CURRICULUM VITAE

NAME

Kim, Hyung-Goo, PhD
Associate Professor / Senior Scientist
Neurological Disorders Research Center
Qatar Biomedical Research Institute
Hamad Bin Khalifa University
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Citizenship: United States of America Citizen

EDUCATION

BE in Chemical Engineering, Korea University, Seoul, Korea, 1981
BS and M.Sc. in Chemistry, University of Goettingen, Germany, 1998
PhD student, Faculty of Biochemistry, Free University of Berlin, Germany, 1999-2001
Research Fellow at Harvard Medical School, Boston, USA, 2001-2006
Research Fellow at Augusta University, USA, 2006-2013
PhD (Dr. rer. nat.), Magna Cum Laude, Biochemistry, Free University of Berlin, Germany, 2012

PROFESSIONAL

Army Ammunition Officer, First Lieutenant, South Korean Army, 1982-1985
Chemist, Korea Oil Refinery, Ulsan, Korea, 1986-1987
Chemist, Kori Nuclear Power Plant, Korea Electric Power Corporation, Korea, 1988-1990
Research Associate, University Hospital, Goettingen, Germany, 1998-1999

HONORS

Molecular Genetics Fellowship, Max-Plank Institute for Molecular Genetics,
Berlin, Germany, 1999-2001

SCIENTIFIC AND PROFESSIONAL SOCIETIES

1999-present:	American Society of Human Genetics
2000:	German Society of Human Genetics
2018:	European Society of Human Genetics
2006-present:	The Endocrine Society

ACADEMIC APPOINTMENT

Associate Professor, Department of Obstetrics & Gynecology, Department of Neuroscience and Regenerative Medicine, Medical College of Georgia at Augusta University, Augusta, GA, USA:

February 1st 2013-April 30th 2018

Senior Scientist / Joint Associate Professor, Neurological Disorders Research Center, Qatar Biomedical Research Institute, Hamad Bin Khalifa University, Doha, Qatar: August 3rd 2018-Present

CONTEST JUDGE

2012 American Society of Human Genetics DNA Day Essay Contest
2013 American Society of Human Genetics DNA Day Essay Contest
2014 American Society of Human Genetics DNA Day Essay Contest
2015 American Society of Human Genetics DNA Day Essay Contest
2016 American Society of Human Genetics DNA Day Essay Contest
2017 American Society of Human Genetics DNA Day Essay Contest
2018 American Society of Human Genetics DNA Day Essay Contest

RESEARCH WEBSITE

Developmental Gene Discovery Project (DGDP) website (www.lonelygene.com. username: hkim; password: farang6123) maintains breakpoint maps, medical records, pedigrees, and other information on families with important chromosomal rearrangements and various neurological disorders including intellectual disability, autism, speech delay, and behavioral disorders.

PRESENTATION AT NATIONAL AND INTERNATIONAL MEETINGS

1. "Molecular characterization of a translocation t(1;19) (q21.3; q13.2) associated with mental retardation and muscular weakness" at the 12th annual meeting of the German Society of Human Genetics held together with the Austrian Society of Human Genetics and the Swiss Society of Medical Genetics in Luebeck, Germany on March 23rd, 2000.
2. "Disease Gene Identification in Human Developmental Disorders" in Grand Rounds of Department of Neurology, Georgia Regents University in Augusta, GA, USA on March 7th 2013.
3. "Disease Gene Discovery with Chromosomal Rearrangements" at Inje University Haeundae Paik Hospital in Busan, Korea on August 23rd, 2013.
4. "Disease Gene Discovery with Chromosomal Rearrangements" at Korea Research Institute for Bioscience and Biotechnology in Daejeon, Korea on August 26th, 2013.
5. "Disease Gene Discovery with Chromosomal Rearrangements" at Chungnam National University in Daejeon, Korea on August 26th, 2013.
6. "Disease Gene Discovery with Chromosomal Rearrangements" at Korea Advanced Institute of Science and Technology in Daejeon, Korea on August 26th, 2013.
7. "Disease Gene Discovery with Chromosomal Rearrangements" at Korea National Institute of Health in Cheongwon-gun, Chungcheongbuk-do, Korea on August 27th, 2013.
8. "Disease Gene Discovery with Chromosomal Rearrangements" at Gachon University of Medicine and Science in Inchon, Korea on August 28th, 2013.
9. "Disease Gene Discovery with Chromosomal Rearrangements" at Asan Medical Center in Seoul, Korea on August 28th, 2013.
10. "Positional Cloning of a New Disease Gene for Potocki-Shaffer Syndrome" at Korea Research Institute of Bioscience & Biotechnology in Daejeon, Korea on May 15th, 2014.
11. "Positional Cloning of a New Disease Gene for Potocki-Shaffer Syndrome" at Gyeongsang National University in Jinju, Korea on May 19th, 2014.
12. "Disease gene discovery for recessive disorders from Pakistani families" at Seoul National University College of Medicine in Seoul, Korea on May 20th, 2014.

13. "Positional Cloning of Autosomal Recessive Disease Genes in Human Developmental Disorders from Consanguineous Marriages" at the Department of Psychiatry at the Hospital Farhat Hached in Sousse, Tunisia on April 14th, 2015.
14. "Molecular Players in Autosomal Dominant Intellectual Disability" at 10th International Meeting on Copy Number Variants and Genes in Intellectual Disability and Autism in Troina, Italy on April 17th, 2015
15. "Molecular Players in Autosomal Dominant Intellectual Disability" at Gyeongsang National University in Jinju, Korea on May 27th, 2015.
16. "Molecular Players in Autosomal Dominant Intellectual Disability" at Chungnam National University in Daejeon, Korea on June 3rd, 2015.
17. "Genetic Mutations and Zebrafish Knockdown Identified *CHD7* as a Cause of DiGeorge syndrome" at Chungnam National University in Daejeon, Korea on April 14th, 2016.
18. "In Search of the Causative Genes for Body Integrity Identity Disorder" at Gyeongsang National University in Jinju, Korea on April 18th, 2016.
19. "Discovery of Disease Genes for Intellectual Disability and Autism" at the University of Sciences, Techniques and Technologies of Bamako (USTTB), Bamako, Mali on June 26th, 2017.
20. "Discovery of Disease Genes for Intellectual Disability and Autism" at the M&M Hospital, Navy Estate, Karshi, Abuja, FCT Nigeria on July 20th, 2017.
21. "Genetic Underpinnings of Autosomal Dominant & Recessive Intellectual Disability and Autism", Qatar Biomedical Research Institute, Hamad bin Khalifa University, Qatar on November 13th, 2017.
22. "In search of Disease Genes for Intellectual Disability and Autism" at the Department of Neurologic Surgery, Mayo Clinic College of Medicine, Mayo Clinic, Rochester, MN on March 13th, 2018.
23. "Identification of Disease Genes in Human Developmental Disorders", University of Okara, Pakistan on September 20, 2019.

TEACHING

Department of OB/GYN, Augusta University, Augusta, USA
Seminar Series to Third Year Medical Students and Fellows: Reproductive Genetics and Advanced Genetics
Feb 2013-Apr 2018

College of Health & Life Sciences, Hamad Bin Khalifa University, Doha, Qatar
Advanced Genetics Course Code: LS 504 to PhD Students
Aug 2018-Present

PEER REVIEWED PUBLICATIONS & REVIEWS

Original Publications

1. Nothwang HG, **Kim HG**, Aoki J, Geisterfer M, Kubart S, Wegner RD, van Moers A, Ashworth LK, Haaf T, Bell J, Arai H, Tommerup N, Ropers HH, Wirth J. *Functional hemizygosity of PAFAH1B3 due to a PAFAH1B3-CLK2 fusion gene in a female with mental retardation, ataxia and atrophy of the brain*. ***Hum Mol Genet*** 2001;10:797-806.
2. Scherer SW, Cheung J, MacDonald JR, Osborne LR, Nakabayashi K, Herbrick JA, Carson AR, Parker-Katiraee L, Skaug J, Khaja R, Zhang J, Hudek AK, Li M, Haddad M, Duggan GE, Fernandez BA, Kanematsu E, Gentles S, Christopoulos CC, Choufani S, Kwasnicka D, Zheng XH, Lai Z, Nusskern D, Zhang Q, Gu Z, Lu F, Zeesman S, Nowaczyk MJ, Teshima I, Chitayat D, Shuman C, Weksberg R, Zackai EH, Grebe TA, Cox SR, Kirkpatrick SJ, Rahman N, Friedman JM, Heng HH, Pelicci PG, Lo-Coco F, Belloni E, Shaffer LG, Pober B, Morton CC, Gusella JF, Bruns GA, Korf BR, Quade BJ, Ligon AH, Ferguson H, Higgins AW, Leach NT, Herrick SR, Lemyre E, Farra CG, **Kim HG**, Summers AM, Gripp KW, Roberts W, Szatmari P, Winsor EJ, Grzeschik KH, Teebi A, Minassian BA, Kere J, Armengol L, Pujana MA, Estivill X, Wilson MD, Koop BF, Tosi S, Moore GE, Boright AP, Zlotorynski E, Kerem B, Kroisel PM, Petek E, Oscier DG, Mould SJ, Döhner H, Döhner K, Rommens JM, Vincent JB, Venter JC, Li PW, Mural RJ, Adams MD, Tsui LC. *Human chromosome 7: DNA sequence and biology*. ***Science*** 2003; 300(5620): 767-772.

3. **Kim HG**, Herrick SR, Lemyre E, Kishikawa S, Salisz JA, Seminara S, MacDonald ME, Bruns GAP, Morton CC, Quade BJ, Gusella JF. *Hypogonadotropic hypogonadism and cleft-lip and palate due to balanced translocation producing haploinsufficiency for FGFR1.* ***J Med Genet*** 2005; 42:666-672.
4. **Kim HG**, Higgins AW, Herrick SR, Kishikawa S, Nicholson L, Kutsche K, Ligon AH, Harris DJ, MacDonald ME, Bruns GAP, Morton CC, Quade BJ, Gusella JF. *Candidate loci for Zimmermann–Laband syndrome at 3p14.3.* ***Am J Med Genet*** A 2007;143A(2):107-111.
5. Lu W, van Eerde AM, Fan X, Quintero-Rivera F, Kulkarni S, Ferguson H, **Kim HG**, Fan Y, Xin Q, Li Q, Sanlaville D, Andrews W, Sundaresan V, Bi W, Yan J, Giltay JC, Wijmenga C, PVM de Jong T, Feather SA, Woolf AS, Rao Y,Lupski JR, Eccles MR, Quade BJ, Gusella JF, Morton CC, Maas RL. *Disruption of ROBO2 is associated with urinary tract anomalies and confers risk of vesicoureteral reflux.* ***Am J Hum Genet*** 2007; 80(4):616-32.
6. Abo-Dalo B*, **Kim HG*** **(*equally contributed first author)**, Stefanova M, Higgins A, Shen Y, Mundlos S, Quade BJ, Gusella JF, and Kutsche K. *Extensive molecular genetic analysis of the 3p14.3 region in patients with Zimmermann-Laband syndrome.* ***Am J Med Genet*** A 2007;143A(22):2668-74.
7. **Kim HG***, Kishikawa S* **(*equally contributed)**, Higgins A, Seong IS, Donovan D, Shen Y, Lally E, Weiss L, Najm J, Kutsche K, Descartes M, Holt L, Braddock S, Troxell R, Kaplan L, Volkmar F, Klin A, Tsatsanis K, Noens I, Pauls D, Daly MJ, MacDonald ME, Morton CC, Quad BJ, Gusella JF. *Disruption of neurexin 1 associated with autism specrum disorder.* ***Am J Hum Genet*** 2008; 82(1):199-207.
8. Higgins AW, Alkuraya FS, Bosco AF, Brown KK, Bruns GAP, Donovan DJ, Eisenman R, Fan Y, Farra CG, Ferguson HL, Gusella JF, Harris DJ, Herrick SR, Kelly C, **Kim HG**, Kishikawa S, Korf BR, Kulkarni S, Lally E, Leach NT, Lemyre E, Lewis J, Ligon AH, Lu W, Maas RH, MacDonald ME, Moore SDP, Peters RE, Quade BJ, Quintero-Rivera F, Saadi I, Shen Y, Shendure J, Williamson RE, Morton CC. *Characterization of Apparently Balanced Chromosomal Rearrangements from the Developmental Genome Anatomy Project*. ***Am J Hum Genet*** 2008; 82(3):712-722.
9. Dibbens LM, Tarpey PS, Hynes K, Bayly B, Scheffer IE, Smith R, Bomar J, Sutton E, Vandeleur L, Shoubridge C, Edkins S, Turner SJ, Stevens C, O'Meara S, Tofts C, Barthorpe S, Buck G, Cole J, Halliday K, Jones D, Lee R, Madison M, Mironenko T, Varian J, West S, Widaa S, Wray P, Teague J, Dicks E, Butler A, Menzies A, Jenkinson A, Shepherd R, Gusella JF, Afawi Z, Mazarib A, Neufeld MY, Kivity S, Lev D, Lerman-Sagie T, Korczyn AD, Derry CP, Sutherland GR, Friend K, Shaw M, Corbett M, **Kim HG**, Geschwind DH, Thomas P, Haan E, Ryan S, McKee S, Berkovic SF, Futreal PA, Stratton MR, Mulley JC, Gécz J. *X-linked protocadherin 19 mutations cause sex-limited epilepsy and cognitive impairment.* ***Nat Genet*** 2008; 40(6):776-781.
10. **Kim HG**, Kurth I, Lan F, Meliciani I, Wenzel W, Eom SH, Kang GB, Rosenberger G, Tekin M, Ozata M, Bick DP, Sherins RJ, Walker SL, Shi Y, Gusella JF, Layman LC. *Mutations in CHD7, Encoding a Chromatin-Remodeling Protein Cause Idiopathic Hypogonadotropic Hypogonadism and Kallmann Syndrome*. ***Am J Hum Genet*** 2008; 83(4):511-519.
11. **Kim HG**, Bhagavath B, Layman LC. *Clinical Manifestations of Impaired GnRH Neuron Development and Function.* ***Neurosignals*** 2008;16(2-3):165-182.
12. Xu N, Bhagavath B*, **Kim HG*** **(*equally contributed),** Halvorson L, Podolsky RS, Chorich LP, Prasad P, Xiong WC, Cameron RC, Layman LC. *NELF Is a nuclear protein involved in hypothalamic GnRH neuronal migration.* ***Mol Cell Endocrinol.*** 2010; 319(1-2):47-55.
13. **Kim HG* (*co-corresponding author)**, Ahn JW, Kurth I, Ullmann R, Kim HT, Kulharya A, Ha KS, Itokawa Y, Meliciani I, Wenzel W, Lee D, Rosenberger G, Ozata M, Bick DP, Sherins RJ, Nagase T, Tekin M, Kim SH, Kim CH, Ropers HH, Gusella JF, Kalscheuer V, Choi CY and Layman LC*****. *WDR11, a WD Protein that Interacts with Transcription Factor EMX1, Is Mutated in Idiopathic Hypogonadotropic Hypogonadism and Kallmann Syndrome.* ***Am J Hum Genet*** 2010; 87(4):465-479.
14. **Kim HG**, Pedersen-White JR, Bhagavath B, Layman LC. *Genotype and phenotype of patients with gonadotropin releasing hormone receptor mutations.* ***Front Horm Res*** 2010;39:94-110.
15. Aminzadeh M, **Kim HG**, Layman LC, Cheetham TD. *Rarer Syndromes Characterized by Hypogonadotropic Hypogonadism.* ***Front Horm Res***. 2010;39:154-167.
16. Xu N*, **Kim HG*** **(*equally contributed)**, Bhagavath B, Cho SG, Lee JH, Ha KS, Meliciani I, Wenzel W, Podolsky RS, Chorich LP, Stackhouse KA, Grove AMH, Odom LN, Ozata M, Bick DP, Sherins RJ, Kim SH, Cameron RS, Layman LC. Nasal Embryonic LHRH Factor (NELF) *Mutations in Patients with Normosmic Hypogonadotropic Hypogonadism and Kallmann Syndrome.* ***Fertility & Sterility*** 2011; 95(5): 1613-1620.

17. Jain S, **Kim HG**, Lacbawan F, Meliciani I, Wenzel W, Kurth I, Sharma J, Schoeneman M, Ten S, Layman LC, Jacobson-Dickman E. *Unique phenotype in a patient with CHARGE syndrome*. ***Int J Pediatr Endocrinol.*** 2011; 11: 1-8.
18. Quaynor SD, **Kim HG**, Cappello EM, Williams T, Chorich LP, Bick DP, Sherins RJ, Layman LC. *The prevalence of digenic mutations in patients with normosmic hypogonadotropic hypogonadism and Kallmann syndrome.* ***Fertility & Sterility*** 2011; 96(6):1424-1430.
19. **Kim HG**, Layman LC. *The Role of CHD7 and the Newly Identified WDR11 Gene in Patients with Idiopathic Hypogonadotropic Hypogonadism and Kallmann Syndrome*. ***Mol Cell Endocrinol.*** 2011; 346(1-2):74-83.

20. Talkowski ME, Rosenfeld JA, Chiang C, Blumenthal I, Pillalamarri V, Heilbut A, Ernst C, Hanscom C, Rossin E, Lindgren A, Pereira S, Ruderfer D, Kirby A, Zhang Y, Ripke S, Harris D, Lee JH, Ha K, **Kim HG**, Solomon BD, Gropman AL, Lucente D, Sims K, Ohsumi TK, Borowsky ML, Loranger S, Quade B, Lage K, Miles J, Wu BL, Shen Y, Neale B, Shaffer LG, Daly MJ, Morton CC, Gusella JF. *Sequencing chromosomal abnormalities reveals neurodevelopmental loci that confer risk across diagnostic boundaries.* ***Cell*** 2012;149(3):525-537.
21. **Kim HG (corresponding author)**, Kim HT, Leach NT, Lan F, Ullmann R, Silahtaroglu A, Kurth I, Nowka A, Seong IS, Shen Y, Talkowski ME, Ruderfer D, Lee JH, Glotzbach C, Ha K, Kjaergaard S, Levin AV, Romeike BF, Kleefstra T, Bartsch O, Elsea SH, Jabs EW,MacDonald ME, Harris DJ, Quade BJ, Ropers HH, Shaffer LG, Kutsche K, Layman LC, Tommerup N, Kalscheuer VM, Shi Y, Morton CC, Kim CC, Gusella JF. *Translocations disrupting PHF21A in the Potocki-Shaffer syndrome region are associated with intellectual disability and craniofacial anomalies*. ***Am J Hum Genet*** 2012 July; 91(1): 56-72.
22. Nguyen LS, **Kim HG**, Rosenfeld JA, Shen Y, Gusella JF, Lacassie Y, Layman LC, Shaffer LG, Gecz J. *Contribution of copy number variants involving nonsense-mediated mRNA decay pathway genes to neuro-developmental disorders*. ***Hum Mol Genet*** 2013 May; 22(9):1816-1825.
23. Newbern K, Natrajan N, **Kim HG**, Chorich LP, Halvorson LM, Cameron RS, Layman LC. *Identification of HESX1 mutations in Kallmann syndrome.* ***Fertility & Sterility*** 2013 June;99(7):1831-1837.
24. Quaynor SD, Stradtman EW Jr., **Kim HG**, Shen Y, Chorich LP, Schreihofer DA, Layman LC. *Delayed puberty and estrogen resistance in a woman with estrogen receptor variant*. ***N Engl J Med.*** 2013;369(2):164-171.
25. Siegel E, **Kim HG**, Nishimoto HK, Layman LC. *The Molecular Basis of Impaired Follicle Stimulating Hormone Action—Evidence from Human Mutations and Mouse Models*. ***Reprod. Sci*** 2013;20(3):211-33.
26. Quaynor SD, Goldberg LY, Ko EK, Stanley RK, Demir D, **Kim HG**, Chorich LP, Cameron RS, Layman LC. *Differential Expression of Nasal Embryonic LHRH Factor (NELF) Variants in Immortalized GnRH Neuronal Cell Lines*. ***Mol Cell Endocrinol.*** 2014 March;383(1-2):32-7.
27. Romeike BFM, Shen Y, Nishimoto HK, Morton CC, Layman LC, **Kim HG**. *Spectrum of genes involved in a unique case of Potocki-Shaffer syndrome with a large chromosome 11 deletion.* ***Clin Neuropathol*** 2014 May-Jun;33(3):238-44.
28. Bhagavath B, Layman LC, Ullmann R, Shen Y, Ha K, Looney S, Rehman K, McDonough PG, **Kim HG**, Carr BR. *Familial 46,XY sex reversal without campomelic dysplasia caused by a deletion upstream of the SOX9 gene.* ***Mol Cell Endocrinol.*** 2014 Aug 5;393(1-2):1-7.
29. Jun KR, Ullmann R, Khan S, Layman LC, **Kim HG (corresponding author)**. *Interstitial Microduplication at 2p11.2 in a Patient with Syndromic Intellectual Disability: 30-year Follow-up.* ***Mol Cytogenet*** 2014 Aug 19; 7:52.
30. Nishimoto HK, Ha K, Jones JR, Dwivedi A, Cho HM, Layman LC, **Kim HG (corresponding author)**. *The Historical Coffin-Lowry Syndrome Family Revisited: Identification of Two Novel Mutations of RPS6KA3 in Three Male Patients.* ***Am J Med Genet*** A 2014 Sep;164(9):2172-9.
31. Minocherhomji S, Hansen C, **Kim HG**, Mang Y, Bak M, Guldberg P, Eiberg H, Doh GD, Møllgård K, Hertz JM, Nielsen JE, Ropers HH, Tümer Z, Tommerup N, Kalscheuer VM, Silahtaroglu A. *Epigenetic re-modelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia*. ***Hum Mol Genet*** 2014 Dec;23(23):6163-76.
32. Dobek WA, **Kim HG**, Walls CA, Chorich LP, Tho SPT, Wang ZX, McDonough PG, Layman LC. *Long-term Follow-up of Females with Unbalanced X;Y Translocations—Reproductive and Nonreproductive Consequences.* ***Mol Cytogenet*** 2015 Feb; 8:13.

33. Liu YF, Sowell SM, Luo Y, Chaubey A, Cameron RS, **Kim HG**, and Srivastava AK. *Autism and Intellectual disability-associated synaptic protein KIRREL3 interacts with MAP1B and MYO16 with roles in neurodevelopment*. ***PLoS One*** 2015 April; 10(4):e0123106.
34. Quaynor SD, Ko EK, Chorich LP, Sullivan ME, Demir D, Waller JL, **Kim HG**, Cameron RS, Layman LC. *NELF knockout is associated with impaired pubertal development and subfertility*. Mol Cell Endocrinol. 2015 May ;407:26-36.
35. Labonne JDJ, Vogt J, Reali R, Kong IK, Layman LC, **Kim HG (corresponding author)**. *A microdeletion encompassing PHF21A in an individual with global developmental delay and craniofacial anomalies.* ***Am J Med Genet*** 2015 Sep 3.
36. Labonne JDJ, Chong MJ, Jones JR, Anand P, Wenzel W, Iacoboni D, Lawman LC, **Kim HG** (**corresponding author**). *Concomitant Partial Exon Skipping by a Unique Missense Mutation of RPS6KA3 causes Coffin-Lowry syndrome*. ***GENE*** 2015 Aug 20.
37. Choi JH, Balasubramanian R, Lee PH, Shaw ND, Hall JE, Plummer L, Buck CL, Kottler ML, Jarzabek K, Wołczynski S, Quinton R, Latronico AC, Dode C, Ogata T, **Kim HG**, Layman LC, Gusella JF, Crowley Jr. WF. *Expanding the Spectrum of Founder Mutations Causing Isolated Gonadotropin-Releasing Hormone Deficiency*. ***J Clin Endocrinol Metab*** 2015 Oct;100(10):E1378-85.
38. Labonne JDJ, Lee KH, Iwase S, Diamond MP, Layman LC, Kim CH, **Kim HG (corresponding author)**. *An atypical 12q24.31 microdeletion implicates six genes including a histone demethylase KDM2B and a histone methyltransferase SETD1B in syndromic intellectual disability*. ***Human Genetics*** 2016 April 22.
39. Labonne JDJ, Shen Y, Diamond MP, Layman LC, Kim CH, **Kim HG (corresponding author)**. *Comparative deletion mapping at 1p31.3-p32.2 implies NFIA responsible for intellectual disability coupled with macrocephaly and the presence of several other genes for syndromic intellectual disability.* ***Mol Cytogenet*** 2016 Mar 17;9:24.
40. Labonne JDJ, Graves TD, Shen Y, Jones JR, Kong IK, Layman LC, **Kim HG (corresponding author)**. *A microdeletion at Xq22.2 implicates a glycine receptor GLRA4 responsible in learning disability, behavioral problems and craniofacial anomalies*. ***BMC Neurology*** 2016 Aug 9;16:132.
41. Ha K, Anand P, Lee JA, Jones JR, Kim CA, Bertola DR, Labonne JDJ, Wenzel W, Layman LC,Kim **HG (corresponding author)**. *Steric clash in the SET domain of histone methyltransferase NSD1 as a cause of Sotos syndrome and its genetic heterogeneity in a Brazilian cohort*. ***GENES (Basel)***. 2016 Nov 9;7(11).
42. Williams LS, **Kim HG**, Kalscheuer VM, Tuck M, Chorich LP, Sullivan ME, Falkenstrom A, Reindollar RH, Layman LC. *A balanced chromosomal translocation involving chromosomes 3 and 16 in a patient with Mayer- Rokitansky-Kuster-Hauser syndrome reveals new candidate genes at 3p22.3 and 16p13.3.* ***Mol Cytogenet*** 2016 Jul 30;9:57.
43. Quaynor SD, Bosley ME, Duckworth CG, Porter KR, Kim SH, **Kim HG**, Chorich LP, Sullivan ME, Choi JH, Cameron RS, Layman LC. *Targeted next generation sequencing approach identifies fifteen new candidate genes in normosmic hypogonadotropic hypogonadism and Kallmann syndrome*. ***Mol Cell Endocrinol***. 2016 Dec 5;437:86-96.
44. Ha K, Shen Y, Graves T, Kim CH, **Kim HG (corresponding author)**. *The presence of two rare genomic syndromes, 1q21 deletion and Xq28 duplication, segregating independently in a family with intellectual disability.* ***Mol Cytogenet***. 2016 Sep 29;9:74.
45. Yoo KW, Thiruvarangan M, Jeong YM, Lee MS, Maddirevula S, Rhee M, Bae YK, **Kim HG**, Kim CH. *Mind Bomb-Binding Partner RanBP9 Plays a Contributory Role in Retinal Development.* ***Mol Cells***. 2017 Apr;40(4):271-279.
46. Porter RS, Murata-Nakamura Y, Nagasu H, **Kim HG**, Iwase S. *Transcriptome analysis revealed impaired cAMP responsiveness in PHF21A-deficient human cells.* ***Neuroscience***. 2017 May 29. pii: S0306-4522(17)30365-2.
47. Williams LS, Demir Eksi D, Shen Y, Lossie AC, Chorich LP, Sullivan ME, Phillips JA 3rd, Erman M, **Kim HG**, Alper OM, Layman LC. *Genetic analysis of Mayer-Rokitansky-Kuster-Hauser syndrome in a large cohort of families.* ***Fertil Steril***. 2017 Jul;108(1):145-151.e2.
48. Kim OH, Cho HJ, Han E, Hong TI, Ariyasiri K, Choi JH, Hwang KS, Jeong YM, Yang SY, Yu K, Park DS, Oh HW, Davis EE, Schwartz CE, Lee JS, **Kim HG (co-corresponding author)**, Kim CH. *Zebrafish knockout of Down syndrome gene, DYRK1A, shows social impairments relevant to autism.* ***Mol Autism.*** 2017 Sep 29;8:50.

49. Choi JH, Jeong YM, Kim S, Lee B, Ariyasiri K, Kim HT, Jung SH, Hwang KS, Choi TI, Park CO, Huh WK, Carl M, Rosenfeld JA, Raskin S, Ma A, Gecz J, **Kim HG**, Kim JS, Shin HC, Park DS, Gerlai R, Jamieson BB, Kim JS, Iremonger KJ, Lee SH, Shin HS, Kim CH. *Targeted knockout of a novel chemokine-like gene increases anxiety and fear behavior.* ***PNAS***. 2018 Jan 30;115(5):E1041-E1050.
50. Kim YJ, Osborn DP, Lee JY, Araki M, Araki K, Mohun T, Känsäkoski J, Brandstack N, Kim HT, Miralles F, Kim CH, Brown NA, **Kim HG**, Martinez-Barbera JP, Ataliotis P, Raivio T, Layman LC, Kim SH. *WDR11-mediated Hedgehog signalling defects underlie a new ciliopathy related to Kallmann syndrome.* ***EMBO Rep***. 2018 Feb;19(2):269-289.
51. Liu ZZ, Wang ZL, Choi TI, Huang WT, Wang HT, Han YY, Zhu LY, Kim HT, Choi JH, Lee JS, **Kim HG**, Zhao J, Chen Y, Lu Z, Tian XL, Pan BX, Li BM, Kim CH, Xu H. *Chd7 Is Critical for Early T-Cell Development and Thymus Organogenesis in Zebrafish.* ***Am J Pathol.*** 2018 Jan 31. pii: S0002-9440(17)30669-7.
52. Demir Eksi D, Shen Y, Erman M, Chorich LP, Sullivan ME, Bilekdemir M, Yılmaz E, Luleci G, Kim HG, Alper OM, Layman LC. *Copy number variation and regions of homozygosity analysis in patients with MÜLLERIAN aplasia.* ***Mol Cytogenet.*** 2018 Feb 3;11:13.
53. Aslanzadeh M, Ariyasiri K, Kim OH, Choi TI, Lim JH, Kim HG, Gerlai R, Kim CH, *The Body Size of Stimulus Conspecifics Affects Social Preference in a Binary Choice Task in Wild-Type, But Not in dyrk1aa Mutant, Zebrafish.* ***Zebrafish.*** 2019 Jun;16(3):262-2
54. **Kim HG* (corresponding author)**, Rosenfeld JA, Scott DA, Bénédicte G, Labonne JD, Brown J, McGuire M, Mahida S, Naidu S, Gutierrez J, Lesca G, des Portes V, Bruel AL, Sorlin A, Xia F, Capri Y, Muller E, McKnight D, Torti E, Rüschendorf F, Hummel O, Islam Z, Kolatkar PR, Layman LC, Ryu D, Kong IK, Madan-Khetarpal S, Kim CH. *Disruption of PHF21A causes syndromic intellectual disability with craniofacial anomalies, epilepsy, hypotonia, and neurobehavioral problems including autism.* ***Molecular Autism***, 2019 Oct 22;10:35.
55. Johnson JL, Stoica L, Liu Y, Zhu PL, Bhattacharya A, Buffington S, Huq R, N. Eissa NT, Larsson O, Porse BT, Domingo D, Nawaz U, Carroll R, Jolly L, Scerri TS, **Kim HG**, Brignell A, Coleman MJ, Braden R, Kini U, Jackson V, Baxter A, Bahlo M, Scheffer IE, Amor DJ, Hildebrand MS, Bonnen PE, Beeton C, Gecz J, Morgan AT, and Costa-Mattioli M. *Inhibition of Upf2-dependent nonsense-mediated decay leads to behavioral and neurophysiological abnormalities by activating the immune response.* ***Neuron***. 2019 Oct 1. pii: S0896-6273(19)30733-0.
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61. Sangare M, Diarra YA, Konate D, Haidara MS, Dolo H, Keita B, Doumbia F, Adama Karembe A, Kouyate M, Sidibe O, Diakite SAS, Kayentao K, Traore D, Doumbia S, Diakite M, **Kim HG**, Awandare GA. "Autism specrtum disorders in Mali: are parental concerns about child's developmental milestones associated with the age of diagnostic?". ***J Autism Dev Disord***. September 2019, Submitted.

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BOOK

1. **Hyung-Goo Kim**. Positional Cloning for Kallmann and Potocki-Shaffer Syndrome---disease gene discovery using breakpoint mapping in human patients with balanced chromosome translocations, Lambert Academic Publishing, 2013.

NEWSPAPER

1. Emily Rose Bennett, “Fish provide proof of genetic defect that leads to brain, skull malformations”, The Augusta Chronicle, July 5, 2012.
2. Tom Corwin, “Augusta University conference looks at disability desire disorder”, The Augusta Chronicle March 30, 2016.

JOURNAL RESPONSIBILITIES

Editorial Board member: Women's Health International / November 2013-present
Editorial Board member: Science Postprint / October 2013-present
Editorial Board member: Journal of Gynecology and Obstetrics Medicine / April 2014-present
Editorial Board member: Scholarena Journal of Genetics / August 2014-present
Editorial Board member: Journal of Cytology and Histology / September 2014-present
Editorial Board member: Journal of Pediatrics and Child Health Care / November 2015-present
Ad Hoc reviewer: Fertility & Sterility: 2012-present
Ad Hoc reviewer: Reproductive Sciences: 2012-present
Ad Hoc reviewer: Journal of Pediatric Genetics: 2013-present
Ad Hoc reviewer: American Journal of Medical Genetics: 2014-present
Ad Hoc reviewer: Clinical Genetics: 2014-present
Ad Hoc reviewer: Journal of Depression and Anxiety: 2015-present
Ad Hoc reviewer: Human Molecular Genetics: 2015-present
Ad Hoc reviewer: European Journal of Medical Genetics: 2015-present
Ad Hoc reviewer: Journal of Molecular Syndromology: 2016-present
Ad Hoc reviewer: Journal of Autism and Epilepsy: 2016-present
Ad Hoc reviewer: Global Journal of Genetics and Gene Therapy: 2016-present
Ad Hoc reviewer: Human Genetics: 2017-present
J Ad Hoc reviewer: JSM Oro Facial Surgeries: 2017-present
Ad Hoc reviewer: International Journal of Molecular Sciences: 2017-present
Ad Hoc reviewer: Molecular Genetics and Genomics: 2018-present
Ad Hoc reviewer: International Journal of Environmental Research & Public Health: 2018-present

RESEARCH SUPPORT

Current Research Funding

Neurological Disorders Research Center, Qatar Biomedical Research Institute (QBRI)
Start-up Package 200K/year started on August 2018
PI: Hyung-Goo Kim at the QBRI

Pending Research Support

Qatar National Research Fund
NPRP12S-0224-190141
Genetic Underpinnings of Autosomal Recessive Intellectual Disability
200K/year for 5 years

Past Research Application Not Funded

1. 1 R01 HD093549-01 submitted in November 2017
   Score at the first submission: 37%
   2018-2022 NIH/NICHD
   Title: Genetic Underpinnings of Autosomal Recessive Intellectual Disability
   PI: Hyung-Goo Kim at the Augusta University

2. 1 R01 HD090668-01A1 resubmitted at August 2016
   2016-2020 NIH/NICHD
   Title: Genetics of Autosomal Dominant Intellectual Disability
   PI: Hyung-Goo Kim at the Augusta University
   Scored at 19%

3. R01HD087403 resubmitted at October 2017
   (PI: Dr. Lawrence Layman at the Augusta University)
   2016-2020 NIH/NICHD
   Title: Molecular Basis of Uterine and Vaginal Development
   Co-Investigator: Hyung-Goo Kim

Completed Research Support

1. R01HD033004 (PI: Dr. Lawrence Layman at the Augusta University)
   2010-2015 NIH/NICHD
   Title: Genetics of Delayed Puberty
   Co-Investigator: Hyung-Goo Kim

2. R21GM119016-01 2016
   (PI: Graydon Gonsalvez at Augusta University)
   2016-2017 NIH/NHGRI
   Title: Using Cell, Fly, and Zebrafish Models to Understand Fam109A Gene Function In Undiagnosed Disease
   Co-Investigator: Hyung-Goo Kim at the Augusta University

3. Philadelphia Scientifics
   Title: Genetic Etiology of Balanced Chromosomal Rearrangements

65K annual directs: April 2016-April 2018
PI: Hyung-Goo Kim at the Augusta University

4. Department of Obstetrics & Gynecology Start-up Package
600K from February 2013 to April 2018
PI: Hyung-Goo Kim at the Augusta University

STUDENTS /RESIDENTS/FELLOWS MENTORED

1. Radhika Subramanian, Third Year Ob/Gyn Resident, MCG, Molecular Reproductive Endocrinology Lab, November-December 2006
2. Anna Grove, MCG STAR Student from Lenoir-Rhyne College, May 2007-July 2007. 2007 J. Charles Eldridge Award for the Outstanding STAR Student
3. Ekta Shah, Medical School Graduate, applying for residency, July 2007- July 2008
4. Javed Patka, Georgia Tech graduate, working in lab, July 2007- June 2008
5. Edward Botse-Baidoo, Internal Medicine Resident, November 2007-June 2008
6. Kathryn Stackhouse, MCG STAR Student from Elon College, May 2008-July 2009, M. Ebad Hasan Memorial Award
7. Juliana Coleman, MCG STAR Student from Clemson University, May 2009-July 2009.
8. Ashley Foster, first year MCG medical student, Summer Dean's Research Grant, June 2009-August 2009; additional times 2010 - 2012
9. Lindsey Goldberg, MCG medical student, July 2009 – May 2010
10. Elizabeth Cappello, MCG STAR Student from Emory University, May 2000-July 2010
11. Samuel Quaynor, MD/PhD Student, Medical College of Georgia, September 2010 – July 2013, successfully defended thesis; graduated May 2015, Neurology Residency, University of Chicago
12. Kate Griewisch, GHSU STAR Student from Lenoir Rhyne College, May 2011-July 2011, J. Charles Eldridge Award for the Outstanding STAR Student
13. Hiromi Koso Nishimoto, MD (Apr 2011-Apr 2013), postdoctoral student
14. Resident advisor for Class of 2014 GHSU Ob/Gyn Residents (Jennifer Tomlinson, Jessica Williams, Anxhela Treska, and Chandricka Rao), July 2011 – Apr 2018
15. Ellison Taylor, GHSU STAR Student from Clemson University, May 2012 –July 2012
16. Quincy Zhong, GHSU Summer Dean's Research Grant, May 2012-August 2012
17. Kayce Newbern, GHSU Summer Dean's Research Grant, May 2012- August 2012
18. Maggie Bosley, GRU STAR Student from Wofford College, May 2013 – July 2013, Charles Eldridge Award for the Outstanding STAR Student, became MUSC medical student
19. Eun Kyung Ko, GRU Graduate Student, May 2013-June 2018 (PhD thesis project)
20. Luke Lee, second year medical student, May 2013 –December 2013
21. Hyun-Min Cho, PhD (Mar. 2013-May 2014), postdoctoral student
22. Kyungsoo Ha, PhD (Mar. 2013-May 2014), postdoctoral student
23. Pinang Shastri, Georgetown University Graduate Student, July 2013-June 2014, became MD of Touro College of Osteopathic Medicine in New York City
24. Jonathan D Labonne, PhD (Apr. 2014-Jun. 2016), postdoctoral student
25. Christina Duckworth, GRU STAR Student, May 2014 – July 2014
26. Allison Falkenstrom, Dean's Summer Research Student, June 2014 – August 2014
27. Lacey Williams, First year medical student, AOA grant submission, December 31,2014, (became ineligible when she converted to MD/PhD Program)
28. Rema Elmoustafa, GRU STAR Student, May 2015—July 2015
29. Tyler Graves, technician, July 2014-June 2015, became medical student of Edward Via College of Osteopathic Medicine, Spartanburg, SC
30. Sarah Seymour, High School Student from Boston, Laboratory Project, June –July 2015
31. Bithika Halder, High School Student from Augusta, Laboratory Project, June –July 2015
32. Marvin Harris, graduate student, Laboratory Project, Feb. 2016-Jul. 2018

33. Jason Brown, undergraduate student of Augusta University, volunteer, became a MD/PhD student at Augusta University, Sep 2016-Apr 2018
34. Vattika Sivised, undergraduate student of Augusta University, volunteer, Sep 2016- Apr 2018
35. Connor Owens, undergraduate student of Augusta University, volunteer, May 2017- Apr 2018
36. Deema Jallad, MS, Qatar Biomedical Research Institute, Research Associate, March 2019-Sep 2019
37. Afif Ben Mahmoud, PhD, Qatar Biomedical Research Institute, Senior Research Associate, September 2019-present

RESEARCH ACCOMPLISHMENTS

I have a long standing interest in neurogenetics and specifically positional cloning of disease genes. My most significant contribution to this field has been the identification of novel disease genes and their underlying mechanisms which lead to understanding the molecular and physiological basis of genetic disorders. I became fascinated with positional cloning using chromosome rearrangements when I was a PhD student with Dr. Hans-Hilger Ropers at Max-Planck-Institute for Molecular Genetics (1991-2001), and then following as a research fellow in Dr. James Gusella's lab at Harvard Medical School (2001-2006).

Autism is a neurodevelopmental disorder of complex etiology in which genetic factors play a major role. Using positional cloning I identified Neurexin 1 (NRXN1) as the causative gene for autism in two independent patients with balanced chromosome translocations involving 2p16.3, where this gene was truncated or dysregulated. Furthermore, after screening this giant gene in a cohort of 74 autism patients, I identified two missense mutations of this gene (**HG Kim et al., AJHG 2008**). In the subsequent eight years, that single gene study has been cited 545 times and has been described in developmental, psychiatric, and basic neuroscience research.

NRXN1 is one of the largest known human genes and encodes a highly polymorphic cell surface receptor that influences synaptic activity and also contributes to intellectual disability and schizophrenia. Continuing investigation has revealed that this gene is in the downstream of PTPRD, a novel gene we have just identified to cause syndromic intellectual disability and autism in 9p23 microdeletion syndrome (manuscript entitled "Loss-of-function mutations of PTPRD cause syndromic intellectual disability and autism in 9p23 microdeletion syndrome" in preparation to *AJHG*)

In order to study genes involved in reproductive disorders, I moved to Dr. Lawrence Layman's lab at the Medical College of Georgia at Augusta University, where I identified two splice site and five missense mutations in the gene CHD7 (chromodomain helicase DNA binding protein 7) in three Kallmann syndrome (KS) patients and four patients with normosmic hypogonadotropic hypogonadism (nHH), indicating that these disorders of delayed puberty are mild allelic variants of CHARGE syndrome. CHD7 represents the first identified chromatin-remodeling protein with a role in human puberty and the second gene to cause both normosmic HH and KS in humans (**HG Kim et al., AJHG 2008**). CHD7 rivals FGFR1 as the most frequent cause of this heterogeneous disorder of delayed puberty and my CHD7 paper has been cited more than 182 times. Further investigation in DiGeorge syndrome patients and zebrafish knockdown has resulted in the identification of CHD7 as a novel disease gene of DiGeorge syndrome (manuscript entitled "Genetic Mutations and Zebrafish Knockdown Identified CHD7 as a Cause of DiGeorge Syndrome" in preparation to *AJHG*).

In 2010, by defining the chromosomal breakpoint of a balanced translocation patient and scanning genes in its vicinity in unrelated hypogonadal subjects, I identified WDR11 with a double propeller structure as a gene involved in human puberty. Furthermore, I discovered that WDR11 interacts with EMX1, a homeodomain transcription factor involved in the development of olfactory neurons, and that missense mutations reduce or abolish this interaction. These findings suggested that a deficiency of productive WDR11 protein interaction is an underlying mechanism of impaired pubertal development in these patients (**HG Kim et al., AJHG 2010**). Currently we are investigating its role in primordial germ cell migration and Hedgehog signal pathway using

Wdr11 KO mice and Stella-GFP mice to uncover the molecular mechanisms of delayed puberty in collaboration with Dr. Soo-Hyun Kim at St George's Medical School, University of London.
Despite the reported identification of two individual genes responsible for multiple exostoses and parietal foramina in Potocki-Shaffer syndrome (PSS), the genetic etiology of the gene associated with intellectual disability and craniofacial anomaly phenotypes has remained elusive. Through characterization of independent subjects with balanced chromosome translocations and comparative deletion mapping of PSS subjects, I have uncovered evidence that the intellectual disability and craniofacial anomalies in PSS are both caused by haploinsufficiency of a single gene, PHF21A, at 11p11.2. PHF21A encodes a plant homeodomain finger protein, and suppression of the zebrafish ortholog led to both craniofacial abnormalities and neuronal apoptosis.

Along with lysine-specific demethylase 1 (KDM1A/ LSD1), PHF21A, also known as BHC80, is a component of the BRAF-histone deacetylase complex that represses target-gene transcription. In lymphoblastoid cell lines from two balanced translocation subjects in whom PHF21A was directly disrupted, I observed derepression of the neuronal gene SCN3A and reduced KDM1A occupancy at the SCN3A promoter, supporting a direct functional consequence of PHF21A haploinsufficiency on transcriptional regulation (**HG Kim et al., AJHG 2012**). We have recently found seven point mutations of *PHF21A* in syndromic intellectual disability patients (Disruption of PHF21A causes syndromic intellectual disability with craniofacial anomalies, epilepsy, hypotonia, and neurobehavioral problems including autism. **Molecular Autism**, October 22 2019 published).