My name is Emma Nott and the particular Patient Group I am involved with is Hope for Hypothalamic Hamartomas. Although we were founded in 2009, our story starts nearly 15 years ago. On July 10 1997 CJ Soeby was born. He had his first gelastic seizure as he was being delivered and continued having 300 a day.

CJ's parents, Lisa and Jon were employed by the US military and were stationed in Turkey at the time. They were swiftly returned to the USA, to the best military hospital where they hoped the very best medical attention could help their son. MRI revealed a large lump at the base of CJ's brain; doctors thought it was a tumour, biopsy showed it was in fact a hypothalamic hamartoma. But in 1997 nothing could be done for HH - it was explained to Lisa and Jon that their son would continue to have hundreds of seizures a day, that these would likely progress in severity and that he would become cognitively impaired to the extent he would be in a care home from the age of 10. Medications were tried but without success - gelastic seizures are generally refractory to anti-epileptic drugs.

Lisa and Jon moved to Phoenix to be near BNI. No real help - no treatment options. They scoured the internet and in 1999 Lisa found a small group of patients had formed a worldwide support group, Hhugs. Through this group she discovered that a surgical team at the RCH Melbourne had pioneered a new surgical approach to HH, with very good preliminary results. Lisa took this to the surgeons at BNI who were sceptical. They said, “If this was any good don’t you think we’d be doing it here?” Unable to find a medical team in the US who would help their rapidly deteriorating son, in 2001 Lisa and Jon flew CJ half way around the world to Melbourne for pioneering surgery. CJ was 4, surgery successfully removed his hamartoma, there were no operative complications and CJ was seizure free for some years. He is now 14, in mainstream school and suffers gelastic episodes periodically. It is not a perfect outcome, but one which the Soebys could not have dreamt of prior to Melbourne. CJ was one of a number of international children accepted by the RCH, many of whom were found through the Hhugs support group. One of the problems in trialling and refining surgery for HH was its comparative rarity - at that time the prevalence was thought to be 1 in a million (now 1 in 200,000) - here an internet support group was finding patients and channelling them to the RCH to the benefit of both, and of course to medical advancement.

But Lisa didn't stop with CJ. After their return to the US she took him to Dr Spetzler, the surgeon at BNI and said “tell me again why we’re not doing this here?” As a result, BNI invited the Melbourne team to BNI to teach them the surgical technique. This happened in 2003, and 12 successful surgeries were performed, the patients in many cases having been found through the Hhugs support group. This in turn led to BNI opening a HH centre, and they have now performed well over 200 surgeries. They have also developed other surgical approaches including an endoscopic approach which can be used for some HHs and which is safer still.
At the same time, back in Europe, in fact right here in Marseille, Prof Regis was busily embarking on his own pioneering HH surgery - the first gamma knife series, and in 2006 he published a preliminary report, the first to suggest that gamma knife could be very effective in the treatment of smaller hamartomas.

And so, medical trials were beginning to focus on the hypothalamic hamartoma, and centres of expertise were beginning to emerge. But for the parent of a recently diagnosed child, options were still very limited, unless you were lucky enough to live in Phoenix, Melbourne or Marseille, or unless you had determination and a USBN line.

My son Charlie was a happy child with some mild speech delay at 2; by 4 he was exhibiting significant behavioural problems and moderate speech delay; by 5 he was diagnosed as suffering from autism and ADHD - the latter because of his inability to sit still, his odd grunts, head-shaking and eye deviations. We were concerned about his frequent unexplained laughter but were told it was a behavioural symptom of autism and to be grateful that we had “one of the happy ones”.

Without the internet Charlie would remain undiagnosed. I read everything I could on autism, but it didn't quite seem to fit. One day when Charlie was seven he had another long episode of laughing which prompted me to type in “uncontrollable laughter” to the google search engine. For the first time I came across the phrase “gelastic seizure” and through that I was led to the Hhugs webpage. We returned to the paediatrician, and MRI later confirmed a small HH. UK hospitals could not help. Great Ormond Street neurosurgeon said he didn't think it was a hamartoma, but even if it was, it was inoperable and advised us to keep off the internet. Too late! I asked the surgeon about the three centres I had read about - Melbourne, Phoenix and Marseille. He said it was all highly experimental, and clearly none of them had actually found a cure as they were all working on different approaches and to an extent working against each other.

I could not get any sensible information about HH from any doctor I saw. I educated myself through medical papers which I sourced on the internet, paying a fortune in subscriptions to medical journals - I built up quite a library. I discovered I was walking an identical path to most other parents who had a child diagnosed. Through searching for help for Charlie, I came across Lisa, and another American lady called Ilene whose son Mark was a similar age to Charlie and who had had gamma knife surgery in the States with some success. Once I felt I had garnered enough information I pushed good manners and protocol to one side and began writing directly to the eminent experts I thought might help. Through Lisa, now working as patient liaison at BNI I sent Charlie's records to BNI who confirmed HH and advised GKS. My parents flew to Melbourne with Charlie's MRI scan and saw Dr Harvey, who confirmed HH, advised GKS and suggested Prof Regis - so much for these Drs working against each other. I wrote directly, by e-mail to Prof Regis, who responded immediately, and in October 2009 Charlie had GKS here in Marseille. His is not yet a perfect outcome - he will likely have a second GK surgery, but although he still has seizures, within a year of surgery his behaviour normalised and his cognition stabilised to the extent that he could leave his autistic unit and now attends an independent school for children with dyslexia - he even went on the school ski trip!. Not something we would have thought possible before surgery here.
Emma Nott Testimony

Stereotactic & Functional Neurosurgery Conference in Marseille, France

February 2012

I should say that in part as a result of Prof Regis’ GK series, Barrow began its own GK series and now also uses the gamma knife as weapon of choice for the smaller hamartomas.

At this time, 2009, Lisa, Ilene and another American mother Erica Webster were setting up a charitable organisation which aimed to take the current support group a step further.

Lisa, Ilene and Erica wanted to post on the web in one place all the medical papers we had each collected over months of research so that other parents would have easy access, they wanted a website that could be a kind of one stop shop, detailing HH types, symptoms, treatment options. They wanted actively to raise awareness of the difficulties in diagnosing and treating HH throughout the medical profession as well as giving patient support. They asked me if I would come on board to give it an international element - they envisaged an organisation without walls. And so Hope for Hypothalamic Hamartomas was born. We set up, thanks to Erica, a strong website where we have our one stop shop. We go, where we can, to epilepsy and neurological conferences to support our doctors and to educate others. We lobby governments, we testify at Public Health Committees. We are, simply, four mothers of HH children who have donated as much time and money as we can spare to establish and run this organisation; however, we don’t see ourselves as a patient organisation, rather a charitable organisation dedicated to the single goal of a successful outcome for all sufferers of HH.

We knew that if we were to have any success in our goal then we needed to engage and support the medical profession, the doctors and surgeons at the forefront of medical advancement. And so we created a Medical Advisory Board. We decided to reach for the stars, literally, and invited those eminent Professors who are changing the face of HH treatment to serve with us. We were delighted that each person we asked accepted without hesitation. Remember this is all on a purely voluntary basis - we can't afford to pay anyone; it is natural that parents would volunteer; but we were all overwhelmed that these doctors have been so generous with their time, and freely sharing of their expertise.

The Hope for HH Medical Advisory Board had its inaugural meeting in December 2011 at the AES Meeting in Baltimore. It was quite something to have all that expertise in one room! The point of our organisation, and its shop window, the website, is that it is designed to engage medics as much as patients and families; and so you will see a high degree of medical content, which we hope to develop, as time goes on. We have already worked with the Journal of Neurosurgery to publish, in February 2011, an online edition of Neurosurgical Focus dedicated solely to Hypothalamic Hamartoma. Dr Rekate of our MAB edited that special edition, and we hope to follow up on that success with further HH supplements as and when appropriate.

We are an organisation in its infancy; we have much to do. Raising awareness is at the top of our list. We want to get these children diagnosed and then get them to the doctors that can help them. Within the last 10 years the prospects for HH sufferers have changed dramatically - from literally no hope to very real hope. It is our aim to facilitate that progress and to support doctors in every way that we can so that progress might continue at the same pace over the next 10 years, and in 2022 we look upon HH as a condition that is always readily identifiable and eminently curable. To that end, please look out for the 2nd International Symposium on Hypothalamic Hamartomas [pencilled in for September 2013] which project arose from discussion at our inaugural Board meeting, which Professor Regis will be chairing and which Hope For HH will be sponsoring.
Finally a few words about CJ Soeby and Grace Webster, our two poster children in the slides we presented at the AES in Baltimore. Shortly after we displayed the slides Grace very sadly passed away suddenly during a seizure. This has devastated Grace's family and our small community too. The picture is a bittersweet reminder of what can be achieved on the one hand, and the very tragic outcome where treatment fails on the other. Much has been achieved, but there remains much to be done.