One Mother's Empirical Report on Glut1

Valerie was born in October 1998. Pregnancy and birth proceeded without any complications. We had our first child, a pretty and trim girl. A new world opened up. We were overjoyed.

On Christmas Eve (!) 1998 we realized for the first time that "something" was not in order: Valerie was 2 ½ months old and her left arm had a spasm out of the blue! It was a classic, so-called "focal convulsive attack" which is described in any medical textbook and which I have been able to observe on numerous occasions in the neonatological intensive care ward during my long professional career at a large university clinic. The first examinations including an EEG did not show anything conspicuous. In the time thereafter conspicuous neurological signals emerged in increasing number and intensity: again and again sudden nodding movements, difficult to describe "rolling" eye movements especially when subjected to cold stimuli and an increasingly obvious reduction in muscle tone which for the layman can best be described as "sloppiness". Since Valerie was on the whole very receptive and cheerful and - still - did not show any clear signs of arrested development, we encountered during our repeated visits to different specialists incredulous looks and behavior which seemed again and again as tending to calm our fears. One time the rolling eye movement, so alarming for us, occurred in the presence of a female pediatrician. I was thinking NOW someone is finally going to believe me that our child had a real neurological problem. Nothing of the kind!!! To my extreme amazement this thoroughly conscientious colleague whom I knew well, assessed the eye movements as typical for a baby at that age and dismissed it as completely harmless. Once again I felt powerless. Both as a mother and in particular as a medical doctor I was quite sure that these occurrences were by no means normal. (Viewed from hindsight my colleague could not have reacted any differently because GLUT 1 was practically unknown in 1999.) In particular, these difficult to describe eye movements are a typical symptom for a Glut1 defect.

After about five months we had reached the point: our fears about our child were sadly confirmed. On a wonderful sunny Saturday morning we held for the very first time in our arms an unconscious, pale, twitching baby. The first big convulsive attack was at hand and it proceeded like a perfect example.

A world collapsed.

In the next few weeks the small conspicuous neurological aspects and the great convulsive attacks occurred in ever shorter intervals. And still, hardly any of my colleagues believed us because Valerie's slow development still lay at a point in time which just fit into the framework of textbook standards! Pediatricians tend to work according to the principle "when in doubt in favor of the accused". Many examinations were indeed conducted on our insistence, but the more inconspicuous the results we had in our hands, the more we encountered the soothing attitude that up to seventy percent of all convulsive attacks in the first year of life remained unexplained short-lived events and that our child would almost assuredly belong to this group because of its otherwise "good" development. To an every increasing extent we felt like we had been abandoned. I was furious in the meantime because if my colleagues didn't take me seriously as a mother, then they should believe me as a colleague. When a female medical doctor as mother cannot be believed when she describes a convulsive attack with all its criteria as a "classic example" and calls it as such, then how are those people going to feel who are not medically trained? In order to finally be able to prove our descriptions and not have to continuously start from the beginning and repeatedly struggle against the unbelieving attitude of our colleagues, we bought a video camera and recorded a few of the "rolling" eye movements and a great convulsive attack!

After nine months our assessment was “finally” confirmed by our pediatrician that her development was arrested due to a disease. Sitting up alone is one of those abilities which every healthy child can do at least at nine months of age even if it is not able to sit up by him/herself. Even when 12 months old there could be no thought of Valerie sitting up alone – a burden that the parents of a healthy child can hardly imagine. The child most definitely wants to experience life because Valerie’s development was not so arrested that she wasn’t interested in different things. She simply was not able to do so herself. She increasingly developed into a fidgety and unruly child who at the same time was very sweet, friendly and open and who always wanted to be held when awake. The time needed for her care was comparable to that of a three or four month old child. You could not put her down and her considerable weight, which in the meantime had reached nine kilos, caused back pains and increasing discontentment especially when simultaneously having to carry all those portable household appliances or baskets and bags. How I envied all those mothers who complained about how strenuous it was to run after a healthy, crawling 7 ½ month old child who with pleasure and interest began to discover the world! In our case, appointments for gymnastics according to Bobath were made three times a week and progress was getting slower and slower. Valerie increasingly displayed an expression of “otherworldliness” as if she were from another planet.

We already had the impression after the third large convulsive attack that her problems could have something to do with her eating because all the attacks occurred in the morning especially after a long break between meals; the first attack was after an uninterrupted night’s sleep. They became more frequent, the more often Valerie slept the night through. The result of this realization for me as mother was not one of being happy about a good night’s sleep, but of being anxious. I set my alarm clock every night at 2:30 AM in order to wake the child and feed her. I hoped to be able to avoid the attack by never letting her get hungry. I was thereafter less and less able to go back to sleep again myself.

Professionally I was working full-time again including strenuous night and weekend shifts since Valerie was no longer being breastfed after 7 months, but I could not avoid working night shifts despite the overall personal situation. I also did not want to admit to myself that our daughter’s increasingly evident neurological impairment would lead to a physical disability which would turn our lives upside down. My colleagues who - still - diagnosed Valerie as having a normal development constantly tried, in their helplessness, to help through soothing words. They did not clarify the situation and attempt to come up with a solution. Chronically lacking sleep, constantly tormented worrying about Valerie and in the free time I had going from examination to therapy appointments carrying a child who became heavier and heavier, I reached the limits of my strength. Critical periods naturally surfaced again and again in our marital relationship, even though my husband was just as concerned as I was. Instead of being there for each other in the few minutes we had together after long and busy day, we only quarreled. How were we to come up with the reserves to be affectionate to each other and to carry on a conversation?

The time between the first attack on Christmas Eve 1998 and the diagnosis when Valerie was one year old was a steady downward spiral in every respect.

In the hope our observations could contribute to finding a diagnosis, I had started relatively early with a diary-like anamnesis documentation and took it with me to the Charité for a hospital stay for the purpose of diagnosing metabolic processes. It was my information that caused Dr. A. von Moers in the Rudolf Virchow Clinic in Berlin to come up with the idea of examining Valerie for the presence of a Glut1 defect. What luck that he had even heard of the symptoms! At this time only 32 people had been diagnosed worldwide with Glut1 and even many experienced neuropediatricians had never heard of it.

We were afraid of the recommended examination not just because a nerve water test was necessary, but that the latter had to be conducted on an empty stomach! It was extremely difficult for us to agree to this examination which, however, corroborated our

suspicion. By means of a time-consuming blood test the Glut1 defect was finally established.

This diagnosis was a relief; we no longer had to grope in the dark. We became acquainted with Dr. Klepper at the University Clinic in Essen who asked us about all the symptoms without having seen the video and having learned about Valerie's medical history. Our odyssey was finally behind us. At last we felt that we were in the right place. We were understood, he took time with us, he answered all our questions and explained so much to us in our very first talk. After his laboratory confirmed the suspected diagnosis, it was perfectly clear what the problem was: lack of energy in the brain. It was also clear as to what had to be done: replace sugar as the supplier of fuel which was not being absorbed with something else: a ketone body.

With great hope, but also worried about how we would cope with everything that we had heard in the meantime about the ketogenic diet, we went on the day after Valerie's first birthday to the clinic for an introduction to the ketogenic diet.

On the first day of the ketosis our child was transformed: the fidgetiness, the restlessness, the nodding movements and the far-away look disappeared from one day to the next and the "rolling" eye movements were gone as if they had just been blown away. In the morning we did not need to worry anymore about large attacks and the middle of the night meals belonged to the past. Valerie started to make progress as soon as the diet was started.

In this respect our daughter exhibited a typical course for this metabolic defect. Unfortunately, attacks occurred to an extent analogous to previously non-existent, self-initiated activities such as crawling which put a strain on her body. It was as if her brain was still doing heavy work in order to fall back upon sugar which could quickly be put to use, but which was now under the ketogenic diet only available in even smaller amounts than before. After a short quiet period, a new pattern of attacks slowly crept in and culminated with numerous attacks every day. We started to doubt the correctness of the Glut1 diagnosis because normally with no attacks under a ketogenic diet you can stop a repeated anti-epileptic therapy and not start such a therapy as we did with a heavy heart. However, we were all astonished when Valerie suddenly did not have any more attacks after taking ¼ of a carbamacepine tablet on November 18, 1999 and has remained free of attacks ever since! For a good year she took an attack inhibitor in such a small dosage that she never reached the therapeutic level until she, like other Glut1 children without attack inhibitors, could continue to be treated only through a diet. In the meantime the molecular-genetic diagnosis on chromosome 1 has been corroborated.

From this day on Valerie has made progress which corresponds to the speed and development of a healthy, one-year old child.

Today Valerie is a cheerful, happy child who runs around like all other children, climbing playground frames, rides a bicycle (with supporting wheels) and wants to learn to swim. She is greatly looking forward to her fifth birthday and to nursery school. That she has a metabolic defect which could lead to an extreme bodily or mental handicap if not treated, or if treated incorrectly, is hardly evident when looking at her. The ketogenic diet has given us back our child and our joy of life. We have found our way back to each other and to a normal life – with one small restriction, however: one of us can only work at most half-days despite an all-day nursery. The additional preparation of meals together with all other things which have to be taken into account still requires, even after four years, a very great expenditure of time.

BUT IT IS WORTH IT!!
In conclusion, allow me to pass on a recommendation for all those who are about to start a ketogenic diet. Make sure you find out how much experience the dieticians have had with a ketogenic diet. The on-going assistance and support of an experienced dietician is absolutely essential for the start, with few problems and free of fear, of such a diet. The

training of the home care person(s) cannot be handled anywhere at any time by anybody, but must be carried out by this team. Also, it is this team that gives you the basic set of recipes to take along and on which you can, and have to, build up yourself.

Lots of luck!

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