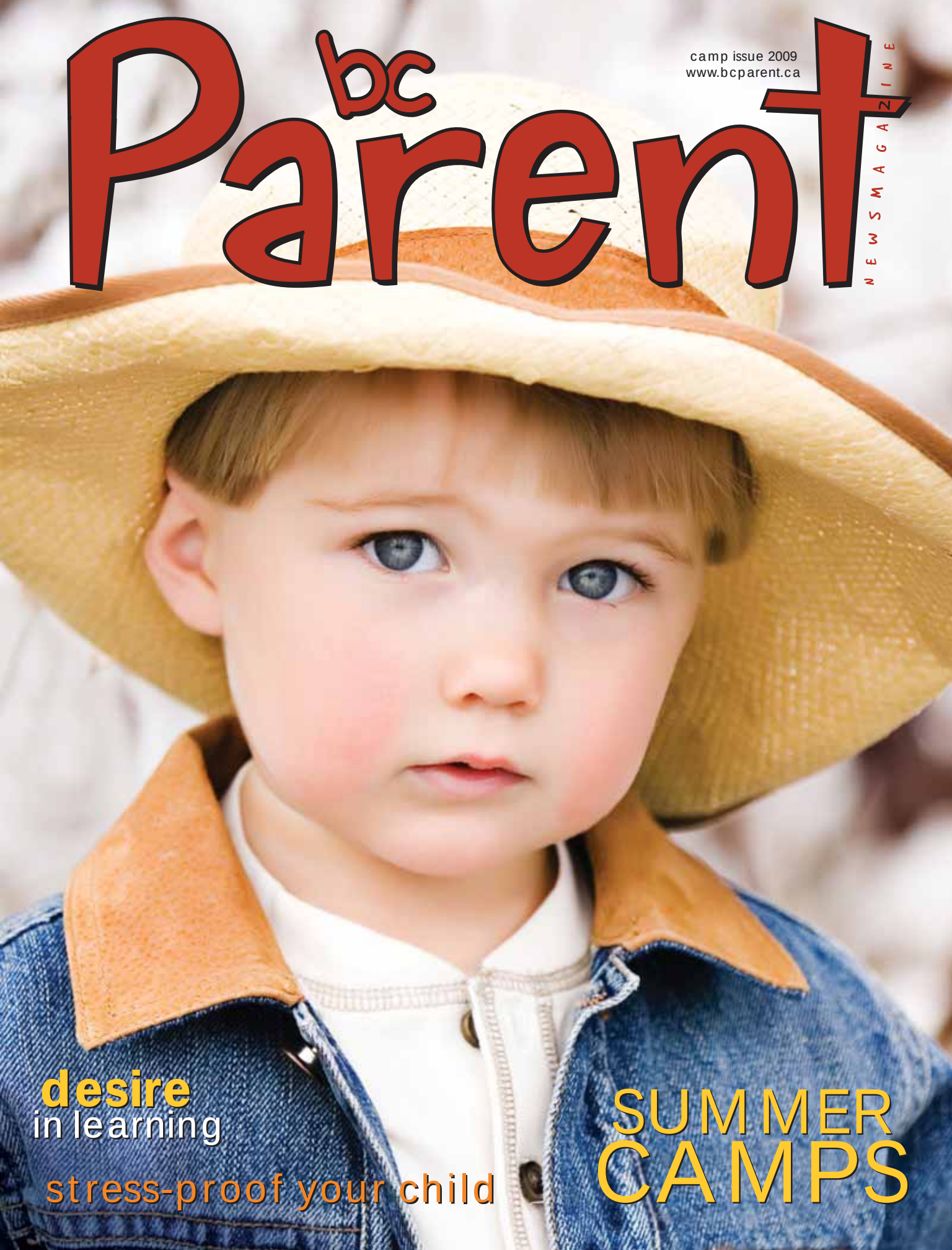
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Against the odds...

Help Give Hope to Families Affected by Rare Diseases

by Deb Purcell

"A rare or 'orphan' disease affects fewer than 1 in 2,000 people. However, there are more than 7,000 rare disorders that, taken together, affect approximately 3 million Canadians. One in ten individuals in Canada have been diagnosed with a rare disorder." —www.cord.ca

I had no idea about any of this until my son, Trey, at the age of two, was diagnosed with a rare disease called Hunter Syndrome (also known as Mucopolysaccharidosis, type II, or MPS II). Many people have not heard of Hunter Syndrome as it is very rare and affects less than 30 people in Canada, however, more well known rare diseases include Tay Sachs, Huntington's, Cystic Fibrosis, and Legionnaires Disease. Chances are, with a statistic as high as 1 in 10, if you or your family is not affected by a rare disease, someone you know is.

Although one in ten Canadians is affected by a rare disease, Canada has no federal plan or policy to deal with orphan diseases and their drugs. Canada is the only developed country in the world that does not have an Orphan Drug Policy (ODP). What is an Orphan Drug Policy and why do we want one? An ODP is a policy that would separate the laws and requirements for rare disease drugs from more common disease drugs. This separation is necessary for many reasons. Since its implementation 25 years ago, the Orphan Drug Act in the United States has provided researchers and companies incentives for developing rare disease drugs so that over 300 drugs have been developed for rare diseases, many of which are life saving.

"Companies in the US are given tax breaks, help with FDA submissions, fast-tracking through the approval process, and market exclusivity as incentives to develop drugs to treat those with rare or 'orphan' diseases. Without these incentives, companies would continue to focus on drugs for common disorders, with a much larger market, instead of novel treatments for the under-served," says Kirsten Harkins, executive director of The Canadian MPS Society. "An ODP in Canada is necessary, not only to provide industry incentives to promote research and innovation, but to provide patients with access to drugs once they are developed and reach the market."

In our situation, the lack of an ODP in Canada meant that Trey could not access the only available (and life saving) treatment for Hunter Syndrome—Elaprase, a drug that had already been proven safe and effective by the FDA, European Union, and other countries—because of where he lived.

Currently, our system caters to treatments for common diseases. Drugs are assessed for safety and efficacy by Health Canada and, once licensed, are passed onto the federal Common Drug Review (CDR) to assess "cost-effectiveness" and recommend to the provinces and territories whether or not drugs should be put on their "public" drug plans. However, because the CDR's framework was developed to deal with drugs for more common diseases, it has recommended that the provinces and territories NOT fund almost every new drug for a rare disease it has encountered, including Elaprase, the drug Trey desperately needed. It is up to each province or territory to decide whether to follow the CDR's recommendation or to fund a drug regardless.

"Because the federal government has not taken the lead on developing a policy or framework for rare disease treatments, some of the provinces have gone ahead and developed their own programs" says Harkins.

Hunter syndrome is a progressive and genetic disease that predominantly affects males. Males who have Hunter Syndrome are missing a vital enzyme that breaks down cellular waste. Unable to break it down, their bodies accumulate this waste, causing an increasing number of symptoms: distinct facial features, a large head, an enlarged abdomen, hearing loss, thickening of the heart valves, obstructive airway disease, sleep apnea, enlargement of the liver and spleen, range of motion and mobility issues, and possible central nervous system involvement. Symptoms and severity vary widely.

This would have been Trey's story had we not gotten him treatment. I began fighting to get Elaprase funded for Trey in Canada after its approval by the FDA on July 24, 2006, which was two months after his diagnosis. I'm sure you can imagine my already completely devastated and overwhelmed emotional state with diagnosis and then being told I would now have to fight for his treatment. Fortunately, we live in BC and although my fight was not what I would describe as easy or tear free, Trey did start ERT seven months after my first efforts to access the drug, on February 5, 2007. He was the first boy in Canada, who was not in a trial, to begin treatment. Other boys and men across the country have not been so fortunate. More than two years after Trey began treatment, only a handful of the dozens of boys and men with Hunter Syndrome are being treated.

Something comprehensive needs to be done. The good news is people who know this are acting. Don Bell, former MP for North Vancouver, is one of these people. On May 7, 2008, North Vancouver Liberal MP Don Bell's Private Members Motion-426 was passed in the House of Commons. This motion addressed the need to create a national strategy for addressing rare diseases. However, Don Bell was not re-elected in the 2008 federal election and his voice for rare diseases in the legislature is no longer heard.

Part of the problem is education and awareness. Most people don't know about our lack of an Orphan Drug Policy in Canada. We can't do anything if we don't know there is something to do. By raising awareness about our situation, I believe we can bring about awareness and change.

Dr. Bill McKellin, is a Board member of the newly formed Rare Disease Foundation (RDF) in BC, chair of the RDF parent group, medical anthropologist and parent of a child with a rare disease. "Most organizations" stated McKellin, like the Canadian MPS Society, "are disease specific. Although beneficial in obvious ways, disease specific organizations cannot always help parents navigate Special Education issues within the school system or access provincial services and tax breaks that are available, in addition to the fact that parents with an undiagnosed child have nowhere to go." The RDF is involved with research, advocacy, and family support and has one goal: "to improve the lives of children with rare diseases."

Our country—one that has always prided itself on taking care of its most vulnerable citizens—is failing people with rare diseases. Hunter Syndrome is progressive. Without treatment, with every second that passes, irreversible and irreparable damage is being done. Most rare diseases affect children and are just as serious and life threatening. With the support of my family, friends and many health care professionals and government workers, I had the confidence to fight for Trey, and the willingness to go public and speak to the media. What about those who can't or don't know how or don't have the support to fight for their rights? ★

Rare Disease Foundation (RDF) in BC

- To help those with MPS II fight for funding of Elaprase in their respective provinces, go to: www.mpssociety.ca and click "Get Involved." A template letter to your local MP has already been drafted. All you have to do is send it!
- To learn more about Trey, MPS II, the research we've funded or to donate, go to: www.treypurcell.com. For information on our upcoming fundraiser, see: www.timeforacure.ca.
- To learn more about RDF visit: www.rarediseasefoundation.org.
- CORD, the Canadian Organization for Rare Disorders, is an organization heading up the fight for a Canadian ODP. To get involved and advocate for those without a voice, you can go to the CORD website, www.cord.ca or www.raredisorders.ca, and send a letter to your local MP.