CDLS experience breeds children's book

BY STEPH WILLEMS
swillems@theemc.ca

EMC News - Ask someone if they know what Cornelia de Lange Syndrome is, and you'll likely be met with a look of puzzlement. That's because the genetic disorder is both rare and frequently misdiagnosed.

Nathalie Wendling and John Glatzmayer of Manotick found that out after spending three years trying to find out what was wrong with their daughter Melanie. The child had many developmental delays, and was, as Ms. Wendling said, "in her own world," displaying symptoms commonly associated with autism. It wasn't until Melanie was three, after numerous trips to numerous hospitals, that a diagnosis of CDLS was made, at which point her parents learned that only 92 cases existed in all of Canada.

Because of the rarity and complexity of the syndrome, Melanie's seven-year-old brother Tommy had trouble explaining to his classmates at St. Leonard Elementary School why his sister, now 10, faced more difficulties in life than they did. Last year Tommy told his mom he wanted to make a picture book that would educate as well as entertain the kids.

"The kids really take care of her at St. Leonard," said Ms. Wendling. "Tommy wanted to bring his friends books about CDLS; we went to the library, Chapters, and got books but they were super depressing. He said, 'let's write our own book,' one that he could give to his friends that wasn't boring or depressing."

The resulting picture book is titled Tommy and Melanie have two pet rats and one syndrome, with professional images provided free of charge by Frank Cava of Photolux Studio. The reference to rats reflects a family tradition (started by John) of having rats as pets.

"They make fantastic pets and are present in almost all of our photos," said Ms. Wendling.

Tommy and Melanie announced the book on June 26, their last day of school. At the back of the book is a list of commonly asked questions and answers, so the kids reading the book aren't left with unanswered queries. Two small videos have also been produced to help create awareness of CDLS.

Living with CDLS means having to endure a number of challenges. Because of its outward similarity to Autism Spectrum Disorder, an accurate diagnosis often takes years.

Symptoms include delayed growth, developmental delays, certain physical characteristics (which vary, but can include missing limbs), severe gastro-esophageal reflux requiring corrective surgery, and seizures.

In her 10 years, Melanie has undergone 11 operations to help her attain a better quality of life. The glasses and hearing aids she received at six years old benefited her greatly, making her more in touch with the people and things around her.

"It was like a miracle," said Ms. Wendling, who added that thousands of men and women who remain undiagnosed have to live without support services, and are misunderstood by those who believe their problems are simply behavioral, rather than a physical genetic flaw that can be traced to an altered chromosome. The actual cause of CDLS was only discovered five years ago.

Ms. Wendling recalled meeting the mother of a 21-year-old CDLS sufferer who didn't know the cause of her son's condition until she saw Melanie's facial characteristics, and put two and two together.

So far the book, which will be available online at www.amazon.ca as of July 19, has been picked up by the Montreal-based Miriam Foundation, which plan to purchase 2,000 copies and distribute them across Canada. The Miriam Foundation (est. 1970) supports services and programs that foster increased socialization and community integration for children and adults living with intellectual disabilities or Autism.

The CHEO Foundation has also agreed to carry the book, with profits being given back to CHEO. The book goes on sale there July 29.

CHEO Foundation president and CEO Fred Bartlett expressed interest in Melanie's syndrome and the book that explains it.

"It's a rare disease," he said. "She's a lovely kid, and we do a fair amount of looking into rare diseases at CHEO."

Mr. Bartlett said he was impressed with the educational aspect of the books, as well as its ability to appeal to children.

PHOTO SUBMITTED

Nathalie Wendling and husband John Glatzmayer are seen with their son Tommy and daughter Melanie, as well as their pet rats. The family has released a informative children's book describing Melanie's experience with CDLS.