A Family Works to Fill Void
As Research Funding Slows
ALBRIGHT'S THERAPY AT STAKE

In his pint-size white doctor's coat, Adam, 4, hands over a check to help fund the research that will help him grow up. In return, he gets a hug from his physician, **Emily Germain-Lee**, in the Pediatric Clinical Research Unit at Hopkins Children's.

The $40,000 check represents the fierce dedication of his parents and supporters to ensure that endocrinologist Germain-Lee can continue the nation's only study of a new treatment for Adam's rare genetic disorder, Albright's hereditary osteodystrophy. In an era of decreasing governmental research funding, private funds are increasingly critical to bridge the gap in translating laboratory findings into actual treatment. "We don't want to risk losing her," says Adam's mother, Debby*. "Without money, she will not be able to continue this important research beyond October, when the current funding ends."

Adam was diagnosed with Albright's at 5-months, after one of its telltale symptoms—bits of bone under his skin—led the family to a dermatologist, a bone expert and eventually to former director of pediatric endocrinology at Hopkins Children's, **Michael Levine,** now at the Children's Hospital of Philadelphia. Albright's is a hormonal resistance syndrome in which a gene key for hormonal activity is defective. When critical hormones fail to function, patients develop multiple biochemical abnormalities, along with formation of bone under the skin and premature bone fusing. Two of the nation's few experts in the field, Levine and Germain-Lee speculated years back whether children with pseudohypoparathyroidism Type 1A, a subset of Albright's, might be growth hormone deficient and whether replacement therapy could prevent the weight gain and short stature caused by this deficiency.

Knowing that his former colleague was studying this hypothesis in children, and that Adam had Type 1A, Levine referred the family to Germain-Lee. Since he's been in her care and on growth hormone, "Adam has been doing so much better," says Debby. "His muscle tone, movements and speech are improving at a faster rate."

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Adam greets Emily Germain-Lee, M.D., with a check.

Because children with Albright's stop growing around the ages of 10 or 12—when their growth plates fuse prematurely. The goal, says Germain-Lee, is to give them the injections while they're still growing. "If it were not for this study, Adam's diagnosis of growth hormone deficiency and subsequent treatment may not have occurred," she says.

To boost the therapy's positive effects and keep his weight down, Debby monitors Adam's diet. Cookies, cake and ice cream are not on the menu. "He doesn't miss what he's never had," she says.

Tireless supporters of Germain-Lee's endeavor, she and her husband, Marc, a New York businessman, have organized a benefit concert in Italy, held raffles and developed teams of volunteer fundraisers with the goal of raising the $100,000 needed to continue the treatment study. "Emily is the nicest human being on the planet," says Debby. "She's brilliant, devoted and tireless. What we're doing to keep her research going is nothing compared to what she's trying to do for us and all the families with Albright's." ■

**No last names used because family wishes to remain anonymous.*

For more information on the study and how you can help, visit **www.jhu.edu/egermainlee.**