

# Presentation to Ontario's Citizen Council

January 30, 2010

## Introduction

Good morning, and thank you for the opportunity to speak today. I would like to start by expressing my appreciation to the 25 members of the Citizen Council for volunteering for this very important public service. My name is Jocelyn, and I am here today to speak about my grandson Michael, the challenge of his rare disease not only for himself but the whole family, the miracle of treatment, and what has become our biggest hurdle, access to treatment.

## Background

When Michael was born, he appeared quite normal and even the rash that he had was considered to be quite typical of newborns. But within a few days, his Mom, Karen, my daughter, recognized that his rash and what could be best described as extreme colic were anything but normal. And that was the beginning of our 20-month journey of pain, fear, hurt and hope. When he was 3 months old, Michael experienced such high fevers and joints that were so swollen and red that he had to be hospitalized. Little did we know that this was to become the pattern: regular high fevers, head banging which led to intracranial pressure, chronic vomiting and diarrhea, painful swollen joints, septic infections, hearing loss and eye problems. Michael spent much of his infancy in hospital and shuffling between specialists. The impact on our family was overwhelming, the emotional and financial strain were crippling, and his mom was slowly losing faith that he was going to get better. We hadn't even been given a diagnosis. It would be at a visit to the Hospital for Sick Kids here in Toronto for his eyes when a physician in training said "Oh, so they think he has NOMID." That was the first time we heard of the disease. Armed with the theory of a diagnosis, Karen turned to the internet and found a support group, after a long discussion with another mom in Texas, Karen was given the number to a researcher at the NIH (National Institutes of Health) in Maryland. Within a week, Michael was admitted to the NIH .

## Treatment (Michaels experience)

Michael underwent a week of intensive testing, where the genetic test confirmed the diagnosis of NOMID. We also learned the extent to which the disease had damaged his small body. Michael was under 2 years of age but he already had decreased bone density, hearing and vision loss, extremely high intracranial pressure, delayed bone growth, and bone dysplasia. The good news was that he was accepted for a clinical trial with a drug called Anakinra. The result was nothing short of a miracle! Michael went from being a 21-month-old confined to his stroller, unable to support his head, and unable to walk. Within days of his very first treatment, the rash that had plagued his body since birth, disappeared; he began walking, had full use of his limbs, and could move his head. He laughed again, even danced on the 4th day of his treatment. Now in

his 4th year of treatment Michael continues to thrive and in fact some of the damage has even been reversed. Michael's hearing has remained stable, his bone density has increased, his intracranial pressure has been controlled. He has not been hospitalized since he began treatment; he attends kindergarten and his teachers say he is nearly at par with his peers.

So, what is the bad news? First, Anakinra has to be injected under the skin every single day. Because the drug remains in the body for only 6 to 12 hours, there is not a single day when you can take a break and, even worse, there is not a day when Michael gets a break. Patients with NOMID over-produce a natural chemical in the body called Interleukin 1 and what Anakinra does is suppress that chemical. If we were to miss even a day, the inflammation would reappear and the chain of damages start all over. As you can imagine, daily injections would be difficult and painful for anyone, let alone a six-year-old child. After over 2000 injections, Michael is developing scar tissue and site reactions as well as mental trauma.

It is important to know that Anakinra was not created for NOMID; it was designed to treat severe arthritis, but researchers understood its effect on Interleukin, and so thought it should work for NOMID patients.

BUT, because the drug was already licensed in the USA for arthritis; because it was already being used to successfully treat NOMID, and because the number of patients was so very, very small, the manufactures were not interested in doing clinical trials for NOMID patients. With no clinical trials, the drug would not be licensed for NOMID and without license for NOMID, our drug plans would not approve paying for it for NOMID patients. So we have the additional stress of knowing that our access to the Anakinra, which is still through the clinical trial program at the NIH, may end at any time.

Which brings us to our big hope. A new drug called Ilaris has been developed specifically for NOMID and has been approved in the USA in the European Union countries. Ilaris stays in the body for up to 26 days, so it needs to be given only once every 8 weeks. We understand that the company has submitted it to Health Canada and it may be approved within the next month. We have spoken with families receiving Ilaris in the states, and they are thrilled. What could Ilaris mean for our family? It means that Michael would get a shot six times a year, instead of 365 times a year. It means we won't have to travel to Washington DC every six months to get the drug. It means that we could have a stable life and be free of a lot of mental and emotional stress. And while Ilaris is not a cure, the drug actually works different from Anakinra. It does just suppress the excess interleukin, it actually washes it out of the body. The benefits are as good without the drawbacks.

I honestly don't know whether Michael could have taken Anakinra every day for the rest of his life. As he gets older, the dosage needs to increase. Now that there is an alternative, our hope for Michael is that Health Canada approves Ilaris very soon and, as importantly, that Ontario makes it available through the Public Drug Plan. With

treatment, Michael will likely have a near-normal life. We don't even want to think about what life for Michael would be like without Ilaris.

Thank you for listening, and thank you for caring about patients with rare disorders, like Michael