



ygnet



The newsletter of
Swan USA

**Our Swan is disjoined, like many families of children who have a
Syndrome Without A Name
Help us make that Connection**

Summer 2006

Issue #4

Better Late Than Never! SWAN USA Update

As I'm sure you all are aware the newsletter is very late. I won't go into detailed excuses, as I'm sure you all know how life gets sometimes. Please accept my apologies.

I will take this opportunity to let you all know what is happening with SWAN USA. We have brought a few people together for a board. Building a board will allow SWAN to become a not for profit organization. This in turn will open the door for many opportunities for our families. We have many great goals for SWAN. The most important goal is getting the word out about SWAN. This will help us find families, just like ours, we all know how scary it can be in that world of the unknown with out any support. A few other goals are building a database of our children's symptoms and networking with the medical community. Accomplishing these goals will take a lot of patients, time and energy. Our goals will also involve some costs, hence the importance of becoming a not for profit organization. If any of you would like to donate, there is a form on the back of our newsletter.

I will continue to keep everyone updated on the structuring of our undiagnosed community.

Your Friend,
Amy Clugston

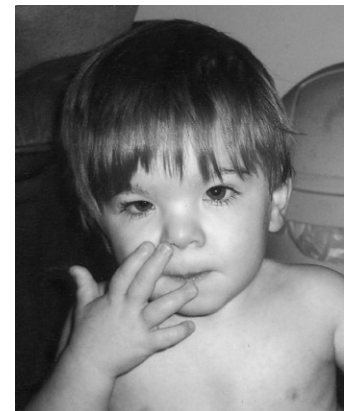
NIH Study

The big conversation on the undiagnosed e-mail group is a NIH Study. Kathryn from our e-mail group has been telling many of us about this study for a few years. It wasn't until recently that many families have taken the steps needed to get into this study.

The name of the study is **Studies of Children With Metabolic and Other Genetic Disorders**. The purpose of the study is to give evaluations to patients with known or suspected metabolic and genetic disorders.

There have been different outcomes for each of the families involved in this study. One family didn't get a diagnosis, but was able to rule many things out. Another family was told they may have no new information to help but they would be willing to see the family anyway. Yet another was given the suggestion of a highly qualified doctor that is closer to the family. Several others are still in the process of being reviewed and tested.

The one family that has given many others hope, is Penny and her son Brison. They made their way to Bethesda Maryland, and on May 30th they were given a clinical diagnosed of Rubenstein Taybi Syndrome. Families involved in this study have mentioned how wonderful Dr. Raygada and her team are.



For more information on this study call 1-800-411-1222 or visit http://clinicalstudies.info.nih.gov/detail/A_2002-CH-0023.html

Getting A Diagnosis for Shiv!



BIRTH: Normal unrelated parents of Indian descent & normal sibling sister, age 9. Pregnancy/delivery normal/vaginal (4 hour labor, mother age 34). He was fine at birth but was a bit gurgly when he breathed. Also had severe reflex & slept for long periods of time.

5 MONTHS: Admitted to INOVA Fairfax Hospital for respiratory distress associated w/ RSV. Was intubated w/ O2 at max setting, was given 3 heart medications & other meds via central line-IV (dopamine, epinephrine, norephrine). Initially they had to give him his IV through his bone, because they couldn't find a vein. His heart slowed down to the point where he required chest compressions. This happened twice in a 1 hour period. He was given a 30% chance of survival by the PICU. He was then flown to Children's National Medical Center for possible Extra-Corporal Membrane Oxygenation (ECMO), but when he reached there, they took new x-rays & compared them w/ the ones from before his 20 minute helicopter journey & there was less white-out in the lungs, so they held off. He never had to have this procedure, but remained intubated for approximately 3 days. He developed hypertension due to micro-embolization of the renal arteries. His brain CT scan was unremarkable & after a total of 3 weeks, he was released from the hospital on GI meds (Zantac, Reglan) & High Blood Pressure meds (Captopril, Clonidine).

He continued breast feeding & later started eating solids. He was also started on weekly Physical Therapy due to his hospitalization & was slightly delayed on his milestones, but his sister was also slightly delayed w/ her milestones as a baby & it didn't seem to be a problem.

7 MONTHS: He was again hospitalized at Fairfax for retching & refluxing a small amount of coffee ground emesis. It was determined during this 5 day stay via endoscopy that he had a duodenal ulcer & esophigitus & was treated w/ Carafate. He was also weaned off of his Clonidine. A brain MRI was also done at this time, due to delayed milestones & hypotonia, & it was fine.

8 MONTHS: He was diagnosed w/ Asthma.

11 MONTHS: He was hospitalized at Fairfax for 3 weeks w/ double pneumonia. After being given a PH probe test, it was determined that he was silently refluxing (no longer visible) 157 times per day. He was also given a swallow study & it was determined that he had delayed swallowing & tended to pool. He also had developed slight ptosis, so was given a Tinsilon test for Myasthenia Gravis but tested negative. He was then operated on & given Nissen Fundoplication surgery, as well as a Gastrostomy tube (G-tube) & NPO from this point. At this time, a rectus abdominus muscle was removed & sent to CNMC & Buffalo, NY to be tested for mitochondrial disorder, but came back negative. Another brain MRI was done, as well as an EEG which were unremarkable.

15 MONTHS: He learned to get into a sitting position from a lying position on his own. One day, we found him in bed grey & unresponsive w/ mucous on his face. We suctioned out the mucous w/ a bulb syringe & administered CPR. Although he was still unconscious, he started slight breathing again. The paramedics took him to Fairfax hospital & he had pulmonary edema & was intubated. He was flown to Johns Hopkins Hospital & after a sleep study & bronchoscopy, it was determined that he had tracheo/laryngo malacia & obstructive sleep apnea. He was extubated after a few days but remained there for 3 weeks. He was sent home w/ a Bi-PAP machine w/ mask. During this hospitalization he was weaned off of his Captopril & no longer had hypertension. He was also given an EMG test that was also negative. He was mistakenly administered a dose of Respiridol instead of Robinol & was unresponsive for 48 hours. An EEG was given & it was normal.

17 MONTHS: I walked away from him for a few minutes & when I came back I found him unconscious. I performed CPR on him & although he was still unconscious he started breathing again. The paramedics took him to the hospital it was determined that he needed a tracheostomy. The surgery was performed & he was sent home after 3 weeks.

18 MONTHS: He started pulling to stand & saying some words. He again went into a quick respiratory arrest & was resuscitated at home. This time when the paramedics took him to the hospital, a bronchoscopy was done which revealed bronchmalacia as well, therefore it was determined that he should be put on CPAP w/ Oxygen 24 hours a day. A Holter monitor was also done & was fine (an EKG was done previously which was also fine).

28 MONTHS: He passed a swallow study, which he had every 6 months until he was 3 & he was Okayed to have 1 Tablespoon of thickened purees while on CPAP. Later, he was allowed to use a Passey Muir Speaking Valve several times a day & weaned off of oxygen after several months. He was on CPAP, pressure 7 (down from 10) when asleep w/ no O2, unless sick. He was evaluated by NIH & they could not find anything. He has had an MR Spectroscopy, CSF spinal tap, bone age, & many DNA (mitochondrial, Rhett's, Angelman's), metabolic, & genetic blood (Ataxia Telangtasia, Acanthocytosis) & urine tests which were negative. Dr. Kelley at Kennedy Krieger found him to have Neutropenia, based on his lab reports.

3 YEARS: His EEG showed brainwaves consistent w/ that of a 12 to 14 month old, but they were trying to organize. No seizures were detected. Also, his TIBC & UIBC were elevated. A sleep study was done for Periodic Limb Myoclonus of Sleep, which was negative. Around this age, he went to the ER for pneumonia.

3 ½ YEARS: He started having apneic incidents. After ambu-bagging him, he regains consciousness & has seizures in which his whole body shakes for about a minute. Sometimes, he has large stools during these episodes. The Neurologist isn't concerned about the seizures, as his previous EEGs were negative, the first seizure was before he started Sinemet (for dystonia), & they are always related to hypoxia.

4 YEARS: He suddenly stopped breathing when he was awake. He did not become conscious immediately after ambu-bagging as usual, but he did start breathing after 1 hour. EMS/ER was not able to get him to take his own breaths though he regained consciousness, he didn't take his own breaths until 2 hours later, so he was put on a respirator. They concluded it was brain-related. He is now mostly on a ventilator w/ 1-2 liters of O2. He does get off of it & stays on 2-3 liters of O2 for up to 3 hour periods. Since then, he has continued to have apneas, sometimes while on the vent – but these days he comes back after ambu-bagging. He sometimes has a seizure afterwards. He has turned both grey & blue.

He started retching. He had a swallow study, upper-GI, & PH-probe test. He aspirated on the swallow study & therefore is NPO & the upper-GI & PH probe were negative. The doctors are assuming his Nissen surgery is loose, but not loose enough to warrant surgery again. Meanwhile, he is on Prevacid.

5 YEARS/CURRENT: He is severely globally developmentally delayed (more like a 6 month old). He started posturing 2 years ago & can no longer get into a sitting position on his own nor pull to standing. He also

used to point to my face parts when I asked him, but no longer does this. He no longer picks up toys nor holds toys on his own. He is average in height & weight but 5th percentile on head size (which has dropped). He also has Broncheictisis. His developmental/neurological health seemed to have started regressing gradually at age 2, but his physical health & immunity improved at this time. His hearing & eyesight had been evaluated & were normal, except for intermittent exophoria in the right eye.

In March of 2005, we took him to Florida, courtesy of Make-a-Wish Foundation, where he must have gotten a mucous plug & went apneic. He was hospitalized at Arnold Palmer Children's & then got tracheitis & pneumonia w/ fever. He was transferred to INOVA Fairfax Hospital PICU (his regular hospital) via air-ambulance & discharged after a total of 10 days (both hospitals). At this time, he was seen at Fairfax by Dr. Virginia Elgin's (his Neurologist) new partner, Dr. William Young for the first time. He asked me if he had ever had a skin biopsy for Batten Disease. I said no. Dr. Elgin & Dr. Amy Lewanda (his Geneticist) found out that there was a new blood test to test for this at Massachusetts General Hospital. **In July we sent his sample there & in October we were told he was positive for CLN6 mutation (Variant Late Infantile Neuronal Ceroid Lipofuscinosis).**

In April 2005, he was taken to the ER for stomach distension & fever that wouldn't go down. He was discharged after 3 days w/ a diagnosis of pneumonia & perhaps some stomach or respiratory virus associated w/ diarrhea.

By Suneeta, Shiv's mom

Planning for the Future

Download a free letter of intent guide. You will need to give your name and e-mail address to subscribe to their newsletter. It will then bring you to a page describing their book. The link to download the free guide will be at the top.

<http://specialneedslegal.com>

SWAN USA Contact Information

Amy Clugston
1745 Lorna Lane
Otsego, MI 49078
269 694 6061 amyclugston@undiagnosed-usa.org
<http://www.undiagnosed-usa.org>
