Curriculum vitae

NAME	Martin Kircher
BORN	1983 in Erfurt, Germany
STUDIES	09/2007 - 07/2011: Dr. rer. nat. (PhD) in Computer Sciences (summa cum laude) at Universität Leipzig, Germany Thesis: *Understanding and improving high-throughput sequencing data production and analysis*
	04/2006 - 07/2007: Master of Science (Honor's Degree) at Saarland University, Germany Computational Molecular Biology Thesis: *In silico analysis of microRNA genes on human chr. 14 and mouse chr. 12*
	10/2003 - 03/2006: Bachelor of Science at Saarland University, Germany Bioinformatics
AFFILIATIONS	since 04/2012: Jay Shendure, PhD, MD Department of Genome Sciences, University of Washington, Seattle, WA, USA Senior fellow
	06/2012 - 11/2015: Center for Mendelian Genomics at the University of Washington, Seattle, WA, USA Lead data analyst
	09/2007 - 03/2012: Janet Kelso, PhD & Prof. Dr. Svante Pääbo Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany PhD student (Bioinformatics, Evolutionary Genetics) (09/2007 - 02/2011) Research scientist (Bioinformatics, Evolutionary Genetics) (03/2011 – 03/2012)
	05/2007 - 07/2007: Professor Dr. Thomas Lengauer, PhD Max Planck Institute for Informatics, Saarbrücken, Germany Student assistant
	09/2006 - 10/2006: Professor Dr. Andreas Zeller Software Engineering Chair, Saarland University, Germany Tutor of practical training in software design
	08/2005 - 01/2006: Professor Dr. Hans-Peter Lenhof Center for Bioinformatics, Saarland University, Germany Student assistant
VISITS	09/2010 - 11/2010, 11/2009: Dr. Philipp Khaitovich CAS-MPG Partner institute for Computational Biology, Shanghai, China Analysis of Illumina Digital Gene Expression data Consultant for high-throughput sequencing
	03/2008: Dr. Dirk Schübeler Friedrich Miescher Institute for Biomedical Research, Basel, Switzerland Measuring DNA methylation using MeDIP

08/2005 - 09/2005: Professor Dr. scient. Inge Jonassen
Computational Biology Unit, Bergen Center for Computational Science, Bergen, Norway
Web application for analyzing expression data

HONORS AND DISTINCTIONS

AAAS Newcomb Cleveland Prize 2009-2010

Doctoral Scholarship (2007-2010) of the International Max Planck Research School of Human Origins, Leipzig, Germany

Advancement award from Sparkassenstiftung Erfurt (2003)

Software Award of the Thuringian Minister of Education (2002)

Albert Schweitzer School Award for mathematics and computer sciences (2002)

2nd Award in the Thuringian state competition of youth research ("Jugend forscht") in geography and space sciences and a special award in software systems technology (2002)

Second Award of the 5th national physics competition (1999)

LANGUAGE SKILLS

German (native speaker)

English (fluent)

French (basic skills)

TECHNICAL SKILLS

Primary processing of Next Generation Sequencing data (mostly Illumina and 454)

Sequence analysis, i.e. Alignment; Comparative Genomics; Population Genetics; Gene expression analysis; Variant Calling; Variant Annotation, Scoring and Interpretation; Epigenetics and DNA modification/fragmentation/methylation; ancient DNA; cell-free DNA

Statistical learning (predictive models and classification)

Massively Parallel Reporter Assays (MPRA)

Python, R, bash, C/C++, HTML, PHP, SQL, LaTeX, PASCAL, and BASIC/VBA programming

Basic wet lab training (e.g. DNA extraction, PCR, gel and capillary electrophoresis, Cloning, NGS-library prep, Immunoprecipitation, Illumina sequencing)

Computer/network administration; usage and installation of software on Windows and Linux-based computer systems; usage of batch-queuing systems

Advanced user of MS Office (serial letters, Excel range formulas and pivot tables, VBA entry forms, etc.)

OTHER SKILLS

Digital and analog SLR photography

Representation of interests and counseling (e.g. student member of University board of examiners, spokesperson during community service, high school class representative)

Organization of events including financing (e.g. high school events, University of Washington PostDoc Association)

Coordination/creation of multimedia discs and print materials

Journal articles

[OA] – The full text is freely available on the publisher's website

First or shared first author publications

2016 F. Inoue, ***M. Kircher***, B. Martin, G.M. Cooper, D.M. Witten, M.T. McManus, N. Ahituv, J. Shendure. *A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity*, **Genome Research**, 2016 Nov 9. pii: gr.212092.116.

M.W. Snyder, ***M. Kircher***, A.J. Hill, R.M. Daza, and J. Shendure, *Cell-free DNA comprises an in vivo nucleosome footprint that informs its tissues-of-origin*, **Cell**, 2016 Jan 14; 164(1-2):57-68.

2015 ***M. Kircher*** & J. Shendure. *Running spell-check to identify regulatory variants*, **Nature Genetics**, 2015 Jul 29;47(8):853-5.

2014 [OA] S. Wunderlich, ***M. Kircher***, B. Vieth, A. Haase, S. Merkert, J. Beier, G. Gohring, S. Glage, A. Schambach, E.C. Curnow, S. Pääbo, , U. Martin, and W. Enard, *Primate iPS cells as tools for evolutionary analyses*, **Stem Cell Research**, 12, 622-629.

M. Kircher, D.M. Witten, P. Jain, B.J. O'Roak, G.M. Cooper, and J. Shendure, *A general framework for estimating the relative pathogenicity of human genetic variants*, **Nature Genetics**, 46, 310-315.

2012 M. Meyer, ***M. Kircher***, M. Gansauge, H. Li, F. Racimo, S. Mallick, J.G. Schraiber, F. Jay, K. Prüfer, C. de Filippo, P.H. Sudmant, C. Alkan, Q. Fu, R. Do, N. Rohland, A. Tandon, M. Siebauer, R.E. Green, K. Bryc, A.W. Briggs, U. Stenzel, J. Dabney, J. Shendure, J. Kitzman, M.F. Hammer, M.V. Shunkov, A.P. Derevianko, N. Patterson, A.M. Andrés, E.E. Eichler, M. Slatkin, D. Reich, J. Kelso, and S. Pääbo. *A high coverage genome sequence from an archaic Denisovan individual*, **Science**, 2012 Aug 31.

***M. Kircher**, Analysis of high-throughput ancient DNA sequencing data*, **Methods in Molecular Biology**, 2012;840:197-228; also as book chapter in "Methods in Molecular Biology: Ancient DNA" edited by Beth Shapiro and Michael Hofreiter.

2011 [OA] ***M. Kircher***, S. Sawyer, and M. Meyer, *Double indexing overcomes inaccuracies in multiplex sequencing on the Illumina platform*, **Nucleic Acids Research**, doi:10.1093/nar/gkr771.

[OA] ***M. Kircher***, P. Heyn, and J. Kelso, *Addressing challenges in the production and analysis of Illumina sequencing data*, **BMC Genomics**, 12(1): p. 382.

2010 [OA] D. Reich, R.E. Green, ***M. Kircher***, J. Krause, N. Patterson, E.Y. Durand, B. Viola, A.W. Briggs, U. Stenzel, P.L.F. Johnson, T. Maricic, J.M. Good, T. Marques-Bonet, C. Alkan, Q. Fu, S. Mallick, H. Li, M. Meyer, E.E. Eichler, M. Stoneking, M. Richards, S. Talamo, M.V. Shunkov, A.P. Derevianko, J.-J. Hublin, J. Kelso, M. Slatkin, and S. Pääbo, *Genetic history of an archaic hominin group from Denisova Cave in Siberia*, **Nature**, 468(7327):1053-1060.

M. Kircher and J. Kelso, *High-throughput DNA sequencing-concepts and limitations*, **BioEssays**, 32(6):524-536.

2009 [OA] ***M. Kircher***, U. Stenzel, and J. Kelso, *Improved base calling for the Illumina Genome Analyzer using machine learning strategies*, **Genome Biology**, 10(8):R83.

2008 [OA] ***M. Kircher,*** C. Bock, and M. Paulsen, *Structural conservation versus functional divergence of maternally expressed microRNAs in the Dlk1/Gtl2 imprinting region*, **BMC Genomics**. Jul 23;9:346.

Additional publications

2016 G. Mirzaa, A.E. Timms, V. Conti, E.A. Boyle, K.M. Girisha, B. Martin, ***M. Kircher***, C. Olds, J. Juusola, S. Collins, K. Park, M. Carter, I. Glass, I. Krägeloh-Mann, D. Chitayat, A.S. Parikh, R. Bradshaw, E. Torti, S. Braddock, L. Burke, S. Ghedia, M. Stephan, F. Stewart, C. Prasad, M. Napier, S. Saitta, R. Straussberg, M. Gabbett, B.C. O'Connor, C.E. Keegan, L.J. Yin, A.H. Lai, N. Martin, M. McKinnon, M.C. Addor, L. Boccuto, C.E. Schwartz, A. Lanoel, R.L. Conway, K. Devriendt, K. Tatton-Brown, M.E. Pierpont, M. Painter, L. Worgan, J. Reggin, R. Hennekam, K. Tsuchiya, C.C. Pritchard, M. Aracena, K.W. Gripp, M. Cordisco, H.V. Esch, L. Garavelli, C. Curry, A. Goriely, H. Kayserilli, J. Shendure, J. Jr Graham , R. Guerrini, and W.B. Dobyns, *PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution*, **JCI Insight**, 2016 Jun 16;1(9). pii: e87623.

P.G. Wheeler, B.G. Ng, L. Sanford, V.R. Sutton, D.W. Bartholomew, M.T. Pastore, M.J. Bamshad, ***M. Kircher***, K.J. Buckingham, D.A. Nickerson, J. Shendure and H.H. Freeze, *SRD5A3-CDG: Expanding the phenotype of a congenital disorder of glycosylation with emphasis on adult onset features*, **American Journal of Medical Genetics Part A**, 2016 Aug 2. doi: 10.1002/ajmg.a.37875.

A.M. Alazami, S.M. Al-Qattan, E. Faqeih, A. Alhashem, M. Alshammari, F. Alzahrani, M.S. Al-Dosari, N. Patel, A. Alsagheir, B. Binabbas, H. Alzaidan, A. Alsiddiky, N. Alharbi, M. Alfadhel, A. Kentab, R.M. Daza, ***M. Kircher***, J. Shendure, M. Hashem, S. Alshahrani, Z. Rahbeeni, O. Khalifa, R. Shaheen and F.S. Alkuraya, *Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue*, **Human Genetics**, 2016 May;135(5):525-40. doi: 10.1007/s00439-016-1660-z

B.G. Ng, S.A. Shiryaev, D. Rymen, E.A. Eklund, K. Raymond, ***M. Kircher***, J.E. Abdenur, F. Alehan, A.T. Midro, M.J. Bamshad, R. Barone, G.T. Berry, J.E. Brumbaugh, K.J. Buckingham, K. Clarkson, F.S. Cole, S. O'Connor, G.M. Cooper, R. Van Coster, L.A. Demmer, L. Diogo, A.J. Fay, C. Ficicioglu, A. Fiumara, W.A. Gahl, R. Ganetzky, H. Goel, L.A. Harshman, M. He, J. Jaeken, P.M. James, D. Katz, L. Keldermans, M. Kibaek, A.J. Kornberg, K. Lachlan, C. Lam, J. Yaplito-Lee, D.A. Nickerson, H.L. Peters, V. Race, L. Régal, J.S. Rush, S.L. Rutledge, J. Shendure, E. Souche, S.E. Sparks, P. Trapane, A. Sanchez-Valle, E. Vilain, A. Vøllo, C.J. Waechter, R.Y. Wang, L.A. Wolfe, D.A. Wong, T. Wood, A.C. Yang, University of Washington Center for Mendelian Genomics, G. Matthijs, and H.H. Freeze, *ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients*, **Human Mutation**, 2016 Jul;37(7):653-60. doi: 10.1002/humu.22983

M. Kuhlwilm, I. Gronau, M.J. Hubisz, C. de Filippo, J. Prado-Martinez, ***M. Kircher***, Q. Fu, H.A. Burbano, C. Lalueza-Fox, M. de la Rasilla, A. Rosas, P. Rudan, D. Brajkovic, Ž. Kucan, I. Gušic, T. Marques-Bonet, A.M. Andrés, B. Viola, S. Pääbo, M. Meyer, A. Siepel, and S. Castellano, *Ancient gene flow from early modern humans into Eastern Neanderthals*, **Nature**, 2016 Feb 25;530(7591):429-33.

B.G. Ng, S.A Shiryaev, D. Rymen, E.A. Eklund, K. Raymond, ***M. Kircher***, J.E. Abdenur, F. Alehan, A.T. Midro, M.J. Bamshad, R. Barone, G.T. Berry, JE Brumbaugh, K.J. Buckingham, K. Clarkson, F.S. Cole, S. O'Connor, G.M. Cooper, R. Van Coster, L.A. Demmer, L. Diogo, A.J. Fay, C. Ficicioglu, A. Fiumara, W.A. Gahl, R. Ganetzky, H Goel, LA Harshman, M He, J Jaeken, PM James, D Katz, L Keldermans, M Kibaek, AJ Kornberg, K. Lachlan, C. Lam, J. Yaplito-Lee, D.A. Nickerson, H.L. Peters, V. Race, L. Régal, J.S. Rush, SL Rutledge, J. Shendure, E. Souche, S.E. Sparks, P. Trapane, A. Sanchez-Valle, E. Vilain, A. Vøllo, C.J Waechter, R.Y. Wang, L.A. Wolfe, D.A. Wong, T. Wood, A.C. Yang, University of Washington Center for Mendelian Genomics, G. Matthijs, and H.H. Freeze, *ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients*, **Human Mutation**, 2016 Mar 2.

2015 R. Shaheen, N. Patel, H. Shamseldin, F. Alzahrani, R. Al-Yamany, A. ALMoisheer, N. Ewida, S. Anazi, M. Alnemer, M. Elsheikh, K. Alfaleh, M. Alshammari, A. Alhashem, A.A. Alangari, M.A. Salih, ***M. Kircher***, R.M. Daza, N. Ibrahim, S.M. Wakil, A. Alaqeel, I. Altowaijri, J. Shendure, A. Al-Habib, E. Faqieh, and F.S. Alkuraya, *Accelerating matchmaking of novel dysmorphology syndromes through clinical and genomic characterization of a large cohort*, **Genetics in Medicine**, 2015 Dec 3.

B.G. Ng, K. Raymond, ***M. Kircher***, K.J. Buckingham, T. Wood, J. Shendure, D.A. Nickerson, M.J. Bamshad, University of Washington Center for Mendelian Genomics, J.T. Wong, F.P. Monteiro, B.H. Graham, S. Jackson, R. Sparkes, A.E. Scheuerle, S. Cathey, F. Kok, J.B. Gibson, and H.H. Freeze, *Expanding the Molecular and Clinical Phenotype of SSR4-CDG*, **Human Mutation**, 2015 Aug 12.

J.X. Chong, K.J. Buckingham, S.N. Jhangiani, C. Boehm, N. Sobreira, J.D. Smith, T.M. Harrell, M.J. McMillin, W. Wiszniewski, T. Gambin, Z.H. Coban Akdemir, K. Doheny, A.F. Scott, D. Avramopoulos, A. Chakravarti, J. Hoover-Fong, D. Mathews, P.D. Witmer, H. Ling, K. Hetrick, L. Watkins, K.E. Patterson, F. Reinier, E. Blue, D. Muzny, ***M. Kircher***, K. Bilguvar, F. López-Giráldez, V.R. Sutton, H.K. Tabor, S.M. Leal, M. Gunel, S. Mane, R.A. Gibbs, E. Boerwinkle, A. Hamosh, J. Shendure, J.R. Lupski, R.P. Lifton, D. Valle, D.A. Nickerson, Centers for Mendelian Genomics, and M.J. Bamshad, *The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities*, **American J. of Human Genetics**, 2015 Jul 8.

H. Fairfield, A. Srivastava, G. Ananda, R. Liu, ***M. Kircher***, A. Lakshminarayana, B.S. Harris, S.Y. Karst, L.A. Dionne, C.C. Kane, M. Curtain, M.L. Berry, P.F. Ward-Bailey, I. Greenstein, C. Byers, A. Czechanski, J. Sharp, K. Palmer, P. Gudis, W. Martin, A. Tadenev, L. Bogdanik, C.H. Pratt, B. Chang, D.G. Schroeder, G.A. Cox, P. Cliften, J. Milbrandt, S. Murray, R. Burgess, D.E. Bergstrom, L.R. Donahue, H. Hamamy, A. Masri, F.A. Santoni, P. Makrythanasis, S.E. Antonarakis, J. Shendure, and L.G. Reinholdt, *Exome sequencing reveals pathogenic mutations in 91 strains of mice with Mendelian disorders*, **Genome Research**, 2015 Apr 27.

A. Garg, ***M. Kircher***, M. Del Campo, R.S. Amato, A.K. Agarwal, and University of Washington Center for Mendelian Genomics, *Whole exome sequencing identifies de novo heterozygous CAV1 mutations associated with a novel neonatal onset lipodystrophy syndrome*, **American Journal of Medical Genetics Part A**, 2015 Apr 21.

[OA] D.A. Hughes, ***M. Kircher***, Z. He, S. Guo, G.L. Fairbrother, C.S. Moreno, P. Khaitovich, and M. Stoneking, *Evaluating intra- and inter-individual variation in the human placental transcriptome*, **Genome Biology**, 2015 Mar 19;16:54.

S.L. Sawyer, L. Tian, M. Kähkönen, J. Schwartzentruber, ***M. Kircher***, University of Washington Centre for Mendelian Genomics, FORGE Canada Consortium, J. Majewski, D.A. Dyment, A.M. Innes, K.M. Boycott, L.A. Moreau, J.S. Moilanen, and R.A. Greenberg, *Biallelic mutations in BRCA1 cause a new Fanconi anemia subtype*, **Cancer Discovery**, 2015 Feb;5(2):135-42.

2014 S.S. Jamuar, A.T. Lam, ***M. Kircher***, A.M. D'Gama, J. Wang, B.J. Barry, X. Zhang, R.S. Hill, J.N. Partlow, A. Rozzo, S. Servattalab, B.K. Mehta, M. Topcu, D. Amrom, E. Andermann, B. Dan, E. Parrini, R. Guerrini, I.E. Scheffer, S.F. Berkovic, R.J. Leventer, Y. Shen, B.L. Wu, A.J. Barkovich, M. Sahin, B.S. Chang, M. Bamshad, D.A. Nickerson, J. Shendure, A. Poduri, T.W. Yu, and C.A. Walsh, *Somatic mutations in cerebral cortical malformations*, **New England Journal of Medicine**, 371(8):733-43.

M. Schubert, L. Ermini, C. Der Sarkissian, H. Jonsson, A. Ginolhac, R. Schaefer, M.D. Martin, R. Fernandez, ***M. Kircher***, M. McCue, E. Willerslev, and L. Orlando, *Characterization of ancient and modern genomes by SNP detection and phylogenomic and metagenomic analysis using PALEOMIX*, **Nature Protocols**, 9, 1056-1082.

S. Castellano, G. Parra, F.A. Sanchez-Quinto, F. Racimo, M. Kuhlwilm, ***M. Kircher***, S. Sawyer, Q. Fu, A. Heinze, B. Nickel, J. Dabney, M. Siebauer, L. White, H.A. Burbano, G. Renaud, U. Stenzel, C. Lalueza-Fox, M. de la Rasilla, A. Rosas, P. Rudan, D. Brajkovic, Z. Kucan, I. Gusic, M.V. Shunkov, A.P. Derevianko, B. Viola, M. Meyer J. Kelso, A.M. Andres, and S. Pääbo, *Patterns of coding variation in the complete exomes of three Neandertals*, **Proceedings of the National Academy of Sciences of the USA**, 111, 6666-6671.

M.R. Knowles, L.E. Ostrowski, M.W. Leigh, P.R. Sears, S.D. Davis, W.E. Wolf, M.J. Hazucha, J.L. Carson, K.N. Olivier, S.D. Sagel, M. Rosenfeld, T.W. Ferkol, S.D. Dell, C.E. Milla, S.H. Randell, W. Yin, A. Sannuti, H.M. Metjian, P.G. Noone, P.J. Noone, C.A. Olson, M.V. Patrone, H. Dang, H.S. Lee, T.W. Hurd, H.Y. Gee, E.A. Otto, J. Halbritter, S. Kohl, ***M. Kircher***, J. Krischer, M.J. Bamshad, D.A. Nickerson, F. Hildebrandt, J. Shendure, and M.A. Zariwala, *Mutations in RSPH1 cause primary ciliary dyskinesia with a unique clinical and ciliary phenotype*, **American Journal of Respiratory and Critical Care Medicine**, 189, 707-717.

[OA] P. Heyn, ***M. Kircher***, A. Dahl, J. Kelso, P. Tomancak, A.T. Kalinka, and K.M. Neugebauer, *The earliest transcribed zygotic genes are short, newly evolved, and different across species*, **Cell Reports**, 6, 285-292.

2013 K. Prüfer, F. Racimo, N. Patterson, F. Jay, S. Sankararaman, S. Sawyer, A. Heinze, G. Renaud, P.H. Sudmant, C. de Filippo, H. Li, S. Mallick, M. Dannemann, Q. Fu, ***M. Kircher***, M. Kuhlwilm, M. Lachmann, M. Meyer, M. Ongyerth, M. Siebauer, C. Theunert, A. Tandon, P. Moorjani, J. Pickrell, J.C. Mullikin, S.H. Vohr, R.E. Green, I. Hellmann, P.L. Johnson, H. Blanche, H. Cann, J.O. Kitzman, J. Shendure, E.E. Eichler, E.S. Lein, T.E. Bakken, L.V. Golovanova, V.B. Doronichev, M.V. Shunkov, A.P. Derevianko, B. Viola, M. Slatkin, D. Reich, J. Kelso, and S. Pääbo, *The complete genome sequence of a Neanderthal from the Altai Mountains*, **Nature**, 505, 43-49.

M.E. Losfeld, B.G. Ng, ***M. Kircher***, K.J. Buckingham, E.H. Turner, A. Eroshkin, J.D. Smith, J. Shendure, D.A. Nickerson, M.J. Bamshad, and H.H. Freeze, *A new congenital disorder of glycosylation caused by a mutation in SSR4, the signal sequence receptor 4 protein of the TRAP complex*, **Human Molecular Genetics**, 23, 1602-1605.

[OA] G. Renaud, ***M. Kircher***, U. Stenzel, and J. Kelso, *freeIbis: an efficient basecaller with calibrated quality scores for Illumina sequencers*, **Bioinformatics**, 2013 Mar 6.

B.G. Ng, K.J. Buckingham, K. Raymond, ***M. Kircher***, E.H. Turner, M. He, J.D. Smith, A. Eroshkin, M. Szybowska, M.E. Losfeld, J.X. Chong, M. Kozenko, C. Li, M.C. Patterson, R.D. Gilbert, D.A. Nickerson, J. Shendure, M.J. Bamshad, and H.H. Freeze, *Mosaicism of the UDP-Galactose Transporter SLC35A2 Causes a Congenital Disorder of Glycosylation*, **American Journal of Human Genetics***,* 92**,** 632-6.

[OA] J.M. Good, V. Wiebe, F.W. Albert, H.A. Burbano, ***M. Kircher***, R.E. Green, M. Halbwax, C. André, R. Atencia, A. Fischer, and S. Pääbo, *Comparative population genomics of the ejaculate in humans and the great apes*, **Molecular Biology and Evolution**, 2013 Apr;30(4):964-76. 2013 Jan 16.

2012 [OA] S.A. Fietz, R. Lachmann, H. Brandl, ***M. Kircher***, N. Samusik, R. Schröder, N. Lakshmanaperumal, I. Henry, J. Vogt, A. Riehn, W. Distler, R. Nitsch, W. Enard, S. Pääbo and W.B. Huttner. *Transcriptomes of germinal zones of human and mouse fetal neocortex suggest a role of extracellular matrix in progenitor self-renewal*, **Proceedings of the National Academy of Sciences of the USA**, 2012 Jul 17;109(29):11836-41.

[OA] L. Hering, M.J. Henze , M. Kohler, A. Kelber, C. Bleidorn, M. Leschke, B. Nickel, M. Meyer, ***M. Kircher***, P. Sunnucks, and G. Mayer. *Opsins in Onychophora (velvet worms) suggest a single origin and subsequent diversification of visual pigments in arthropods*, **Molecular Biology and Evolution**, June 7, mss148.

2011 [OA] N. Nagaraj, J.R. Wisniewski, T. Geiger, J. Cox, ***M. Kircher***, J. Kelso, S. Pääbo, and M. Mann, *Deep proteome and transcriptome mapping of a human cancer cell line*, **Molecular Systems Biology**, Nov 8;7:548.

Reich, D., N. Patterson, ***M. Kircher***, F. Delfin, M. R. Nandineni, I. Pugach, A. M. Ko, Y. C. Ko, T. A. Jinam, M. E. Phipps, N. Saitou, A. Wollstein, M. Kayser, S. Pääbo, and M. Stoneking, *Denisova admixture and the first modern human dispersals into Southeast Asia and Oceania*, **American Journal of Human Genetics**, 89:516-528.

D. Brawand, M. Soumillon, A. Necsulea, P. Julien, G. Csardi, P. Harrigan, M. Weier, A. Liechti, A. Aximu-Petri, ***M. Kircher***, F.W. Albert, U. Zeller, P. Khaitovich, F. Grützner, S. Bergmann, R. Nielsen, S. Pääbo, and H. Kässmann, *The evolution of gene expression levels in mammalian organs*, **Nature**, 478(7369):343-348.

2010 [OA] P. Heyn, U. Stenzel, A.W. Briggs, ***M. Kircher***, M. Hofreiter, and M. Meyer, *Road blocks on paleogenomes - polymerase extension profiling reveals the frequency of blocking lesions in ancient DNA*, **Nucleic Acids Research**, 38(16):e161.

M. Meyer and ***M. Kircher***, *Illumina sequencing library preparation for highly multiplexed target capture and sequencing*, **Cold Spring Harbor protocols**, 2010(6).

[OA] R. E. Green, J. Krause, A. W. Briggs, T. Maricic, U. Stenzel, ***M. Kircher***, N. Patterson, H. Li, W. Zhai, M.H. Fritz, N.F. Hansen, E.Y. Durand, A.S. Malaspinas, J.D. Jensen, T. Marques-Bonet, C. Alkan, K. Prüfer, M. Meyer, H.A. Burbano, J.M. Good, R. Schultz, A. Aximu-Petri, A. Butthof, B. Höber, B. Höffner, M. Siegemund, A. Weihmann, C. Nusbaum, E.S. Lander, C. Russ, N. Novod, J. Affourtit, M. Egholm, C. Verna, P. Rudan, D. Brajkovic, Z. Kucan, I. Gusic, V.B. Doronichev, L.V. Golovanova, C. Lalueza-Fox, M. de la Rasilla, J. Fortea, A. Rosas, R.W. Schmitz, P.L. Johnson, E.E. Eichler, D. Falush, E. Birney, J.C. Mullikin, M. Slatkin, R. Nielsen, J. Kelso, M. Lachmann, D. Reich, and S. Pääbo, *A draft sequence of the Neandertal genome*, **Science**, 328(5979):710-22. *(Shared second author)*

2009 [OA] A. W. Briggs, U. Stenzel, M. Meyer, J. Krause, ***M. Kircher***, and S. Pääbo, *Removal of deaminated cytosines and detection of in vivo methylation in ancient DNA*, **Nucleic Acids Research**, 38(6):e87.

J. Krause, A. W. Briggs, ***M. Kircher***, T. Maricic, N. Zwyns, A. Derevianko, and S. Pääbo, *A complete mtDNA genome of an early modern human from Kostenki, Russia*, **Current Biology**, Feb 9;20(3):231-6.

Scientific service

- Program Committee member of the European Conference on Computational Biology (ECCB) 2012, 2014, and 2016
- Reviewer for:
  - *American Association for the Advancement of Science (AAAS)*: Science
  - *BioMed Central (BMC)*: Bioinformatics, Genome Biology, Genomics
  - *Cell Press*: Trends in Genetics, Cell Systems
  - *Elsevier*: Journal of Human Evolution
  - *Public Library of Science (PLoS)*: ONE
  - *Multidisciplinary Digital Publishing Institute*: Biology
  - *National Academies*: Proceedings of the National Academy of Sciences of the United States of America, Proceedings of the National Academy of Sciences of India
  - *Nature Publishing Group*: Nature Methods, Nature Genetics
  - *Oxford Journals*: Briefings in Bioinformatics, Bioinformatics, Human Molecular Genetics, Nucleic Acids Research
  - *Springer Journals:* Human Genetics
  - *Wiley Periodicals:* Human Mutation

Patents

- "A framework for determining the relative effect of genetic variants" (PCT/US2014/056701)
- "Methods of determining tissues and/or cell types giving rise to cell-free DNA, and methods of identifying a disease or disorder using same" (PCT/US2015/042310)

Attendance of workshops

- Future Faculty Fellows Workshop, August 26 & 27, 2015, UW School of Medicine, University of Washington, Seattle, USA
- Reproducible Research for Biomedical Big Data, July 20-22, 2015, Summer Institute in Statistics for Big Data, University of Washington, Seattle, WA
- Building Next Generation Sequencing platforms and pipeline solutions, November 18-20, 2009, Rome, Italy
- Workshop on Molecular Evolution (Europe), January 12-23, 2009, Ceský Krumlov, Czech Republic
- Autumn School on Epigenetics, 2007, Humboldt University & Charité, Berlin, Germany

Teaching

2015 **ACAD Advanced Bioinformatics Early Career Researcher Workshop**, *University of Adelaide*, November 9-13, 2015, Adelaide, Australia
Bioinformatics of NGS sequencing, *Max Planck Institute for the Science of Human History*, October 26-27, 2015, Jena, Germany
UW Center for Mendelian Genomics Data Analysis Workshop, University of Washington, August 10-14, 2015, Seattle (WA), USA

2014 **UW Center for Mendelian Genomics Data Analysis Workshop**, University of Washington, August 11-15, 2014, Seattle (WA), USA

2013 **UW Center for Mendelian Genomics Data Analysis Workshop**, University of Washington, August 5-9, 2013, Seattle (WA), USA

2012 **ACAD Bioinformatics Early Career Researcher Workshop**, *University of Adelaide*, November 5-9, 2012, Adelaide, Australia

Unix command line tools and scripting for the analysis of high-throughput sequencing data, *Max Planck Institute for Evolutionary Anthropology*, February 27-March 2, 2012, Leipzig, Germany

2011 **Palaeogenomics Summer School**, *French network of palaeogenetics, RTP paléogénétique de l'Homme et de son environnement (CNRS-INEE)*, October 17-21 2011, Cargese, France

2010 **Next Generation Sequence Analysis**, *International Max Planck Research School of Human Origins*, August 3-4, 2010, Leipzig, Germany

2008 **Introduction to Python**, *Max Planck Institute for Evolutionary Anthropology*, March-June 2008, Leipzig, Germany

List of invited talks and lectures

2016 "Advancing massively parallel reporter assays for interpreting regulatory variants", **Genome Informatics**, 19-22 September 2016, Hinxton, Cambridge, UK

"Next Generation Sequencing: Diverse applications of Sequence, Counts and Structure", **Standards, Precautions and Advances in Ancient Metagenomics**, March 16 2016, MPI for Science of Human History, Jena, Germany

2015 "Tracing cell death throughout the body from cell-free DNA in blood", **Australian Centre for Ancient DNA**, November 12 2015, Adelaide, Australia

"Differences in objectives and implementations of variant effect scores" and "Developments to the Combined Annotation Dependent Depletion (CADD) framework for estimating deleteriousness of human genetic variation", **ISMB/ECCB 2015**, July 12 2015, Dublin, IRE

2014 "Combined Annotation Dependent Depletion (CADD) framework", **Future of Variant Annotation workshop**. September 20 2014, Cambridge, UK

"Integration of ENCODE data in the Combined Annotation Dependent Depletion (CADD) framework", **ENCODE Consortium Meeting 2014**. July 9 2014, Stanford (CA), USA

2013 "A general framework for estimating the relative pathogenicity of human genetic variants", **SNP-SIG @ ISMB/ECCB 2013**. July 19 2013, Berlin, Germany

"Identification of disease alleles from whole genome and exome sequencing data using a unified framework for ranking genetic variation", **TCGC: The Clinical Genome Conference**. June 26 2013, San Francisco (CA), USA

"An Integrated Approach for Prioritizing Causal Variants in Whole Exome and Whole Genome Sequencing", **ABRF 2012**. March 3 2012, Palm Springs (CA), USA

2012 "A high coverage Denisovan genome", **Australian Centre for Ancient DNA**, November 5 2012, Adelaide, Australia

2011 "Studying modern human origins from ancient DNA" (June 14 2011) and "Illumina Basecaller Ibis and the importance of quality scores" (June 15 2011), **Early Researcher Next-Generation Sequencing Symposium**. Saarbrücken, Germany

"Challenges in the analysis of high-throughput sequencing data", **PennState University. Beth Shapiro, PhD**. May 26 2011, State College (PA), USA

"Studying the transcriptome with next generation sequencing technologies", **Max Planck Institute of Molecular Cell Biology and Genetics**. March 30 2011, Dresden, Germany

"Challenges in the analysis of high-throughput sequencing data", **Dresden University, Institute for Medical Informatics and Biometry, Prof. Dr. Ingo Röder**. March 29 2011, Dresden, Germany

"High throughput sequencing - concepts and limitations....", **5th International Symposium on the Biology and Immunology of Cutaneous Lymphoma 2011**. January 14 2011, Berlin, Germany

2010 "Applying high-throughput sequencing to ancient and modern DNA samples for studying the genetic history of humankind", **Illumina Inc**. September 14 2010, Little Chesterford, UK

"Studying Modern Human Origins from Neandertal DNA", **Illumina Next Generation Seminar**. September 9 2010, Berlin, Germany

"Applying high-throughput sequencing to ancient and modern DNA samples", **Universität Leipzig, Faculty of Biosciences, Pharmacy and Psychology, Prof. Dr. Mario Mörl**. June 11 2010, Leipzig, Germany

"MPI EVA: High-throughput sequencing of ancient and modern DNA samples", **The First Galaxy Developer Conference**. May 16 2010, Cold Spring Harbor, New York, USA

2009 "One and a half years of Illumina processing pipelines", **Department of Evolutionary Genetics, Max Planck for Evolutionary Anthropology**. December 3 2009, Leipzig, Germany

"Improving Illumina Genome Analyzer data quality by alternative base calling", **Universität Leipzig, Interdisciplinary Centre for Bioinformatics, PD Dr. habil. Hans Binder**. December 1 2009, Leipzig, Germany

"Improving data quality of the Illumina Genome Analyzer platform", **Bioinformatics Autumn Seminar, Chair for Bioinformatics, Universität Leipzig**, October 24 2009, Vysoká Lípa, Decín, Czech Republic

"Applying high-throughput sequencing to ancient and modern DNA samples for studying the genetic history of humankind", **Genomics with Illumina: Arrays and Next Generation Sequencing**. September 22 2009, Leibniz-Institut für Pflanzengenetik und Kulturpflanzenforschung, Gatersleben, Germany

"High throughput sequencing", **Universität Leipzig, Faculty of Biosciences, Pharmacy and Psychology, Prof. Dr. Mario Mörl**. June 5 2009, Leipzig, Germany

"High throughput sequencing - concepts and limitations". **Deutsches Rheuma-Forschungszentrum Berlin**. May 26 2009, Berlin, Germany

"Improving Illumina base calling using Statistical Learners", **Department of Evolutionary Genetics, Max Planck for Evolutionary Anthropology**. January 29 2009, Leipzig, Germany

2008 "Illumina sequencing - A technology growing up...", **Saarland University, Faculty Natural Sciences and Technology III, Genetics / Epigenetics, Prof. Dr. Jörn Walter**. December 12 2008, Saarbrücken, Germany

"Solexa sequencing - a technology growing up...", **Bioinformatics Autumn Seminar, Chair for Bioinformatics, Universität Leipzig**. November 3 2008 Ceská Kamenice, Decín, Czech Republic

2007 "Coin flipping for selection - finding regions of positive selection in Human using the Neanderthal genome", **Department of Evolutionary Genetics, Max Planck for Evolutionary Anthropology**. December 20 2007, Leipzig, Germany

List of posters

2015 *M. Kircher*, F. Inoue, B. Martin, D.M. Witten, G.M. Cooper, N. Ahituv, J. Shendure. Computational and functional assessment of noncoding mutations in the human genome. **The Biology of Genomes**, Cold Spring Harbor (NY), USA

2014 *M. Kircher*, D.M. Witten, G.M. Cooper, J. Shendure. Integration of diverse annotation data in a general framework for estimating the relative pathogenicity of human genetic variants. **CSH/WT Genome Informatics**, Cambridge, UK

M. Kircher, D.M. Witten, G.M. Cooper, J. Shendure. Integration of diverse annotation data in a general framework for estimating the relative pathogenicity of human genetic variants. **European Conference on Computational Biology (ECCB)**, Strasbourg, France

2013 *M. Kircher*, D.M. Witten, G.M. Cooper, J. Shendure. A general framework for estimating the relative pathogenicity of human genetic variants. **Annual meeting of the American Society of Human Genetics (ASHG)**, Boston (MA), USA

M. Kircher, D.M. Witten, G.M. Cooper, J. Shendure. A general framework for estimating the relative pathogenicity of human genetic variants. **Annual International Conference on Intelligent Systems for Molecular Biology (ISMB) & European Conference on Computational Biology (ECCB),** Berlin, Germany

M. Kircher, D.M. Witten, G.M. Cooper, J. Shendure. A general framework for estimating the relative pathogenicity of human genetic variants. **The Biology of Genomes**, Cold Spring Harbor (NY), USA

2012 *M. Kircher* and The Denisova Genome Analysis Consortium. Analysis of a high coverage genome of a Denisovan individual. **Annual International Conference on Intelligent Systems for Molecular Biology (ISMB)**, Long Beach (CA), USA

2011 *M. Kircher* and J. Kelso. Computational challenges in the production and analysis of Illumina Genome Analyzer data. **Annual International Conference on Intelligent Systems for Molecular Biology (ISMB) & European Conference on Computational Biology (ECCB)**, Vienna, Austria

M. Kircher, J. Kelso, S. Pääbo, and The Neandertal & Denisova Genome Analysis Consortia. Identifying recent evolutionary changes on the human lineage. **The Biology of Genomes**, Cold Spring Harbor (NY), USA

2010 *M. Kircher*, E. Lizano, T. Giger, S. Pääbo, and J. Kelso. Challenges in the comparative analysis of gene expression in apes using Illumina Digital Gene Expression. **CSH/WT Genome Informatics**, Hinxton, Cambridge, UK

J. Kelso and *M. Kircher*. High-throughput DNA sequencing - concepts and limitations. **Annual International Conference on Intelligent Systems for Molecular Biology (ISMB)**, Boston (MA), USA

M. Kircher, U. Stenzel, and J. Kelso. Increasing the Genome Analyzer's output using IBIS. **Annual International Conference on Intelligent Systems for Molecular Biology (ISMB)**, Boston (MA), USA

U. Stenzel, A. Aximu-Petri, A. Butthof, B. Höber, B. Höffner, M. Siegemund, A. Weihmann, T. Maricic, M. Meyer, J. Kelso, and *M. Kircher*. Increasing the Genome Analyzer's output using IBIS - with or without a dedicated control lane. **2010 Illumina Europe User Symposium**, Sitges (Barcelona), Spain

M. Kircher and J. Kelso. Illumina Genome Analyzer: Artifacts and good analysis practice. **The Biology of Genomes**, Cold Spring Harbor (NY), USA

2009 *M. Kircher*, U. Stenzel, and J. Kelso. IBIS – Improved base calling for the Illumina Genome Analyzer. **German Conference on Bioinformatics**, Halle (Saale), Germany

M. Kircher, U. Stenzel, and J. Kelso. Improved base calling for the Illumina Genome Analyzer using machine learning strategies. **Annual International Conference on Intelligent Systems for Molecular Biology (ISMB) & European Conference on Computational Biology (ECCB)**, Stockholm, Sweden

M. Kircher, U. Stenzel, and J. Kelso. Improved base calling for the Illumina Genome Analyzer using machine learning strategies. **The Biology of Genomes**, Cold Spring Harbor (NY), USA