CURRICULUM VITAE

Huntington Faxon Willard, Ph.D.

Current Appointment: Director, Geisinger National Precision Health
Associate Chief Scientific Officer
Professor of Precision Health
Geisinger
North Bethesda, MD | Danville, PA

Education	**Institution**	**Date**	**Degree**
Baccalaureate	Harvard University	1975	A.B.
Doctorate	Yale University	1979	Ph.D.

Scholarly Societies:

National Academy of Medicine (*elected,* 2016)
National Academy of Sciences (*elected*, 2013)
American Academy of Arts & Sciences (*elected*, 2008)
American Association for the Advancement of Science (*elected*, 2005)
Genetics Society of America
American Society of Human Genetics

Academic Career:

Institution	**Position/Title**	**Dates**
Geisinger	Associate Chief Scientific Officer	2018 – present
Geisinger National Precision Health	Director	
Marine Biological Laboratory	President and Director	2015 – 2017
University of Chicago		
Department of Human Genetics	Professor	
Committee on Evolutionary Biology		
Duke University		
Department of Biology	Arts & Sciences Professor	2014
Institute for Genome Sciences & Policy	Institute Director	2003 – 2014
Department of Molecular Genetics & Microbiology	Nanaline H. Duke Professor	2004 – 2014
Case Western Reserve University		
Department of Genetics	Henry Willson Payne Professor and Chairman	1992 - 2001
	Professor	2001 - 2002

Center for Human Genetics	Director	1992 - 2002
University Hospitals of Cleveland		
Research Institute	Director and President	1999 - 2002
Stanford University		
Department of Genetics	Associate Professor	1989 - 1992
University of Toronto		
Department of Molecular Genetics	Assistant Professor	1982 - 1987
Faculty of Medicine | Arts & Science	Associate Professor	1987 - 1989
Johns Hopkins University School of Medicine		
Division of Medical Genetics	Fellow	1979 - 1981

Administrative, Executive and Professional Experience:

Academic Leadership and Executive Positions

2018 – present	Director, Geisinger National Precision Health Associate Chief Scientific Officer
2015 – 2017	President and Director, Marine Biological Laboratory
2003 – 2014	Director, Institute for Genome Sciences & Policy, Duke University
2010 – 2012	Senior Advisor, Office of the Vice Provost and Dean for Undergraduate Education
2009 – 2012	Member, Appointments, Promotion and Tenure Committee, Duke University
2009 – 2010	Member, Undergraduate Education Strategic Planning Committee, Duke University
2003 – 2009	Vice Chancellor for Genome Sciences, Duke University Health System and Duke University
1999 – 2002	President and Director, Research Institute, University Hospitals of Cleveland
1992 – 2002	Director, Center for Human Genetics, University Hospitals of Cleveland
1992 – 2001	Chairman, Department of Genetics, Case Western Reserve University

Educational Program Leadership, Awards and Honors:

2015	Recipient (joint), Education Prize, American Society of Human Genetics
2008 – 2014	Director, Undergraduate Program in Genome Sciences & Policy, Duke University
2009, 2010, 2011	Arts & Sciences Faculty "Dean's List" (top 5% of student evaluations), Duke University
2006 – present	Appointed HHMI Professor, Howard Hughes Medical Institute
2005 – 2008	Director, Graduate Program in Computational Biology & Bioinformatics, Duke University
2001	Recipient, Outstanding Faculty Award, Biomedical Sciences Doctoral Training Programs, Case Western Reserve University
1992 – 2001	Director, Graduate Program in Genetics, Case Western Reserve University

Professional Awards, Named Lectures and Honors:

2016	Elected, Member, National Academy of Medicine
2013	Elected, Member, National Academy of Sciences
2013	James V. Neel Memorial Lecture, University of Michigan
2011	Wilbur Cross Medal, Yale Graduate School
2009	William Allan Award, American Society of Human Genetics
2008	Elected, Fellow, American Academy of Arts and Sciences
2007	Elected, Society of Scholars, Johns Hopkins University
2006	Catherine Birch McCormick Distinguished Lecture, George Washington University
2006	David L. Rimoin Lecture, UCLA
2005	Elected, Fellow, American Association for the Advancement of Science
2004	Abelson Family Lecture, Washington State University, Pullman, WA
2001	President, American Society of Human Genetics
1999	Pruzansky Lecture, American College of Medical Genetics
1998 – 2003	Franklin Delano Roosevelt Investigator, March of Dimes Birth Defects Foundation
1988 – 1993	Scientist Award, Medical Research Council of Canada (resigned, 1989)
1983 – 1988	Scholar Award, Medical Research Council of Canada
1982 – 1984	Basil O'Connor Award, March of Dimes Birth Defects Foundation

National Institutes of Health | Department of Health and Human Services

2003 – 2007	Secretary's Advisory Committee on Genetics, Health & Society, Department of Health and Human Services
1999 – 2002	Mammalian Genetics Study Section, National Institutes of Health (Chair, 2000-2002)
1987 – 1991	NICHD Mental Retardation Research Committee, National Institutes of Health (Chair, 1990-1991)

American Society of Human Genetics | Genetics Society of America

2016 – 2018	Board of Directors, Genetics Society of America
2005 – 2006	Nominating Committee, American Society of Human Genetics (Chair, 2006)
2000 – 2003	Board of Directors, American Society of Human Genetics
2001	President, American Society of Human Genetics
1997 – 2001	Awards Committee, American Society of Human Genetics
1994 – 1996	Board of Directors, American Society of Human Genetics
1990 – 1994	Program Committee, American Society of Human Genetics (Chair, 1994)
1986 – 1987	Nominating Committee, American Society of Human Genetics (Chair, 1987)

Other Scientific Review and Advisory Committees

1997 – present	Scientific Review Board, Howard Hughes Medical Institute
2011 – present	Scientific Advisory Board, Simons Foundation Autism Research Initiative
2011 – 2017	Chair, Scientific Advisory Board, Geisinger Health System
2012	Academic Program Review Committee, Graduate Programs in Biomedical and Biological Sciences, University of Southern California

2012	External Review Committee, Interdisciplinary Graduate Program in Molecular Biosciences, Washington State University
2009 – 2011	Scientific Review Panel, Simons Foundation
2007	External Review Committee, Institute for Systems Medicine Planning Authority, Spokane, WA
2006	External Review Committee, Department of Biology, Boston College
2004 – 2010	Chair, Basil O'Connor Advisory Committee, March of Dimes Birth Defects Foundation
2004, 2006	Review Panel (Chair, 2004), Genome Canada
2002	External Review, Graduate Programs in Human Genetics, University of California, Los Angeles
2001	Chair, Human Genetics External Review Committee, University of California, San Francisco
1992 – 1998	Basil O'Connor Advisory Committee, March of Dimes Birth Defects Foundation

Other National and International Organizations

2018 – present	Member, Roundtable on Genomics & Precision Medicine, National Academies of Science, Engineering, and Medicine
2017	Scientific Executive Committee, GP-write Initiative, Center of Excellence for Engineering Biology, NYU
2002 – 2005	Member, Excellence in Science Award Committee, Federation of American Societies for Experimental Biology (FASEB)
2000 – 2001	Member, Committee on Understanding the Biology of Sex and Gender Differences, Institute of Medicine
1997 – 2000	Member, Council of Academic Societies, American Association of Medical Colleges (AAMC)
1996 - 1999	Member, Board of Councilors, Association of Professors of Human/Medical Genetics
1992 - 1995	Co-editor, Genetic Constitution of the Human X Chromosome, Human Gene Mapping Workshops
1993 - 1995	Human Genome Mapping Committee, Human Genome Organization (HUGO)
1988 - 1991	Committee on Genetic Constitution of the Human X Chromosome, International Workshop on Human Gene Mapping
1982 - 1989	Committee on Human Gene Mapping Using Recombinant DNA Techniques, International Workshop on Human Gene Mapping

Academic Publications

2016 – present	Series Editor (with G. Ginsburg), *Genomic and Precision Medicine*
2011 – 2015	Editorial Committee, *Annual Review of Genomics and Human Genetics*
2005 – present	Consulting Editor, *Human Molecular Genetics*
2005 – 2010	Editorial Board, *BMC Genomics*
1991 – 2005	Co-Founder and Executive Editor, *Human Molecular Genetics*
1990 – 1999	Editor, *Chromosoma*
1987 - 1991	Editorial Board and Subject Area Editor, *Genomics*
1989 - 1991	Associate Editor, *The American Journal of Human Genetics*
1984 - 1989	Editorial Board, *Cytogenetics and Cell Genetics*

Other Organizations (Academic, Corporate and Community):

2011 – 2017	Chair, Scientific Advisory Board, Geisinger Health System, Danville, PA
2014 – 2015	Consultant, Third Rock Ventures, Boston, MA
2011 – 2014	Board of Directors, AptamiR Therapeutics, Inc.
2009	Co-Founder, Athleticode, Inc., San Francisco, CA
2007 – 2008	Chair, Strategic Advisory Board, Institute for Systems Medicine, Spokane, WA
2004 – 2009	Scientific Advisory Board, LabCorp Inc, Research Triangle Park, NC
2004 – 2008	Advisory Board, McLaughlin Centre for Molecular Medicine, University of Toronto
2004 – 2005	Board of Directors, N. Carolina Biotechnology Center, Research Triangle Park, NC
2002 – 2005	Advisory Board, Genetics Prize, Peter Gruber Foundation
2001 – 2002	Board of Directors, Cleveland BioEnterprise Corporation, Cleveland, OH
2001 – 2002	Board of Trustees, Great Lakes Science Center, Cleveland, OH
2000 – 2003	Strategic Advisory Board, NineSigma, Inc., Cleveland, OH
1999 – 2003	Technology Advisory Council, Biomec, Inc., Cleveland, OH
1999 – 2002	Board of Trustees, Edison Biotechnology Center, Inc.
1995	Co-Founder, Athersys, Inc., Cleveland, OH
1988 – 1998	Consultant, Oncor, Inc. and OncorMed, Inc., Gaithersburg, MD

Publications
ORCHID I.D. 0000-0002-7755-0727

Books:

Thompson MW, McInnes RR, **Willard HF** (1991). *Genetics in Medicine*, 5th Edition. Philadelphia: WB Saunders Co., 500 pp.

Nussbaum RL, McInnes RR, **Willard HF** (2001). *Genetics in Medicine*, 6th Edition. Philadelphia: WB Saunders Co., 444 pp.

Nussbaum RL, McInnes RR, **Willard HF** (2007). *Genetics in Medicine*, 7th Edition. Philadelphia: WB Saunders Co., 585 pp.

Nussbaum RL, McInnes RR, **Willard HF** (2015). *Genetics in Medicine*, 8th Edition. Philadelphia: WB Saunders Co., 546 pp.

Willard HF, Ginsburg G (eds) (2008). *Genomic and Personalized Medicine* (2 vols). New York: Elsevier, 1558 pp.

Ginsburg G, **Willard HF** (eds) (2009). *Essentials of Genomic and Personalized Medicine*. New York: Elsevier, 785 pp.

Ginsburg G, **Willard HF** (eds) (2012). *Genomic and Personalized Medicine,* 2nd edition (2 vols). New York: Elsevier, 1305 pp.

Ginsburg G, **Willard HF** (eds) (2016). *Genomic and Precision Medicine: Foundations, Translation, and Implementation*, 3rd edition. New York: Elsevier, 399 pp.

Original Contributions in Refereed Journals:

1. Latt SA, Stetten G, Juergens LA, **Willard HF**, Scher CD (1975). Recent developments in the detection of DNA synthesis by 33258 Hoechst fluorescence. J. Histochem. Cytochem. 23:493-505.
2. **Willard HF**, Latt SA (1976). Analysis of DNA replication in human X chromosomes by fluorescence microscopy. Am. J. Hum. Genet. 28:213-227.
3. Latt SA, **Willard HF**, Gerald PS (1976). BrdU-33258 Hoechst analysis of DNA replication in human lymphocytes with supernumerary or structurally abnormal X chromosomes. Chromosoma 57:135-153.
4. **Willard HF**, Ambani LM, Hart AC, Mahoney MJ, Rosenberg LE (1976). Rapid prenatal and postnatal detection of inborn errors of propionate, methylmalonate and cobalamin metabolism: a sensitive assay using cultured cells. Hum. Genet. 34:277-283.
5. **Willard HF** (1977). Tissue-specific heterogeneity in DNA replication patterns of human X chromosomes. Chromosoma 61:61-73.
6. Mellman IS, Youngdahl-Turner P, **Willard HF**, Rosenberg LE (1977). Intracellular binding of radioactive hydroxycobalamin to cobalamin-dependant apoenzymes in rat liver. Proc. Natl. Acad. Sci. USA 74:916-920.
7. **Willard HF**, Rosenberg LE (1977). Inherited deficiencies of human methylmalonyl CoA mutase activity: reduced affinity of mutant apoenzyme for adenosylcobalamin. Biochem. Biophys. Res. Comm. 78:927-934.

8. **Willard HF**, Mellman IS, Rosenberg LE (1978). Genetic complementation among inherited deficiencies of methylmalonyl CoA activity: evidence for a new class of human cobalamin mutant. Am. J. Hum. Genet. 30:1-13.

9. Mellman IS, **Willard HF**, Rosenberg LE (1978). Cobalamin binding and cobalamin-dependent enzyme activity in normal and mutant human fibroblasts. J. Clin. Invest. 62:952-960.

10. **Willard HF**, Rosenberg LE (1979). Inborn errors of cobalamin metabolism: effect of cobalamin supplementation in culture on methylmalonyl CoA mutase activity in normal and mutant human fibroblasts. Biochem. Genet.17:57-75.

11. Mellman IS, **Willard HF**, Youngdahl-Turner P, Rosenberg LE (1979). Cobalamin coenzyme synthesis in normal and mutant human fibroblasts: evidence for a processing enzyme activity deficient in cbl C cells. J. Biol. Chem. 254:11847-11853.

12. Wolf B, **Willard HF**, Rosenberg LE (1980). Kinetic analysis of genetic complementation in heterokaryons of propionyl CoA carboxylase-deficient human fibroblasts. Am. J. Hum. Genet. 32:16-25.

13. **Willard HF**, Rosenberg LE (1980). Interactions of methylmalonyl CoA mutase from human fibroblasts with adenosylcobalamin and methylmalonyl CoA: evidence for non-equivalent active sites. Arch. Biochem. Biophys. 200:130-139.

14. **Willard HF**, Breg WR (1980). Human X chromosomes: synchrony of DNA replication in diploid and triploid fibroblasts with multiple active or inactive X chromosomes. Somat. Cell Genet. 6:187-198.

15. **Willard HF**, Rosenberg LE (1980). Inherited methylmalonyl CoA mutase apoenzyme deficiency in human fibroblasts: evidence of allelic heterogeneity, genetic compounds, and codominant expression. J. Clin. Invest. 65:690-698.

16. Schmeckpeper BJ, **Willard HF**, Smith KD (1981). Isolation and characterization of cloned human DNA fragments carrying reiterated sequences common to both autosomes and the X chromosome. Nucl. Acids Res. 9:1853-1872.

17. Fenton WA, Hack A, **Willard HF**, Gertler A, Rosenberg LE (1982). Purification and properties of methylmalonyl CoA mutase from normal human liver. Arch. Biochem. Biophys. 214:815-823.

18. **Willard HF**, Smith KD, Sutherland J (1983). Isolation and characterization of a major tandem repeat family from the human X chromosome. Nucl. Acids Res. 11:2017-2033.

19. **Willard HF**, Holmes MT (1984). Sensitive and dependable assay for distinguishing hamster and human X-linked steroid sulfatase activity in somatic cell hybrids. Hum. Genet. 66:272-275.

20. Worton RG, Duff C, Sylvester JE, Schmickel RD, **Willard HF** (1984). Duchenne muscular dystrophy involving translocation of the dmd gene next to ribosomal RNA genes. Science 224:1447-1449.

21. Caccia N, Kronenberg M, Saxe D, Haars R, Bruns G, Goverman J, Malissen M, **Willard HF**, Yoshikai J, Simon M, Hood L, Mak T (1984). The T cell receptor ß chain genes are located on chromosome 6 in mouse and chromosome 7 in humans. Cell 37:1091-1099.

22. Korneluk RG, Quan F, Lewis W, Guise KS, **Willard HF**, Holmes MT, Gravel RA (1984). Isolation of human fibroblast catalase cDNA clones: Sequence of clones derived from spliced and unspliced mRNA. J. Biol. Chem. 259:13819-13823.

23. Rubin JS, Prideaux VR, **Willard HF**, Dulhanty AM, Whitmore GF, Bernstein A (1985). Molecular cloning and chromosomal localization of DNA sequences associated with a human DNA repair gene. Mol. Cell Biol. 5:398-405.

24. O'Dowd B, Quan F, **Willard HF**, Korneluk RG, Lowden JA, Gravel RA, Mahuran DJ (1985). Isolation and characterization of cDNA clones encoding the ß-subunit of human ß-hexosaminidase. Proc. Natl. Acad. Sci. USA 82:1184-1188.

25. Ingle C, Williamson R, de la Chapelle A, Herva RR, Haapala K, Bates G, **Willard HF**, Davies KE (1985). Mapping DNA sequences in a human X chromosome deletion which extends across the region of the Duchenne muscular dystrophy mutation. Am. J. Hum. Genet. 37:451-472.

26. **Willard HF** (1985). Chromosome-specific organization of human alpha satellite DNA. Am. J. Hum. Genet. 37:524-532.

27. Wolfe J, Darling SM, Erickson RP, Craig I, Buckle V, Rigby P, **Willard HF**, Goodfellow P (1985). Isolation and characterization of an alphoid centromeric repeat family from the human Y chromosome. J. Mol. Biol. 182:477-485.

28. Bakker E, Hofker MH, Goor N, Mandel JL, Davis KE, Kunkel LM, **Willard HF**, Fenton WA, Sandkuyl L, Majoor-Krakauer D, v. Essen AJ, Jahoda MGJ, Sachs ES, Van Ommen GJB, Pearson PL (1985). Prenatal diagnosis and carrier-detection of Duchenne muscular dystrophy with closely linked RFLPs. Lancet 2:655-658.

29. Waye JS, **Willard HF** (1985). Chromosome-specific alpha satellite DNA: Nucleotide sequence analysis of the 2.0 kilobasepair repeat from the human X chromosome. Nucl. Acids Res. 12:2731-2743.

30. Michalopoulos EE, Bevilacqua PJ, Stokoe N, Powers VE, **Willard HF**, Lewis WH (1985). Molecular analysis of gene deletion in aniridia-Wilms tumor association. Hum. Genet. 70:157-162.

31. **Willard HF**, Meakin S, Tsui LC, Breitman M (1985). Assignment of the human gamma crystallin multigene family to chromosome 2. Somat. Cell Mol. Genet. 11:511-516.

32. **Willard HF**, Goss SJ, Holmes MT, Munroe DL (1985). Regional localization of the phosphoglycerate kinase gene and pseudogene on the human X chromosome and assignment of a related DNA sequence to chromosome 19. Hum. Genet. 71:138-143.

33. Schmeckpeper B, Davis J, **Willard HF**, Smith K (1985). An anonymous single-copy X chromosome RFLP for DXS72 from Xq13-Xq22. Nucl. Acids Res. 13:5724.

34. **Willard HF**, Riordan JR (1985). Assignment of the gene for myelin proteolipid protein to the X chromosome: implications for X-linked inherited disorders of myelin. Science 230:940-942.

35. Lewis WH, Goguen JM, Powers VE, **Willard HF**, Michalopoulos EE (1985). Gene order on the short arm of human chromosome 11: regional assignment of the LDHA gene distal to catalase in two translocations. Hum. Genet. 71:249-253.

36. Lamhonwah AM, Barankiewicz TJ, **Willard HF**, Mahuran DJD, Quan F, Gravel RA (1986). Isolation of cDNA clones coding for the alpha and beta chains of human propionyl CoA carboxylases: chromosomal assignments and DNA polymorphisms associated with PCCA and PCCB genes. Proc. Natl. Acad. Sci. USA 83:4864-4868.

37. Buchwald M, Zsiga M, Markiewicz D, Plavsic N, Kennedy D, Zengerling S, **Willard HF**, Tsipouras P, Schmiegelow K, Schwartz M, Eiberg H, Mohr J, Donis-Keller H, Tsui L-C (1986). Linkage of cystic fibrosis to the pro a2 (I) collagen gene, COL1A2, on chromosome 7. Cytogenet. Cell. Genet. 41:234-239.

38. Korneluk RG, Mahuran DJ, Neote K, Klavins MH, O'Dowd BF, Tropak M, **Willard HF**, Anderson MJ, Lowden JA, Gravel RA (1986). Isolation of cDNA clones coding for the α subunit of human

β-hexosaminidase: extensive homology between the α and β subunits and studies on Tay-Sachs disease. J. Biol. Chem. 261:8407-8413.

39. Williams BRG, Saunders M, **Willard HF** (1986). The interferon-induced human 2-5A synthetase gene is on chromosome 12. Somat. Cell Mol. Genet. 12:403-408.

40. **Willard HF**, Waye JS, Skolnick MH, Schwartz CE, Powers VE, England SB (1986). Restriction fragment length polymorphisms at the centromeres of human chromosomes using chromosome-specific alpha satellite DNA: implications for development of centromere-based genetic linkage maps. Proc. Natl. Acad. Sci. USA 83:5611-5615.

41. Squire J, Dryja TP, Dunn JJ, Goddard A, Hoffman T, Musarella M, **Willard HF**, Becker AJ, Gallie BL, Phillips RA (1986). Cloning of the esterase D gene: a polymorphic gene probe closely linked to the retinoblastoma locus on chromosome 13. Proc. Natl. Acad. Sci. USA 83:6573-6577.

42. Glaser T, Lewis WH, Bruns GAP, Watkins PC, Roger LE, Shows TB, Powers VE, **Willard HF**, Goguen JM, Simola KOJ, Housman DE (1986). Beta subunit of follicle stimulating hormone is deleted in patients with aniridia and Wilm's tumor: a further definition of the WAGR locus. Nature 321:882-887.

43. Waye JS, **Willard HF** (1986). Structure, organization, and sequence of alpha satellite DNA from human chromosome 17: evidence for evolution by unequal crossing-over and an ancestral pentamer repeat shared with the human X chromosome. Mol. Cell. Biol. 6:3156-3165.

44. Waye JS, **Willard HF** (1986). Molecular analysis of a deletion polymorphism in alpha satellite of human chromosome 17: evidence of homologous unequal crossing-over and subsequent fixation. Nucl. Acids Res. 14:6915-6927.

45. Cooke NE, **Willard HF**, David EV, George DL (1986). Direct regional assignment of the gene for vitamin D-binding protein (Gc-globulin) to human chromosome 4q11-q13 and identification of associated DNA polymorphisms. Hum. Genet. 73:225-229.

46. O'Dowd BF, Klavins MH, **Willard HF**, Gravel RA, Lowden A, Mahuran DJ (1986). Molecular heterogeneity in the infantile and juvenile forms of Sandhoff disease. J. Biol. Chem. 261:12680-12685.

47. Durfy SJ, Clark SC, Williams BRG, **Willard HF** (1986). RFLP detected by an X-linked cDNA encoding erythroid-potentiating activity / tissue inhibitor of metalloproteinase (EPA/TIMP). Nucl. Acids Res. 14:9226.

48. Waye JS, England SB, **Willard HF** (1987). Genomic organization of alpha satellite DNA on human chromosome 7: evidence for two distinct alphoid domains on a single chromosome. Mol. Cell. Biol. 7:349-356.

49. Bell DR, Trent JM, **Willard HF**, Riordan JR, Ling V (1987). Chromosomal location of human P-glycoprotein gene sequences. Cancer Genet. Cytogenet. 25:141-148.

50. Wu J-S, Riordan JR, **Willard HF**, Milner R, Kidd KK (1987). MSP RFLP for X-linked proteolipid protein gene (PLP) identified with either rat or human PLP cDNA clone. Nucl. Acids Res. 15:1882.

51. Barker D, Wright E, Nguyen K, Cannon L, Fain P, Goldgar D, Bishop DT, Carey J, Baty B, Kivlin J, **Willard HF**, Waye JS, Greig G, Leinwand L, Nakamura Y, O'Connell P, Leppert M, Lalouel JM, White R, Skolnick M (1987). Gene for von Recklinghausen neurofibromatosis is in the pericentromeric region of chromosome 17. Science 236:1100-1102.

52. Waye JS, Creeper LA, **Willard HF** (1987). Organization and evolution of alpha satellite DNA from human chromosome 11. Chromosoma 95:182-188.

53. **Willard HF**, Waye JS (1987). Hierarchical order in chromosome-specific human alpha satellite DNA. Trends in Genetics 3:192-198.

54. Mengle-Gaw L, **Willard HF**, Smith CIE, Hammarstrom L, Fischer P, Sherrington P, Lucas G, Thompson PW, Baer R, Rabbitts TH (1987). Human T-Cell tumours containing chromosome 14 inversion or translocation with breakpoints proximal to immunoglobulin joining regions at 14q32. EMBO J. 6:2273-2280.

55. **Willard HF**, Waye JS (1987). Chromosome-specific subsets of human alpha satellite DNA: Analysis of sequence divergence within and between chromosomal subsets and evidence for an ancestral pentameric repeat. J. Mol. Evol. 25:207-214.

56. Durfy SJ, **Willard HF** (1987). Molecular analysis of a polymorphic domain of X chromosome alpha satellite DNA. Am. J. Hum. Genet. 41:391-401.

57. MacLennan DH, Brandl CJ, Champaneria S, Holland PC, Powers VE, **Willard HF** (1987). Fast-twitch and slow-twitch / cardiac Ca^{2+} ATPase genes map to human chromosomes 16 and 12. Somat. Cell Mol. Genet. 13:341-346.

58. O'Dowd B, Neote K, Munroe DLG, Gravel RA, Mahuran D, **Willard HF** (1987). PstI RFLP in the human hexosaminidase (*HEXB*) gene on chromosome 5. Nucl. Acids Res. 15:3194.

59. Waye JS, Durfy SJ, Pinkel D, Kenwrick S, Patterson M, Davies KE, **Willard HF** (1987). Chromosome-specific alpha satellite DNA from human chromosome 1: Hierarchical structure and genomic organization of a polymorphic domain spanning several hundred kilobasepairs of centromeric DNA. Genomics 1:43-51.

60. Waye JS, **Willard HF** (1987). Nucleotide sequence heterogeneity of alpha satellite repetitive DNA: a survey of alphoid sequences from different human chromosomes. Nucl. Acids Res. 15:7549-7580.

61. Waye JS, Greig GM, **Willard HF** (1987). Detection of novel centromeric polymorphisms associated with alpha satellite DNA from human chromosome 11. Hum. Genet. 77:151-156.

62. Barker D, Green P, Knowlton R, Schumm J, Lander E, Oliphant A, **Willard HF**, Akots G, Brown V, Gravius T, Helms C, Nelson C, Parker C, Rediker K, Watt D, Weiffenbach B, Donis-Keller H (1987). A genetic linkage map of 63 chromosome 7 DNA markers. Proc. Natl. Acad. Sci. USA 84:8006-8010.

63. Barker D, Wright E, Nguyen K, Cannon L, Fain P, Goldgar D, Bishop DT, Carey J, Kivlin J, **Willard HF**, Nakamura Y, O'Connell P, Leppert P, White R, Skolnick M (1987). A genomic search for linkage of NF to RFLP's. J. Med. Genet. 24:536-538.

64. **Willard HF**, Greig GM, Powers VE, Waye JS (1987). Molecular organization and haplotype analysis of centromeric DNA from human chromosome 17: implications for linkage in neurofibromatosis. Genomics 1: 368-373.

65. Vogelstein B, Fearon ER, Hamilton SR, Preisinger AC, **Willard HF**, Michelson AM, Riggs AD, Orkin S (1987). Clonal analysis using recombinant DNA probes from the human X chromosome. Cancer Res. 47:4806-4813.

66. Brown CJ, **Willard HF** (1987). MspI RFLP detected with chromosome-walk clone pXUT23-SE3.2L from DXS16 in Xp22.1-22.3. Nucl. Acids Res. 15:9614.

67. Fain PR, Barker DF, Goldgar DE, Wright E, Nguyen K, Carey J, Johnson J, Kivlin J, **Willard HF**, Mathew C, Ponder B, Skolnick M (1987). Genetic analysis of NF1: identification of close flanking markers on chromosome 17. Genomics 1:340-345.

68. Spence JF, Perciaccante RG, Greig GM, **Willard HF**, Ledbetter DH, Hejtmancik JF, Pollack MS, O' Brien WE, Beaudet AL (1988). Uniparental disomy as a mechanism for human genetic disease. Am. J. Hum. Genet. 42:217-226.

69. Worton RG, Sutherland J, Sylvester JE, **Willard HF**, Bodrug S, Dube I, Duff C, Kean V, Ray PN, Schmickel RD (1988). Human ribosomal RNA genes: orientation of the tandem array and conservation of the 5' end. Science 239:64-68.

70. Tsipouras P, Schwartz RC, Phillips JA III, **Willard HF** (1988). A centromere-based linkage group on the long arm of chromosome 17. Cytogenet. Cell Genet. 47:109-111.

71. Waye JS, Mitchell AR, **Willard HF** (1988). Organization and genomic distribution of '82H' alpha satellite DNA: Evidence for a low-copy or single-copy alphoid domain located on human chromosome 14. Hum. Genet. 78:27-32.

72. Waye JS, Gravel RA, **Willard HF** (1988). Two PstI RFLPs in the PCCB gene on the long arm of chromosome 3. Nucl. Acids Res. 16:2362.

73. Allore R, O'Hanlon D, Price R, Neilson K, **Willard HF**, Cox D, Marks A, Dunn RJ (1988). Gene encoding the β subunit of S100 protein is on chromosome 21: implications for Down Syndrome. Science 239:1311-1313.

74. Lubahn DB, Joseph DR, Sullivan DM, **Willard HF**, French FS, Wilson EM (1988). Cloning of human androgen receptor cDNA and localization to the X chromosome. Science 240:327-330.

75. Otulakowski G, Robinson BH, **Willard HF** (1988). The gene for lipoamide dehydrogenase maps to human chromosome 7. Somat. Cell Mol. Genet. 14:411-414.

76. Mahtani MM, **Willard HF** (1988). A primary genetic map of the pericentromeric region of the human X chromosome. Genomics 2:294-301.

77. Devilee P, Kievits T, Waye JS, Pearson PL, **Willard HF** (1988). Chromosome-specific alpha satellite DNA: isolation and mapping of a polymorphic alphoid repeat from human chromosome 10. Genomics 3:1-7.

78. Mitchell GA, Looney JE, Brody LC, Steel G, Suchanek M, Engelhardt JF, **Willard HF**, Valle D (1988). Human ornithine-δ-aminotransferase: cDNA cloning and analysis of the structural gene. J. Biol. Chem. 263:14288-14295.

79. Brown CJ, Mahtani MM, **Willard HF** (1988). Genetic mapping of four DNA markers (DXS16, DXS43, DXS85 and DXS143) from the p22 region of the human X chromosome. Hum. Genet. 80:296-298.

80. Devilee P, Thierry RF, Kievits T, Kolluri R, Hopman AHN, **Willard HF**, Pearson PL, Cornelisse CJ (1988). Detection of chromosome aneuploidy in interphase nuclei from human primary breast tumours using chromosome-specific repetitive DNA probes. Cancer Res. 48:5825-5830.

81. Brown CJ, Sekiguchi T, Nishimoto T, **Willard HF** (1989). Regional localization of the CCG1 gene which complements the hamster cell cycle mutation BN462 to Xq11-Xq13. Somat. Cell Mol. Genet. 15:93-96.

82. Stephens K, Green P, Riccardi VM, Ng S, Rising M, Barker D, Darby J, Falls KM, Collins FS, **Willard HF**, Donis-Keller H (1989). Genetic analysis of eight loci tightly linked to neurofibromatosis. Am. J. Hum. Genet. 44:13-19.

83. Fain PR, Wright E, **Willard HF**, Stephens K, Barker DF (1989). The order of loci in the pericentric region of chromosome 17, based on evidence from physical and genetic breakpoints. Am. J. Hum. Genet. 44:68-72.

84. **Willard HF**, Durfy SJ, Mahtani MM, Dorkins H, Davies KE, Williams BRG (1989). Regional localization of the *TIMP* gene on the human X chromosome: extension of a conserved synteny and linkage group on proximal Xp. Hum. Genet. 81:234-238.

85. Logan C, **Willard HF**, Rommens JM, Joyner AL (1989). Chromosomal localization of the human homeo box-containing genes, *EN1* and *EN2*. Genomics 4:206-209.

86. Brown CJ, Goss SJ, Lubahn DB, Joseph DR, Wilson EM, French FS, **Willard HF** (1989). Androgen receptor locus on the human X chromosome: Regional localization to Xq11-12 and description of a DNA polymorphism. Am. J. Hum. Genet. 44:264-269.

87. Waye JS, **Willard HF** (1989). Chromosome-specificity of satellite DNAs: short and long-range organization of a diverged dimeric subset of human alpha satellite from chromosome 3. Chromosoma 95:275-280.

88. Brown CJ, Powers VE, Munroe DL, Sheinin R, **Willard HF** (1989). A gene on the short arm of the human X chromosome complements the murine tsA1S9 DNA synthesis mutation. Somat. Cell Mol. Genet. 15:173-178.

89. Waye JS, **Willard HF** (1989). Human beta satellite DNA: genomic organization and sequence definition of a class of highly repetitive tandem DNA. Proc. Natl. Acad. Sci. USA 86:6250-6254.

90. Waye JS, **Willard HF** (1989). Concerted evolution of alpha satellite DNA: evidence for species specificity and a general lack of sequence conservation among alphoid sequences of higher primates. Chromosoma 98:273-279.

91. Alitalo T, **Willard HF**, de la Chapelle A (1989). Determination of the breakpoint of 1;7 translocations in myelodyoplastic syndrome by in situ hybridization using chromosome-specific alpha satellite DNA from human chromosomes 1 and 7. Cytogenet. Cell Genet. 50:49-53.

92. Greer WL, Mahtani MM, Kwong PC, Rubin LA, Peacocke M, **Willard HF**, Siminovitch KA (1989). Linkage studies of the Wiskott-Aldrich syndrome: polymorphisms at TIMP and the X chromosome centromere are informative markers for genetic prediction. Hum. Genet. 83:227-230.

93. Brown CJ, **Willard HF** (1989). Non-inactivation of a selectable human X-linked gene that complements a murine temperature-sensitive cell cycle defect. Am. J. Hum. Genet. 45:592-598.

94. Durfy SJ, **Willard HF** (1989). Patterns of intra- and interarray sequence variation in alpha satellite from the human X chromosome: Evidence for short-range homogenization of tandemly repeated DNA sequences. Genomics 5:810-821.

95. Wevrick R, **Willard HF** (1989). Long-range organization of tandem arrays of alpha satellite at the centromeres of human chromosomes: High frequency array length polymorphism and meiotic stability. Proc. Natl. Acad. Sci. USA 86:9394-9398.

96. Lafreniere RG, Mahtani MM, **Willard HF** (1989). An X-linked Dra I RFLP recognized by cpX23 (DXS132). Nucl. Acids Res. 17:6754.

97. Greig GM, England SB, Bedford HM, **Willard HF** (1989). Chromosome-specific alpha satellite DNA from the centromere of human chromosome 16. Am. J. Hum. Genet. 45:862-872.

98. Tran-Paterson R, **Willard HF**, Letarte M (1989). The common acute lymphoblastic leukemia antigen (neutral endopeptidase-3.4.24.11) gene is located on human chromosome 3. Cancer Genet. Cytogenet. 42:129-134.

99. Brown CJ, **Willard HF** (1990). Localization of a gene that escapes inactivation to the X chromosome proximal short arm: implications for X inactivation. Am. J. Hum. Genet. 45:273-279.

100. Koch J, Kolvraa S, Hobolt N, Petersen GB, **Willard HF**, Waye JS, Gregersen N, Bolund L (1990). A case of 46,XX,r(X) (p1q1) diagnosed by in situ hybridization. Clin. Genet. 37:216-220.

101. Luty JA, Guo Z, **Willard HF**, Ledbetter DH, Ledbetter S, Litt M (1990). Five polymorphic microsatellite VNTRs on the human X chromosome. Am. J. Hum. Genet. 46:776-783.

102. Fujii J, **Willard HF**, MacLennan DH (1990). Characterization and localization to human chromosome 1 of the human fast-twitch skeletal muscle calsequestrin gene. Somat. Cell Mol. Genet. 16:185-189.

103. Clarke JTR, **Willard HF**, Teshima I, Chang PL, Skomorowski MA (1990). Hunter disease (mucopolysaccharidosis type II) in a karyotypically normal girl. Clin. Genet. 37:355-362.

104. Lafreniere RG, **Willard HF** (1990). A RsaI RFLP at the DXS467 locus. Nucl. Acids Res. 18:3673.

105. Lafreniere RG, **Willard HF** (1990). A BstEII RFLP at the DXS153 locus. Nucl. Acids Res. 18:3673.

106. MacKenzie AE, Korneluk RG, Zorzato F, Fujii J, Phillips M, Iles D, Wieringa B, Leblond S, Bailly J, **Willard HF**, Duff C, Worton RG, MacLennan DH (1990). The human ryanodine receptor gene: its mapping to 19q13.1, placement in a chromosome 19 linkage group, and exclusion as the gene causing myotonic dystrophy. Am. J. Hum. Genet. 46: 1082-1089.

107. Mahtani MM, **Willard HF** (1990). Pulsed-field gel analysis of alpha satellite DNA at the human X chromosome centromere: high-frequency polymorphisms and array size estimate. Genomics 7:607-613.

108. Brown CJ, Flenniken AM, Williams BRG, **Willard HF** (1990). X chromosome inactivation of the human TIMP gene. Nucl. Acids Res. 18:4191-4195.

109. Ozcelik T, Lafreniere RG, Archer BT, Johnston PA, **Willard HF**, Francke U, Sudhof TC (1990). Synaptophysin: structure of the human gene and assignment to the X chromosome in man and mouse. Am. J. Hum. Genet. 42:551-561.

110. Sharp CB, Bedford HM, **Willard HF** (1990). Pericentromeric structure of human X "isochromosomes": evidence for molecular heterogeneity. Hum. Genet. 85:330-336.

111. Norum RA, Lafreniere RG, O'Neal LW, Nikolai TF, Delaney JP, Sisson JC, Sobol H, Lenoir GM, Ponder BA, **Willard HF**, Jackson CE (1990). Linkage of the multiple endocrine neoplasia type 2B gene (MEN2B) to chromosome 10 markers linked to MEN2A. Genomics 8:313-317.

112. Ten Hagen KG, Gilbert DM, **Willard HF**, Cohen SN (1990). Replication timing of DNA sequences associated with human centromeres and telomeres. Mol. Cell Biol. 10:6348-6355.

113. Wevrick R, Earnshaw WC, Howard-Peebles PN, **Willard HF** (1990). Partial deletion of alpha satellite DNA associated with reduced amounts of the centromere protein CENP-B in a mitotically stable human chromosome rearrangement. Mol. Cell. Biol. 10:6374-6380.

114. Warburton PE, **Willard HF** (1990). Genomic analysis of sequence variation in tandemly repeated DNA: evidence for localized homogeneous sequence domains within arrays of alpha satellite DNA. J. Mol. Biol. 216:3-16.

115. **Willard HF** (1990). Centromeres of mammalian chromosomes. Trends in Genetics 6:410-416.

116. Watt VM, **Willard HF** (1990). The human aminopeptidase N gene: isolation, chromosome localization, and DNA polymorphism analysis. Hum. Genet. 85:651-654.

117. Shier P, **Willard HF**, Watt VM (1990). Localization of insulin receptor-related receptor gene, IRR, to human chromosome 1. Cytogenet. Cell Genet. 54:80-81.

118. Fisher EMC, Beer-Romero P, Brown LG, Ridley A, McNeil JA, Lawrence JB, **Willard HF**, Bieber FR, Page DC (1990). Homologous ribosomal protein genes on the human X and Y chromosomes: escape from X inactivation and implications for Turner syndrome. Cell 63: 1205-1218.

119. Durfy SJ, **Willard HF** (1990). Concerted evolution of primate alpha satellite DNA: evidence for an ancestral sequence shared by gorilla and human X chromosome alpha satellite. J. Mol. Biol. 216:555-566.

120. Dietz-Band JN, Turco AE, **Willard HF**, Vincent A, Skolnick MH, Barker DF (1990). Isolation, characterization, and physical localization of 33 human X chromosome RFLP markers. Cytogenet. Cell Genet. 54: 137-141.

121. Chun K, Mackay N, **Willard HF**, Robinson BH (1990). Isolation, characterization and chromosomal localization of cDNA clones for the $E_1\beta$ subunit of the pyruvate dehydrogenase complex. Eur. J. Biochem. 194:587-592.

122. Brown CJ, Ballabio A, Rupert JL, Lafreniere RG, Grompe M, Tonlorenzi R, **Willard HF** (1991). A gene from the region of the human X inactivation centre is expressed exclusively from the inactive X chromosome. Nature 349:38-44.

123. Brown CJ, Lafreniere RG, Powers VE, Sebastio G, Ballabio A, Pettigrew AL, Ledbetter DH, Levy E, Craig IW, **Willard HF** (1991). Localization of the X inactivation centre on the human X chromosome in Xq13. Nature 349:82-84.

124. Trifiro M, Gottlieb B, Pinsky L, Kaufman M, Prior L, Belsham DD, Wrogemann K, Brown CJ, **Willard HF**, Trapman J, Brinkmann AO, Chang C, Liao S, Sergovich F, Jung J (1991). The 56/58 androgen-binding protein in male genital skin fibroblasts with a deleted androgen receptor gene. Mol. Cell. Endocrin. 75:37-47.

125. Dracopoli NC, O'Connell P, Elsner T I, Lalouel J-M, White RL, Buetow KH, Nishimura DY, Murray JC, Helms C, Mishra SK, Donis-Keller H, Hall JM, Lee MK, King M-C, Attwood J, Morton NE, Robson EB, Mahtani M, **Willard HF**, Royle NJ, Patel I, Jeffreys AJ, Verga V, Jenkins T, Weber JL, Mitchell AL, Bale A (1991). The CEPH consortium linkage map of human chromosome 1. Genomics 9:686-700.

126. Lafreniere RG, Geraghty MT, Valle D, Shows TB, **Willard HF** (1991). Ornithine aminotransferase-related sequences map to two non-adjacent intervals on the human X chromosome short arm. Genomics 10:276-279.

127. Wevrick R, **Willard HF** (1991). Physical map of the centromeric region of human chromosome 7: relationship between two distinct alpha satellite arrays. Nucl. Acids Res. 19:2295-2301.

128. Borsani G, Tonlorenzi R, Simmler MC, Dandolo L, Arnaud D, Capra V, Grompe M, Pizzuti A, Muzny D, Lawrence C, **Willard HF**, Avner P, Ballabio A (1991). Characterization of a murine gene expressed from the inactive X chromosome. Nature 351:325-329.

129. Greig GM, Parikh S, George J, Powers VE, **Willard HF** (1991). Molecular cytogenetics of alpha satellite DNA from chromosome 12: fluorescence in situ hybridization and description of DNA and array length polymorphisms. Cytogenet. Cell Genet. 56: 144-148.

130. Taylor RG, Garcia-Heras J, Sadler SJ, Lafreniere RG, **Willard HF**, Ledbetter DH, McInnes RR (1991). Localization of histidase to human chromosome 12q22-24.1 and mouse chromosome 10C2-D1. Cytogenet. Cell Genet. 56: 178-181.

131. Mahtani MM, Lafreniere RG, Kruse T, **Willard HF** (1991). An 18-locus linkage map of the pericentromeric region of the human X chromosome: genetic framework for mapping X-linked disorders. Genomics 10:849-857.

132. Losada AP, Wessman M, Tiainen M, Hopman AHN, **Willard HF**, Sole F, Casballin MR, Woessner S, Knuutila S (1991). Trisomy 12 in chronic lymphocytic leukemia: an interphase cytogenetic study. Blood 78:775-779.

133. Kirchgessner CU, Trofatter JA, Mahtani MM, **Willard HF**, DeGennaro LJ (1991). A highly polymorphic dinucleotide repeat on the proximal short arm of the human X chromosome: linkage mapping of the synapsin I/A-raf-1 genes. Am. J. Hum. Genet. 49:184-191.

134. Fujii J, Zarain-Herzberg A, **Willard HF**, Tada M, MacLennan DH (1991). Structure of the rabbit phospholamban gene, cloning of the human cDNA and assignment of the gene to human chromosome 6. J. Biol. Chem. 266:11669-11675.

135. Warburton PE, Greig GM, Haaf T, **Willard HF** (1991). PCR amplification of chromosome-specific alpha satellite DNA: definition of centromeric STS markers and polymorphic analysis. Genomics 11:324-333.

136. Lafreniere RG, Brown CJ, Powers VE, Carrel L, Davies KE, Barker DF, **Willard HF** (1991). Physical mapping of 60 DNA markers in the p21.1 → q21.3 region of the human X chromosome. Genomics 11:352-363.

137. Franco B, Guioli S, Pragliola A, Incerti B, Bardoni B, Tonlorenzi R, Carrozzo R, Maestrini E, Pieretti M, Taillon-Miller P, Brown C, **Willard HF**, Lawrence C, Persico G, Camerino G, Ballabio A (1991). A gene deleted in Kallmann's syndrome shares homology with neural cell adhesion and axonal pathfinding molecules. Nature 353:529-536.

138. Coleman MP, Murray JC, **Willard HF**, Nolan KF, Reid KBM, Blake DJ, Lindsay S, Bhattacharya SS, Wright A, Davies KE (1991). Genetic and physical mapping around the properdin P gene. Genomics 11:991-996.

139. Greig GM, **Willard HF** (1992). β satellite DNA: Characterization and localization of two subfamilies from the distal and proximal short arms of the human acrocentric chromosomes. Genomics 12:573-580.

140. Quigley CA, Friedman KJ, Johnson A, Lafreniere RG, Silverman LM, Lubahn DB, Brown TR, Wilson EM, **Willard HF**, French FS (1992). Complete deletion of the androgren receptor gene: Definition of the null phenotype of the androgen insensitivity syndrome and determination of carrier status. J. Clin. Endocrin. Metab. 74:927-933.

141. Haaf T, **Willard HF** (1992). Organization, polymorphism, and molecular cytogenetics of chromosome-specific α-satellite DNA from the centromere of chromosome 2. Genomics 13:122-128.

142. Cotter PD, **Willard HF**, Gorski JL, Bishop DF (1992). Assignment of human erythroid d-aminolevulinate synthase (ALAS2) to a distal subregion of band Xp11.21 by PCR analysis of somatic cell hybrids containing X; autosome translocations. Genomics 13:211-212.

143. Haaf T, Sumner AT, Köhler J, **Willard HF**, Schmid M (1992). A microchromosome derived from chromosome 11 in a patient with the CREST syndrome of scleroderma. Cytogenet. Cell Genet. 60:12-17.

144. Corcos IA, Lafreniere RG, Begy CR, Loch-Caruso R, **Willard HF**, Glover TW (1992). Refined localization of human connexin 32 gene locus, GJB1, to Xq13.1. Genomics 13:479-480.

145. Haaf T, Warburton PE, **Willard HF** (1992). Integration of human alpha satellite DNA into simian chromosomes: centromere protein binding and disruption of normal chromosome segregation. Cell 70:681-696.

146. Mandel J-L, Monaco AP, Nelson DL, Schlessinger D, **Willard HF** (1992). Genome analysis and the human X chromosome. Science 258:103-109.

147. Brown CJ, Hendrich BD, Rupert JL, Lafreniere RG, Xing Y, Lawrence J, **Willard HF** (1992). The human *XIST* gene: analysis of a 17 kb inactive X-specific RNA that contains conserved repeats and is highly localized within the nucleus. Cell 71: 527-542.

148. Bascom RA, García-Heras J, Hsieh C-L, Gerhard DS, Jones C, Francke U, **Willard HF**, Ledbetter DH, McInnes RR (1992). 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.

149. Warburton PE, **Willard HF** (1992). PCR amplification of tandemly repeated DNA: analysis of intra- and interchromosomal sequence variation and homologous unequal crossing-over in human alpha satellite DNA. Nucl. Acids Res. 20: 6033-6042.

150. Lorber BJ, Grantham M, Peters J, **Willard HF**, Hassold TJ (1992). Nondisjunction of chromosome 21: comparisons of cytogenetic and molecular studies of the meiotic stage and parent of origin. Am. J. Hum. Genet. 51: 1265-1276.

151. Wevrick R, Willard VP, **Willard HF** (1992). Structure of DNA near long tandem arrays of alpha satellite DNA at the centromere of human chromosome 7. Genomics 14: 912-923.

152. Mahtani MM, **Willard HF** (1993). A polymorphic X-linked tetranucleotide repeat locus displaying a high rate of new mutation: implications for mechanisms of mutation at short tandem repeat loci. Hum. Mol. Genet. 2: 431-437.

153. Hendrich BD, Brown CJ, **Willard HF** (1993). Evolutionary conservation of possible functional domains of the human and murine *XIST* genes. Hum. Mol. Genet. 2: 663-672.

154. Haaf T, Schmid M, Steinlein C, Galetti PM, **Willard HF** (1993). Organization and molecular cytogenetics of a satellite DNA family from Hoplias malabaricus (Pisces, Erythrinidae). Chromosome Res. 1: 77-86.

155. Leppig KA, Brown CJ, Bressler SL, Gustashaw K, Pagon RA, **Willard HF**, Disteche CM (1993). Mapping of the distal boundary of the X inactivation center in a rearranged X chromosome from a female expressing XIST. Hum. Mol. Genet. 2: 883-887.

156. Lafreniere RG, **Willard HF** (1993). Pulsed field map of Xq13 in the region of the X inactivation center. Genomics 17: 502-506.

157. Puck JM, Deschenes SM, Porter JC, Dutra AS, Brown CJ, **Willard HF**, Henthorn PS (1993). The interleukin-2 receptor γ chain maps to Xq13.1 and is mutated in X-linked severe combined immunodeficiency, SCIDX1. Hum. Mol. Genet. 2: 1099-1104.

158. Lafreniere RG, Brown CJ, Rider S, Chelly J, Taillon-Miller P, Chinault AC, Monaco AP, **Willard HF** (1993). 2.6 Mb YAC contig of the human X inactivation center region in Xq13: physical linkage of the RPS4X, PHKA1, XIST, and DXS128E genes. Hum. Mol. Genet. 2: 1105-1115.

159. Greig GM, Sharp CB, Carrel L, **Willard HF** (1993). Duplicated zinc finger protein genes on the proximal short arm of the human X chromosome: isolation, characterization, and X inactivation studies. Hum. Mol. Genet. 2: 1611-1618.

160. Warburton PE, Waye JS, **Willard HF** (1993). Non-random localization of recombination events in human alpha satellite repeat unit variants: implications for higher-order structural characteristics within centromeric heterochromatin. Mol. Cell. Biol. 13: 6520-6529.

161. Greig GM, Warburton PE, **Willard HF** (1993). Organization and evolution of an alpha satellite DNA subset shared by human chromosomes 13 and 21. J. Mol. Evol. 37: 464-475.

162. Duncan AMV, Macdonald A, Brown CJ, Wolff D, **Willard HF**, Sutton B (1993). Characterization of a small supernumerary ring X chromosome by fluorescence in situ hybridization. Am. J. Med. Genet. 47: 1153-1156.

163. Reed V, Laval SH, McCabe V, **Willard HF**, Boyd Y (1994). Mapping of loci and translocation breakpoints in Xq13: isolation of a conserved locus that maps close to CCG1 in human and mouse. Mammalian Genome 5:237-240.

164. Brown CJ, **Willard HF** (1994). The human X inactivation centre is not required for maintenance of X chromosome inactivation. Nature 368: 646-648.

165. Rack KA, Chelly J, Gibbons RJ, Rider S, Benjamin D, Lafreniere RG, Oscier D, Hendriks RW, Craig IW, **Willard HF**, Monaco AP, Buckle VJ (1994). Absence of the XIST gene from late-replicating isodicentric X chromosomes in leukaemia. Hum. Mol. Genet. 3: 1053-1060.

166. Lafreniere RG, Carrel L, **Willard HF** (1994). A novel transmembrane transporter encoded by the XPCT gene in Xq13.2. Hum. Mol. Genet. 3: 1133-1140.

167. Wolff D, Brown CJ, Schwartz S, Duncan AMV, Surti S, **Willard HF** (1994). Small marker X chromosomes lack the X inactivation center: implications for karyotype/phenotype correlations. Am. J. Hum. Genet. 55: 87-95.

168. Gustashaw KM, Zurcher V, Dickerman LH, Stallard R, **Willard HF** (1994). Partial X chromosome trisomy with functional disomy of Xp due to failure of X inactivation. Am. J. Med. Genet. 53:39-45.

169. Brown CJ, Miller AP, Carrel L, Rupert JL, Davies KE, **Willard HF** (1995). The DXS423E gene in Xp11.21 escapes X chromosome inactivation. Hum. Mol. Genet. 4: 251-255.

170. Sultana R, Adler DA, Edelhoff S, Carrel L, Lee K, Chapman VC, **Willard HF**, Disteche CM (1995). The mouse Sb1.8 gene located at the distal end of the X chromosome is subject to X inactivation. Hum. Mol. Genet. 4: 257-263.

171. Alitalo T, Francis F, Kere J, Lehrach H, Schlessinger D, **Willard HF** (1995). A 6 Mb YAC contig in Xp22.1-p22.2 spanning the DXS69E, XE59, GLRA2, PIGA, GRPR, CALB3, and PHKA2 genes. Genomics 25: 691-700.

172. Miller AP, Gustashaw K, Wolff DJ, Rider SH, Monaco AP, Eble B, Schlessinger D, Gorski JL, van Ommen GJ, Weissenbach J, **Willard HF** (1995). Three genes that escape X chromosome inactivation are clustered within a 6 Mb YAC contig and STS map in Xp11.21 - p11.22. Hum. Mol. Genet. 4: 731-739.

173. Hendrich BD, **Willard HF** (1995). Epigenetic regulation of gene expression: the effect of altered chromatin structure from yeast to mammals. Hum. Mol. Genet. 4: 1765-1777.

174. Kirchgessner CU, Warren ST, **Willard HF** (1995). X inactivation of the *FMR1* fragile X mental retardation gene. J. Med. Genet. 32: 925-929.

175. Warburton PE, **Willard HF** (1995). Interhomologue sequence variation of alpha satellite DNA from human chromosome 17: evidence for concerted evolution along haplotypic lineages. J. Mol. Evol. 41: 1006-1015.

176. Rupert JL, Brown CJ, **Willard HF** (1995). Direct detection of non-random X chromosome inactivation by use of a transcribed polymorphism in the *XIST* gene. Eur. J. Hum. Genet. 3: 333-343.

177. Wolff DJ, Miller AP, VanDyke DL, Schwartz S, **Willard HF** (1996). Molecular definition of breakpoints associated with human Xq isochromosomes: implications for mechanisms of formation. Am. J. Hum. Genet. 58: 154-160.

178. Coleman MP, Ambrose HJ, Carrel L, Nemeth AH, **Willard HF**, Davies KE (1996). A novel gene, DXS8237E, lies within 20 kb upstream of UBE1 in Xp11.23 and has a different X inactivation status. Genomics 31: 135-138.

179. Thibonnier M, Graves MK, Wagner MS, Auzan C, Clauser E, **Willard HF** (1996). Structure, sequence, expression, and chromosomal localization of the human V1a vasopressin receptor gene. Genomics 31: 327-334.

180. Carrel L, Clemson CM, Dunn JM, Miller AP, Hunt PA, Lawrence JB, **Willard HF** (1996). X inactivation analysis and DNA methylation studies of the ubiquitin activating enzyme E1 and PCTAIRE-1 genes in human and mouse. Hum. Mol. Genet. 5: 391-401.

181. Clemson CM, McNeil JA, **Willard HF**, Lawrence JB (1996). *XIST* RNA paints the inactive X chromosome at interphase: evidence for a novel RNA involved in nuclear/chromosome structure. J. Cell Biol. 132: 259-275.

182. Warburton PE, Haaf T, Gosden J, Lawson D, **Willard HF** (1996). Characterization of a chromosome-specific chimpanzee alpha satellite subset: evolutionary relationship to subsets on human chromosomes. Genomics 33: 220-228.

183. Carrel L, **Willard HF** (1996). An assay for X inactivation based on differential methylation at the fragile X locus, *FMR1*. Am. J. Med. Genet. 64: 27-30.

184. Naumova AK, Plenge RM, Bird LM, Leppert M, Morgan K, **Willard HF**, Sapienza C (1996). Heritability of X chromosome inactivation phenotype in a large family. Am. J. Hum. Genet. 58: 1111-1119.

185. Carrel L, Hunt PA, **Willard HF** (1996). Tissue- and lineage-specific variation in inactive X chromosome expression of the murine Smcx gene. Hum. Mol. Genet. 5: 1361-1366.

186. Harrington JJ, Van Bokkelen G, Mays RW, Gustashaw K, **Willard HF** (1997). Formation of de novo centromeres and construction of first-generation human artificial microchromosomes. Nature genetics 15: 345-355.

187. Brown CJ, Carrel L, **Willard HF** (1997). Expression of genes from the human active and inactive X chromosomes. Am. J. Hum. Genet. 60: 1333-1343.

188. Hendrich BD, Plenge RM, **Willard HF** (1997). Identification and characterization of the human *XIST* gene promoter: implications for models of X chromosome inactivation. Nucl. Acids Res. 25: 2661-2671.

189. Depinet TW, Zackowski JL, Earnshaw WC, Kaffe S, Sekhon GS, Stallard R, Sullivan BA, Vance GH, Van Dyke DL, **Willard HF**, Schwartz S (1997). Characterization of neo-centromeres in marker chromosomes lacking detectable alpha satellite DNA. Hum. Mol. Genet. 6: 1195-1204.

190. Ishikawa-Brush Y, Brush JF, Bolton P, Miller AP, Francis F, **Willard HF**, Lehrach H, Monaco AP (1997). Autism and multiple exostoses associated with an X;8 translocation occurring within the GRPR gene and 3' to the SDC2 gene. Hum. Mol. Genet. 6: 1241-1250.

191. Haaf T, **Willard HF** (1997). Chromosome-specific alpha satellite DNA from the centromere of chimpanzee chromosome 4. Chromosoma 106: 226-232.

192. Plenge RM, Hendrich BD, Schwartz C, Arena JF, Naumova A, Sapienza C, Winter RM, **Willard HF** (1997). A promoter mutation in the *XIST* gene in two unrelated families with skewed X chromosome inactivation. Nature genetics 17: 353-356.

193. Wolff DJ, Gustashaw KM, Zurcher V, Ko L, White W, Weiss L, Van Dyke DL, Schwartz S, **Willard HF** (1997). Deletions in Xq26.3-q27.3 including FMR1 result in a severe phenotype in a

male and variable phenotypes in females depending upon X inactivation pattern. Hum. Genet. 100: 256-262.

194. James RS, Dalton P, Gustashaw K, Wolff DJ, **Willard HF**, Mitchell C, Jacobs PA (1997). Molecular characterization of isochromosomes of Xq. Ann. Hum. Genet. 61: 485-490.

195. Dal Zotto L, Quaderi NA, Elliott R, Lingerfelter PA, Carrel L, Valsecchi V, Montini E, Yen CH, Chapman V, Kalcheva I, Arrigo G, Zuffardi O, Thomas S, **Willard HF**, Ballabio A, Disteche CM, Rugarli EI (1998). The mouse *Mid1* gene: implications for the pathogenesis of Opitz syndrome and the evolution of the mammalian pseudoautosomal region. Hum. Mol. Genet. 7: 489-499.

196. Mahtani MM, **Willard HF** (1998). Physical and genetic mapping of the human X chromosome centromere: repression of recombination. Genome Res. 8: 100-110.

197. Greenfield A, Carrel L, Pennis D, Philippe C, Quaderi N, Siggers P, Steiner K, Tam PPL, Monaco AP, **Willard HF**, Koopman P (1998). The *UTX* gene escapes X inactivation in mice and humans. Hum. Mol. Genet. 7: 737-742.

198. White WM, **Willard HF**, Van Dyke DL, Wolff DJ (1998). X inactivation spread into autosomal material of an X;autosome translocation: evidence for a difference between autosomal and X chromosomal DNA. Am. J. Hum. Genet. 63: 20-28.

199. Haaf T, **Willard HF** (1998). Orangutan alpha satellite monomers are closely related to the human consensus sequence. Mamm. Genome 9: 440-447.

200. Miller AP, **Willard HF** (1998). Chromosomal basis for X chromosome inactivation: identification of a multigene domain in Xp11.21-11.22 that escapes inactivation. Proc. Natl. Acad. Sci. USA 95: 8709-8714.

201. Sullivan BA, **Willard HF** (1998). Stable dicentric X chromosomes with two active centromeres. Nature Genet. 20: 227-228.

202. Plenge RM, Tranebjaerg L, Jensen PKA, Schwartz C, **Willard HF** (1999). Evidence that mutations in the X-linked *DDP* gene cause incompletely penetrant and variable skewed X inactivation. Am. J. Hum. Genet. 64: 759-767.

203. Carrel L, **Willard HF** (1999). Heterogeneous gene expression from the inactive X chromosome: an X-linked gene that escapes X inactivation in some human cell lines but is inactivated in others. Proc. Natl. Acad. Sci. USA 96: 7364-7369.

204. Higgins AW, Schueler MG, **Willard HF** (1999). Chromosome engineering: generation of mono- and dicentric isochromosomes in a somatic cell hybrid system. Chromosoma 108: 256-265.

205. Tentler D, Gustavsson P, Leisti J, Schueler M, Chelly J, Timonen E, Anneren G, **Willard HF**, Dahl N (1999). Deletion including the *oligophrenin-1* gene associated with enlarged cerebral ventricles, cerebellar hypoplasia, seizures and ataxia. Eur. J. Hum. Genet. 54: 1-8.

206. Carrel L, Cottle A, Goglin KC, **Willard HF** (1999). A first-generation X inactivation profile of the human X chromosome. Proc. Natl. Acad. Sci. USA 96: 14440-14444.

207. Plenge RM, Percec I, Nadeau JH, **Willard HF** (2000). Expression-based assay of an X-linked gene to examine effects of the X-controlling element (Xce) locus. Mammal. Genome 11: 405-408.

208. Schueler MG, Higgins AW, Nagaraja R, Tentler D, Dahl N, Gustashaw K, **Willard HF** (2000). Large-insert clone / STS contigs in Xq11-q12, spanning deletions in patients with androgen insensitivity and mental retardation. Genomics 66: 104-109.

209. Thibonnier M, Graves MK, Wagner MS, Chatelain N, Soubrier F, Corvol P, **Willard HF**, Jeunemaitre X (2000). Study of V1-vascular vasopressin receptor gene microsatellite polymorphisms in human essential hypertension. J. Mol. Cell. Cardio. 32: 557-564.

210. Tsuchiya KD, **Willard HF** (2000). Chromosomal domains and escape from X inactivation: comparative X inactivation analysis in mouse and human. Mammal. Genome 11: 849-854.

211. Chadwick BP, **Willard HF** (2001). A novel chromatin protein, distantly related to histone H2A, is largely excluded from the inactive X chromosome. J. Cell Biol. 152: 375-384.

212. Chadwick BP, **Willard HF** (2001). Histone H2A variants and the inactive X chromosome: identification of a second macroH2A variant. Hum. Mol. Genet. 10: 1101-1113.

213. Chadwick BP, Valley CM, **Willard HF** (2001). Histone variant macroH2A contains two distinct macrochromatin domains capable of directing macroH2A to the inactive X chromosome. Nucl. Acids Res. 29: 2699-2705.

214. Schueler MG, Higgins AW, Rudd MK, Gustashaw K, **Willard HF** (2001). Genomic and genetic definition of a functional human centromere. Science 294: 109-115.

215. Percec I, Plenge RM, Nadeau JH, Bartolomei MS, **Willard HF** (2002). Autosomal dominant mutations affecting X inactivation choice in the mouse. Science 296: 1136-1139.

216. Grimes BR, Rhoades AA, **Willard HF** (2002). Alpha satellite and vector composition influence rates of human artificial chromosome formation. Mol. Therapy 5: 798-805.

217. Plenge RM, Stevenson RA, Lubs HA, Schwartz CE, **Willard HF** (2002). Skewed X chromosome inactivation is a common feature of X-linked mental retardation disorders. Am. J. Hum. Genet. 71: 168-173.

218. Chadwick BP, **Willard HF** (2002). Cell cycle-dependent localization of macroH2A in chromatin of the inactive X chromosome. J. Cell Biol. 157: 1113-1123.

219. Hall LL, Byron M, Sakai K, Carrel L, **Willard HF**, Lawrence JB (2002). An ectopic human XIST gene can induce chromosome inactivation in postdifferentiation human HT1080 cells. Proc. Natl. Acad. Sci. USA 99: 8677-8692.

220. Percec I, Thorvaldsen JL, Plenge RM, Krapp CJ, Nadeau JH, **Willard HF**, Bartolomei MS (2003). An ENU mutagenesis screen for epigenetic mutations in the mouse. Genetics 164: 1481-1494.

221. Chadwick BP, **Willard HF** (2003). Chromatin of the Barr Body: Histone and non-histone proteins associated with or excluded from the inactive X chromosome. Hum. Mol. Genet. 12: 2167-2178.

222. Rudd MK, Mays RW, Schwartz S, **Willard HF** (2003). Human artificial chromosomes with alpha satellite-based de novo centromeres show increased frequency of nondisjunction and anaphase lag. Mol. Cell. Biol. 23: 7689-7697.

223. Chadwick BP, **Willard HF** (2003). Barring gene expression after XIST: maintaining facultative heterochromatin on the inactive X. Seminars Cell Dev. Biol. 14: 359-367.

224. Rudd MK, Schueler MG, **Willard HF** (2004). Characterization and functional annotation of human centromeres. Cold Spring Harb. Symp. Quant. Biol. 68: 141-150.

225. Rudd MK, **Willard HF** (2004). Analysis of the centromeric regions of the human genome assembly. Trends Genet. 20: 529-533.

226. Grimes BR, Babcock J, Rudd, MK, Chadwick B, **Willard HF** (2004). Assembly and characterization of heterochromatin and euchromatin on human artificial chromosomes. Genome Biol. 5: R89.1 – R89.14.

227. Chadwick BP, **Willard HF** (2004). Multiple spatially distinct types of facultative heterochromatin on the human inactive X chromosome. Proc. Natl. Acad. Sci. USA 101: 17450-17455.

228. Basu J, Stromberg G, Compitello G, **Willard HF**, Van Bokkelen G (2005). Rapid creation of BAC-based human artificial chromosome vectors by transposition with synthetic alpha satellite arrays. Nucl. Acids Res. 33: 587-596.

229. Ross MT et al. (with ~250 authors including Schueler MG, Carrel L, and **Willard HF**) (2005). Sequence of the human X chromosome. Nature 434: 325-337.

230. Carrel L, **Willard HF** (2005). X inactivation profile reveals extensive variability in X-linked gene expression in females. Nature 434: 400-404.

231. Basu J, **Willard HF** (2005). Artificial and engineered chromosomes: non-integrating vectors for gene therapy. Trends Mol. Med. 11: 251-258.

232. **Willard HF**, Angrist M, Ginsburg GS (2005). Genomic medicine: genetic variation and its impact on the future of health care. Phil. Trans. R. Soc. Lond. B 360: 1543-1550.

233. Schueler MG, Dunn JM, Bird CP, Ross MT, Viggiano L, Rocchi M, **Willard HF**, Green ED (2005). Progressive proximal expansion of the primate X chromosome centromere. Proc. Natl. Acad. Sci. USA 102: 10563-10568.

234. Basu J, Compitello G, Stromberg G, **Willard HF**, Van Bokkelen G (2005). Efficient assembly of de novo human artificial chromosomes from large genomic loci. BMC Biotech. 5: 21.

235. Chadwick LH, **Willard HF** (2005). Genetic and parent-of-origin influences on X chromosome choice in *Xce* heterozygous mice. Mammal. Genome 16: 691-699.

236. Higgins AW, Gustashaw KM, **Willard HF** (2005). Engineered human dicentric chromosomes show centromere plasticity. Chromosome Res. 13: 745-762.

237. Rudd MK, Wray GA, **Willard HF** (2006). The evolutionary dynamics of alpha satellite. Genome Res. 16: 88-96.

238. Scott KC, Merrett SL, **Willard HF** (2006). A heterochromatin barrier partitions the fission yeast centromere into discrete chromatin domains. Current Biol. 16: 119-129.

239. Chadwick LH, Pertz LM, Broman KW, Bartolomei MS, **Willard HF** (2006). Genetic control of X chromosome inactivation in mice: definition of the *Xce* candidate interval. Genetics 173: 2103-2110.

240. Valley CM, **Willard HF** (2006). Genomic and epigenomic approaches to the study of X chromosome inactivation. Curr. Opin. Genet. Dev. 16: 240-245.

241. Valley CM, Pertz LM, Balakumaran BS, **Willard HF** (2006). Chromosome-wide, allele-specific analysis of the histone code on the human X chromosome. Hum. Mol. Genet. 15: 2335-2347.

242. Amos-Landgraf JM, Cottle A, Plenge RM, Friez M, Schwartz CE, Longshore J, **Willard HF** (2006). X chromosome inactivation patterns of 1005 phenotypically normal females. Am. J. Hum. Genet. 79:493-499.

243. Wang Z, **Willard HF**, Mukherjee S, Furey TS (2006). Evidence of influence of genomic DNA sequences on human X chromosome inactivation. PLoS Comp. Biol. 2: e113, 979-988.

244. Haga SB, **Willard HF** (2006). Defining the spectrum of genome policy. Nature Rev. Genet. 7: 966-972.

245. Horvath JE, **Willard HF** (2007). Primate comparative genomics: lemur biology and evolution. Trends Genet. 23:173-182.

246. Scott KC, White CV, **Willard HF** (2007). An RNA polymerase III-dependent heterochromatin barrier at fission yeast centromere 1. PLoS ONE 10: e1099.

247. Horvath JE, Weisrock DW, Embry SL, Foirentino I, Balhoff JP, Kappeler P, Wray GA, **Willard HF**, Yoder AD (2008). Development and application of a phylogenomic toolkit: resolving the evolutionary history of Madagascar's lemurs. Genome Res. 18: 489-499.

248. Kawamoto K, Lobach DF, **Willard HF**, Ginsburg GS (2009). A national clinical decision support infrastructure to enable the widespread and consistent practice of genomic and personalized medicine. BMC Med. Inform. Decis. Making 9: 17.1-14.

249. Wheeler B, Blau J, **Willard HF**, Scott KC (2009). Impact of local genome sequence on definition of heterochromatin domains. PLoS Genet. 5: e1000453.1-15.

250. Ginsburg GS, **Willard HF** (2009). Genomic and personalized medicine: foundations and applications. Transl. Res. 154: 277-287.

251. McDaniell R, Lee B, Song L, Liu Z, Boyle AP, Erdos MR, Scott LJ, Morken MA, Kucera KS, Battenhouse A, Keefe D, Collins FS, **Willard HF**, Lieb JD, Furey TS, Crawford GE, Iyer VR, Birney E (2010). Heritable individual-specific and allele-specific chromatin signatures in humans. Science 328: 235-239.

252. O'Daniel JM, Haga SB, **Willard HF** (2010). Considerations for the impact of personal genome information: a study of genomic profiling among genetics and genomics professionals. J. Genet. Couns. 19: 387-401.

253. Horvath JE, Sheedy CB, Merrett SL, Biallo AB, Swofford DL, NISC Comparative Sequencing Program, Green ED, **Willard HF** (2011). Comparative analysis of the primate X inactivation center region and reconstruction of the ancestral primate XIST locus. Genome Res. 21: 850-862.

254. Gertz J, Varley KE, Reddy TE, Bowling KM, Pauli F, Parker SL, Kucera KS, **Willard HF**, Myers RM (2011). Analysis of DNA methylation in a three-generation family reveals widespread genetic influence on epigenetic regulation. PLoS Genet. 7, e1002228.

255. Kucera KS, Reddy TE, Pauli F, Gertz J, Logan JE, Myers RM, **Willard HF** (2011). Allele-specific distribution of RNA polymerase II on female X chromosomes. Hum. Mol. Genet. 20: 3964-3973.

256. Lee H-R, Hayden KE, **Willard HF** (2011). Organization and molecular evolution of CENP-A-associated satellite DNA families in a basal primate genome. Genome Biol. Evol. 3: 1136-1149.

257. Wheeler BS, Ruderman BT, **Willard HF,** Scott KC (2012). Uncoupling of genomic and epigenetic signals in the maintenance and inheritance of heterochromatin domains. Genetics 190: 549-557.

258. Reddy TE, Gertz J, Pauli F, Kucera KS, Newberry KM, Marinov GK, Mortazavi A, Williams BA, Wold BJ, **Willard HF**, Myers RM (2012). Effects of sequence variation on differential allelic transcription factor occupancy and gene expression. Genome Res. 22: 860-869.

259. Hayden KE, **Willard HF** (2012). Composition and organization of active centromere sequences in complex genomes. BMC Genomics 13: 324.

260. Wang Z, **Willard HF** (2012). Evidence for sequence biases associated with patterns of histone methylation. BMC Genomics 13: 367.

261. Thorvaldsen JL, Krapp C, **Willard HF**, Bartolomei MS (2012). Multiple closely linked regions on the murine X chromosome affect nonrandom X chromosome inactivation. Genetics 192: 1095-1107.

262. Hayden KE, Strome ED, Merrett SE, Lee H-R, Rudd MK, **Willard HF** (2013). Sequences associated with centromere competency in the human genome. Mol. Cell. Biol. 33: 763-772.

263. Haga SB, Barry WT, Mills R, Ginsburg GS, Svetkey L, Sullivan J, **Willard HF** (2013). Public knowledge of and attitudes toward genetics and genetic testing. Genet. Test. Mol. Biomarkers 17: 327-335.

264. Haga SB, Barry WT, Mills R, Svetkey L, Suchindran S, **Willard HF**, Ginsburg GS (2014). Impact of delivery models on understanding genomic risk for type 2 diabetes. Public Health Genomics 17: 95-104.

265. Miga KH, Newton Y, Jain M, Altemose N, **Willard HF**, Kent WJ (2014). Centromere reference models for human chromosomes X and Y satellite arrays. Genome Res. 24: 697-707.

266. Brownstein CA, Beggs AH…**Willard HF**...Kohane IS, Margulies SM (2014). An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY challenge. Genome Biol 15: R53.

267. Altemose N, Miga KH, Maggioni M, **Willard HF** (2014). Genomic characterization of large heterochromatic gaps in the human genome assembly. PLoS Comp. Biol. 10: e1003628.

268. Carman AJ, Ferguson R, Cantu R,…(+21 authors)…**Willard H**, Fillit HM (2015). Mind the gaps – advancing research into short-term and long-term neuropsychological outcomes of youth sports-related concussions. Nature Rev. Neurol. 11: 230-244.

269. Jain M, Olsen HE, Turner DJ, Stoddart D, Bulazel KV, Paten B, Haussler D, **Willard HF**, Akeson M, Miga KH (2017). Linear assembly of a human Y centromere using Nanopore long reads. Nature Biotech. (in press).

Chapters in Books:

270. **Willard HF**, Rosenberg LE (1979). Inherited deficiencies of methylmalonyl CoA mutase activity: biochemical and genetic studies in cultured skin fibroblasts. In: *Models for the Study of Inborn Errors of Metabolism* (F.A. Hommes, ed.) Elsevier/North-Holland Biomedical Press, pp. 297-310.
271. **Willard HF** (1983). Replication of human X chromosomes in cultured fibroblasts and in somatic cell hybrids: Cytogenetic analysis and a molecular perspective. In: *Cytogenetics of Mammalian X Chromosomes* (A.A. Sandberg, ed.). Alan R. Liss, Inc., New York, pp. 427-448.
272. **Willard HF** (1983). Molecular and cytogenetic mapping studies of mammalian X chromosomes: Ohno's hypothesis revisited. In: *Cytogenetics of Mammalian X Chromosomes* (A.A. Sandberg, ed.). Alan R. Liss, Inc., New York, pp. 463-478.
273. **Willard HF** (1985). Molecular organization of repeated DNA sequences on the human Y chromosome. In: *The Y Chromosome*, (A.A. Sandberg, ed.), Alan R. Liss Inc., New York, pp. 125-140.
274. O'Dowd BF, Klavins MH, **Willard HF**, Gravel R, Lowden JA, Mahuran DJ (1986). Molecular heterogeneity in O-variant GM2 gangliosidosis. In: *Enzymes of Lipid Metabolism* (R. Massarelli and S. Gatt, eds.), Plenum Publishing Corp., NY, pp.779-784.
275. **Willard HF**, Wevrick R, Warburton PE (1989). Human centromere structure: organization and potential role of alpha satellite DNA. In: *Aneuploidy: Mechanisms of Origin* (MA Resnick, ed.). Alan R. Liss, Inc., New York, pp. 9-18.
276. **Willard HF** (1989). The genomics of long tandem arrays of satellite DNA in the human genome. In: Proceedings of the Sixteenth International Congress of Genetics. Genome 31:737-744.
277. **Willard HF** (1990). Molecular cytogenetics of centromeres of human chromosomes. In*: Chromosomes Today*, vol. 10:47-60 (Fredga K, ed.), Unwin Hyman, London.
278. **Willard HF** (1990). Alpha and beta satellite sequences on chromosome 21: the possible role of centromere and chromosome structure in nondisjunction. In: *The Molecular Genetic Analysis of Chromosome 21* (Epstein CJ, Patterson D, eds.), Alan R. Liss, New York, pp. 39-52.

279. **Willard HF** (1994). Human genetics. In: *Introduction to Molecular Medicine*, Leder P, Clayton D, Rubinstein E (eds). New York: Scientific American Medicine, pp. 1-26.
280. **Willard HF** (1995). Sex chromosomes and X chromosome inactivation, In: *The Metabolic and Molecular Bases of Inherited Disease*, 7th ed., Scriver CR, Beaudet AL, Sly WS, Valle D (eds). New York: McGraw-Hill Publishing Co., pp. 719-737.
281. Pyeritz RE, Fearon ER, Seidman C, **Willard HF** (1995). Genetics in clinical medicine, In: *MKSAP 10*, Goldman L (ed). Philadelphia: American College of Physicians, pp. 1113-1150.
282. Tilghman SM, **Willard HF** (1995). Epigenetic regulation in mammals. In: *Chromatin Structure and Gene Expression* (Elgin S, ed.). Oxford: Oxford Univ. Press, pp. 197-222.
283. Warburton PE, **Willard HF** (1996). Evolution of centromeric alpha satellite DNA: molecular organization within and between human and primate chromosomes. In: *Human Genome Evolution*, Jackson M, Strachan T, Dover G, eds. Oxford: BIOS Scientific Publishers, pp. 121-145.
284. **Willard HF** (1997). X chromosome inactivation. In: *Encyclopedia of Human Biology*, 2nd edition, vol. 8. Academic Press, New York, pp. 785-789.
285. **Willard HF** (2000). The sex chromosomes and X chromosome inactivation. In: *The Metabolic and Molecular Bases of Inherited Disease*, 8th ed. Edited by Scriver CR, Beaudet AL, Sly WS, Valle D, Childs B, Vogelstein B. New York: McGraw-Hill, pp. 1191-1221.
286. **Willard HF** (2000). Chromonomics: structure and function of natural and artificial human chromosomes. In: *Current Options for the Human Genome Project*. Edited by S. Grisolia. Madrid: Fundacion BBV, pp. 253-262.
287. **Willard HF**, Rudd MK (2006). Genomic organization and function of human centromeres. In: *Genomic Disorders: The Genomic Basis of Disease*. Edited by Stankiewicz P, Lupski J., pp. 115-129.
288. Basu J, **Willard HF** (2006). Human artificial chromosomes: potential applications and clinical considerations. Ped. Clinics N. America 53: 843-853.
289. Basu J, **Willard HF** (2007). Human artificial chromosome assembly by transposon-based retrofitting of genomic BACs with synthetic alpha satellite arrays. Curr Protoc Hum Genet 5: unit 5.18.
290. **Willard HF** (2008). Organization, variation and expression of the human genome as a foundation of genomic and personalized medicine. In: *Genomic and Personalized Medicine*. Edited by H.F. Willard, G. Ginsburg. New York: Elsevier, pp. 4-21.
291. Ginsburg GS, **Willard HF** (2009). The foundations of genomic and personalized medicine. In: *Essentials of Genomic and Personalized Medicine*. Edited by G.S. Ginsburg, H.F. Willard. New York: Elsevier, pp. 1-10.
292. **Willard HF** (2009). Organization, variation and expression of the human genome. In: *Essentials of Genomic and Personalized Medicine*. Edited by G.S. Ginsburg, H.F. Willard. New York: Elsevier, pp. 13-25.
293. **Willard HF** (2012). The human genome: a window on human genetics, biology and medicine. In: *Genomic and Personalized Medicine, 2nd edition*. Edited by G.S. Ginsburg, H.F. Willard. New York: Elsevier, pp. 4-27.

Other Reviews and Commentaries:

294. Tsui L-C, Zengerling S, **Willard HF**, Buchwald M (1987). Mapping of the cystic fibrosis locus on chromosome 7. Cold Spring Harb. Symp. Quant. Biol. 51:325-336.
295. **Willard HF** (1991). Evolution of alpha satellite. Curr. Opin. Genet. Dev. 1:509-514.
296. Ballabio A, **Willard HF** (1992). Mammalian X chromosome inactivation and the *XIST* gene. Curr. Opin. Genet. Dev. 2:439-447.

297. Brown CJ, **Willard HF** (1993). Molecular and genetic studies of human X chromosome inactivation. Adv. Dev. Biol. 2: 37-72.

298. Tyler-Smith C, **Willard HF** (1993). Mammalian chromosome structure. Curr. Opin. Genet. Dev. 3: 390-397.

299. **Willard HF**, Brown CJ, Carrel L, Hendrich B, Miller AP (1993). Epigenetic and chromosomal control of gene expression: molecular and genetic analysis of X chromosome inactivation. Cold Spring Harbor Symp. Quant. Biol. 58: 315-322.

300. **Willard HF** (1996). X chromosome inactivation and X-linked mental retardation. Am. J. Med. Genet. 64: 21-26.

301. **Willard HF** (1996). Chromosome manipulation: a systematic approach toward understanding human chromosome structure and function. Proc. Natl. Acad. Sci. USA 93: 6847-6850.

302. **Willard HF** (1996). X chromosome inactivation, *XIST*, and pursuit of the X inactivation center. Cell 86: 5-7.

303. Sullivan BA, Schwartz S, **Willard HF** (1996). Centromeres of human chromosomes. Envir. Mol. Mutagen. 28: 182-191.

304. **Willard HF**, Salz HK (1997). Dosage compensation: remodeling chromatin with RNA. Nature 386: 228-229.

305. Puck JM, **Willard HF** (1998). X inactivation in females with X-linked disease. New Engl. J. Med. 338: 291-295.

306. **Willard HF** (1998). Centromeres: the 'missing link' in the development of human artificial chromosomes. Curr. Opin. Genet. Dev. 8: 219-225.

307. **Willard HF** (1998). Human artificial chromosomes coming into focus. Nature Biotechnol. 16: 415-416.

308. Carrel L, **Willard HF** (1998). Counting on Xist [News & Views]. Nature Genet. 19: 211-212.

309. **Willard HF**, Hendrich BD (1999). Breaking the silence in Rett syndrome [News & Views]. Nature Genet. 23: 127-128.

310. **Willard HF** (2000). Artificial chromosomes coming to life [Perspectives]. Science 290: 1308-1309.

311. **Willard HF**, Carrel L (2001). Making sense (and antisense) of the X inactivation center [Commentary]. Proc. Natl. Acad. Sci. USA 98: 10025-10027.

312. **Willard HF** (2001). Neocentromeres and human artificial chromosomes: an unnatural act [Commentary]. Proc. Natl. Acad. Sci. USA 98: 5374-5376.

313. **Willard HF** (2001). You say tomato and I say tomahto: human genetics and gene therapy. Mol. Therapy 4: 514

314. **Willard HF** (2002). ASHG Presidential Address – On black boxes and storytellers: lessons learned in human genetics. Am. J. Hum. Genet. 70: 285-296.

315. Chadwick BP, **Willard HF** (2003). SETting the stage: Eed-Enx1 leaves an epigenetic signature on the inactive X chromosome. Dev. Cell 4: 445-447.

316. **Willard HF** (2003). Genome biology: Tales of the Y chromosome [News & Views]. Nature 423: 810-812.

317. Shianna KV, **Willard HF** (2006). Human genomics: in search of normality [News & Views]. Nature 444: 428-429.

318. **Willard HF** (2007). Honest Jim talks manners [review of *Avoid Boring People* by James D. Watson]. Nature 449: 787.

319. Haga SB, **Willard HF** (2008). Letting the genome out of the bottle. New Engl. J. Med. 358: 2184-2185.

320. **Willard HF** (2010). Allan Award Address – Life in the Sandbox: unfinished business. Am. J. Hum. Genet. 86: 318-327.

321. Anderson WA et al. (including **Willard HF**) (2011). Competencies: A cure for the pre-med curriculum. Science 334: 760-761.

322. Earnshaw, WC et al. (including **Willard HF**) (2013). Esperanto for histones: CENP-A, not CenH3, is the centromeric histone H3 variant. Chromosome Res. 21: 101-106.

Public Opinion / Editorials (Op-Eds):

Willard HF. "Hollywood Gives Real Scientists a Bad Rep"
The Herald Sun (Durham, NC*)*, May 24, 2004
Willard HF. "The Way to Double Our Bet"
News & Observer (Raleigh, NC), June 4, 2004
Willard HF. "Evolution Sticker Shock"
The Seattle Times and *News & Observer*, December 16, 2004
Goldstein D, **Willard HF**. "Dr. King's Legacy in Our Genomic Future"
The Boston Globe and *The Contra Costa Times*, January 17, 2005
Willard HF. "The Race Against Gene Doping"
The Orlando Sentinel and *The Newark Star-Ledger*, July 13, 2005
The Boston Globe and *News & Observer*, July 14, 2005
Haga S, **Willard HF**. "Act Now to Prevent Genetic Discrimination"
The Washington Post, December 28, 2005
Goldstein DB, **Willard HF**. "Is she just what Japan needs?"
News & Observer, April 21, 2006
Willard HF. "What a pig! – after a new genome"
News & Observer, May 26, 2006
Willard HF. "Genes and a Sporting Chance"
News & Observer, July 11, 2010

Patents:

U.S Patent No. 5,695,967
December 9, 1997
Method for stably cloning large repeating units of DNA
Van Bokkelen G, Harrington JJ, Willard HF

U.S Patent No. 5,869,294
February 9, 1999
Method for stably cloning large repeating DNA sequences
Harrington JJ, Van Bokkelen G, Willard HF

U.S. Patent No. 6,348,353
February 19, 2002
Artificial mammalian chromosome
Harrington JJ, Van Bokkelen G, Willard HF

Teaching

Undergraduate

1985 – 1987	MGB 470H - "Cellular and Molecular Mammalian Genetics", Faculty of Arts and Science, University of Toronto (6-10 hrs)
1987 – 1989	MGB 470H - "Molecular Genetics of Complex Genomes", Faculty of Arts and Science, University of Toronto (26 hrs)
1993 – 1996	BIO 214 - "Genetics", School of Arts and Sciences, Case Western Reserve University (2 hrs)
2004 – 2014	BIO 195S/194S/210S – “Genomes, Biology and Medicine”, Duke University (40 hrs)
2007 – 2009, 2014	BIO 289S/410S – “Advanced Topics in Genome Sciences Research,” Duke University (40 hrs)
2009, 2010	GENOME 148 – “Genetics, Genomics and Society”, Duke University (40 hrs)
2009 – 2013	GENOME 198S – “Genome Sciences & Policy Capstone Course”, Duke University (40 hrs)

Graduate

1982	MBP 1012-1014L - 'Molecular Organization of the Human Genome', Department of Molecular Genetics, University of Toronto (12 hrs)
1987	MBP 1014L - 'Human Molecular Genetics', Department of Molecular Genetics, University of Toronto (6 hrs of 24 hr course, with Drs. R. Worton, L. Tsui and D. Cox)
1991	GEN 210 - 'Advanced Human Genetics', Department of Genetics, Stanford University (with Dr. D. Botstein)
1992 – 2002	GENE 500/504 - 'Advanced Eukaryotic Genetics', Department of Genetics, Case Western Reserve University (with other faculty)

1995, 1997	GENE 515 - 'Chromosome Structure and Function', Department of Genetics, Case Western Reserve University
1996, 1998, 2000	GENE 510 - 'Advanced Human Genetics', Department of Genetics, Case Western Reserve University
1997, 1998	C3MB - 'Cell and Molecular Biology', Human Genetics Section, Case Western Reserve University (with Dr. M. Warman)
2002	GENE 512 – 'Analysis of Complex Genomes', Department of Genetics, Case Western Reserve University
2005 – 2006	MGM 300 – 'Biological Regulatory Mechanisms', Department of Molecular Genetics & Microbiology, Duke University (6 hrs of 30 hr course)
2006 – 2007	UPGEN 301 – 'Advanced Topics in Genetics/Genomics', University Program in Genetics and Genomics, Duke University (15 hrs)

Medical Students

1983 - 1988	Course Director, Genetics for Second Year Medical Students (30 hrs), Faculty of Medicine, University of Toronto
1982 - 1988	Lecturing in Genetics for Second Year Medical Students, University of Toronto (10-18 hrs)
1989 - 1992	Lecturing in Genetics 201, Stanford University (4 - 10 hrs)
1990 - 1992	Course Director, Genetics 201 (36 hrs), Stanford University
1992 - 1996	Lecturing in First Year Committee course for medical students, Case Western Reserve University School of Medicine (~5 hrs)
2004 - 2007	Lecturing in First Year "Molecules and Cells" segment, Duke University School of Medicine (4 hrs)

Graduate Students Supervised

University of Toronto

1982 - 1985	Leslie A. Creeper, M.Sc. Thesis: *Functional analysis of a mammalian chromosomal origin in yeast*
1984 - 1987	John S. Waye, Ph.D. Thesis: *Organization and evolution of chromosome-specific human alpha satellite DNA*
1984 - 1990	Sharon J. Durfy, Ph.D. Thesis: *Nucleotide sequence variation, homogenization, and evolution of X chromosome alpha satellite DNA*
1987 - 1990	Carolyn J. Brown, Ph.D. Thesis: *Studies of human X chromosome inactivation*
1987 - 1992	Rachel Wevrick, Ph.D. Thesis: *Organization and behavior of alpha satellite DNA at the centromeres of human chromosomes*
1986 - 1993	Peter E. Warburton, Ph.D. Thesis: *Evolution of tandemly repeated DNA: repeat unit variation of human alpha satellite DNA*

Stanford University

1988 - 1993	Melanie M. Mahtani, Ph. D. Thesis: *Physical and genetic mapping studies of the human X chromosome: repression of recombination at the centromere*
1990 - 1995	Laura Carrel, Ph.D. Thesis: *Developmental and chromosomal basis of gene regulation on the mammalian X chromosome*
1991 - 1995	Brian Hendrich, Ph.D. Thesis: *Characterization of the human XIST gene and its promoter: implications for mammalian X chromosome inactivation*
1991 - 1995	Andrew Miller, Ph.D. Thesis: *Physical mapping studies and X inactivation analysis on the short arm of the human X chromosome*

Case Western Reserve University

1994 – 1998	Robert M. Plenge, M.D., Ph.D. Thesis: *Genetic control of skewed X chromosome inactivation*
1995 – 2000	Anne W. Higgins, Ph.D. Thesis: *Centromere structure and behavior: insight from engineered dicentric chromosomes*
1996 – 2001	Mary G. Schueler, Ph.D. Thesis: *Structure and evolution of the pericentromeric region of the human X chromosome*
1998 – 2002	Ivona Percec, M.D., Ph.D. [jointly with University of Pennsylvania; Marisa Bartolomei, advisor] Thesis: *Genetic dissection reveals multiple autosomal elements involved in X chromosome inactivation in the mouse*
1997 – 2004	James Amos-Landgraf, Ph.D. Thesis: *A human population study of the genetic control of X inactivation*
1999 – 2004	M. Katharine Rudd, Ph.D. Thesis: *Organization, evolution and function of alpha satellite DNA at human centromeres*
1999 – 2005	Lisa Helbling Chadwick, Ph.D. Thesis: *Genetic control of X chromosome choice in X inactivation*

Duke University

2000 – 2007	Cory Valley, Ph.D. Thesis: *Genomic analysis of chromatin organization on mammalian X chromosomes*
2005 – 2010	Bayly Wheeler, Ph.D. Thesis: *Role of genomic sequence in the spatial and temporal propagation of heterochromatin in fission yeast*
2006 – 2011	Katerina S. Kucera, Ph.D. Thesis: *States of allelic imbalance on the X chromosomes in human females*

2005 – 2011	Karen Hayden, Ph.D. Thesis: *A genomic definition of centromeres in complex genomes*
2006 – 2012	Christina Sheedy, Ph.D. Thesis: *Genomic characterization and comparative analysis of the Xce candidate region in mouse*

Postdoctoral Fellows / Research Associates / Senior Research Associates

1988 - 1991	Cecil B. Sharp, D.M.D., Ph.D.
1990 - 1992	Thomas Haaf, M.D., Ph.D.
1991 - 1994	Tiina Alitalo, Ph.D.
1991 - 1994	Carolyn J. Brown, Ph.D.
1991 - 1992	Cordula Kirchgessner, Ph.D.
1992 - 1994	Daynna Wolff, Ph.D.
1994 - 1997	John Harrington, Ph.D.
1994 - 1996	Gil van Bokkelen, Ph.D.
1994 - 1998	Kosuke Sakai, Ph.D.
1995 - 1996	Beth Sullivan, Ph.D.
1995 - 1999	Laura Carrel, Ph.D.
1996 - 1998	R. Willie Mays, Ph.D.
1996 – 2000	Karen Tsuchiya, M.D.
1998 – 2001	Christie Gunter, Ph.D.
1999 – 2003	Brian P. Chadwick, Ph.D.
1999 – 2003	Brenda Grimes, Ph.D.
2000 – 2005	Satkunanathan (Bala) Balakumaran, Ph.D.
2004 – 2007	Zhong Wang, Ph.D.
2000 – 2008	Kristin C. Scott, Ph.D.
2004 – 2008	Joydeep Basu, Ph.D.
2004 – 2008	Julie Horvath, Ph.D.
2008 – 2010	Kira Bulazel, Ph.D.
2008 – 2011	Erin Strome, Ph.D.
2008 – 2011	Hye-Ran Lee, PhD.

Areas of research interests:

Genetics	Genome Biology
Genomics and Precision Health	Epigenetics
Genetics, Genomics and Society	Chromosome Structure and Function