Stanford

Anna L Gloyn

Professor of Pediatrics (Endocrinology) and of Genetics

Pediatrics - Endocrinology

CONTACT INFORMATION

- **Administrative Associate**

  Nora Arrizon-Ruiz

  Email narrizon@stanford.edu

  Tel 650 721 2100

Bio

BIO

Dr Anna Gloyn joined the faculty at Stanford University in February 2020 after sixteen years at the University of Oxford, UK. Dr Gloyn completed her DPhil at the University of Oxford under the supervision of the late Professor Robert Turner. Her post-doctoral training was carried out at the University of Exeter under the mentorship of Professors Andrew Hattersley & Sian Ellard and at the University of Pennsylvania in Philadelphia under the mentorship of Professor Franz Matschinsky. In 2004 she returned to Oxford with a Diabetes UK RD Lawrence Career Development Fellowship and established an independent research group focused on understanding beta-cell function through the investigation of genetic variants causally implicated in monogenic diabetes. In 2011 she was awarded a prestigious Wellcome Senior Fellowship in Basic Biomedical Science which she successfully renewed in 2016 and transferred to Stanford when she relocated.

ACADEMIC APPOINTMENTS

- Professor, Pediatrics - Endocrinology
- Professor, Genetics
- Member, Bio-X
- Member, Cardiovascular Institute
- Member, Maternal & Child Health Research Institute (MCHRI)
- Faculty Fellow, Sarafan ChEM-H

ADMINISTRATIVE APPOINTMENTS

- Associate Chair for Basic Science Research, Department of Pediatrics, (2020- present)
- Associate Director for Basic Science, Stanford Diabetes Research Center, (2026- present)
- Co-Director of Enrichment Program, Stanford Diabetes Research Center, (2023- present)
- Co-Lead of Pancreas & Islet Affinity Group, Stanford Diabetes Research Centre, (2020- present)
- Co-Director of the Pilot & Feasibility Program, Stanford Diabetes Research Center, (2023-2026)
- Member, Stanford Diabetes Research Centre, (2020- present)

- Co-Director of Recruitment for Physician-Scientist Training Program, Pediatrics Residency Program, (2022- present)
- Co-Lead for Basic & Translational Science Scholarly Concentration, Pediatrics Residency Program, (2020- present)

HONORS AND AWARDS

- Honorary Doctorate, University of Bergen, Norway (May 2026)
- Transatlantic Alliance Award, Endocrine Society & European Society for Endocrinology (May 2026)
- Fellow, Academy of Medical Sciences (May 2025)
- Outstanding Scientific Achievement Award, American Diabetes Association (June 2022)
- Dorothy Hodgkin Named Lecture, Diabetes UK (March 2019)
- G.B. Morgagni Prize Silver Medal, University of Padua Medical School, Padua, Italy (October 2014)
- Minkowski Award, European Association for the Study of Diabetes (October 2014)
- RD Lawrence Named Lecture, Diabetes UK (March 2009)
- Rising Star Award, European Association for the Study of Diabetes (September 2005)
- Senior Fellowship in Basic Biomedical Science, Wellcome (2011-2022)
- New Investigator, Medical Research Council (UK) (2007-2011)
- RD Lawrence Career Development Fellow, Diabetes UK (2004-2008)

BOARDS, ADVISORY COMMITTEES, PROFESSIONAL ORGANIZATIONS

- Associate Editor, Diabetes Care (2024 - present)
- Editorial Advisory Board, Cell Genomics (2021 - present)
- Chair of Working Group Precision Diagnostics for Monogenic Diabetes, American Diabetes Association Precision Medicine Consensus Working Group (2020 - present)
- Member of the Executive Committee, Atlas of Variant Effects Alliance (2020 - present)
- Member of the Flagship Disease (Diabetes) Working Group, International Common Disease Alliance (2020 - present)
- Member of the Working Group (Mechanisms), International Common Disease Alliance (2020 - present)
- Member of the Working Group (Medicines), International Common Disease Alliance (2019 - present)
- Panel Member, ClinGen Monogenic Diabetes Variant Curation Expert Panel (2019 - present)
- Associate Editor, Endocrine Reviews (2018 - 2021)
- Editorial Advisory Board, Diabetologia (2018 - 2021)
- Trustee, European Diabetology (2017 - 2021)
- Expert Review Panel Member for Genetics Genomics & Population Health, Wellcome Trust (2016 - 2021)

PROFESSIONAL EDUCATION

- DPhil, University of Oxford , Molecular Genetics of Type 2 Diabetes (2000)
- BSc (Hons), University of Surrey , Biochemistry (Medical) (1996)

LINKS

- Stanford Lab Website: https://med.stanford.edu/genomics-of-diabetes.html

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

The consistent focus of Anna's research has been using naturally occurring mutations in humans as tools to identity critical regulatory pathways and insights into normal physiology. Her early post-doctoral research led to the identification a new genetic aetiology for permanent and transient neonatal diabetes due to KCNJ11 mutations and resulted in one of the first examples of precision medicine, where the determination of the molecular genetic aetiology lead to improved treatment options for patients. Whilst in Oxford, Anna's team discovered a novel genetic cause of constitutive insulin sensitivity in humans due to mutations in the PTEN gene highlighting the complex interplay between pathways involved in cell-growth and metabolism.

Her early independent work focused on the clinical and functional characterization of glucokinase mutations in monogenic forms of hyperinsulinemia of infancy, hyperglycemia and neonatal diabetes. Anna maintains an active research program in monogenic forms of diabetes and how genetics can be used to assist diabetes diagnosis and treatment. Her lab has expertise in variant characterization for multiple genes involved in monogenic diabetes and she supports clinicians with the interpretation of variants of unknown significance from genetic testing. A major focus of her current work is focused on how deep mutational scanning and maps of variant effects for diabetes relevant genes can be incorporated into guidelines for variant interpretation for monogenic diabetes through her involvement in the Clin Gen Monogenic Diabetes Variant Expert Review Panel and the Atlas of Variant Effects (AVE) consortium. Recently she has co-led the working group on Precision Diagnostics in monogenic diabetes for the and the ADA/EASD Precision Medicines Initiative which has uncovered a number of gaps in our knowledge.

Anna's research is not limited to monogenic forms of diabetes she is also an active member of multiple international consortia for genetic discovery for type 2 diabetes including the Accelerated Medicines Partnership for Common Metabolic Disease (AMP-CMD) where she uses her expertise in islet biology, functional genomics and cell and molecular physiology to bridge the gap between genetic discovery and biological and clinical insight. One of her areas of interest is in how genetics can be used for stratified medicine. Dr Gloyn is involved in several efforts to integrate genetic data on diabetes heterogeneity into human islet research within the Human Islet Research Network (HIRN). She is currently responsible for the genetic characterization of human islet donors for both the Integrated Islet Distribution Program (IIDP) where she heads the Human Genotyping Initiative (HIGI) and the Human Pancreas Analysis Program (HPAP) where she is responsible for the genotyping all donors. She has developed tools and methods to make genetic data available to islet users on ancestry and genetic risk for type 1 and type 2 diabetes.

Anna is an active member of multiple internal genetic discovery efforts including: NIH/Pharma funded Accelerated Medicines Partnership, DIAGRAM (Diabetes Genetics Replication and Meta-analysis), MAGIC (Meta-analysis of Glucose and Insulin traits Consortium), Type 2 Diabetes Genetic Exploration by Next-generation sequencing in multi-Ethnic Samples (T2D-GENES) and the Genetics of Type 2 Diabetes (GoT2D). She was also involved in the IMI funded STEMBANCC project which focused on delivering human IPS cell derived beta-cell models for drug discovery efforts.

Teaching

STANFORD ADVISEES

Doctoral Dissertation Reader (AC)

Kyle Trinh, Lucy Zhang

Postdoctoral Faculty Sponsor

Tamadher Alghunaim, Claire Kim, Yunkyeong Lee, Mei-Lin Okino, Lu Zhang

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Endocrinology (Fellowship Program)
- Genetics (Phd Program)
- Medical Genetics (Fellowship Program)
- Medicine (Masters Program)
- Pediatric Endocrinology (Fellowship Program)

Publications

PUBLICATIONS

- **Heterogeneous endocrine cell composition defines human islet functional phenotypes.** *Nature communications*
  Evans-Molina, C., Pettway, Y. D., Saunders, D. C., Sharp, S. A., Bate, T. S., Sun, H., Durai, H., Mei, S., Coldren, A., Davis, C., Reihsmann, C. V., Hopkirk, A. L., Taylor, et al
  2026; 17 (1)

- **Type 2 diabetes risk alleles in peptidyl-glycine alpha-amidating monooxygenase influence GLP-1 levels and response to GLP-1 receptor agonists.** *Genome medicine*
  Umapathysivam, M. M., Araldi, E., Hastoy, B., Dawed, A. Y., Vatandaslar, H., Mayrhofer, J. E., Lindquist, P., Silva, P. N., Goga, A., Trüllinger, G. O., Godbersen, S., Sengupta, S., Kaufmann, et al
  2026

- **Functional Characterization of Glucokinase Variants to Aid Clinical Interpretation of Monogenic Diabetes** *INTERNATIONAL JOURNAL OF MOLECULAR SCIENCES*
  Rajesh, V., Ibarra, D., Yang, J., Zhang, H., Barrett, A., Kaplan, E. G., Kumthekar, A., Sunden, F., Sun, H., Addala, A., Misakian, A., Letourneau-Freiberg, L. R., Jodarski, et al
  2025; 27 (1)

- **Complete Loss of PAX4 causes Transient Neonatal Diabetes in Humans.** *Molecular metabolism*
  Russ-Silsby, J., Lee, Y., Rajesh, V., Amoli, M., Mirhosseini, N. A., Godbole, T., Johnson, M. B., Ibarra, D. E., Sun, H., Krentz, N. A., Wakeling, M. N., Flanagan, S. E., Hattersley, et al
  2025: 102201

- **Diabetes mellitus polygenic risk scores: heterogeneity and clinical translation.** *Nature reviews. Endocrinology*
  Ortega, H. I., Udler, M. S., Gloyn, A. L., Sharp, S. A.
  2025

- **Accelerating Medicines Partnership in Type 2 Diabetes and Common Metabolic Diseases: Collaborating to Maximize the Value of Genetic and Genomic Data.** *Diabetes*
  Costanzo, M. C., Akolkar, B., Claussnitzer, M., Florez, J. C., Gloyn, A. L., Grant, S. F., Kaestner, K. H., Manning, A. K., Mohlke, K. L., Parker, S. C., Titchenell, P. M., Udler, M. S., Jones, et al
  2025

- **Standardized Measurement of Type 1 Diabetes Polygenic Risk Across Multiancestry Population Cohorts.** *Diabetes care*
  Luckett, A. M., Oram, R. A., Deutsch, A. J., Ortega, H. I., Fraser, D. P., Ashok, K., Manning, A. K., Mercader, J. M., Rivas, M. A., Udler, M. S., Weedon, M. N., Gloyn, A. L., Sharp, et al
  2025

- **Peptidylglycine alpha-amidating monooxygenase is important in mice for beta-cell cilia formation and insulin secretion but promotes diabetes risk through beta-cell independent mechanisms.** *Molecular metabolism*
  Chen, Y., Back, N. E., Zhen, J., Xiong, L., Komba, M., Gloyn, A. L., MacDonald, P. E., Mains, R. E., Eipper, B. A., Verchere, C. B.
  2025: 102123

- **Effects of coding variants in the glucokinase regulatory protein gene on hepatic glucose and triglyceride metabolism suggest a gene regulatory function of glucokinase.** *Metabolism: clinical and experimental*
  Langer, S., Jagdhuhn, D., Waterstradt, R., Gromoll, J., Müller, M., Rees, M. G., Gloyn, A. L., Baltrusch, S.
  2025: 156150

- **Prediction of metabolic subphenotypes of type 2 diabetes via continuous glucose monitoring and machine learning.** *Nature biomedical engineering*
  Metwally, A. A., Perelman, D., Park, H., Wu, Y., Jha, A., Sharp, S., Celli, A., Ayhan, E., Abbasi, F., Gloyn, A. L., McLaughlin, T., Snyder, M. P.
  2024
- **Rare variant analyses in 51,256 type 2 diabetes cases and 370,487 controls reveal the pathogenicity spectrum of monogenic diabetes genes.** *Nature genetics*
  Huerta-Chagoya, A., Schroeder, P., Mandla, R., Li, J., Morris, L., Vora, M., Alkanaq, A., Nagy, D., Szczerbinski, L., Madsen, J. G., Bonas-Guarch, S., Mollandin, F., Cole, et al
  2024
- **HumanIslets.com: Improving accessibility, integration, and usability of human research islet data.** *Cell metabolism*
  Ewald, J. D., Lu, Y., Ellis, C. E., Worton, J., Kolic, J., Sasaki, S., Zhang, D., Dos Santos, T., Spigelman, A. F., Bautista, A., Dai, X., Lyon, J. G., Smith, et al
  2024
- **Characteristics of glucokinase regulatory protein missense mutations with distinct effects on hepatic lipid metabolism suggest a gene regulatory function of glucokinase**
  Langer, S., Jagdhuhn, D., Waterstradt, R., Gromoll, J., Mueller, M., Rees, M. G., Gloyn, A. L., Baltrusch, S. E.
  SPRINGER.2024: S270-S271
- **Diabetes mellitus-Progress and opportunities in the evolving epidemic.** *Cell*
  Abel, E. D., Gloyn, A. L., Evans-Molina, C., Joseph, J. J., Misra, S., Pajvani, U. B., Simcox, J., Susztak, K., Drucker, D. J.
  2024; 187 (15): 3789-3820
- **A global initiative to deliver precision health in diabetes.** *Nature medicine*
  Cefalu, W. T., Franks, P. W., Rosenblum, N. D., Zaghloul, N. A., Florez, J. C., Giorgino, F., Ji, L., Ma, R. C., Mathieu, C., Misra, S., Ramirez, A. H., Roden, M., Scherer, et al
  2024
- **Proteomic predictors of individualized nutrient-specific insulin secretion in health and disease.** *Cell metabolism*
  Kolic, J., Sun, W. G., Cen, H. H., Ewald, J. D., Rogalski, J. C., Sasaki, S., Sun, H., Rajesh, V., Xia, Y. H., Moravcova, R., Skovsø, S., Spigelman, A. F., Manning Fox, et al
  2024; 36 (7): 1619-1633.e5
- **CD39 delineates chimeric antigen receptor regulatory T cell subsets with distinct cytotoxic & regulatory functions against human islets** *FRONTIERS IN IMMUNOLOGY*
  Wu, X., Chen, P., Whitener, R. L., MacDougall, M. S., Coykendall, V. M. N., Yan, H., Kim, Y., Harper, W., Pathak, S., Iliopoulou, B. P., Hestor, A., Saunders, D. C., Spears, et al
  2024; 15: 1415102
- **Electrophysiological characterisation of iPSC-derived human β-like cells and an SLC30A8 disease model.** *Diabetes*
  Jaffredo, M., Krentz, N. A., Champon, B., Duff, C. E., Nawaz, S., Beer, N., Honore, C., Clark, A., Rorsman, P., Lang, J., Gloyn, A. L., Raoux, M., Hastoy, et al
  2024
- **Multiplexed CRISPR gene editing in primary human islet cells with Cas9 ribonucleoprotein.** *iScience*
  Bevacqua, R. J., Zhao, W., Merheb, E., Kim, S. H., Marson, A., Gloyn, A. L., Kim, S. K.
  2024; 27 (1): 108693
- **Heterogeneity of increased biological age in type 2 diabetes correlates with differential tissue DNA methylation, biological variables, and pharmacological treatments.** *GeroScience*
  Cortez, B. N., Pan, H., Hinthorn, S., Sun, H., Neretti, N., Gloyn, A. L., Aguayo-Mazzucato, C.
  2023
- **Large-scale exome array summary statistics resources for glycemic traits to aid effector gene prioritization.** *Wellcome open research*
  Willems, S. M., Ng, N. H., Fernandez, J., Fine, R. S., Wheeler, E., Wessel, J., Kitajima, H., Marenne, G., Sim, X., Yaghootkar, H., Wang, S., Chen, S., Chen, et al
  2023; 8: 483
- **Second international consensus report on gaps and opportunities for the clinical translation of precision diabetes medicine.** *Nature medicine*

Tobias, D. K., Merino, J., Ahmad, A., Aiken, C., Benham, J. L., Bodhini, D., Clark, A. L., Colclough, K., Corcoy, R., Cromer, S. J., Duan, D., Felton, J. L., Francis, et al
2023

- **The use of precision diagnostics for monogenic diabetes: a systematic review and expert opinion.** *Communications medicine*
  Murphy, R., Colclough, K., Pollin, T. I., Ikle, J. M., Svalastoga, P., Maloney, K. A., Saint-Martin, C., Molnes, J., Misra, S., Aukrust, I., de Franco, E., Flanagan, S. E., Njølstad, et al
  2023; 3 (1): 136

- **PAX4 loss of function increases diabetes risk by altering human pancreatic endocrine cell development.** *Nature communications*
  Lau, H. H., Krentz, N. A., Abaitua, F., Perez-Alcantara, M., Chan, J. W., Ajeian, J., Ghosh, S., Lee, Y., Yang, J., Thaman, S., Champon, B., Sun, H., Jha, et al
  2023; 14 (1): 6119

- **Safe use of the ketogenic diet in an infant with microcephaly, epilepsy, and diabetes syndrome: a case report.** *BMC pediatrics*
  Zegarra, W. A., Gallentine, W. B., Ruzhnikov, M. R., McAndrews, C. A., Gloyn, A. L., Addala, A.
  2023; 23 (1): 453

- **Management of Neonatal Diabetes due to a KCNJ11 Mutation with Automated Insulin Delivery System and Remote Patient Monitoring.** *Case reports in endocrinology*
  Lee, M. Y., Gloyn, A. L., Maahs, D. M., Prahalad, P.
  2023; 2023: 8825724

- **An Atlas of Variant Effects to understand the genome at nucleotide resolution.** *Genome biology*
  Fowler, D. M., Adams, D. J., Gloyn, A. L., Hahn, W. C., Marks, D. S., Muffley, L. A., Neal, J. T., Roth, F. P., Rubin, A. F., Starita, L. M., Hurles, M. E.
  2023; 24 (1): 147

- **A comprehensive map of human glucokinase variant activity.** *Genome biology*
  Gersing, S., Cagiada, M., Gebbia, M., Gjesing, A. P., Coté, A. G., Seesankar, G., Li, R., Tabet, D., Weile, J., Stein, A., Gloyn, A. L., Hansen, T., Roth, et al
  2023; 24 (1): 97

- **Small but mighty: microexons in glucose homeostasis.** *Trends in genetics : TIG*
  Garcia, K., Gloyn, A. L.
  2023

- **Type 2 Diabetes risk alleles in Peptidyl-glycine Alpha-amidating Monooxygenase influence GLP-1 levels and response to GLP-1 Receptor Agonists.** *medRxiv : the preprint server for health sciences*
  Umapathysivam, M. M., Araldi, E., Hastoy, B., Dawed, A. Y., Vatandaslar, H., Sengupta, S., Kaufmann, A., Thomsen, S., Hartmann, B., Jonsson, A. E., Kabakci, H., Thaman, S., Grarup, et al
  2023

- **The Type 2 Diabetes Knowledge Portal: An open access genetic resource dedicated to type 2 diabetes and related traits.** *Cell metabolism*
  Costanzo, M. C., von Grotthuss, M., Massung, J., Jang, D., Caulkins, L., Koesterer, R., Gilbert, C., Welch, R. P., Kudtarkar, P., Hoang, Q., Boughton, A. P., Singh, P., Sun, et al
  2023

- **Loss of RREB1 in pancreatic beta cells reduces cellular insulin content and affects endocrine cell gene expression.** *Diabetologia*
  Mattis, K. K., Krentz, N. A., Metzendorf, C., Abaitua, F., Spigelman, A. F., Sun, H., Ikle, J. M., Thaman, S., Rottner, A. K., Bautista, A., Mazzaferro, E., Perez-Alcantara, M., Manning Fox, et al
  2023

- **Inferring causal genes at type 2 diabetes GWAS loci through chromosome interactions in islet cells.** *Wellcome open research*
  Torres, J. M., Sun, H., Nylander, V., Downes, D. J., van de Bunt, M., McCarthy, M. I., Hughes, J. R., Gloyn, A. L.
  2023; 8: 165

- **A genome-wide CRISPR screen identifies CALCOCO2 as a regulator of beta cell function influencing type 2 diabetes risk.** *Nature genetics*
  Rottner, A. K., Ye, Y., Navarro-Guerrero, E., Rajesh, V., Pollner, A., Bevacqua, R. J., Yang, J., Spigelman, A. F., Baronio, R., Bautista, A., Thomsen, S. K., Lyon, J., Nawaz, et al
  2022

- **Zmiz1 is required for mature β-cell function and mass expansion upon high fat feeding.** *Molecular metabolism*
  Alghamdi, T. A., Krentz, N. A., Smith, N., Spigelman, A. F., Rajesh, V., Jha, A., Ferdaoussi, M., Suzuki, K., Yang, J., Manning Fox, J. E., Sun, H., Sun, Z., Gloyn, et al
  2022: 101621

- **The contribution of functional HNF1A variants and polygenic susceptibility to risk of type 2 diabetes in ancestrally diverse populations.** *Diabetologia*
  Stalbow, L. A., Preuss, M. H., Smit, R. A., Chami, N., Bjørkhaug, L., Aukrust, I., Gloyn, A. L., Loos, R. J.
  2022

- **Genetic regulation of RNA splicing in human pancreatic islets.** *Genome biology*
  Atla, G., Bonas-Guarch, S., Cuenca-Ardura, M., Beucher, A., Crouch, D. J., Garcia-Hurtado, J., Moran, I., T2DSystems Consortium, Irimia, M., Prasad, R. B., Gloyn, A. L., Marselli, L., Suleiman, M., et al
  2022; 23 (1): 196

- **Every islet matters: improving the impact of human islet research.** *Nature metabolism*
  Gloyn, A. L., Ibberson, M., Marchetti, P., Powers, A. C., Rorsman, P., Sander, M., Solimena, M.
  2022

- **Multi-ancestry genetic study of type 2 diabetes highlights the power of diverse populations for discovery and translation.** *Nature genetics*
  Mahajan, A., Spracklen, C. N., Zhang, W., Ng, M. C., Petty, L. E., Kitajima, H., Yu, G. Z., Rueger, S., Speidel, L., Kim, Y. J., Horikoshi, M., Mercader, J. M., Taliun, et al
  2022

- **TIGER: The gene expression regulatory variation landscape of human pancreatic islets.** *Cell reports*
  Alonso, L., Piron, A., Moran, I., Guindo-Martinez, M., Bonas-Guarch, S., Atla, G., Miguel-Escalada, I., Royo, R., Puiggros, M., Garcia-Hurtado, X., Suleiman, M., Marselli, L., Esguerra, et al
  2021; 37 (2): 109807

- **Genetics of Type 2 Diabetes: Opportunities for Precision Medicine: JACC Focus Seminar.** *Journal of the American College of Cardiology*
  Kim, D. S., Gloyn, A. L., Knowles, J. W.
  2021; 78 (5): 496-512

- **A brief history of diabetes genetics: insights for pancreatic beta-cell development and function.** *The Journal of endocrinology*
  Ikle, J. M., Gloyn, A. L.
  2021

- **There is more than one way to reach type 2 diabetes.** *Nature metabolism*
  Gloyn, A. L., Powers, A. C.
  2021

- **The trans-ancestral genomic architecture of glycemic traits.** *Nature genetics*
  Chen, J., Spracklen, C. N., Marenne, G., Varshney, A., Corbin, L. J., Luan, J., Willems, S. M., Wu, Y., Zhang, X., Horikoshi, M., Boutin, T. S., Magi, R., Waage, et al
  2021

- **Monogenic diabetes: a gateway to precision medicine in diabetes.** *The Journal of clinical investigation*
  Zhang, H., Colclough, K., Gloyn, A. L., Pollin, T. I.
  2021; 131 (3)

- **A Multi-omic Integrative Scheme Characterizes Tissues of Action at Loci Associated with Type 2 Diabetes.** *American journal of human genetics*
  Torres, J. M., Abdalla, M., Payne, A., Fernandez-Tajes, J., Thurner, M., Nylander, V., Gloyn, A. L., Mahajan, A., McCarthy, M. I.
  2020

- **Response to Comment on Misra et al. Homozygous Hypomorphic HNF1A Alleles Are a Novel Cause of Young-Onset Diabetes and Result in Sulfonylurea-Sensitive Diabetes. Diabetes Care 2020;43:909-912.** *Diabetes care*
  Misra, S., Hassanali, N., Bennett, A. J., Juszczak, A., Caswell, R., Colclough, K., Valabhji, J., Ellard, S., Oliver, N. S., Gloyn, A. L.
  2020; 43 (10): e155–e156

- **Identification of type 2 diabetes loci in 433,540 East Asian individuals.** *Nature*

Spracklen, C. N., Horikoshi, M., Kim, Y. J., Lin, K., Bragg, F., Moon, S., Suzuki, K., Tam, C. H., Tabara, Y., Kwak, S., Takeuchi, F., Long, J., Lim, et al
2020

- **Endocrine-Exocrine Signaling Drives Obesity-Associated Pancreatic Ductal Adenocarcinoma.** *Cell*
  Chung, K. M., Singh, J., Lawres, L., Dorans, K. J., Garcia, C., Burkhardt, D. B., Robbins, R., Bhutkar, A., Cardone, R., Zhao, X., Babic, A., Vayrynen, S. A., Dias Costa, et al
  2020
- **From Genetic Association to Molecular Mechanisms for Islet-cell Dysfunction in Type 2 Diabetes** *JOURNAL OF MOLECULAR BIOLOGY*
  Mattis, K. K., Gloyn, A. L.
  2020; 432 (5): 1551–78
- **Editorial Overview: "Islet Biology in Type 2 Diabetes"** *JOURNAL OF MOLECULAR BIOLOGY*
  Gaisano, H. Y., Jonas, J., Gloyn, A. L.
  2020; 432 (5): 1307–9
- **Insights into pancreatic islet cell dysfunction from type 2 diabetes mellitus genetics.** *Nature reviews. Endocrinology*
  Krentz, N. A., Gloyn, A. L.
  2020
- **Deep learning models predict regulatory variants in pancreatic islets and refine type 2 diabetes association signals.** *eLife*
  Wesolowska-Andersen, A. n., Zhuo Yu, G. n., Nylander, V. n., Abaitua, F. n., Thurner, M. n., Torres, J. M., Mahajan, A. n., Gloyn, A. L., McCarthy, M. I.
  2020; 9
- **Unsupervised Clustering of Missense Variants in HNF1A Using Multidimensional Functional Data Aids Clinical Interpretation.** *American journal of human genetics*
  Althari, S. n., Najmi, L. A., Bennett, A. J., Aukrust, I. n., Rundle, J. K., Colclough, K. n., Molnes, J. n., Kaci, A. n., Nawaz, S. n., van der Lugt, T. n., Hassanali, N. n., Mahajan, A. n., Molven, et al
  2020
- **Genetic variant effects on gene expression in human pancreatic islets and their implications for T2D.** *Nature communications*
  Viñuela, A. n., Varshney, A. n., van de Bunt, M. n., Prasad, R. B., Asplund, O. n., Bennett, A. n., Boehnke, M. n., Brown, A. A., Erdos, M. R., Fadista, J. n., Hansson, O. n., Hatem, G. n., Howald, et al
  2020; 11 (1): 4912
- **Homozygous Hypomorphic HNF1A Alleles Are a Novel Cause of Young-Onset Diabetes and Result in Sulphonylurea-Sensitive Diabetes.** *Diabetes care*
  Misra, S. n., Hassanali, N. n., Bennett, A. J., Juszczak, A. n., Caswell, R. n., Colclough, K. n., Valabhji, J. n., Ellard, S. n., Oliver, N. S., Gloyn, A. L.
  2020
- **Analysis of Differentiation Protocols Defines a Common Pancreatic Progenitor Molecular Signature and Guides Refinement of Endocrine Differentiation.** *Stem cell reports*
  Wesolowska-Andersen, A. n., Jensen, R. R., Alcántara, M. P., Beer, N. L., Duff, C. n., Nylander, V. n., Gosden, M. n., Witty, L. n., Bowden, R. n., McCarthy, M. I., Hansson, M. n., Gloyn, A. L., Honore, et al
  2020; 14 (1): 138–53
- **Exocrine or endocrine? A circulating pancreatic elastase that regulates glucose homeostasis.** *Nature metabolism*
  Gloyn, A. L.
  2019; 1 (9): 853-855
- **METABOLIC SYNDROME Exocrine or endocrine? A circulating pancreatic elastase that regulates glucose homeostasis** *NATURE METABOLISM*
  Gloyn, A. L.
  2019; 1 (9): 853–55
- **Fostering improved human islet research: a European perspective** *DIABETOLOGIA*
  Marchetti, P., Schulte, A. M., Marselli, L., Schoniger, E., Bugliani, M., Kramer, W., Overbergh, L., Ullrich, S., Gloyn, A. L., Ibberson, M., Rutter, G., Froguel, P., Groop, et al
  2019; 62 (8): 1514–16

- **Developing a network view of type 2 diabetes risk pathways through integration of genetic, genomic and functional data** *GENOME MEDICINE*
  Fernandez-Tajes, J., Gaulton, K. J., van de Bunt, M., Torres, J., Thurner, M., Mahajan, A., Gloyn, A. L., Lage, K., McCarthy, M. I.
  2019; 11: 19

- **Loss of ZnT8 function protects against diabetes by enhanced insulin secretion.** *Nature genetics*
  Dwivedi, O. P., Lehtovirta, M. n., Hastoy, B. n., Chandra, V. n., Krentz, N. A., Kleiner, S. n., Jain, D. n., Richard, A. M., Abaitua, F. n., Beer, N. L., Grotz, A. n., Prasad, R. B., Hansson, et al
  2019; 51 (11): 1596–1606

- **Plasma Fucosylated Glycans and C-Reactive Protein as Biomarkers of HNF1A-MODY in Young Adult-Onset Nonautoimmune Diabetes** *DIABETES CARE*
  Juszczak, A., Pavic, T., Vuckovic, F., Bennett, A. J., Shah, N., Medvidovic, E., Groves, C. J., Sekerija, M., Chandler, K., Burrows, C., Putarek, N., Lovrencic, M., Knezevic, et al
  2019; 42 (1): 17–26

- **A CRISPR/Cas9 genome editing pipeline in the EndoC-βH1 cell line to study genes implicated in beta cell function.** *Wellcome open research*
  Grotz, A. K., Abaitua, F. n., Navarro-Guerrero, E. n., Hastoy, B. n., Ebner, D. n., Gloyn, A. L.
  2019; 4: 150

- **Translational genomics and precision medicine: Moving from the lab to the clinic.** *Science (New York, N.Y.)*
  Zeggini, E. n., Gloyn, A. L., Barton, A. C., Wain, L. V.
  2019; 365 (6460): 1409–13

- **Fine-mapping type 2 diabetes loci to single-variant resolution using high-density imputation and islet-specific epigenome maps.** *Nature genetics*
  Mahajan, A., Taliun, D., Thurner, M., Robertson, N. R., Torres, J. M., Rayner, N. W., Payne, A. J., Steinthorsdottir, V., Scott, R. A., Grarup, N., Cook, J. P., Schmidt, E. M., Wuttke, et al
  2018

- **Understanding human fetal pancreas development using subpopulation sorting, RNA sequencing and single-cell profiling** *DEVELOPMENT*
  Ramond, C., Beydag-Tasoz, B., Azad, A., van de Bunt, M., Petersen, M., Beer, N. L., Glaser, N., Berthault, C., Gloyn, A. L., Hansson, M., McCarthy, M., Honore, C., Grapin-Botton, et al
  2018; 145 (16)

- **Patterns of differential gene expression in a cellular model of human islet development, and relationship to type 2 diabetes predisposition** *DIABETOLOGIA*
  Perez-Alcantara, M., Honore, C., Wesolowska-Andersen, A., Gloyn, A. L., McCarthy, M. I., Hansson, M., Beer, N. L., van de Bunt, M.
  2018; 61 (7): 1614–22

- **NKX6.1 induced pluripotent stem cell reporter lines for isolation and analysis of functionally relevant neuronal and pancreas populations** *STEM CELL RESEARCH*
  Gupta, S., Wesolowska-Andersen, A., Ringgaard, A. K., Jaiswal, H., Song, L., Hastoy, B., Ingvorsen, C., Taheri-Ghahfarokhi, A., Magnusson, B., Maresca, M., Jensen, R. R., Beer, N. L., Fels, et al
  2018; 29: 220–31

- **Regulatory variants at KLF14 influence type 2 diabetes risk via a female-specific effect on adipocyte size and body composition** *NATURE GENETICS*
  Small, K. S., Todorcevic, M., Civelek, M., Moustafa, J., Wang, X., Simon, M. M., Fernandez-Tajes, J., Mahajan, A., Horikoshi, M., Hugill, A., Glastonbury, C. A., Quaye, L., Neville, et al
  2018; 50 (4): 572-+

- **A Partial Loss-of-Function Variant in AKT2 Is Associated With Reduced Insulin-Mediated Glucose Uptake in Multiple Insulin-Sensitive Tissues: A Genotype-Based Callback Positron Emission Tomography Study** *DIABETES*
  Latva-Rasku, A., Honka, M., Stancakova, A., Koistinen, H. A., Kuusisto, J., Guan, L., Manning, A. K., Stringham, H., Gloyn, A. L., Lindgren, C. M., Collins, F. S., Mohlke, K. L., Scott, et al
  2018; 67 (2): 334–42

- **Sequence data and association statistics from 12,940 type 2 diabetes cases and controls (vol 4, 170179, 2017)** *SCIENTIFIC DATA*
  Flannick, J., Fuchsberger, C., Mahajan, A., Teslovich, T. M., Agarwala, V., Gaulton, K. J., Caulkins, L., Koesterer, R., Ma, C., Moutsianas, L., McCarthy, D. J., Rivas, M. A., Perry, et al

2018; 5: 180002

- **Type 2 diabetes risk alleles in PAM impact insulin release from human pancreatic β-cells.** *Nature genetics*
  Thomsen, S. K., Raimondo, A. n., Hastoy, B. n., Sengupta, S. n., Dai, X. Q., Bautista, A. n., Censin, J. n., Payne, A. J., Umapathysivam, M. M., Spigelman, A. F., Barrett, A. n., Groves, C. J., Beer, et al
  2018; 50 (8): 1122–31

- **Maturity onset diabetes of the young due to HNF1A variants in Croatia** *BIOCHEMIA MEDICA*
  Pavic, T., Juszczak, A., Medvidovic, E., Burrows, C., Sekerija, M., Bennett, A. J., Knezevic, J., Gloyn, A. L., Lauc, G., McCarthy, M., Gornik, O., Owen, K. R.
  2018; 28 (2): 020703

- **Electrophysiological properties of human beta-cell lines EndoC-βH1 and -βH2 conform with human beta-cells.** *Scientific reports*
  Hastoy, B. n., Godazgar, M. n., Clark, A. n., Nylander, V. n., Spiliotis, I. n., van de Bunt, M. n., Chibalina, M. V., Barrett, A. n., Burrows, C. n., Tarasov, A. I., Scharfmann, R. n., Gloyn, A. L., Rorsman, et al
  2018; 8 (1): 16994

- **Integration of human pancreatic islet genomic data refines regulatory mechanisms at Type 2 Diabetes susceptibility loci.** *eLife*
  Thurner, M. n., van de Bunt, M. n., Torres, J. M., Mahajan, A. n., Nylander, V. n., Bennett, A. J., Gaulton, K. J., Barrett, A. n., Burrows, C. n., Bell, C. G., Lowe, R. n., Beck, S. n., Rakyan, et al
  2018; 7

- **Precision medicine in the management of type 2 diabetes.** *The lancet. Diabetes & endocrinology*
  Gloyn, A. L., Drucker, D. J.
  2018; 6 (11): 891–900

- **Data Descriptor: Sequence data and association statistics from 12,940 type 2 diabetes cases and controls** *SCIENTIFIC DATA*
  Flannick, J., Fuchsberger, C., Mahajan, A., Teslovich, T. M., Agarwala, V., Gaulton, K. J., Caulkins, L., Koesterer, R., Ma, C., Moutsianas, L., McCarthy, D. J., Rivas, M. A., Perry, et al
  2017; 4: 170179

- **Genes Associated with Pancreas Development and Function Maintain Open Chromatin in iPSCs Generated from Human Pancreatic Beta Cells** *STEM CELL REPORTS*
  Thurner, M., Shenhav, L., Wesolowska-Andersen, A., Bennett, A. J., Barrett, A., Gloyn, A. L., McCarthy, M. I., Beer, N. L., Efrat, S.
  2017; 9 (5): 1395–1405

- **Prioritising Causal Genes at Type 2 Diabetes Risk Loci** *CURRENT DIABETES REPORTS*
  Grotz, A. K., Gloyn, A. L., Thomsen, S. K.
  2017; 17 (9): 76

- **Human genetics as a model for target validation: finding new therapies for diabetes** *DIABETOLOGIA*
  Thomsen, S. K., Gloyn, A. L.
  2017; 60 (6): 960–70

- **Variant Enriched in the Finnish Population is Associated With Fasting Insulin Levels and Type 2 Diabetes Risk.** *Diabetes*
  Manning, A., Highland, H. M., Gasser, J., Sim, X., Tukiainen, T., Fontanillas, P., Grarup, N., Rivas, M. A., Mahajan, A., Locke, A. E., Cingolani, P., Pers, T. H., Viñuela, et al
  2017

- **Decreased STARD10 Expression Is Associated with Defective Insulin Secretion in Humans and Mice** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Carrat, G. R., Hu, M., Nguyen-Tu, M., Chabosseau, P., Gaulton, K. J., van de Bunt, M., Siddiq, A., Falchi, M., Thurner, M., Canouil, M., Pattou, F., Leclerc, I., Pullen, et al
  2017; 100 (2): 238–56

- **The importance of Context: Uncovering Species- and Tissue-Specific Effects of Genetic Risk Variants for Type 2 Diabetes** *FRONTIERS IN ENDOCRINOLOGY*
  Thomsen, S. K., McCarthy, M. I., Gloyn, A. L.
  2016; 7: 112

- **The genetic architecture of type 2 diabetes** *NATURE*

Fuchsberger, C., Flannick, J., Teslovich, T. M., Mahajan, A., Agarwala, V., Gaulton, K. J., Ma, C., Fontanillas, P., Moutsianas, L., McCarthy, D. J., Rivas, M. A., Perry, J. R., Sim, et al
2016; 536 (7614): 41-?

- **Insights into metabolic disease from studying genetics in isolated populations: stories from Greece to Greenland** *DIABETOLOGIA*
  Zeggini, E., Gloyn, A. L., Hansen, T.
  2016; 59 (5): 938–41
- **Systematic Functional Characterization of Candidate Causal Genes for Type 2 Diabetes Risk Variants.** *Diabetes*
  Thomsen, S. K., Ceroni, A. n., van de Bunt, M. n., Burrows, C. n., Barrett, A. n., Scharfmann, R. n., Ebner, D. n., McCarthy, M. I., Gloyn, A. L.
  2016; 65 (12): 3805–11
- **Insights into islet development and biology through characterization of a human iPSC-derived endocrine pancreas model** *ISLETS*
  van de Bunt, M., Lako, M., Barrett, A., Gloyn, A. L., Hansson, M., McCarthy, M. I., Beer, N. L., Honore, C.
  2016; 8 (3): 83–95
- **Loss-of-Function Mutations in the Cell-Cycle Control Gene CDKN2A Impact on Glucose Homeostasis in Humans.** *Diabetes*
  Pal, A. n., Potjer, T. P., Thomsen, S. K., Ng, H. J., Barrett, A. n., Scharfmann, R. n., James, T. J., Bishop, D. T., Karpe, F. n., Godsland, I. F., Vasen, H. F., Newton-Bishop, J. n., Pijl, et al
  2016; 65 (2): 527–33
- **Genome-edited human stem cell-derived beta cells: a powerful tool for drilling down on type 2 diabetes GWAS biology.** *F1000Research*
  Beer, N. L., Gloyn, A. L.
  2016; 5
- **Transcript Expression Data from Human Islets Links Regulatory Signals from Genome-Wide Association Studies for Type 2 Diabetes and Glycemic Traits to Their Downstream Effectors** *PLOS GENETICS*
  van de Bunt, M., Fox, J., Dai, X., Barrett, A., Grey, C., Li, L., Bennett, A. J., Johnson, P. R., Rajotte, R. V., Gaulton, K. J., Dermitzakis, E. T., MacDonald, P. E., McCarthy, et al
  2015; 11 (12): e1005694
- **Genetic fine mapping and genomic annotation defines causal mechanisms at type 2 diabetes susceptibility loci** *NATURE GENETICS*
  Gaulton, K. J., Ferreira, T., Lee, Y., Raimondo, A., Maegi, R., Reschen, M. E., Mahajan, A., Locke, A., Rayner, N. W., Robertson, N., Scott, R. A., Prokopenko, I., Scott, et al
  2015; 47 (12): 1415-?
- **Isocitrate-to-SENP1 signaling amplifies insulin secretion and rescues dysfunctional beta cells** *JOURNAL OF CLINICAL INVESTIGATION*
  Ferdaoussi, M., Dai, X., Jensen, M. V., Wang, R., Peterson, B. S., Huang, C., Ilkayeva, O., Smith, N., Miller, N., Hajmrle, C., Spigelman, A. F., Wright, R. C., Plummer, et al
  2015; 125 (10): 3847–60
- **When is it MODY? Challenges in the Interpretation of Sequence Variants in MODY Genes.** *The review of diabetic studies : RDS*
  Althari, S., Gloyn, A. L.
  2015; 12 (3-4): 330–48
- **Human islet function following 20 years of cryogenic biobanking** *DIABETOLOGIA*
  Fox, J., Lyon, J., Dai, X., Wright, R. C., Hayward, J., van de Bunt, M., Kin, T., Shapiro, A., McCarthy, M. I., Gloyn, A. L., Ungrin, M. D., Lakey, J. R., Kneteman, et al
  2015; 58 (7): 1503–12
- **Recognition and Management of Individuals With Hyperglycemia Because of a Heterozygous Glucokinase Mutation** *DIABETES CARE*
  Chakera, A. J., Steele, A. M., Gloyn, A. L., Shepherd, M. H., Shields, B., Ellard, S., Hattersley, A. T.
  2015; 38 (7): 1383–92
- **Glucokinase regulatory protein: complexity at the crossroads of triglyceride and glucose metabolism** *CURRENT OPINION IN LIPIDOLOGY*
  Raimondo, A., Rees, M. G., Gloyn, A. L.
  2015; 26 (2): 88–95
- **Identification and Functional Characterization of G6PC2 Coding Variants Influencing Glycemic Traits Define an Effector Transcript at the G6PC2-ABCB11 Locus** *PLOS GENETICS*

Mahajan, A., Sim, X., Ng, H. J., Manning, A., Rivas, M. A., Highland, H. M., Locke, A. E., Grarup, N., Im, H. K., Cingolani, P., Flannick, J., Fontanillas, P., Fuchsberger, et al

2015; 11 (1)

- **The pancreatic beta cell: recent insights from human genetics** *TRENDS IN ENDOCRINOLOGY AND METABOLISM*

  Thomsen, S. K., Gloyn, A. L.

  2014; 25 (8): 425–34

- **Reclassification of Diabetes Etiology in a Family With Multiple Diabetes Phenotypes** *JOURNAL OF CLINICAL ENDOCRINOLOGY & METABOLISM*

  Kavvoura, F. K., Raimondo, A., Thanabalasingham, G., Barrett, A., Webster, A. L., Shears, D., Mann, N. P., Ellard, S., Gloyn, A. L., Owen, K. R.

  2014; 99 (6): E1067–E1071

- **Analysis of the co-operative interaction between the allosterically regulated proteins GK and GKRP using tryptophan fluorescence** *BIOCHEMICAL JOURNAL*

  Zelent, B., Raimondo, A., Barrettt, A., Buettger, C. W., Chen, P., Gloyn, A. L., Matschinsky, F. M.

  2014; 459: 551–64

- **A Panel of Diverse Assays to Interrogate the Interaction between Glucokinase and Glucokinase Regulatory Protein, Two Vital Proteins in Human Disease** *PLOS ONE*

  Rees, M. G., Davis, M. I., Shen, M., Titus, S., Raimondo, A., Barrett, A., Gloyn, A. L., Collins, F. S., Simeonov, A.

  2014; 9 (2): e89335

- **Pancreatic islet enhancer clusters enriched in type 2 diabetes risk-associated variants** *NATURE GENETICS*

  Pasquali, L., Gaulton, K. J., Rodriguez-Segui, S. A., Mularoni, L., Miguel-Escalada, I., Akerman, I., Tena, J. J., Moran, I., Gomez-Marin, C., van de Bunt, M., Ponsa-Cobas, J., Castro, N., Nammo, et al

  2014; 46 (2): 136-+

- **Argonaute2 Mediates Compensatory Expansion of the Pancreatic beta Cell** *CELL METABOLISM*

  Tattikota, S. G., Rathjen, T., McAnulty, S. J., Wessels, H., Akerman, I., van de Bunt, M., Hausser, J., Esguerra, J. L. S., Musahl, A., Pandey, A. K., You, X., Chen, W., Herrera, et al

  2014; 19 (1): 122–34

- **Phenotypic severity of homozygous GCK mutations causing neonatal or childhood-onset diabetes is primarily mediated through effects on protein stability.** *Human molecular genetics*

  Raimondo, A. n., Chakera, A. J., Thomsen, S. K., Colclough, K. n., Barrett, A. n., De Franco, E. n., Chatelas, A. n., Demirbilek, H. n., Akcay, T. n., Alawneh, H. n., Flanagan, S. E., Van De Bunt, M. n., Hattersley, et al

  2014; 23 (24): 6432–40

- **Genetics in Diabetes Type 2 Diabetes and Related Traits Preface** *GENETICS IN DIABETES: TYPE 2 DIABETES AND RELATED TRAITS*

  Gloyn, A. L., McCarthy, M. I.

  edited by Gloyn, A. L., McCarthy, M. I.

  2014; 23: VII

- **Translating Genetic Association Signals for Diabetes and Metabolic Traits into Molecular Mechanisms for Disease** *GENETICS IN DIABETES: TYPE 2 DIABETES AND RELATED TRAITS*

  Rees, M. G., Gloyn, A. L.

  edited by Gloyn, A. L., McCarthy, M. I.

  2014; 23: 133–45

- **Translating Advances in Our Understanding of the Genetics of Diabetes into the Clinic** *GENETICS IN DIABETES: TYPE 2 DIABETES AND RELATED TRAITS*

  Gardner, D. S., Owen, K. R., Gloyn, A. L.

  edited by Gloyn, A. L., McCarthy, M. I.

  2014; 23: 173–86

- **Inheritance of rare functional GCKR variants and their contribution to triglyceride levels in families.** *Human molecular genetics*

  Rees, M. G., Raimondo, A. n., Wang, J. n., Ban, M. R., Davis, M. I., Barrett, A. n., Ranft, J. n., Jagdhuhn, D. n., Waterstradt, R. n., Baltrusch, S. n., Simeonov, A. n., Collins, F. S., Hegele, et al

  2014; 23 (20): 5570–78

- **Role of K-ATP Channels in Glucose-Regulated Glucagon Secretion and Impaired Counterregulation in Type 2 Diabetes** *CELL METABOLISM*
  Zhang, Q., Ramracheya, R., Lahmann, C., Tarasov, A., Bengtsson, M., Braha, O., Braun, M., Brereton, M., Collins, S., Galvanovskis, J., Gonzalez, A., Groschner, L. N., Rorsman, et al
  2013; 18 (6): 871–82

- **Bridging the Gap Between Genetic Associations and Molecular Mechanisms for Type 2 Diabetes** *CURRENT DIABETES REPORTS*
  Ng, H., Gloyn, A. L.
  2013; 13 (6): 778–85

- **Genome-Wide Association Study Identifies a Novel Locus Contributing to Type 2 Diabetes Susceptibility in Sikhs of Punjabi Origin From India** *DIABETES*
  Saxena, R., Saleheen, D., Been, L. F., Garavito, M. L., Braun, T., Bjonnes, A., Young, R., Ho, W., Rasheed, A., Frossard, P., Sim, X., Hassanali, N., Radha, et al
  2013; 62 (5): 1746–55

- **Mutations in HNF1A Result in Marked Alterations of Plasma Glycan Profile** *DIABETES*
  Thanabalasingham, G., Huffman, J. E., Kattla, J. J., Novokmet, M., Rudan, I., Gloyn, A. L., Hayward, C., Adamczyk, B., Reynolds, R. M., Muzinic, A., Hassanali, N., Pucic, M., Bennett, et al
  2013; 62 (4): 1329–37

- **Genome-Wide Association Study for Type 2 Diabetes in Indians Identifies a New Susceptibility Locus at 2q21** *DIABETES*
  Tabassum, R., Chauhan, G., Dwivedi, O., Mahajan, A., Jaiswal, A., Kaur, I., Bandesh, K., Singh, T., Mathai, B., Pandey, Y., Chidambaram, M., Sharma, A., Chavali, et al
  2013; 62 (3): 977–86

- **TCF7L2 and Diabetes: A Tale of Two Tissues, and of Two Species** *CELL METABOLISM*
  McCarthy, M. I., Rorsman, P., Gloyn, A. L.
  2013; 17 (2): 157–59

- **Apolipoprotein M can discriminate HNF1A-MODY from Type 1 diabetes** *DIABETIC MEDICINE*
  Mughal, S. A., Park, R., Nowak, N., Gloyn, A. L., Karpe, F., Matile, H., Malecki, M. T., McCarthy, M. I., Stoffel, M., Owen, K. R.
  2013; 30 (2): 246–50

- **The miRNA Profile of Human Pancreatic Islets and Beta-Cells and Relationship to Type 2 Diabetes Pathogenesis** *PLOS ONE*
  van de Bunt, M., Gaulton, K. J., Parts, L., Moran, I., Johnson, P. R., Lindgren, C. M., Ferrer, J., Gloyn, A. L., McCarthy, M. I.
  2013; 8 (1): e55272

- **Insights into the molecular mechanism for type 2 diabetes susceptibility at the KCNQ1 locus from temporal changes in imprinting status in human islets.** *Diabetes*
  Travers, M. E., Mackay, D. J., Dekker Nitert, M. n., Morris, A. P., Lindgren, C. M., Berry, A. n., Johnson, P. R., Hanley, N. n., Groop, L. C., McCarthy, M. I., Gloyn, A. L.
  2013; 62 (3): 987–92

- **Small molecular glucokinase activators: has another new anti-diabetic therapeutic lost favour?** *BRITISH JOURNAL OF PHARMACOLOGY*
  Rees, M. G., Gloyn, A. L.
  2013; 168 (2): 335–38

- **SSTR2 is the functionally dominant somatostatin receptor in human pancreatic beta- and alpha-cells** *AMERICAN JOURNAL OF PHYSIOLOGY-ENDOCRINOLOGY AND METABOLISM*
  Kailey, B., van de Bunt, M., Cheley, S., Johnson, P. R., MacDonald, P. E., Gloyn, A. L., Rorsman, P., Braun, M.
  2012; 303 (9): E1107–E1116

- **Human beta Cell Transcriptome Analysis Uncovers lncRNAs That Are Tissue-Specific, Dynamically Regulated, and Abnormally Expressed in Type 2 Diabetes** *CELL METABOLISM*
  Moran, I., Akerman, I., van de Bunt, M., Xie, R., Benazra, M., Nammo, T., Arnes, L., Nakic, N., Garcia-Hurtado, J., Rodriguez-Segui, S., Pasquali, L., Sauty-Colace, C., Beucher, et al
  2012; 16 (4): 435–48

- **A tale of two glucose transporters: how GLUT2 re-emerged as a contender for glucose transport into the human beta cell** *DIABETOLOGIA*
  van de Bunt, M., Gloyn, A. L.
  2012; 55 (9): 2312–15

- **Metabolic Profiling in Maturity-Onset Diabetes of the Young (MODY) and Young Onset Type 2 Diabetes Fails to Detect Robust Urinary Biomarkers** *PLOS ONE*
  Gloyn, A. L., Faber, J. H., Malmodin, D., Thanabalasingham, G., Lam, F., Ueland, P., McCarthy, M. I., Owen, K. R., Baunsgaard, D.
  2012; 7 (7): e40962

- **Reduced Insulin Exocytosis in Human Pancreatic beta-Cells With Gene Variants Linked to Type 2 Diabetes** *DIABETES*
  Rosengren, A. H., Braun, M., Mandi, T., Andersson, S. A., Travers, M. E., Shigeto, M., Zhang, E., Almgren, P., Ladenvall, C., Axelsson, A. S., Edlund, A., Pedersen, M., Jonsson, et al
  2012; 61 (7): 1726–33

- **A genome-wide approach accounting for body mass index identifies genetic variants influencing fasting glycemic traits and insulin resistance** *NATURE GENETICS*
  Manning, A. K., Hivert, M., Scott, R. A., Grimsby, J. L., Bouatia-Naji, N., Chen, H., Rybin, D., Liu, C., Bielak, L. F., Prokopenko, I., Amin, N., Barnes, D., Cadby, et al
  2012; 44 (6): 659-U81

- **Identification and Functional Characterisation of Novel Glucokinase Mutations Causing Maturity-Onset Diabetes of the Young in Slovakia** *PLOS ONE*
  Valentinova, L., Beer, N. L., Stanik, J., Tribble, N. D., van de Bunt, M., Huckova, M., Barrett, A., Klimes, I., Gasperikova, D., Gloyn, A. L.
  2012; 7 (4): e34541

- **Novel Loci for Adiponectin Levels and Their Influence on Type 2 Diabetes and Metabolic Traits: A Multi-Ethnic Meta-Analysis of 45,891 Individuals** *PLOS GENETICS*
  Dastani, Z., Hivert, M., Timpson, N., Perry, J. R., Yuan, X., Scott, R. A., Henneman, P., Heid, I. M., Kizer, J. R., Lyytikainen, L., Fuchsberger, C., Tanaka, T., Morris, et al
  2012; 8 (3)

- **Low-Frequency Variants in HMGA1 Are Not Associated With Type 2 Diabetes Risk** *DIABETES*
  Marquez, M., Huyvaert, M., Perry, J. R. B., Pearson, R. D., Falchi, M., Morris, A. P., Vivequin, S., Lobbens, S., Yengo, L., Gaget, S., Pattou, F., Poulain-Godefroy, O., Charpentier, et al
  2012; 61 (2): 524–30

- **PTEN mutations as a cause of constitutive insulin sensitivity and obesity.** *The New England journal of medicine*
  Pal, A. n., Barber, T. M., Van de Bunt, M. n., Rudge, S. A., Zhang, Q. n., Lachlan, K. L., Cooper, N. S., Linden, H. n., Levy, J. C., Wakelam, M. J., Walker, L. n., Karpe, F. n., Gloyn, et al
  2012; 367 (11): 1002–11

- **Meta-analysis of genome-wide association studies identifies eight new loci for type 2 diabetes in east Asians.** *Nature genetics*
  Cho, Y. S., Chen, C., Hu, C., Long, J., Ong, R. T., Sim, X., Takeuchi, F., Wu, Y., Go, M. J., Yamauchi, T., Chang, Y., Kwak, S. H., Ma, et al
  2012; 44 (1): 67-72

- **Insights into the pathogenicity of rare missense GCK variants from the identification and functional characterization of compound heterozygous and double mutations inherited in cis.** *Diabetes care*
  Beer, N. L., Osbak, K. K., van de Bunt, M. n., Tribble, N. D., Steele, A. M., Wensley, K. J., Edghill, E. L., Colcough, K. n., Barrett, A. n., Valentínová, L. n., Rundle, J. K., Raimondo, A. n., Grimsby, et al
  2012; 35 (7): 1482–84

- **Correlation of rare coding variants in the gene encoding human glucokinase regulatory protein with phenotypic, cellular, and kinetic outcomes.** *The Journal of clinical investigation*
  Rees, M. G., Ng, D. n., Ruppert, S. n., Turner, C. n., Beer, N. L., Swift, A. J., Morken, M. A., Below, J. E., Blech, I. n., Mullikin, J. C., McCarthy, M. I., Biesecker, L. G., Gloyn, et al
  2012; 122 (1): 205–17

- **Cellular characterisation of the GCKR P446L variant associated with type 2 diabetes risk.** *Diabetologia*
  Rees, M. G., Wincovitch, S. n., Schultz, J. n., Waterstradt, R. n., Beer, N. L., Baltrusch, S. n., Collins, F. S., Gloyn, A. L.
  2012; 55 (1): 114–22

- **GLUT2 (SLC2A2) is not the principal glucose transporter in human pancreatic beta cells: Implications for understanding genetic association signals at this locus** *MOLECULAR GENETICS AND METABOLISM*
  McCulloch, L. J., van de Bunt, M., Braun, M., Frayn, K. N., Clark, A., Gloyn, A. L.
  2011; 104 (4): 648–53

- **A large multi-centre European study validates high-sensitivity C-reactive protein (hsCRP) as a clinical biomarker for the diagnosis of diabetes subtypes** *DIABETOLOGIA*
  Thanabalasingham, G., Shah, N., Vaxillaire, M., Hansen, T., Tuomi, T., Gasperikova, D., Szopa, M., Tjora, E., James, T. J., Kokko, P., Loiseleur, F., Andersson, E., Gaget, et al
  2011; 54 (11): 2801–10
- **Genome-wide association study identifies loci influencing concentrations of liver enzymes in plasma** *NATURE GENETICS*
  Chambers, J. C., Zhang, W., Sehmi, J., Li, X., Wass, M. N., Van der Harst, P., Holm, H., Sanna, S., Kavousi, M., Baumeister, S. E., Coin, L. J., Deng, G., Gieger, et al
  2011; 43 (11): 1131–38
- **Genome-Wide Association Identifies Nine Common Variants Associated With Fasting Proinsulin Levels and Provides New Insights Into the Pathophysiology of Type 2 Diabetes** *DIABETES*
  Strawbridge, R. J., Dupuis, J., Prokopenko, I., Barker, A., Ahlqvist, E., Rybin, D., Petrie, J. R., Travers, M. E., Bouatia-Naji, N., Dimas, A. S., Nica, A., Wheeler, E., Chen, et al
  2011; 60 (10): 2624-2634
- **The previously reported T342P GCK missense variant is not a pathogenic mutation causing MODY** *DIABETOLOGIA*
  Steele, A. M., Tribble, N. D., Caswell, R., Wensley, K. J., Hattersley, A. T., Gloyn, A. L., Ellard, S.
  2011; 54 (8): 2202–5
- **High-Sensitivity CRP Discriminates HNF1A-MODY From Other Subtypes of Diabetes** *DIABETES CARE*
  McDonald, T. J., Shields, B. M., Lawry, J., Owen, K. R., Gloyn, A. L., Ellard, S., Hattersley, A. T.
  2011; 34 (8): 1860–62
- **Identification of an imprinted master trans regulator at the KLF14 locus related to multiple metabolic phenotypes** *NATURE GENETICS*
  Small, K. S., Hedman, A. K., Grundberg, E., Nica, A. C., Thorleifsson, G., Kong, A., Thorsteindottir, U., Shin, S., Richards, H. B., Soranzo, N., Ahmadi, K. R., Lindgren, C. M., Stefansson, et al
  2011; 43 (6): 561–U90
- **Comprehensive Human Adipose Tissue mRNA and MicroRNA Endogenous Control Selection for Quantitative Real-Time-PCR Normalization** *OBESITY*
  Neville, M. J., Collins, J. M., Gloyn, A. L., McCarthy, M. I., Karpe, F.
  2011; 19 (4): 888–92
- **Discovery of a novel site regulating glucokinase activity following characterization of a new mutation causing hyperinsulinemic hypoglycemia in humans.** *The Journal of biological chemistry*
  Beer, N. L., van de Bunt, M. n., Colclough, K. n., Lukacs, C. n., Arundel, P. n., Chik, C. L., Grimsby, J. n., Ellard, S. n., Gloyn, A. L.
  2011; 286 (21): 19118–26
- **Genetically Programmed Defects in beta-Cell Function** *BETASYS: SYSTEMS BIOLOGY OF REGULATED EXOCYTOSIS IN PANCREATIC BETA-CELLS*
  Pal, A., Gloyn, A. L.
  edited by BoossBavnbek, B., Klosgen, B., Larsen, J., Pociot, F., Renstrom, E.
  2011; 2: 299–326
- **A role for coding functional variants in HNF4A in type 2 diabetes susceptibility** *DIABETOLOGIA*
  Jafar-Mohammadi, B., Groves, C. J., Gjesing, A. P., Herrera, B. M., Winckler, W., Stringham, H. M., Morris, A. P., Lauritzen, T., Doney, A. F., Morris, A. D., Weedon, M. N., Swift, A. J., Kuusisto, et al
  2011; 54 (1): 111–19
- **From Genetic Association to Molecular Mechanism** *CURRENT DIABETES REPORTS*
  van de Bunt, M., Gloyn, A. L.
  2010; 10 (6): 452–66
- **Variation across the allele frequency spectrum** *NATURE GENETICS*
  Gloyn, A. L., McCarthy, M. I.
  2010; 42 (8): 648–50
- **Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis** *NATURE GENETICS*

Voight, B. F., Scott, L. J., Steinthorsdottir, V., Morris, A. P., Dina, C., Welch, R. P., Zeggini, E., Huth, C., Aulchenko, Y. S., Thorleifsson, G., McCulloch, L. J., Ferreira, T., Grallert, et al
2010; 42 (7): 579-U155

- **Global microRNA expression profiles in insulin target tissues in a spontaneous rat model of type 2 diabetes** *DIABETOLOGIA*
  Herrera, B. M., Lockstone, H. E., Taylor, J. M., Ria, M., Barrett, A., Collins, S., Kaisaki, P., Argoud, K., Fernandez, C., Travers, M. E., Grew, J. P., Randall, J. C., Gloyn, et al
  2010; 53 (6): 1099–1109

- **New genetic loci implicated in fasting glucose homeostasis and their impact on type 2 diabetes risk** *NATURE GENETICS*
  Dupuis, J., Langenberg, C., Prokopenko, I., Saxena, R., Soranzo, N., Jackson, A. U., Wheeler, E., Glazer, N. L., Bouatia-Naji, N., Gloyn, A. L., Lindgren, C. M., Magi, R., Morris, et al
  2010; 42 (2): 105-U32

- **Evaluation of Serum 1,5 Anhydroglucitol Levels as a Clinical Test to Differentiate Subtypes of Diabetes** *DIABETES CARE*
  Pal, A., Farmer, A. J., Dudley, C., Selwood, M. P., Barrow, B. A., Klyne, R., Grew, J. P., McCarthy, M. I., Gloyn, A. L., Owen, K. R.
  2010; 33 (2): 252–57

- **Assessment of high-sensitivity C-reactive protein levels as diagnostic discriminator of maturity-onset diabetes of the young due to HNF1A mutations.** *Diabetes care*
  Owen, K. R., Thanabalasingham, G. n., James, T. J., Karpe, F. n., Farmer, A. J., McCarthy, M. I., Gloyn, A. L.
  2010; 33 (9): 1919–24

- **Naturally Occurring Glucokinase Mutations Are Associated with Defects in Posttranslational S-Nitrosylation** *MOLECULAR ENDOCRINOLOGY*
  Ding, S., Tribble, N. D., Kraft, C. A., Markwardt, M., Gloyn, A. L., Rizzo, M. A.
  2010; 24 (1): 171–77

- **Species-Specific Differences in the Expression of the HNF1A, HNF1B and HNF4A Genes** *PLOS ONE*
  Harries, L. W., Brown, J. E., Gloyn, A. L.
  2009; 4 (11): e7855

- **RD Lawrence Lecture 2009 Old genes, new tricks: learning about blood glucose regulation from naturally occurring genetic variation in humans** *DIABETIC MEDICINE*
  Gloyn, A. L.
  2009; 26 (11): 1083–89

- **Update on Mutations in Glucokinase (GCK), Which Cause Maturity-Onset Diabetes of the Young, Permanent Neonatal Diabetes, and Hyperinsulinemic Hypoglycemia** *HUMAN MUTATION*
  Osbak, K. K., Colclough, K., Saint-Martin, C., Beer, N. L., Bellanne-Chantelot, C., Ellard, S., Gloyn, A. L.
  2009; 30 (11): 1512–26

- **Coexpression of the Type 2 Diabetes Susceptibility Gene Variants KCNJ11 E23K and ABCC8 S1369A Alter the ATP and Sulfonylurea Sensitivities of the ATP-Sensitive K+ Channel** *DIABETES*
  Hamming, K. S. C., Soliman, D., Matemisz, L. C., Niazi, O., Lang, Y., Gloyn, A. L., Light, P. E.
  2009; 58 (10): 2419–24

- **Low Frequency Variants in the Exons Only Encoding Isoform A of HNF1A Do Not Contribute to Susceptibility to Type 2 Diabetes** *PLOS ONE*
  Jafar-Mohammadi, B., Groves, C. J., Owen, K. R., Frayling, T. M., Hattersley, A. T., McCarthy, M. I., Gloyn, A. L.
  2009; 4 (8): e6615

- **Partial lipodystrophy and insulin resistant diabetes in a patient with a homozygous nonsense mutation in CIDEC** *EMBO MOLECULAR MEDICINE*
  Rubio-Cabezas, O., Puri, V., Murano, I., Saudek, V., Semple, R. K., Dash, S., Hyden, C. S. S., Bottomley, W., Vigouroux, C., Magre, J., Raymond-Barker, P., Murgatroyd, P. R., Chawla, et al
  2009; 1 (5): 280–87

- **Type 2 Diabetes Susceptibility Gene TCF7L2 and Its Role in beta-Cell Function** *DIABETES*
  Gloyn, A. L., Braun, M., Rorsman, P.
  2009; 58 (4): 800–802

- **Update of Mutations in the Genes Encoding the Pancreatic Beta-Cell K-ATP Channel Subunits Kir6.2 (KCNJ11) and Sulfonylurea Receptor 1 (ABCC8) in Diabetes Mellitus and Hyperinsulinism** *HUMAN MUTATION*
  Flanagan, S. E., Clauin, S., Bellanne-Chantelot, C., de Lonlay, P., Harries, L. W., Gloyn, A. L., Ellard, S.
  2009; 30 (2): 170–80

- **Severe insulin resistance and intrauterine growth deficiency associated with haploinsufficiency for INSR and CHN2: new insights into synergistic pathways involved in growth and metabolism.** *Diabetes*
  Suliman, S. G., Stanik, J. n., McCulloch, L. J., Wilson, N. n., Edghill, E. L., Misovicova, N. n., Gasperikova, D. n., Sandrikova, V. n., Elliott, K. S., Barak, L. n., Ellard, S. n., Volpi, E. V., Klimes, et al
  2009; 58 (12): 2954–61

- **Mutations in the third gene shown to alter fasting glucose levels in the population (G6PC2) are not a common cause of monogenic forms of pancreatic B-cell dysfunction** *DIABETIC MEDICINE*
  Edghill, E. L., McCulloch, L., Fulton, P., Beer, N., Hattersley, A. T., Gloyn, A. L.
  2009; 26 (1): 113–14

- **Prevalence of GCK mutations in individuals screened for fasting hyperglycaemia** *DIABETOLOGIA*
  Gloyn, A. L., van de Bunt, M., Stratton, I. M., Lonie, L., Tucker, L., Ellard, S., Holman, R. R.
  2009; 52 (1): 172–74

- **The P446L variant in GCKR associated with fasting plasma glucose and triglyceride levels exerts its effect through increased glucokinase activity in liver.** *Human molecular genetics*
  Beer, N. L., Tribble, N. D., McCulloch, L. J., Roos, C. n., Johnson, P. R., Orho-Melander, M. n., Gloyn, A. L.
  2009; 18 (21): 4081–88

- **Identification of a novel beta-cell glucokinase (GCK) promoter mutation (-71G>C) that modulates GCK gene expression through loss of allele-specific Sp1 binding causing mild fasting hyperglycemia in humans.** *Diabetes*
  Gasperíková, D. n., Tribble, N. D., Staník, J. n., Hucková, M. n., Misovicová, N. n., van de Bunt, M. n., Valentínová, L. n., Barrow, B. A., Barák, L. n., Dobránsky, R. n., Bereczková, E. n., Michálek, J. n., Wicks, et al
  2009; 58 (8): 1929–35

- **Genetics: how the UKPDS contributed to determining the genetic landscape of Type 2 diabetes** *DIABETIC MEDICINE*
  Gloyn, A. L., McCarthy, M. I.
  2008; 25: 35–40

- **Activating glucokinase (GCK) Mutations as a cause of medically responsive congenital hyperinsulinism: prevalence in children and characterisation of a novel GCK mutation** *EUROPEAN JOURNAL OF ENDOCRINOLOGY*
  Christesen, H. B. T., Tribble, N. D., Molven, A., Siddiqui, J., Sandal, T., Brusgaard, K., Ellard, S., Njolstad, P. R., Alm, J., Jacobsen, B., Hussain, K., Gloyn, A. L.
  2008; 159 (1): 27–34

- **Permanent neonatal diabetes mellitus caused by a novel homozygous (T168A) glucokinase (GCK) mutation: Initial response to oral sulphonylurea therapy** *JOURNAL OF PEDIATRICS*
  Turkkahraman, D., Brcan, I., Tribble, N. D., Akcurin, S., Ellard, S., Gloyn, A. L.
  2008; 153 (1): 122–26

- **Glucokinase (GCK) and other susceptibility genes for beta-cell dysfunction: the candidate approach**
  Gloyn, A. L., Tribble, N. D., van de Bunt, M., Barrett, A., Johnson, P. R. V.
  PORTLAND PRESS LTD.2008: 306–11

- **Gene duplications resulting in over expression of glucokinase are not a common cause of hypoglycaemia of infancy in humans** *MOLECULAR GENETICS AND METABOLISM*
  van de Bunt, M., Edghill, E. L., Hussain, K., Ellard, S., Gloyn, A. L.
  2008; 94 (2): 268–69

- **Prevalence and clinical characteristics of maternally inherited diabetes and deafness caused by the mt3243A > G mutation in young adult diabetic subjects in Sri Lanka** *DIABETIC MEDICINE*
  Katulanda, P., Groves, C. J., Barrett, A., Sheriff, R., Matthews, D. R., McCarthy, M. I., Gloyn, A. L.
  2008; 25 (3): 370–74

- **Maturity onset diabetes of the young without gene mutation (MODYX) is rare in children with autosomal dominant diabetes in the UK**

Porter, J. R., Ellardw, S., Hattersleyw, A., Gloyn, A., Clarky, P., Barrett, T. G.
WILEY.2008

- **Permanent neonatal diabetes mellitus caused by a novel homozygous (T168A) glucokinase (GCK) mutation: response to oral sulphonylurea therapy**
  Gloyn, A. L., Bircanw, I., Tribble, N. D., Akcurinw, S., Ellard, S., Turkkahramanw, D.
  WILEY.2008

- **Congenital Hyperinsullnism: Prevalence Prevalence of activating glucokinase mutations and a novel mutation**
  Christesen, H., Tribble, N. D., Molven, A., Siddiqi, J., Sandal, T., Brusgaard, K., Ellard, S., Njolstad, P. R., Alm, J., Jacobsen, B., Hussain, K., Gloyn, A. L.
  KARGER.2008: 41

- **Heterogeneity in disease severity in a family with a novel G68V GCK activating mutation causing persistent hyperinsulinaemic hypoglycaemia of infancy** *DIABETIC MEDICINE*
  Wabitsch, M., Lahr, G., Van de Bunt, M., Marchant, C., Lindnert, M., von Puttkamer, J., Fenneberg, A., Debatin, K. M., Klein, R., Ellard, S., Clark, A., Gloyn, A. L.
  2007; 24 (12): 1393–99

- **Mutations in HHEX are not a common cause of monogenic forms of beta cell dysfunction** *DIABETOLOGIA*
  Minton, J. L., van de Bunt, M., Boustred, C., Hussain, K., Hattersley, A. T., Ellard, S., Gloyn, A. L.
  2007; 50 (9): 2019–22

- **Monogenic disorders of the pancreatic beta-cell: personalizing treatment for rare forms of diabetes and hypoglycemia.** *Personalized medicine*
  van de Bunt, M., Gloyn, A. L.
  2007; 4 (3): 247–59

- **Cell biology assessment of glucokinase mutations V62M and G72R in pancreatic beta-cells - Evidence for cellular instability of catalytic activity** *DIABETES*
  Arden, C., Trainer, A., de la Iglesia, N., Scougall, K. T., Gloyn, A. L., Lange, A. J., Shaw, J. A. M., Matschinsky, F. M., Agius, L.
  2007; 56 (7): 1773–82

- **Mutations in ATP-sensitive K+ channel genes cause transient neonatal diabetes and permanent diabetes in childhood or adulthood** *DIABETES*
  Flanagan, S. E., Patch, A., Mackay, D. J. G., Edghill, E. L., Gloyn, A. L., Robinson, D., Shield, J. P. H., Temple, K., Ellard, S., Hattersley, A. T.
  2007; 56 (7): 1930–37

- **Relationship between E23K (an established type II diabetes-susceptibility variant within KCNJ11), polycystic ovary syndrome and androgen levels** *EUROPEAN JOURNAL OF HUMAN GENETICS*
  Barber, T. M., Bennett, A. J., Gloyn, A. L., Groves, C. J., Sovio, U., Ruokonen, A., Martikainen, H., Pouta, A., Taponen, S., Weedon, M. N., Hartikainen, A., Wass, J. A. H., Jarvelin, et al
  2007; 15 (6): 679–84

- **Origin of de novo KCNJ11 mutations and risk of neonatal diabetes for subsequent siblings** *JOURNAL OF CLINICAL ENDOCRINOLOGY & METABOLISM*
  Edghill, E. L., Gloyn, A. L., Goriely, A., Harries, L. W., Flanagan, S. E., Rankin, J., Hattersley, A. T., Ellard, S.
  2007; 92 (5): 1773–77

- **Asian MODY: are we missing an important diagnosis?** *DIABETIC MEDICINE*
  Porter, J. R., Rangasami, J. J., Ellard, S., Gloyn, A. L., Shields, B. M., Edwards, J., Anderson, J. M., Shaw, N. J., Hattersley, A. T., Frayling, T. M., Plunkett, M., Barrett, T. G.
  2006; 23 (11): 1257–60

- **Defining the genetic aetiology monogenic diabetes can improve treatment** *EXPERT OPINION ON PHARMACOTHERAPY*
  Gloyn, A. L., Ellard, S.
  2006; 7 (13): 1759–67

- **Assessment of the role of common genetic variation in the transient neonatal diabetes mellitus (TNDM) region in type 2 diabetes - A comparative genomic and tagging single nucleotide polymorphism approach** *DIABETES*
  Gloyn, A. L., Mackay, D. J. G., Weedon, M. N., McCarthy, M. I., Walker, M., Hitman, G., Knight, B. A., Owen, K. R., Hattersley, A. T., Frayling, T. M.

2006; 55 (8): 2272–76

- **KCNJ11 activating mutations are associated with developmental delay, epilepsy and neonatal diabetes syndrome and other neurological features** *EUROPEAN JOURNAL OF HUMAN GENETICS*
  Gloyn, A. L., Diatloff-Zito, C., Edghill, E. L., Bellanne-Chantelot, C., Nivot, S., Coutant, R., Ellard, S., Hattersley, A. T., Robert, J.
  2006; 14 (7): 824–30
- **Mutations in KCNJ11, which encodes Kir6.2, are a common cause of diabetes diagnosed in the first 6 months of life, with the phenotype determined by genotype** *DIABETOLOGIA*
  Flanagan, S. E., Edghill, E. L., Gloyn, A. L., Ellard, S., Hattersley, A. T.
  2006; 49 (6): 1190–97
- **Mutations in the genes encoding the pancreatic beta-cell K-ATP channel subunits Kir6.2 (KCNJ11) and SUR1 (ABCC8) in diabetes mellitus and hyperinsullinlism** *HUMAN MUTATION*
  Gloyn, A. L., Siddiqui, J., Ellard, S.
  2006; 27 (3): 220–31
- **A gating mutation at the internal mouth of the Kir6.2 pore is associated with DEND syndrome** *EMBO REPORTS*
  Proks, P., Girard, C., Haider, S., Gloyn, A. L., Hattersley, A. T., Sansom, M. S., Ashcroft, F. M.
  2005; 6 (5): 470–75
- **Relapsing diabetes can result from moderately activating mutations in KCNJ11.** *Human molecular genetics*
  Gloyn, A. L., Reimann, F. n., Girard, C. n., Edghill, E. L., Proks, P. n., Pearson, E. R., Temple, I. K., Mackay, D. J., Shield, J. P., Freedenberg, D. n., Noyes, K. n., Ellard, S. n., Ashcroft, et al
  2005; 14 (7): 925–34
- **KCNJ11 activating mutations in Italian patients with permanent neonatal diabetes** *HUMAN MUTATION*
  Massa, O., Iafusco, D., D'Amato, E., Gloyn, A. L., Hattersley, A. T., Pasquino, B., Tonini, G., Dammacco, F., Zanette, G., Meschi, F., Porzio, O., Bottazzo, G., Crino, et al
  2005; 25 (1): 22–27
- **Permanent neonatal diabetes in an Asian infant** *JOURNAL OF PEDIATRICS*
  Porter, Shaw, N. J., Barrett, T. G., Hattersley, A. T., Ellard, S., Gloyn, A. L.
  2005; 146 (1): 131–33
- **Insights into the structure and regulation of glucokinase from a novel mutation (V62M), which causes maturity-onset diabetes of the young.** *The Journal of biological chemistry*
  Gloyn, A. L., Odili, S. n., Zelent, D. n., Buettger, C. n., Castleden, H. A., Steele, A. M., Stride, A. n., Shiota, C. n., Magnuson, M. A., Lorini, R. n., d'Annunzio, G. n., Stanley, C. A., Kwagh, et al
  2005; 280 (14): 14105–13
- **Molecular basis of Kir6.2 mutations associated with neonatal diabetes or neonatal diabetes plus neurological features** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
  Proks, P., Antcliff, J. F., Lippiat, J., Gloyn, A. L., Hattersley, A. T., Ashcroft, F. M.
  2004; 101 (50): 17539–44
- **Mutations in PTF1A cause pancreatic and cerebellar agenesis** *NATURE GENETICS*
  Sellick, G. S., Barker, K. T., Stolte-Dijkstra, Fleischmann, C., Coleman, R. J., Garrett, C., Gloyn, A. L., Edghill, E. L., Hattersley, A. T., Wellauer, P. K., Goodwin, G., Houlston, R. S.
  2004; 36 (12): 1301–5
- **Permanent neonatal diabetes due to mutations in KCNJ11 encoding Kir6.2 - Patient characteristics and initial response to sulfonylurea therapy** *DIABETES*
  Sagen, J. V., Raeder, H., Hathout, E., Shehadeh, N., Gudmundsson, K., Baevre, H., Abuelo, D., Phornphutkul, C., Molnes, J., Bell, G. I., Gloyn, A. L., Hattersley, A. T., Molven, et al
  2004; 53 (10): 2713–18
- **Kir6.2 mutations are a common cause of permanent neonatal diabetes in a large cohort of French patients** *DIABETES*
  Vaxillaire, M., Populaire, C., Busiah, K., Cave, H., Gloyn, A. L., Hattersley, A. T., Czernichow, P., Froguel, P., Polak, M.
  2004; 53 (10): 2719–22

- **Permanent neonatal diabetes due to paternal germline mosaicism for an activating mutation of the KCNJ11 gene encoding the Kir6.2 subunit of the beta-cell potassium adenosine triphosphate channel** *JOURNAL OF CLINICAL ENDOCRINOLOGY & METABOLISM*
  Gloyn, A. L., Cummings, E. A., Edghill, E. L., Harries, L. W., Scott, R., Costa, T., Temple, I. K., Hattersley, A. T., Ellard, S.
  2004; 89 (8): 3932–35

- **Activating mutations in the KCNJ11 gene encoding the ATP-sensitive K+ channel subunit Kir6.2 are rare in clinically defined type 1 diabetes diagnosed before 2 years.** *Diabetes*
  Edghill, E. L., Gloyn, A. L., Gillespie, K. M., Lambert, A. P., Raymond, N. T., Swift, P. G., Ellard, S. n., Gale, E. A., Hattersley, A. T.
  2004; 53 (11): 2998–3001

- **Activating mutations in the gene encoding the ATP-sensitive potassium-channel subunit Kir6.2 and permanent neonatal diabetes.** *The New England journal of medicine*
  Gloyn, A. L., Pearson, E. R., Antcliff, J. F., Proks, P. n., Bruining, G. J., Slingerland, A. S., Howard, N. n., Srinivasan, S. n., Silva, J. M., Molnes, J. n., Edghill, E. L., Frayling, T. M., Temple, et al
  2004; 350 (18): 1838–49

- **Genetics for Endocrinologists: The Molecular Genetic Basis of Endocrine Disorders.** *Clinical endocrinology*
  McCarthy, M. I., Gloyn, A. L.
  2003; 59 (6): 826

- **Identification of 21 novel glucokinase (GCK) mutations in UK and European Caucasians with maturity-onset diabetes of the young (MODY).** *Human mutation*
  Thomson, K. L., Gloyn, A. L., Colclough, K., Batten, M., Allen, L. I., Beards, F., Hattersley, A. T., Ellard, S.
  2003; 22 (5): 417

- **Quantitative traits associated with the Type 2 diabetes susceptibility allele in Kir6.2** *DIABETOLOGIA*
  Weedon, M. N., Gloyn, A. L., Frayling, T. M., Hattersley, A. T., Smith, G. D., Ben-Shlomo, Y.
  2003; 46 (7): 1021-1023

- **The search for type 2 diabetes genes** *AGEING RESEARCH REVIEWS*
  Gloyn, A. L.
  2003; 2 (2): 111–27

- **Glucokinase (GCK) mutations in hyper- and hypoglycemia: maturity-onset diabetes of the young, permanent neonatal diabetes, and hyperinsulinemia of infancy.** *Human mutation*
  Gloyn, A. L.
  2003; 22 (5): 353–62

- **Insights into the biochemical and genetic basis of glucokinase activation from naturally occurring hypoglycemia mutations.** *Diabetes*
  Gloyn, A. L., Noordam, K. n., Willemsen, M. A., Ellard, S. n., Lam, W. W., Campbell, I. W., Midgley, P. n., Shiota, C. n., Buettger, C. n., Magnuson, M. A., Matschinsky, F. M., Hattersley, A. T.
  2003; 52 (9): 2433–40

- **Large-scale association studies of variants in genes encoding the pancreatic beta-cell KATP channel subunits Kir6.2 (KCNJ11) and SUR1 (ABCC8) confirm that the KCNJ11 E23K variant is associated with type 2 diabetes.** *Diabetes*
  Gloyn, A. L., Weedon, M. N., Owen, K. R., Turner, M. J., Knight, B. A., Hitman, G. n., Walker, M. n., Levy, J. C., Sampson, M. n., Halford, S. n., McCarthy, M. I., Hattersley, A. T., Frayling, et al
  2003; 52 (2): 568–72

- **A putative functional polymorphism in the IGF-I gene - Association studies with type 2 diabetes, adult height, glucose tolerance, and fetal growth in UK populations** *DIABETES*
  Frayling, T. M., Hattersley, A. T., McCarthy, A., Holly, J., Mitchell, S. M., Gloyn, A. L., Owen, K., Davies, D., Smith, G. D., Ben-Shlomo, Y.
  2002; 51 (7): 2313–16

- **The role of the HNF4 alpha enhancer in type 2 diabetes variants of the HNF4 alpha enhancer are not a common cause of susceptibility to type 2 diabetes** *MOLECULAR GENETICS AND METABOLISM*
  Mitchell, S. M., Gloyn, A. L., Owen, K. R., Hattersley, A. T., Frayling, T. M.
  2002; 76 (2): 148–51

- **Maturity-onset diabetes of the young caused by a balanced translocation where the 20q12 break point results in disruption upstream of the coding region of hepatocyte nuclear factor-4alpha (HNF4A) gene.** *Diabetes*

Gloyn, A. L., Ellard, S. n., Shepherd, M. n., Howell, R. T., Parry, E. M., Jefferson, A. n., Levy, E. R., Hattersley, A. T.
2002; 51 (7): 2329–33

- **Human calcium/calmodulin-dependent protein kinase II gamma gene (CAMK2G): cloning, genomic structure and detection of variants in subjects with type II diabetes.** *Diabetologia*
  Gloyn, A. L., Desai, M. n., Clark, A. n., Levy, J. C., Holman, R. R., Frayling, T. M., Hattersley, A. T., Ashcroft, S. J.
  2002; 45 (4): 580–83
- **Complete glucokinase deficiency is not a common cause of permanent neonatal diabetes.** *Diabetologia*
  Gloyn, A. L., Ellard, S. n., Shield, J. P., Temple, I. K., Mackay, D. J., Polak, M. n., Barrett, T. n., Hattersley, A. T.
  2002; 45 (2): 290
- **The genetics of type 2 diabetes** *BEST PRACTICE & RESEARCH CLINICAL ENDOCRINOLOGY & METABOLISM*
  Gloyn, A. L., McCarthy, M. I.
  2001; 15 (3): 293–308
- **The beta-cell Ca2+/calmodulin-dependent protein kinase II (CaM kinase II) beta3 isoform containing a proline-rich tandem repeat in the association domain can be found in the human genome.** *Diabetologia*
  Gloyn, A. L., Ashcroft, S. J.
  2001; 44 (6): 787
- **Association studies of variants in promoter and coding regions of beta-cell ATP-sensitive K-channel genes SUR1 and Kir6.2 with Type 2 diabetes mellitus (UKPDS 53).** *Diabetic medicine : a journal of the British Diabetic Association*
  Gloyn, A. L., Hashim, Y. n., Ashcroft, S. J., Ashfield, R. n., Wiltshire, S. n., Turner, R. C.
  2001; 18 (3): 206–12