Stanford

Uta Francke

Professor of Genetics and of Pediatrics, Emerita

Curriculum Vitae available Online

CLINICAL OFFICE (PRIMARY)

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Bio

BIO

Uta Francke is professor of Genetics Emerita and Professor of Pediatrics - Medical Genetics at Stanford University. Her research has ranged from human and mouse chromosome identification and gene mapping to the discovery of genes involved in heritable disorders, studies of their functions and of disease-causing mechanisms. Applying genomic technologies to mammalian genetics research, her laboratory developed mouse models for human microdeletions, such as Prader-Willi and Williams-Beuren syndrome.

Dr. Francke holds an M.D. from the University of Munich, Germany. She is board-certified in Pediatrics, and in Clinical and Molecular Genetics and Cytogenetics. She has been an Investigator of the Howard Hughes Medical Institute and the recipient of the Antoine Marfan Award from the National Marfan Foundation, the Colonel Harland Sanders Lifetime Achievement Award in Genetics from the March of Dimes Birth Defects Foundation and the William Allan Award from the American Society of Human Genetics. In 2014,, she received the Association for Molecular Pathology Award for Excellence in Molecular Diagnostics. She has been elected to the Institute of Medicine of the National Academies, the American Association for Advancement of Science and the American Academy of Arts and Sciences. She is a past-president of the American Society for Human Genetics and of the International Federation of Human Genetics Societies, and a founding member of the American College of Medical Genetics.
Dr. Francke teaches medical and molecular genetics and sees patients in the Multidisciplinary Cardiovascular Connective Tissue Disorders Clinic at Stanford.

Dr. Francke has served as a consultant to 23andMe.Inc since 2007, and as a part-time employee from 2010-2013. In her role a Senior Medical Director she was involved in the preparation of health report and in various research projects, foremost the study of consumer responses to receiving BRCA mutation results.

CLINICAL FOCUS

- Neurogenetics
- Clinical Genetics

- Marfan syndrome
- Williams syndrome
- Heritable connective tissue disorders

ACADEMIC APPOINTMENTS

- Professor Emeritus-Hourly, Genetics
- Professor, Pediatrics - Medical Genetics

HONORS AND AWARDS

- Original Member, Highly Cited Researchers database, ISI (2002)
- Antoine Marfan Award, National Marfan Foundation (1996)
- President, International Federation of Human Genetics Societies (2000-2002)
- Elected President, American Society of Human Genetics (1999)
- Elected Member, American Academy of Arts and Sciences (1997)
- Elected Fellow, American Association for the Advancement of Science (1995)
- Elected Member, Institute of Medicine (National Academies) (1990)
- Elected Associate Member, European Molecular Biology Organization (2009)
- Colonel Harland Sanders Lifetime Achievement Award in Genetics, March of Dimes Birth Defects Foundation (2001)
- William Allan Award, American Society for Human Genetics (2012)
- Award for Excellence in Molecular Diagnostics, Association for Molecular Pathology (2014)

BOARDS, ADVISORY COMMITTEES, PROFESSIONAL ORGANIZATIONS

- Chair, Awards Committee, American Society of Human Genetics (2020 - present)
- Founding Member, American College of Medical Genetics (1993 - present)

PROFESSIONAL EDUCATION

- Board Certification: Clinical Cytogenetics and Genomics, American Board of Medical Genetics and Genomics (1982)
- Board Certification: Clinical Genetics, American Board of Medical Genetics and Genomics (1982)
- Board Certification: Pediatrics, American Board of Pediatrics (1981)
- Fellowship: UCSD Medical Genetics Fellowship (1973) CA
- Fellowship: UCLA Medical Genetics and Genomics Fellowship (1971) CA
- Residency: Childrens Hospital Los Angeles Pediatric Residency (1970) CA
- Internship: Klinikum Rechts Der Isar (1969) Germany
- Medical Education: Ludwig Maximilians Universitat Munchen (1966) Germany
- Board certification, Clinical Molecular Genetics , American Board of Medical Genetics (2009)
- M.D. (Dr. med.), Universitaet Munchen, Germany , Medicine (1967)

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

My laboratory at Stanford is closed. My ongoing research activities are collaborative.

Teaching

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Genetics (Phd Program)

Publications

PUBLICATIONS

- **Optical genome mapping with genome sequencing identifies subtelomeric Xq28 deletion and inserted 7p22.3 duplication in a male with multisystem developmental disorder.** *American journal of medical genetics. Part A*
  Rodriguez-Gil, J. L., Nagy, P. L., Francke, U.
  2024: e63814

- **Direct-to-consumer genetic testing for factor V Leiden and prothrombin 20210G>A: the consumer experience.** *Molecular genetics & genomic medicine*
  Elson, S. L., Furlotte, N. A., Hromatka, B. S., Wilson, C. H., Mountain, J. L., Rowbotham, H. M., Varga, E. A., Francke, U.
  2020: e1468

- **Report of the Phenotype of a Patient with Roberts Syndrome and a Rare ESCO2 Variant.** *Journal of pediatric genetics*
  da Costa Almeida, C. B., Welter, A. T., Abech, G. D., Brandão, G. R., Flores, J. A., Schüle, B. n., Francke, U. n., Fiegenbaum, M. n., Zen, P. R., Rosa, R. F.
  2020; 9 (1): 58–62

- **Isolation of a Novel Gene Mutated in Wiskott-Aldrich Syndrome** *JOURNAL OF IMMUNOLOGY*
  Derry, J. J., Ochs, H. D., Francke, U.
  2018; 200 (11): 3671–80

- **Germ line variants predispose to both JAK2 V617F clonal hematopoiesis and myeloproliferative neoplasms.** *Blood*
  Hinds, D. A., Barnholt, K. E., Mesa, R. A., Kiefer, A. K., Do, C. B., Eriksson, N., Mountain, J. L., Francke, U., Tung, J. Y., Nguyen, H. M., Zhang, H., Gojenola, L., Zehnder, et al
  2016; 128 (8): 1121-1128

- **A genome-wide association meta-analysis of self-reported allergy identifies shared and allergy-specific susceptibility loci** *NATURE GENETICS*
  Hinds, D. A., McMahon, G., Kiefer, A. K., Do, C. B., Eriksson, N., Evans, D. M., St Pourcain, B., Ring, S. M., Mountain, J. L., Francke, U., Davey-Smith, G., Timpson, N. J., Tung, et al
  2013; 45 (8): 907-U292

- **How will genomic information become integrated into the health care system?** *Molecular genetics & genomic medicine*
  Francke, U.
  2013; 1 (2): 67-70

- **Genome-Wide Association Analysis Implicates Elastic Microfibrils in the Development of Non-Syndromic Striae Distensae.** *The Journal of investigative dermatology*
  Tung, J. Y., Kiefer, A. K., Mullins, M., Francke, U., Eriksson, N.
  2013

- **2012 William Allan Award: Adventures in Cytogenetics** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Francke, U.
  2013; 92 (3): 325-337

- **Dealing with the unexpected: consumer responses to direct-access BRCA mutation testing** *PEERJ*
  Francke, U., Dijamco, C., Kiefer, A. K., Eriksson, N., Moiseff, B., Tung, J. Y., Mountain, J. L.
  2013; 1

- **Genome-Wide Analysis Points to Roles for Extracellular Matrix Remodeling, the Visual Cycle, and Neuronal Development in Myopia** *PLOS GENETICS*
  Kiefer, A. K., Tung, J. Y., Do, C. B., Hinds, D. A., Mountain, J. L., Francke, U., Eriksson, N.

2013; 9 (2)

- **Correlation of Symptom Assessment with Genotyping Analysis of Saliva Samples in a Large Cohort of Myeloproliferative Neoplasm Patients** *54th Annual Meeting and Exposition of the American-Society-of-Hematology (ASH)*
  Nguyen, H. (., Hinds, D. A., Barnholt, K. E., Kiefer, A. K., Do, C. B., Eriksson, N., Mountain, J. L., Francke, U., Tung, J. A., Levine, R. L., Zehnder, J. L., Gotlib, J., Mesa, et al
  AMER SOC HEMATOLOGY.2012

- **Estimation of JAK2 V617F Prevalence by Detection of the Mutation in Saliva Samples From Online MPN and General Population Cohorts** *54th Annual Meeting and Exposition of the American-Society-of-Hematology (ASH)*
  Barnholt, K. E., Hinds, D. A., Kiefer, A. K., Do, C. B., Eriksson, N., Mountain, J. L., Francke, U., Tung, J. A., Nguyen, H. (., Levine, R. L., Mesa, R. A., Gotlib, J., Zehnder, et al
  AMER SOC HEMATOLOGY.2012

- **Comparison of Family History and SNPs for Predicting Risk of Complex Disease** *PLOS GENETICS*
  Do, C. B., Hinds, D. A., Francke, U., Eriksson, N.
  2012; 8 (10)

- **Genetic variants associated with breast size also influence breast cancer risk** *BMC MEDICAL GENETICS*
  Eriksson, N., Benton, G. M., Do, C. B., Kiefer, A. K., Mountain, J. L., Hinds, D. A., Francke, U., Tung, J. Y.
  2012; 13

- **The new Ghent criteria for Marfan syndrome: what do they change?** *CLINICAL GENETICS*
  Faivre, L., Collod-Beroud, G., Ades, L., Arbustini, E., Child, A., Callewaert, B. L., Loeys, B., Binquet, C., Gautier, E., Mayer, K., Arslan-Kirchner, M., Grasso, M., Beroud, et al
  2012; 81 (5): 433-442

- **Novel Associations for Hypothyroidism Include Known Autoimmune Risk Loci** *PLOS ONE*
  Eriksson, N., Tung, J. Y., Kiefer, A. K., Hinds, D. A., Francke, U., Mountain, J. L., Do, C. B.
  2012; 7 (4)

- **Reduction of NADPH-Oxidase Activity Ameliorates the Cardiovascular Phenotype in a Mouse Model of Williams-Beuren Syndrome** *PLOS GENETICS*
  Campuzano, V., Segura-Puimedon, M., Terrado, V., Sanchez-Rodriguez, C., Coustets, M., Menacho-Marquez, M., Nevado, J., Bustelo, X. R., Francke, U., Perez-Jurado, L. A.
  2012; 8 (2)

- **Skeletogenic phenotype of human Marfan embryonic stem cells faithfully phenocopied by patient-specific induced-pluripotent stem cells** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
  Quarto, N., Leonard, B., Li, S., Marchand, M., Anderson, E., Behr, B., Francke, U., Reijo-Pera, R., Chiao, E., Longaker, M. T.
  2012; 109 (1): 215-220

- **Efficient Replication of over 180 Genetic Associations with Self-Reported Medical Data** *PLOS ONE*
  Tung, J. Y., Do, C. B., Hinds, D. A., Kiefer, A. K., Macpherson, J. M., Chowdry, A. B., Francke, U., Naughton, B. T., Mountain, J. L., Wojcicki, A., Eriksson, N.
  2011; 6 (8)

- **Web-Based Genome-Wide Association Study Identifies Two Novel Loci and a Substantial Genetic Component for Parkinson's Disease** *PLOS GENETICS*
  Do, C. B., Tung, J. Y., Dorfman, E., Kiefer, A. K., Drabant, E. M., Francke, U., Mountain, J. L., Goldman, S. M., Tanner, C. M., Langston, J. W., Wojcicki, A., Eriksson, N.
  2011; 7 (6)

- **Control of bone formation by the serpentine receptor Frizzled-9** *JOURNAL OF CELL BIOLOGY*
  Albers, J., Schulze, J., Beil, F. T., Gebauer, M., Baranowsky, A., Keller, J., Marshall, R. P., Wintges, K., Friedrich, F. W., Priemel, M., Schilling, A. F., Rueger, J. M., Cornils, et al
  2011; 192 (6): 1057-1072

- **Prognosis Factors in Probands With an FBN1 Mutation Diagnosed Before the Age of 1 Year** *PEDIATRIC RESEARCH*
  Stheneur, C., Faivre, L., Collod-Beroud, G., Gautier, E., Binquet, C., Bonithon-Kopp, C., Claustres, M., Child, A. H., Arbustini, E., Ades, L. C., Francke, U., Mayer, K., Arslan-Kirchner, et al
  2011; 69 (3): 265-270

- **Induced Chromosome Deletion in a Williams-Beuren Syndrome Mouse Model Causes Cardiovascular Abnormalities** *JOURNAL OF VASCULAR RESEARCH*
  Goergen, C. J., Li, H., Francke, U., Taylor, C. A.
  2011; 48 (2): 119-129

- **Cardiovascular manifestations in men and women carrying a FBN1 mutation** *EUROPEAN HEART JOURNAL*
  Detaint, D., Faivre, L., Collod-Beroud, G., Child, A. H., Loeys, B. L., Binquet, C., Gautier, E., Arbustini, E., Mayer, K., Arslan-Kirchner, M., Stheneur, C., Halliday, D., Beroud, et al
  2010; 31 (18): 2223-2229

- **Neonatal Maternal Deprivation Response and Developmental Changes in Gene Expression Revealed by Hypothalamic Gene Expression Profiling in Mice** *PLOS ONE*
  Ding, F., Li, H. H., Li, J., Myers, R. M., Francke, U.
  2010; 5 (2)

- **On the bumpy road towards 'personalized medicine'** *EMBO MOLECULAR MEDICINE*
  Francke, U.
  2010; 2 (1): 1-2

- **Widespread Changes in Dendritic and Axonal Morphology in Mecp2-Mutant Mouse Models of Rett Syndrome: Evidence for Disruption of Neuronal Networks** *JOURNAL OF COMPARATIVE NEUROLOGY*
  Belichenko, P. V., Wright, E. E., Belichenko, N. P., Masliah, E., Li, H. H., Mobley, W. C., Francke, U.
  2009; 514 (3): 240-258

- **Pathogenic FBN1 Mutations in 146 Adults Not Meeting Clinical Diagnostic Criteria for Marfan Syndrome: Further Delineation of Type 1 Fibrillinopathies and Focus on Patients With an Isolated Major Criterion** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Faivre, L., Collod-Beroud, G., Callewaert, B., Child, A., Loeys, B. L., Binquet, C., Gautier, E., Arbustini, E., Mayer, K., Arslan-Kirchner, M., Kiotsekoglou, A., Comeglio, P., Grasso, et al
  2009; 149A (5): 854-860

- **Clinical and mutation-type analysis from an international series of 198 probands with a pathogenic FBN1 exons 24-32 mutation** *EUROPEAN JOURNAL OF HUMAN GENETICS*
  Faivre, L., Collod-Beroud, G., Callewaert, B., Child, A., Binquet, C., Gautier, E., Loeys, B. L., Arbustini, E., Mayer, K., Arslan-Kirchner, M., Stheneur, C., Kiotsekoglou, A., Comeglio, et al
  2009; 17 (4): 491-501

- **Induced chromosome deletions cause hypersociability and other features of Williams-Beuren syndrome in mice** *EMBO MOLECULAR MEDICINE*
  Li, H. H., Roy, M., Kuscuoglu, U., Spencer, C. M., Halm, B., Harrison, K. C., Bayle, J. H., Splendore, A., Ding, F., Meltzer, L. A., Wright, E., Paylor, R., Deisseroth, et al
  2009; 1 (1): 50-65

- **Clinical and Molecular Study of 320 Children With Marfan Syndrome and Related Type I Fibrillinopathies in a Series of 1009 Probands With Pathogenic FBN1 Mutations** *PEDIATRICS*
  Faivre, L., Masurel-Paulet, A., Collod-Beroud, G., Callewaert, B. L., Child, A. H., Stheneur, C., Binquet, C., Gautier, E., Chevallier, B., Huet, F., Loeys, B. L., Arbustini, E., Mayer, et al
  2009; 123 (1): 391-398

- **Severe congenital encephalopathy caused by MECP2 null mutations in males: central hypoxia and reduced neuronal dendritic structure** *CLINICAL GENETICS*
  Schule, B., Armstrong, D. D., Vogel, H., Oviedo, A., Francke, U.
  2008; 74 (2): 116-126

- **Contribution of molecular analyses in diagnosing Marfan syndrome and type I fibrillinopathies: an international study of 1009 probands** *JOURNAL OF MEDICAL GENETICS*
  Faivre, L., Collod-Beroud, G., Child, A., Callewaert, B., Loeys, B. L., Binquet, C., Gautier, E., Arbustini, E., Mayer, K., Arslan-Kirchner, M., Stheneur, C., Kiotsekoglou, A., Comeglio, et al
  2008; 45 (6): 384-390

- **Comparative study of brain morphology in Mecp2 mutant mouse models of Rett syndrome** *JOURNAL OF COMPARATIVE NEUROLOGY*
  Belichenko, N. P., Belichenko, P. V., Li, H. H., Mobley, W. C., Francke, U.

2008; 508 (1): 184-195

- **SnoRNA Snord116 (Pwcr1/MBII-85) Deletion Causes Growth Deficiency and Hyperphagia in Mice** *PLOS ONE*
  Ding, F., Li, H. H., Zhang, S., Solomon, N. M., Camper, S. A., Cohen, P., Francke, U.
  2008; 3 (3)
- **Reduced aortic wall motion and cyclic strain in a Williams-Beuren syndrome mouse model due to changes in vessel structure** *80th Annual Scientific Session of the American-Heart-Association (AHA)*
  Goergen, C. J., Li, H. H., Abilez, O. J., Francke, U., Taylor, C. A.
  LIPPINCOTT WILLIAMS & WILKINS.2007: 33–33
- **A Marfan syndrome gene expression phenotype in cultured skin fibroblasts** *BMC GENOMICS*
  Yao, Z., Jaeger, J. C., Ruzzo, W. L., Morale, C. Z., Emond, M., Francke, U., Milewicz, D. M., Schwartz, S. M., Mulvihill, E. R.
  2007; 8
- **DLX5 and DLX6 expression is biallelic and not modulated by MeCP2 deficiency** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Schuele, B., Li, H. H., Fisch-Kohl, C., Purmann, C., Francke, U.
  2007; 81 (3): 492-506
- **Effect of mutation type and location on clinical outcome in 1,013 probands with Marfan syndrome or related phenotypes and FBN1 mutations: An international study** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Faivre, L., Collod-Beroud, G., Loeys, B. L., Child, A., Binquet, C., Gautier, E., Callewaert, B., Arbustini, E., Mayer, K., Arslan-Kirchner, M., Kiotsekoglou, A., Comeglio, P., Marziliano, et al
  2007; 81 (3): 454-466
- **Cerebellar gene expression profiles of mouse models for Rett syndrome reveal novel MeCP2 targets** *BMC MEDICAL GENETICS*
  Jordan, C., Li, H. H., Kwan, H. C., Francke, U.
  2007; 8
- **An atypical deletion of the Williams-Beuren syndrome interval implicates genes associated with defective visuospatial processing and autism** *JOURNAL OF MEDICAL GENETICS*
  Edelmann, L., Prosnitz, A., Pardo, S., Bhatt, J., Cohen, N., Lauriat, T., Ouchanov, L., Gonzalez, P. J., Manghi, E. R., Bondy, P., Esquivel, M., Monge, S., Delgado, et al
  2007; 44 (2): 136-143
- **Ube3a expression is not altered in Mecp2 mutant mice** *HUMAN MOLECULAR GENETICS*
  Jordan, C., Francke, U.
  2006; 15 (14): 2210-2215
- **NFAT dysregulation by increased dosage of DSCR1 and DYRK1A on chromosome 21** *NATURE*
  Arron, J. R., Winslow, M. M., Polleri, A., Chang, C., Wu, H., Gao, X., Neilson, J. R., Chen, L., Heit, J. J., Kim, S. K., Yamasaki, N., Miyakawa, T., Francke, et al
  2006; 441 (7093): 595-600
- **Identification of cis-regulatory elements for MECP2 expression** *HUMAN MOLECULAR GENETICS*
  Liu, J., Francke, U.
  2006; 15 (11): 1769-1782
- **Mechanisms of disease: neurogenetics of MeCP2 deficiency** *NATURE CLINICAL PRACTICE NEUROLOGY*
  Francke, U.
  2006; 2 (4): 212-221
- **Inactivating mutations in ESCO2 cause SC phocomelia and Roberts syndrome: No phenotype-genotype correlation** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Schule, B., Oviedo, A., Johnston, K., Pai, S., Francke, U.
  2005; 77 (6): 1117-1128
- **Lack of Pwcr1/MBII-85 snoRNA is critical for neonatal lethality in Prader-Willi syndrome mouse models** *MAMMALIAN GENOME*
  Ding, F., Prints, Y., Dhar, M. S., Johnson, D. K., Garnacho-Montero, C., Nicholls, R. D., Francke, U.
  2005; 16 (6): 424-431

- **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*
  Schule, B., Albalwi, M., Northrop, E., Francis, D. I., Rowell, M., Slater, H. R., Gardner, R. J., Francke, U.
  2005; 6

- **Frizzled 9 knock-out mice have abnormal B-cell development** *BLOOD*
  Ranheim, E. A., Kwan, H. C., Reya, T., Wang, Y. K., Weissman, I. L., FRANCKE, U.
  2005; 105 (6): 2487-2494

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

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

- **Absence of frizzled 9 affects self-renewal signals in developing murine B cells and alters mature B cell and plasma cell function.** *45th Annual Meeting and Exhibition of the American-Society-of-Hematology*
  Ranheim, E. A., Kwan, H. C., Wang, Y. K., Reya, T., FRANCKE, U., Weissman, I. L.
  AMER SOC HEMATOLOGY.2003: 49A–49A

- **Frizzled 9 knock-out mice have intrinsic defect in B cell development and function.** *53rd Annual Meeting of the American-Society-of-Human-Genetics*
  Kwan, H., Ranheim, E., Wang, Y., Reya, T., Weissman, I., FRANCKE, U.
  CELL PRESS.2003: 167–67

- **Frizzled 9 knockout mice have defects in B cell development and function** *90th Annual Meeting of the American-Association-for-Immunologists*
  Ranheim, E. A., Kwan, H. C., Wang, Y. K., Reya, T., Weissman, I. L., FRANCKE, U.
  FEDERATION AMER SOC EXP BIOL.2003: C95–C96

- **Gene expression patterns vary in clonal cell cultures from Rett syndrome females with eight different MECP2 mutations.** *BMC medical genetics*
  Traynor, J., Agarwal, P., Lazzeroni, L., Francke, U.
  2002; 3: 12-?

- **Evidence for the role of PWCR1/HBII-85 C/D box small nucleolar RNAs in Prader-Willi syndrome** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Gallagher, R. C., Pils, B., Albalwi, M., Francke, U.
  2002; 71 (3): 669-678

- **Premature termination mutations in FBN1: Distinct effects on differential allelic expression and on protein and clinical phenotypes** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Schrijver, I., Liu, W. G., Odom, R., Brenn, T., Oefner, P., FURTHMAYR, H., Francke, U.
  2002; 71 (2): 223-237

- **Spontaneous spinal cerebrospinal fluid leaks and minor skeletal features of Marfan syndrome: a microfibrillopathy** *JOURNAL OF NEUROSURGERY*
  Schrijver, I., Schievink, W. I., Godfrey, M., Meyer, F. B., FRANCKE, U.
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- **Spectrum of MECP2 mutations in Rett syndrome** *World Congress on Rett Syndrome*
  Lee, S. S., Wan, M. M., Francke, U.
  ELSEVIER SCIENCE BV.2001: S138–S143

- **frizzled 9 is expressed in neural precursor cells in the developing neural tube** *DEVELOPMENT GENES AND EVOLUTION*
  Van Raay, T. J., Wang, Y. K., Stark, M. R., Rasmussen, J. T., FRANCKE, U., Vetter, M. L., Rao, M. S.
  2001; 211 (8-9): 453-457

- **Guidelines for reporting clinical features in cases with MECP2 mutations** *BRAIN & DEVELOPMENT*
  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, et al
  2001; 23 (4): 208-211

- **Evolutionary relationships among Rel domains indicate functional diversification by recombination** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
  Graef, I. A., Gastier, J. M., FRANCKE, U., Crabtree, G. R.
  2001; 98 (10): 5740-5745

- **MECP2 truncating mutations cause histone H4 hyperacetylation in Rett syndrome** *HUMAN MOLECULAR GENETICS*
  Wan, M. M., Zhao, K. J., Lee, S. S., FRANCKE, U.
  2001; 10 (10): 1085-1092

- **Human genetics around the world** *GENETICS IN MEDICINE*
  FRANCKE, U.
  2001; 3 (3): 167-167

- **Association of acetylated histones with paternally expressed genes in the Prader-Willi deletion region** *HUMAN MOLECULAR GENETICS*
  Fulmer-Smentek, S. B., FRANCKE, U.
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- **Williams (Williams Beuren) syndrome: A distinct neurobehavioral disorder** *JOURNAL OF CHILD NEUROLOGY*
  Kaplan, P., Wang, P. P., FRANCKE, U.
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- **The human genome project: implications for the endocrinologist.** *Journal of pediatric endocrinology & metabolism*
  FRANCKE, U.
  2001; 14: 1395-1408

- **Multi-exon deletions of the FBN1 gene in Marfan syndrome.** *BMC medical genetics*
  Liu, W., Schrijver, I., Brenn, T., FURTHMAYR, H., FRANCKE, U.
  2001; 2: 11-?

- **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** *AMERICAN JOURNAL OF HUMAN GENETICS*
  de los Santos, T., Schweizer, J., Rees, C. A., Francke, U.
  2000; 67 (5): 1067-1082

- **Discovery of the Rett syndrome gene and its function** *TURKISH JOURNAL OF PEDIATRICS*
  FRANCKE, U.
  2000; 42 (4): 271-271

- **Rett syndrome is caused by mutations in the X-linked MECP2 gene encoding methyl-CpG-binding protein**
  Zoghbi, H. Y., Amir, R. E., Wan, M., Lee, S. S., Van den Veyver, I. B., Tran, C. Q., Malicki, D., Schanen, N. C., FRANCKE, U.
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