We spoke today with Mrs Liistro who gave us information about her son Sammy-Joe who has trichothiodystrophy (TTD). This is a rare genetic disease with multisystem involvement. We saw him here at NIH in the 2003. A copy of our summary letter from that visit is attached to this e-mail.

After returning home and beginning medications his behaviour improved markedly, he became more interactive and ate more food. His weight improved and he was able to attend special education classes.

Since October of 2005 his mother reports that Sammy-Joe has been regressing. His appetite has worsened. He has lost weight became more difficult to manage. In December 2005 it became clear that he was having difficulty swallowing and was noted to aspirate saliva. Subsequently he began choking on his food and had a episode of food aspiration with hospitalisation.

Recently he has become progressively weaker with difficulty walking and even holding up his head. A feeding peg was placed and he receives nutrition through the tube.

He also has experienced progressive difficult in urinating. According to the mother he now has a urinary tract infection.

I realize that it is almost impossible to diagnose a problem from this distance and that we have not directly examined Sammy-Joe. Neither do we have extensive experience in long term follow-up of patients with TTD. However, we have been observing patients with a related disorder, xeroderma pigmentosum (XP), who unfortunately often develop progressive neurological degeneration. Some of Sammy-Joe's features seem to mimic those we have seen in XP patients. The XP patients frequently develop problems with swallowing due to weakness of the muscles involved in swallowing. Several of the XP patients have had severe choking episodes. At least two of the patients died from aspiration and one nearly choked to death on her food. Placing of the peg tube for feeding is an important therapeutic intervention.

Two of the XP patients also developed inability to empty their bladders. They had to be cauterised to remove the urine. Perhaps this is also what is happening with Sammy-Joe. We recommend that he be examined for this possibility.

We would be happy to review any of the lab tests and other studies that have been carried out at home. This is certainly a challenging patient to manage. Please do not hesitate to contact us if you have any questions. We will try to help you to the best of our ability.

Sincerely yours,

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