

CURRICULUM VITAE

NAME: DAVID H. LEDBETTER, Ph.D., FACMG

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BIRTH DATE AND PLACE: July 10, 1953, Lakehurst, NJ

CITIZENSHIP: United States

CURRENT TITLE:
2010- present Executive Vice President and
Chief Scientific Officer
Geisinger Health System

PREVIOUS ACADEMIC AND PROFESSIONAL EXPERIENCE:

- 1978-1981 Visiting Predoctoral Fellow (cytogenetics training with Dr. T.C. Hsu),
The University of Texas System Cancer Center, M.D. Anderson Hospital
and Tumor Institute, Houston, TX
- 1981-1993 Assistant Professor (1981), Associate Professor (with tenure, 1986), Full Professor (with
tenure, 1990), and Director, Kleberg Cytogenetics and Prenatal Diagnosis Laboratory,
Department of Human and Molecular Genetics, Baylor College of Medicine, Houston, TX
- 1993-1996 Member, Senior Executive Service (SES6) of the United States of America, and
Chief, Diagnostic Development Branch, National Center for Human Genome Research,
National Institutes of Health, Bethesda, MD.
- 1996-2003 Marjorie I. and Bernard A. Mitchell Professor and Founding Chair,
Department of Human Genetics, The University of Chicago.
- 2003-2010 Robert W. Woodruff Professor of Human Genetics and Director, Division of Medical
Genetics, Department of Human Genetics, Emory University School of Medicine,
Atlanta, GA

PREVIOUS LEADERSHIP EXPERIENCE

1993-1996 Branch Chief (equivalent to Department Chair in academic structure), Diagnostic Development Branch, National Center for Human Genome Research (now National Human Genome Research Institute), National Institutes of Health.

Recruited by Francis Collins, M.D., Ph.D. as a Founding Branch Chief (along with Robert Nussbaum, M.D.) to design and initiate the Intramural Research program at NCHGR to include 6 Branches and Laboratories. Faculty recruitments to the Diagnostic Development Branch included Thomas Ried, M.D., Mark Hughes, M.D., Ph.D., and Eric Green, M.D., Ph.D. (now Director, National Human Genome Research Institute).

1996-2003 Founding Chair, Department of Human Genetics (clinical and basic science department), University of Chicago.

Recruited by Dr. Glenn Steele, Dean of Biological Sciences and the Pritzker School of Medicine, to initiate a new department to integrate and enhance human genetics research and clinical activities. Grew from 4 faculty members (including Nancy Cox and Carole Ober), to 12 faculty (including Bill Dobyns, Bruce Lahn, and Jonathon Pritchard) with the highest percentage faculty salary recovery from external grants and the highest ratio of grant dollars per square foot of research space of any basic science or clinical department in the medical school.

2003-2010 Director, Division of Medical Genetics, Department of Human Genetics, Emory University School of Medicine.

Recruited by Dr. Steve Warren to transform a moderate sized clinical division into a strong academic program of translational research while providing outstanding clinical care to genetics patients and in genetic diagnostic laboratory services. Successfully recruited 10 new faculty members with strong clinical and translational research programs or potential, increasing grant and contract portfolio to over \$5 M/year and clinical revenues to over \$10 M/year. The full service genetics diagnostics laboratories of Emory Genetics (including biochemical genetics, cytogenetics and molecular genetics) are widely recognized as one of the top 3 academically-based programs in the country (with Baylor College of Medicine and the Mayo Clinic).

LICENSURE/BOARDS:

1984 Clinical Cytogeneticist (#1745), American Board of Medical Genetics

EDUCATION:

1971-1975 B.S., magna cum laude (Psychology), Tulane University, New Orleans, LA

1975-1981 Ph.D. (Behavioral Genetics-Psychology), The University of Texas, Austin, TX

GOVERNMENT SERVICE:

1993-1996 Member, Senior Executive Service (SES6) of the United States of America

COMMITTEE MEMBERSHIPS:

1985- Scientific Advisory Board, Prader-Willi Syndrome Association (USA), St. Louis, MO

1991-1993 Member and Chairman (1993), NIGMS Human Genetic Mutant Cell Repository Working Group, NIH

1990-1993 Charter Member, Genome Research Review Committee, National Center for Human Genome Research (NCHGR), NIH

1991-1992 Member, Scientific Advisory Board, Oncor, Inc., Gaithersburg, MD

1993-2003 Scientific Director, The Lissencephaly Network, Inc., Fort Wayne, IN

1995-2000 Member, Basil O'Connor Starter Research Grant Review Committee, March of Dimes

1995-2007 Member and Chair, Scientific Advisory Board, Vysis, Inc., Downer's Grove, IL

1996-2006 Member, International Standing Committee on Human Cytogenetic Nomenclature

1996-1998 Member, Mammalian Genetics Study Section, Division of Research Grants, National Institutes of Health

CONSULTANTSHIPS:

1990-1992 Consultant, Oncor, Inc., Gaithersburg, MD

1993-1997 Consultant, Genzyme Genetics, Inc., Framingham, MA

1995- 2005 Consultant and Chair, Scientific Advisory Board, Vysis, Inc., Downer's Grove, IL

2002- 2010 Consultant and Chair, Scientific Advisory Board, Geisinger Health Systems, Danville, PA

2010-2011 Consultant, GeneDx (BioReference Laboratories), Gaithersburg, MD

2010- present Consultant, Roche Nimblegen, Madison, WI

2010- present Consultant, Celula, Inc., La Jolla, CA

2010- present Consultant, Combimatrix, Inc., Irvine, CA

EDITORIAL BOARDS:

- 1989-1992 American Journal of Human Genetics
- 1992-1994 Genomics
- 1993-2001 Human Molecular Genetics
- 1995-1998 American Journal of Medical Genetics
- 2003-present Prenatal Diagnosis

HONORS AND AWARDS:

- 1975 Phi Beta Kappa
 The Aaron Hartman Prize for Psychology, Tulane University
- 1995 The 12th Carter Lecturer and Medal Recipient,
 British Clinical Genetics Society
- 1998 William C. and Ruth P. Griffith Visiting Professor and Lecturer
 Indiana University School of Medicine
- 2003 Named by the Institute for Scientific Information (ISI) as a
 “Highly Cited Researcher in the field of Molecular Biology and Genetics”

SOCIETY MEMBERSHIPS:

- 1979- American Society of Human Genetics (ASHG)
 Program Committee (1990-1992)
 Board of Directors (1992-1994)
- 1979- American Association for the Advancement of Science
- 1979- Association of Cytogenetics Technologists
- 1989- Human Genome Organization (HUGO)
 Board of Trustees, HUGO Americas (2001-2004)
- 1993- American College of Medical Genetics (ACMG), Founding Fellow
 Board of Directors (1999-2004)
 Laboratory Practice Committee (1999-2004)
 Chair, Intellectual Property Committee (2008-present)
- 1994-2000 ASHG-ACMG Test and Technology Transfer Committee

RESEARCH FOCUS:

Research is focused on autism and other developmental disorders, particularly the identification of specific genetic etiologies (genes and copy number variants) and technology development for prevention, early diagnosis and early intervention. Broader research and policy interests include personalized and genomic medicine, genetic testing, gene patents and licensing, newborn screening.

GRANT SUPPORT (CURRENT):

2R01 MH074090-06 (D.H. Ledbetter-PI, 20% effort) 07/15/10-07/14/15
NIH/NIMH “Gene Dosage Imbalance in Neurodevelopmental Disorders”
\$449,174 (annual d.c.)

This project is aimed at identification of human genes involved in developmental brain disorders. Specifically, we are using whole genome cytogenomic array analysis to identify copy number variants (CNVs) and DNA sequencing strategies to identify specific genes that show haploinsufficiency resulting in developmental delay, intellectual disability, autism, epilepsy or schizophrenia.

1U01HG006382-01 (D.H.Ledbetter and D.J. Carey-PIs, 20% effort) 08/15/11-07/31/15
Geisinger eGenomic Medicine (GeM) Program
\$542,164 (annual d.c.)

Geisinger recently joined Phase II of the Electronic Medical Records and Genomics (eMERGE) network, consisting of seven member organizations and one coordinating center. All member organizations have extensive EMRs in their healthcare organization for data-mining purposes to perform genome-wide association studies (GWAS) and other genotype-phenotype correlative studies as a collaborative group for discovery research as well as clinical validation of genomics-guided patient care.

1RC2HD064525-01 (D.H. Ledbetter-PI, 20% effort) 09/30/09-08/31/11
NIH/NICHHD “CNV Atlas of Human Development” \$1,417,216 (annual d.c.)

This Grand Opportunity (GO) grant will support infrastructure and software tool development to allow efficient collection of clinical and genetic data in standardized formats on 50-100,000 pediatric patients/year undergoing genetic diagnostic testing as part of their routine evaluation for autism and other unexplained developmental disorders (intellectual disability, epilepsy, etc.). It will also support development of advanced data integration and visualization tools to provide researchers and clinicians with user-friendly access to these large datasets for data mining and clinical interpretation of genetic test results.

R01 MH081754-03 (D.H. Geschwind-PI; C.L. Martin – subcontract PI) 04/01/08-01/31/13
NIH/NIMH “A Comprehensive Approach to Identification of Autism Susceptibility Genes”
\$170,536 (annual d.c.) (D. H. Ledbetter, 10% effort)

This is a multicenter Autism Center of Excellence (ACE) grant including investigators at UCLA, Emory, U. Chicago, Yale, and Johns Hopkins. It is a broad-based, multidisciplinary approach to identify genes involved in autism etiology and/or predisposition. The goal of this study is to collect simplex families and characterize them through detailed phenotyping and genetic analysis to identify autism susceptibility genes.

R01 HD055651-04 (R. Wapner-PI; C.L. Martin, – subcontract PI) 04/01/07-03/31/12
NIH/NICHHD “Prenatal Cytogenetic Diagnosis using Comparative Genomic Hybridization Microarray”
\$151,664 (annual d.c.) (D. H. Ledbetter, 5% effort)

This study is a large multi-center collaborative study to evaluate the accuracy, efficacy and clinical advantages of prenatal diagnosis using array-based comparative genomic hybridization (aCGH) as compared with conventional cytogenetic analysis (G-banded karyotype).

Simons Foundation, New York (D.H. Ledbetter, PI, 20% effort) 5/01/10-4/30/12
Simons VIP Project
\$302,261 (annual d.c.)

This project is aimed at the identification and recruitment of patients with specific copy number variants (CNVs), such as del 16p11.2, which have a high risk of autism/ASD and other behavioral and psychiatric disorders. Our site will be tasked with recruitment through clinical genetics testing labs (via the ISCA Consortium based at Emory) as well as development of a patient/family website and registry.

GRANT SUPPORT (PAST, SELECTED LIST):

NIH RO1 MH64547: “A Genomewide Search for Autism Susceptibility Loci”; D.H. Ledbetter, subcontract P.I.; 1/0 – 12/07.

NIH RO1 HD36111; “Mechanism and Consequences of Chromosomal Abnormalities”; D. H. Ledbetter, P.I.; 7/98 – 6/04.

NIH PO1 NS39404; Project I. “The Genetic Basis of Lissencephaly”; D.H. Ledbetter, P.I.; 4/99 – 3/04.

NIH PO1 NS39404; Core B. “Molecular Diagnostic Core”; D.H. Ledbetter, P.I.; 4/99 – 3/04.

NIH 1 RO1 HD36715; “Cryptic Telomere Abnormalities in Mental Retardation”; D.H. Ledbetter, P.I.; 3/99 – 2/03.

NICHD 1 RO1 HD20619; “Molecular Analysis of Human Chromosome 17”; D. H. Ledbetter, P.I.; 7/86-3/94.

NIH 1 RO1 HG00024; “High-resolution physical map of human chromosome 17”; D. H. Ledbetter, P.I.; 5/90-4/94.

NICHD 1 RO1 HD22478: “Mechanisms of Fragile X Expression”; D. H. Ledbetter, P.I.; 3/87-2/90.

NICHD 1 U01 HD19881; “Comparison of Chorion Villus Sampling and Amniocentesis”; D. H. Ledbetter, P.I.; 7/84-6/92.

FORMAL TEACHING:

Medical student teaching

1981-1993 Lecturer in Medical Genetics (Pathophysiology course for medical students), Baylor College of Medicine

1997-1999 Course Director and Lecturer for Medical Genetics 339 (22 hours basic science genetics core to first year medical students), University of Chicago

Graduate student teaching

1986-1993 Co-Director and Lecturer for Human Genetics II (graduate student course), Baylor College of Medicine

2001-2002 Course Director and Lecturer, Human Genetics 470, University of Chicago. Introductory course for first year graduate students in Molecular Biosciences Cluster of the Biological Sciences Division.

Other significant teaching

1990-present Co-Organizer and Lecturer for American College of Medical Genetics Review Course. National board review course in Medical Genetics held every 2 years prior to the American Board of Medical Genetics certification exams. Initiated in 1990 by Baylor College of Medicine Genetics Program (“The Baylor Course”) and adopted by the ACMG in 1999.

1992-present Faculty, Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, ME. Initiated in 1959 by Dr. Victor McKusick, this two week intensive course in human and mouse genetics is offered every year to advanced students and professionals from the U. S. and abroad.

1993-1995 Faculty, European School of Medical Genetics, Sestri Levante, Italy. A one-week intensive course in human genetics modeled after the Bar Harbor Short Course held on an annual basis.

SUPERVISORY TEACHING:

Ph.D. students directly supervised:

Apiwat Mutirangura, M.D., Ph.D. (1993), Associate Professor, Faculty of Medicine, Chulalongkorn University, Bangkok, Thailand

Stephanie Mewborn, Ph.D. (2004), University of Chicago.

Postdoctoral fellows directly supervised:

Susanne Gollin, Ph.D., Professor (tenured), University of Pittsburgh, Pittsburgh, PA

Eduardo Cantu, Ph.D., Laboratory Director, Cytogenetics, Genzyme Genetics, Phoenix, AZ

Peter vanTuinen, Ph.D., Director, Medical College of Wisconsin, Milwaukee, WI

Robert F. Stratton, M.D., Clinical Associate Professor, University of Texas Health Sciences Center, San Antonio, TX

Anjana Pettigrew, M.D., Director, Cytogenetics Lab, Associate Professor, University of Kentucky Medical Center

Linda Taylor Cooley, M.D., Assistant Professor, University of Texas Health Sciences Center, Houston

Jacqueline Batanian, Ph.D., Assistant Professor, University of St. Louis Mental Health and Mental Retardation

Jamie Garcia-Heras, M.D., Ph.D., Director, Cytogenetics Laboratory, Texas Dept of Mental Health and and Mental Retardation

Timothy H.-M. Huang, Ph.D., Professor, Ohio State University Cancer Center, Columbus, OH

Akira Kuwano, M.D., Ph.D., Associate Professor, Ehime University, Japan

Lisa G. Shaffer, Ph.D., President, Signature Genomic Laboratories, LLC, Spokane, WA

Romeo Carrozzo, M.D., Assistant Professor, G. Gaslini Institute, Genoa, Italy
Akira Tanigami, M.D., Ph.D., Associate Professor, National Cancer Center Research Institute, Tokyo, Japan
Bing Huang, M.D., Ph.D., Director, Genzyme Genetics, Orange, CA
Yi Ning, M.D., Ph.D., Associate Professor and Director, University of Maryland, Baltimore, MD
Samuel S. Chong, Ph.D., Associate Professor, National University of Singapore, Singapore
Takeo Kubota, M.D., Ph.D., Associate Professor, National Center for Neurology and Psychiatry, Tokyo, Japan
Christa Lese Martin, Ph.D., Associate Professor, Emory University, Atlanta, GA
Daniela Pilz, M.D., Clinical Geneticist, University Hospital, Wales
Naomichi Matsumoto, M.D., Ph.D., Professor, Yokohama City University, Yokohama, Japan
Richard Leventer, PhD, Senior Lecturer, Royal Children's Hospital Melbourne, Victoria, Australia
Carlos Cardoso, Ph.D., Assistant Professor, Faculte de Medecine La Timone, France
Susan Christian, Ph.D., Research Associate (Assistant Professor), University of Chicago
Laura B. Kubicek Herzing, Ph.D., Assistant Professor, Northwestern University, Chicago, IL
Andrew Wong, Ph.D., Instructor, Emory University
Mitsuhiro Kato, M.D., Research Associate, University of Chicago
Erin Baldwin, Ph.D., Instructor, Emory University
Daniel Moreno De Luca, M.D., Research Associate, Emory University, Atlanta, GA
Andres Moreno De Luca, M.D., Research Associate, Emory University, Atlanta, GA
Chunshu Yang, M.D., Ph.D., Research Associate, Emory University, Atlanta, GA

BIBLIOGRAPHY

h-index = 75 (Hirsch J.E.: An index to quantify an individual's scientific research output. Proceedings of the National Academy of Sciences 102:16569-16572, 2005).

Peer-Reviewed Articles

- A1. Gallup, G.G., Jr., **Ledbetter, D.H.** and Maser, J.D.: Strain differences among chickens in tonic immobility: Evidence for an emotionality component. *Journal of Comparative and Physiological Psychology* 90:1075-1081, 1976. PMID: 993390.
- A2. **Ledbetter, D.H.**, Riccardi, V.M., Au, W.W., Wilson, D.P. and Holmquist, G.P.: Ring chromosome 15: Phenotype, Ag-NOR analysis, secondary aneuploidy, and associated chromosome instability. *Cytogenetics and Cell Genetics* 27:111-122, 1980. PMID: 6156798.
- A3. Riccardi, V.M., Hittner, H.M., Francke, U., Yunis, J.J., **Ledbetter, D.H.** and Borges, W.: The aniridia-Wilms tumor association: The critical role of chromosome band 11p11.3-14.1. *Cancer Genetics and Cytogenetics* 2:131-137, 1980.
- A4. Strobel, R.J., Riccardi, V.M., **Ledbetter, D.H.** and Hittner, H.M.: Duplication 11p11.3 leads to 14.1 to meiotic crossing-over. *American Journal of Medical Genetics* 7:15-20, 1980. PMID: 7211949.
- A5. Hittner, H.M., Riccardi, V.M., Ferrell, R.E., Strobel, R.J., **Ledbetter, D.H.**, Strong, L. and Lebo, R.: Genetic heterogeneity of aniridia: Negative linkage data. *Metabolic and Pediatric Ophthalmology* 4:179-182, 1980. PMID: 6259463.
- A6. **Ledbetter, D.H.**, Riccardi, V.M., Airhart, S.D., Strobel, R.J., Keenan, B.S. and Crawford, J.D.: Deletions of chromosome 15 as a cause of the Prader-Willi syndrome. *New England Journal of Medicine* 304:325-329, 1981. PMID 7442771. PMID: 7442771.
- A7. Michels, V.V., Driscoll, D.J., **Ledbetter, D.H.** and Riccardi, V.M.: Phenotype associated with ring 10 chromosome: Report of patient and review of literature. *American Journal of Medical Genetics* 9:231-237, 1981. PMID: 7025632.
- A8. Melton, D.W., Konecki, D.S., **Ledbetter, D.H.**, Hejtmancik, J.F. and Caskey, C.T.: In vitro translation of hypoxanthine/guanine phosphoribosyltransferase mRNA: Characterization of a mouse neuroblastoma cell line that has elevated levels of hypoxanthine/guanine phosphoribosyltransferase protein. *Proceedings of the National Academy of Sciences of the United States of America* 78:6977-6980, 1981. PMID: 6947270.
- A9. **Ledbetter, D.H.**, Mascarello, J.T., Riccardi, V.M., Harper, V.D., Airhart, S.D. and Strobel, R.J.: Chromosome 15 abnormalities and the Prader-Willi syndrome: A follow-up report of 40 cases. *American Journal of Human Genetics* 34:278-285, 1982. PMID: 7072717.
- A10. **Ledbetter, D.H.** and Basen, J.A.: Failure to demonstrate self-recognition in gorillas. *American Journal of Primatology* 2:307-310, 1982.
- A11. Hittner, H.M., King, R.A., Riccardi, V.M., **Ledbetter, D.H.**, Borda, R.P., Ferrell, R.E. and Kretzer, F.L.: Oculocutaneous albinoidism as a manifestation of reduced neural crest derivatives in the Prader-Willi syndrome. *American Journal of Ophthalmology* 94:328-337, 1982. PMID: 6812426.

- A12. Dobyns, W.B., Stratton, R.F., Parke, J.T., Greenberg, F., Nussbaum, R.L. and **Ledbetter, D.H.**: Miller-Dieker syndrome: Lissencephaly and monosomy 17p. *Journal of Pediatrics* 102:552-558, 1983. PMID 6834189.
- A13. Fuscoe, J.C., Fenwick, R.G., Jr., **Ledbetter, D.H.** and Caskey, C.T.: Deletion and amplification of the HGPRT locus in Chinese hamster cells. *Molecular and Cellular Biology* 3:1086-1096, 1983. PMID: 6877239.
- A14. van Tuinen, P. and **Ledbetter, D.H.**: Cytogenetic comparison and phylogeny of three species of Hylobatidae. *American Journal of Physical Anthropology* 61:453-466, 1983. PMID: 6624889.
- A15. Nussbaum, R.L., Airhart, S.D. and **Ledbetter, D.H.**: Expression of the fragile (X) chromosome in an interspecific somatic cell hybrid. *Human Genetics* 64:148-150, 1983. PMID: 6885049.
- A16. Greenberg, F., Carpenter, R.J. and **Ledbetter, D.H.**: Cystic hygroma and hydrops fetalis in a fetus with trisomy 13. *Clinical Genetics* 24:389-391, 1983. PMID: 6652950.
- A17. Greenberg, F., Crowder, W.E., Paschall, V., Colon-Linares, J., Lubianski, B. and **Ledbetter, D.H.**: Familial DiGeorge syndrome and associated partial monosomy of chromosome 22. *Human Genetics* 65:317-319, 1984. PMID: 6693120.
- A18. Stratton, R.F., Dobyns, W.B., Airhart, S.D. and **Ledbetter, D.H.**: New chromosomal syndrome: Miller-Dieker syndrome and monosomy 17p13. *Human Genetics* 67:193-200, 1984. PMID: 6745939.
- A19. Patel, P.I., Nussbaum, R.L., Gramson, P.E., **Ledbetter, D.H.**, Caskey, C.T. and Chinault, A.C.: Organization of the HPRT gene and related sequences in the human genome. *Somatic Cell and Molecular Genetics* 10:483-493, 1984. PMID: 6089358.
- A20. Su, T.S., Nussbaum, R.L., Airhart, S., **Ledbetter, D.H.**, Mohandas, T., O'Brien, W.E. and Beaudet, A.L.: Human chromosomal assignments for 14 argininosuccinate synthetase pseudogenes: Cloned DNAs as reagents for cytogenetic analysis. *American Journal of Human Genetics* 36:954-964, 1984. PMID: 6093508.
- A21. Gollin, S.M., Holmquist, G.P. and **Ledbetter, D.H.**: Fra(10)(q25): The BrdU effect is substitution-dependent. *American Journal of Human Genetics* 37:208-214, 1985. PMID: 3976659.
- A22. Gollin, S.M., Bock, H.G., Caskey, C.T. and **Ledbetter, D.H.**: A new family with fra(10)(q25): Spontaneous expression and 100% expression with 100 microM BrdU. *American Journal of Medical Genetics* 21:643-648, 1985. PMID: 4025395.
- A23. Cantu, E.S., Nussbaum, R.L., Airhart, S.D. and **Ledbetter, D.H.**: Fragile (X) expression induced by FUdR is transient and inversely related to levels of thymidylate synthase activity. *American Journal of Human Genetics* 37:947-955, 1985. PMID: 2931977.
- A24. Chan, L., van Tuinen, P., **Ledbetter, D.H.**, Daiger, S.P., Gotto, A.M., Jr. and Chen, S.H.: The human apolipoprotein B-100 gene: A highly polymorphic gene that maps to the short arm of chromosome 2. *Biochemical and Biophysical Research Communications* 133:248-255, 1985. PMID: 4074366.

- A25. Nussbaum, R.L., Walmsley, R.M., Lesko, J.G., Airhart, S.D. and **Ledbetter, D.H.**: Thymidylate synthase-deficient Chinese hamster cells: A selection system for human chromosome 18 and experimental system for the study of thymidylate synthase regulation and fragile X expression. *American Journal of Human Genetics* 37:1192-1205, 1985. PMID: 3002173.
- A26. Yamaoka, L.H., Bartlett, R.J., Ross, D.A., Fey, G.H., **Ledbetter, D.H.**, Bruns, G., Pericak-Vance, M.A., Herbstreith, M.H. and Roses, A.D.: Localization of cloned unique DNA to three different regions of chromosome 19: Screen for linkage probes for myotonic dystrophy. *Journal of Neurogenetics* 2:403-412, 1985. PMID: 3001264.
- A27. **Ledbetter, D.H.**, Airhart, S.D. and Nussbaum, R.L.: Somatic cell hybrid studies of fragile (X) expression in a carrier female and transmitting male. *American Journal of Medical Genetics* 23:429-443, 1986. PMID: 2937298.
- A28. **Ledbetter, D.H.**, Airhart, S.D. and Nussbaum, R.L.: Caffeine enhances fragile (X) expression in somatic cell hybrids. *American Journal of Medical Genetics* 23:445-455, 1986. PMID: 2937299.
- A29. Nussbaum, R.L., Airhart, S.D. and **Ledbetter, D.H.**: A rodent-human hybrid containing Xq24-qter translocated to a hamster chromosome expresses the Xq27 folate-sensitive fragile site. *American Journal of Medical Genetics* 23:457-466, 1986. PMID: 2937300.
- A30. Nussbaum, R.L., Airhart, S.D. and **Ledbetter, D.H.**: Recombination and amplification of pyrimidine-rich sequences may be responsible for initiation and progression of the Xq27 fragile site: An hypothesis. *American Journal of Medical Genetics* 23:715-721, 1986. PMID: 3456708.
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- A32. Goode, M.E., van Tuinen, P., **Ledbetter, D.H.** and Daiger, S.P.: The anonymous polymorphic DNA clone D1S1, previously mapped to human chromosome 1p36 by in situ hybridization, is from chromosome 3 and is duplicated on chromosome 1. *American Journal of Human Genetics* 38:437-446, 1986. PMID: 3010710.
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- A36. Greenberg, F., Valdes, C., Rosenblatt, H.M., Kirkland, J.L. and **Ledbetter, D.H.**: Hypoparathyroidism and T cell immune defect in a patient with 10p deletion syndrome. *Journal of Pediatrics* 109:489-492, 1986. PMID: 3509963.

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- A38. Greenberg, F., Gresik, M.V., Carpenter, R.J., Law, S.W., Hoffman, L.P. and **Ledbetter, D.H.**: The Gardner-Silengo-Wachtel or genito-palato-cardiac syndrome: Male pseudohermaphroditism with micrognathia, cleft palate, and conotruncal cardiac defect. American Journal of Medical Genetics 26:59-64, 1987. PMID: 3812578.
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