

CURRICULUM VITAE

February 25, 2026

NAME		James Lewis Weber
DATE OF BIRTH		October 19, 1952
MARITAL STATUS		Married, two children
EDUCATION	1970-1972	University of Wisconsin-Milwaukee
	1972-1974	University of Wisconsin-Madison B.S. with Honors in Chemistry
	1975-1980	University of California-Berkeley Ph.D. in Biochemistry
RESEARCH AND PROFESSIONAL POSITIONS	1975	Research assistant, University of Wisconsin, M. Sundaralingam. Model building of nucleic acids, especially tRNA.
	1976-1980	Graduate research, University of California, R. D. Cole. Purification and characterization of chromatin fragments containing bovine satellite DNA.
	1980-1982	Postdoctoral Research, University of Wisconsin, Jack Gorski. Repetitive DNA structure and DNA methylation of the rat prolactin gene.
	1983-1986	Investigator, Walter Reed Army Institute of Research. Molecular biology of human malaria parasites.
	1986-2005	Senior Research Scientist, Marshfield Medical Research Foundation. Human genetics.
	1994-2005	Director, Center for Medical Genetics, Marshfield Medical Research Foundation.
	1994-2005	Director, NHLBI Mammalian Genotyping Service
	2004-2021	Founder, President and CEO, PreventionGenetics LLC
	2006-2018	Adjunct Scientist, Marshfield Clinic Research Foundation
	2011-2017	Member, Board of Trustees, Marshfield Clinic Research Foundation
	2019-	Clinical Adjunct Assistant Professor, University of Wisconsin-Madison
HONORS	1974	Helpaer scholarship, Department of Chemistry, University of Wisconsin
	1980-1981	Postdoctoral fellowship, American Cancer Society
	1981-1982	Postdoctoral fellowship, National Institutes of Health

James L. Weber, Ph.D.

1984 Army Commendation Medal for work on the malaria circumsporozoite protein gene
1991 Sebold Award from Marshfield Clinic for outstanding researcher
2002 ISI Highly Cited Researcher (< 0.5% of researchers)
2003 Lancet Paper of the Year (Rosenberg et al. Genetic structure of human populations. **Science** 298:2381-2385, 2002)
2012 Marshfield Clinic Heritage Award
2020 Wisconsin Biohealth Business Achievement Award

GRANTS AND CONTRACTS

1987 U. S. Army
Cloning and sequencing the 3' end of the serine-rich antigen gene of the human malaria parasite *Plasmodium falciparum*. \$18,400
1989-1990 Tourette Syndrome Association
Mapping of Tourette Genes Through Linkage Analysis. \$19,700
1989-1992 National Institutes of Health
Analysis of an abundant class of human DNA polymorphisms. \$580,000
1990-1991 Tourette Syndrome Association
Linkage Mapping of Tourette Syndrome Gene(s) using Microsatellite DNA Polymorphisms. \$30,000
1991-1992 National Institutes of Health
Analysis of an abundant class of human DNA polymorphisms -- Supplement. \$485,000
1991-1992 Tourette Syndrome Association
Linkage Mapping of Tourette Syndrome Gene(s). \$25,000.
1991-1995 National Institutes of Health
Epilepsy Program Project (Co-Investigator). \$215,000.
1992-1996 National Institutes of Health
Cooperative Human Linkage Center (CHLC) (Co-Principal Investigator). \$1,425,000.
1992-1997 National Institutes of Health
Asthma Genetics (Co-Investigator). \$406,000.
1994-1998 Glaxo-Wellcome Pharmaceuticals
Type II diabetes gene mapping. \$1,800,000.
1994-1995 Tourette Syndrome Association
Patch mapping of Tourette Syndrome genes. \$6,500.
1994-1999 National Institutes of Health
Mammalian Genotyping Service. \$3,912,000.
1996-1999 National Institutes of Health
Instrumentation for DNA Detection. \$476,000.

James L. Weber, Ph.D.

1996-1999	National Institutes of Health Supplement, Mammalian Genotyping Service. \$2,415,000.
1998-1999	Tourette Syndrome Association Tourette Gene Mapping in Afrikaners. \$33,460.
1998-2002	National Institutes of Health Insertion/Deletion Polymorphisms. \$1,906,000.
1999-2006	National Institutes of Health Mammalian Genotyping Service. \$22,562,000.

PATENTS

Length Polymorphisms in (dC-dA)_n:(dG-dT)_n Sequences #5,075,217, issued December 24, 1991.
Length Polymorphisms in (dC-dA)_n:(dG-dT)_n Sequences and Methods of Using the Same #5,582,979, issued December 10, 1996.
Apparatus and Method for Testing and Continuously Reading Low-Volume Samples, #7,232,547, issued June 19, 2007.

PEER REVIEW

1984-1985	Review Committees for Research Proposals, U. S. Agency for International Development
1987-1988	Consultant to American Institute of Biological Sciences for review of malaria grant proposals
1990-1995	Site Visit Committees for NIH Human Genome Initiative Center Grants
1991	Ad-hoc member, NIH Genome Study Section
1991-1993	Editorial Board, PCR Methods and Applications Cold Spring Harbor Press
1991	Site Visit Committee for Genome Data Base
1991-1995	Member, NIH Genome Research Review Committee (Study Section)
1995-2005	Chair, NHLBI Mammalian Genotyping Service Advisory Panel
1996-1998	Associate Editor, American Journal of Human Genetics
1996	Ad-hoc member NIH Genome Study Section
1997	Review Panel, NHGRI Technologies for Genome Analysis
1997	U.S. Army Breast Cancer Research Review Panel
1997-1999	Member, NHGRI Scientific Advisory Panel for Human Genome Sequencing Centers
1999	Review Committee, NHGRI Mouse Genome Sequencing
1999	Chair, Review Panel, NHGRI Quality Assessment for Genomic Sequence
2000	Ad-hoc member NIH Genome Study Section
2001	Review Committee, Juvenile Diabetes Research Foundation

James L. Weber, Ph.D.

		Review Committee, NHGRI Centers of Excellence in Genomic Science
	2002	Review Committees: NHGRI Genome Sequencing Centers, NIDDK diabetes gene hunting, and USDA bovine sequencing
	2003-2004	Editorial Board, BMC Biology
	1988-2008	Review Committee: NHGRI Sequencing Technology Manuscript review for many journals including Nature, Nature Genetics, American Journal of Human Genetics, Nature Reviews Genetics, Genome Research.
	2011	Review Committee: NHGRI Genome Sequencing Centers
OTHER PROFESSIONAL COMMITTEES	1992-1993	Wisconsin State Legislative Council on Genetic and Medical Information.
	1992	NIGMS Human Genetic Mutant Cell Repository Evaluation
	1995	Ad-hoc member, NIH Genome Advisory Council
	1999	Planning committee, NHLBI Centers for Genomic Applications
	2003	Planning committee, DOE JGI Sequencing Service
	2003	Scientific Advisory Board, Finnish Genome Center
	2010-2011	Chair, Wisconsin Genomics Initiative Scientific Advisory Board
	2019-2022	ClinGen Expert Panel on Malignant Hyperthermia
SOCIETY MEMBERSHIP		American Society of Human Genetics Human Genome Organization (HUGO) American Association for the Advancement of Science

James L. Weber, Ph.D.

PUBLICATIONS

1. Milman, G., Anton, D. L., and Weber, J. L. Chinese Hamster purine-nucleoside phosphorylase: purification, structural, and catalytic properties. **Biochemistry** 15:4967-4973, 1976.
2. Weber, J. L. Purification and characterization of bovine satellite chromatin. **Ph.D. Thesis**, University of California-Berkeley, 1980.
3. Weber, J. L. and Cole, R. D. Chromatin fragments containing bovine 1.715 g/ml satellite DNA: purification by chromatography on malachite green DNA affinity resin. **J. Biol. Chem.** 257:11774-11783, 1982.
4. Weber, J. L. and Cole, R. D. Chromatin fragments containing bovine 1.715 g/ml satellite DNA: nucleosome structure and protein composition. **J. Biol. Chem.** 257:11784-11790, 1982.
5. Schuler, L. A., Weber, J. L. and Gorski, J. Polymorphism near the rat prolactin gene caused by insertion of an Alu-like element. **Nature** 305:159-160, 1983.
6. Durrin, L. K., Weber, J. L. and Gorski, J. Chromatin structure, transcription, and methylation of the prolactin gene domain in pituitary tumors of Fischer 344 rats. **J. Biol. Chem.** 259:7086-7093, 1984.
7. Weber, J. L., Durrin, L. K. and Gorski, J. Repetitive DNA sequences within and around the prolactin gene. **Mol. Cell. Biochem.** 65:171-179, 1984.
8. Dame, J. B., Williams, J. L., McCutchan, T. F., Weber, J. L., Wirtz, R. A., Hockmeyer, W. T., Maloy, W. T., Haynes, J. D., Schneider, I., Roberts, D., Sanders, G. S., Reddy, E. P., Diggs, C. L. and Miller, L.H. Structure of the gene encoding the immunodominant surface antigen on the sporozoite of the human malaria parasite *Plasmodium falciparum*. **Science** 225:593-599, 1984.
9. Weber, J. L. and Hockmeyer, W. T. Structure of the circumsporozoite protein gene in 18 strains of *Plasmodium falciparum*. **Mol. Biochem. Parasitol.** 15:305-316, 1985.
10. Gorski, J., Shull, J., Weber, J. and Durrin, L. Estrogen regulation of prolactin gene transcription and chromatin structure. **In: Prolactin. Basic and Clinical Correlates**, MacLeod, R. M., Thorner, M. O. and Scapagnini, U. (eds), Fida Research Series. Vol. 1, Liviana Press, Padova, pp. 259-269, 1985.
11. Lyon, J. A., Geller, R. H., Haynes, J. D., Chulay, J. D. and Weber, J. L. Epitope map and processing scheme for the 195,000 dalton surface glycoprotein of *Plasmodium falciparum* merozoites deduced from cloned overlapping segments of the gene. **Proc. Natl. Acad. Sci.** 83:2989-2993, 1986.

James L. Weber, Ph.D.

12. Weber, J. L., Leininger, W. M. and Lyon, J. A. Variation in the gene encoding a major merozoite surface antigen of the human malaria parasite *Plasmodium falciparum*. **Nucleic Acids Res.** 14:3311-3323, 1986.
13. Weber, J. L., Lyon, J. A. and Camus, D. Blood stage antigen genes of *Plasmodium falciparum*, in Molecular Strategies of Parasitic Invasion, **UCLA Symposia on Molecular and Cellular Biology**, New Series, Vol. 42, Agabian, N., Goodman, H. and Noguiera, N. editors, Alan R. Liss, New York, 1987, pp. 379-388.
14. Weber, J. L. Analysis of sequences from the extremely AT-rich genome of *Plasmodium falciparum*. **Gene** 52:103-109, 1987.
15. Weber, J. L., Egan, J. E., Lyon, J. A., Wirtz, R. A., Charoenvit, Y., Maloy, W. L. and Hockmeyer, W. T. *Plasmodium berghei*: Cloning of the circumsporozoite protein gene. **Exp. Parasitol.** 63:295-300, 1987.
16. Egan, J. E., Weber, J. L., Ballou, W. R., Majarian, W. R., Gordon, D. M., Hoffman, S. L., Wirtz, R. A., Schneider, I., Woollett, G. R., Hollingdale, M. R., Young, J. F., and Hockmeyer, W. T. Efficacy of murine malaria sporozoite vaccines: Implications for human vaccine development. **Science** 236:453-456, 1987.
17. Campbell, G. H., Aley, S. B., Ballou, W. R., Hall, T., Hockmeyer, W. T., Hoffman, S. L., Hollingdale, M. R., Howard, R. J., Lyon, J. A., Nardin, E. H., Nussenzweig, R. S., Nussenzweig, V., Tsang, V. C. W., Weber, J. L., Wellem, T. E., Young, J. F., and Zavala, F. Use of synthetic and recombinant peptides in the study of host-parasite interactions in the malarial. **Am. J. Trop. Med. Hyg.** 37:428-444, 1987.
18. Lyon, J. A. and Weber, J. L. Preparation and use of monospecific antibodies selected using recombinant expression proteins adsorbed to nitrocellulose. **In: CRC Handbook of Immunoblotting of Proteins**, Vol. 2, Experimental and Clinical Applications, Bjerrum, O. J. and Heegard, N. H. H. (eds), CRC Press, Boca Raton, Florida, 1988, pp 95-104.
19. Weber, J. L. A Review: Molecular biology of malaria parasites, **Exp. Parasitol.** 66:143-170, 1988.
20. Chulay, J. D., Lyon, J. A., Wolff, R. H., Hall, T., Nagasawa, H., Aikawa, M., and Weber, J. L. Primary structure of a lysine and glutamate rich *Plasmodium falciparum* antigen located at the merozoite surface and in the parasitophorous vacuole, in Technological Advances in Vaccine Development, **UCLA Symposia on Molecular and Cellular Biology**, Laskey, L. editor, AR Liss, New York, 1988, pp. 35-43.
21. Weber, J. L., Sim, B. K. L., Lyon, J. A., and Wolff, R. Merozoite surface protein sequence from the Camp strain of the human malaria parasite *Plasmodium falciparum*. **Nucleic Acids Res.** 16:1206, 1988.

James L. Weber, Ph.D.

22. Weber, J. L. *Plasmodium falciparum*: Mapping genes to nine parasite chromosomes. **Exp. Parasitol.** 65:148-153, 1988.
23. Weber, J. L. Interspersed repetitive DNA from *Plasmodium falciparum*. **Mol. Biochem. Parasitol.** 29:117-124, 1988.
24. Weber, J. L., Lyon, J. A., Wolff, R. H., Hall, T., and Chulay, J. D. Primary structure of a *Plasmodium falciparum* malaria antigen located at the merozoite surface and within the parasitophorous vacuole. **J. Biol. Chem.** 263:11421-11425, 1988.
25. Delplace, P., Bhatia, A., Cagnard, M., Camus, D., Colombet, G., Debrabant, A., Dubremetz, J. F., Dubreuil, N., Prensier, G., Fortier, B., Haq, A., Weber, J., and Vernes, A. Protein p126: a parasitophorous vacuole antigen associated with the release of *Plasmodium falciparum* merozoites. **Biol Cell** 64:215-221, 1988.
26. Weber, J. L. and May, P. E. Abundant class of human DNA polymorphisms which can be typed using the polymerase chain reaction. **Am. J. Hum. Genet.** 44:388-396, 1989.
27. Weber, J.L. Human DNA polymorphisms based on length variations in simple sequence tandem repeats. **In: Genome Analysis Series**, Vol. 1: Genetic and Physical Mapping, Tilghman, S., and Davies, K. (eds.), Cold Spring Harbor Laboratory Press, pp 159-181, 1990.
28. Weber, J. L. Human DNA polymorphisms and methods of analysis. **Curr. Opin. Biotechnol.** 1:166-171, 1990.
29. Weber, J. L., May, P. E., and Kappel, C. Dinucleotide repeat polymorphism at the D19S49 locus. **Nucleic Acids Res.** 18:1927, 1990.
30. Heutink, P., van de Wetering, B. J. M., Breedveld, G. J., Weber, J., Sandkuyl, L. A., Devor, E. J., Heiberg, A., Niermeijer, M. F., and Oostra, B. A. No evidence for genetic linkage of Gilles de la Tourette Syndrome on chromosome 7 and 18. **J. Med. Genet.** 27:433-436, 1990.
31. Weber, J. L., Kwitek, A. E., and May, P. E. Dinucleotide repeat polymorphism at the D1S102 locus. **Nucleic Acids Res.** 18:2199, 1990.
32. Weber, J. L., Kwitek, A. E., and May, P. E. Dinucleotide repeat polymorphism at the D1S103 locus. **Nucleic Acids Res.** 18:2199, 1990.
33. Weber, J. L. and May, P. E. Dinucleotide repeat polymorphism at the PENK locus. **Nucleic Acids Res.** 18:2200, 1990.

James L. Weber, Ph.D.

34. Weber, J. L. and May, P. E. Dinucleotide repeat polymorphism at the D2S72 locus. **Nucleic Acids Res.** 18:2200, 1990.
35. Weber, J. L. and May, P. E. Dinucleotide repeat polymorphism at the D18S34 locus. **Nucleic Acids Res.** 18:2201, 1990.
36. Weber, J. L. and May, P. E. Dinucleotide repeat polymorphism at the D3S240 locus. **Nucleic Acids Res.** 18:2201, 1990.
37. Weber, J. L. and May, P. E. Dinucleotide repeat polymorphism at the D4S171 locus. **Nucleic Acids Res.** 18:2202, 1990.
38. Weber, J. L. and May, P. E. Dinucleotide repeat polymorphism at the D20S27 locus. **Nucleic Acids Res.** 18:2202, 1990.
39. Weber, J. L. and May, P. E. Dinucleotide repeat polymorphism at the D9S43 locus. **Nucleic Acids Res.** 18:2203, 1990.
40. Weber, J. L. and May, P. E. Dinucleotide repeat polymorphism at the D2S71 locus. **Nucleic Acids Res.** 18:2203, 1990.
41. Weber, J. L. Informativeness of human (dC-dA)_n:(dG-dT)_n polymorphisms. **Genomics** 7:524-530, 1990.
42. Weber, J. L. and May, P. E. Dinucleotide repeat polymorphism at the D19S76 locus. **Nucleic Acids Res.** 18:2835, 1990.
43. Weber, J. L., Kwitek, A. E., and May, P. E. Dinucleotide repeat polymorphism at the D1S104 locus. **Nucleic Acids Res.** 18:2835, 1990.
44. Lewis, J. G., Weber, J. L., Petersen, M. B., Slaughaupt, S. A., Kwitek, A., May, P. E., Warren, A. C., Chakravarti, A., and Antonarakis, S. E. Linkage mapping of the highly informative DNA marker D21S156 to human chromosome 21 using a polymorphic GT dinucleotide repeat. **Genomics** 8:400-402, 1990.
45. Weber, J. L., Kwitek, A. E., and May, P. E. Dinucleotide repeat polymorphisms at the D16S260, D16S261, D16S265, D16S266, and D16S267 loci. **Nucleic Acids Res.** 18:4034, 1990.
46. Weber, J. L., Kwitek, A. E., and May, P. E. Dinucleotide repeat polymorphisms at the D5S107, D5S108, D5S111, D5S117, and D5S118 loci. **Nucleic Acids Res.** 18:4035, 1990.
47. Weber, J. L., Kwitek, A. E., and May, P. E. Dinucleotide repeat polymorphisms at the D11S419 and CD3D loci. **Nucleic Acids Res.** 18:4036, 1990.

James L. Weber, Ph.D.

48. Weber, J. L., Kwitek, A. E., May, P. E., Polymeropoulos, M. H., and Ledbetter, S. Dinucleotide repeat polymorphisms at the DXS453, DXS454, and DXS458 loci. **Nucleic Acids Res.** 18:4037, 1990.
49. Weber, J. L., Kwitek, A. E., May, P. E., Patterson, D., and Drabkin, H. Dinucleotide repeat polymorphisms at the D8S85, D8S87, and D8S88 loci. **Nucleic Acids Res.** 18:4038, 1990.
50. Weber, J. L., Kwitek, A. E., and May, P. E. Dinucleotide repeat polymorphisms at the D7S435 and D7S440 loci. **Nucleic Acids Res.** 18:4039, 1990
51. Wijmenga, C., Frants, R. R., Brouwer, O. F., Moerer, P., Weber, J. L., and Padberg, G. W. Location of the fascioscapulohumeral muscular dystrophy gene on chromosome 4. **Lancet**, 336:651-653, 1990.
52. Weber, J. L., Kwitek, A. E., and May, P. E. Dinucleotide repeat polymorphism at the CRP locus. **Nucleic Acids Res.** 18:4635, 1990.
53. Weber, J. L., May, P. E., Patterson, D., Drabkin, H., and Killary, A. M. Dinucleotide repeat polymorphism at the D3S196 locus. **Nucleic Acids Res.** 18:4635, 1990.
54. Weber, J. L., Kwitek, A. E., and May, P. E. Dinucleotide repeat polymorphism at the D4S174 locus. **Nucleic Acids Res.** 18:4636, 1990.
55. Weber, J. L., Kwitek, A. E., and May, P. E. Dinucleotide repeat polymorphism at the D6S87 locus. **Nucleic Acids Res.** 18:4636, 1990.
56. Weber, J. L. and May, P. E. Dinucleotide repeat polymorphism at the D10S89 locus. **Nucleic Acids Res.** 18:4637, 1990.
57. Weber, J. L., Kwitek, A. E., May, P. E., and Polymeropoulos, M. H. Dinucleotide repeat polymorphism at the D12S43 locus. **Nucleic Acids Res.** 18:4637, 1990.
58. Weber, J. L., Kwitek, A. E., and May, P. E. Dinucleotide repeat polymorphism at the D13S71 locus. **Nucleic Acids Res.** 18:4638, 1990.
59. Weber, J. L., Kwitek, A. E., and May, P. E. Dinucleotide repeat polymorphism at the D14S34 locus. **Nucleic Acids Res.** 18:4638, 1990.
60. Weber, J. L., Kappel, C., May, P. E., and Kwitek, A. E. Dinucleotide repeat polymorphism at the D19S75 locus. **Nucleic Acids Res.** 18:4639, 1990.
61. Weber, J. L. and May, P. E. Dinucleotide repeat polymorphism at the D22S156 locus. **Nucleic Acids Res.** 18:4639, 1990.

James L. Weber, Ph.D.

62. Weber, J. L., Kwitek, A. E., and May, P. E. Dinucleotide repeat polymorphism at the D15S87 locus. **Nucleic Acids Res.** 18:4640, 1990.
63. Weber, J. L., Kwitek, A. E., May, P. E., Wallace, M. R., Collins, F. S., and Ledbetter, D. H. Dinucleotide repeat polymorphisms at the D17S250 and D17S261 loci. **Nucleic Acids Res.** 18:4640, 1990.
64. Weber, J. L. and May, P. E. Dinucleotide repeat polymorphism at the D18S35 locus. **Nucleic Acids Res.** 18:6465, 1990.
65. Pakstis, A. J. et al. Progress in the search for genetic linkage with Tourette Syndrome: An exclusion map covering more than 50% of the autosomal genome. **Am. J. Hum. Genet.** 48:281-294, 1991.
66. Weber, J. L., Kwitek, A. E., May, P. E., and Zoghbi, H. Y. Dinucleotide repeat polymorphism at the D6S105 locus. **Nucleic Acids Res.** 19:968, 1991.
67. Herring, W. J., Litwer, S., Weber, J. L., and Danner, D. J. Molecular genetic basis of maple syrup urine disease in a family with two defective alleles for branched chain acyltransferase and localization of the gene to human chromosome 1. **Am. J. Hum. Genet.** 48:342-350, 1991.
68. Wijmenga, C., Padberg, G. W., Moerer, P., Wiegant, J., Liem, L., Brouwer, O. F., Milner, E. C. B., Weber, J. L., Sandkuyl, L. A., van Ommen, G. B. and Frants, R. R. Mapping of facioscapulohumeral muscular dystrophy gene to chromosome 4q35-qter by multipoint linkage analysis and in situ hybridization. **Genomics** 9:570-575, 1991.
69. Heutink, P., Sandkuyl, L. A., Van de Wetering, B., Oostra, B. A., Weber, J., Wilkie, P., Devor, E. J., Pakstis, A. J., Pauls, D., Kidd, K. K. Linkage and Tourette syndrome (letter). **Lancet** 337:122, 1991.
70. Dracopoli, N. C., et al. The CEPH consortium linkage map of human chromosome 1. **Genomics** 9:686-700, 1991.
71. Ning, Y., Weber, J. L., Killary, A. M., Ledbetter, D. H., Smith, J. R., and Pereira-Smith, O. M. Introduction of a normal human chromosome 4 reverses the immortal phenotype of Hela Cells. **Proc. Natl. Acad. Sci.** 88:5635-5639, 1991.
72. Peterson, M. B., Weber, J. L., Slaughterhaupt, S. A., Kwitek, A. E., McInnis, M. G., Chakravarti, A., and Antonarakis, S. E. Linkage mapping of D21S171 to the distal long arm of human chromosome 21 using a polymorphic (AC)_n dinucleotide repeat. **Hum. Genet.** 87:401-404, 1991.

James L. Weber, Ph.D.

73. Jabs, E. W., Li, X., Coss, C.A., Taylor, E. W., Meyers, D. A., and Weber, J. L. Mapping the Treacher Collins syndrome locus to 5q31.3-5q33.3. **Genomics** 11:193-198, 1991.
74. Weber, J. L., Polymeropoulos, M., May, P., Kwitek, A., Xiao, H., McPherson, J. D., and Wasmuth, J. J. Mapping of human chromosome 5 microsatellite polymorphisms. **Genomics** 11:695-700, 1991.
75. Small, K. W., Weber, J. L., Hung, W., Vance, J., Roses, A., Pericak-Vance, M. North Carolina macular dystrophy: exclusion map using RFLPs and microsatellites. **Genomics** 11:763-766, 1991.
76. Wilkie, P. J., Ahmann, P. A., Hardacre, J., LaPlant, R. J., Hiner, B. C., and Weber, J. L. Application of microsatellite DNA polymorphisms to linkage mapping of Tourette Syndrome gene(s). 1992. **In: Advances in Neurology**, Vol. 58, T. N. Chase, A. J. Friedhoff, and D. J. Cohen (eds.), Raven Press, NY, pp. 173-180.
77. Beckmann, J. S. and Weber, J. L. Survey of human and rat microsatellites. **Genomics** 12:627-631, 1992.
78. Decker, R. A., Moore, J., Ponder, B. and Weber, J. L. Linkage mapping of human chromosome 10 microsatellite polymorphisms. **Genomics** 12:604-606, 1992.
79. Wilkie, P. J., Krizman, D., and Weber, J. L. A linkage map of human chromosome 9 microsatellite DNA polymorphisms. **Genomics** 12:607-609, 1992.
80. Hall, J. M., Friedman, L., Guenther, C., Lee, M. K., Weber, J. L., Black, D. M., and King, M.-C. Closing in on a breast cancer gene on chromosome 17q. **Am. J. Hum. Genet.** 50:1235-1242, 1992.
81. Goto, M., Rubenstein, M., Weber, J. L., Woods, K., and Drayna, D. Genetic linkage of the Werner's syndrome gene to five markers on chromosome 8. **Nature** 355:735-738, 1992.
82. Wang, Z., and Weber, J. L. Continuous linkage map of human chromosome 14 short tandem repeat polymorphisms. **Genomics** 13:532-536, 1992.
83. Speer, M. C., Yamaoka, L. H., Gilchrist, J. M., Gaskell, C. P., Stajich, J. M., Vance, J. M., Kazantsev, A., Lastra, A. A., Haynes, C. S., Beckmann, J. S., Cohen, D., Weber, J. L., Roses, A. D., and Pericak-Vance, M. A. Confirmation of genetic heterogeneity in limb-girdle muscular dystrophy: linkage of an autosomal dominant form to chromosome 5q. **Am. J. Hum. Genet.** 50:1211-1217, 1992.
84. Purohit, K., Weber, J. L., Ward, L. J., and Keats, B. J. B. The Kell blood group locus is close to the cystic fibrosis locus on chromosome 7. **Hum. Genet.** 89:457-458, 1992.

James L. Weber, Ph.D.

85. Small, K. W., Weber, J. L., Roses, A., Lennon, F., Vance, J. M., Pericak-Vance, M. A. North Carolina macular dystrophy is assigned to chromosome 6. **Genomics** 13:681-685, 1992.
86. Heutink, P., van der Mey, A. G. L., Sandkuijl, L. A., van Gils, A. P. G., Bardoel, A., Breedveld, G. J., van Vliet, M., van Ommen, G. B., Cornelisse, C. J., Oostra, B. A., Weber, J. L., and Devilee, P. A gene subject to genomic imprinting and responsible for hereditary paragangliomas maps to chromosome 11q23-qter. **Hum. Mol. Genet.** 1:7-10, 1992.
87. Matsutani, A., Hing, A., Steinbrueck, T., Janssen, R., Weber, J., Permutt, M. A., and Donis-Keller, H. Mapping the human liver/islet glucose transporter (GLUT-2) gene within a genetic linkage map of chromosome 3q using a (CA)_n dinucleotide repeat polymorphism and characterization of the polymorphism in 3 racial groups. **Genomics** 13:495-501, 1992.
88. Mills, K. A., Beutow, K. H., Weber, J. L., Altherr, M. R., Wasmuth, J. J., and Murray, J. C. Genetic and physical maps of human chromosome 4 based on dinucleotide repeats. **Genomics**, 14:209-219, 1992.
89. Tomfohrde, J., Wood, S., Schertzer, M., Wagner, M. J., Wells, D. E., Parrish, J., Sadler, L. A., Blanton, S. H., Daiger, S. P., Wang, Z., Wilkie, P. J., and Weber, J. L. Human chromosome 8 linkage map based on short tandem repeat polymorphisms: effect of genotyping errors. **Genomics** 14:144-152, 1992.
90. Kandt, R. S., Haines, J. L., Smith, M., Northrup, H., Gardner, R. J. M., Short, M. P., Dumars, K., Roach, E. S., Steingold, S., Wall, S. L., Blanton, S. H., Flodman, P., Kwiatkowski, D. J., Jewell, A., Weber, J. L., Roses, A. D., and Pericak-Vance, M. A. Linkage of a major gene locus for tuberous sclerosis to a chromosome-16 marker for polycystic kidney disease. **Nature Genet.** 2:37-41, 1992.
91. Hubert, R., Weber, J. L., Schmitt, K., Lin, X., and Arnheim, N. A new source of polymorphic DNA markers for sperm typing: analysis of microsatellite repeats in single cells. **Am. J. Hum. Genet.** 51:985-991, 1992.
92. Murray, J. C., Bennett, S. R., Kwitek, A. E., Small, K. W., Schinzel, A., Alward, W. L. M., Weber, J. L., Bell, G. I., and Buetow, K. H. Linkage of Rieger Syndrome in the region of epidermal growth factor on chromosome 4. **Nature Genet.** 2:46-49, 1992.
93. NIH/CEPH Collaborative Mapping Group. A Comprehensive Genetic Linkage Map of the Human Genome. **Science** 258:67-86, 1992.
94. Nancarrow, D. J., Walker, G. J., Weber, J. L., Walters, M. K., Palmer, J. M., and Hayward, N. K. Linkage mapping of melanoma (MLM) using 172 microsatellite markers. **Genomics** 14:939-947, 1992.

James L. Weber, Ph.D.

95. Schellenberg, G. D., Bird, T. D., Wijsman, E. M., Orr, H. T., Anderson, L., Nemens, E., White, J. A., Bonnycastle, L., Weber, J. L., Alonso, E., Potter, H., Heston, L. L., and Martin, G. M. Genetic linkage evidence for a familial Alzheimer Disease locus on chromosome 14. **Science** 258:668-671, 1992.
96. Decker, R. A., Ponder, B. A., Mole, S., and Weber, J. L. Dinucleotide repeat polymorphisms at the D10S183 and D10S245 loci. **Hum. Mol. Genet.** 1:777, 1992.
97. Plaetke, R., Weber, J., Wood, S., Dean, M., Jeffreys, A. J., Weiffenbach, B., Vergnaud, G., Vogelstein, B., and White, R. Report of the Second International Workshop for chromosome 5: Consensus genetic map. **Cytogenet. Cell Genet.** 61:226-231, 1992.
98. Spurr, N. K. et. al. The CEPH consortium linkage map of human chromosome 2. **Genomics** 14:1055-1063, 1992.
99. Engelstein, M., Hudson, T. J., Lane, J. M., Leverone, B., Landes, G. M., Peltonen, L., Weber, J. L., and Dracopoli, N. C. A PCR based linkage map of human chromosome 1. **Genomics** 15:251-258, 1993.
100. Wilkie, P. J., Polymeropoulos, M. H., Trent, J. M., Small, K. W. and Weber, J. L. Genetic and physical map of short tandem repeat polymorphisms on human chromosome 6. **Genomics** 15:225-227, 1993.
101. Müller, U. Weber, J. L., Berry, P. and Kupke, K. G. Second polar body incorporation into a blastomere results in 46,XX/69,XXX mixoploidy. **J. Med. Genet.** 30:597-600, 1993.
102. Gispert, S., Twells, R., Orozco, G., Brice, A., Weber, J., Heredero, L., Scheufler, K., Riley, B., Allotey, R., Nothers, C., Hillermann, R., Lunkes, A., Khati, C., Stevanin, G., Hernandez, A., Margarino, C., Klockgether, T., Durr, A., Chneiweiss, H., Enczmann, J., Farrall, M., Beckmann, J., Mullan, M., Wernet, P., Agid, Y., Freund, H.-J., Williamson, R., Auburger, G., and Chamberlain, S. Chromosomal assignment of the second (Cuban) locus for autosomal dominant cerebellar ataxia (SCA2) to human chromosome 12q23-24.1. **Nature Genet.** 4:295-299, 1993.
103. Müller, U., Warman, M. L., Mulliken, J. B., and Weber, J. L. Assignment of a gene locus involved in craniosynostosis to chromosome 5qter. **Hum. Mol. Genet.** 2:119-122, 1993.
104. Peltomaki, P., Aaltonen, L. A., Sistonen, P., Pylkkanen, L., Mecklin, J., Jarvinen, H., Green, J. S., Jass, J. R., Weber, J. L., Leach, F. S., Peterson, G. M., Hamilton, S. R., de la Chapelle, A., and Vogelstein, B. Genetic mapping of a locus predisposing to human colorectal cancer. **Science** 260:810-812, 1993.

James L. Weber, Ph.D.

105. Dawson, E., Shaikh, S., Weber, J. L., Wang, Z., Weissenbach, J., Powell, J. F., and Gill, M. A continuous linkage map of 22 short tandem repeat polymorphisms on human chromosome 12. **Genomics** 17:245-248, 1993.
106. Olson, T. M., Michels, V. V., Lindor, N. M., Pastores, G. M., Weber, J. L., Schaid, D. J., Driscoll, D. J., Feldt, R. H., Thibodeau, S. N. Autosomal dominant supraaortic stenosis: Localization to chromosome 7. **Hum. Mol. Genet.** 2:869-873, 1993.
107. Litt, M., Kramer, P., Hauge, X. Y., Weber, J. L., Wang, Z., Wilkie, P. J., Holt, M. S., Mishra, S., Donis-Keller, H., Warnich, L., Retief, A. E., Jones, C., Weissenbach, J. A microsatellite-based index map of human chromosome 11. **Hum. Mol. Genet.** 2:909-913, 1993.
108. Weber, J. L. and Wong, C. Mutation in human short tandem repeat polymorphisms. **Hum. Mol. Genet.** 2:1123-1128, 1993.
109. Weber, J. L., Wang, Z., Hansen, K., Stevenson, M., Kappel, C., Salzman, S., Wilkie, P. J., Keats, B., Dracopoli, N. C., Brandriff, B. F., and Olsen, A. S. Evidence for human meiotic crossover interference obtained through construction of a short tandem repeat polymorphism linkage map of chromosome 19. **Am. J. Hum. Genet.** 53:1079-1095, 1993.
110. Nancarrow, D. J., Mann, G. J., Holland, E. A., Walker, G. J., Beaton, S. C., Walters, M. K., Luxford, C., Palmer, J. M., Donald, J. A., Weber, J. L., Fountain, J. W., Kefford, R. F., Hayward, N. K. Confirmation of chromosome 9p linkage in familial melanoma. **Am. J. Hum. Genet.** 53:936-942, 1993.
111. Gruis, N. A., Sandkuijl, L. A., Weber, J. L., van der Zee, A., Borgstein, A.-M., Bergman, W., and Frants, R. R. Linkage analysis in Dutch familial atypical multiple mole-melanoma (FAMMM) syndrome families. Effect of naevus count. **Melanoma Res.** 3:271-277, 1993.
112. Hildebrandt, F., Singh-Sawhney, I., Schnieders, B., Centofante, L, Omran, H., Pohlmann, A., Schmaltz, C., Wedekind, H., Schubotz, C. Antignac, C., Weber, J. L., Brandis, M., and Members of the APN Study Group. Mapping of a gene for familial juvenile nephronophthisis: refining the map and defining flanking markers on chromosome 2. **Am. J. Hum. Genet.** 53:1256-1261, 1993.
113. Briggs, M. D., Rasmussen, I. M., Weber, J. L., Yuen, J., Rimoin, D. L., and Cohn, D. H. Genetic linkage of mild pseudoachondroplasia (PSACH) to markers in the pericentromeric region of chromosome 19. **Genomics** 18:656-660, 1993.
114. Graeber, M. B., Müller, U., Monaco, A. P., and Weber, J. L. Four dinucleotide repeat polymorphisms close to the human collagen gene locus COL1A2 on chromosome 7q21. **Hum. Mol. Genet.** 2:2195, 1993.

James L. Weber, Ph.D.

115. Small, K. W., Weber, J., Roses, A., and Pericak-Vance, P. North Carolina macular dystrophy (MCDR1). A review and refined mapping to 6q14-q16.2. **Ophthalmic Paediatr. Genet.** 14:143-150, 1993.
116. Weber, J. L., Wang, Z., Wilkie, P. J., Bennesch, S., David, D. D., and Salzman, S. Linkage mapping using short tandem repeat polymorphisms. 1994. **In: Genetic Approaches to Mental Disorders**, Gershon, E. S. and Cloninger, C. R., (eds.), American Psychiatric Press, Washington, D.C., pp. 109-122.
117. Hughes, A. E., Shearman, A. M., Weber, J. L., Barr, R. J., Wallace, R. G. H., Osterberg, P. H., Nevin, N. C., and Mollan, R. A. B. Genetic linkage of familial expansile osteolysis to chromosome 18q. **Hum. Mol. Genet.** 3:359-361, 1994.
118. Wilkie, P. J. and Weber, J. L. Mapping of short tandem repeat polymorphisms on human chromosome 3. **Genomics** 19:167-169, 1994.
119. Attwood, J., Chiano, M., Collins, A., Donis-Keller, H., Dracopoli, N., Falk, C., Goudie, D., Gusella, J., Haines, J., Armour, J. A. L., Jeffreys, A., Kwiatkowski, D., Lathrop, M., Matisse, T., Moreno, F., Northrup, H., Pericak-Vance, M., Phillips, J., Retief, A., Robson, E., Shields, D., Slaugenhaupt, S., Vergnaud, G., Weber, J., Weissenbach, J., White, R., Yates, J., and Povey, S. CEPH consortium map of chromosome 9. **Genomics** 19:203-214, 1994.
120. Buetow, K. H., Weber, J. L., Ludwigsen, S., Scherpbier-Heddema, T., Duyk, G. M., Sheffield, V. C., and Murray, J. C. Integrated genome-wide maps constructed using the CEPH reference panel. **Nature Genet.** 6:391-393, 1994.
121. Schwengel, D. A., Jedlicka, A. E., Nanthakumara, E. J., Weber, J. L., and Levitt, R. C. Comparison of fluorescence-based semi-automated genotyping of multiple microsatellite loci with autoradiographic techniques. **Genomics** 22:46-54, 1994.
122. Perez Jurado, L. A., Phillips, J. A., III, Summar, M. L., Mao J., Weber, J. L., Schaefer, F. V., Hazan, J., Argente, J. Genetic mapping of the human growth hormone-releasing factor gene (GHRF) using two intragenic polymorphisms detected by PCR amplification. **Genomics** 20:132-134, 1994.
123. Wang, Z., Weber, J. L., Zhong, G., Tanksley, S. D. Survey of plant short tandem DNA repeats. **Theor. Appl. Genet.** 88:1-6, 1994.
124. Weber, J. L. Know thy genome. **Nature Genet.** 7:343-344, 1994.
125. Bonnycastle, L. L. C., Yu, C., Hunt, C. R., Trask, B. J., Clancy, K. P., Weber, J. L., Patterson, D., and Schellenberg, G. D. Cloning, sequencing and mapping of the human chromosome 14 heat shock protein gene (HSPA2). **Genomics** 23:85-93, 1994.

James L. Weber, Ph.D.

126. Oshima, J., Yu, C., Boehnke, M., Weber, J. L., Edelhoff, S., Wagner, M. J., Wells, D. E., Wood, S., Distech, C. M., Martin, G. M., and Schellenberg, G. D. Integrated mapping analysis of the Werner's Syndrome region of chromosome 8. **Genomics** 23:100-113, 1994.
127. Nakura, J., Wijsman, E. M., Miki, T., Kamino, K., Yu, C.-E., Oshima, J., Fukuchi, K.-I., Weber, J. L., Piussan, C., Melaragno, M. I., Epstein, C. J., Scappaticci, S., Fraccaro, M., Matsumura, T., Murano, S., Yoshida, S., Fujiwara, Y., Saida, T., Ogihara, T., Martin, G. M., Schellenberg, G. D. Homozygosity mapping of the Werner's syndrome locus. **Genomics** 23:600-608, 1994.
128. Murray, J. C., Buetow, K. H., Weber, J. L., Ludwigsen, S., Scherpbier-Heddema, T., Manion, F., Quillen, J., Sheffield, V. C., Sunden, S., Duyk, G. M., Weissenbach, J., Gyapay, G., Dib, C., Morrissette, J., Lathrop, G. M., Vignal, A., White, R., Matsunami, N., Gerken, S., Melis, R., Albertsen, H., Plaetke, R., Odelberg, S., Ward, D., Dausset, J., Cohen, D. and Cann, H. A comprehensive human linkage map with centimorgan density. **Science** 265:2049-2054, 1994.
129. Twells, R., Yenchitsomanus, P., Sirinavin, C., Allotey, R., Pongvarin, N., Viriyavejakul, A., Cemal, C., Weber, J., Farrall, M., Rodprasert, P., Prayoonwiwat, N., Williamson, R., and Chamberlain, S. Autosomal dominant cerebellar ataxia with dementia: evidence for a fourth disease locus. **Hum. Mol. Genet.** 3:177-180, 1994.
130. O'Connell, P., Leach, R. J., Rains, D., Taylor, T., Gargia, D., Ballard, L., Holik, P., Weissenbach, S., Sherman, P., Wilkie, P., Weber, J. L. and Naylor, S. L. A PCR-based genetic map for human chromosome 3. **Genomics** 24:557-567, 1994.
131. Walker, G. J., Nancarrow, D. J., Walters, M. K., Palmer, J. M., Weber, J. L., Hayward, N. K. Linkage analysis in familial melanoma kindreds to markers on chromosome 6p. **Int. J. Cancer** 59:771-775, 1994.
132. Friedman, T. B., Liang, Y., Weber, J. L., Hinnant, J. T., Barber, T. D., Winata, S., Arhya, I. N., and Asher, J. H. Jr. A gene for congenital, recessive deafness DFNB3 maps to the pericentromeric region of chromosome 17. **Nature Genet.** 9:86-91, 1995.
133. Morell, R., Liang, Y., Asher, J. H. Jr., Weber, J. L., Hinnant, J. T., Winata, S., Arhya, I. N., and Friedman, T. B. Analysis of short tandem repeat (STR) allele frequency distributions in a Balinese population. **Hum. Mol. Genet.** 4:85-91, 1995.
134. Leutelt, J., Oehlmann, R., Younus, F., van den Born, L.I., Weber, J.L., Denton, M.J., Mehdi, S.Q., and Gal A. Autosomal recessive retinitis pigmentosa locus maps on chromosome 1q in a large consanguineous family from Pakistan. **Clin. Genet.** 47:122-124, 1995.

James L. Weber, Ph.D.

135. Dubovsky, J., Sheffield, V. C., Duyk, G. M., and Weber, J. L. Sets of short tandem repeat polymorphisms for efficient linkage screening of the human genome. **Hum. Mol. Genet.** 4:449-452, 1995.
136. Dubovsky, J., Zabramsky, J. M., Kurth, J., Spetzler, R. F., Rich, S. S., Orr, H. T., Weber, J. L. A gene responsible for cavernous malformations of the brain maps to chromosome 7q. **Hum. Mol. Genet.** 4:453-458, 1995.
137. Fossdal, R., Magnússon, L., Weber, J. L., and Jensson, O. Mapping the locus of atrophica areata, a helicoid peripapillary chorioretinal degeneration with autosomal dominant inheritance, to chromosome 11p15. **Hum. Mol. Genet.** 4:479-483, 1995.
138. Berry, R., Stevens, T. J., Walter, N. A. R., Wilcox, A. S., Rubano, T., Hopkins, J. A., Weber, J., Goold, R., Soares, M. B., Sikela, J. M. Gene-based STSs as the basis for a human gene map. **Nature Genet.** 10:415-423, 1995.
139. Litt, M., Kramer, P., Kort, E., Fain, P., Cox, S., Root, D., White, R., Weissenbach, J., Donis-Keller, H., Gatti, R., Weber, J., Nakamura, Y., Julier, C., Hayashi, K., Spurr, N., Dean, M., Mandel, J., Kidd, K., Kruse, T., Retief, A., Bale, A., Meo, T., Vergnaud, G., Warren, S., and Willard, H. F. The CEPH consortium linkage map of human chromosome 11. **Genomics** 28:101-112, 1995.
140. Cox, D. W., Billingsley, G. D., Bale, A. E., Cooperative Human Linkage Center, Donis-Keller, H., Edwards, J. H., Litt, M., McBride, W., Persichetti, F., Spurr, N. K., Weber, J. L., Weissenbach, J., and White, R. L. CEPH Consortium Map of Chromosome 14. **Cytogenet. Cell Genet.** 69:175-178, 1995.
141. Christodoulou, K., Kyriakides, T., Hristova, A. H., Georgiou, D., Kalaydjieva, L., Yshpekova, B., Ivanova, T., Weber, J.L., and Middleton, L. Mapping of a distal form of spinal muscular atrophy with upper limb predominance to chromosome 7p. **Hum. Mol. Genet.** 4:1629-1632, 1995.
142. Lunkes, A., Hartung, U., Magarino, C., Rodriguez, M., Palmero, A., Rodriguez, L., Heredero, L., Weissenbach, J., Weber, J., and Auburger, G. Refinement of the OPA1 gene locus on chromosome 3q28-q29 to a region of 2 to 8 centimorgans in one Cuban pedigree with autosomal dominant optic atrophy type Kjer. **Am. J. Hum. Genet.** 57:968-970, 1995.
143. Levy-Lahad, E., Wijsman, E. M., Nemens, E., Anderson, L., Goddard, K. A. B., Weber, J. L., Bird, T. D., Schellenberg, G. D. A familial Alzheimer's disease locus on chromosome 1. **Science** 269:970-973, 1995.
144. Sheffield, V. C., Weber, J. L., Buetow, K. H., Murray, J. C., Even, D. A., Wiles, K., Gastier, J. M., Pulido, J. C., Yandava, C., Sunden, S. L., Mattes, G., Businga, T., McClain, A., Beck, J., Scherpier, T., Gilliam, J., Zhong, J., and Duyk, G. M. A collection

James L. Weber, Ph.D.

- of tri- and tetranucleotide repeat markers used to generate high quality, high resolution human genome-wide linkage maps. **Hum. Mol. Genet.** 4:1837-1844, 1995.
145. Arcot, S.S., Wang, Z., Weber, J.L., Deininger, P.L., and Batzer, M.A. Alu Repeats: A source for the genesis of microsatellites. **Genomics** 29:136-144, 1995.
 146. Gastier, J. M., Pulido, J. C., Brody, T., Sheffield, V. C., Weber, J. L., Buetow, K. H., Murray, J. C., Hudson, T. J., and Duyk, G. M. Survey of trinucleotide repeats in the human genome: assessment of their utility as genetic markers. **Hum. Mol. Genet.** 4:1829-1836, 1995.
 147. Johnson, E. W., Smith, L. M., Rich, S. S., Orr, H. T., Gil-Nagel, A., Kurth, J. H., Zabramski, J. M., Marchuk, D. A., Weissenbach, J., Clericuzio, C. L., Davis, L. E., Hart, B. L., Gusella, J. F., Kosofsky, B. E., Louis, D. N., Morrison, L. A., Green, E. D., and Weber, J. L. Refined localization of the cerebral cavernous malformation gene (CCM1) to a 4 cM interval of chromosome 7q contained in a well defined YAC contig. **Genome Res.** 5:368-380, 1995.
 148. Gil-Nagel, A., Dubovsky, J., Wilcox, K. J., Stewart, J. M., Anderson, V. E., Leppik, I. E., Orr, H. T., Johnson, E.W., Weber, J. L., and Rich, S. S. Familial cerebral cavernous angioma: a gene localized to a 15 cM interval on human chromosome 7q. **Ann. Neurol.**, 39:807-810, 1996.
 149. Blumenthal, M. N., Wang, Z., Weber, J. L., and Rich, S. S. Absence of linkage between 5q markers and serum IgE levels in four large atopic families. **Clin. Exp. Allergy**, 26:892-896, 1996.
 150. Rusch, T.L., Petsinger, J., Christensen, C., Vaske, D.A., Brumley, R. L., Luckey, J.A., and Weber, J.L. Scanning fluorescence detector for high-throughput DNA genotyping. Proceedings, Advances in DNA Monitoring Technologies, Biomedical Optics Society, **Int. Soc. Optical Eng.** (SPIE) 2680:316-325, 1996.
 151. Sunden, S.L.F., Businga, T., Beck, J., McClain, A., Gastier, J.M., Pulido, J.C., Yandava, C.N., Brody, T., Ghazizadeh, J., Weber, J.L., Duyk, G.M., Murray, J.C., Buetow, K.H., and Sheffield, V.C. Chromosomal assignment of 2900 tri- and tetranucleotide repeat markers using NIGMS somatic cell Hybrid Panel #2. **Genomics** 32:15-20, 1996.
 152. Gleeson, C. M., Sloan, J. M., McGuigan, J. A., Ritchie, A. J., Weber, J. L. and Russell, S. E. Widespread microsatellite instability occurs infrequently in adenocarcinoma of the gastric cardia. **Oncogene** 18:1653-1662, 1996.
 153. Gleeson, C. M., Sloan, J. M., McGuigan, J. A., Ritchie, A. J., Weber, J. L., Russell, S. E. Ubiquitous somatic alterations at microsatellite alleles occur infrequently in Barrett's-associated esophageal adenocarcinoma. **Cancer Res.** 56:259-263, 1996.

James L. Weber, Ph.D.

154. Mohrenweiser, H., Olsen, A., Archibald, A., Beattie, C., Burmeister, M., Lamerdin, J., Stewart, E., Stubbs, L., Weber, J.L., Johnson, K. Report of the Third International Workshop on Human Chromosome 19 Mapping 1996. **Cytogenet Cell Genet.**, 74:161-186, 1996.
155. Matthews, D., Fry, L., Powles, Weber, J., McCarthy, M., Fisher, E., Davies, K., Williamson, R. Evidence for a locus for familial psoriasis mapping to chromosome 4q. **Nature Genet.** 14:231-233, 1996.
156. Endo, K., Sasaki, H., Wakisaka, A., Tanaka, H., Saito, M., Igarashi, S., Takiyama, Y., Sanpei, K., Iwabuchi, K., Suzuki, Y., Onari, K., Suzuki, T., Weissenbach, J., Weber, J. L., Nomura, Y., Segawa, M., Nishizawa, M., Tsuji, S. Strong linkage disequilibrium and haplotype analysis in Japanese pedigrees with Machado-Joseph Disease. **Am. J. Med. Genet.** 67:437-444, 1996.
157. Yuan, B., Vaske, D., Weber, J. L., Beck, J., and Sheffield, V.C. Improved set of short tandem repeat polymorphisms for screening the human genome. **Am. J. Hum. Genet.** 60:459-460, 1997.
158. The Collaborative Study of the Genetics of Asthma (many authors) A genome-wide search for asthma susceptibility loci in ethnically diverse populations. **Nature Genet.** 15:389-392, 1997.
159. Weber, J.L. and Myers, E.M. Human whole genome shotgun sequencing. **Genome Res.** 7:401-409, 1997.
160. Gleeson, C. M., Sloan, J. M., McGuigan, J. A., Ritchie, A. J., Weber, J. L., Russell, S. E. Allelotype analysis of adenocarcinoma of the gastric cardia. **Br. J. Cancer** 76:1455-1465, 1997.
161. Johnson, E. W., Dubovsky J., Rich, S. S., Donovan, C. A., Orr, H. T., Anderson, V. E., Gil-Nagel, A., Ahmann, P., Dokken, C. G., Schneider, D. T., and Weber, J.L. Evidence for a novel gene for familial febrile convulsions, FEB2, linked to chromosome 19p in an extended family from the Midwest. **Hum. Molec. Genet.** 7:63-67, 1998.
162. Stone, D., Agarwala, R., Schäffer, A. A., Weber, J. L., Vaske, D., Oda, T., Chandrasekharappa, S. C., Francomano, C. A., and Biesecker, L. G. Genetic and physical mapping of the McKusick-Kaufman Syndrome. **Hum. Molec. Genet.** 7:475-481, 1998.
163. Finckh, U., Xu, S., Kumaramanickavel, G., Schurmann, M., Mukkadan, J. K., Fernandez, S. T., John, S., Weber, J. L., Denton, M. J., Gal, A. Homozygosity mapping of autosomal recessive retinitis pigmentosa locus (RP22) on chromosome 16p12.1-p12.3. **Genomics** 48:341-345, 1998.

James L. Weber, Ph.D.

164. Fenske, C. D., Jeffery, S., Weber, J. L., Houlston, R., Leonard, J. V., Lee, P. J. Localisation of the gene for glycogen storage disease type 1C by homozygosity mapping to 11q. **J. Med. Genet.** 35:269-272, 1998.
165. Dietrich, W. F., Weber, J. L., Kwok, P.-Y. and Nickerson, D. A. Isolation and analysis of DNA polymorphisms. In: **Genome Analysis: A Laboratory Manual** (Myers, R., et al., Eds.). Cold Spring Harbor Laboratory Press, Cold Spring Harbor, NY. 1998.
166. Blumenthal, M. N., Rich, S. S., King, R., Weber, J., Collaborative Study for the Genetics of Asthma (CSGA). Approaches and issues in defining asthma and associated phenotypes map to chromosome susceptibility areas in large Minnesota families. **Clin. Exp. Allergy**, 28 Suppl 1:51-55, 1998.
167. Weber, J. L. et al. Marshfield Center for Medical Genetics Web Site with data on human DNA polymorphisms, human genetic maps, instruments and lab protocols. **<http://www.marshmed.org/genetics>**.
168. Cho, J. H., Nicolae, D., Gold, L., Fields, C., Labuda, M., Rohal, P., Pickles, M., Qin, L., Fu, Y., Mann, J., Kirschner, B. S., Jabs, E. W., Weber, J., Hanauer, S. B., Bayless, T. M., Brant, S. R. Identification of novel susceptibility loci for inflammatory bowel disease on chromosomes 1p, 3q and 4q. **Proc. Natl. Acad. Sci.** 95:7502-7507, 1998.
169. Hizawa, N., Freidhoff, L. R., Chieu, Y.-F., Ehrlich, E., Luehr, C. A., Anderson, J. L., Duffy, D. L., Dunston, G. M., Weber, J. L., Huang, S.-K., Barnes, K. C., Marsh, D. G., Beaty, T. H. Genetic regulation of *Dermatophagoides pteronyssinus*-specific IgE responsiveness: A genome-wide multipoint linkage analysis in families recruited through 2 asthmatic sibs. **J. Allergy Clin. Immunol.** 102:436-442, 1998.
170. Hizawa, N., Collins, G., Rafnar, T., Huang, S.-K., Duffy, D. L., Weber, J. L., Freidhoff, L. R., Ehrlich, E., Marsh, D. G., Beaty, T. H., Barnes, K. C., Collaborative Study on the Genetics of Asthma. Linkage analysis of *Dermatophagoides pteronyssinus*-specific IgE responsiveness with polymorphic markers on chromosome 6p21 (HLA-D region) in Caucasian families by the transmission/disequilibrium test. **J. Allergy Clin. Immunol.** 102:443-448, 1998.
171. Broman, K. W., Murray, J. C., Sheffield, V. C., White, R. L. and Weber, J. L. Comprehensive human genetic maps: individual and sex-specific variation in recombination. **Am. J. Hum. Genet.** 63:861-869, 1998.
172. Simonik, I., Gericke, G. S., Ott, J., Weber, J.L. Identification of genetic markers associated with Gilles de la Tourette Syndrome in an Afrikaner population. **Am. J. Hum. Genet.** 63:839-846, 1998.
173. Broman, K.W. and Weber, J.L. Estimation of pairwise relationships in the presence of genotyping errors. **Am. J. Hum. Genet.** 63:1563-1564, 1998.

James L. Weber, Ph.D.

174. Gleeson, C.M., Sloan, J.M., McGuigan, J.A., Ritchie, A.J., Weber, J.L., Russell, S.E. Barrett's oesophagus: microsatellite analysis provides evidence to support the proposed metaplasia-dysplasia-carcinoma sequence. **Genes Chrom. Cancer** 21:49-60, 1998.
175. Broman, K.W. and Weber, J.L. Method for constructing confidently ordered linkage maps. **Genet. Epidemiol.** 16:337-343, 1999.
176. Tourette Syndrome Genetics Consortium (many authors). A complete genome screen in sib-pairs affected with the Gilles de la Tourette Syndrome. **Am. J. Hum. Genet.** 65:1428-1436, 1999.
177. Broman, K.W. and Weber, J.L. Long homozygous chromosomal segments in the CEPH families. **Am. J. Hum. Genet.** 65:1493-1500, 1999.
178. Rosenberg, M.J., Vaske, D., Killoran, C.E., Ning, Y., Wargowski, D., Hudgins, L., Tiffit, C.J., Meck, J., Blancato, J.K., Rosenbaum, K., Pauli, R.M., Weber, J., and Biesecker, L.G. Detection of chromosomal aberrations by a whole genome microsatellite screen. **Am. J. Hum. Genet.** 66:419-427, 2000.
179. Suarez, B.K., Lin, J., Burmester, J.K., Broman, K.W., Weber, J.L., Banerjee, T.K., Goddard, K.A.B., Witte, J.S., Elston, R.C., Catalona, W.J. A genome screen of multiplex prostate cancer sibships. **Am J. Hum. Genet.** 66:933-944, 2000.
180. Yuan, B., Neuman, R., Duan, S.H., Weber, J.L., Kwok, P.-Y., Saccone, N.L., Wu, J.S., Liu, K.-Y., Schonfeld, G. Linkage of a gene for familial hypobetalipoproteinemia to chromosome 3p21.1-22. **Am J. Hum. Genet.** 66:1699-1704, 2000.
181. Ehm, M.G., Karnoub, M.C., Sakul, H., Gottschalk, K., Holt, D.C., Weber, J.L., Vaske, D., Briley, D., Briley, L., Kopf, J., McMillen, P., Nguyen, Q., Reisman, M., Lai, E.H., Joslyn, G., Shepard, N.S., Bell, C., Wagner, M.J., and Burns, D.K. Genome-wide search for type 2 diabetes susceptibility genes in four American populations. **Am J. Hum. Genet.** 66:1871-1881, 2000.
182. Broman, K. W. and Weber, J.L. Characterization of human crossover interference. **Am J. Hum. Genet.** 66:1911-1926, 2000.
183. Reinartz, G.E., Karron, J.D., Phillips, R.B., and Weber, J.L. Patterns in microsatellite polymorphism in the range-restricted Bonobo (*Pan paniscus*). **Molec. Ecology** 9:315-328, 2000.
184. Witte, J.S., Goddard, K.A.B., Conti, D.V., Elston, R.C., Lin, J., Suarez, B.K., Broman, K.W., Burmester, J.K., Weber, J.L., Catalona, W.J. Genomewide scan for prostate cancer - aggressiveness loci. **Am J. Hum. Genet.** 67:92-99, 2000.

James L. Weber, Ph.D.

185. Matsumoto, N., David, D.E., Johnson, E.W., Konecki, D., Burmester, J., Ledbetter, D.H., and Weber, J.L. Breakpoint sequences of an 1;8 translocation in a family with Gilles de la Tourette Syndrome. **Eur. J. Hum. Genet.** 8:875-883, 2000.
186. Kissebah, A.H., Sonnenberg, G.E., Myklebust, J., Goldstein, M., Broman, K., James, R.G., Marks, J.A., Krakower, G.R., Jacob, H.J., Weber, J.L., Martin, L., Blangero, J., Comuzzie, A.G. Quantitative trait loci on chromosomes 3 and 17 influence phenotypes of the metabolic syndrome. **Proc. Natl. Acad. Sci.** 97:14478-14483, 2000.
187. Vitale, E., Brancolini, V., de Rienzo, A., Bird, L., Allada, V., Sklansky, M., Chae, C. U., Ferrero, G.B., Weber, J., Devoto, M., Casey, B. Suggestive linkage of *situs inversus* and other left-right axis anomalies to chromosome 6p. **J. Med. Genet.** 38:182-185, 2001.
188. Weber, J.L. and Broman, K.W. Genotyping for human whole genome scans: past, present and future. In **Advances in Genetics**, v. 42, D.C. Rao and M.A. Province, eds. Academic Press, pp. 77-96, 2001.
189. Simonic, I., Gericke, G.S., Nyholt, D.R., Matsumoto, N., Ledbetter, D.H., Gordon, D., Ott, J., Weber, J.L. Confirmation of Gilles de la Tourette Syndrome (GTS) susceptibility loci on chromosomes 2p11, 8q22 and 11q23-24 in South African Afrikaners. **Am. J. Med. Genet. – Neuropsychiatric Genet.** 105:163-167, 2001.
190. Yu, A., Zhao, C., Fan, Y., Jang, W., Mungall, A., Deloukas, P., Olsen, A., Doggett, N., Ghebranious, N., Broman, K.W. and Weber, J.L. Comparison of human genetic and sequence-based physical maps. **Nature** 409:951-953, 2001.
191. Giglio, S., Broman, K.W., Matsumoto, N., Calvari, V., Gimelli, G., Neumann, T., Ohashi, H., Voullaire, L., Larizza, D., Giorda, R., Weber, J.L., Ledbetter, D.H., Zuffardi, O. Olfactory receptor (OR) gene clusters, genomic inversion polymorphisms and common chromosome rearrangements. **Am J. Hum. Genet.** 68:874-883, 2001.
192. Collins-Schramm, H.E., Phillips, C.M., Operario, D.J., Lee, J.S., Weber, J.L., Hanson, R.L., Knowler, W.C., Cooper, R., Li, H., Seldin, M.F. Ethnic difference markers for use in mapping by admixture linkage disequilibrium. **Am J. Hum. Genet.** 70:737-750, 2002.
193. Cann H. M. et al. (many authors). A human genome diversity cell line panel (letter to editor). **Science** 296:261-262, 2002.
194. Weber, J.L. The Iceland Map. News and Views. **Nature Genet.** 31:225-226, 2002.
195. Rosenberg, M.J., Agarwala, R., Bouffard, G., Davis, J., Fiermonte, G., Hilliard, M.S., Koch, T., Kalikin, L.M., Makalowska, I., Morton, D.H., Petty, E.M., Weber, J.L., Palmieri, F., Kelley, R.I., Schäffer, A.A., Biasecker, L.G. Mutant deoxynucleotide carrier is associated with congenital microcephaly. **Nature Genet.** 32:175-179, 2002.

James L. Weber, Ph.D.

196. Weber, J.L., David, D., Heil, J., Fan, Y., Zhao, C., and Marth, G. Human diallelic insertion/deletion polymorphisms. **Am J. Hum. Genet.** 71:854-862, 2002.
197. Collins-Schramm, H.E., Kittles, R.A., Operario, D.J., Weber, J.L., Criswell, L.A., Cooper, R.S., Seldin, M.F. Markers that discriminate between European and African ancestry show limited variation within Africa. **Human Genet.** 111:566-569, 2002.
198. Rosenberg, N.A., Pritchard, J.K., Weber, J.L., Cann, H.M., Kidd, K.K., Zhivotovsky, L.A., Feldman, M.W. Genetic structure of human populations. **Science** 298:2381-2385, 2002.
199. Ruf, R., Rensing, C., Topaloglu, R., Guay-Woodford, L., Klein, C., Vollmer, M., Otto, E., Beekmann, F., Haller, M., Wiedensohler, A., Leumann, E., Antignac, C., Rizzoni, G., Filler, G., Brandis, M., Weber, J. L., Hildebrandt, F. Confirmation of the ATP6B1 gene as responsible for distal renal tubular acidosis. **Pediatric Nephrology** 218:105-109, 2003.
200. Broman, K.W., Matsumoto, N., Giglio, S., Martin, C.L., Roseberry, J.A., Zuffardi, O., Ledbetter, D.H. and Weber, J.L. Common long human inversion polymorphism on chromosome 8p. In: Goldstein, D.R. (ed). Science and Statistics: A Festschrift for Terry Speed. **IMS Lecture Notes-Monograph Series.** 40:237-245, 2003.
201. Ghebranious, N., Vaske, D., Yu, A., Zhao, C., Marth, G., and Weber, J.L. STRP Screening Sets for the human genome at 5 cM density. **BMC Genomics** 4:6, 2003.
202. Niemann, S., Zhao, C., Pascu, F., Stahl, U., Aulepp, U., Niswander, L., Weber, J.L., and Müller, U. Homozygous WNT3 mutation causes tetra-amelia in a large consanguineous family. **Am J. Hum. Genet.** 74:558-563, 2004
203. Giampietro, P.F., Raggio, C.L., Reynolds, C.E., Shukla, S.K., McPherson, E., Ghebranious, N., Jacobsen, F.S., Kumar, V., Faciszewski, T., Pauli, R.M., Rasmussen, K., Burmester, J.K., Zaleski, C., Merchant, S., David, D., Weber, J.L., Glurich, I. and Blank, R.D. An analysis of *PAX1* in the development of vertebral malformations. **Clin Genet.** 68:448-453, 2005.
204. Carothers, A.D., Rudan, I., Kolcic, I., Polasek, O., Hayward, C., Wright, A.F., Campbell, H., Teague, P., Hastie, N.D. and Weber J.L. Estimating human inbreeding coefficients: comparison of genealogical and marker heterozygosity approaches. **Ann Hum Genet.** 70:666-676, 2006.
205. Xiao, P., Liu, P., Weber, J.L., Papasian, C.J., Recker, R.R., and Deng, H.-W. Paternal uniparental isodisomy of the entire chromosome 3 revealed in a person with no apparent phenotypic disorders. **Hum Mutat.** 27:133-137, 2006.

James L. Weber, Ph.D.

206. Su Z, Li Y, James JC, McDuffie M, Matsumoto AH, Helm GA, Weber JL, Lusis AJ, Shi W. Quantitative trait locus analysis of atherosclerosis in an intercross between C57BL/6 and C3H mice carrying the mutant apolipoprotein E gene. **Genetics** 172:1799-1807, 2006.
207. Chudyk JP, Rusch TL, Fieweger KM, Dobrin SE, Weber JL. Automating microsatellite genotyping with array tape. **J Assoc Lab Automation** 11:260-267, 2006.
208. Weber, J.L. Clinical applications of genome polymorphism scans. **Biol Direct** 1:16, 2006.
209. Doggett, N.A., Xie, G., Meincke, L.J., Sutherland, R.D., Mundt, M.O., Berbari, N.S., Davy, B.E., Robinson, M.L., Weber, J.L., Stallings, R.L., and Han, C. A 360 Kb interchromosomal duplication of the human HUDIN locus. **Genomics** 88:762-771, 2006.
210. Rosenberg, N.A., Mahajan, S., Gonzales-Quevedo, C., Nino-Rosales, L., Ninis, V., Das, P., Hegde, M., Molinari, L., Zapata, G., Weber, J.L., Belmont, J.W., and Patel, P.I. Low levels of genetic divergence across geographically and linguistically diverse populations from India. **PLOS Genetics** 2:e215 2006.
211. Campbell, H., Carothers, A.D., Rudan, I., Hayward, C., Biloglav, Z., Barac, L., Pericic, M., Janicijevic, B., Smolej-Narancic, N., Polasek, O., Kolcic, I., Weber, J.L., Hastie, N.D., Rundan, P., and Wright, A.F. Effects of genome-wide heterozygosity on a range of biomedically relevant human quantitative traits. **Hum. Mol. Genet.** 16:233-241, 2007.
212. Ocaña, L., Zhao, C., Reed, J.A., Ebenezer, N.D., Brice, G., Morley, T., Mehta, M., O'Dowd, J., Weber, J.L., Hardcastle, A.J., Child, A.H. Assignment of two loci for autosomal dominant Adolescent Idiopathic Scoliosis (AIS) to chromosomes 9q31.2-q34.2 and 17q25.3-qtel. **J. Med. Genet.** 45:87-92, 2007.
213. Weber, J. (2007), "Human Microsatellite and Minisatellite DNA Polymorphisms", in Cavalli-Sforza, L. and Feldman, M. (eds), *Human Population Genetics: Evolution and Variation*, The Biomedical & Life Sciences Collection, Henry Stewart Talks Ltd, London (online at <http://www.hstalks.com/?t=BL0251558-Weber>).
214. Betanzos-Cabrera, G., Harker, B.W., Doktycz, M.J., Weber, J.L., and Beattie, K.L. A comparison of hybridization efficiency between flat glass and channel glass solid supports. **Mol. Biotechnol.** 38:71-80, 2008.
215. Betanzos-Cabrera, G., Harker, B.W., Doktycz, M.J., Weber, J.L., and Beattie, K.L. Channel glass-based detection of human short insertion/deletion polymorphisms by tandem hybridization. **Mol. Biotechnol.** 38:145-153, 2008.

James L. Weber, Ph.D.

216. Friedlaender, J.S., Friedlaender, F.R., Reed, F.R., Kidd, K.K., Kidd, J.R., Chambers, G., Lea, R., Loo, J.-H., Koki, G., Hodgson, J.A., Merriwether, D.A., and Weber, J.L. The Genetic Structure of Pacific Islanders. **PLOS Genetics** 4:e19, 2008.
217. Rudan, I., Carothers, A.D., Polasek, O., Hayward, C., Vitart, V., Biloglav, Z., Kolcic, I., Zgaga, L., Ivankovic, D., Vorko-Jovic, A., Wilson, J.F., Weber, J.L., Hastie, N., Wright, A., and Campbell, H. Quantifying the increase in average human heterozygosity due to urbanization. **Eur. J. Hum. Genet.** 16:1097-1102, 2008.
218. Payseur, B.A., Place, M., and Weber, J.L. Linkage disequilibrium between STRPs and SNPs across the human genome. **Am. J. Hum. Genet.** 82:1039-1050, 2008.
219. Sammalisto, S., Hiekkalinna, T., Schwander, K., Kardia, S., Weder, A.B., Rodriguez, B.L., Doria, A., Kelly, J.A., Bruner, G.R., Harley, J.B., Redline, S., Larkin, E.K., Patel, S.R., Ewan, A.J., Weber, J.L., Perola, M., and Peltonen, L. Genome-wide linkage screen for stature and body-mass index in 3,032 families: evidence for sex- and population-specific genetic effects. **Eur. J. Hum. Genet.** 17:258-266, 2008.
220. Raggio, C.L., Giampietro, P.F., Dobrin, S., Zhao, C., Dorshorst, D., Ghebranious, N., Weber, J.L. and Blank, R.D. A novel locus for adolescent idiopathic scoliosis on chromosome 12p. **J. Orthopaed. Res.** 27:1366-1372, 2009.
221. Tishkoff, S.A., Reed, F.A., Friedlaender, F.R., Froment, A., Ehret, C., Dobrin, S., Doumbo, O., Hirbo, J.B., Ibrahim, M., Juma, A.T., Kotze, M.J., Lema, G., Moore, J.H., Nyambo, T.B., Omar, S.A., Pretorius, G.S., Ranciaro, A., Smith, M.W., Thera, M., Wambebe, C., Weber, J.L., Williams, S.M. The Genetic Structure and History of Africans and African Americans. **Science** 324:1035-1044, 2009.
222. Gallione, C.J., Solatycki, A., Awad, I.A., Weber, J.L., Marchuk, D.A. A Founder Mutation in the Ashkenazi Jewish Population Affecting mRNA Splicing of the CCM2 Gene Causes Cerebral Cavernous Malformations. **Genetics in Medicine** 13:662-666, 2011.
223. Wu, Y., Weber, J.L., Vladutiu, G.D., Tarnopolsky, M.A. Six novel mutations in the myophosphorylase gene in patients with McArdle's disease and a family with pseudo-dominant inheritance pattern. **Molec. Genet. Metab.** 104:587-591, 2011.
224. Sarangi, S.N., Golightly, M., Weber, J., Chan, E.L. A Family with Bolzano Type Bernard-Soulier Syndrome Carrier a Benign A1939T MYH9 Mutation. **Platelets** 24:81-84, 2013.
225. Fisher, O.S., Liu, W., Zhang, R., Stiegler, A.L., Ghedia, S., Weber J.L., Boggon, T.J. Structural Basis for the Disruption of the Cerebral Cavernous Malformations 2 (CCM2) Interaction with Krev Interaction Trapped 1 (KRIT1) by Disease-associated Mutations. **J. Biol. Chem.** 290:2842-2853, 2015.

James L. Weber, Ph.D.

226. Biesecker, L.G., Dirksen, R.T., Girard, T., Hopkins, P.M., Riazi, S., Rosenberg, H., Stowell, K., Weber, J. Genomic Screening for Malignant Hyperthermia Susceptibility. **Anesthesiology** 133:1277-1282, 2020.
227. Johnston, J.J., Dirksen, R.T., Girard, T., Gonsalves, S.G., Hopkins, P.M., Riazi, S., Saddic, L.A., Sambuughin, N., Saxena, R., Stowell, K., Weber, J., Rosenberg, H., and Biesecker, L.G. Variant Curation Expert Panel Recommendations for RYR1 Pathogenicity Classifications in Malignant Hyperthermia Susceptibility. **Genetics in Medicine** 23:1288-1295, 2021.
228. Chanes, N., Tumolo, J., Weber, J. L., Sampath, S., Gómez, R., Vargas, A., Gedalia, A., León, A., Bernal, M.D., Sernich, S., Sanderson, T.M., Bronfin, D.R., Zambrano, R.M., Keshavamurthy, C.B., Woolridge, K.A., Urrea, S., and Lacassie, Y. Mild Ocular Findings and Skeletal Anomalies Producing a Marfanoid Appearance, Along with Autoimmune/Inflammatory Findings Due to SNV of Unknown Significance in Genes ABCC6 and OPA1 Inherited from the Mother. **World Journal of Ophthalmology & Vision Research** DOI: 10.33552/WJOVR.2022.04.000578, 2022.
229. Johnston, J.J., Dirksen, R.T., Girard, T., Hopkins, P.M., Kraeve, N., Oguno, M., Radenbaugh, K.B., Riazi, S., Robinson, R.L., Saddic, L.A., Sambuughin, N., Saxena, R., Shepherd, S., Stowell, K., Weber, J., Yoo, S., Rosenberg, H., and Biesecker, L.G. Updated Variant Curation Expert Panel Criteria and Pathogenicity Classifications for 251 Variants for RYR1-related Malignant Hyperthermia Susceptibility. **Human Molecular Genetics** 31:4087-4093, 2022.
230. Pantrangi, M., Rath, J., Kaetterhenry, N., Branham, K., Talsness, D., and Weber J.L. Clinical Sequencing of the Retinitis Pigmentosa Gene *RPGR* in Over 1,000 Cases of Vision Loss. **Molecular Vision** 30:49-57, 2024.
231. Weber, J.L. Healthcare Records Need to Be Preserved. Ten Generations Press. 2026.
232. Weber, J.L. Genomes and the Gene Pool. Ten Generations Press. 2026.
233. Weber, J.L. Suggestions for Long Term Goals for Humanity. Ten Generations Press. 2026.

James L. Weber, Ph.D.

FIFTEEN MOST SIGNIFICANT PUBLICATIONS
(Listed subjectively from highest to lowest impact)

1. Weber, J.L. Genomes and the Gene Pool. Ten Generations Press. 2026.
2. Weber, J.L. Suggestions for Long Term Goals for Humanity. Ten Generations Press. 2026.
3. Weber, J. L. and May, P. E. Abundant class of human DNA polymorphisms which can be typed using the polymerase chain reaction. **Am. J. Hum. Genet.** 44:388-396, 1989.
4. Wijmenga, C., Frants, R. R., Brouwer, O. F., Moerer, P., Weber, J. L., and Padberg, G. W. Location of the fascioscapulohumeral muscular dystrophy gene on chromosome 4. **Lancet** 336:651-653, 1990.
5. Broman, K. W., Murray, J. C., Sheffield, V. C., White, R. L. and Weber, J. L. Comprehensive human genetic maps: individual and sex-specific variation in recombination. **Am. J. Hum. Genet.**, 63:861-869, 1998.
6. Rosenberg, N.A., Pritchard, J.K., Weber, J.L., Cann, H.M., Kidd, K.K., Zhivotovsky, L.A., Feldman, M.W. Genetic structure of human populations. **Science** 298:2381-2385, 2002.
7. Weber, J.L. and Myers, E.M. Human whole genome shotgun sequencing. **Genome Res.** 7:401-409, 1997.
8. Egan, J. E., Weber, J. L., Ballou, W. R., Majarian, W. R., Gordon, D. M., Hoffman, S. L., Wirtz, R. A., Schneider, I., Woollett, G. R., Hollingdale, M. R., Young, J. F., and Hockmeyer, W. T. Efficacy of murine malaria sporozoite vaccines: Implications for human vaccine development. **Science** 236:453-456, 1987.
9. Weber, J. L. and Wong, C. Mutation of human short tandem repeats. **Hum. Mol. Genet.** 2:1123-1128, 1993.
10. Broman, K.W. and Weber, J.L. Long homozygous chromosomal segments in reference families from the Centre d'Etude du polymorphisme humain. **Am. J. Hum. Genet.** 65:1493-1500, 1999.
11. Weber, J.L. Healthcare Records Need to Be Preserved. Submitted for publication. 2026.
12. Broman, K. W. and Weber, J.L. Characterization of human crossover interference. **Am J. Hum. Genet.** 66:1911-1926, 2000.
13. Weber, J.L., David, D., Heil, J., Fan, Y., Zhao, C., and Marth, G. Human diallelic insertion/deletion polymorphisms. **Am J. Hum. Genet.** 71:854-862, 2002.

James L. Weber, Ph.D.

14. Sheffield, V. C., Weber, J. L., Buetow, K. H., Murray, J. C., Even, D. A., Wiles, K., Gastier, J. M., Pulido, J. C., Yandava, C., Sunden, S. L., Mattes, G., Businga, T., McClain, A., Beck, J., Scherpier, T., Gilliam, J., Zhong, J., and Duyk, G. M. A collection of tri- and tetranucleotide repeat markers used to generate high quality, high resolution human genome-wide linkage maps. **Hum. Mol. Genet.** 4:1837-1844, 1995.
15. Weber, J. L. Informativeness of human (dC-dA)_n·(dG-dT)_n polymorphisms. **Genomics** 7:524-530, 1990.