From: Jeremy L Freeman
To: kraemerk@nih.gov
Cc: sammyjo2@bigpond.com
Sent: Friday, June 23, 2006 3:14 AM
Subject: Sammyjoe Liistro

Dear Dr Kraemer,

Thank you for your very informative email and summary about Sammyjoe .
I have been minimally involved with his care this week when I was asked to provide an opinion about repeating his MR imaging.
Apparently it had been suggested to Mrs Liistro that a scan done with a 3-Tesla MRI was superior to that done with a 1.5-Tesla machine and could show a small brainstem stroke that might be missed at 1.5T.
I reviewed his case and the numerous investigations performed in order to determine a cause for his declining bulbar function.
These included CSF studies, metabolic studies, nutritional evaluation, Acetylcholine receptor antibodies and a trial of pyridostigmine, in addition to neuroimaging at 1.5T.
No contributory conditions other than his TTD were found and no improvement was noted with pyridostigmine.
As you described in your email, patients with XP develop bulbar dysfunction similar to the problem that Sammyjoe is having.
The current view is that Sammyjoe's problems represent a deterioration of his condition. This is difficult for his parents to accept because they have seen him avoid some of the problems that younger children with TTD develop.
I have offered to correspond with you to discover if there are patients with TTD that you care for who have had findings on a 3T MRI scan that were not detectable with a 1.5T scan.
I am currently of the opinion that our clinical imaging at 1.5T is superior to that performed elsewhere with high field magnets, including at most US centres.
I would be willing to rethink this position with your help, given your expertise with TTD.

I do appreciate the time and effort you have devoted to Sammyjoe and his family and the assistance you have offered.

Best regards,

Jeremy Freeman

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Thank you for your e-mail.

Mrs Liistro is an amazingly dedicated woman. She has literally travelled around the world to find help for her son. She has obtained support in Australia and has set up communications via e-mail with other families throughout the world. As a non-medical person she has become very knowledgeable about TTD. She has shared her knowledge with many families who later come to us with stories as to how they were helped by her.

We have been studying xeroderma pigmentosum here at NIH for several decades. Recently it became clear that patients with trichothiodystrophy also have defects in the same genes as XP patients despite their very different clinical features. XP patients have a 1000-fold increase in skin cancer while TTD patients have normal skin cancer risk. Consequently we began studying TTD patients in order to try to understand the basis of these differences. When she brought her son here in 2003 we had never seen a patient like him before. His severe autistic features, extremely marked sun sensitivity and long hair were unusual for TTD patients.

We performed an extensive work-up and consulted with Dr. Maryland Pao, a paediatric psychiatrist. She recommended treatment with Prozac after initial calming with Valium. He had a remarkably good response. He gained weight, was more communicative and attended school. We had periodic reports of his progress. In fact he was scheduled to come back here next week in follow-up.

As you know, since last October his condition has deteriorated. We spoke with his mother on the phone yesterday. He has lost considerable weight, become weaker and has marked difficulty walking and swallowing. In Dec 2005 he was brought to the ER with an episode of choking on his food and nearly asphyxiated. He has continued to aspirate his saliva. Recently a feeding peg was placed into his stomach. He was in and out of the hospital during the past few months. Yesterday he developed a urinary tract infection. His mother indicated that he had difficulty urinating and moving his bowels. We suggested that he be checked for a distended bladder.

The workup you describe is very appropriate and extensive. There was a question about use of MRI. I spoke with the neuroradiologist here, Dr. Nick Patronas, and he indicated that the 3 Telsa MRI is not necessary for anatomic studies but would be useful for functional MRI or spectral MRI. It is doubtful that these specialized studies would be indicated at this time. Dr Patronas reviewed the earlier MRI during the visit in 2003. He has also seen MRIs of other TTD and XP patients that we have examined here. Dr Patronas is willing to review the new MRI from Sammy-Joe if it is sent to us. We are also willing to review any of the other medical records.

Please let me know if there is any way that we can assist you in understanding this difficult situation.

Sincerely yours,

Ken Kraemer

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