

Yong Xie

From: Sheil, Amy <Amy.Sheil@cchmc.org>
Sent: Thursday, April 23, 2015 7:54 PM
To: Yong Xie
Subject: RE: Thanks and Status?

Hello, Mr. Xie,

I apologize for the delayed reply - I have been out of town. I did fax the request for Jianhua's pathology materials to SCH; we are waiting on receipt of these materials.

Thank you for your concerted efforts, and the papers you attached to your last email. We will do our best to define Jianhua's liver condition. I have asked that the materials be sent to Dr. Bove, our most experienced liver pathologist, as I am leaving Cincinnati Children's next week to take a different job. Even after I leave, I will make sure Jianhua's pathology receives the attention deserved.

Congratulations on your impending addition to your family.

Take care, and best regards,

Amy Sheil, MD

From: Yong Xie [yong.yx.xie@gmail.com]
Sent: Sunday, April 19, 2015 11:19 PM
To: Sheil, Amy
Subject: Thanks and Status?

Dear Dr. Sheil,

Thank you very much for your report using the 4 slides we sent to Dr. Balistreri. We are wondering if you have received the biopsy material from SCH including one paraffin block and an epon bullet. When can we know how precisely you can narrow down the diagnosis?

So far, we think infantile hepatic CPT2 deficiency can explain everything the best though we also have concerns on ACAD9 deficiency and CACT deficiency. There might be other possibilities that you know a lot more, please try your best to narrow down as much as possible so that Jianhua can receive the best treatments from Dr. Balistreri.

Because we only know FAODs, we have read a lot on FAODs. It's interesting that Dr. Shneider thought many pediatric acute liver failure (PALF) patients had metabolic conditions. His paper from PALFSG is attached. Actually, Jianhua was diagnosed with PALF in ER; later, Dr. Horslen denied PALF and told us Jianhua had a chronic liver disease, not acute liver failure. Thus, we didn't join the PALF study program.

Another paper is also attached, in which the doctor said "Of particular interest was the absence of fatty infiltration of the liver in our patients, a finding that was initially interpreted as incompatible with an underlying disorder of fatty-acid oxidation...." It may explain why it's so hard to get a clear-cut diagnosis. Maybe we have to do the FAO functional test using his muscle sample or to do the acylcarnitine profile with his liver tissue sample, or another biopsy. Anyway, please use them with your best experience, don't be influenced by our email or papers.

The right diagnosis is the best for Jianhua. Two years ago today, he was put onto liver transplant waiting list without a diagnosis and his doctor said it's not important because his liver will be replaced. Now we are doing our best to save his liver. The diagnosis is invaluable for him, and it will benefit the health of Liying and Jianhua's incoming sibling as well.

Please convey our thanks to Dr. Bove for his help.

Best regards and thanks again.

Yong