DESSH
DeSanto-Shinawi Syndrome

A Guide to Research Participation for DeSanto-Shinawi Syndrome Families

We understand that including your loved-one in research may be intimidating for some families. The enclosed information serves to:

- Explain why research is important
- Summarize different options for research participation
- Describe how clinical data will be used

1. Why is research important?

"Genetics research studies how individual genes or groups of genes are involved in health and disease. Understanding genetic factors and genetic disorders is important in learning more about promoting health and preventing disease." (https://www.cdc.gov/genomics/about/basics.htm)

Specifically, in our studies we wish to understand how genetic changes (also known as mutations) in the *WAC* gene cause the signs and symptoms of DESSH syndrome. These preliminary studies will lay the groundwork for future research that will be aimed at intervention. Intervention involves the development of tools to assist in early diagnosis, and therapies (including pharmacological and non-pharmacological) that will attempt to mitigate one or more aspect of DESSH syndrome.

In addition, understanding whether all *WAC* mutations act in the same way or whether different mutations lead to different signs and symptoms in individuals (termed genotype-phenotype correlations) are key to elucidate. This allows clinical geneticists to provide accurate information to current and newly diagnosed patients and their families about health issues which may be of the most importance to them.

We are hoping that collection of large amounts of data from many patients with DESSH syndrome will enable our team to understand the natural history of this disorder. We are planning to use this information to provide anticipatory guidance and recommendations to medical professionals who are managing patients with DESSH syndrome.

2. Different types of research.

There are two main types of research we wish to undertake. The first is clinical research. The second is pre-clinical research.

a. Clinical research involves the collection of patient data such as age, physical characteristics, intellectual or behavioral difficulties, medications being used, ongoing illnesses and anecdotal observations made by close family and friends. Descriptions may be provided as written word or other data types such as photographs and video provided by family or medical providers.

These data help inform clinicians around the world on what to look out for when they see new patients that may be undiagnosed, or for current patients with a DESSH diagnosis and how best to help them manage specific facets of DESSH.

b. Pre-clinical research involves the use of animal models (for example mice and zebrafish) as well as cell lines grown in flasks that are derived from patients' actual tissues (such as blood). This helps us understand the mechanisms of how *WAC* mutations alone cause DESSH without confounding and complicating factors faced in clinical research such as environmental factors, different genetic backgrounds, locations, ages etc. They also provide excellent models to test interventions aimed at treating one or more aspects of DESSH. We need to understand whether something is likely to work and provide a proof-of-principle before a tool can be developed for use clinically.

3. How will clinical data be used?

As outlined above, clinical data may include written information from a patient's family/guardians or medical professionals. Providing photographs are highly desirable but are not a requirement for participation in our studies. These data can be used in several ways. It may be used clinically to help create a database of information that will be *disseminated* within the medical community through the form of publication in medical journals (example provided) or presentation at medical conferences or family support meetings. Both publications and conferences are key for communicating medical discoveries with other health professionals (clinicians and research scientists). Furthermore, the data will be used by geneticists and other specialist health professionals involved in frontline DESSH patient care to understand and track the medical history of patients. The data collected will help us provide a comprehensive plan of care and management recommendations that will ultimately maximize the developmental potential and happiness of individuals with DESSH syndrome.

4. Are you and your family willing to participate in research?

There are several ways in which your family can be involved in research and, after reviewing the above information, we ask you to contact us if you have more questions. There are varying levels of participation and decisions made by you, the families, will be respected and understood. If at any time you wish to increase or decrease participation level, you are free to contact Dr. Shinawi (information below) and the data collected will be amended appropriately.

For more information about research opportunities please contact:
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