

Family maintains positive attitude despite devastating disease

At a time in their lives when they should have been proudly cheering on their young sons' first steps, Roger Laforest and Lucie Boudreau were instead sadly puzzled as to why Francis and Simon kept falling with each attempt to walk. It seemed to be a milestone they could not reach.

“When we took the kids to their pediatrician to ask why they could not learn to walk, he suspected Morquio disease right away,” says Roger. “The disease was confirmed after a visit to the genetic department of the Montreal Children’s Hospital of the McGill University Health Centre.”

For Francis, now 16 years old, and Simon, now 14 years old, their diagnosis was just the beginning of a life filled with operation after operation and struggles most other children their age would not face. Morquio Syndrome is a very rare genetic disease. It is estimated to occur in one of every 200,000 births. As with other recessively inherited genetic disorders like cystic fibrosis or phenylketonuria, both parents have to carry the same defective gene to pass the condition on to their child. The chance of having a child with this disease if both parents are carriers is one in four. Roger and Lucie have an older son who did not inherit the disease.

Morquio disease comes in two different forms, but the basic premise is people with this disease lack an enzyme that breaks down certain sugar molecules, therefore abnormally large amounts of that complex sugar build up leading to mostly skeletal involvement in type A and combined skeletal and neurodegenerative involvement in type B. Symptoms usually start between ages one and three and these can include mildly coarse facial features, a large head (macrocephaly), short stature with a particularly short trunk, misshapen hands and feet with hyperextensible joints.

With time, spinal, cardiorespiratory and corneal involvement will be observed. Both boys are about one metre tall. Francis weighs 23 kgs and Simon, 30 kgs, about the size of your average six-year-old. “Simon is currently in a wheelchair because of his bowed-legs, which make it hard for him to walk. He is also a bit heavy now, so his frame can’t hold the weight,” says Roger. “Francis recently had an operation to straighten his legs so he is walking on his own but because he is still growing they will most likely revert back to being bow-legged.”

The boys also have strength and motor problems with their ankles and wrists, which causes difficulty with opening jars, going to the washroom and dressing themselves. Over the past 15 years, together they have undergone 25 operations to correct bone structure, heart problems, eyesight and more. And each time they have an operation they lose 2-3 months of school so they are currently a bit behind. The disease does not affect their mental ability, it just limits them physically. And because they are small in stature their father says they play like 10 year olds, preferring games for smaller bodies. “They play some sports, like hockey in their wheelchairs and they love to swim,” says Roger. “They also play X-box, watch movies and they have an electric tricycle. It goes up to 20 km/hr and Simon usually goes that fast—he is not afraid of anything!”

The boys' orthopedic surgeon, Dr. Francois Fassier of the Montreal Children’s Hospital and the Shriners’ Hospital for Children, has followed the boys since they were diagnosed.

“Unfortunately we treat the deformities so we run behind the problem,” he says. “To prevent the deformities would be fantastic, but that means a type of cure.”

Currently Dr. Serge Melancon, chief of Biochemical Genetics at the MCH, is in the beginning phases of conducting a large scale clinical study that could hopefully lead to participation in the first trial of enzyme replacement therapy ever for Morquio type A disease. “Until there is a miracle drug produced, enzyme replacement should be sufficient in stabilizing its progression,” he says. “Enzyme replacement is not perfect, but it does improve the patients day to day life generally by slowing it down. It isn’t perfect because the enzyme is not able to reach every part of the body that is affected.”

Dr. Melancon is also in the process of trying to set up a special clinic at the MCH for Morquio syndrome to be able to clinically assess patients. “The amazing thing is the boys never cry or feel sorry for themselves. They are so strong despite the fact that their bones hurt them 24 hours a day,” says Roger. “Even the doctors say they don’t know why they don’t complain because it’s a bone disease, but I guess they are just used to it.” Roger says that it helps that the boys have each other. “They could have their own bedrooms but they want to be together,” he says. “When Francis was operated on recently, Simon slept in Francis’ bed. They really are strong guys....they help us cope.”

Francis loves hockey—he would go to every game at the Bell Centre if he could and he tries to never miss a game on TV—and Simon loves to get outside and enjoy life and he jokes all the time. Roger said he and Lucie cried a lot when they first found out about the boys’ disease and asked often “why us?” There is no answer. “It’s life,” he says. “I am happy to have kids like this...it makes me appreciate everything more. I never feel sorry for myself. I just get on with it. My job in life is to parent and care for these boys.”

According to Dr. Fassier, the kids have a great attitude and their father is a miracle. “Roger is not always happy to come to the hospital but he is always happy with what we are trying to do for his children,” he says. “Each one is a hero, because these kids have to live with this disease and the father is fantastic because he is able to cope with the situation and he does it with optimism and with a great attitude.”

Roger knows his kids don’t have a long life ahead of them, or the quality of life he could imagine for them, so he gives 500 per cent to them every day. “I look at it like this,” he says, “tomorrow is tomorrow and yesterday is gone, so today I will do my best.”

Cutline: Simon, left, Francis, right, Roger Laforest (dad), behind.