

It was August 2009, and I was visiting my 31-year old daughter, Marika, who had been diagnosed with Chorea Acanthocytosis 8 months earlier. She was now ambulating with the assistance of a cane, her diet consisting of soft foods such as applesauce and puddings. After she had showered and dressed for dinner that first night, I sat down on her bed for the chat that I anticipated would be difficult yet necessary.

“Honey, we need to talk,” I began. “I have been asked to help write a letter to people who have a diagnosis similar to yours. It’s a letter asking them to consider donating post-mortem brain tissue. That means after they die, they would agree to send their brain tissue to Dr. Bader and Prof. Danek – the researchers in Germany who confirmed Dr. Shale’s diagnosis – in order to further research into this rare and elusive neurological disorder that you have.” My beautiful daughter nodded, encouraging me to continue. “I need your help because I cannot know what it is like to have what you have. I can’t possibly write this letter without your insight. Would you be willing to help me?” She readily agreed. I continued, “Sweetheart, even though your neurologist assured us that you will likely get to celebrate many more birthdays, when you do pass away, would you be willing to donate your brain tissue for research?” She looked at me steadily and said, “Oh, yes! Of course!”

Little did we know then that a mere 3 months later, she would choke on a hot dog, an episode that ultimately would end her life. The accident occurred on a Wednesday evening in November. I had spoken with her at least 3 times already that day and had left a message on her voice mail – as it turns out – about the same time that the ambulance arrived on the scene to rush her to the Emergency Room.

By the time I arrived at her bedside in the Intensive Care Unit, she was heavily sedated. The doctors had told us not to expect any miracles, as her brain had been deprived of oxygen for approximately 10 minutes. However, they didn’t know Marika! By the weekend, she did regain consciousness. Nonetheless, by Monday afternoon we were in conversation with the palliative care team...

“My daughter wants to live. However, she wants to live a social life,” I told them. “I know this because, unlike most mothers and daughters, we have had this conversation. And, if this is the end of her life, I know that she wants her brain tissue sent to Germany for research purposes so that other families in the future can benefit.”

As I look back on those days, the last few of my daughter’s life, I cherish the clarity that I had, despite my sadness and grief. That she and I had had the conversations helped me make some of the most difficult decisions with which I have ever been confronted. My daughter gave me a most beautiful gift in that regard – certainty about her wish to donate post-mortem brain tissue for research. Brain donations such as Marika’s provide researchers with the invaluable opportunity to examine the origin of NA symptoms. Such donations of post-mortem brain tissue are a deeply valued and scarce resource for researchers seeking to understand how the gene mutation interferes with the brain’s function in movement control. NA is not seen in other species, so human brain

examination is vital. Such research offers hope that we can someday understand the pathology of this rare neurological disorder.

Below is the email that Dr. Benedikt Bader sent me on Tuesday, two days before Marika breathed her last breath. I share it so that other family members may know what to do, should they find themselves in a situation similar to ours.

Dear Ann,

The sad news and also your request reached us here in Germany. Tissue donation is the ultimate contribution patients like Marika can make to support research. We are very grateful, especially since her disease is extremely rare and patients' tissue is vital to advance our understanding of the mechanisms leading to disease. Despite the sad news, if it comes to the worst, we need to be prepared. If there is consent with Marika and her parents, the autopsy needs to be done as quickly as possible. Can you tell me, the name of the doctor caring for Marika at the moment? I would contact him and advise him about the autopsy if needed. In any case, if an autopsy was already performed, it is necessary to preserve at least paraffinated tissue blocks of the following regions (as complete as possible):

*Frontal,
temporal,
parietal,
occipital (area 17),
cingulate + corpus callosum,
Amygdala,
anterior hippocampus,
posterior hippocampus,
thalamus,
cerebellum (vermis region),
cerebellum (dentate nucleus),
mesencephalon inc. substantia nigra,
pons,
medulla oblongata (inferior olive region)*

I would try to arrange the transfer of tissue to Germany directly together with the pathologist in charge.

In any case, I do hope that Marika is not suffering!

All the best and please, if you have the chance, pass our deepest condolences to the rest of Marika's family! Benedikt