A RARE DISEASE RESOURCE GUIDE
FOR INDIVIDUALS, FAMILIES
AND CAREGIVERS IN ALABAMA

PRESENTED BY THE ALABAMA RARE DISEASE ADVISORY COUNCIL
SUPPORTING PEOPLE WITH RARE DISEASES AND THEIR FAMILIES

Living with a rare disease is a unique experience that, for many, can often feel like travelling an uncharted path with unexpected twists and turns. You or a loved one may have lived with a rare disease for years. Or, you may be newly diagnosed or currently seeking a diagnosis. The good news is that hundreds of resources are available – both nationally and within Alabama – to help you find information, support, and understanding for the journey ahead.

The Alabama Rare Disease Advisory Council developed this Rare Disease Resource Guide to provide helpful information and resources to assist and support people with rare diseases and their families. We hope that this resource booklet will help you learn more about your rare disease and connect with a supportive rare disease patient community. Also, this guide includes resources to help you locate experienced clinicians and peer advocates, identify state and federal resources, and find important research and clinical trials. With information, support, and experienced medical providers, you will have the most valuable tools for navigating your journey successfully.

We hope that this resource booklet will help you learn more about your rare disease and connect with a supportive rare disease patient community.
ABOUT THE ALABAMA RARE DISEASE ADVISORY COUNCIL

Alabama Governor Kay Ivey established the Alabama Rare Disease Advisory Council in 2017 to serve as an advisory body to the governor and the Alabama Legislature. Our focus is on research, diagnosis, treatment, and education regarding rare diseases and their economic impact on the state. Chaired by Associate Dean for Genomic Medicine and Chief Genomics Officer of UAB Medicine Bruce R. Korf, MD, PhD, our Council comprises 17 members representing distinct areas of expertise in medicine, research, advocacy, and government policy. The governor nominates each member to fulfill a three-year term of service.

Bruce R. Korf, MD, PhD
Council Chair

Dr. Korf is a medical geneticist with board certification in clinical genetics, clinical cytogenetics, clinical molecular genetics, pediatrics, and neurology (child neurology). He serves as Associate Dean for Genomic Medicine and Chief Genomics Officer of UAB Medicine and Director of Rare Disorders in the UAB Hugh Kaul Personalized Medicine Institute. Over the past seven years, he has led the UAB Undiagnosed Diseases Program, which is modeled on the original Undiagnosed Diseases Program at the NIH. His role at UAB involves working with clinicians across the health system to integrate genetics and genomics into clinical practice. Dr. Korf serves as co-PI for a project in the Clinical Sequencing Evidence-Generating Research Consortium aimed at establishing the clinical utility of whole genome sequencing in diagnosis of sick newborns. He also leads the Alabama Genomic Health Initiative, which is a state-funded effort to introduce genomic medicine to citizens and health practitioners in the state. Finally, he is the contact PI for the Southern All of Us Network, which is part of the national All of Us research initiative. He has also been active nationally and internationally in genetics and genomic medicine. He has served as president of the Association of Professors of Human and Medical Genetics and the American College of Medical Genetics and Genomics (ACMG), and now is the president of the ACMG Foundation for Genetic and Genomic Medicine. He is also on the steering committee of the Global Genomic Medical Collaborative. Finally, he is the editor-in-chief of the American Journal of Human Genetics, the official journal of the American Society of Human Genetics (ASHG).

Matthew Alexander, PhD

Dr. Alexander currently serves as Assistant Professor in Pediatric Neurology and Genetics and Center for Exercise Medicine (UCEM) Education Chair at the University of Alabama at Birmingham (UAB), where he maintains a research laboratory focused on identifying novel epigenetic and genetic regulators of human neuromuscular diseases and generating novel zebrafish models of disease for drug screening purposes. Named a Pittman Scholar in 2019, he obtained a PhD in genetics and development from the University of Texas Southwestern Medical Center at Dallas where he studied the functional roles of forkhead transcription factors in the regulation of muscle cells. He later completed a postdoctoral fellowship at Harvard Medical School where his work focused on the roles of epigenetic and genetic modifiers of human neuromuscular disease with an emphasis on Duchenne muscular dystrophy (DMD), myotonic dystrophy (DM), and Limbgirdle muscular dystrophy (LGMD).
Kristin Anthony

Ms. Anthony serves as President of the PTEN Hamartoma Tumor Syndrome Foundation, an organization she founded in 2013 after a personal diagnostic odyssey led to her diagnosis of Cowden Syndrome, a rare disease associated with a mutation in the tumor-suppressor gene PTEN that increases the risk of certain cancers in affected individuals. A three-time cancer survivor, Ms. Anthony advocates for research funding for PTEN as well as other rare diseases, lobbying for policy that will benefit affected patients and families. Her efforts to support PTEN syndrome research have been recognized by several national organizations including Stand Up to Cancer, the National Organization for Rare Disorders (NORD) and the American Cancer Society.

Elizabeth Barnby, DNP, ACNP-BC, FNP-BC, CRNP

Dr. Barnby currently serves as Clinical Associate Professor of Nursing at the University of Alabama in Huntsville (UAH), and she has extensive clinical expertise in acute care, primary care, and pediatric nursing as well as informatics and genetics/genomics. Her research interests include inborn errors of metabolism, metabolomics, genomics, and tyrosinemia type I. As the mother of two children with tyrosinemia type 1, a rare genetic metabolic disorder, Dr. Barnby is familiar with the challenges faced by patients and families affected by rare diseases. She also serves as President of the Tyrosinemia Society, an international fundraising and advocacy organization dedicated to serving individuals and families affected by all forms of tyrosinemia.

Martina Bebin, MD

As a Professor of Neurology and Pediatrics at the University of Alabama at Birmingham (UAB), Dr. Bebin utilizes her extensive clinical expertise in providing care to patients with Tuberous Sclerosis Complex (TSC), a rare genetic disorder associated with benign tumor growth in the brain and other vital organs. She completed pediatric neurology fellowship training at the Mayo Clinic and since 2007 has served as Co-Director of the UAB Tuberous Sclerosis Clinic that provides multidisciplinary care for TSC patients throughout their lives; the Clinic has been recognized as a TSC Center of Excellence by the TS Alliance. For the past 20 years, Dr. Bebin has been involved in clinical drug development and has served as site Principal Investigator for numerous pediatric antiepileptic clinical trials as well as Director of the Pediatric Expanded Access Program for Cannabidiol in the State of Alabama. More recently, Dr. Bebin has participated in whole genome sequencing projects with the HudsonAlpha Institute for Biotechnology and the UAB Department of Genetics in children with unexplained developmental delays and various epilepsies.
David P. Bick, MD

David P. Bick, MD, serves as the Chief Medical Officer and a Faculty Investigator at the HudsonAlpha Institute for Biotechnology, the Medical Director of the Smith Family Clinic for Genomic Medicine, and as a Laboratory Director in the HudsonAlpha Clinical Services Laboratory. Dr. Bick received his medical degree from George Washington University School of Medicine in 1981 and completed a residency in pediatrics at Yale-New Haven Hospital in New Haven, Conn. At the Yale University School of Medicine, Dr. Bick completed a fellowship in human genetics and pediatrics in 1986 followed by a postdoctoral research fellowship in human genetics in 1987. Prior to joining HudsonAlpha, Dr. Bick served in numerous roles at the Medical College of Wisconsin, including Professor in the Department of Pediatrics and the Department of Obstetrics and Gynecology, Chief of the Division of Genetics in the Department of Pediatrics, and Director of the Clinical Sequencing Laboratory; he also previously served in several roles at the Children’s Hospital of Wisconsin including Director of the Advanced Genomics Laboratory and Medical Director of the Genetics Clinic. Dr. Bick’s laboratories at the Medical College of Wisconsin and Children’s Hospital of Wisconsin were the first in the world to offer whole genome sequencing as a clinical test. A leader in the field of genomic medicine, Dr. Bick has published numerous peer-reviewed articles, chapters, and reviews.

Senator Linda Coleman-Madison

Senator Coleman-Madison is currently serving a 4th term in the Alabama Senate representing District 20. Prior to her election to the Senate, she served one term in the House (Dist. 60) and also served for 12 years on the Birmingham City Council. A former special education teacher and graduate of Alabama A & M and the University of Alabama at Birmingham (UAB), she currently serves as the Americans with Disabilities Administrator for the city of Birmingham. She is also a licensed realtor.

Karen A. Fagan, MD

Dr. Fagan is Professor of Medicine and Pharmacology at the University of South Alabama (USA) and Chief of the Division of Pulmonary and Critical Care Medicine at USA University Hospital. She also serves as the Director of the USA Pulmonary Hypertension Center. Dr. Fagan has spent her career in research and patient care for individuals with pulmonary arterial hypertension, a life-shortening illness that causes significant disability in people of any age, gender, or racial background in Alabama. Despite treatment advances, none are curative but are used to slow the progression of disease.
Crayton A. Fargason, Jr., MD, MM

Dr. Fargason is the Chief Medical Officer of Children’s of Alabama as well as a tenured Professor of Pediatrics in the Department of Pediatrics at the University of Alabama at Birmingham (UAB). He currently serves as the Director of Neurodevelopmental Initiatives, a collaborative effort by Children’s of Alabama and the University of Alabama at Birmingham to improve the services to children with developmental disabilities. As the father of a child on the autism spectrum, Dr. Fargason has firsthand experience with the challenges faced by children and families with rare and complex diseases.

Mark N. Gillespie, PhD

Dr. Gillespie currently serves as a SAMSF-Locke Endowed Professor, Chair of Pharmacology, and Professor of Internal Medicine at the University of South Alabama (USA). After completing undergraduate and doctoral degrees at the University of Kentucky, he completed a postdoctoral fellowship at the University of Colorado’s Cardiovascular-Pulmonary Research Laboratory. Prior to joining the University of South Alabama in 1995, Dr. Gillespie served as Professor and Chairperson of Pharmacology and Experimental Therapeutics in the College of Pharmacy at the University of Kentucky. For the past several decades, he has served as an ad hoc or regular member or chairperson of multiple NIH study sections and held leadership positions in organizations dedicated to lung health and disease. His research program, supported continuously by the NIH and other agencies for more than 30 years, focuses on translational aspects of vascular biology and pathology using model systems ranging in complexity from cultured cells to human subjects. He is a founding member of a startup biotechnology company with a mission to develop pharmacologic agents to repair oxidative damage to the mitochondrial genome as a therapeutic strategy in oxygen radical-mediated disease.

Scott Griffin

Mr. Griffin is the President and Chief Executive Officer of the non-profit Hope for Gabe Foundation (H4G), a national organization committed to patient advocacy and research support for Duchenne muscular dystrophy (DMD), a rare genetic disorder that results in progressive muscle degeneration and weakness. A resident of Birmingham and father of four, Mr. Griffin founded the organization in 2010 when his then 3-year-old son, Gabe, received a diagnosis of DMD. Since that time, the H4G Foundation has accomplished a wide array of advocacy, awareness, and research efforts including the Gabe Griffin Right to Try Law passed in Alabama in 2015 to allow those with terminal diseases to have access to drugs not yet approved by the FDA. In 2016, Mr. Griffin served as one of a panel of speakers at the FDA Advisory Committee in Washington that led to approval of the first FDA-approved drug for DMD, and in 2017 he was appointed by Governor Kay Ivey to serve on the Health Coordinating Council. The H4G Foundation has helped to fund research at Nationwide Children’s Hospital for a drug call Follistatin and has also partnered with other foundations to fund drug clinical trials and explore gene therapies for DMD.
Scott Harris, MD, MPH, FACP, FIDSA

Dr. Harris was appointed Acting State Health Officer for the Alabama Department of Public Health (ADPH) in September 2017 and was formally designated as Alabama’s 21st State Health Officer in February 2018. A graduate of Harding University in Arkansas, Dr. Harris attended medical school at the University of Alabama at Birmingham (UAB) School of Medicine and served an internal medicine internship and residency at Carraway Methodist Medical Center in Birmingham before returning to UAB to complete a fellowship in adult infectious diseases. He earned his MPH from the UAB School of Public Health. In 1996, he began his practice in general infectious diseases and HIV medicine in Decatur, Alabama, and began serving as tuberculosis consultant with ADPH. In 2004, he helped to establish the non-profit Decatur-Morgan Community Free Clinic, serving 13 years as the medical director as well as a board member and board chair. As a volunteer physician, Dr. Harris has served on many international medical missions to Central America, South America, and Africa. In addition to his board certification in internal medicine and infectious disease, he has additional certificates of qualification in tropical medicine from the American Society of Tropical Medicine and Hygiene, and in travel medicine from the International Society of Travel Medicine.

Matthew Might, PhD

Dr. Might has served as the Director of the Hugh Kaul Precision Medicine Institute at the University of Alabama at Birmingham (UAB) since 2017. At UAB, Dr. Might also serves as the Hugh Kaul Endowed Chair of Personalized Medicine, a Professor of Internal Medicine, and a Professor of Computer Science. Prior to joining UAB, Dr. Might joined the faculty of the Department of Biomedical Informatics at the Harvard Medical School in 2015 where his research focused on rare disease discovery and diagnosis as well as the development of personalized therapeutics for rare disease. From 2016 to 2018, Dr. Might was a Strategist in the Executive Office of the President in The White House where he worked primarily on President Obama’s Precision Medicine Initiative with both the NIH and the Department of Veterans Affairs. Dr. Might’s journey from computer science to medicine was inspired by his son, Bertrand, who in 2012 became the first patient in the world to be diagnosed with NGLY1 deficiency. This diagnosis inspired Dr. Might to use social media to discover other patients and form a community; through the community’s efforts in science, two therapeutics for NGLY1 deficiency have been identified since its discovery, and more are under active development. Dr. Might is co-founder and Chief Scientific Officer of NGLY1.org, a non-profit dedicated to finding treatments for NGLY1 deficiency, and he was a co-founder and Scientific Advisor to Pairnomix, a start-up which identifies potential patient-specific therapies for rare disorders, particularly epilepsies. Dr. Might holds a BS, MS, and PhD in Computer Science from the Georgia Institute of Technology.
Samuel Perna, III, DO

Dr. Perna has more than 10 years of medical experience in palliative medicine. He currently serves as the Medical Director of the Pediatric Palliative Medicine Team at Children’s of Alabama and is also the lead physician for Aspire Health in Central and North Alabama. Dr. Perna is an Associate Professor of Medicine at the University of Alabama at Birmingham (UAB) with secondary academic appointments in nursing and pediatrics. He is board certified in both family medicine and hospice and palliative medicine and has distinguished himself as a Fellow of the American Academy of Hospice and Palliative Medicine.

Senator Dan Roberts

Senator Roberts represents district 15, which includes Jefferson, Shelby, and Talladega counties is Alabama. After graduating from Auburn University with a bachelor of science in building construction, Senator Roberts worked for Blount International in Alabama, South Korea, and Saudi Arabia. He earned a master of science in real estate development and urban affairs from Georgia State University, and while in graduate school he started a successful international trading company with an office in South Korea. After a season with Jim Wilson and Associates in Montgomery, he founded a real estate sales and development firm in Birmingham. Senator Roberts’ years of community leadership and service include the Entrepreneurs Round Table, Alabama Policy Institute, Racerunners, Young Business Leaders, and Leachman Ministries in Great Falls, Virginia (serving as Chairman of the Board).

Tammi L. Skelton, MSN, CRNP

Ms. Skelton is a nurse practitioner at the University of Alabama at Birmingham (UAB) with experience in both internal medicine and pediatric primary care. In 2015, Ms. Skelton joined the UAB Department of Genetics where she works with Dr. Bruce Korf in providing care to patients in both the Neurofibromatosis Clinic and the Undiagnosed Diseases Program. She received a B.S. in nursing from the University of Alabama at Birmingham (UAB) in 1992 and afterward joined Children’s of Alabama in the Pediatric Intensive Care Unit where she was a member of the Critical Care Transport Team, served as a Pediatric Advanced Life Support Instructor, and helped to initiate the Trauma Prevention Program that provides education for teens with traffic violations. She later became interested in pediatric clinical research and received certification as a Clinical Research Coordinator from the Association of Clinical Research Professionals (ACRP) in 2004. Ms. Skelton completed a master’s degree in nursing at UAB in 2012 and is certified through the American Association of Nurse Practitioners (AANP) as a family nurse practitioner.
Stephen Olufemi Sodeke, PhD

Dr. Sodeke is Resident Bioethicist and Professor of Allied Health and Bioethics at the Tuskegee University Center for Biomedical Research. He also currently chairs the Tuskegee University Institutional Review Board and serves as a bioethicist for the Alabama Genomic Health Initiative (AGHI) and for the Southern All of Us Research Network; he additionally serves as Resource Expert for NGO Bioethics Beyond Borders. At various periods during past 15 years, Dr. Sodeke has served as Associate Director and Interim Director of the Tuskegee University National Center for Bioethics in Research and Health Care. He holds a PhD in curriculum and instruction in allied health education, supervision, and administration from the University of Florida, a master’s degrees in clinical laboratory science education and guidance from Texas Christian University and in bioethics and health policy from Loyola University of Chicago. Dr. Sodeke holds certifications in bioethics from the Kennedy Institute of Ethics at Georgetown University as well as in health care ethics from the University of Washington in Seattle. He participates in ethics consultations and speaking engagements that promote the goals and purposes of bioethics in medical, biomedical, and behavioral research locally, nationally, and internationally.

Katelyn Staley, MSN, CRNP

Ms. Staley serves as the Discharge Coordinator for Cardiovascular Services at Children’s of Alabama. After obtaining a BS in Nursing at the University of Alabama at Birmingham (UAB), she began her nursing career at UAB Hospital in the Regional Newborn Intensive Care Unit (RNICU). Ms. Staley later obtained a master’s degree in nursing at UAB, specializing in pediatric primary and acute care. She is committed to serving patients in the rare disease community along with her husband, who serves on the board of the Hope for Gabe Foundation to end Duchenne muscular dystrophy.
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WHAT IS A RARE DISEASE?

In the United States, the National Institutes of Health (NIH) defines a rare disease as a condition that affects fewer than 200,000 people. Rare diseases may include many single gene disorders, childhood and adult diseases, and some childhood cancers and infectious diseases. They also include the diseases on state newborn screening tests. Rare diseases can cause long-term illness, physical and intellectual disabilities, and reduced lifespan. Some individuals and families affected by rare diseases face challenges related to the financial cost of medical management and treatment. Also, many rare disease patients and families, especially those who are newly diagnosed, may feel overwhelmed when searching for information and resources related to their disease.

WHAT IS THE PREVALENCE OF RARE DISEASES IN THE U.S. AND IN ALABAMA?

Although individual diseases may be rare, it is estimated that there are more than 7,000 rare diseases that affect 25 to 30 million Americans. One out every 10 Americans lives with a rare disease. In Alabama, about 400,000 residents are affected by rare diseases. This number translates to about 8.2% of the state’s population that is living with a rare disease.

These numbers are only estimates, however. This is because only a few types of rare diseases are tracked in the U.S. Because most rare diseases are not tracked, it can be difficult to determine the exact number of rare diseases and how many people are affected.

WHAT CAUSES RARE DISEASES?

Rare diseases have many different causes, although a high percentage are thought to be genetic. A genetic disease is directly caused by a change to a gene or chromosome. Genetic changes that cause disease are sometimes called mutations or genetic variants. These genetic changes are often passed from one generation to the next. This is known as an inherited genetic variant. In other cases, the genetic change occurs randomly in a person who is the first in a family to be diagnosed; this is called a spontaneous mutation. Many rare diseases do not have a known genetic cause. These include infections, some rare cancers, and some autoimmune diseases. Researchers continue to learn more about rare diseases each year, although the cause of many rare diseases is unknown.
RARE DISEASE FAST FACTS

- In the U.S., a rare disease is defined as a condition that affects fewer than 200,000 people.
- There are estimated to be 7,000 rare diseases.
- 30 million Americans are estimated to be living with a rare disease. This figure is equivalent to one in 10 people in the U.S. (Source: NIH).
- In Alabama, 400,000 people are living with a rare disease. This figure is equivalent to 8.2% of the state’s population (Source: Extrapolated from NIH data).
- More than 90% of rare diseases do not yet have a treatment approved by the FDA (Source: NORD).
- One in 33 babies in the U.S. is born with birth defects, which include rare diseases (Source: CDC).
- The Alabama Newborn Screening program screens about 60,000 babies annually. The program identifies on average 150 to 200 babies with a metabolic, endocrine, hematological or other congenital disorder.
ACHIEVING A DIAGNOSIS

For many people living with a rare disease, getting a diagnosis can be a long, stressful experience that can take years. Navigating the healthcare system for months or years in search of a diagnosis can leave a person feeling frustrated and exhausted from a seemingly never-ending quest for answers. The term “diagnostic odyssey” is often used to describe the unpredictable journey many people affected by rare diseases face. An accurate and timely diagnosis is an important first step in receiving the appropriate treatment to improve symptoms and prevent progression of rare diseases.

There are several reasons why a rare disease can be difficult to diagnose. Many primary care physicians, and even some specialists, are often unfamiliar with the variety of symptoms that can be associated with rare diseases. Also, some rare diseases have nonspecific symptoms, such as weakness, pain, or dizziness. These symptoms can be misdiagnosed as more common conditions. Also, because more than 80% of rare diseases have a genetic cause, genetic testing is often the only way a definite diagnosis can be made. However, if a physician does not include genetic testing as part of the diagnostic process, the diagnosis may be missed.

THE VALUE OF A DIAGNOSIS

Achieving an accurate diagnosis is the first step in finding the right treatment and improving care for people with rare diseases and their families. However, delays in diagnosis can lead to inappropriate management of a rare disease and disease progression. Also, a misdiagnosis can lead to interventions that are not appropriate for the underlying disease or condition.

A correct diagnosis ensures that people with rare diseases receive the clinical care and counseling appropriate for their disease. Also, a diagnosis removes a significant psychological weight from those who have been struggling with a long diagnostic odyssey. It also gives people and families affected by a rare disease the chance to connect with a rare disease community. Improving the diagnostic journey for people with rare diseases is an important area of focus for rare disease patient advocacy organizations.
FINDING A CLINIC OR DIAGNOSTIC PROGRAM

It is important to know that help and resources are available for people who are currently searching for a diagnosis. Some simply have not been referred to the correct specialist, while others have a disease that is either unknown to the medical community or does not yet have a known test. Finding a clinic or diagnostic program that specializes in undiagnosed patients is an important way to connect with specialists who have expertise in diagnosing rare disorders. The following section features a list of diagnostic programs, including those within Alabama, focused on uncovering a diagnosis for people with rare diseases.

The Undiagnosed Diseases Network (UDN)
The Undiagnosed Diseases Network (UDN) is the leading clinical research study in the U.S. for undiagnosed patients (https://commonfund.nih.gov/diseases). The study, funded by the National Institutes of Health (NIH), bridges the gap between clinical care and research by assembling experts from around the country to collaborate in seeking diagnoses for the most complex undiagnosed diseases.

The UDN comprises 12 clinical sites across the United States including a coordinating center, a sequencing core, a metabolomics core, two model organism screening centers, and a central biorepository. Please visit https://undiagnosed.hms.harvard.edu/ for more information on the UDN and how to apply.

The Undiagnosed Diseases Program (UDP) at the University of Alabama at Birmingham
Undiagnosed patients in Alabama also have access to the Undiagnosed Diseases Program (UDP) at the University of Alabama at Birmingham (UAB). This leading-edge program addresses the needs of patients with severe chronic medical conditions for whom a diagnosis has not been made despite extensive efforts by the referring physician. The UDP strives to meet the needs of patients of all ages. The program serves those with rare diseases, those with conditions not previously known to exist, and those with atypical presentations of common diseases.

The UDP is led by a multidisciplinary team of experienced UAB clinicians that includes:

- Bruce R. Korf, MD, PhD
- Martin Rodriguez, MD
- Anna Hurst, MD

The team also includes certified genetic counselor Kirstin Smith, MS, CGC, and certified registered nurse practitioners Tammi Skelton, MSN, CRNP, NP-C, and Kaitlin Callaway, MSN, CRNP, NP-C. The program benefits from the expertise of clinicians and investigators from UAB Medicine, Children’s of Alabama, and HudsonAlpha Institute for Biotechnology. These experienced professionals work together in using medical expertise and testing, as well as genetic and genomic technologies, to uncover a diagnosis and determine an effective treatment. For an evaluation, patients need a physician referral and must also meet essential criteria. For more information about the UDP, please visit the program website: www.uab.edu/medicine/genetics/undiagnosed-diseases.

Smith Family Clinic for Genomic Medicine
The Smith Family Clinic for Genomic Medicine is the first clinic in the world designed to provide diagnoses to patients with undiagnosed diseases by using whole genome sequencing data when other genetic technologies are unable to reach a diagnosis. Led by Medical Director David Bick, MD, the Clinic is unique in sequencing the whole genome, rather than part of the genome, in the diagnostic process. The Clinic is located on the campus of the HudsonAlpha Institute for Biotechnology in Huntsville. For more information, please visit the Clinic website at: www.smithfamilyclinic.org
The Undiagnosed Disease Registry

The National Organization for Rare Disorders (NORD) is the leading patient advocacy organization dedicated to improving the lives of individuals and families living with rare diseases. NORD has launched the Undiagnosed Rare Disease Registry to support research on rare diseases and how they progress over time. The online registry securely collects and stores data for medical research. The goal of the study is to limit the diagnostic odyssey for people with undiagnosed rare diseases by building a rare disease community. Other goals include collecting relevant demographic and symptom-related data and informing researchers, clinicians, and regulatory agencies. To join the Undiagnosed Rare Disease Registry, please visit: www.rarediseases.org/iamrare-registry-program/

For a full list of resources for undiagnosed patients, please refer to Appendix A.

THE ROLE OF GENETIC TESTING AND GENOME SEQUENCING

Because a high percentage of rare diseases have a genetic component, most undiagnosed patients benefit from evaluation at a clinical genetics center where they can receive genetic testing (for a search tool to locate clinical genetics centers in the U.S., please visit the American College of Medical Genetics and Genomics website at: https://clinics.acmg.net.) Genetic testing involves examining a person’s DNA, the structure in cells that carries the genetic code. This testing can reveal variants, or changes, in genes that can help to diagnose some rare diseases.

Genetic tests can be performed on samples of blood, skin, saliva, or other tissue. These tests can help to confirm a rare disease diagnosis and identify treatment options, including clinical trials. Most genetic tests focus on examining one gene at a time, based on a clinician’s judgement of the most likely cause for a person’s condition. Although genetic testing often provides a diagnosis, it may be difficult to predict which of the approximately 22,000 genes in the human genome is the cause of a person’s condition.

Genome Sequencing

It is now possible to examine most or all of these genes in a single test called genome sequencing. Genome sequencing comes in two forms: whole exome sequencing (WES) and whole genome sequencing (WGS). WES focuses just on that part of the genome that encodes for proteins. This is where most genetic variants that cause disease are located. WGS looks at the entire genome, including regions that do not encode protein. Currently, most clinical laboratories that do genome sequencing offer WES, which is much less expensive and easier to interpret than WGS. WGS is offered on a clinical or research basis in some settings; it may be able to detect a larger number of genetic variants associated with disease, but it is also considerably more expensive to perform.

Genome sequencing identifies a genetic explanation for rare disease in 25-50% of cases. The diagnostic rate depends in part on how much information there is to support a genetic cause for a person’s rare disease in the first place. The genetic variant that is identified may be a unique one that has never been seen before. Sometimes this results in discoveries of new genetic conditions, especially if more than one person is found with a change in the same gene. Laboratories can share information on a confidential basis to try to find similar cases that help to validate these new findings. However, sometimes genome sequencing results in finding a genetic variant that cannot be definitively concluded to be the cause of a person’s condition, though it may have features that suggest it is possibly related. Geneticists call these “variants of unknown significance,” or VUSs.
It’s important to note that a VUS is not the definitive cause of a person’s condition. Some VUSs are eventually determined to be the cause, while others are eventually found not to cause disease. Therefore, clinical decisions, such as starting treatment, should not be based on the finding of a VUS.

Even with advances in genome sequencing technology, we are still not able to detect all possible genetic changes that cause disease. Because of this, a negative result from genome sequencing does not mean that there is no genetic cause of a person’s condition. For this reason, one should not assume that genetic transmission of the condition will not occur if the sequencing was not informative. In some cases, re-analysis of a genome sequence a year or two after the initial test reveals a variant that was not initially appreciated. This is usually because more information has been gained to help interpret variants. Genome sequencing is gradually being improved to detect specific kinds of genetic variants. If genome sequencing does not result in a diagnosis, it can be useful to maintain contact with a genetics physician to periodically explore whether there are new approaches that may shed light on the underlying diagnosis.

**Alabama Genomic Health Initiative**

Undiagnosed patients in Alabama may benefit from participation in the Alabama Genomic Health Initiative (AGHI). This program is one of the first statewide efforts in the nation to use the power of genomic analysis (genome sequencing) in helping to identify those who are at risk for disease and genomic abnormalities. AGHI includes members from UAB, HudsonAlpha Institute for Biotechnology, and Tuskegee University, all of which are nationally recognized institutions and leaders in bioethics and genomic medicine.

The program includes a component of testing adults for several rare genetic conditions. These mostly include conditions that increase risk of cancer or some kinds of heart disease. The program also offers WGS for individuals with rare undiagnosed diseases. For more information about the AGHI, please visit: [www.uabmedicine.org/aghi](http://www.uabmedicine.org/aghi).

**GENETICS PROFESSIONALS**

Most clinical genetics centers and diagnostic programs offer teams of experienced physician medical geneticists and genetic counselors who can help families learn more about genetic testing and provide support to assist in adjusting to a new diagnosis. This genetics professional team can help families determine whether genetic testing is appropriate and provide information about interpretation of a genetic test. Services can include:

- Providing information about how a genetic disease can be passed through families and determining your risk of having children with the disease
- Explaining the testing options available and discussing the benefits and limits of genetic testing
- Providing information about specific tests that are available for prenatal and preimplantation genetic diagnosis
- Interpreting and explaining the results of genetic testing
- Implementing a management plan based on a genetic diagnosis
CONNECTING WITH OTHER PATIENTS AND FAMILIES

If you or a loved one has been diagnosed with a rare disease, you may feel alone and overwhelmed at first. It’s important to know that these feelings are normal. It may be helpful to give yourself some time to adjust to the news and reflect on your diagnosis before beginning the search for information about your rare disease. Also, remember that you are not alone. Resources are available to help you and your family navigate this new and unfamiliar journey.

When you are ready, you can find support and understanding by connecting with a rare disease community that is familiar with the unique challenges related to your rare disease. Patient advocacy organizations can help you make these connections and provide other forms of support. The National Organization for Rare Disorders (NORD) is a great resource for finding advocacy organizations for rare diseases. It lists more than 1,000 organizations on its website: https://rarediseases.org/for-patients-and-families/connect-others/find-patient-organization/.
USING SOCIAL MEDIA

The use of social media can be another effective way to connect with or create a rare disease community. Facebook groups, blog posts, some websites, and a variety of other social media platforms can be useful in finding other individuals and families who share similar challenges and concerns.

The following are just a few social media sites that can help rare disease patients and families connect with an online community:

- **GenomeConnect** ([www.genomeconnect.org](http://www.genomeconnect.org)) This online tool allows people to share their genetic test results and health information with researchers and healthcare providers as well as to connect with other individuals who have a similar diagnosis or related symptoms.

- **GlobalGenes** ([www.globalgenes.org](http://www.globalgenes.org)) This site provides patient education and advocacy programs and helps people with rare diseases find a sense of community and support through connecting with one another.

- **Inspire** ([www.inspire.com](http://www.inspire.com)) Users can search the site by condition to find information and online communities.

- **MyGene2** ([https://mygene2.org/MyGene2/](https://mygene2.org/MyGene2/)) This consortium connects individuals and families with rare conditions to clinicians and researchers who are interested in sharing health and genetic information with each other.

- **RareConnect** ([www.rareconnect.org/en/communities](http://www.rareconnect.org/en/communities)) This site features online communities for patients and families with rare medical conditions. The project is a collaboration between the European Rare Disease Organisation (EURORDIS) and the National Organization for Rare Disorders (NORD).

- **RareShare** ([www.rareshare.org](http://www.rareshare.org)). This online social hub is dedicated to individuals, families, and healthcare professionals affected by rare medical disorders.

CONNECTING WITH A NATIONAL OR STATE-BASED ORGANIZATION FOR YOUR DISEASE

Once you’ve connected with a patient advocacy organization that can provide support and encouragement after your diagnosis, consider connecting with a national or state-based organization for your disease. This is an important and helpful way to learn more about your disease from a reliable, trusted source. A general internet search can usually help to identify a national organization associated with a specific disease. Also, there are several websites that can be helpful in identifying national, disease-specific advocacy organizations.

For example, the NORD website provides a library of information for more than 1,200 rare diseases, including national and international advocacy and research organizations for each disease ([https://rarediseases.org/for-patients-and-families/information-resources/rare-disease-information/](https://rarediseases.org/for-patients-and-families/information-resources/rare-disease-information/)). Also, the National Institutes of Health (NIH) Genetic and Rare Disease Information Center (GARD) website contains an easy-to-use search tool that lists national and international advocacy organizations for specific diseases ([https://rarediseases.info.nih.gov](https://rarediseases.info.nih.gov)).
In Alabama, the grassroots organization, Alabama Rare (www.alabamarare.org), focuses on uniting Alabamians around the rare disease population within our state. It provides support for individuals and families as well as educates the broader community about rare diseases. Also, the organization collaborates with stakeholders in Alabama to create awareness of the rare disease population’s needs and advocate for necessary change to improve healthcare delivery. Alabama Rare is involved in key policy initiatives including:

- Medicaid expansion in Alabama
- Expanding the state Newborn Screening Panel (NSP) of genetic disorders
- Comprehensive insurance coverage of medical nutrition
- Coverage of whole genome and whole exome sequencing genetic testing

*For a full list of rare disease and disease-specific advocacy organizations, please refer to Appendix B.*

**NAVIGATING AND EVALUATING OTHER ONLINE RESOURCES**

Learning about your disease is an important way to empower yourself. With information, you can make informed decisions about your healthcare choices regarding treatment, providers, and participation in research. A vast amount of health information is available on the internet. However, some of this information has not been vetted by trustworthy sources and therefore may not always be considered accurate and reliable. Also, information on the internet often highlights the most extreme cases and may give a skewed view of the severity of the condition. Accurate and reliable sources of health information may include national disease organizations, health organizations, healthcare practitioners, universities, or government agencies.

Resources are available to help you determine the most reliable sources of online healthcare information. MEDLINEplus, a service of the National Library of Medicine (NLM) and the National Institutes of Health (NIH), has posted a series of documents and links called *Evaluating Health Information*. This resource provides helpful guidance on how to find reliable information on the Web. It also includes tips for evaluating the quality and accuracy of internet sources and information. To access this resource, please visit: https://medlineplus.gov/evaluatinghealthinformation.html

*Please refer to Appendix B for a full list of websites with vetted, reliable health information.*
FINDING THE MOST EXPERIENCED PROVIDERS FOR YOUR DISEASE

Once you have achieved a diagnosis, it is important to find an experienced healthcare provider with training and expertise in the management and treatment of your disease. These providers have the most up-to-date knowledge and information about the most effective treatments as well as current research and clinical trials. It sometimes can be challenging to find specialists for rare diseases, especially for people who do not live near major academic medical centers. However, resources are available to help in your search for providers.

Following are resources, including those recommended by the National Institutes of Health (NIH) Genetic and Rare Disease Information Center (GARD) website, for locating a healthcare professional for your disease:

- **Disease Advocacy Organizations.** Many of these organization have physician locator services or patient networks that may help you find a healthcare professional who is familiar with your condition. Some may also have a list of doctors recommended by their members. Also, medical advisory boards for these organizations are comprised of experts in the field. Sometimes, disease advocacy organizations establish Centers of Excellence for their condition; contact a support organization to determine if they are familiar with a Center of Excellence.
  
  - To find disease advocacy organizations, the NORD website provides a library of information for more than 1,200 rare diseases, including national and international advocacy and research organizations for each disease (www.rarediseases.org/for-patients-and-families/information-resources/rare-disease-information/).
  - Also, the National Institutes of Health (NIH) Genetic and Rare Disease Information Center (GARD) website contains an easy-to-use search tool that lists national and international advocacy organizations for specific diseases (www.rarediseases.info.nih.gov/).
  - The Genetic Alliance website also contains a list of advocacy and patient organizations for specific diseases (www.geneticalliance.org).

- **List of Specialty Clinics in Alabama.** A list of specialty clinics in Alabama has been compiled by UAB medical researcher Matthew Alexander, PhD. *Please refer to Appendix C for the specialty clinics list, including clinic websites and phone numbers.*

- **Major Teaching Hospitals and Academic Health Centers.** These institutions are often located in larger cities and metropolitan areas. They are more likely to have medical staff with expertise in the treatment and management of rare diseases and physicians involved in clinical trials.
Clinical Genetics Centers. Genetics professionals can be a valuable source of information for people with rare diseases and their families. The following organizations and websites are good sources of information:

- **The American College of Medical Genetics and Genomics** – Features a Genetics Clinics Database to assist with locating a U.S. genetics center ([https://acmg.net](https://acmg.net)).
- **The American Society of Human Genetics (ASHG)** – Professional organization of research and clinical geneticists that maintains a database of its members, some of whom live outside the U.S. Some geneticists listed may be researchers only and may not offer medical care ([www.ashg.org](http://www.ashg.org)).
- **The National Society of Genetic Counselors** – Features a database of genetics counseling services that are searchable by location, name, institution, type of practice, or specialty ([www.nsgc.org](http://www.nsgc.org)).
- **MEDLINEPlus** – Website maintained by the U.S. National Library of Medicine that provides a link to various websites and directories to assist in finding health professionals, services, and facilities ([www.medlineplus.gov/directories.html](http://www.medlineplus.gov/directories.html)).

Medical Researchers. These professionals can be a helpful source for identifying an expert for a specific disease. Frequently, researchers conducting clinical trials are medical doctors who may also see patients who are not enrolled in a study. These physicians may also provide a referral to a colleague who sees patients for a specific disease. The following organizations and websites are useful in locating medical researchers who may be helpful:

- **The Rare Diseases Clinical Research Network (RDCRN)** comprises 22 distinctive consortia working to improve the availability of rare disease information, treatment, clinical studies, and general awareness for patients and the medical community. The RDCRN is focused on providing up-to-date information for patients and connecting them with advocacy groups, expert doctors, and clinical research opportunities ([www.rarediseasesnetwork.org](http://www.rarediseasesnetwork.org)).
- **The Centers for Mendelian Genomics** is a research program that focuses on discovering causes of rare genetic disorders ([www.mendelian.org](http://www.mendelian.org)).
- **ClinicalTrials.gov** is a website that was developed by the U.S. National Institutes of Health (NIH) through the National Library of Medicine. The purpose of the site is to provide clinicians, patients, family members, and the public with current information on clinical research studies in the U.S. This resource may be helpful in locating researchers who are studying a specific condition. A basic search can be conducted using the condition name as the search term; click on a study to review the eligibility criteria and obtain the study’s contact information.
- **The Patient Recruitment and Public Liaison (PRPL) Office** at the NIH provides information about opportunities to participate in research conducted at the NIH Clinical Center Hospital. You may call the PRPL office toll-free at 1-800-411-1222 or send an email to prpl@mail.cc.nih.gov to ask about clinical trials being conducted for a specific disease and contact information for research involved in these trials.

For a full list of resources to help in locating healthcare providers and clinics, please refer to Appendix D.
WHAT TO CONSIDER WHEN LOOKING FOR AN EXPERT

One of the challenges in dealing with a rare disease is that there may be very few clinicians who have experience with providing care for affected individuals. Often, patient advocacy groups for specific conditions list centers of excellence for care of the disorder. However, it is possible that there will be only a few such centers for some conditions. It may not be practical to be seen or followed at a site that is distant from home and not covered by a person’s insurance. When considering a specialist, you should feel you can build a rapport with the clinician. Here are a few other points to consider in selecting a specialist:

- Have they cared for patients with this condition before – and, if so, how many?
- Do they participate in clinical trials or clinical research on the disorder?
- Have they published studies on the disorder?
- Are they affiliated with a relevant patient support group?

A medical home, where patients have a dedicated provider and healthcare team, is an important way to ensure that care is coordinated among the various specialists involved in providing care.

THE VALUE OF A MEDICAL HOME

Most people with rare diseases have complex healthcare needs that often require the coordination of multiple specialists and diagnostic tests. A medical home, where patients have a dedicated provider and healthcare team, is an important way to ensure that care is coordinated among the various specialists involved in providing care.

The clinic in which your disease specialist and healthcare team provide care may serve as your medical home. This works best if the clinic is located close to where you live. If your disease specialists are located out of town or in another state, the best option for your medical home is probably the primary care physician and clinic in the city where you live. In either case, a medical home is a place where a disease specialist or primary care physician can follow patients consistently over time and coordinate care with specialists as needed. This is the best way to ensure that patients receive consistent care from a dedicated healthcare team who is familiar with the rare disease diagnosis and health challenges.

The benefits of having a medical home include:

- Patients have a relationship with a personal physician
- A practice-based care team manages your ongoing healthcare needs
- Care is coordinated across care settings and specialists
- You have enhanced access to and communication with your healthcare providers
THE IMPORTANCE OF MEDICAL RECORD KEEPING

Many people with a rare disease diagnosis visit multiple doctors as part of the treatment and management of their disease. As a result, keeping track of personal medical information can be a challenge. However, accurate record-keeping is important for ensuring successful management of your disease. It is also an important way to enhance communication of timely information with your healthcare providers. The best way to accomplish accurate record keeping is to maintain a personal health record (PHR). This document allows you to gather and manage all your health information in one easily accessible location.

ELECTRONIC PERSONAL HEALTH RECORD

Most health providers these days use electronic medical record systems to record information about medical tests and visits. Unfortunately, these rarely communicate between one health system and another. If a person is seen by health providers at different institutions or private practices, the medical records at one are usually not visible by the other. Many electronic health record systems include an online portal. This allows a patient to see some of their medical information and test results; they can print or save them as an electronic copy to share with other providers. Patients can also provide a signed release of medical records form that authorizes a provider at one practice to send or fax medical records to the other. That said, it is often a good idea for a person with a rare disorder to keep a personal file of relevant medical information. These can be kept in electronic form or as paper records. Imaging studies can be copied onto CDs and shared with providers as needed.

You may wish to include the following information in your personal health record:

- Information about your diagnosis, including the date you were diagnosed, copies of imaging or other diagnostic tests, and pathology/laboratory reports
- Complete treatment information, including start and end dates as well as results and any complications/side effects
- Details of past physical exams, including relevant clinic notes, laboratory reports, and diagnostic testing
- Current immunization records
- Dates and details of other major illnesses, chronic health conditions, and hospitalizations
- Contact information for the doctors and clinics involved in your diagnosis and treatment
- Information about your family medical history
- List of current and previous treatments, procedures, and medications
STRATEGIES FOR KEEPING UP-TO-DATE MEDICAL RECORDS

Following are a few strategies that can be helpful in collecting the latest copies of your records on an ongoing basis:

- Ask your doctor or nurse for a copy of the results or report any time you have a diagnostic test or procedure
- At each appointment, ask your doctor or nurse for a copy of any clinic notes added to your file or electronic medical record
- Use a secure online patient portal to access your current medical records
- If you spent time in the hospital, request a copy of your records when discharged
- Keep copies of medical bills and insurance claims as they occur
- Talk to your doctor for guidance about what records to include
HOW TO TALK TO OTHERS ABOUT YOUR DISEASE

Once you’ve had some time to adjust to your diagnosis, you’ll probably start to consider when and how to share information about your disease with others. These individuals might include close friends, family members, employers, or even a child. Deciding with whom to share the news – and how much to tell – can be one of the most difficult aspects of dealing with a new diagnosis. The information you share about your disease is a highly personal decision. There is no right or wrong approach to how much, or with whom, you choose to share. Here are some suggestions that may help make the process a bit easier.

- **Whom to Tell is Up to You.** Some people with rare diseases have obvious symptoms, making it more likely they will be asked about their condition. For others, symptoms are not as visible, which makes it easier to be more selective about whom to tell. A general guideline is that many people reveal their diagnosis to others only if it seems important to their relationship.

  - In some cases, it may be necessary to talk about how your disease impacts you with people other than close friends or family members. These individuals might include employers, teachers, or professors. In K-12 educational settings, a 504 plan or an Individual Education Plan (IEP) provides learning accommodations to people with special needs, including those with chronic disease or serious illness, under the Individuals with Disabilities Education Act ([https://sites.ed.gov/idea](https://sites.ed.gov/idea)).

    The Americans With Disabilities Act provides protections and accommodations to those who are eligible within college and work settings ([https://www.dol.gov/general/topic/disability/ada](https://www.dol.gov/general/topic/disability/ada)). You may also consider seeking advice from a disability attorney about protecting your rights and ensuring equal access to education, employment, housing, and transportation.

- **You Are in Control of the Information You Share.** When you decide to share your diagnosis with others, or the diagnosis of a family member who has given specific consent, you may only want to share in a limited way. For example, you might only discuss your most common symptom and how it impacts your daily activities. Other times, you may choose to talk to someone with whom you can share more details. These details might include the potential challenges that go along with the diagnosis. It is important to remember that you know your story best, and you are in control of what you want to share.

- **Get Advice from Professionals.** You may find it helpful to talk to a licensed professional counselor, disability attorney, genetic counselor, or your doctor to get advice and guidance about what information to share about your diagnosis and with whom. A licensed professional counselor or a genetic counselor can also provide valuable insight and suggestions about how and when to tell children about their own rare disease diagnosis or that of a parent, sibling, or other family member.
MANAGING THE FINANCIAL COSTS OF A RARE DISEASE

The overall financial cost of managing a rare disease diagnosis can be hard for patients and families, even with the best health insurance coverage. For example, managing a rare disease diagnosis often includes buying prescription drugs, which may include expensive orphan drugs. Rare disease patients and families might also have to pay for specialized medical equipment, travel to multiple specialists, and full-time or part-time caregiving. While the financial challenges for people living with a rare disease can be difficult, state and federal resources are available to help manage the costs for those who qualify.

ALABAMA RESOURCES

- **Alabama Department of Rehabilitation Services (ADRS)** is the primary state agency that serves Alabamians with disabilities from birth throughout their lives through four main programs:
  
  - **Early Intervention (EI)** – Serves children from birth to age three by coordinating services statewide for infants and toddlers with developmental delays.
  
  - **Children’s Rehabilitation Service (CRS)** – Serves children and teens from birth to 21 with special healthcare needs and their families.
  
  - **Independent Living** – Assists individuals with the most significant disabilities by providing a wide range of education, assistive technology, training, and home-based services to allow them to gain more independence.
  
  - **Vocational Rehabilitation (VR) Services** – ADRS’ largest program; serves teens and adults to provide specialized employment and education-related services and training. Programs within VR include:
    
    - **VRS-Blind/Deaf Program** – Provides Blind services, Deaf services, and OASIS (Older Alabamians System of Information and Services) programs.
    
    - **Business Enterprise Program** – Enables qualified blind individuals to achieve independence through self-employment; partners with businesses to connect them with reliable and cost-efficient vendors.
    
    - **Business Relations Program** – Works with thousands of Alabama businesses on disability-related issues. This include providing recruitment assistance, disability management, training, and employee retention services.
    
    - **Lakeshore Rehabilitation Clinic** – Accredited program in Birmingham that helps people with significant disabilities achieve their educational goals through vocational training, college, or direct employment.

*To learn more about the programs ARDS provides, please visit their website: [www.rehab.alabama.gov](http://www.rehab.alabama.gov)*
Alabama Home and Community-Based Services (HCBS) Medicaid Waivers provide services to support people with intellectual or significant physical and medical disabilities. These services provide support that helps people live at home or in other community-based settings like group homes. The services provided can include the following:

- Personal care
- Skilled nursing
- Behavioral supports
- Respite services
- Assistive technology
- Homemaker services

Alabama’s seven waiver programs are administered by the Alabama Medicaid Agency and its state partners:

- The Alabama Department of Mental Health (ADMH)
- The Alabama Department of Senior Services (ADSS)
- The Alabama Department of Rehabilitation Services (ADRS).

For more information about Alabama’s HCBS Medicaid Waivers, please visit the Alabama Medicaid website: www.medicaid.alabama.gov/content/6.0_LTC_waivers/6.1_HCBS_waivers.aspx

Also, the Alabama Disabilities Advocacy Program has posted a resource entitled Alabama’s Home & Community-Based Medicaid Waivers that you can find by visiting: www.adap.ua.edu/uploads/5/7/8/9/57892141/medicaid_community_waiver_aug_2015.pdf

The Alabama Health Insurance Premium Payment (HIPP) Program is designed to save money for Medicaid families with high healthcare costs. The program pays most out-of-pocket medical expenses for qualifying Medicaid recipients and reimburses policyholders for the cost of health insurance provided by an employer or COBRA. Some recipients may also qualify to receive reimbursement for the cost of a family health insurance policy premium. To apply or learn more about the Alabama Medicaid HIPP Program, visit: www.myalhipp.com.

ALL Kids is an Alabama Children’s Health Insurance Program (CHIP) administered by the Alabama Department of Public Health. This low-cost, comprehensive healthcare coverage program for children under age 19 includes benefits such as regular checkups and immunizations, sick child doctor visits, prescriptions, vision and dental care, hospitalization, mental health and substance abuse services, and other services. ALL Kids uses Blue Cross Blue Shield of Alabama to provide medical and mental health services through their preferred provider network (PPO). To learn more about ALL Kids or to apply for the program, visit: www.alabamapublichealth.gov/allkids/.

UAB Hospital’s Charity Care Program is designed for uninsured or underinsured patients of the Kirklin Clinic of UAB Hospital or affiliated clinics. Applicants may qualify for financial assistance, which is based on household size and household income. To receive an application, ask your UAB healthcare provider.
FEDERAL RESOURCES

Consolidated Omnibus Budget Reconciliation Act (COBRA) is government legislation that gives workers and their families who lose their health benefits through job loss and other circumstances the right to choose to continue group health benefits provided by their employer for limited periods of time. For more information, please visit: www.dol.gov/general/topic/health-plans/cobra

The Medicare Insurance Program (www.medicare.gov) is designed for people who are 65 years or older, certain younger people with disabilities and people with end-stage renal disease (ESRD).

The Medicaid Insurance Program (www.medicaid.gov) is a federally sponsored, state-administered health insurance program for families and individuals with low incomes.

The Affordable Care Act/Obamacare (www.healthcare.gov) provides access to the federal government’s insurance marketplace for health insurance policies. These policies are provided by private insurance companies for individuals, families, and small businesses. Health Insurance Marketplace call center: 1-800-318-2596

USA.gov – Provides links to federal resources that can help with the cost of medical care and prescription drugs (www.usa.gov/help-with-bills#item-36707). You can also contact them directly at: 844-872-4681.

NON-PROFIT PATIENT ASSISTANCE PROGRAMS

NORD RareCare® offers assistance programs that provide medication, financial assistance with insurance premiums and co-pays, diagnostic testing assistance, and travel assistance for clinical trials or consultation with disease specialists (www.rarediseases.org/for-patients-and-families/help-access-medications/patient-assistance-programs-2/).

Patient Advocate Foundation (PAF) provides professional case management services to people with chronic, life-threatening, and debilitating illnesses (www.patientadvocate.org).

Patient Services, Inc. provides financial support and guidance for qualified patients with specific, rare chronic diseases (www.patientservicesinc.org).

ADVICE FOR DEALING WITH INSURANCE PROVIDERS

For people with rare diseases, dealing with insurance providers can be a time-consuming, complex, and often stressful experience. Determining what treatments and procedures are covered and advocating for your rights as an insured patient are important keys to successfully navigating the health insurance landscape. This section provides a few tips to help make the process a bit easier.

- Keep thorough, up-to-date records. Store all policy statements, claims statements, and notes of conversations with insurance representatives in a binder or electronic health record. Keep these records well-organized and accessible so that you can refer to them each time you speak with your insurance company representative.

Determining what treatments and procedures are covered and advocating for your rights as an insured patient are important keys to successfully navigating the health insurance landscape.
Request an insurance case manager. Because rare disease patients often have complex healthcare needs, a case manager can serve as the dedicated liaison to help patients manage their benefits, arrange care, and find resources.

- Case managers are usually registered nurses or social workers with special training in helping patients navigate the health insurance landscape to better understand the covered treatments and resources available to them.
- As a patient, you benefit from having a single point of contact dedicated to managing your account.
- If your health insurance company has not assigned a case manager to your account, ask for one.

Understand prior authorizations. A prior authorization is also called a precertification or prior approval. This is a process many health insurance companies require. It involves having your physician or healthcare provider secure expressed approval for a specific drug, treatment, or procedure. Without this approval, the insurer will not cover the cost.

- Prior authorization can increase wait times for patients to receive drugs or treatments. Sometimes the process results in denial of coverage.
- A benefit to the prior authorization process is that it helps to reduce surprise medical billing by ensuring coverage for drugs and treatments ahead of time.
- Always ask your doctor or pharmacist if a prior authorization is needed for a drug, treatment, or procedure.
- Your healthcare provider has the responsibility for submitting a prior authorization request. Always check with your insurance company if you need to confirm coverage for a specific treatment or procedure.

Be proactive in appealing a denial of coverage. Health insurance companies sometimes deny requests for prior authorization of a specific drug, treatment, or procedure. As an insured patient, you have the right to file an appeal to this decision. The appeal usually must be completed within a specified time. This involves sending a detailed letter to the insurance company outlining the reasons why the treatment is medically necessary.

- To strengthen your case, ask your doctor to write a supporting letter providing evidence of medical necessity.
- Send your letter and supporting documentation through certified mail or online to a designated appeals address.
- Within seven to 10 days, follow up with your insurance company to confirm that your appeal was received and that all necessary documents needed for a decision have been submitted.

The patient advocacy organization, Global Genes (www.globalgenes.org), has developed a toolkit for rare disease patients and families entitled Navigating Health Insurance. To access this resource, please visit: www.globalgenes.happyfox.com/kb/article/26-navigating-health-insurance/
In some cases, the Food and Drug Administration (FDA) allows pharmaceutical companies to provide their experimental drugs to people outside of clinical trials. This practice is known as compassionate use, or expanded access. The FDA created compassionate use in 1987 to allow people with life-threatening diseases to request access to investigational new drugs outside of clinical trials. Investigational drugs have not been approved by the FDA, and the FDA has not established that they are safe and effective for their specific use.

Gaining access to experimental medications through a compassionate use request can be a long and difficult process. However, it is a reasonable option for some people with rare diseases. It is important to know that the cost of these medications can be high. Also, few insurance companies will cover the cost of these drugs due to their experimental classification. A physician and other medical resources are required to administer these drugs, which further increases out-of-pocket costs for patients.

Talk to your doctor if you are interested in gaining access to an experimental medication through compassionate use dispensation. To be approved for the program, your doctor must contact the pharmaceutical company and submit a request to the FDA. The FDA requires the following criteria to be met to consider your request:

- Your disease is serious or immediately life-threatening.
- No treatment is available, or treatments approved for your disease have not helped.
- You are not eligible for clinical trials of the experimental drug.
- Your doctor verifies that you have no other options, and the experimental treatment may help.
- Your doctor believes the benefit outweighs the potential risks of the treatment.
- The pharmaceutical company that makes the drug agrees to provide it.

To learn more about the rules regarding compassionate use, please visit the patient information page on the FDA website: [www.fda.gov/news-events/public-health-focus/expanded-access](http://www.fda.gov/news-events/public-health-focus/expanded-access)

Expanded Access Studies

Participation in expanded access studies is another way to gain access to experimental treatments. In these studies, experimental drugs in the later stages of clinical trials are offered to people who do not qualify for the clinical trials. To determine if an experimental drug is available through an expanded access study, contact the drug’s manufacturer or visit [www.clinicaltrials.gov](http://www.clinicaltrials.gov) and search “expanded access studies.”

*Please refer to Appendix E for a complete list of insurance and financial assistance resources.*
Medical research is essential for developing new diagnostic tests and treatments to improve the lives of people with rare diseases. Many people living with rare diseases have an interest in advancing research for their disease through participation in clinical trials and clinical research. Clinical trials are a type of clinical research designed to evaluate and test new interventions, such as medications or medical devices, using human volunteers who meet specific study criteria. A clinical research study is any type of clinical research involving people, regardless of whether it is testing a specific intervention; clinical studies might investigate other aspects of care, such as improving quality of life.

Great advances are being made in understanding the basis for many rare diseases. This has the potential to generate new approaches for treatment. In some cases, a drug may be tested that is already approved by the FDA for some other indication. New information may suggest that it also could be helpful for people with a rare disorder. Physicians can prescribe an FDA-approved drug for reasons other than those for which it was initially approved. This is referred to as “off-label use.” In such cases, it is unknown whether the drug will work or whether side effects may occur; however, the physician and patient may judge that the risks are worth taking to see if there may be a benefit. Sometimes, new knowledge about the biological basis for a rare disorder might lead to the development of entirely new drugs that have never been used to treat any disease. Clinical trials can be conducted with existing FDA-approved drugs to see if they are safe and effective in treating a rare disease. Clinical trials are mandatory if FDA approval is being sought for a brand new drug.
RESOURCES FOR FINDING CLINICAL TRIALS

Stringent rules govern clinical trials, which are monitored by the National Institutes of Health (www.nih.gov) and the U.S. Food and Drug Administration (www.fda.gov). Before the FDA approves a clinical trial, scientists perform laboratory tests and studies in animals to determine a potential therapy's safety and efficacy. If these studies show favorable results, the FDA gives permission for the medication or device to be tested in humans.

Following are resources that can be helpful in finding clinical trials that are being conducted for a specific disease or condition:

The website www.clinicaltrials.gov, maintained by the NIH U.S. National Library of Medicine, is the primary resource to learn about current and upcoming clinical trials in the U.S. and internationally. Starting at the home page, users of the site can search for clinical trials by disease or condition, location, and key terms, such as a drug or investigator name. The federal government requires trials to be registered and to include detailed information on the site about eligibility criteria, site location, primary outcome measures, and other information.

The Rare Diseases Clinical Research Network (RDCRN) program, supported by the NIH National Center for Advancing Translational Sciences, advances medical research on rare diseases by providing support for clinical studies, collaboration, study enrollment, and data sharing. The RDCRN comprises 20 individual clinical research consortia and a Data Management and Coordinating Center. Each consortium focuses on at least three related rare diseases or conditions, participates in multiple studies, and actively involves patient advocacy groups as research partners. To learn more about RDCRN consortia and clinical trials, please visit: www.rarediseasesnetwork.org.

ResearchMatch is an organization that brings together people trying to find research studies and clinical trials, and researchers associated with registered IRB-approved studies on clinicaltrials.gov who are looking for volunteers. For more information, please visit: www.researchmatch.org.

National Patient Advocacy and Rare Disease Organizations are a good source of information about clinical trials and clinical research being conducted for a specific disease. The NORD website provides a library of information for more than 1,200 rare diseases, including national and international advocacy and research organizations for each disease www.rarediseases.org/for-patients-and-families/information-resources/rare-disease-information). Also, the National Institutes of Health (NIH) Genetic and Rare Disease Information Center (GARD) website contains an easy-to-use search tool that lists national and international advocacy organizations for specific diseases (https://rarediseases.info.nih.gov).

Disease specialists and clinicians are often knowledgeable about clinical trials and research currently in the pipeline for specific diseases. Frequently, researchers conducting clinical trials are medical doctors who may also see patients who are not enrolled in a study. These researchers may be able to refer you to a colleague who sees patients for your disease.
ELIGIBILITY FOR PARTICIPATION IN A CLINICAL TRIAL

Clinical studies have specific requirements, called eligibility criteria. These criteria outline who can participate in the study. Inclusion criteria are the requirements that allow someone to participate in a clinical study. Exclusion criteria are the factors that disqualify an individual from participating. Inclusion criteria for a trial might include factors such as:

- Age
- Stage of disease
- Sex
- Genetic profile
- Family history

Exclusion criteria might include factors such as specific health conditions or medications that could interfere with the treatment being tested. The website www.clinicaltrials.gov explains inclusion and exclusion criteria for each study in the “Eligibility Criteria” section.

Inclusion and exclusion criteria for clinical trials must be strictly followed. In some cases, these criteria may seem arbitrary, especially if a person is excluded because of one factor and otherwise is eager to participate in the trial. However, it is important to understand that the criteria are not arbitrary. It is critical to strictly follow the protocol, for at least two reasons. The first is for the safety of participants. Many of the criteria are designed to avoid potential adverse effects of the treatment being tested by excluding individuals who may be at high risk of complications. Another reason is to preserve the integrity of the trial. If individuals are included in a trial who don’t precisely fit the criteria stated in the protocol, there is a risk that the trial will fail to adequately test the effectiveness of the treatment. In the long run, this hurts everyone with the rare disease because it could prevent researchers from determining whether the treatment works or not.

CLINICAL TRIAL PHASES

Clinical trials are divided into several phases, and each phase can be thought of as its own study. It may take many years for a treatment to pass through the various phases. Preclinical testing in model systems, such as animal models, is usually conducted before clinical trials involving humans. The phases of human testing are:

- **Phase 0** – Study how the drug interacts with the disease tissue and affects the body in a small number of subjects.
- **Phase I** – Determine the safety of the treatment and the most appropriate dosage (of a medication) that can be safely given. Although it is possible that some benefits may be realized from Phase I treatment, this is not the primary aim of the study.
- **Phase II** – Determine the efficacy of the treatment. Specific criteria are established in advance to judge whether the treatment is working; side effects are also monitored.
- **Phase III** – Compare the efficacy of the treatment to other existing treatments for the disease.
- **Phase IV** – Monitor the treatment after FDA approval for long-term outcomes and rare side effects.
Following completion of Phase III, clinical trial data may be submitted to the FDA for approval, generally for indications that were tested in the clinical trial. For rare diseases, however, it is possible that Phase II data may need to be submitted, for two reasons. First, there may not be enough affected individuals to do a complete Phase III study for a rare condition. Second, for rare diseases, there may not be a known effective treatment to compare with a new treatment.

N OF 1 TRIALS

As more is learned about the cause of specific rare diseases, it sometimes is possible to implement a treatment that has never been tested before and where it is not possible to organize a randomized clinical trial with a large number of participants. In special circumstances, it might be possible to conduct a trial just in a single patient, comparing symptoms with and without treatment. If it is an existing FDA-approved drug, it may be used clinically, as described above, as an off-label use. If it is a new drug, special FDA approval must be obtained to test it in a patient.

RISKS AND BENEFITS OF PARTICIPATION

There are advantages of participating in a clinical trial. However, it is important to understand that some risks are involved in clinical research. Most clinical studies pose risks of minor discomfort that last for a short time. Some volunteer subjects may experience complications that require medical attention. The specific risks associated with a clinical trial are described in detail in the consent document that participants are asked to sign before taking part in the research. Also, the primary risks of participating in a study are explained by a member of the research team. This individual will also provide an opportunity to answer questions about the study. A medical ethics committee oversees each clinical trial to make sure all participants are treated appropriately. Also, if a participant is dissatisfied at any time during the conduct of a clinical trial, he or she is free to leave the study.

Following are some questions you may consider asking the research team when thinking about participation in a clinical trial:

- What is this study trying to determine?
- What treatment or test will I have, and will I receive the test or lab results?
- What are the chances that I will get the experimental treatment or placebo?
- What are the possible risks, side effects, and benefits of the study treatment?
- How long will the clinical trial last?
- Where will the study take place, and will transportation to the study site be provided if needed, such as a rideshare service?
- Can I do any part of the trial with my regular doctor? Is there a clinical trial closer to where I live?
- How will the study impact my everyday life?
- How will my privacy be protected?
How will my health be protected during the study?

What happens if my health problems get worse during the study?

Can I take my regular medications during the trial?

Who will administer my care while I am in the study? Will I be able to see my own doctor?

Does participation in the study cost anything? If so, how will I be reimbursed for expenses such as travel, parking, or lodging?

Will my insurance pay for costs not covered by the research trial, or will I need to pay out of pocket? Am I still eligible to participate if I do not have insurance?

Will you follow up on my health after the end of the study?

Will I learn the results of the study?

In addition to the risks involved, there are several benefits of participating in a clinical trial for individuals with rare diseases. A few of the most important benefits include:

- Access to promising new treatments often not available outside the setting of a clinical trial
- Close monitoring, advice, care, and support by a research team of doctors and other healthcare professionals who understand your disease or condition
- The opportunity to be the first to benefit from a new medication or method under study
- The chance to play a more active role in your own healthcare and gain a greater understanding of your disease
- The opportunity to help society by contributing to medical research. You may not benefit directly from participating in a clinical trial. However, your participation helps others and makes you a partner in scientific discovery.

For a complete list of resources to help you locate clinical trials and clinical research for a specific disease, please refer to Appendix F.

RIGHT TO TRY ACT

The Right to Try Act, signed into law in 2018, gives patients diagnosed with life-threatening diseases or conditions access to certain unapproved treatments. Pharmaceutical companies may sometimes grant permission for an investigational drug or biologic to be given to a patient as an eligible investigational drug as part of the Right to Try Act. To qualify, patients must have tried all approved treatment options. Also, they must be unable to participate in a clinical trial involving the eligible investigational drug. If you are interested in Right to Try, you should first discuss this option with your physician or disease specialist. He or she can determine whether a drug or biological product meets the criteria for use under the Right to Try Act.

For more information about the Right to Try Act, and to download the patient fact sheet, please visit: www.fda.gov/patients/learn-about-expanded-access-and-other-treatment-options/right-try
APPENDIX A
RESOURCES FOR UNDIAGNOSED PATIENTS AND FAMILIES

ALABAMA

Alabama Genomic Health Initiative (AGHI) – This program is one of the first statewide efforts in the nation to use the power of genomic analysis (genome sequencing) in helping to identify those who are at risk for disease and genomic abnormalities. AGHI leadership includes members from UAB, HudsonAlpha Institute for Biotechnology, and Tuskegee University, all of which are nationally recognized institutions and leaders in bioethics and genomic medicine. The program offers whole genome sequencing (WGS) for individuals with rare undiagnosed diseases. www.uabmedicine.org/agli

Smith Family Clinic for Genomic Medicine – Located on the campus of the HudsonAlpha Institute for Biotechnology in Huntsville, the Clinic provides diagnoses to patients with undiagnosed diseases using whole genome sequencing data when other genetic technologies are unable to reach a diagnosis. www.smithfamilyclinic.org.

The Undiagnosed Diseases Program (UDP) at the University of Alabama at Birmingham – A leading-edge program that addresses the needs of patients with severe chronic medical conditions for whom a diagnosis has not been made despite extensive efforts by the referring physician. www.uab.edu/medicine/genetics/undiagnosed-diseases.

NATIONAL PROGRAMS AND RESOURCES

American College of Medical Genetics – Site provides a search tool to locate medical genetic clinics within the U.S. https://clinics.acmg.net

Columbia University Discover Program for Undiagnosed Diseases www.cumc.columbia.edu/discover

NORD Undiagnosed Rare Disease Registry – A study that aims to limit the diagnostic odyssey of people with undiagnosed rare diseases through building a community of affected individuals and collecting relevant data about participant demographics, symptoms, and quality of life. www.rarediseases.org/iamrare-registry-program/

Scripps Idiopathic Diseases of Man (IDIOM) www.scripps.edu/science-and-medicine/translational-institute/translational-research/genomic-medicine/idiom/

The Undiagnosed Diseases Network (UDN) – The leading clinical research study in the U.S. for undiagnosed patients, funded by the National Institutes of Health (NIH) Common Fund www.commonfund.nih.gov/Diseases

Global Genes (www.globalgenes.org) – This organization has developed a resource for people with undiagnosed conditions called Becoming an Empowered Patient: A Toolkit for the Undiagnosed. Please access this resource online at: www.globalgenes.happyfox.com/kb/article/18-becoming-an-empowered-patient-a-toolkit-for-the-undiagnosed/
UNDIAGNOSED PATIENT ADVOCACY ORGANIZATIONS

Genetic Alliance Rare Disease & Genetic Conditions Support Community – A partnership between the Genetic Alliance and Inspire.com to create an online community that connects patients, family members, friends, and caregivers. www.inspire.com/groups/rare-disease-and-genetic-conditions/

National Organization for Rare Disorders (NORD) – Provides information and support for patients and families coping with undiagnosed rare medical conditions. www.rarediseases.org

Syndromes Without A Name (SWAN) – A supportive organization for families of children who have undiagnosed, unnamed conditions, or who are still looking for a diagnosis. www.swanusa.org
APPENDIX B
RARE DISEASE ADVOCACY ORGANIZATIONS AND RESOURCES

Alabama Rare – State-based advocacy organization that provides support for individuals with rare diseases and their families. This organization educates the broader community, creates awareness, and advocates for necessary change to improve healthcare delivery by collaborating with stakeholders within Alabama. www.alabamarare.org

Genetic and Rare Disease Information Center (GARD) – Website featuring an easy-to-use search tool that lists national and international advocacy organizations for specific diseases. www.rarediseases.info.nih.gov

Genetic Alliance – A broad-based collation that builds partnerships to promote healthy lives for those living with genetic conditions. www.geneticalliance.org

Global Genes – An organization focused on patient education and advocacy programs as well as helping people with rare diseases find a sense of community and support through connecting with one another. www.globalgenes.org

National Organization for Rare Disorders (NORD) – Advocacy organization dedicated to improving the lives of individuals and families living with rare diseases. www.rarediseases.org

NORD’s website provides a library of information for more than 1,200 rare diseases, including national and international advocacy and research organizations for each disease. www.rarediseases.org/for-patients-and-families/information-resources/rare-disease-information/

PTEN Hamartoma Tumor Syndrome Foundation – Nonprofit organization founded to find treatments or therapies for PTEN Syndromes by funding research, providing education, supporting patients, and raising awareness. https://ptenfoundation.org or email: kristin@ptenfoundation.org

Rare Action Network (RAN) – Advocacy organization dedicated to empowering a unified network of individuals and organizations with the resources to become effective advocates for rare diseases through national and state-based initiatives across the U.S. www.rareaction.org/about/rare-action-network/

SOURCES OF RELIABLE HEALTH INFORMATION

Genetics Home Reference – The National Library of Medicine’s website for consumer information about genetic conditions and the genes or chromosomes related to those conditions. https://medlineplus.gov/about/general/genetics/newhome/

MEDLINEplus – A service of the National Library of Medicine (NLM) and the National Institutes of Health (NIH) that offers high-quality information on more than 600 diseases and conditions. www.nlm.nih.gov/medlineplus/

Evaluating Health Information – MEDLINEplus has posted a series of documents and links that provide helpful guidance on how to find reliable information on the Web as well as tips for evaluating the quality and accuracy of internet sources. www.medlineplus.gov/evaluatinghealthinformation.html

National Human Genome Research Institute (NHGRI) – Provides helpful information on genetic and/or rare diseases research, genetic and rare conditions, patient support groups, and more. Please visit the NHGRI site at: www.genome.gov.
National Human Genome Research Institute (NHGRI) Talking Glossary of Genetic Terms – Designed to help everyone understand the terms and concepts used in genetic research; features specialists in the field of genetics sharing their descriptions of terms with images, animation, and links to related terms. www.genome.gov/genetics-glossary


SOCIAL MEDIA WEBSITES FOR CONNECTING PEOPLE WITH RARE DISEASES

GenomeConnect – Online tool that allows people to share their genetic test results and health information with researchers and healthcare providers as well as to connect with other individuals who have a similar diagnosis or related symptoms. www.genomeconnect.org

Global Genes – A rare diseases patient advocacy organization that helps people with rare diseases find a sense of community and support through connecting with one another. www.globalgenes.org

Inspire – Users can search the site by condition to find information and online communities. www.inspire.com

MyGene² – A consortium that connects families with rare conditions to clinicians and researchers who are interested in publicly sharing health and genetic information with one another. https://mygene2.org/MyGene2/

RareConnect – Features online communities for patients and families with rare medical conditions. The project is a collaboration between the European Rare Disease Organisation (EURORDIS) and the National Organization for Rare Disorders (NORD). www.rareconnect.org/en/communities

RareShare - Online social hub dedicated to patients, families, and healthcare professionals affected by rare medical disorders. www.rareshare.org
### APPENDIX C

**STANDING LIST OF SPECIALTY CLINICS IN THE STATE OF ALABAMA (AS OF 2020)**

It is best for patients to contact the clinics directly, as some contact names may have changed.

<table>
<thead>
<tr>
<th>CLINIC</th>
<th>CITY</th>
<th>AFFILIATION</th>
<th>CONTACT NAME</th>
<th>CONTACT PHONE</th>
<th>CONTACT EMAIL</th>
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<tbody>
<tr>
<td>East Alabama Medical Center (all clinics)</td>
<td>Auburn</td>
<td>East Alabama Medical Center</td>
<td>Peter King, MD</td>
<td>(334) 749-3411</td>
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<tr>
<td>ALS Clinic</td>
<td>Birmingham</td>
<td>UAB</td>
<td>Sarah O’Kelley</td>
<td>(205) 934-5471</td>
<td><a href="mailto:sokkelley@uab.edu">sokkelley@uab.edu</a></td>
</tr>
<tr>
<td>Autism Spectrum Disorders (ASDs)</td>
<td>Birmingham</td>
<td>UAB/Children’s</td>
<td>Alan Percy, MD</td>
<td>(205) 934-5471</td>
<td><a href="mailto:apercy@peds.uab.edu">apercy@peds.uab.edu</a></td>
</tr>
<tr>
<td>Chiari Malformation Clinic</td>
<td>Birmingham</td>
<td>UAB/Children’s</td>
<td>Steven Rowe, MD, MPH</td>
<td>(205) 996-5864</td>
<td><a href="mailto:smrowe@uab.edu">smrowe@uab.edu</a></td>
</tr>
<tr>
<td>Civitan-Sparks (Dev. Delay Clinic)</td>
<td>Birmingham</td>
<td>UAB/Children’s</td>
<td>Avi Maden-Swain, PhD</td>
<td>(205) 638-9285</td>
<td></td>
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<tr>
<td>Craniofacial Clinic</td>
<td>Birmingham</td>
<td>UAB/Children’s</td>
<td>Victor Sung, MD</td>
<td>(205) 801-8986</td>
<td><a href="mailto:vsung@uab.edu">vsung@uab.edu</a></td>
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<tr>
<td>Cystic Fibrosis Clinic</td>
<td>Birmingham</td>
<td>UAB</td>
<td>Bradley Troxler, MD</td>
<td>(205) 638-9941</td>
<td><a href="mailto:btroxler@peds.uab.edu">btroxler@peds.uab.edu</a></td>
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<tr>
<td>Endocrinology and Metabolism Clinic</td>
<td>Birmingham</td>
<td>UAB</td>
<td>Ken McCormick, MD</td>
<td>(205) 996-9369</td>
<td><a href="mailto:kenmc@uab.edu">kenmc@uab.edu</a></td>
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<tr>
<td>Fetal Medicine Clinic</td>
<td>Birmingham</td>
<td>UAB/Children’s</td>
<td>Drew Davis, MD</td>
<td>(205) 638-9941</td>
<td><a href="mailto:dDavis@peds.uab.edu">dDavis@peds.uab.edu</a></td>
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<tr>
<td>Huntington’s Disease</td>
<td>Birmingham</td>
<td>UAB</td>
<td>Bruce Korf, MD, PhD</td>
<td>(205) 934-4983</td>
<td><a href="mailto:bkorf@uabmc.edu">bkorf@uabmc.edu</a></td>
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<tr>
<td>MDA Clinic (all MDs)</td>
<td>Birmingham</td>
<td>UAB/Children’s</td>
<td>T. Brooks Vaughan, MD</td>
<td>(205) 934-0994</td>
<td><a href="mailto:byoder@uab.edu">byoder@uab.edu</a></td>
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<td>Metabolic Bone Clinic</td>
<td>Birmingham</td>
<td>UAB/Children’s</td>
<td>Brad Yoder, PhD</td>
<td>(205) 934-0994</td>
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<tr>
<td>Movement Disorder/Spasticity Clinic</td>
<td>Birmingham</td>
<td>UAB/Children’s</td>
<td>Alan Percy, MD; Jane Lane, RN (contact)</td>
<td>(205) 996-6406</td>
<td><a href="mailto:apercy@peds.uab.edu">apercy@peds.uab.edu</a></td>
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<td>Spina Bifida</td>
<td>Birmingham</td>
<td>UAB/Children’s Hospital</td>
<td>Betsy Hopsin, RSN</td>
<td>(205) 638-5281</td>
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<td>Transverse Myelitis</td>
<td>Birmingham</td>
<td>UAB</td>
<td>Khurram Bashir, MD</td>
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<td>Martina Bebine, MD</td>
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<td>Dothan Specialty Clinic (all clinics)</td>
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<td>Dothan Specialty Clinic</td>
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<td>Flowers Hospital (all clinics)</td>
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<td>Flowers Clinic</td>
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<td>Noland Hospital (all clinics)</td>
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<td>Noland Hospital</td>
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<td>(205) 783-8440</td>
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<td>SE Alabama Medical Center (all clinics)</td>
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<td>ALS Clinic</td>
<td>Huntsville</td>
<td>Crestwood Medical Center</td>
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<td>(256) 429-4113</td>
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<tr>
<td>Cancer Center of Huntsville</td>
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<td>The Cancer Center of Huntsville</td>
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<td>(256) 265-1822</td>
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<td>Clearview Cancer Institute (all cancers)</td>
<td>Huntsville</td>
<td>Huntsville Hospital</td>
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<td>Fetal Medicine Clinic</td>
<td>Huntsville</td>
<td>Huntsville Woman &amp; Children's</td>
<td>Margaret Carter, MD</td>
<td>(256) 265-0880</td>
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<td>Pediatric Endocrinology Clinic</td>
<td>Huntsville</td>
<td>Huntsville Hospital</td>
<td>Linnea Larson-Williams, MD</td>
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<td>Kimberly Limbo, MD</td>
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<td>USA Children’s and Women’s Hospital (all clinics)</td>
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<td>MDA Clinic (all MDs)</td>
<td><a href="https://www.childrensal.org/muscular-dystrophy-care-center">https://www.childrensal.org/muscular-dystrophy-care-center</a></td>
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<td>Metabolic Bone Clinic</td>
<td><a href="https://www.childrensal.org/metabolic-bone-clinic">https://www.childrensal.org/metabolic-bone-clinic</a></td>
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<td>Movement Disorder/Spasticity Clinic</td>
<td><a href="https://www.childrensal.org/rehab-medicine-outpatient-clinics">https://www.childrensal.org/rehab-medicine-outpatient-clinics</a></td>
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<td>Pediatric Hematology Disorders Clinic</td>
<td><a href="https://www.childrensal.org/hematology">https://www.childrensal.org/hematology</a></td>
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<td>Pituitary Disorders Clinic</td>
<td><a href="https://www.uab.edu/medicine/endocrinology/patient-care">https://www.uab.edu/medicine/endocrinology/patient-care</a></td>
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<td>Rett Syndrome</td>
<td><a href="https://www.uab.edu/civitansparks/rett-syndrome">https://www.uab.edu/civitansparks/rett-syndrome</a></td>
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<td>Spina Bifida</td>
<td><a href="https://www.childrensal.org/rehab-medicine-outpatient-clinics">https://www.childrensal.org/rehab-medicine-outpatient-clinics</a></td>
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<td>Transverse Myelitis</td>
<td><a href="https://www.uabmedicine.org/patient-care/conditions/transverse-myelitis">https://www.uabmedicine.org/patient-care/conditions/transverse-myelitis</a></td>
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<td>Tubular Sclerosis Clinic</td>
<td><a href="https://www.uabmedicine.org/locations/tuberous-sclerosis-clinic">https://www.uabmedicine.org/locations/tuberous-sclerosis-clinic</a></td>
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<td>Dothan Specialty Clinic (all clinics)</td>
<td><a href="http://dothanspecialty.com/office-locations/">http://dothanspecialty.com/office-locations/</a></td>
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<td>Flowers Hospital (all clinics)</td>
<td><a href="https://www.flowershospital.com/">https://www.flowershospital.com/</a></td>
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<td>Noland Hospital (all clinics)</td>
<td><a href="https://nolandhospitals.com">https://nolandhospitals.com</a></td>
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<td>SE Alabama Medical Center (all clinics)</td>
<td><a href="https://www.samc.org/">https://www.samc.org/</a></td>
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<td>ALS Clinic</td>
<td><a href="https://www.crestwoodmedcenter.com/als-clinic">https://www.crestwoodmedcenter.com/als-clinic</a></td>
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<td>Cancer Center of Huntsville</td>
<td><a href="http://www.tcchsv.com/contact_us.aspx">http://www.tcchsv.com/contact_us.aspx</a></td>
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<td>Clearview Cancer Institute (all clinics)</td>
<td><a href="http://www.clearviewcancer.com/about/hours-and-locations/huntsville-main">http://www.clearviewcancer.com/about/hours-and-locations/huntsville-main</a></td>
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<td>Fetal Medicine Clinic</td>
<td><a href="https://www.huntsvillehospital.org/maternal-fetal-medicine">https://www.huntsvillehospital.org/maternal-fetal-medicine</a></td>
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<td>Pediatric Endocrinology Clinic</td>
<td><a href="https://www.huntsvillehospital.org/pediatric-endocrinology-diabetes-clinic">https://www.huntsvillehospital.org/pediatric-endocrinology-diabetes-clinic</a></td>
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<td>Pediatric Neurology Clinic</td>
<td><a href="https://www.huntsvillehospital.org/huntsville-hospital-pediatric-neurology">https://www.huntsvillehospital.org/huntsville-hospital-pediatric-neurology</a></td>
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<td>Spine and Neuro Clinic</td>
<td><a href="https://www.huntsvillehospital.org/services/spine-and-neuro">https://www.huntsvillehospital.org/services/spine-and-neuro</a></td>
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<td>St. Jude Affiliate Clinic (Cancers)</td>
<td><a href="http://www.hhwomenandchildren.org/st-jude-affiliate-clinic">http://www.hhwomenandchildren.org/st-jude-affiliate-clinic</a></td>
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<td>Providence Hospital (all clinics)</td>
<td><a href="https://www.providencehospital.org/">https://www.providencehospital.org/</a></td>
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<td>USA Children’s &amp; Women’s Hospital (all clinics)</td>
<td><a href="http://www.usahealthsystem.com/usacwh">http://www.usahealthsystem.com/usacwh</a></td>
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<td>USA Medical Center (all clinics)</td>
<td><a href="https://www.usahealthsystem.com/locations/university-hospital">https://www.usahealthsystem.com/locations/university-hospital</a></td>
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<td>Montgomery Children’s Specialty Center (all clinics)</td>
<td><a href="http://www.montgomerychildrenscare.com/">http://www.montgomerychildrenscare.com/</a></td>
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<td>UAB Multispecialty Clinic</td>
<td><a href="https://uabmedicine-baptist.com/">https://uabmedicine-baptist.com/</a></td>
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APPENDIX D
RESOURCES TO LOCATE HEALTHCARE PROVIDERS AND SPECIALTY CLINICS

DISEASE ADVOCACY ORGANIZATIONS

Alabama Rare – State-based advocacy organization that provides support for individuals with rare diseases and their families. The organization educates the broader community, creates awareness, and advocates for necessary change to improve healthcare delivery by collaborating with stakeholders within Alabama. www.alabamarare.org.

Genetic Alliance – Website contains a list of advocacy and patient organizations for specific diseases. www.geneticalliance.org

Genetic and Rare Disease Information Center (GARD) – Website contains an easy-to-use search tool that lists national and international advocacy organizations for specific diseases that can be helpful in locating healthcare providers and clinics. www.rarediseases.info.nih.gov

Global Genes – An organization focused on patient education and advocacy programs as well as helping people with rare diseases find a sense of community and support through connecting with one another. www.globalgenes.org

National Organization for Rare Disorders (NORD) – Website provides a library of information that includes national and international advocacy and research organizations for each disease. www.rarediseases.org

PTEN Hamartoma Tumor Syndrome Foundation – Nonprofit organization founded to find treatments or therapies for PTEN Syndromes by funding research, providing education, supporting patients, and raising awareness. https://ptenfoundation.org or email kristin@pten.org

CLINICAL GENETICS CENTERS

The American College of Medical Genetics – Features a Genetics Clinics Database to assist with locating a U.S. genetics center. https://acmg.net

The American Society of Human Genetics (ASHG) – Professional organization of research and clinical geneticists that maintains a database of its members, some of whom live outside the U.S. Some geneticists listed may be researchers only and may not offer medical care. www.ashg.org

The National Society of Genetic Counselors – Features a database of genetics counseling services that are searchable by location, name, institution, type of practice, or specialty. www.nsgc.org

MEDLINEPlus – Website maintained by the U.S. National Library of Medicine that provides a link to various websites and directories to assist in finding health professionals, services, and facilities. www.medlineplus.gov/directories.html
MEDICAL RESEARCHER PROGRAMS AND NETWORKS

The Rare Diseases Clinical Research Network (RDCRN) – Comprises 22 distinctive consortia working to improve the availability of rare disease information, treatment, clinical studies, and general awareness for patients and the medical community. The RDCRN provides up-to-date information for patients and connects them with advocacy groups, expert doctors, and clinical research opportunities. [www.rarediseasesnetwork.org](http://www.rarediseasesnetwork.org)

The Centers for Mendelian Genomics – Program collaborates with healthcare professionals and researchers working with people who have Mendelian disorders with a focus on discovering causes of rare genetic disorders. [www.mendelian.org](http://www.mendelian.org)

ClinicalTrials.gov – Website that was developed by the U.S. National Institutes of Health (NIH) through the National Library of Medicine to provide patients, family members, and the public with current information on clinical research studies in the U.S. This resource may be helpful in locating researchers who are studying your condition. A basic search can be conducted using the condition name as the search term; click on a study to review the eligibility criteria and obtain the study’s contact information. [www.clinicaltrials.gov](http://www.clinicaltrials.gov)

The Patient Recruitment and Public Liaison (PRPL) – Office at the National Institutes of Health (NIH) that provides information about participating in research conducted at the NIH Clinical Center Hospital. You may call the PRPL office toll-free at 1-800-411-1222 or send an email to prpl@mail.cc.nih.gov to ask about clinical trials being conducted for a specific disease and contact information for research involved in these trials.
APPENDIX E
INSURANCE AND FINANCIAL ASSISTANCE RESOURCES

ALABAMA

Alabama Department of Rehabilitation Services (ADRS) – The primary state agency that serves Alabamians with disabilities from birth throughout their lives through four main programs: early intervention (EI); children’s rehabilitation services (CRS); independent living; and vocational rehabilitation services (VR). www.rehab.alabama.gov

Alabama Disabilities Program (ADAP) – This program provides legal service to Alabamians with disabilities to protect, promote, and expand their rights. ADAP is part of the National Disability Rights Network (NDRN). www.adap.ua.edu

Alabama Health Insurance Premium Payment (HIP) Program – Designed to save money for Medicaid families with high healthcare costs. The program pays most out-of-pocket medical expenses for qualifying Medicaid recipients and reimburses policyholders for the cost of health insurance provided by an employer or COBRA. Some recipients may also qualify to receive reimbursement for the cost of a family health insurance policy premium. www.myalhipp.com

Alabama Home and Community-Based Services (HCBS) Medicaid Waivers - Provide services to support people with intellectual or significant physical and medical disabilities. These services provide support that helps people live at home or in other community-based settings like group homes. The services provided can include the following: personal care; skilled nursing; behavioral supports; respite services; assistive technology; and homemaker services. Alabama’s seven waiver programs are administered by the Alabama Medicaid Agency and its state partners: the Alabama Department of Mental Health (ADMH); the Alabama Department of Senior Services (ADSS); and the Alabama Department of Rehabilitation Services (ADRS). For more information about Alabama’s HCBS Medicaid Waivers, please visit the Alabama Medicaid website: www.medicaid.alabama.gov/content/6.0_LTC_waivers/6.1_HCBS_waivers.aspx

ALL Kids - Alabama Children’s Health Insurance Program (CHIP) administered by the Alabama Department of Public Health, is a low-cost, comprehensive healthcare coverage program for children under age 19. The program includes benefits such as regular checkups and immunizations, sick child doctor visits, prescriptions, vision and dental care, hospitalization, mental health and substance abuse services, and other services. ALL Kids uses Blue Cross Blue Shield of Alabama to provide medical and mental health services through their preferred provider network (PPO). www.alabamapublichealth.gov/allkids

The Alabama Health Insurance Premium Payment (HIP) Program - Designed to save money for Medicaid families with high healthcare costs. The program eliminates most out-of-pocket medical expenses for qualifying Medicaid recipients and reimburses policyholders for the cost of health insurance provided by an employer or COBRA. Some recipients may also qualify to receive reimbursement for the cost of a family health insurance policy premium. www.myalhipp.com

UAB Hospital’s Charity Care Program – Designed for uninsured or underinsured patients of the Kirklin Clinic of UAB Hospital or affiliated clinics. Applicants may qualify for financial assistance, which is based on household size and household income. To receive an application, please ask your UAB healthcare provider.
FEDERAL

COBRA (Consolidated Omnibus Budget Reconciliation Act) – Government legislation that gives workers and their families who lose their health benefits through job loss and other circumstances the right to choose to continue group health benefits provided by their employer for limited periods when leaving employment for a specific amount of time.  www.dol.gov/general/topic/health-plans/cobra

The Medicaid Insurance Program – Designed for people who are 65 years or older, certain younger people with disabilities and people with end-stage renal disease (ESRD).  www.medicaid.gov

The Medicare Insurance Program – Federal-sponsored, state-administered health insurance program for families and individuals with low incomes.  www.medicare.gov

The Affordable Care Act/Obamacare – Provides access to the federal government’s insurance marketplace for health insurance policies. These are provided by private insurance companies for individuals, families, and small businesses. Health Insurance Marketplace call center: 1-800-318-2596 www.healthcare.gov

USA.gov – Provides links to federal resources that can help with the cost of medical care and prescription drugs (www.usa.gov/help-with-bills#item-36707). You can also contact them directly at: 844-872-4681.

NON-PROFIT PATIENT ASSISTANCE PROGRAMS

Family Voices – Program that aims to achieve family-centered care for all children and youth with special healthcare needs and/or disabilities. The website features a map to assist in locating the Family-to-Family Health Information Center in each state.  www.familyvoices.org

Needy Meds – Non-profit organization that lists programs to help those who cannot afford medications and healthcare costs. NeedyMeds has information about government programs, low-cost or free medical and dental clinics, and prescription assistance. The program also offers disease-specific financial aid programs.  www.needymeds.org

NORD RareCare® – Assistance programs that provide medication, financial assistance with insurance premiums and copays, diagnostic testing assistance, and travel assistance for clinical trials or consultation with disease specialists.  www.rarediseases.org/for-patients-and-families/help-access-medications/patient-assistance-programs-2/

Patient Advocate Foundation (PAF) – Provides case management assistance for the uninsured or underinsured with life-threatening or debilitating illnesses. Services include help with access to care, copay assistance, social security disability applications, and insurance appeals. PAF also has a National Financial Resource Directory that allows patients to find resources within a specific state.  www.patientadvocate.org

Patient Services, Inc. - Provides financial support and guidance for qualified patients with specific, rare chronic diseases.  www.patientservicesinc.org

The National Human Genome Research Institute (NHGRI) – Program at the National Institutes of Health (NIH) that provides information about financial assistance resources for people who need help paying for medical care.  www.genome.gov/11008842/

The Pharmaceutical Research and Manufacturers of America (PhRMA) – Website features a tool that allows patients to search for financial assistance resources offered through various biopharmaceutical industry programs to help cover the cost of medications.  www.medicineassistance tool.org/Who-We-Are
ORGANIZATIONS PROVIDING DISEASE-SPECIFIC FUNDS

**Good Days** – Provides help to patients with specific life-altering conditions. Assistance includes help with the cost of medications and travel. [www.mygooddays.org](http://www.mygooddays.org)

**HealthWellFoundation** – Provides financial assistance for those with specific chronic or life-altering conditions. [www.healthwellfoundation.org](http://www.healthwellfoundation.org)

**Patient Services, Inc.** – Offers assistance programs for various conditions; programs may include help with copayments, premiums, ancillary services, infusion, nursing services, or travel. [www.patientservicesinc.org](http://www.patientservicesinc.org)

**The Assistance Fund** – Provides various services to help patients with specific chronic or serious illnesses cover the cost of FDA-approved medications. Visit [www.tafcares.org/](http://www.tafcares.org/) or [www.tafcares.org/program-listing/](http://www.tafcares.org/program-listing/) to view program listings. You can also contact them directly at: 855-845-3663.

**The Patient Access Network Foundation (PAN Foundation)** – Helps the underinsured cover out-of-pocket costs associated with medication and treatments for specific diseases. [www.panfoundation.org](http://www.panfoundation.org)

RESOURCES FOR DEALING WITH INSURANCE PROVIDERS


**The Patient Advocate Foundation** – Website features an Education Resource Library that offers free downloadable brochures and webinars on a range of health insurance topics. [www.patientadvocate.org/explore-our-resources/education-resource-library/](http://www.patientadvocate.org/explore-our-resources/education-resource-library/)
APPENDIX F
RESOURCES TO LOCATE CLINICAL TRIALS AND CLINICAL RESEARCH

Clinicaltrials.gov – Website maintained by the NIH U.S. National Library of Medicine that serves as the primary resource to learn about current and upcoming clinical trials in the U.S. and internationally. www.clinicaltrials.gov

Genetic and Rare Disease Information Center (GARD) – Website contains an easy-to-use tool that lists national and international advocacy organizations for specific diseases. These organizations are a good source of information about clinical trials and clinical research. www.rarediseases.info.nih.gov

ResearchMatch – Organization that brings together people trying to find research studies and clinical trials, and researchers associated with registered IRB-approved studies on clinicaltrials.gov who are looking for volunteers. www.researchmatch.org

The National Organization for Rare Diseases (NORD) – Website provides a library of information that includes a directory of national and international advocacy and research organizations for each disease. These organizations are a good source of information about clinical trials and clinical research. www.rarediseases.org

The Rare Diseases Clinical Research Network (RDCRN) – Program supported by the NIH National Center for Advancing Translations Sciences. The program advances medical research on rare diseases by providing support for clinical studies, collaboration, study enrollment, and data sharing. The RDCRN comprises 20 individual clinical research consortia and a Data Management and Coordinating Center; each consortium focuses on at least three related rare diseases or conditions, participates in multiple studies, and actively involves patient advocacy groups as research partners. www.rarediseasenetwork.org
Prepared by:
Alabama Rare Disease Advisory Council (ARDAC)

Special thanks to the following rare disease patients and family members for their review of this Rare Disease Resource Guide:

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