



Neal C. Larson / Dispatch

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Dublin girl refuses to back down from genetic childhood disease

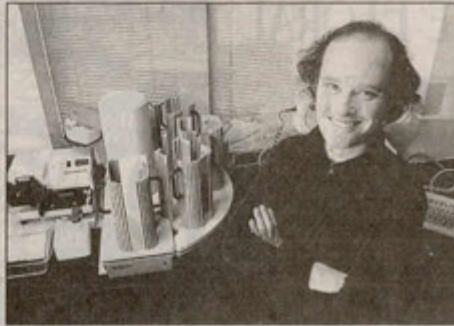
OSU researcher has hopes lifted by breakthrough

By David Lore
Dispatch Science Reporter

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Chris Russell / Dispatch

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Both Madison Rose Reed and Arthur Burghes are up against steep odds in their fight against a deadly childhood disease called spinal muscular atrophy.

Both are doing better than expected.

Madison celebrated her third birthday yesterday even though she was born with Type I spinal muscular atrophy, which kills most children before age 2.

She seems to be holding her own against the genetic disease, which affects nerve cells controlling such things as walking and swallowing. Her parents, Carl and Annette Reed of Dublin, are preparing to enroll her in preschool.

Burghes, associate professor of medical biochemistry and neurology at Ohio State University, thinks recent breakthroughs in genetics are giving scientists at least a 50-50 chance of developing a drug to treat the disease. His research is supported in part by the Miracle for Madison Fund established by Madison's parents.

"Maybe within three years we'll be ready for human trials," Burghes said last week.

Until recently, he would have rated chances of developing a treatment drug at only about one in 10, he said. "Now I'd raise that to 50 or 60 percent. In dealing with a disease like this, a 50 percent chance is pretty good."

Burghes is lead author of a report on the research in this month's issue of Human Molecular Genetics.

Spinal muscular atrophy affects about one out of 10,000 children. It is the leading inherited cause of death in early childhood, although it can afflict adults as well.

The disease is caused by the absence of a gene, SMN1, that controls the production of a protein that nerve cells need to survive. The breakthrough, Burghes said, was isolating that gene as well as a mysterious twin called SMN2 through the Human Genome Project.

The structure of the SMN2 "copy" gene is unusual, and the purpose of the gene remains something of a mystery. It's not found in other animals and seemingly evolved only in man.

SMN2 also produces the nerve- cell protein but in much smaller amounts.

Still, by using this backup gene to compensate for a missing SMN1 gene, doctors might be able to head off -- or possibly even reverse -- spinal muscular atrophy in children. The trick, Burghes said, is to find a compound that stimulates the copy gene to increase protein production fivefold or sixfold, the minimum amount needed to maintain nerve- cell function.

"We've got 40,000 compounds in the freezer waiting to be screened," he said.

Madison, however, is on borrowed time because there is no way to reverse the damage to her nervous system.

"Seven months after she was born, when she was diagnosed with this, pretty much the doctors said we should just take Madison home and make her comfortable," her father said. "The meaning of that was clear, but we pretty much refused to accept that."

Mr. Reed said he and his wife, a nurse at Doctor's Hospital West, concentrate on keeping Madison's lungs clear and protecting her from infection. She can't sit up by herself and has lost some strength, he said, but she has gained weight in the past year and has become more animated and talkative.

She has always been mentally alert -- "as sharp as a tack and stubborn as a mule," according to her father.

Madison celebrated her birthday yesterday at Dublin Coffman High School, where her father teaches government and international relations. The student senate sponsored the party.

Madison celebrated her second birthday there, too, and everybody is hoping she'll be back for cake and candles next Feb. 4.

With community support, the Reeds have raised \$130,000 for spinal-muscular-atrophy research at Ohio State through the Miracle for Madison Fund.

Mr. Reed is aware of Burghes' research, he said.

"I'd like to think it could help Madison, but the reality is I'm not sure."

Information about the Miracle for Madison Fund can be found at www.miracleformadison.org.

Caption: (1) Neal C. Lauron / Dispatch

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(2) Chris Russell / Dispatch

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