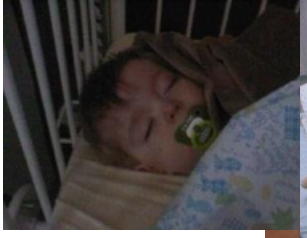
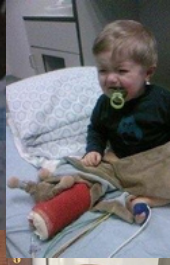
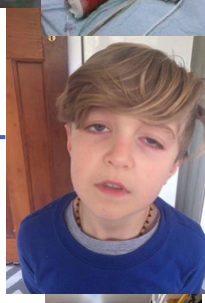
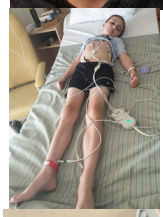
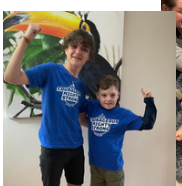
Letter of Support for Amplo Bio Tech – Amp 101





November 15, 2024

Mackay Jayden
CMS DOK 7
15 yrs old

Dear members of the Application Review Subcommittee,

I would like to write to you and give you a bit more of an insight what it has been like living with CMS, Dok 7. From as long as I can remember my parents have had to fight to first convince doctors I was not ok.You see, I was not just a baby not meeting any milestones,, never crawling , not sitting by age 14 months, not walking by 2 yrs old, ...And getting told he is just slow, after my first respiratory arrest where mum found me and had to revive me, again they had an explanation. Evertime there was a new pneumonia, which we now know was from aspirating, he is just sickly however... I would regress and become vague, loose any thing I might have learned..... since I was born I spent 4-5 times in hospital a year and mum and dad knew there was something potentially serious going on. ...Then when the diagnosis was found, after a gruelling long time it is still a constant battle. You see, you might have read that people with this condition have muscle weakness, which is worsened upon exertion. The age of onset, severity of presenting symptoms, and distribution of muscle weakness can vary from one patient to another. A variety of additional symptoms affecting other organ systems can be present in differeny subtypes. Severity can range from minor symptoms such as mild exercise intolerance to severe, disabling ones. You will find in the literature how we have these daily treatments, and by managing our CMS and "energy batteries" we should be able to live I guess a reasonable life? Now , my reality has been, very different. This is my experience of living daily life and how it controls every aspect of my life since diagnosis despite trying to avoid deteriorations or 'crashes' , watching the amount of energy, adaptations, avoiding heat exhaustion, making sure not to have a boom-bust life style and also avoiding infections as they are a huge trigger for me as proven when mum found me having a respiratory arrest age 1 when I had a double ear infection and she had to revive me. It was not to first time I spent in the ICU. Or that mum and dad thought they might loose me. I have spent several times there. I actually heard my parents only a few weeks back talk quietly about their fears again, due to my latest challenges. And I know they live with fear every day, my mum checks on me multiple times a night. Yes, I am 15. And no, this is not weird. I am so glad she does as mum has literally had to make quick decisions on so many occasions.

Despite my desire to be included at school and be with my friends in what their world is... no matter what I try, how careful I am- all has failed due to constant reminders and the heavy price I pay during and after trying to be part of their world.
Without excerting myself, just by being. I would fall asleep in class, not being able to be woken up - ambulances called. Multiple times.

I would have conversations with my friends or anybody, with my speech clear and defined one minute, and the other minute, unable to form words, people staring at me or walking away. I would walk around, trying to be with my friends, but not be able to keep up- combined with loosing the ability to form words. By the time a teacher noticed me, I had collapsed as I was struggling to call for help and nobody heard me that I needed help to take my jumper of on the warmer day.
I am known to my teachers as intelligent, inquisitive, motivational, inspiring, mature, , loves to learn, lots of interest etc... however because of my CMS can also cause such severe brain fog and a cognitive decline that I have also been described by as urgent assessment needed, could be neuro diverse, cognitive impaired, learning difficulties, , etc. ..(All tested - all negative!)

It took me years to swim, and so when the diagnosis came and I appeared better on the medications , all should be ok? Well, in reality in my situation, all changed after a few seconds, my brain seemed liked my limbs were doing the movements, but they were not. Again, shouting for help is not my best skil, nor is breathing when feeling panicked. My mum jumped in with her clothes and all when I was on the bottom... I had swam fine before and seemed fine.
I am 15 years old, every child should have the right to education right? Unfortunately not if you have what I guess I call ' unmanageable CMS'.
, IN theory, there are daily treatments?
In my case, they have trialled Ephedrine, which made me completely loose all appetite, stopped my growth, I lost 5 kgs in weight, have severe migraines, that severe that I could not tolerate anyone around me. And I developed chest pains and cardiology got involved.

Then, they trialled 3;4 Dap. First I did a severe reaction, which again led to my parents believing they would loose me. The dosage was increased to rapidly.
Mum, an intensive care nurse followed her gut feeling and wrote to Dr Engel. He advised against using it. She never expected him to reply but he was extremely helpful.
We tried without it for a year however I by that time had lost all skills I finally had learned again like swimming, going to school for 3 , days a week despite having to use an electric wheel chair and give up on walking became to much, and I spent multiple times in hospital with crashes even tho I had not excercised. I might have gotten a cold, perhaps summer was tough for me, or perhaps mum checked in the middle of the night and found me in a compromised position, unable to roll myself back, unable to wake me. You see, when I deteriorate, one of my signs will always be a very deep sleep, where my GCS drops and It is not safe for me anymore. I am unable to take deep breaths, my oxygen levels drop and this might take days, sometimes longer.

The medical team convinced them 4 years ago to try 3,4 dap as a last resort. The risk for long term damage to my muscles was only 'slight', and maybe 20 years away.

it did help for 3 years, and I got better. High school started and I even attempted basketball. I got injured 8 times in 4 months, ended up in a moon boot,had a wrist brace, was in constant pain but I felt like I was Michael Jordan. I loved school. I did end up in the intensive care again and had 2-3 hospital admissions a year. But, ... I played basket ball... at least 10 mins per game before getting swapped out, If I was not injured. I managed High school without my wheel chair!

Until last year, The last 10 months I have again lost all my skills. I have not been able to go to school since January. meaning, I have missed my entire year 10. I developed really painful problems with my hips and knees, flanks... and all of the sudden my hands lost lots of strength to. My head is to heavy for my neck and It feels like it will roll off any minute. When standing my legs tremble and burn, and even tho I have been working on strengthening, stretching and rehab I have been stuck at 6- 11 mins walking before I get or injured or my legs give away.

My ptosis and double vision is so bad at times , everything looks distorted...which is not great if all you can do is spent your daily life in your room and your view is distorted and blurry.
I would like to think I'm not bad looking, but when everyone stares at you with concern or what the? On their faces, it is not the best feeling.

All my friends disappeared, 15 yr olds have a life to live you know. Just not the ones like me. Stuck in their room or at hospital. Having to use the wheelchair again, was very confronting. Mum not only checking me but helping to get me comfortable multiple times a night. Being 15 needing help with daily care, dressing, cutting food, ...

My independence is gone. I feel I am disappearing to. But I know I can really be someone truly amazing and inspiring! I just need to be giving the chance!

3 hospital visits in they had maxxed out the salbutamol dosage to the absolute max.(4x4mgs) Of course when you take that much you can imagine the racing heart, tremors etc. plus the inability to sleep. Lying awake , and then realising it all starts again.
If I get sick, which is when they normally increase the salbutamol- we have no more back up. And the 3,4 dap made me so much worse and is being weaned off atm. I am so lost, angry and to be honest do not feel I can trust my doctors because if this was known why did I have to go through all this. Why guess? Why give me a taste of what life could be and then take it away like this ?
They say they can not figure out what to do next ? But you are my doctors?The rehab team refuses to help with rehab as they feel my CMS is to unstable and any attempts to do rehab making me deteriorate. We are being shovved around in an endless loop. Where it feels there is no hope?
It also scares me how little the hospitals know about Congenital Myasthenia vs Myasthenia gravis. And despite of countless admissions per year, every time, countless drug errors occur or meds are not administered because, salbutamol given in such big dosage crashes the administration system or the nurses still think it is for asthma...

The dangers of them still not recognising symptoms of myasthenia crashes despite looking after me for 8 yrs since diagnosis worries me. They of course try and are kind, but it is because it is rare...
The times I have ended in up in intensive care is the times I got sick. I am terrified. My mum and dad are to. We do not know what to do next. The only medicine available is maxxed out.and there is no plan b.

I want to live life, Have a chance to actually be a teenager and go to school, be with friends and run around, have fun without fear that that lands me in ICU.

Have sleepovers, school camps ... think about what I want to do at university.
I missed the majority of primary school. Now it is happening at my high school to... doesn't.t ever child have the right on education?

i do know this, together with so many of others with this condition , we need the gene therapy.THAT IS OUR HOPE for living life. The ONLY HOPE - We live in fear every day. And 40 mls of salbutamol a day- is not the treatment that is sustainable for this 15 year old fwo has a lot of big dreams.

Our daily life is not what you read or how the condition is defined by literature and research. It is not just a case of limit exercise, take the medicine available and you will be ok. The treatments now available are failing many. ..I hope by telling you my story, you realise how the CMS community desperately needs help.