Expert Opinion on Orphan Drugs

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EXPERT OPINION

Knowing more about Noonan, by Samantha Scheer

Katie Thortenson of Chicago is a proud mother of two healthy sons: her eldest, Jake, is 6, and her youngest, Drew, is 2, but often mistaken to be younger. When people ask why he's so small, Katie explains that he has Noonan syndrome (NS). "Most people have no idea what it is, so it's important to raise awareness whenever I can".

NS is an autosomal genetic disorder, caused by mutations in one of several different genes. The mutation is caused either through inheritance of a dominant gene or is spontaneous. The syndrome affects physical appearance in a variety of ways, including distinctive facial features, a webbed neck, sunken sternum, low posterior hairline and short stature. NS can also affect development, vision and bleeding. Around two-thirds of people with NS also have congenital heart defects. It is considered a rare syndrome, affecting only 1/1000 – 2500.

Drew was diagnosed with NS before he was even born. "At 12 weeks, doctors knew something was 'wrong'", Katie explained. After a series of genetic testing, and 7 weeks of uncertainty, it was determined that Katie's child was going to be born with NS. In this case, the genetic mutation was spontaneous, and not passed down from a parental gene.

Drew was born with two heart issues: the first, atrial septal defect, characterized by an opening in the wall separating the left and right atria, and the second, mild pulmonary valve stenosis, characterized by an obstructed flow from the right ventricle to the pulmonary artery. Drew was experiencing 'failure to thrive', meaning that his rate of growth was much below that of other male infants of his age.

Nothing seemed terribly wrong with Drew when he went in for a routine echocardiogram. Luckily, the technician noticed shadowy spots in his lungs that would later be identified in a chest x-ray as food particles; Drew was experiencing pulmonary aspiration. One week later, Drew lapsed into respiratory distress. He was placed on ECMO life support for 7 days at the Lurie Children's Hospital in Chicago. "We thought we would lose him", Katie reflected. "It was a miracle that he pulled through that".

Drew stayed at the Lurie Children's Hospital for 3 long months. He was placed on a nasogastric tube for feeding, to prevent issues arising from his underdeveloped lymphatic system. The medical staff quickly realized how sensitive and fragile a child could be with NS and operated with the utmost patience and caution. While Drew's time in the hospital was difficult, it also provided the family with an invaluable source of information about NS and how it specifically affected Drew. "We are so grateful for the doctors and technicians that helped our son. This experience might even pave the way for other children with severe complications from Noonan disorder at this hospital".

However, Drew's hospital stay only marked the beginning of his family's education about NS. It is challenging to find help and information about this rare syndrome, because as Katie said, "most doctors just don't know". To get her son the most expert medical care, Katie sought out the Noonan Syndrome Foundation (NSF). "I just met Dr. Noonan herself last July", said Katie. Every 2 years, the NSF hosts a conference; this year, it will take place in Clearwater, FL. "I got to sit down with a group of doctors who are well versed on Noonan, with Drew's medical chart. It's been an invaluable help".

Along with the NSF's expert medical panel, and Drew's knowledgeable pediatrician, Katie found a wealth of information from other parents of children with NS.

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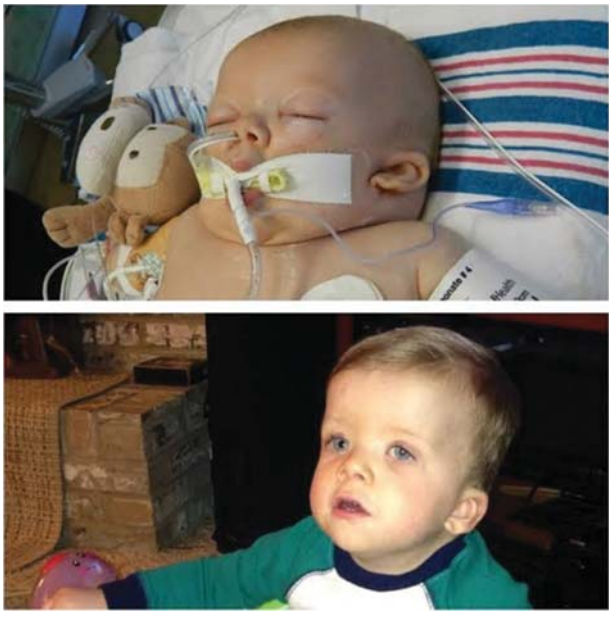
"I've learned so much; for example, a parent told me they'd started their baby with Noonan syndrome on growth hormone already", Katie said. "I had no idea you could start that so early." Sure enough, after a visit to the endocrinologist, Drew started the treatment as well. "He's been on it for two months, and has already grown 2 inches", Katie exclaimed. "It's truly amazing".

Katie's desire to spread knowledge about NS led her to join the Board of Directors at the NSF. "It's nice to know there are people out there, going through exactly what you're going through", said Katie. The NSF aims to foster support groups, but also to raise awareness; Katie is similarly passionate. "Parents don't want to find anything wrong with their child… they want them to be perfect. But sometimes, that doesn't work out". With earlier diagnosis of NS in children, and a wider knowledge base among the medical community, necessary precautions can be taken, appropriate treatments can be administered and lives can be saved.

Now, Drew is a happy and healthy 2-year-old. He is still on a feeding tube and has eight specialty doctors alongside therapy, but his life is becoming less encumbered by medical emergency. "A while back, one family told us that things get easier", Katie said. "I'm finally starting to understand; some months are harder, but we're finally coming towards normalcy".

To learn more about NS, please visit The NSF at www.teamnoonan.org, or the full report of NS at the National Organization for Rare Disorders https://www.rarediseases.org/rare-disease-information/rare-diseases/byID/412/viewFullReport. For NS online support group, please visit www.facebook.com/groups/124712997720295/.



Top: Drew at two months old, on ECMO life support at the Lurie Children's Hospital.
Bottom: Drew right before his second birthday.