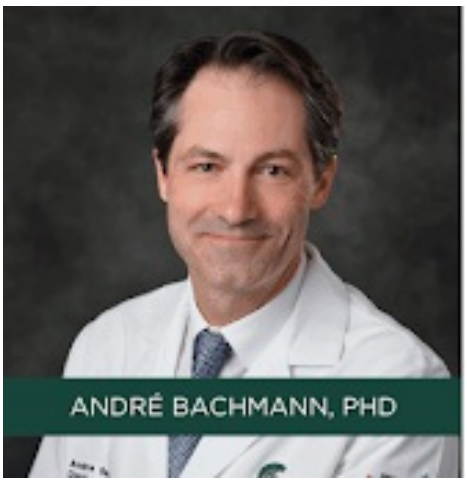
Unraveling BABS: A Rare Disease, A Remarkable Discovery From Michigan State University And Corewell Health

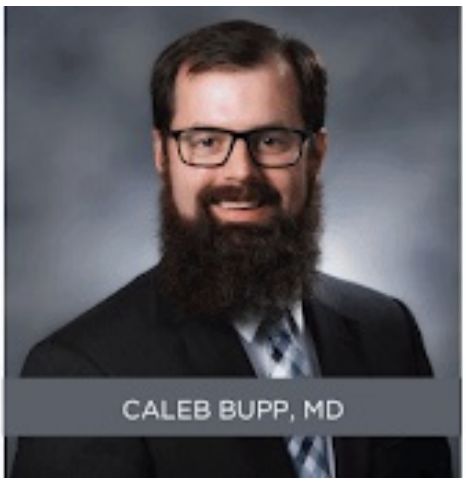
Corewell Health
Michigan State University

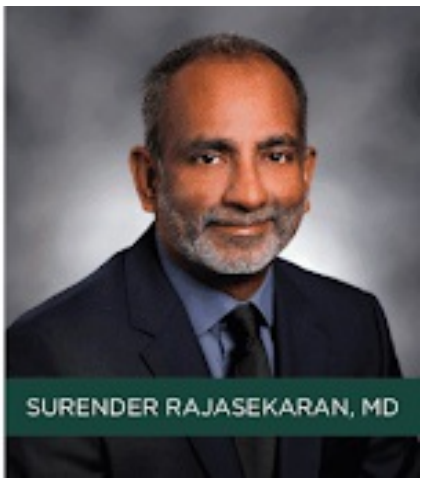
Living with rare diseases can be extremely difficult and isolating not only for the patient but also for their families. There may be very little information available about the condition, and possibly few, if any, treatments that exist. This is the situation Marley's family faced when their three-year-old baby girl was diagnosed with an ultra-rare pediatric disease now known as Bachmann-Bupp Syndrome (BABS).

The disease was discovered and named after a physician-scientist duo: Dr. André Bachmann, a professor at Michigan State University (MSU) College of Human Medicine, and Dr. Caleb Bupp, a board-certified medical geneticist with Corewell Health® and Helen DeVos Children's

Hospital (HDVCH), and an Assistant Professor at MSU. They eventually discovered its treatment along with MSU Assistant Professor Dr. Surender Rajasekaran, DeVos Endowed Chair for Pediatrics with Corewell Health and HDVCH. The repurposing treatment involves oral administration of the drug Eflornithine (DFMO) and is claimed in a U.S. patent owned by MSU and Corewell Health (formerly Spectrum Health).







In 2015, Marley was born with health issues that baffled doctors and her parents. She suffered developmental delays, couldn't regulate her temperature or blood sugar, and strikingly lost all her hair, including her eyebrows and eyelashes. Marley's genetic testing revealed that a gene involved in the polyamine pathway (known as *ODC1*) had a mutation with devastating effects. A series of scientific discussions led MSU inventors to connect some critical biochemical dots, and Eflornithine was administered to Marley. Within six months of her treatment, Marley's hair, eyebrows, and eyelashes had grown back. She was able to sit up by herself, scoot around, trade high fives, and has recently been reported to have tried sledding and made her first attempts at walking.

Since Marley's story was published, the number of documented BABS cases have risen to twelve, and the success of Eflornithine in three other treated BABS patients has spurred additional research into BABS and other rare diseases.

Additionally, the discovery attracted interest from pharmaceutical companies operating in the rare disease space. In 2023, MSU and Corewell Health announced the execution of license agreement with Orbus Therapeutics Inc., a privately held, late-stage biopharmaceutical company focused on developing and commercializing therapies that treat rare diseases. "We are delighted to enter into this agreement with Orbus Therapeutics," said Anupam Jhingran,

Technology Manager with MSU Technologies, the technology transfer office of MSU. “We believe that Orbus Therapeutics' unique strength in developing new treatment options in the rare disease space and their current clinical development of eflornithine makes them the ideal partner to move this effort forward." The license grants Orbus exclusive rights to intellectual property related to the method of use of Eflornithine in the treatment of BABS.

In June of 2023, the New York Intellectual Property Law Association (NYIPLA) awarded the Inventor of the Year Award to Bachmann, Bupp, and Rajasekaran praising them “for your valuable contributions to medicine, life sciences, and human health, and for your appreciation for and support of the U.S. Patent System”.

Further testament to the importance of this discovery, Dr. Bachmann and his colleagues at Corewell Health secured a multi-institutional, five-year NIH/R01 grant award which provides more than $4 million to further study the rare BABS genetic condition and other polyamine-related genetic disorders.

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