Exceptional Family TV

The One In A Billion Genetic Condition

By Nathan Charlan

Most parents consider their children "one in a million." It may be cliché, but they are each special and unique.

For one exceptional family from St. Louis, MO, they discovered this parental cliché actually went deeper than they ever could have imagined. Their daughter was more like "one in a billion."

Even though all of us and our children are each individually unique, we all find common ground making us "just like" a particular group of people. With our children's conditions, or special needs, we often can find many others to connect with who also have the same conditions like cerebral palsy, or Down syndrome, or autism. When it comes to genetic disorders, the commonalities can often be fewer and far between.

For Todd and Michelle Oswald, they've only found one other person alive on the planet who has the same genetic condition as their daughter, Katy.

Michelle gave birth to Katy on March 5, 2005. Although in good health, both Katy's hips were dislocated due to the stress of a breech position in the womb. After braces, surgery and three long months in a cast stretching from belly button to ankles, Katy's doctors expected her to develop normally, albeit slightly delayed, and learn to roll and crawl.

"We were still working under the assumption, at this point, that Katy was just delayed due to her surgery and the cast that kept her immobile for three months," Todd said.

But Katy did not learn to roll and she did not learn to crawl. Todd and Michelle became concerned. At one year of age, Katy was enrolled in an early intervention program and began speech, physical and occupational therapy. She had difficulty eating, was non-verbal, had no mobility and could not pick up small items without help.

After two more years involving an erroneous diagnosis of cerebral palsy, then an abnormal electroencephalograph, and still no answers and no correct diagnosis of Katy's condition, her neurologist finally ordered a major blood test to determine if there was some sort of genetic deficiency.

"Test results confirmed Katy suffers from a deletion on chromosome 6," Todd said. "Her official karyotype is 46,xx, del (6) q13q14.2. What this means in simple terms is that a part of chromosome 6 has

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been wiped out between sections 13 and 14.2. What this means in terms of Katy's future, no one really knows."

Todd and Michelle were sent home with no information, no prognosis, no direction on where to go and what to do. As any of us 21st century parents do when learning about a diagnosis for the first time, the Oswalds turned to the Internet and searched, and searched, and searched.

"There's just no information out there. You're used to being able to type in a question and get 50 million answers to it. With this, you can't find a single one. It threw me for a loop," Todd said.

After hours upon hours of researching the Internet in every way possible, the Oswalds were able to contact one other living person with this exact defect. Through two years of extensive searching through medical papers going back to the 1970s, they believe they've found three others in the world with the exact condition.

"In short, we know basically as much about Katy's condition as her doctors do," Todd said. "It gets more than difficult. It gets frustrating. There's just nothing for us to know what to do."

Today, Katy is five years old. While she still cannot walk or talk, she continues to reach developmental milestones. She communicates by sign language and Todd and Michelle estimate she knows between 500-600 words. The Oswalds have her enrolled in physical, occupational, speech and music therapies through an Early Childhood Development Center. Katy also thrives with swimming, music and horse therapy. Recently, the Oswalds filmed Katy walking for the first time in the pool.

Every therapy, procedure or treatment all have unknown benefits for Katy's genetic condition.

"Everything for her is experimental," Michelle said. "So the insurance companies don't want to pay for anything. We don't know if it will work, but we're not going to stop trying. You have to fight for every little thing that you try to do."

The Oswalds' journey with Katy's future is a constantly evolving path of discovery. There's no medical history for Katy's genetic condition, no historical basis for which to map out a possible future. According to Todd in his writings on Katy's story, "it's both a blessing and a curse to have no historical basis to go on. On one hand, we have no preset stereotypes of what she can or can't accomplish; but we also don't know what will work best."

Although Todd and Michelle may not have a medical history for what will work best, they are constantly on the move to make sure Katy has the best life possible. So, while the parental cliché holds true that Katy is definitely "one in a billion," she has a set of exceptional parents who also fit the bill.

To watch the full story on the Oswald family and their daughter Katy, visit Exceptional Family TV [www.exceptionalfamilytv.com] to watch their family episodes. •

Nathan is father to Zachary, who has spastic quadriplegia cerebral palsy, husband to wife Renee, and the host of an online web-TV series, *Exceptional Family TV*, covering the stories and personal journeys of exceptional families.