The Division of Pediatric Genetics and Metabolism, under the direction of Ralph DeBerardinis, M.D., Ph.D., is responsible for the evaluation, diagnosis, and treatment of children with genetic disorders, including birth defects, malformation syndromes, geneticallydefined developmental delays, and inborn errors of metabolism. Approximately one in four admissions to tertiary care in pediatric hospitals result from conditions with a genetic basis. Although many genetic conditions are rare, there are hundreds of these diseases and they collectively account for a disproportionate amount of illness in children and are the leading cause of death in infancy. Furthermore, identifying the genetic basis of rare conditions often leads to specific treatments that dramatically improve the health of the patient.

Ralph DeBerardinis, M.D., Ph.D.
Division Chief

There are three major components to the Division's mission: Patient Care, Research, and Education.

Patient Care

With a large and growing team of physicians, genetic counselors, nurse practitioners, dieticians, and social workers, we are a major regional resource for children and families with genetic diseases. Our team evaluates more than 250 patients each month with known or suspected genetic diseases. Particular strengths of our clinical program include:

- We have the largest regional practice specializing in the diagnosis and treatment of children with inborn errors of metabolism.
- We provide 24/7 coverage for our patients, with an M.D. Medical Geneticist on call at all times.
- We have several clinics specializing in malformation syndromes and genetic forms of developmental delay.
- We are experts in the use and interpretation of advanced genetic diagnostics, including tests involving next-generation sequencing.
- We are the only clinic in Dallas, and currently the only one in North Texas, that accepts referrals from the Texas Department of Health's newborn screening program for biochemical disorders.
- We participate in multidisciplinary clinics specializing in relatively common disorders such as Down syndrome and 22q11.2 deletion syndrome.

Research

Our clinical team is unique in that it is fully synchronized with a state-of-the-art research program in the Children's Research Institute (CRI), a joint venture between UT Southwestern and Children's Health. The Genetic and Metabolic Disease Program (GMDP) within the CRI is comprised of a team of scientists dedicated to identifying new genetic diseases and developing new ways to treat children with genetic disorders. We use advanced technologies to evaluate each patient's genetic and metabolic individuality. Laboratory-based approaches in cellular and molecular biology are then used to understand the precise consequences of the DNA mutations identified in our patients. Our research team is funded through federal, state, and private grant support. Specific research goals within the GMDP include efforts to:

- Discover new genetic causes of childhood diseases.
- Understand the genetic basis of metabolic diversity and its relationship to health and disease in children.
- Develop new diagnostic methods to detect genetic diseases in patients.
- Establish clinical trials to assess the effect of new treatments.
- Construct new disease models using geneticallymodified mice and use them to test the effect of experimental therapies.
- Use multidisciplinary approaches (chart review, public health databases, two- and three-dimensional image analyses, etc.) to identify and characterize novel malformation syndromes.

Education

We are a vital part of UT Southwestern Medical Center's mission to train medical students, residents, fellows, and allied health professionals in pediatrics, genetics, and metabolism. We teach medical students and pediatric residents throughout their training, manage an accredited residency program to train the next generation of physicians in Medical Genetics, co-direct a fellowship program in Laboratory Genetics and Genomics, and provide continuing medical education in genetics and metabolism to the Dallas-Fort Worth medical community.

Faculty

The Division has six full-time faculty members (four with primary appointments in Pediatrics, one in Internal Medicine, and one in the Children's Research Institute), all with interests in the diagnosis and management of a variety of genetic conditions such as inborn errors of metabolism, newborn screening, lysosomal storage disorders, craniofacial malformation syndromes, and incontinentia pigmenti.

Laura Mackay, M.D., M.P.H.
Assistant Professor

M.D.
Texas Tech University Health Sciences Center, El Paso, TX, 2017
M.P.H.
University of Texas at Houston School of Public Health, El Paso, TX, 2017
Postdoctoral Training
Residency, Pediatrics and Medical Genetics
Baylor College of Medicine, Houston, TX, 2017 – 2021
Fellowship, Medical Biochemical Genetics
Baylor College of Medicine, Houston, TX 2021-2022
Interests
Metabolic disorders, newborn screening, intellectual disability syndromes

Honors / Awards

Best Pediatric Specialists, *D Magazine*

- Ralph DeBerardinis
- Garrett Gotway
- Angela Scheuerle
- Luis Umaña

Texas Super Doctor, Texas Monthly

- Angela Scheuerle
- Luis Umaña

Ralph DeBerardinis

- Clarivate Highly-Cited Researcher

Luis Umaña

- Promoted to Associate Professor

Invited Lectures

Ralph DeBerardinis

- Department of Nutritional Sciences Graduate Student Association Seminar, University of Texas at Austin, 2022
  - *"The role of metabolic reprogramming in human cancer and monogenic diseases"*
- Agios - Genetically Defined Diseases Learning Series, Virtual, 2022
  - *"Human Metabolic Outliers"*
- Georgetown-Lombardi Oncology Grand Rounds, Virtual, 2022
  - *"Metabolic reprogramming in human cancer"*
- Forbeck Forem: Diet and Metabolic Therapeutics in Cancer- Towards a Molecular Understanding, Pacific Grove, CA, 2022
  - *"Human tumor metabolism & cancer progression"*
- The Science of Childhood Cancer Lecture Series; St. Jude, Virtual, 2022
  - *"Human Metabolic Outliers."*
- Paul Marks Prize Symposium, New York City, NY, 2022
  - *"Metabolic reprogramming in cancer and other diseases"*
- University of Rochester Grand Rounds, Virtual, 2022
  - *"Metabolic outliers in human disease: deep metabolic phenotyping in mendelian diseases"*
- American Society of Biochemistry and Molecular Biology, Philadelphia, PA, 2022
  - *"Metabolic Outliers in Human Disease"*
- Northwestern University, David W. Cugell Honorary Lectureship, Internal Medicine Grand Rounds, Virtual, 2022
  - *"Metabolic Outliers in Human Disease: deep metabolic phenotyping in Mendelian Disorders"*

- University of Alabama – Burmingham, O'Neal Research Seminar, Virtual, 2022
    - *"Metabolic Phenotypes and Liabilities in Human Cancer"*
- Children's Hospital of Philadelphia, Michael Palmieri Lectureship in Metabolism, Pediatrics Grand Rounds, Philadelphia, PA, 2022
    - *"Metabolic Outliers in Human Disease: deep metabolic phenotyping in Mendelian Disorders"*
- Academy of Kidney Cancer Investigators, Virtual, 2022
    - *"Metabolic phenotypes and cancer progression in humans"*
- Susan Swerling Lecture, Dana Farber Cancer Institute, Virtual, 2022
    - *"Metabolic Phenotypes and Liabilities in Human Cancer"*
- Weill-Cornell Cancer Metabolism and Inflammation Symposium, New York, NY, 2022
    - *"Metabolic Phenotypes and Liabilities in Human Cancer"*
- Dutch Translational Medicine, Virtual, 2022
    - *"Metabolic Outliers in Human Disease"*
- EMBL Symposium on Inter-Organ Communication, Virtual, 2022
    - *"Metabolic Outliers in Human Disease"*
- FUSION Conference on Metabolism in Health and Disease, Cancun, Mexico, 2022
    - *"Mendelian anomalies in human growth and development"*
- Lake Como Cancer Meeting, Lake Como, Italy, 2022
    - *"Metabolic phenotypes and cancer progression in humans"*

Angela Scheuerle

- Webinar, May 2022
    - *"Is it Safe? Safety Surveillance through the Antiretroviral Pregnancy Registry."*
- Hope Cottage Family Education, Dallas, TX, September 2022
    - *"Teratogens"*

Luis Umaña

- IV simposio latinoamericano de Genetica Medica, Bogota Colombia, September 2022 Asociacion Colombiana de Medicos Genetistas ACMGEN
    - *"Presimposio Jornada en Tamizacion neonatal: Ruta de atencion para Biotinidasa y Galactosmeia"*

Other Conferences

Benjamin RH, Mitchell LE, **Scheuerle AE**, Langlois PH, Canfield MA, Drummond-Borg M, Nguyen JM, Agopian AJ
International Clearinghouse for Birth Defects Surveillance and Research, Bologna, Italy, September 2022
Poster Presentation, *"Syndrome identification in surveillance systems using CDC/BPA coding"*

Du H, Jolly A, Grochowski CM, Yuan B, Dawood M, Jhangiani SN, Fatih JM, Coban-Akdemir Z, Carlin ME, **Scheuerle AE**, Witzl K, Posey JE, Pendleton M, Harrington E, Juul S, Hastings PJ, Bi W, Gibbs RA, Sedlazeck FJ, Lupski JR, Carvalho CMB, Liu P
Genomics of Rare Disease, Wellcome Genome Campus, UK, March 2022
Poster Presentation, *"The multiple de novo copy number variant (MdnCNV) phenomenon presents with peri-zygotic DNA mutational signatures and multilocus pathogenic variation"*

Morrison M, Patel S, Saukam S, Willard A, Santiago MG, Martinez D, Miller V, Jacobs M, **Scheuerle AE**, Koduru P
American College of Medical Genetic (ACMG), Nashville, TN, March 2022
Poster Presentation, *"Isodicentric (Y)(p11.2) mosaicism in newborn with 46,XX cells"*

Renwick A, Schraw JM, Desrosiers TA, Janitz AE, Scheurer ME, Canfield MA, Langlois PH, **Scheuerle AE**, Plon SE, Lupo PJ
American Association for Cancer Research, New Orleans, LA, April 2022
Poster Presentation, *"A population-based assessment of cancer risk in children with recurrent multiple congenital anomalies"*

Scheuerle AE, Ni M, Ahmad AA, Boothe M
David W Smith Workshop on Malformations and Morphogenesis, Norfolk, VA, August 2022
Poster Presentation, *"Biallelic variants n NUDCD2 cause a multiple malformation syndrome with cholestasis and renal failure"*

Education and Training

The Division of Pediatric Genetics and Metabolism is committed to providing quality medical education for medical students, residents, and fellows.

Medical Student Education

Genetics is an essential component of all facets of medicine, and the Division of Pediatric Genetics and Metabolism is proud to play a major role in the education of medical students and other trainees within the UT Southwestern system.

First-Year Medical Students

We are highly involved in the first-year medical school curriculum, including:

- Macromolecules Course: protein and amino acid metabolism, hyperammonemia and urea cycle defects, defects in amino acid metabolism (PKU, MSUD, etc.), purine and pyrimidine metabolism, and treatment of inborn errors of metabolism

Second-Year Medical Students

We offer a competitive, paid Summer Genetics Fellowship for rising second-year medical students that is an intense Clinical Genetics experience that includes:

- Two weeks in the cytogenetics lab and one in the molecular diagnostic lab
- Three weeks rotating through various clinics focusing on genetic diseases
- Two weeks attending the Cold Spring Harbor Human Genetics Course (paid for by the program)
- Completion of a formal project for presentation in Austin
- Trip to Austin for conference and tour of the Newborn Screening Laboratory services

Third-Year Medical Students

Third-year medical students participate in pediatrics rotations involving:

- Case studies in clinical genetics
- Genetics clinic outpatient rotations
- Clinical genetics consultations

Fourth-Year Medical Students

We offer an elective in clinical genetics to fourth-year medical students involving outpatient genetics clinics and inpatient genetics consultations.

Dr. Gotway is also a co-director in the Frontiers in Medicine course. He organizes and teaches a section that focuses on new developments in clinical genetics and human development for graduating students about to start residencies in Pediatrics and Obstetrics and Gynecology.

Medical Genetics Interest Group

We provide mentorship to UT Southwestern medical students considering a career in medical genetics. This highly successful interest group meets periodically to discuss new developments in clinical and research-based genetics. We seek to provide an environment to educate students about career opportunities in this exciting and rapidly expanding area of pediatrics.

Resident Education

We play a major role in the education of residents at UT Southwestern. Some of our activities include:

Medical Genetics Residency Program

The Department of Pediatrics, through the Division of Pediatric Genetics and Metabolism, is the sponsoring clinical department for our Accreditation Council for Graduate Medical Education-certified training program in Medical Genetics. Medical Genetics is a specialty of its own, rather than being a subspecialty of Pediatrics, Internal Medicine, or Obstetrics/Gynecology. The training program encompasses many clinical departments at UT Southwestern, including Pediatrics, Internal Medicine, Obstetrics/Gynecology (prenatal diagnosis), Neurology, and Pathology (Clinical Molecular Genetics, Cytogenetics, and Biochemical Genetics), among others. The program is directed by Garrett Gotway, M.D., Ph.D., a board-certified pediatrician and medical geneticist. The program has recently changed administrative support to the Department of Pediatrics. Learn more about the Medical Genetics program.

Pediatrics Residency Program

The Division provides didactic teaching for the pediatric residents, including but not limited to:

- Clinical dysmorphology
- Teratology
- Cause and evaluation of birth defects
- Common chromosome anomalies
- Newborn screening
- Acute metabolic disorders
- Genetic storage disorders

We provide direct teaching for the residents in the regular departmental clinical conferences, as well as part of our inpatient consultation service.

Finally, there is a Clinical Genetics elective available for second- and third-year pediatric residents. The residents see outpatients in our clinics and inpatients for consultation services under the supervision of one of the faculty. We encourage the residents to participate in clinical research projects if they are interested.

Other Specialties

Trainees in other departments also spend time in our clinics. Residents in Neurology and other specialties may receive some of their training through our Division. This includes the Laboratory Genetics and Genomics Fellow and Molecular Pathology Fellow in the Department of Pathology, who have dedicated clinical rotations in both Metabolic Genetics and General Genetics.

Graduate Student and Postdoctoral Fellow Education

We participate in a variety of courses for students pursuing Ph.Ds. and post-doctoral training, including seminars on:

- Human genetics and genomics
- Mendelian genetic diseases
- The use of metabolic tracers and metabolomics in the evaluation of human diseases
- Cancer metabolism
- Regulation of metabolic pathways in health and disease
- Informatic analysis of high-content genomic and metabolomic data sets

The Clinical Genetics Division is an important component of the Laboratory Genetics and Genomics (LGG) Fellowship in the Department of Pathology. The fellow is required to spend a total of six weeks in clinical genetics and biochemical genetics (in addition to rotations in cancer and prenatal genetics). Previously, the fellow would spend a dedicated month in clinic with us, but this led to over-representation of common disorders that might not have the highest relevance to the fellow's training agenda. This year, we are notifying the fellow when specific patients of interest are due in clinic so that she may be present either in person or virtually as appropriate. This has the advantage of allowing the fellow to concentrate on patients with cytogenetic or molecular diagnosis, testing, and counseling, thus providing a better, broader, and more relevant, clinical experience for her. It also helps with the overall LGG program scheduling.

Research Activities

Our faculty are involved in clinical, epidemiologic, translational, and basic research projects. Our overarching goal is to better understand the genetic basis of human disease and to advance new knowledge into new approaches to diagnose and treat our patients. Our large and varied patient population inspires research in numerous areas.

The laboratory of Dr. DeBerardinis is interested in how perturbed metabolic states contribute to tissue dysfunction and disease. The lab studies two diseases related to cellular metabolic anomalies: cancer and inborn errors of metabolism. In both cases, the lab seeks to develop hypotheses about disease mechanisms from the direct examination of patients and then use experimental models to test these hypotheses. In cancer, the lab pioneered the use of stable isotopes tracers (e.g. heavy carbon, ^{13}C) to analyze metabolic pathways in tumors in vivo, both in mice and in patients. To examine tumor metabolism in patients, isotope labeled nutrients (e.g., ^{13}C-glucose) are infused into cancer patients during surgical tumor resection and then metabolites are extracted from the tumor and adjacent tissue. These metabolites are analyzed by mass spectrometry, and computational methods are used to detect labeling differences between the tumor and adjacent tissue or between tumors with different properties. Hypotheses about the role of particular metabolic pathways on cancer progression are then studied in mouse models of cancer, where the pathway can be pharmacologically or genetically silenced to test its requirement for maximal tumor growth rate, therapy resistance, metastasis, etc. These approaches have led to international recognition for the DeBerardinis lab's contributions to the field of cancer metabolism, including the 2021 Paul Marks Prize in Cancer Research.

In inborn errors of metabolism, the DeBerardinis laboratory uses clinical resources generated through the Genetic and Metabolic Disease Program described above. An integrated analysis of genomics and metabolomics data from patients suspected to have genetic diseases allows the laboratory to pinpoint the mutation causing the disease. Mutations of particular interest, especially ones that point to genes not previously connected with human diseases, are then modeled in genetically-modified mice to help uncover disease mechanisms and to explore therapeutic opportunities.

Dr. Gotway is collaborating with the McDermott Center for Human Growth and Development to enhance the discovery of new gene-disease associations in patients with novel clinical presentations. The Human Gene Discovery Laboratory will analyze whole exome and genome data from patients with unknown clinical syndromes to identify variants in novel genes that will expand our knowledge and understanding of human genetics.

Dr. Scheuerle is a co-investigator on A.J. Agopian's study from The University of Texas School of Public Health titled "A Multidisciplinary Approach for Identifying and Characterizing Novel Congenital Malformation Syndromes" (NIH 1R01HD093660-01A1). This study uses a combination of Texas Department of State Health Services Birth Defects Registry data and chart review with the goal of identifying previously unrecognized malformation associations. Additionally, this study links birth defects with other health databases, such as cancer, to evaluate potential associations. Dr. Scheuerle is also involved in research exploring the VATER/VACTERL association (a collaboration with Dr. Agopian); the developmental consequences of ribosomal dysfunction (a collaboration with Michael Buszczak in the Department of Molecular Biology); and Incontinentia Pigmenti, particularly elucidating the adult phenotype and advancing understanding of the molecular and clinical aspects of this disease.

Clinical Activities

We accept referrals from all pediatricians and children's hospitals in the Dallas-Fort Worth metroplex, as well as from more distant areas within and beyond Texas. The Division's clinical activities at Children's Medical Center Dallas are focused in the following areas:

Metabolic Disease Clinics

The Metabolic Diseases Clinic provides evaluation and testing for children with known or suspected inborn errors of metabolism (IEMs). IEMs are a family of hundreds of rare diseases caused by mutations in the genes that allow the body to produce energy and grow. We are a regional center of excellence in these diseases, establishing the diagnosis in affected children, counseling and educating their families about these conditions, and optimizing therapy tailored to the needs of each child. Blood, urine, enzyme, and DNA analyses are performed for diagnosis. Patients with a confirmed diagnosis are then provided with nutritional evaluation, genetic counseling, and psychosocial assessment, as well as long-term care.

The Metabolic Disease Clinic is closely associated with the Newborn Screening Clinic. We are a major referral center for the Texas Newborn Screening Program. This statewide program seeks to identify newborn babies with any of over 30 different treatable diseases, many of which are genetic/metabolic in nature. A large fraction of the approximately 400,000 babies born in Texas each year are evaluated through our Division. When a baby in North Texas is found to have a metabolic abnormality on the newborn screen, the family is referred to our team for definitive diagnosis, treatment, and long-term care if necessary. Through the Texas Newborn Screening Program, more than 75 children with genetic metabolic diseases are identified each year, and the coordinated care of these children by the Metabolic Disease Clinic at Children's significantly improves their development and survival. Efforts in newborn screening are led by Dr. Luis Umaña.

A dedicated clinic is also provided for teenagers with IEMs transitioning into adult medicine. This clinic at Children's is staffed by Markey McNutt, M.D., Ph.D, who is board certified in both Medical Genetics and Internal Medicine, and follows these patients after age 18 at a clinic in the Aston Center.

Genetics/Dysmorphology Clinic

Children with conditions involving birth defects, developmental delay or mental retardation, or other known or suspected genetic disorders receive evaluation and testing in the Genetics/Dysmorphology Clinic. Chromosomal and DNA analysis for the diagnosis of genetic disease is provided, as well as psychosocial assessment, counseling, and comprehensive case management with referral to medical specialists, community resources, and support groups. Family history analysis and risk counseling to discuss reproductive options also are available through a team of board-certified genetic counselors. As of August 2016, this clinic has been available at the Children's Specialty Center at THR Presbyterian in addition to the Children's Health Dallas campus.

Down Syndrome Clinic

Faculty and staff with the Down Syndrome Clinic have decades of experience in caring for children with Down syndrome and provide comprehensive treatment for children and their families, including medical management, genetic counseling, physical, speech, and motor development evaluation and recommendations, psychosocial support, screening and referral for behavioral and psychiatric problems, and referral to community agencies for educational intervention or therapies. New patients are seen at the Children's Health Dallas campus with follow-up available there and by telemedicine. There is also a Transition Clinic for adolescents with Down syndrome to help them transition to adulthood.

Interdivisional and Interdepartmental Collaborations

The genetic basis of many human diseases, and the broad utility of genetic testing across numerous subspecialties of Pediatrics and Internal Medicine, make the consultative services of our physicians essential to the clinical and academic missions of UT Southwestern.

In 2021, the Division of Pediatric Genetics and Metabolism led an effort to have UT Southwestern designated as a Rare Disease Center of Excellence (COE) by the National Organization of Rare Disorders (NORD). NORD advocates for patients of all ages suffering from about 7,000 rare diseases, many of which have genetic causes and manifest in childhood. UT Southwestern is among an inaugural group of 31 medical centers participating in this network. The new COE is directed by Dr. Scheuerle and involves over 30 physicians from seven clinical departments. The COE designation recognizes UT Southwestern's long-standing excellence in both clinical care and research in rare disorders and is expected to attract patients seeking advanced treatment for these diseases. UTSW faculty across departments are active in nationallevel working groups within the COE network. The designation also boosts our efforts to recruit the best clinicians, researchers and trainees in medical genetics and related specialties. Under Dr. Scheuerle's direction, the COE supports academic retreats focused on rare disease clinical care and research and campus-wide educational efforts in rare diseases.

Initial activities of the COE in 2022 include: the organization of Rare Disease Day activities on the UTSW campus, the establishment of a rare disease clinic rotation for pediatric residents, and the presentation of our first annual Rare Disease Symposium. UTSW has established a rare disease website within swmed.org dedicated to the CoE. There is a Twitter account managed by one of the graduate students. Currently anticipated activities for 2023 include a UTSW "Day at the Zoo" for Rare Disease Day, expansion of the Symposium, and hosting the 2023 fall in-person COE Directors Meeting (Oct. 13-14).

Dr. Scheuerle sees adult patients referred from both UTSW and community obstetricians and maternal fetal medicine specialists for concerns about genetic disease risk. These include women for whom the concern of a congenital malformation was raised by prenatal imaging or other diagnostic testing. Many of these encounters are coordinated through the Children's FETAL program. She also serves on the Parkland Hospital Stillbirth Committee, an organ of the Obstetrics & Gynecology Department that reviews all the Parkland Hospital stillbirths. Previous discussions with obstetricians and neonatologists at Clements University Hospital have resulted in increased referrals for prenatal counseling as well as the establishment of the Genetics eConsult at that hospital.

The 22q11.2 multidisciplinary clinic is expanding. We have modified the criteria for managing patients that are new to the Genetics and Metabolism Division. If the patient is less than a year from diagnosis (regardless of age), they will be seen as a separate new patient visit in the Genetics clinic. If the patient is more than a year from diagnosis, their first visit will be with the 22q11.2 multidisciplary clinic on the second Wednesday afternoon of the month. All established patients will continue to be seen in the multidisciplinary clinic at defined follow-up time intervals. A separate appointment in the Genetics clinic will be arranged for any patient/parent who requests it or who would benefit from it.

Current Grant Support

Ralph DeBerardinis

Grantor: Howard Hughes Medical Institute
Title of Project: HHMI Investigator Program
Role: Principal Investigator
Dates: 09/2018 – 08/2025

Grantor: Lawrence Steinberg Endowment
Title of Project: Joel B. Steinberg, M.D. Distinguished Chair in Pediatrics
Role: Principal Investigator
Dates: 12/2018 – Ongoing

Grantor: Robert L. Moody, Sr. Faculty Scholar Endowment
Title of Project: Moody Faculty Scholar
Role: Principal Investigator
Dates: 10/2018 – Ongoing

Grantor: NIH-National Cancer Institute - 2P50CA070907-21A1
Title of Project: The University of Texas SPORE (Special Program of Research Excellence) in Lung Cancer
Role: PI of Project 1 (Overall PIs: John Minna and Jack Roth)
Dates: 09/2020 – 08/2025

Grantor: NIH-National Cancer Institute
Title of Project: The UT Southwestern Medical Center SPORE in Kidney Cancer
Role: Co-PI of Project 3 (Overall PI: James Brugarolas)
Dates: 08/2022 – 07/2027

Grantor: NIH-National Cancer Institute
Title of Project: Investigating the Role of Metabolic Reprogramming in Cancer Cell Death Sensitivity
Role: Principal Investigator
Dates: 04/2020 – 03/2024

Angela Scheuerle

Grantor: National Institutes of Health/National Institute of Child Health and Human Development (R01)
Title of Project: A Multidisciplinary approach for identifying and characterizing novel congenital malformation syndromes
Role: Co-Investigator – Local/UTSW Principal Investigator
Dates: 09/2018 – 09/2023

Peer-Reviewed Publications

1. Aurora AB, Khivansara V, Leach A, Gill JG, Martin-Sandoval M, Yang C, Kasitinon SY, Bezwada D, Tasdogan A, Gu W, Mathews TP, Zhao Z, **DeBerardinis RJ**, Morrison SJ. Loss of glucose 6-phosphate dehydrogenase function increases oxidative stress and glutaminolysis in metastasizing melanoma cells. *Proc Natl Acad Sci U S A.* 2022 Feb 8;119(6):. PMID:35110412

2. Baytas O, Davidson SM, **DeBerardinis RJ**, Morrow EM. Mitochondrial enzyme GPT2 regulates metabolic mechanisms required for neuron growth and motor function in vivo. *Hum Mol Genet.* 2022 Feb 21;31(4):587-603. PMID:34519342

3. Benjamin RH, **Scheuerle AE**, Scott DA, Navarro Sanchez ML, Langlois PH, Canfield MA, Northrup H, Schaaf CP, Ray JW, McLean SD, Chen H, Swartz MD, Lupo PJ, Agopian AJ. Birth defect co-occurrence patterns in the Texas Birth Defects Registry. *Pediatr Res.* 2022 Apr;91(5):1278-1285. PMID:34193968

4. Bull MJ, Trotter T, Santoro SL, Christensen C, Grout RW, COUNCIL ON GENETICS, Burke LW, Berry SA, Geleske TA, Holm I, Hopkin RJ, Introne WJ, Lyons MJ, Monteil DC, **Scheuerle A**, Stoler JM, Vergano SA, Chen E, Hamid R, Downs SM, Grout RW, Cunniff C, Parisi MA, Ralston SJ, Scott JA, Shapira SK, Spire P. Health Supervision for Children and Adolescents With Down Syndrome. *Pediatrics.* 2022 May 1;149(5):. PMID:35490285

5. Cai L, **DeBerardinis RJ**, Xiao G, Minna JD, Xie Y. A Pan-Cancer Assessment of RB1/TP53 Co-Mutations. *Cancers (Basel).* 2022 Aug 30;14(17):. PMID:36077736

6. Chen R, Wang J, Gradinaru I, Vu HS, Geboers S, Naidoo J, Ready JM, Williams NS, **DeBerardinis RJ**, Ross EM, Collins JJ 3rd. A male-derived nonribosomal peptide pheromone controls female schistosome development. *Cell.* 2022 Apr 28;185(9):1506-1520.e17. PMID:35385687

7. Clowse M, Fischer-Betz R, Nelson-Piercy C, **Scheuerle AE**, Stephan B, Dubinsky M, Kumke T, Kasliwal R, Lauwerys B, Förger F. Pharmacovigilance pregnancy data in a large population of patients with chronic inflammatory disease exposed to certolizumab pegol. *Ther Adv Musculoskelet Dis.* 2022;14():1759720X221087650. PMID:35464812

8. **DeBerardinis RJ**, Keshari KR. Metabolic analysis as a driver for discovery, diagnosis, and therapy. *Cell.* 2022 Jul 21;185(15):2678-2689. PMID:35839759

9. Du H, Jolly A, Grochowski CM, Yuan B, Dawood M, Jhangiani SN, Li H, Muzny D, Fatih JM, Coban-Akdemir Z, Carlin ME, **Scheuerle AE**, Witzl K, Posey JE, Pendleton M, Harrington E, Juul S, Hastings PJ, Bi W, Gibbs RA, Sedlazeck FJ, Lupski JR, Carvalho CMB, Liu P. The multiple de novo copy number variant (MdnCNV) phenomenon presents with peri-zygotic DNA mutational signatures and multilocus pathogenic variation. *Genome Med.* 2022 Oct 27;14(1):122. PMID:36303224

10. Kaushik AK, Tarangelo A, Boroughs LK, Ragavan M, Zhang Y, Wu CY, Li X, Ahumada K, Chiang JC, Tcheuyap VT, Saatchi F, Do QN, Yong C, Rosales T, Stevens C, Rao AD, Faubert B, Pachnis P, Zacharias LG, Vu H, Cai F, Mathews TP, Genovese G, Slusher BS, Kapur P, Sun X, Merritt M, Brugarolas J, **DeBerardinis RJ**. In vivo characterization of glutamine metabolism identifies therapeutic targets in clear cell renal cell carcinoma. *Sci Adv.* 2022 Dec 16;8(50):eabp8293. PMID:36525494

11. Kaymak I, Luda KM, Duimstra LR, Ma EH, Longo J, Dahabieh MS, Faubert B, Oswald BM, Watson MJ, Kitchen-Goosen SM, DeCamp LM, Compton SE, Fu Z, **DeBerardinis RJ**, Williams KS, Sheldon RD, Jones RG. Carbon source availability drives nutrient utilization in CD8(+) T cells. *Cell Metab.* 2022 Sep 6;34(9):1298-1311.e6. PMID:35981545

12. Kilgour MK, MacPherson S, Zacharias LG, LeBlanc J, Babinszky S, Kowalchuk G, Parks S, Sheldon RD, Jones RG, **DeBerardinis RJ**, Hamilton PT, Watson PH, Lum JJ. Principles of reproducible metabolite profiling of enriched lymphocytes in tumors and ascites from human ovarian cancer. *Nat Protoc.* 2022 Nov;17(11):2668-2698. PMID:35986218

13. Lesner NP, Wang X, Chen Z, Frank A, Menezes CJ, House S, Shelton SD, Lemoff A, McFadden DG, Wansapura J, **DeBerardinis RJ**, Mishra P. Differential requirements for mitochondrial electron transport chain components in the adult murine liver. *Elife.* 2022 Sep 26;11():. PMID:36154948

14. Liao C, Glodowski CR, Fan C, Liu J, Mott KR, Kaushik A, Vu H, Locasale JW, McBrayer SK, **DeBerardinis RJ**, Perou CM, Zhang Q. Integrated Metabolic Profiling and Transcriptional Analysis Reveals Therapeutic Modalities for Targeting Rapidly Proliferating Breast Cancers. *Cancer Res.* 2022 Feb 15;82(4):665-680. PMID:34911787

15. MacPherson S, Keyes S, Kilgour MK, Smazynski J, Chan V, Sudderth J, Turcotte T, Devlieger A, Yu J, Huggler KS, Cantor JR, **DeBerardinis RJ**, Siatskas C, Lum JJ. Clinically relevant T cell expansion media activate distinct metabolic programs uncoupled from cellular function. *Mol Ther Methods Clin Dev.* 2022 Mar 10;24():380-393. PMID:35284590

16. **McNutt MC**, Foreman N, **Gotway G**. Arginase 1 Deficiency in Patients Initially Diagnosed with Hereditary Spastic Paraplegia. Mov Disord Clin Pract. 2022 Nov 22;10(1):109-114. PMID: 36698992

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