).

Blonden, L.A.J., Grootsholten, P.M., den Dunnen, J.T., Bakker, E., Abbs, S., Bobrow, M., Boehm, C., van Broeckhoven, C., Baumbach, L., Chamberlain, J., Caskey, C.T., Denton, M., Felicetti, L., Galluzi, G., Fischbeck, K.H., Francke, U., Darras, B., Gilgenkrantz, H., Kaplan, J.-C., Herrmann, F.H., Junien, C., Boileau, C., Liechti-Gallati, S., Lindlöf, M., Matsumoto, T., Niikawa, N., Müller, C.R., Poncin, J., Malcolm, S., Robertson, E., Romeo, G., Covone, A.E., Scheffer, H., Schröder, E., Schwartz, M., Verellen, C., Walker, A., Worton, R., Gillard, E., van Ommen, G.J.B.: 242 breakpoints in the 200-kb deletion-prone p20 region of the DMD gene are widely spread. *Genomics* **10**:631-639 (1991).

Matsuo, M., Nishio, H., Kitoh, Y., Francke, U., Nakamura, H.: Partial deletion of a dystrophin gene leads to exon skipping and to loss of an intra-exon hairpin structure from the predicted mRNA precursor. *Biochem. Biophys Res. Comm.* **182**:495-500 (1992).

Müller, B., Dechant, C., Meng, G., Liechti-Gallati, S., Doherty, R.A., Hejtmancik, J.F., Bakker, E., Read, A.P., Jeanpierre, M., Fischbeck, K.H., Romeo, G., Francke, U., Wilichoski, E., Greenberg, C.R., van Broeckhoven, C., Junien, C., Müller, C.R., Grimm, T.: Estimation of the male and female mutation rates in Duchenne muscular dystrophy (DMD). *Hum. Genet.* **89**:204-206 (1992).

D. Growth Hormone Insensitivity Syndrome (Laron Syndrome)

Berg, M.A., Guevara-Aguirre, J.G., Rosenbloom, A.L., Rosenfeld, R.G., Francke, U.: Mutation creating a new donor splice site in the growth hormone receptor genes of 37 Ecuadorean patients with Laron syndrome. *Hum. Mutat.* 1:24-34 (1992).

Berg, M.A., Guevara-Aguirre, J., Rosenbloom, A.L., Rosenfeld, R.G., Francke, U.: Laron syndrome in Ecuador: Point mutation in the growth hormone receptor gene causes deletion of 8 amino acids from the extracellular domain. *Pediatr. Adolesc. Endocrinol.* **24**:140-152 (1993).

Rosenbloom, A.L., Guevara-Aguirre, J., Fielder, P.J., Gargosky, S., Rosenfeld, R.G., Berg, M.A., Francke, U., Diamond, Jr., F.B., Vaccarello, M.A.: Growth hormone receptor deficiency/Laron syndrome in Ecuador: Clinical and biochemical characteristics. *Pediatr. Adolesc. Endocrinol.* **24**:34-52 (1993).

Berg, M.A., Argente, J., Chernauek, S., Gracia, R., Guevara-Aguirre, J., Hopp, M., Pérez-Jurado, L., Rosenbloom, A., Toledo, S.P.A., Francke, U.: Diverse growth hormone receptor gene mutations in Laron syndrome. *Am. J. Hum. Genet.* 52:998-1005 (1993).

Francke, U., Berg, M.A.: Genetic heterogeneity in Laron Syndrome. *Acta Paediatr. Suppl.* **391**:3-7 (1993).

Berg, M.A., Peoples, R., Pérez-Jurado, L., Guevara-Aguirre, J., Rosenbloom, A.L., Laron, Z., Milner, R.D.G., Francke, U.: Receptor mutations and haplotypes in growth hormone receptor deficiency: A global survey and identification of the Ecuadorean E180 splice mutation in an Oriental Jewish patient. *Acta Paediatr. Suppl.* **399**:112-114 (1994).

Rosenbloom, A.L., Berg, M.A., Kasatkina E.P., Volkova, T.N., Skorobogatova, V.F., Sokolovskya, V.N., Francke, U.: Severe growth hormone insensitivity (Laron Syndrome) due to nonsense mutation of the GH receptor in brothers from Russia. *J. Pediatr. Endocrinol. Metab.* **8**:159-165 (1995).

Rosenbloom, A.L., Guevara-Aguirre, J., Berg, M.A., Francke, U.: Stature in Ecuadorians heterozygous for the growth hormone receptor gene E180 splice mutation does not differ from that of homozygous normal relatives. *J. Clin. Endocrinol. Metab.* **83**:2373-2375 (1998).

Wojcik, J., Berg, M.A., Esposito, N., Geffner, M.E., Sakati, N., Reiter, E.O., Dower, S., Francke, U., Postel-Vinay, M.-C., Finidori, J.: Four contiguous amino-acid substitutions, identified in patients with Laron syndrome, differently affect the binding affinity and intracellular trafficking of the growth hormone receptor. *J. Clin. Endocrinol. Metab.* **83:4481-4489 (1998).**

Rosenbloom, A.L., Guevara-Aguirre, J., Rosenfeld, R.G., Francke, U.: Growth hormone receptor deficiency in Ecuador. *J. Clin. Endocrinol. Metab.* **84**:4436-4443 (1999).

Gastier, J.M., Berg, M.A., Verterhus, P., Reiter, E.O., Francke, U.: Diverse deletions in the growth hormone receptor gene cause growth hormone insensitivity syndrome. *Hum. Mutat.* **16: 323-333 (2000).**

E. Roberts Syndrome

Krassikoff, N.E., Cowan, J.M., Parry, D.M., Francke, U.: Chromatid repulsion associated with Roberts/SC phocomelia syndrome is reduced in malignant cells and not expressed in interspecies somatic-cell hybrids. *Am. J. Hum. Genet.* **39**:618-630 (1986).

Van Den Berg, D., Francke, U.: Roberts syndrome: A review of 100 cases and a new rating system for severity. *Am. J. Med. Genet.* **47**:1104-1123 (1993).

Van Den Berg, D., Francke, U.: Sensitivity of Roberts syndrome cells to gamma radiation, mitomycin C and protein synthesis inhibitors. *Somat. Cell Molec. Genet.* **19**:377-392 (1993).

Schüle B, Oviedo A, Johnston K, Pai S, Francke U. Inactivating mutations in *ESCO2* cause SC Phocomelia and Roberts Syndrome: No phenotype-genotype correlation. *Am. J. Hum. Genet* **77**:1117-1128 (2005).

F. Prader Willi Syndrome

Özçelik, T., Leff, S., Robinson, W., Donlon, T., Lalande, M., Sanjines, E., Schinzel, A., Francke, U.: Small nuclear ribonucleoprotein polypeptide N (*SNRPN*), an expressed gene in the Prader-Willi syndrome critical region. *Nature Genet.* **2:265-269 (1992).**

Leff, S.E., Brannan, C.I., Reed, M.L., Özçelik, T., Francke, U., Copeland, N.G., Jenkins, N.A.: Maternal imprinting of the mouse *Snrpn* gene and conserved linkage homology with Prader-Willi syndrome region of humans. *Nature Genet.* 2:259-264 (1992).

Giacalone, J., Francke, U.: Single nucleotide dimorphism in the transcribed region of the SNRPN gene at 15q12. *Hum. Mol. Genet.* 3:379 (1994).

Wevrick, R., Kerns, J.A., Francke, U.: Identification of a novel paternally expressed gene in the Prader-Willi syndrome region. *Hum. Molec. Genet.* 3:1877-1882 (1994).

Francke, U.: Prader Willi Syndrome: Chromosomal and Gene Aberrations. *Growth, Genetics and Hormones* 10:4-7 (1994).

Brilliant, M.H., King, R., Francke, U., Schuffenhauer, S., Meitinger, T., Gardner, J.M., Durham Pierre, D., Nakatsu, Y.: The mouse pink-eyed dilution gene: Association with hypopigmentation in Prader-Willi and Angelman syndromes and with human OCA2. *Pigment Cell Res.* 7:398-402 (1994).

Francke, U., Kerns, J.A., Giacalone, J.: The *SNRPN* gene and Prader Willi syndrome. In *Genomic Imprinting* (R. Ohlsson, K. Hall and M. Ritzen, Eds.), pp. 309-323, Cambridge University Press, Cambridge, UK (1995).

Wevrick, R., Kerns, J.A., Francke, U.: The IPW gene is imprinted and is not expressed in the Prader-Willi syndrome. *Acta Genet. Med. Gemellol.* 45:191-197 (1996).

Wevrick, R., Francke, U.: Diagnostic test for the Prader-Willi syndrome by SNRPN expression in blood. *The Lancet* 348:1068-1069 (1996).

Wevrick, R. and Francke, U.: An imprinted mouse transcript homologous to the human imprinted in Prader-Willi syndrome (IPW) gene. *Hum. Mol. Genet.* 6:325-332 (1997).

Francke, U.: Imprinted genes in the Prader Willi deletion. *Novartis Found. Symp.* 214:264-279 (1998).

Yang, T., Adamson, T.E., Resnick, J.L., Leff, S., Wevrick, R., Francke, U., Jenkins, N.A., Copeland, N.G., Brannan, C.I.: A mouse model for Prader-Willi syndrome imprinting-centre mutations. *Nature Genet.* 19:25-31 (1998).

Schweizer, J., Zynger, D., Francke, U.: *In vivo* nuclease hypersensitivity studies reveal multiple sites of parental origin-dependent differential chromatin conformation in the 150 kb SNRPN transcription unit. *Hum. Mol. Genet.* 8:555-566 (1999).

De Los Santos T, Schweizer J, Rees CA, Francke U.: Small evolutionarily conserved RNA, resembling C/D box small nucleolar RNA, is transcribed from PWCR1, a novel imprinted gene in the Prader-Willi deletion region, which is highly expressed in brain. *Am. J. Hum. Genet.* 67:1067-1082 (2000).

Fulmer-Smentek, S.B., Francke, U.: Association of acetylated histones with paternally expressed genes in the Prader-Willi deletion region. *Hum. Molec. Genet.* 10:645-652 (2001).

Gallagher, R.C., Pils, B., Albalwi, M., Francke, U. Evidence for the role of *PWCR1/HBII-85* C/D box small nucleolar RNAs in Prader-Willi syndrome. *Amer. J. Hum. Genet.* 71: 669-678 (2002).

Schüle B, Albalwi M, Northrop E, Francis DI, Rowell M, Slater HR, McKinlay Gardner RJ, Francke U. Molecular breakpoint cloning and gene expression studies of a novel translocation t(4;15)(q27;q11.2) associated with Prader-Willi syndrome. *BMC Medical Genetics* 2005, 6:18.

Ding F, Prints Y, Dhar MS, Johnson DK, Garnacho-Montero C, Nicholls RD, Francke U. Lack of *Pwcr1/MBII-85* snoRNA is critical for neonatal lethality in Prader-Willi syndrome mouse models. *Mamm. Genome* 16: 424-431 (2005).

Ding F, Li HH, Zhang S, Solomon NM, Camper SA, Francke U. SnoRNA *Snord116* (*Pwcr1/MBII-85*) deletion causes growth deficiency and hyperphagia in mice. *PloS ONE* 3(3): e1709. doi:10.1371/journal.pone.0001709 (2008)

G. Marfan Syndrome and Fibrillinopathies

Francke, U., Furthmayr, H.: Genes and gene products involved in Marfan syndrome. *Sem. Thoracic Cardiovasc. Surg.* 5:3-10 (1993).

Pyeritz, R.E., Francke, U.: Conference Report: The Second International Symposium on the Marfan Syndrome. *Am. J. Med. Genet.* 47:127-142 (1993).

Tynan, K.M., Comeau, K., Pearson, M., Wilgenbus, P., Levitt, D., Gasner, C., Berg, M.A., Miller, D.C., Francke, U.: Mutation screening of complete fibrillin-1 coding sequence: Report of five new mutations, including two in 8 cysteine domains. *Hum. Molec. Genet.* 2:1813-1821 (1993).

Aoyama, T., Tynan, K., Dietz, H.C., Francke, U., Furthmayr, H.: Missense mutations impair intracellular processing of fibrillin and microfibril assembly in Marfan syndrome. *Hum. Molec. Genet.* 2:2135-2140 (1993).

Aoyama, T., Francke, U., Dietz, H.C., Furthmayr, H.: Quantitative differences in biosynthesis and extracellular deposition of fibrillin in cultured fibroblasts distinguish five groups of Marfan syndrome patients and suggest distinct pathogenetic mechanisms. *J. Clin. Invest.* 94:130-137 (1994).

Francke, U., Furthmayr, H.: Marfan's syndrome and other disorders of fibrillin. Editorial, *N. Engl. J. Med.* 330:1384-1385 (1994).

Francke, U., Tynan, K., Aoyama, T., Gasner, C., Furthmayr, H.: Marfan syndrome and related heritable connective tissue disorders: fibrillin abnormalities, FBN1 mutations and pathogenetic mechanisms. In *Advances in Gene Technology: Molecular Biology and Human Disease*, Proceedings of the 1994 Miami Biotechnology Winter Symposium; Miami Short Reports 4:7 (1994).

Faraco, J., Bashir, M., Rosenbloom, J., Francke, U.: Characterization of the human gene for microfibril-associated glycoprotein (MAGP), assignment to chromosome 1 p36.1-p35 and linkage to D1S170. *Genomics* 25:630-637 (1995).

Francke, U., Berg, M.A., Tynan, K., Brenn, T., Liu, W., Aoyama, T., Gasner, C., Miller, D.C., Furthmayr, H.: A Gly1127Ser mutation in an EGF-like domain of the fibrillin-1 gene is a risk factor for ascending aortic aneurysm and dissection. *Am. J. Hum. Genet.* 56:1287-1296 (1995).

Aoyama, T., Francke, U., Gasner, C., Furthmayr, H.: Fibrillin abnormalities and prognosis in Marfan syndrome and related disorders. *Am. J. Med. Genet.* 58:169-176 (1995).

Dietz H, Francke U, Furthmayr H, Francomano C, De Paepe A, Devereux R, Ramirez F, Pyeritz R.: The question of heterogeneity in Marfan syndrome. *Nat Genet* 9:228-231 (1995)

Brenn, T., Aoyama, T., Francke, U., Furthmayr, H.: Dermal fibroblast culture as a model system for studies of fibrillin assembly and pathogenetic mechanisms: defects in distinct groups of individuals with Marfan's syndrome. *Lab. Invest.* 75:389-402 (1996)

Liu, W., Qian, C., Comeau, K., Brenn, T., Furthmayr, H., Francke, U.: Mutant fibrillin-1 monomers lacking EGF-like domains disrupt microfibril assembly and cause severe Marfan syndrome. *Hum. Mol. Genet.* 5:1581-1587 (1996).

Liu, W., Faraco, J., Qian, C., Francke, U.: The gene for microfibril-associated protein-1 (MFAP1) is located several megabases centromeric to FBN1 and is not mutated in Marfan syndrome. *Hum. Genet.* 99:578-584 (1997).

Schrijver, I., Liu, W., Francke, U.: The pathogenicity of the Pro1148Ala substitution in the FBN1 gene: causing or predisposing to Marfan syndrome and aortic aneurysm, or clinically innocent? *Hum. Genet.* 99:607-611 (1997).

Furthmayr, H., Francke, U.: Ascending aortic aneurysm with or without features of Marfan syndrome and other fibrillinopathies: New insights. *Sem. Thorac. Cardiovasc. Surg.* 9:191-205 (1997).

Liu, W., Qian, C., Francke, U.: Silent mutation induces exon skipping of fibrillin-1 gene in Marfan syndrome. *Nature Genet.* 16:328-329 (1997).

Collod-Bérout, G., Bérout, C., Ades, L., Black, C., Boxer, M., Brock, D.J.H., Holman, K.J., de Paepe, A., Francke, U., Grau, U., Hayward, C., Klein, H.G., Liu, W., Nuytinck, L., Peltonen, L., Alvarez Perez, A.B., Rantamäki, T., Junien, C., Boileau, C.: Marfan Database (third edition): new mutations and new routines for the software. *Nucleic Acids Res.* 26:229-233 (1998).

Babcock, D., Gasner, C., Francke, U., Maslen, C.: A single mutation that results in an Asp to His substitution and partial exon skipping in a family with congenital contractural arachnodactyly. *Hum. Genet.* 103:22-28 (1998).

Liu, W., Oefner, P., Qian, C., Odom, R., Francke, U.: Denaturing HPLC-identified novel FBN1 mutations, polymorphisms, and sequence variants in Marfan syndrome and related connective tissue disorders. *Genetic Testing* 1:237-242 (1998).

Schrijver, I., Liu, W., Brenn, T., Furthmayr, H., Francke, U.: Cysteine substitutions in epidermal growth factor-like domains of fibrillin-1: effects on biochemical and clinical phenotypes. *Am. J. Hum. Genet.* 65:1007-1020 (1999).

Francke, U.: Heritable Disorders of Connective Tissue. In *Kelley's Textbook of Internal Medicine*, 4th ed., H.D. Humes, et al., (eds). Philadelphia, PA: Lippincott Williams & Wilkins 2000:1439-1445.

Schrijver, I., Alcorn, D.M., Francke, U.: Marfan Syndrome. In *Management of Genetic Syndromes*, S.B. Cassidy and J.E. Allanson (eds). New York, John Wiley & Sons, pp. 207-228 (2001).

Liu, W., Schrijver, I., Brenn, T., Furthmayr, H., Francke, U.: Multi-exon deletions of the *FBNI* gene in Marfan syndrome. *BMC Medical Genetics* **2**:11 (2001)

Schrijver, I., Schievink, W.I., Godfrey, M., Meyer, F.B., Francke, U. Spontaneous spinal cerebrospinal fluid leaks and minor skeletal features of the Marfan syndrome. A microfibrilopathy. *J Neurosurg.* **96**:483-489 (2002).

Schrijver, I., Liu, W., Odom, R., Brenn, T., Oefner, P., Furthmayr, H., Francke, U.: Premature termination mutations in *FBNI*: Distinct effects on differential allelic expression and on protein and clinical phenotypes. *Amer. J. Hum. Genet.* **71: 223-237 (2002).**

Schrijver, I., Alcorn, D.M., Francke, U.: Marfan Syndrome. In *Management of Genetic Syndromes*, S.B. Cassidy and J.E. Allanson (eds). Second Edition. pp. 335-349. John Wiley & Sons, Hoboken, New Jersey (2005).

Faivre L, Collod-Beroud G, Loeys BL, Child A, Binquet C, Gautier E, Callewaert B, Arbustini E, Mayer K, Arslan-Kirchner M, Kiotsekoglou A, Comeglio P, Marziliano N, Dietz HC, Halliday D, Beroud C, Bonithon-Kopp C, Claustres M, Muti C, Plauchu H, Robinson PN, Ades LC, Biggin A, Benetts B, Brett M, Holman KJ, De Backer J, Coucke P, Francke U, De Paepe A, Jondeau G, Boileau C. Effect of mutation type and location on clinical outcome in 1,013 probands with Marfan syndrome or related phenotypes and *FBNI* mutations: an international study. *Am J Hum Genet.* **81**:454-66 (2007).

Yao Z, Jaeger JC, Ruzzo WL, Morales CZ, Emond M, Francke U, Milewicz DM, Schwartz SM, Mulvihill ER. A Marfan syndrome gene expression phenotype in cultured skin fibroblasts. *BMC Genomics* **8**:319 (2007).

Faivre L, Masurel-Paulet A, Collod-Beroud G, Loeys BL, Child A, Binquet C, Gautier E, Stheuner C, Chevallier P, Callewaert B, Arbustini E, Mayer K, Arslan-Kirchner M, Kiotsekoglou A, Comeglio P, Marziliano N, Halliday D, Beroud C, Bonithon-Kopp C, Claustres M, Robinson PN, Adès L, De Backer J, Coucke P, Francke U, De Paepe A, Boileau C, Jondeau G. Contribution of molecular screening in diagnosing Marfan syndrome and type I fibrillinopathies: an international study of 1009 probands. *J. Med. Genet.* **45**:384-390 (2008)

Faivre L, Masurel-Paulet A, Collod-Beroud G, Loeys BL, Child A, Binquet C, Gautier E, Stheuner C, Chevallier P, Callewaert B, Arbustini E, Mayer K, Arslan-Kirchner M, Kiotsekoglou A, Comeglio P, Marziliano N, Halliday D, Beroud C, Bonithon-Kopp C, Claustres M, Robinson PN, Adès L, De Backer J, Coucke P, Francke U, De Paepe A, Boileau C, Jondeau G. Clinical and molecular study of 320 children with Marfan syndrome and related

type I fibrillinopathies out of a series of 1009 probands with a pathogenic FBN1 Mutation. *Pediatrics* (in press 2008)

H. Rett Syndrome

Archidiacono, N., Lerone, M., Rocchi, M., Anvret, M., Özçelik, T., Francke, U., Romeo, G.: Rett syndrome: exclusion mapping following the hypothesis of germinal mosaicism for new X-linked mutations. *Hum. Genet.* **86**:604-606 (1991).

Francke, U., Schanen, C.: Book review: Rett Syndrome--Clinical and Biological Aspects. *Am. J. Hum. Genet.* **55**:596-597 (1994).

Hofferbert, S., Schanen, N.C., Budden, S.S., Francke, U.: Is Rett syndrome caused by a triplet repeat expansion? *Neuropediatrics* 28:179-183 (1997).

Schanen, N.C., Roth Dahle, E.J., Capozzoli, F., Holm, V.A., Zoghbi, H.Y., Francke, U.: A new Rett syndrome family consistent with X-linked inheritance expands the X chromosome exclusion map. *Am. J. Hum. Genet.* 61:634-641 (1997).

Wan, M., Francke, U.: Evaluation of two X chromosomal candidate genes for Rett syndrome: glutamate dehydrogenase-2 (GLUD2) and rab GDP-dissociation inhibitor (GDI1). *Am. J. Med. Genet.* **78**:169-172 (1998).

Heidary, G., Hampton, L.L., Schanen, N.C., Rivkin, M.J., Darras, B.T., Battey, J., Francke, U.: Exclusion of the gastrin-releasing peptide receptor (GRPR) locus as a candidate gene for Rett syndrome. *Am. J. Med. Genet.* **78**:173-175 (1998).

Schanen, C., Francke, U.: A severely affected male born into a Rett syndrome kindred supports X-linked inheritance and allows extension of the exclusion map. *Am. J. Hum. Genet.* **63**:267-269 (1998).

Amir, R.E., Van den Veyver, I.B., Wan, M., Tran, C.Q., Francke, U., Zoghbi, H.Y.: Rett syndrome is caused by mutations in X-linked MECP2, encoding methyl-CpG-binding protein 2. *Nature Genet.* 23:185-188, 1999.

Wan, M., Lee, S.S.J., Zhang, X., Houwink-Manville, I., Song, H.-R., Amir, R.E., Budden, S., Naidu, S., Pereira, J.L.P., Lo, I.F.M., Zoghbi, H.Y., Schanen, N.C., Francke, U.: Rett syndrome and beyond: recurrent spontaneous and familial MECP2 mutations at CpG hotspots. *Am. J. Hum. Genet.* 65:1520-1529 (1999).

Kerr, A.M., Nomura, Y., Armstrong, D., Anvret, M., Belichenko, P.V., Budden, S., Cass, H., Christodoulou, J., Clarke, A., Ellaway, C., d'Esposito, M., Francke, U., Hulten M., Julu, P., Leonard, H., Naidu, S., Schanen, C., Webb, T., Engerstrom, I.W., Yamashita, Y., Segawa, M.: Guidelines for reporting clinical features in cases with MECP2 mutations. *Brain & Dev.* **23**:208-211 (2001).

Lee, S.S.J., Wan, M., Francke, U.: Spectrum of MECP2 mutations in Rett syndrome. *Brain & Dev.* **23**:S138-S143 (2001).

Wan, M., Zhao, K., Lee, S.S.J., Francke, U.: MECP2 truncating mutations cause histone H4 hyperacetylation in Rett syndrome. *Hum. Molec. Genet.* 10:1085-1092 (2001).

Traynor, J., Agarwal, P., Lazzeroni, L., Francke, U. Gene expression patterns vary in clonal cell cultures from Rett syndrome females with eight different MECP2 mutations. *BMC Medical Genetics* 3:12 (2002).

Gartler, S.M., Varadarajan, K.R., Luo, P., Canfield, T.K., Traynor, J., Francke, U., Hansen, R.S. Normal histone modifications on the inactive X chromosome in ICF and Rett syndrome cells: implications for methyl-CpG binding proteins. *BMC Biology* 2004, 2:21 (2004)

Francke U. Mechanisms of Disease: neurogenetics of MeCP2 deficiency. *Nature Clinical Practice Neurology* 2, 212-221 (2006).

Liu J, Francke U. Identification of cis-regulatory elements for MECP2 expression. *Hum Molec Genet.* 15:1769-1782 (2006)

Jordan C, Francke U. Ube3a expression is not altered in Mecp2 mutant mice. *Hum Mol Genet.* 15:2210-2215 (2006)

Jordan C, Li HH, Kwan HC, Francke U. Cerebellar gene expression profiles of mouse models for Rett syndrome reveal novel MeCP2 targets. *BMC Medical Genetics* 8:36 (2007).

Schüle B, Li HH, Fisch-Kohl C, Purmann C, Francke U. DLX5 and DLX6 expression is bi-allelic and not modulated by MeCP2 deficiency. *Amer J Hum Genet* 81:492-506 (2007).

Belichenko NP, Belichenko PV, Li HH, Mobley WC, Francke U. Comparative study of brain morphology in Mecp2 mutant mouse models of Rett syndrome. *J Comp. Neurology* 508:148-195 (2008)

Schüle B, Armstrong D, Vogel H, Oviedo A, Francke U. Severe congenital encephalopathy caused by MECP2 null mutations in males: central hypoxia and reduced neuronal dendritic structure. *Clin Genet.* 74: 116-126 (2008)

I. Williams Beuren Syndrome

Pérez-Jurado, L.A., Li, X., Francke, U.: The human calcitonin receptor gene (CALCR) at 7q21.3 is outside the deletion associated with the Williams syndrome. *Cytogenet. Cell Genet.* 70:246-249 (1995).

Peoples, R., Pérez-Jurado, L., Wang, Y.-K., Kaplan, P., Francke, U.: The gene for replication factor C subunit 2 (RFC2) is within the 7q11.23 Williams syndrome deletion. *Am. J. Hum. Genet.* 58:1370-1373 (1996).

Pérez-Jurado, L.A., Peoples, R., Kaplan, P., Hamel, B.C.J., Francke, U.: Molecular definition of the chromosome 7 deletion in Williams syndrome and parent-of-origin effects on growth. *Am. J. Hum. Genet.* 59:781-792 (1996).

Wang, Y.-K., Harryman Samos, C., Peoples, R., Pérez-Jurado, L.A., Nusse, R., Francke, U.: A novel human homologue of the *Drosophila frizzled* wnt receptor gene binds wingless protein and is in the Williams syndrome deletion at 7q11.23. *Hum. Mol. Genet.* 6:465-472 (1997).

Wedemeyer, N., Peoples, R., Himmelbauer, H., Lehrach, H., Francke, U., Wanker, E.E.: Localization of the human *HIP1* gene close to the elastin (ELN) locus on 7q11.23. *Genomics* 46:313-315 (1997).

Pérez-Jurado, L.A., Wang, Y.-K., Peoples, R., Coloma, A., Cruces, J., Francke, U.: A duplicated gene in the breakpoint regions of the 7q11.23 Williams-Beuren syndrome deletion encodes the initiator binding protein TFII-I and BAP-135, a phosphorylation target of BTK. *Hum. Mol. Genet.* 7:325-334 (1998).

Wang, Y.-K., Pérez-Jurado, L.A., Francke, U.: A mouse single-copy gene *Gtf2i*, the homolog of human *GTF2I*, that is duplicated in the Williams-Beuren syndrome deletion region. *Genomics* 48:163-170 (1998).

Peoples, R., Cisco, M.J., Kaplan, P., Francke, U.: Identification of the WBSCR9 gene, encoding a novel transcriptional regulator, in the Williams-Beuren syndrome deletion at 7q11.23. *Cytogenet. Cell Genet.* 82: 238-246 (1998).

Paperna, T., Peoples, R., Wang, Y.-K., Kaplan, P., Francke, U.: Genes for the CPE receptor (*CPETRI*) and the human homolog of RVP1 (*CPETR2*) are localized within the Williams Beuren syndrome deletion. *Genomics* 54:453-459 (1998).

Wang, Y.-K., Spörle, R., Paperna, T., Schughart, K., Francke, U.: Characterization and expression pattern of the *frizzled* gene *Fzd9*, the mouse homolog of *FZD9* which is deleted in Williams-Beuren syndrome. *Genomics* 57:235-248 (1999).

Francke, U.: Williams-Beuren Syndrome: Genes and Mechanisms. *Hum. Molec. Genet.* 8:1947-1954 (1999).

Franke, Y., Peoples, R.J., Francke, U.: Identification of GTF2IRD1 a putative transcription factor within the Williams-Beuren syndrome deletion at 7q11.23. *Cytogenet. Cell Genet.* 86:296-304 (1999).

Pérez-Jurado, L.A., Wang, Y.-K., Francke, U., Cruces, J.: TBL2, a novel transducin family member in the WBS deletion: characterization of the complete sequence, genomic structure, transcriptional variants and the mouse ortholog. *Cytogenet. Cell Genet.* 86:277-284 (1999).

Peoples, R., Franke, Y., Wang, Y.-K., Pérez-Jurado, L.A., Paperna, T., Cisco, M., Francke, U.: A physical map, including a BAC/PAC clone contig, of the Williams-Beuren syndrome deletion region at 7q11.23. *Am. J. Hum. Genet.* 66:47-68 (2000).

Kaplan, P., Wang, P., Francke, U.: Williams (Williams Beuren) Syndrome: a distinct neurobehavioral disorder. *J. Child Neurology* 16:177-190 (2001).

Van Raay, T.J., Wang, Y.-K., Stark, M.R., Rasmussen, J.T., Francke, U., Vetter, M.L., Rao, M.S.: *Frizzled 9* is expressed in neural precursor cells in the developing neural tube. *Dev. Genes & Evol.* 211:453-457 (2001).

Ranheim EA, Kwan HC, Reya T, Wang YK, Weissman IL, Francke U. Frizzled 9 knockout mice have abnormal B cell development. *Blood* 105:2487-2494 (2005)

Edelmann L, Prosnitz A, Pardo S, Bhatt J, Cohen N, Lauriat T, Ouchanov L, Jimenez Gonzalez P, Manghi ER, Bondy P, Esquivel M, Monge S, Fallas M, Splendore A, Francke U, Burton BK, McInnes LA. An atypical deletion of the Williams-Beuren Syndrome interval implicates genes associated with defective visuospatial processing and autism. *J Med Genet.* 44:136-143 (2007).

J. Wiskott Aldrich Syndrome

Derry, J.M.J., Ochs, H.D., Francke, U.: Isolation of a novel gene mutated in Wiskott Aldrich syndrome. *Cell* 78:635-644 (1994).

Derry, J.M.J., Kerns, J.A., Weinberg, K.I., Ochs, H.D., Volpini, V., Estivil, X., Walker, A.P., Francke, U.: WASP gene mutations in Wiskott-Aldrich syndrome and X-linked thrombocytopenia. *Hum. Molec. Genet.* 4:1127-1135 (1995).

Zhu, Q., Zhang, M., Blaese, R.M., Derry, J.M.J., Junker, A., Francke, U., Chen, S.-H., Ochs, H.D.: The Wiskott-Aldrich syndrome and X-linked congenital thrombocytopenia are caused by mutations of the same gene. *Blood* 86:3797-3804 (1995).

Derry, J.M.J., Wiedemann, P., Blair, P., Wang, Y.-K., Kerns, J.A., Lemahieu, V., Godfrey, V.L., Wilkinson, J.E., Francke, U.: The mouse homolog of the Wiskott Aldrich Syndrome Protein (WASP) gene is highly conserved and maps near the *scurfy* (*sf*) mutation on the X chromosome. *Genomics* 29:471-477 (1995).

Symons, M., Derry, J.M.J., Karlak, B., Jiang, S., Lemahieu, V., McCormick, F., Francke, U., Abo, A.: Wiskott-Aldrich syndrome protein, a novel effector for the GTPase CDC42Hs, is implicated in actin polymerization. *Cell* 84:723-734 (1996).

Lemahieu, V., Gastier, J.M., Francke, U.: Novel mutations in the Wiskott-Aldrich syndrome protein gene and their effects on transcriptional, translational, and clinical phenotypes. *Hum. Mutat.* 14:54-66 (1999).

Ferguson, P.J., Blanton, S.H., Saulsbury F.T., McDuffie, M.J., Lemahieu, V., Gastier, J.M., Francke, U., Borowitz, S. M., Sutphen, J.L., Kelly, T.E.: Manifestations and linkage analysis in X-linked autoimmunity-immunodeficiency syndrome. *Am. J. Med. Genet.* 90:390-397 (2000).

Huang, M.-M., Tsuboi, S., Wong, A., Yu, X.J., Oh-Eda, M., Derry, J.M., Francke, U., Fukuda, M., Weinberg, K.I., Kohn, D.B.: Expression of human Wiskott-Aldrich syndrome protein in patients' cells leads to partial correction of a phenotypic abnormality of cell surface glycoproteins. *J. Gene Therapy* 7:314-320 (2000).

Francke, U. WASP (Wiskott-Aldrich Syndrome Protein). In *Wiley Encyclopedia of Molecular Medicine*, John Wiley & Sons, Inc., New York, pp 3396-3398. (2002).

VII. HUMAN MOLECULAR GENETICS

De Martinville, B., Blakemore, K.J., Mahoney, M.J., Francke, U.: DNA analysis of first-trimester chorionic villous biopsies: Test for maternal contamination. *Am. J. Hum. Genet.* **36**:1357-1368 (1984).

Barton, D.E., Francke, U.: Activation of human α 1-antitrypsin genes in rat hepatoma x human fibroblast hybrid cell lines is correlated with demethylation. *Somat. Cell Molec. Genet.* **13:635-644 (1987).**

Swaroop, A., Hogan, B.L.M., Francke, U.: Molecular analysis of the cDNA for human SPARC/Osteonectin/BM-40: Sequence, expression, and localization of the gene to chromosome 5q31-q33. *Genomics* **2:37-47 (1988).**

Lux, S.E., Tse, W.T., Menninger, J.C., John, K.M., Harris, P., Shalev, O., Chilcote, R.R., Marchesi, S.L., Watkins, P.C., Bennett, V., McIntosh, S., Collins, F.S., Francke, U., Ward, D.C., Forget, B.G.: Hereditary spherocytosis associated with deletion of the human erythrocyte ankyrin gene on chromosome 8. *Nature* **345**:736-739 (1990).

Spritz, R.A., Strunk, K.M., Hsieh, C.-L., Sekhon, G., Francke, U.: Homozygous tyrosinase gene mutation in an American Black with tyrosinase-negative (type IA) oculocutaneous albinism. *Am. J. Hum. Genet.* **48**:318-324 (1991).

Cali, J.J., Hsieh, C.-L., Francke, U., Russell, D.W.: Mutations in the bile acid biosynthetic enzyme sterol 27-hydroxylase underlie cerebrotendinous xanthomatosis. *J. Biol. Chem.* **266**:7779-7783 (1991).

Giacalone, J., Francke, U.: Common sequence motifs at the rearrangement sites of a constitutional X/autosome translocation and associated deletion. *Am. J. Hum. Genet.* **50:725-741 (1992).**

Giacalone, G., Friedes, J., Francke, U.: A novel GC-rich human macrosatellite VNTR in Xq24 is differentially methylated on active and inactive X chromosomes. *Nature Genet.* **1:137-143 (1992).**

Patel, P.I., Roa, B., Welcher, A.A., Schoener-Scott, R., Trask, B.J., Pentao, L., Snipes, G.J., Garcia, C.A., Francke, U., Shooter, E.M., Lupski, J.R., Suter, U.: The gene for the peripheral myelin protein PMP-22 is a candidate for Charcot-Marie-Tooth disease type 1A. *Nature Genet.* **1:159-165 (1992).**

Pritchard, C., Zhu, N., Zuo, J., Bull, L., Pericak-Vance, M.A., Vance, J.M., Roses, A.D., Milatovich, A., Francke, U., Cox, D.R., Myers, R.M.: Recombination of 4p16 DNA markers in an unusual family with Huntington disease. *Am. J. Hum. Genet.* **50**:1218-1230 (1992).

Ahuja, S.K., Özçelik, T., Milatovich, A., Francke, U., Murphy, P.M.: Molecular evolution of the human interleukin-8 receptor gene cluster. *Nature Genetics* **2:31-36 (1992).**

Aruffo, A., Farrington, M., Hollenbaugh, D., Li, X., Milatovich, A., Nonoyama, S., Bajorath, J., Grosmaire, L.S., Stenkamp, R., Neubauer, M., Roberts, R.L., Noelle, R.J., Ledbetter, J.A., Francke, U., Ochs, H.D.: The CD40 ligand, gp39, is defective in activated T cells from patients with X-linked hyper-IgM syndrome. *Cell* **72:291-300 (1993).**

Pérez-Jurado, L.A., Francke, U.: Dinucleotide repeat polymorphism at the human pituitary adenylate cyclase activating polypeptide (PACAP) gene. *Hum. Molec. Genet.* **2**:827 (1993).

Chang, E., Welch, S., Luna, J., Giacalone, J., Francke, U.: Generation of a human chromosome 18-specific YAC clone collection and mapping of 55 unique YACs by FISH and fingerprinting. *Genomics* **17:393-402 (1993).**

Chang, E., Luna, J., Giacalone, J., Uyar, D., Silverman, G.A., Francke, U.: Regional localization of 56 new human chromosome 18-specific yeast artificial chromosomes. *Cytogenet. Cell Genet.* **65**:136-139 (1993).

Pérez-Jurado, L.A., Phillips, III, J.A., Francke, U.: Exclusion of growth hormone-releasing hormone gene mutations in familial isolated growth hormone deficiency by linkage and single strand conformation analysis. *J. Clin. Endocrinol. Metab.* **78**:622-628 (1994).

Francke, U., Chang, E., Comeau, K., Geigl, E.-M., Giacalone, J., Li, X., Luna, J., Moon, A., Welch, S., Wilgenbus, P.: A radiation hybrid map of human chromosome 18. *Cytogenet. Cell Genet.* **66:196-213 (1994).**

Durham-Pierre, D., Gardner, J.M., Nakatsu, Y., King, R.A., Francke, U., Ching, A., Aquaron, R., del Marmol, V., Brilliant, M.H.: African origin of an intragenic deletion of the human P gene in tyrosinase positive oculocutaneous albinism (OCA2). *Nature Genet.* **7**:176-179 (1994).

Pérez-Jurado, L.A., Phillips, III, J.A., Francke, U.: Exclusion of growth hormone (GH)-releasing hormone gene mutations in familial isolated GH deficiency by linkage and single strand conformation analysis. *J. Clin. Endocrinol. Metab.* **78**:622-628 (1994).

Wiemer E.A.C., Nuttley, W.M., Bertolaet, B.L., Li, X., Francke, U., Wheelock, M.J., Anné, U.K., Johnson, K.R., Subramani, S.: Human peroxisomal targeting signal-1 receptor restores peroxisomal protein import in cells from patients with fatal peroxisomal disorders. *J. Cell Biol.* **130**:51-65 (1995).

Sampaio, S.O., Li, X., Takeuchi, M., Mei, C., Francke, U., Butcher, E.C., Briskin, M.J.: Organization, regulatory sequences, and alternatively spliced transcripts of the mucosal addressin cell adhesion molecule-1 (MAdCAM-1) gene. *J. Immunol.* **155**:2477-2486 (1995).

Derry, J.M.J., Jess, U., Francke, U.: Cloning and characterization of a novel zinc finger gene in Xp11.2. *Genomics* **30:361-365 (1995).**

Derry, J.M.J., Kerns, J.A., Francke, U.: RBM3, a novel human gene in Xp11.23 with a putative RNA-binding domain. *Hum. Molec. Genet.* **4:2307-2311 (1995).**

Hofferbert, S., Schanen, N.C., Chehab, F., Francke, U.: Trinucleotide repeats in the human genome: size distributions for all possible triplets and detection of expanded disease alleles in a group of Huntington disease individuals by the Repeat Expansion Detection method. *Hum. Mol. Genet.* **6:77-83 (1997).**

Zhou, Y.-D., Barnard, M., Tian, H., Li, X., Ring, H.Z., Francke, U., Shelton, J., Richardson, J., Russell, D.W., McKnight, S.: Molecular characterization of two mammalian bHLH-PAS domain proteins selectively expressed in the central nervous system. *Proc. Natl. Acad. Sci. USA* **94**:713-718 (1997).

Pérez-Jurado, L.A., Argente, J., Barrios, V., Pozo, J., Muñoz, M.T., Hernández, M., Francke, U.: Molecular diagnosis and endocrine evaluation of a patient with a homozygous 7.0 kilobase deletion of the growth hormone gene cluster: Response to biosynthetic GH therapy. *J. Pediatr. Adol. Endocrinol.* **10**:185-190 (1997).

Hwang, B.J., Toering, S., Francke, U., Chu, G.: p48 activates a UV-damaged-DNA binding factor and is defective in Xeroderma pigmentosum group E cells that lack binding activity. *Molec. Cell. Biol.* **18**:4391-4399 (1998).

Ring, H.Z., Chang, H., Guilbot, A., LeGuern, E., Brice, A., Francke, U.: The human neuregulin-2 (*NRG2*) gene: cloning, mapping and evaluation as a candidate for the autosomal recessive form of Charcot-Marie-Tooth disease linked to 5q. *Hum. Genet.* **104**:326-332 (1999).

Francke, U. The human genome project: Implications for the endocrinologist. *J. Pediatr. Endocrinol. Metab.* **14**:1395-1408 (2001).

Arron JR, Winslow MM, Polleri A, Chang CP, Wu H, Gao X, Neilson JR, Chen L, Heit JJ, Kim SK, Yamasaki N, Miyakawa T, Francke U, Graef IA, Crabtree GR. NFAT dysregulation by increased dosage of DSCR1 and DYRK1A on chromosome 21. *Nature* 441:595-600 .(2006)