Social Media: A Rare Disease Lifeline for Patients and Their Caregivers

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Microsoft eyes patient engagement, emerging care scenarios for HIMSS16

The software giant is working to tie together products including Skype, Windows, Office, Surface Hub and Band to connect doctors with patients.

By Tom Sullivan | January 14, 2016 | 03:53 PM

The top 50 social media influencers in orphan drugs and rare disease

With over 36,000 tweets, and 3,500 followers, Ben’s Friends is a great handle to follow for information on rare disease patient communities and support groups.
GRIN1  glutamate ionotropic receptor NMDA type subunit 1 [Homo sapiens (human)]

Gene ID: 2902, updated on 14-Apr-2016

Summary  The protein encoded by this gene is a critical subunit of N-methyl-D-aspartate receptors, members of the glutamate receptor channel superfamily which are heteromeric protein complexes with multiple subunits arranged to form a ligand-gated ion channel. These subunits play a key role in the plasticity of synapses, which is believed to underlie memory and learning. Cell-specific factors are thought to control expression of different isoforms, possibly contributing to the functional diversity of the subunits. Alternatively spliced transcript variants have been described. [provided by RefSeq, Jul 2008]
We Have a diagnosis: GRIN1

Laura took a deep breath and mustered up the courage to call the neurology clinic.

Families of sick children were supposed to wait patiently. The clinic nurse had made this clear eight weeks earlier when Laura first called to check up on Bryson’s lab results.

But parents quickly learn that it pays to be pushy. And what was supposed to be a four-month wait had stretched to half a year of waiting to find out if Bryson had tested positive for a degenerative disease that would prevent him from reaching adulthood. So Laura ignored the ‘don’t call us; we’ll call you’ directive and dialed again.

“The results still aren’t back,” the nurse said. “We’ll call you when they are.”

When Laura pressed, the nurse reluctantly agreed to check on the file. A few minutes later, the nurse returned to the phone to sheepishly acknowledge that an error had been made. Bryson’s blood was never sent to the US lab for testing.
Christina said:

March 1, 2016 at 11:44 pm

Keith,
I am so happy to have found your blog! My husband and I have 2 year old triplets, all girls. Two of them are identical twins and there's a fraternal sister as well. They were born at 32 weeks and the twins have had severe delays since birth. The fraternal sister has developed normally. They did the micro array testing on the twins when they were a few months old, but nothing came back. For two years we have been in and out of doctor's offices, but no one was able to give us an affirmative diagnosis or explain the cause of their delays. They simply labeled them as having Cerebral Palsy. Back in August, the geneticist told us about the whole exome sequencing test and we decided to have it done on one of the twins. When the results came back in Mid-January, we were told the diagnosis was GRIN1 Related Disorder. The genetic counselor told us that only about 10 other people in the entire world have this diagnosis (that have been tested so far) and said that it would be difficult to connect with other families. The other twin is having targeted testing done now to affirm that she also has GRIN1 disorder, but since they are identical and she has the same symptoms as her sister, it is already almost definite. The testing just makes it official.
Hi Keith. I was so excited to come across your blog. Our stories sound very similar. My son was diagnosed with GRIN 1 about 1 1/2 years ago. You mentioned being part of a community on social media. Could you please give me some more information. I would love to join!

My 18 month old daughter just received GRIN1 diagnosis. Any advice? Is there a support group/website that I can get more information?
What does GRIN1 look like?

- Hypotonia
- Moderate to severe developmental delay
- Physical disabilities
- Laughter (frequently manic) & outbursts
- Seizures
- Speech delay
- Flexibility
- Infinite hugs
Thank You!