



Complex Child E-Magazine

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The NIH Undiagnosed Diseases Program: Diagnosing Rare Diseases by Susan Agrawal

The National Institutes of Health (NIH), the US agency responsible for medical research, recently began a new program through the Office of Rare Diseases. This program, called the Undiagnosed Diseases Program, invites 50-100 individuals per year with longstanding undiagnosed disorders to the NIH's Clinical Center in Bethesda, Maryland, for further evaluation.

While the ultimate goal of this program from the perspective of the patient family is diagnosis, the NIH also hopes to learn more about the basis of disease, genetic markers of the disease, and so forth. Acceptance to the program does not guarantee a diagnosis. In fact, a large percentage of individuals who participate will remain undiagnosed. The NIH also will not provide ongoing or follow-up care, whether a diagnosis is determined or not.

In order to be considered for the program, your child must be referred by his or her doctor. Your doctor must submit the following materials:

- A letter describing your child's illness, when it started, and how your child is doing currently
- A list of attempted treatments (medications or procedures) and their impact, either positive or negative
- Medical records, including diagnostic tests, imaging, and other relevant materials

Materials should be submitted to:

National Human Genome Research Institute
National Institutes of Health
Undiagnosed Diseases Program
10 Center Drive
MSC 1851, Building 10, Room 10C103
Bethesda, Maryland 20892-1851

Contact 866-444-8806 for more information.

After these items are submitted, your child's case will be reviewed over the next six to eight weeks. If accepted, additional medical records may be requested. Then your child will be asked to come to the Clinical Center for a multi-disciplinary evaluation by top

specialists. This may include any and all specialists, from hematology to infectious disease and beyond. Evaluations typically last from two to five days. Services, including some travel and lodging expenses, are provided at no cost to the child and family. Children are often hosted at the Children's Inn at NIH [<http://www.childrensinn.org>]

Unfortunately, only a small number of cases are accepted per year. Hopefully its funding will increase over time, allowing for more patients to take part in the program.

For more information on the program, see
<http://rarediseases.info.nih.gov/Resources.aspx?PageID=31>

If your child is not accepted to the program, there are other resources that may help your child get a diagnosis. Several of these are listed below:

- Syndromes Without a Name (SWAN): <http://www.undiagnosed-usa.org/>
- In Need of Diagnosis: <http://www.inod.org/index.html>
- National Association for Rare Diseases (NORD): <http://www.rarediseases.org/>
- Parent-2-Parent Undiagnosed Forum: <http://parent-2-parent.com/forum/forumdisplay.php?f=61>
- Madisons Foundation: <http://www.madisonsfoundation.org>