

BIOGRAPHICAL SKETCH

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NAME Joel Naom Hirschhorn, MD, PhD		POSITION TITLE Associate Professor of Genetics, Children's Hospital/Harvard Medical School	
eRA COMMONS USER NAME joelhirschhorn			
EDUCATION/TRAINING (Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.)			
INSTITUTION AND LOCATION	DEGREE (if applicable)	YEAR(s)	FIELD OF STUDY
Harvard College, Cambridge, MA	A.B.	1982-86	Biochemistry
Harvard Medical School, Boston, MA	M.D.	1986-95	Medicine
Harvard University, Graduate School of Arts and Sciences, Cambridge, MA	Ph.D.	1988-95	Genetics

A. Positions and Honors

Fellowships and Honors

1982 Finalist, Westinghouse Science Talent Search
1982 Captain, Stuyvesant HS and New York City Math Teams (national champions)
1982 2nd in Nation, Mathematics Association of America Exam
1986 *summa cum laude*, Biochemistry and Molecular Biology
1987-9, 92-5 Fellow, Medical Scientist Training Program
1989-92 Fikes Fellowship
1995 New England Pediatrics Society, Pediatrics Prize
1998-2001 Howard Hughes Medical Institute Postdoctoral Fellow
2001-2004 Burroughs Wellcome Career Award in the Biomedical Sciences
2004 Young Investigator Award, Society for Pediatrics Research
2006 Member, American Society of Clinical Investigation

Clinical Appointments

1995-Present Attending Physician, Children's Hospital, Department of Medicine

Board Certifications

2001-Present Certified, Pediatric Endocrinology

Academic and Professional Appointments

2000-2001 Harvard Medical School, Instructor in Pediatrics
2001-2007 Harvard Medical School, Assistant Professor in Genetics (Pediatrics)
2007-present Harvard Medical School, Associate Professor in Genetics
2001-2004 Whitehead Institute/MIT Center for Genome Research, Director of Endocrine Genetics
2004-2006 Broad Institute of MIT and Harvard, Associate Member; Coordinator of Metabolism Initiative
2006-Present Broad Institute, Senior Associate Member; Coordinator of Metabolism Initiative

Research and Professional Experience:

1988-1995 Ph.D. Research, *Chromatin structure and gene expression in yeast*, Harvard Univ.; Fred Winston, advisor
1995-1997 Intern and Resident in Pediatrics, Department of Medicine, Children's Hospital, Boston
1997-2000 Clinical and Research Fellow in Pediatric Endocrinology, Children's Hospital, Boston
1998-2001 Post-doctoral Fellowship, *Genetic analysis of complex endocrine traits* Whitehead /MIT Center for Genome Research; Eric Lander, mentor
2001-present Independent investigator, *Genetics of complex traits and endocrine disorders*, Children's Hospital, Boston and Broad Institute of MIT and Harvard

B. Selected original peer-reviewed publications (from 73)

1. Altshuler D⁺, **Hirschhorn JN⁺**, Klannemark M, Lindgren CM, Vohl M-C, Nemesh J, Lane C, Schaffner SF, Bolk S, Brewer C, Tuomi T, Gaudet D, Hudson TJ, Daly M, Groop L, Lander ES. The common Pro12Ala variant in PPAR γ is associated with decreased risk of type 2 diabetes. *Nature Genet.* 2000; 26: 76-80

2. **Hirschhorn JN**, Lohmueller K, Byrne E, Hirschhorn K. A comprehensive review of genetic association studies. *Genet. Med.* 2002; 4: 45-61.
- +These authors contributed equally to this work.
3. Lohmueller KE, Pearce CL, Pike M, Lander ES, **Hirschhorn JN**. Meta-analysis of genetic association studies supports a contribution of common variants to susceptibility to common disease. *Nature Genet.* 2003; 33:177-82.
4. Bersaglieri T, Sabeti PC, Patterson N, Vanderploeg T, Schaffner SF, Drake JA, Rhodes M, Reich DE, **Hirschhorn JN**. Genetic signatures of strong recent positive selection at the lactase gene. *Am. J. Hum. Genet.* 2004; 74:1111-20.
5. Freedman ML+, Reich D+, Penney K, McDonald GJ, Mignault AA, Patterson N, Gabriel SB, Topol EJ, Smoller JW, Pato CN, Pato MT, Petryshen T, Kolonel L, Lander ES, Sklar P, Henderson B, **Hirschhorn JN**, Altshuler D. Assessing the impact of population stratification on genetic association studies. *Nature Genet.* 2004; 36:388-93.
6. Campbell CD, Ogburn EL, Lunetta KL, Lyon HN, Freedman ML, Groop LC, Altshuler D, Ardlie KG, **Hirschhorn JN**. Demonstrating stratification in a European-American population. *Nature Genet.* 2005; 37:868-72.
7. Drake JA+, Bird C+, Nemesh J+, Thomas DJ+, Newton-Cheh C, Reymond A, Excoffier L, Attar H, Antonarakis SE, Dermitzakis E*, **Hirschhorn JN***. Conserved noncoding sequences are selectively constrained and not mutation cold spots. *Nature Genet.* 2006; 38:223-7.
8. Herbert A, Gerry NP, McQueen MB, Heid IM, Pfeufer A, Illig T, Wichmann HE, Meitinger T, Hunter D, Hu FB, Colditz G, Hinney A, Hebebrand J, Koberwitz K, Zhu X, Cooper R, Ardlie K, Lyon H, **Hirschhorn JN**, Laird NM, Lenburg ME, Lange C and Christman MF. A common genetic variant is associated with adult and childhood obesity. *Science* 2006; 312:279-83.
9. Lyon HN+, Florez JC+, Bersaglieri T, Saxena R, Winckler W, Almgren P, Lindblad U, Tuomi T, Gaudet D, Zhu X, Cooper R, Ardlie KG, Daly MJ, Altshuler D, Groop L*, **Hirschhorn JN***. Common variants in the ENPP1 gene are not reproducibly associated with diabetes or obesity. *Diabetes* 2006; 55:3180-3184.
10. Campbell CD, Lyon HN, Nemesh J, Drake JA, Tuomi T, Gaudet D, Zhu X, Cooper RS, Ardlie KG, Groop LC, **Hirschhorn JN**. Association studies of body mass index and type 2 diabetes in the Neuropeptide Y pathway: a possible role for NPY2R as a candidate gene for type 2 diabetes in men. *Diabetes* 2007; 56:1460-7.
11. Lettre G, Lange C, **Hirschhorn JN**. Genetic model testing and statistical power in population-based association studies of quantitative traits. *Genetic Epidemiol.* 2007; 31:358-62.
12. Diabetes Genetics Initiative (member of study design, writing and clinical characterization/phenotype teams). Genome-wide association analysis identifies loci for type 2 diabetes and triglyceride levels. *Science.* 2007 316:1331-6
13. Lyon HN+, Emilsson V+, Hinney A+, Heid IM+, Lasky-Su J+, ... Celedon JC*, Wichmann HE*, Hebebrand J*, Stefansson K*, Lange C*, **Hirschhorn JN***. The association of a SNP upstream of INSIG2 with Body Mass Index is reproduced in several but not all cohorts. *PLoS Genetics* 2007; 3:e61.
14. Weedon MN+, Lettre G+, Freathy RM+, Lindgren CM+, ..., McCarthy MI*, **Hirschhorn JN***, Frayling TM*. A common variant of HMGA2 is associated with adult and childhood height in the general population. *Nature Genet.* 2007; 39:1245-50.
15. Lettre G, Butler JL, Ardlie KG, **Hirschhorn JN**. Common genetic variation in eight genes of the GH/IGF1 axis does not contribute to adult height variation. *Hum. Genet.* 2007; 122:129-139.
16. Campbell CD, Kirby A, Nemesh J, Daly MJ, **Hirschhorn JN**. A survey of allelic imbalance in F1 mice. *Genome Res.* 2008; 18:555-563.
17. Lettre G, Jackson AU*, Gieger C*, Schumacher FR*, Berndt SI*, ..., **Hirschhorn JN**. Genome-wide association studies identify ten novel loci for height and highlight new biological pathways in human growth. *Nature Genet.* 2008; Epub April 6.
18. Willer CJ+, Speliotes EK+, Loos RJ+, Li S+, Lindgren CM, Heid IM, ..., McCarthy MI*, Boehnke M*, Barroso I*, Abecasis GR*, **Hirschhorn JN***; Genetic Investigation of ANthropometric Traits Consortium. Six

new loci associated with body mass index highlight a neuronal influence on body weight regulation. *Nature Genet.* 2009; 41:25-34.

+*These authors contributed equally to this work.

Selected chapters, invited editorials and reviews (from 22)

1. **Hirschhorn JN**, Daly MJ. Genome-wide association studies for common diseases and complex traits. *Nat. Rev. Genet.* 2005; 6:95-108.
2. McCarthy MI, Abecasis GR, Cardon LR, Goldstein DB, Little J, Ioannidis JPA, **Hirschhorn JN**. Genome wide association studies for complex traits: consensus, uncertainty and challenges. *Nature Rev. Genet.* 2008; Epub April 9.