

UNDIAGNOSED DISEASES PROGRAM

The UAB Undiagnosed Diseases Program (UDP) seeks to meet the needs of patients with severe chronic medical conditions for whom a diagnosis has not been made. The UDP team is led by four UAB Medicine faculty physicians and also includes a certified genetic counselor and two clinical nurse coordinators. UAB physicians from various medical and pediatric subspecialties serve as consultants and provide their expertise in each case as necessary. *(See opposite page for more information about our team.)* **The program uses genome sequencing as one of its major diagnostic approaches.**

Patient Population

The UDP strives to meet the needs of **patients of all ages**, including those with rare diseases, those with conditions not previously known to exist, and those with atypical presentations of common diseases. The program is designed to explore conditions amenable to diagnostic work-up and intervention and is not appropriate for evaluation of chronic conditions such as fatigue, weight loss, fibromyalgia, and diffuse pain.

Determination for Evaluation

Only a limited number of patients who are referred will be evaluated. The principal deciding factor regarding the decision to evaluate a patient is that the UDP has the expertise and resources necessary to possibly achieve a diagnosis. In some cases, the staff may determine that it will be unable to add to an already extensive work-up, in which case we are often able to arrange evaluations in alternative UAB programs. Despite evaluation in the UDP, it is possible that a diagnosis will not be made.

Prospective patients must meet the following criteria:

- Are referred by a physician (self-referrals are not accepted)
- Have had an extensive evaluation not resulting in a diagnosis
- The condition prompting the referral must be causing significant impairment to quality of life (organ dysfunction, constitutional symptoms, or loss of function) and must be of at least six months duration
- Are clinically stable and not acutely ill (requiring urgent evaluation and/or management in an acute setting)



Children's
of Alabama®

To refer a PEDIATRIC patient, please call:
205.975.6337.

UAB MEDICINE
UNDIAGNOSED DISEASES PROGRAM

To refer an ADULT patient, please fax the requested information *(see opposite page)* to 205.934.4111

Some of this information can also be submitted online:
uab.edu/medicine/genetics/undiagnosed-diseases

For records > 30 pages, please mail to:
UAB Kaul Human Genetics Building
Suite 230 720 20th Street South
Birmingham, AL 35233

For more information about the program, call 205.996.6583. Physicians may call UAB MIST at 1.800.822.6478 to be connected with our UAB Medicine faculty and specialists. UAB MIST is a 24/7 physician to-physician consultation and referral service.

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Meet Our Team!

UAB Program Physicians



Bruce Korf, MD, PhD
Professor & Chair
UAB Department
of Genetics



Maria Descartes, MD
Professor
UAB Department
of Genetics



Anna Hurst, MD
Assistant Professor
UAB Department
of Genetics



Martin Rodriguez, MD
Associate Professor
UAB Department
of Medicine

Pediatric Subspecialists



IMMUNOLOGY
Prescott Atkinson, MD, PhD
Professor



GASTROENTEROLOGY
Reed Dimmitt, MD, MSPH
Associate Professor



NEPHROLOGY
Daniel Feig, MD, PhD, MPH
Professor



CARDIOLOGY
Walter Johnson, MD
Professor



PULMONOLOGY
David Lozano, MD
Associate Professor



NEUROLOGY
Jayne Ness, MD, PhD
Associate Professor



RADIOLOGY
Stuart Royal, MS, MD, FACR
Clinical Professor



DERMATOLOGY
Amy Theos, MD
Associate Professor



RHEUMATOLOGY
Peter Weiser, MD
Assistant Professor

Referral & Evaluation Process

For patients meeting the criteria outlined on the opposite page, please supply the following elements needed for the referral; these can be faxed or mailed (*see contact information provided on the opposite page*):

- Referring physician's first and last name, practice address/telephone/fax numbers, and email address
- Physician's Summary of Present Illness Form: can be submitted online or a written summary to include nature and onset of condition(s), lab and imaging abnormalities, results of prior evaluations, treatments used and their outcomes, and patient's current condition
- List of current medications and drug allergies
- Patient's contact and insurance information
- For pediatric patients, provide prenatal, birth, and family history, and growth curves

Once the above elements are received, a letter of acknowledgement will be sent to the referring physician, including a request for missing elements if applicable. The estimated time from receipt of the referral to determination for evaluation is six to eight weeks. The expectation is that the patient will be evaluated in the outpatient setting; should extensive and/or additional invasive testing be required, hospital admission will be arranged.

The UDP should not be envisioned as a referral site due to lack of availability of basic diagnostic or evaluation resources to the referring physician. Similarly, the program is not intended for management of patients who have already been diagnosed with complex medical conditions. Upon diagnosis, treatment and management recommendations will be made, to be coordinated in our facility and/or per the patient's local providers, according to each patient's needs and the available resources. The UDP does not assume ongoing care for patients, either during the evaluation or following determination of a diagnosis.