

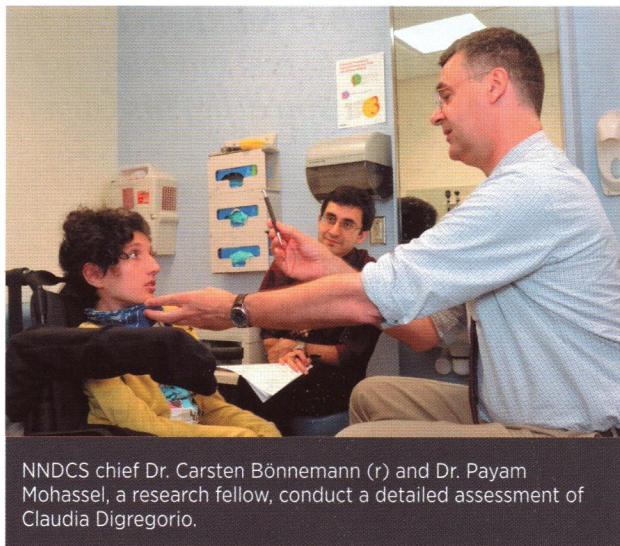
NINDS Intramural Scientists Participate In International Humanitarian Effort

BY SHANNON E. GARNETT

Fifteen year old Claudia Digregorio recently traveled from her hometown in Santaremo, Puglia, Italy to NIH on an important mission. She wanted to find a name and diagnosis for her neurological disorder, learn about possible treatments and, perhaps, even find a cure. Her trip was sponsored by the National Police Defense Foundation's Operation Kids Program.

Digregorio, who has suffered from a rare, yet undiagnosed, neurological disorder since age 5, visited NIH in early September to participate in a clinical research study led by Dr. Carsten Bönnemann, chief of the neuromuscular and neurogenetic disorders of childhood section (NNDCS), Neurogenetics Branch, at NINDS. Digregorio's doctors in Italy have not been able to provide her with a diagnosis or effective treatment.

While Digregorio was at NIH, Bönnemann's NINDS team—including Dr. Reghan Foley, a staff clinician; Dr. Payam Mohassel, a fellow in the Neurogenetics Branch; Sandra Donkervoot, a genetic



NNDCS chief Dr. Carsten Bönnemann (r) and Dr. Payam Mohassel, a research fellow, conduct a detailed assessment of Claudia Digregorio.

counselor; and Christopher Mendoza, a patient care coordinator—conducted a detailed assessment of Digregorio's condition involving a battery of tests and procedures to determine the underlying cause of her disease as well as any potential treatments.

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Before coming to NIH, Digregorio also met with Pope Francis, who pledged his support and prayers for her health.

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Digregorio, who traveled to NIH with her mother and father, stayed at the Children's Inn during her week-long visit. At the inn, Van Mitchell, secretary of the Maryland department of health and mental hygiene, and Francesco Cascione, an NPDF representative, visited the family.

Although it's fairly common for children with rare, undiagnosed neurological illnesses to visit NIH for help—directly at NINDS or through the NIH Undiagnosed Diseases Program—Digregorio's journey was somewhat unconventional. Her story first came to the attention of the NPDF through a YouTube video that “went viral” asking internationally for help “to find a diagnosis for Claudia.”

As a result of the video, the NPDF contacted their counterparts at the Italian Police Defense Foundation and agreed to help Digregorio through the Operation Kids program—a congressionally recognized child

safety program committed to protecting children from criminals and life-threatening illnesses. To date the program has arranged eight lifesaving operations for critically ill children worldwide.

The NPDF then worked with NIH, specifically Bönnemann and Foley, to secure an appointment for Digregorio. Before coming to NIH, Digregorio also met with Pope Francis, who pledged his support and prayers for her health.

Digregorio's NIH stay ended with a meeting to discuss early results of her medical evaluations and tests. According to Bönnemann, the NINDS medical team will continue to follow her as part of the clinical study and as further results become available in the ongoing effort to establish her underlying diagnosis. **R**



ON THE COVER: Detail of artwork used in poster for 2011 NIMH Innovation Lecture Series.

IMAGE: NIH MEDICAL ARTS

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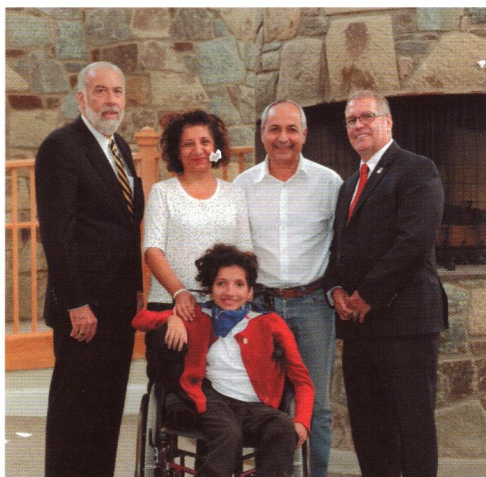
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Van Mitchell (l), secretary of the Maryland department of health and mental hygiene, and Francesco Cascione (r), an NPDF representative, visit the Digregorio family at the Children's Inn.

PHOTOS: ERNIE BRANSON, MICHAEL SPENCER

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