BIOGRAPHICAL SKETCH

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NAME Thakuria, Joseph Vincent	POSITION TITLE Attending Physician in Clinical and Biochemical Genetics / Instructor of Pediatrics
eRA COMMONS USER NAME (credential, e.g., agency login) JTHAKURIA	

EDUCATION/TRAINING

INSTITUTION AND LOCATION	DEGREE *(if applicable)*	YEAR(s)	FIELD OF STUDY
Pennsylvania State University, State College, PA	B.S.	1986-1990	Psychology / Anthropology
St. Georges University, Grenada, West Indies	M.D.	1990-1995	Medicine
University of Pennsylvania Health System, Philadelphia, PA	Internship and Residency	1996-2000	Internal Medicine
Harvard Medical School, Boston, MA	Residency	2005-2008	Clinical and Biochemical Genetics
Harvard Medical School, Boston, MA	M.M.Sc.	2006-2008	Masters in Medical Science / HMS Clinical Scholars Program

A. Personal Statement:

I have background training in internal medicine and clinical and biochemical genetics with expertise in individualized interpretation of genomic and other -omic datasets. While still in my clinical genetics training, I discovered a new syndrome of Wolff-Parkinson-White arrhythmia with developmental delay, along with the causative gene. Since 2005, I have worked as co-investigator and Medical Director of the Personal Genome Project (PGP) led by PI, Dr. George Church – an HMS IRB approved study for enrollment of 100,000 participants for comprehensive sequencing, phenotyping, tissue collection, generation of individualized adult stem cell lines, and biobanking. I have been instrumental in developing GET (Genetic, Environmental, and Traits) Evidence, and Trait-o-matic for automated analysis of whole genome and exome data. More recently, colleagues and I have developed the Genome Parsing System (GPS), a genomic analyses tool for user-specified, project-customized, clinical bioinformatics. I aided in clinical genomic analyses published in several high-impact journals including Nature, Science, and Lancet. I work as a staff physician in clinical and biochemical genetics at Massachusetts General Hospital and have been involved in NHLBI and NHGRI Working Groups to develop consensus statements on the return of genomic results to research participants. I also review allocation of inpatient and outpatient resources for clinical genetic/genomic trials conducted system-wide across Harvard affiliated hospitals through my work with the Harvard Catalyst Clinical Research Center Protocol Review Committee.

B. Positions and Honors:

1996-2000: Internal Medicine, Intern and Resident, University of Pennsylvania Health System
2000-2002: Assistant Director of Medical Education, Frankford Hospital, Jefferson Health System
2002-2005: Instructor in Medicine/Inpatient Specialist, North Shore – Long Island Jewish Health System
2005- 2008: Resident in Clinical and Biochemical Genetics, Harvard Medical School
2006-2008: Clinical Scholar, Harvard Medical School Scholars in Clinical Science program
2008-Present: Attending Physician in Clinical and Biochemical Genetics/Instructor of Pediatrics, Massachusetts General Hospital/ Harvard Medical School
1997-2000: Harrison Fellowship Award
2002: PCOM Physician Recognition Award for Academic Excellence
2007: Genzyme Clinical Biochemical Fellowship Award
2007: Society of Inherited Metabolic Disorders Fellowship Award

Professional Society Memberships:
American Medical Association
American College of Physicians

American College of Medical Genetics
American Society of Human Genetics
Society of Inherited Metabolic Disorders
New England Regional Genetics Group
New York State Medical Society
Thomas Bond Society
2009-2010: Member, NIH/NHLBI Working Group on Guidelines for Reporting of Genetic Research Results to Study Participants

C. Selected peer-reviewed publications:

1. Oh A, Thakuria JV, Kimonis V, Mulliken J, "Subglossopalatal Synechia in Association with Cardiac and Digital Anomalies," *Cleft Palate-Craniofacial Journal*, 2008 Mar;45(2):217-21. Review.

2. Miller DT, Shen Y, Weiss LA, Korn J, Anselm I, Bridgemohan C, Cox GF, Dickinson H, Gentile J, Harris DJ, Hegde V, Hundley R, Khwaja O, Kothare S, Luedke C, Nasir R, Poduri A, Prasad K, Raffalli P, Reinhard A, Smith S, Sobeih M, Soul J, Stoler J, Takeoka M, Tan W, Thakuria JV,Wolff R, Yusupov R, Gusella JF, Daly MJ, Wu B, "Microdeletion/duplication at 15q13.2-q13.3 among individuals with features of autism and other neuropsychiatric disorders," *Journal of Medical Genetics*, 2009 Apr;46(4):242-8. Epub 2008 Sep 19.

3. Lalani SR*, Thakuria JV*, Cox GF, Wang X, Weimin B, Bray M, Shaw C, Cheung SW, Chinault C, Boggs B, Ou Z, Lupski J, Beaudet A, Pursley A, Zapata G, Friedman R, Stankiewicz P, Towbin J, Schnittger S, Hansmann I, Belmont J, Potocki L, "20p12.3 Microdeletion predisposes to Wolff-Parkinson-White syndrome with variable neurocognitive deficits," *Journal of Medical Genetics*, 2009 Mar;46(3):168-75. Epub 2008 Sep 23. (*= co-first authors)

4. Kim JI, Ju YS, Park H, Kim S, Lee S, Yi JH, Mudge J, Miller NA, Hong D, Bell CJ, Kim HS, Chung IS, Lee WC, Lee JS, Seo SH, Yun JY, Woo HN, Lee H, Suh D, Lee S, Kim HJ, Yavartanoo M, Kwak M, Zheng Y, Lee MK, Park H, Kim JY, Gokcumen O, Mills RE, Zaranek AW, Thakuria JV, Wu X, Kim RW, Huntley JJ, Luo S, Schroth GP, Wu TD, Kim H, Yang KS, Park WY, Kim H, Church GM, Lee C, Kingsmore SF, Seo JS., "A highly annotated whole-genome sequence of a Korean individual," *Nature,* 2009 Aug 20;460(7258):1011-5. Epub 2009 Jul 8.

5. Drmanac R, Sparks AB, Callow MJ, Halpern AL, Burns NL, Kermani BG, Carnevali P, Nazarenko I, Nilsen GB, Yeung G, Dahl F, Fernandez A, Staker B, Pant KP, Baccash J, Borcherding AP, Brownley A, Cedeno R, Chen L, Chernikoff D, Cheung A, Chirita R, Curson B, Ebert JC, Hacker CR, Hartlage R, Hauser B, Huang S, Jiang Y, Karpinchyk V, Koenig M, Kong C, Landers T, Le C, Liu J, McBride CE, Morenzoni M, Morey RE, Mutch K, Perazich H, Perry K, Peters BA, Peterson J, Pethiyagoda CL, Pothuraju K, Richter C, Rosenbaum AM, Roy S, Shafto J, Sharanhovich U, Shannon KW, Sheppy CG, Sun M, Thakuria JV, Tran A, Vu D, Zaranek AW, Wu X, Drmanac S, Oliphant AR, Banyai WC, Martin B, Ballinger DG, Church GM, Reid CA., "Human genome sequencing using unchained base reads on self-assembling DNA nanoarrays," *Science*, 2010 Jan 1;327(5961):78-81. Epub 2009 Nov 5.

6. Ashley EA, Butte AJ, Wheeler MT, Chen R, Klein TE, Dewey FE, Dudley J, Ormond KE, Pavlovic A, Hudgins L, Gong L, Hodges LM, Berlin DS, Thorn CF, Sangkuhl K, Hebert JM, Woon M, Sagreiya H, Whaley R, Morgan AA, Pushkarev D, Neff NF, Knowles JW, Chou M, Thakuria JV, Rosenbaum A, Zaranek AW, Church GM, Greely HT, Quake SR, Altman RB, "Clinical evaluation incorporating a personal genome," *Lancet,* 2010 May 1;375(9725):1525-35.

7. Lunshof JE, Bobe J, Aach J, Angrist M, Thakuria JV, Vorhaus DB, Hoehe MR, Church GM, "Personal genomes in progress: from the human genome project to the personal genome project," *Dialogues in Clinical Neuroscience,* 2010;12(1):47-60

8. Fabsitz RR, McGuire A, Sharp RR, Puggal M, Beskow LM, Biesecker LG, Bookman E, Burke W, Burchard EG, Church GM, Clayton EW, Eckfeldt JH, Fernandez CV, Fisher R, Fullerton SM, Gabriel S, Gachupin F, James C, Jarvik GP, Kittles R, Leib JR, O'Donnell C, O'Rourke PP, Rodriguez LL, Schully SD, Shuldiner AR, Sze RKF, Thakuria JV, Wolf SM, Burke GL, "Ethical and Practical Guidelines for Reporting Genetic Research Results to Study Participants:

Updated Guidelines From a National Heart, Lung, and Blood Institute Working Group," *Circ Cardiovasc Genet.* 2010 Dec 1;3(6):574-80

9. Dewey FE, Chen R, Cordero SP, Ormond KE, Caleshu C, Karczewski KJ, Whirl-Carrillo M, Wheeler MT, Dudley JT, Byrnes JK, Cornejo OE, Knowles JW, Woon M, Sangkuhl K, Gong L, Thorn CF, Hebert JM, Capriotti E, David SP, Pavlovic A, West A, Thakuria JV, Ball MP, Zaranek AW, Rehm HL, Church GM, West JS, Bustamante CD, Snyder M, Altman RB, Klein TE, Butte AJ, Ashley EA, "Phased whole-genome genetic risk in a family quartet using a major allele reference sequence,"*PLoS Genet*, 2011 Sept 7;9

10. Fradley MG, Collins AB, Moore SA, Thakuria JV, Stone JR, "The utility of direct tissue assessment using immunofluoresence in the diagnosis of hereditary transthyretin cardiac amyloidosis," *Tex Heart Inst J.* 2012;39(1):71-5

11. Thakuria JV, Zaranek AW, Church GM, Berry GT, "Back to the Future: From Genome to Metabolome," *Human Mutat.* 2012 May;33(5):809-12

12. Ball MP*, Thakuria JV*, Zaranek AW*, Clegg T, RosenbaumAM, Wu X, Angrist M, Bhak J, Bobe J, Callow M, Cano C, Chou MF, Chung WK, Douglas SM, Estep PW, Gore A, Hulick P, Labarga A, Lee J, Lunshof J, Kim BC, Kim J, Li Z, Murray MF, Nilsen G, Peters B, Raman AM, Rienhoff HY, Robasky K, Wheeler M, Vandewege W, Vorhaus D, Yang JL, Yang L, Aach J, Ashley EA, Drmanac R, Kim S, Li JB, Peshkin L, Seidman CE, Seo J, Zhang K, Rehm HL, Church GM, "A Public Resource Facilitating Clinical Use of Genomes," Proc Natl Acad Sci U S A. 2012 Jul 24;109(30):11920-7. (*= co-first authors)

13. Thakuria JV, Murray M, (in press for November, 2013), Clinical Interpretation of Genomic Data, chapter in *Clinical Genomics: Practical Applications in Adult Patient Care*, Mc Graw Hill, Columbus, OH

D. Research Support:

Personal Genome Project (2005-2013): Co-investigator and Medical Director of the Personal Genome Project (PGP) – an HMS IRB approved study for enrollment of 100,000 participants for comprehensive sequencing, phenotyping, tissue collection, biobanking, generation and distribution of individualized cell lines (including iPS). PI is Dr. George Church. NHGRI, NHLBI, and private funding.

Next Generation Sequencing Panel for Neurodegenerative Disorders: awarded HMS Catalyst Pilot Grant along with Kathie Sims (MGH), Winnie Xie (MGH), and Mike Chou (HMS).

IRB review in progress for 2 industry sponsored trials (in collaboration with Mike Murray (BWH): a) Identification of Undiagnosed Gaucher Disease (Genzyme), b) Identification of Undiagnosed Lysosomal Acid Lipase Deficiency (Synageva BioPharma Corporation)