Families of children with rare genetic disorder affecting one in every 50,000 newborns aim to share the most up-to-date diagnosis and care information.

**Flagler Beach, Florida (May ____)** – The doctor walked into the examination room and said to the anxious parents waiting inside: “I’m sorry to tell you, your baby suffers from an incurable genetic disorder called Cri du Chat Syndrome. She will never walk or talk. She may have a very short life and you should consider institutionalizing her.”

For the child’s parents, it was like time stood still. They looked at each other, terrified about what may become of their precious little girl and started to panic about what they would do next.

Unfortunately, the doctor, who relied on outdated information from 30 years ago, greatly exasperated one of the most difficult pieces of information these young parents may ever receive in their lifetimes.

“For many parents whose children are diagnosed with Cri du Chat, this is exactly what they have experienced,” says Stephen Furnari, 40, a resident of Flagler Beach, Florida and father to Katie, a three year old with the syndrome. “But, it doesn’t have to be that way.”

Cri du Chat (also called 5p- Syndrome) is a rare genetic disorder caused by a deletion in the “P” region of the fifth chromosome. French for “cry of the cat”, the syndrome is named for the distinctive kitten-like sound that infants affected by the syndrome make when they cry.

The syndrome affects approximately one in every 50,000 newborns, making it quite rare. Even in densely populated metropolitan areas like New York or Los Angeles, it’s not uncommon for medical professionals to have never seen a case or even be aware the syndrome exists.

According to Furnari, Cri du Chat frequently goes undiagnosed. “You have children suffering from unexplained health issues and developmental delays, often for prolonged periods of time. It’s incredibly frustrating for parents who want the best care for their child,” he said.

This is particularly so in cases like Furnari’s where there was a normal pregnancy and the baby was born with no life threatening medical problems. Mr. Furnari’s daughter Katie had a few of minor issues when she was born, including a small skin tag, low muscle tone and a heart murmur. Independently, none of these things were cause for concern.

“In retrospect, there were enough signs,” said Furnari. “In particular, Katie’s kitten-like cry. Any medical professional armed with the most basic information about the syndrome should have noticed and recommended genetic screening.”

Furnari’s daughter was not diagnosed until she was three months old. Her new pediatrician happened to see one case thirty years ago and pieced together the clues that none of the other doctors caring for Katie could. “We were very lucky,” says Furnari, “we know other families whose children went undiagnosed for more than three years.”
The other problem is that once the syndrome is diagnosed, some medical professionals are relying on outdated information about the syndrome. “You would be surprised at how many doctors are still distributing incorrect information from the 1970’s, a time when most children with the syndrome were institutionalized.”

Now, with the help of early intervention programs, including speech, occupational and physical therapies, children with Cri du Chat live with their parents and are expected to live healthy, normal lifespans.

**Furnari** is a member of the 5p- Society, the U.S. based support group for parents and caretakers of family members with Cri du Chat. Starting on May 4, 2014, the 5p- Society is sponsoring the first annual International Cri du Chat Awareness Week, together with other Cri du Chat support groups from around the world.

“Our goal is to share the most up-to-date diagnosis and care information about the syndrome to as many healthcare professionals as possible. We particularly want to reach pediatricians, OBGYN doctors and maternity ward nurses who are the first line of defense when it comes to diagnosis,” said **Furnari**.

“Katie is doing great,” beamed the proud dad. “With the help of the therapies she receives in the ESL program at her elementary school, Katie is very close to walking independently and her vocabulary of words and ASL signs continues to grow each day. Her mom and I couldn’t be happier.”

**About The 5p- Society and International Cri Du Chat Awareness Week**

The 5p- Society (five p minus) is the support group for families and caretakers of children affected by Cri du Chat Syndrome (also known as 5p- Syndrome or Cat Cry Syndrome) and a co-sponsor of the First Annual International Cri du Chat Awareness Week. The Society encourages and facilitates communication throughout the Cri du Chat community and spreads awareness and education about the syndrome and treatment best practices for those affected by Cri du Chat Syndrome.

For more information about International Cri du Chat Awareness Week, including information for medical professionals about diagnosis and treatment of the syndrome, visit [http://www.criduchat.org](http://www.criduchat.org).

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