yong.xie@outlook.com

From: Cella, Ellen <Ellen.Cella@seattlechildrens.org>
Sent: Friday, May 10, 2013 4:59 PM
To: 'yong.xie@outlook.com'
Cc: Kuiper, Victoria
Subject: FW: J. Xie

Dr. Horslen is the Medical Director of Solid Organ Transplantation of SCH.

Yong,

I forwarded your email to Dr. Horslen. He separated out your concerns and put his responses in red. I hope this helps to answer your questions.

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Our biggest concern is the reason of his liver damage. The biopsy shows severe fibrotic changes and tells us that this is not a liver that will recover. The pattern is of a developmental nature with the liver and the injury probably started before Jianhua was born. The genes that determine liver development in utero are only gradually being discovered and the mechanisms of fetal liver injury are not well understood yet. But we can say that the pattern of injury is not what one sees with infection, autoimmune injury, vascular insufficiency, toxic effect or metabolic disease.

Without the reason, the damage in his body may not be limited to his liver only (spleen is still big while liver data are improving), and there might be other things happening while he is waiting. There is no evidence of any other organ system in his body being primarily affected, this does not mean that if his liver function worsens again that other organs may not be secondarily affected. Kidneys, heart and lungs function can all be compromised by advanced liver disease of any cause. This is why he needs a liver transplant, but presently there is no evidence of other organ involvement.

"Fat in Stool" is still observed everyday at home, could this be a serious problem? All patients with advanced liver disease have fat in their stool, because bile is required for fat digestion in the intestine and will chronic liver disease bile flow is compromised. This is also the reason that we are supplementing fat soluble vitamins - source CF (contains vitamins A, D, E & K) and extra vitamin D (cholecalciferol) and vitamin E. The fat in the stool is not harmful in itself it just means that he his not absorbing it from his formula.

His blood test is continuously improving slowly while taking his medicine and Pregestimil, which is given to babies with metabolic diseases. The formula (Pregestimil) and medicines are not given to children with metabolic diseases unless they have problems with bile flow or fat malabsorption. There are no metabolic diseases that are improved with what he is taking.

We didn't see many metabolic testings in the lab list, how can we be certain that it's not related to metabolic things?

- Blood gases show no acidosis
- Ammonia levels not progressively increasing and without protein restriction this rules out a defect of the urea cycle
- Normal electrolytes rules out defect of steroid hormone metabolism
- Alpha1 antitrypsin deficiency rule out
- Defects of fatty acid metabolism ruled out by acylcarnitine profile
- Defects of peroxisomal metabolism ruled out by normal urinary bile acid levels and liver biopsy
- Defects of mitochondrial function and energy generation ruled out by normal lactate and pyruvate levels and liver biopsy, no acidosis and normal acylcarnitine profile
- Defects of aminoacid and organic acid metabolism rule out by acylcarnitine profile and plasma amino acid profile
- Niemann-Pick type C rule out with normal acid lipase level
- Defects of glycosylation pathways ruled out by transferrin testing
- Cholesterol, triglycerides, HDL and LDL do not indicate any defect of lipid and lipoprotein metabolism

What other types of metabolic disease are they worried about ?

Maybe he is not a common case, some symptoms are not so apparent on him? We checked his pictures, jaundice is never apparent on him until several days before being accepted in ER on Apr 9. There are no common cases in infant liver diseases

Before the biopsy, Dr. Horslen told us that the biopsy could either telling us the damage source or pointing to the right direction for further testings. We just wonder what testings are we going to do and when. See answer to first question above in regard to what the biopsy told us. There are no more tests to do, he has cirrhosis secondary to a presumed defect in fetal or embryonic development of the liver – some such as ARPKD and Alagille (and no he does not have either of these conditions) we know the defective gene but for most we do not.

The important thing to concentrate on now is keeping Jianhua as well as possible so that he is ready to undergo transplant when a suitable organ becomes available.

From: Cella, Ellen
Sent: Friday, May 10, 2013 14:31
To: Horslen, Simon
Subject: FW: J. Xie

Simon,
I received this email from Jianhua's father. I am not very familiar with this child since he is followed by Vickie and he was seen by Jorge yesterday in clinic. Are you able to answer or decipher his questions. If not I will let him know Vickie will address his concerns when she is back on Monday.

Thanks.

Ellen

From: 永 谢 [mailto:yong.xie@outlook.com]
Sent: Friday, May 10, 2013 10:29
To: Cella, Ellen
Subject: FW: contact info

Ellen,

Could you please convey our questions to Dr. Hsu or Dr. Horslen?

Thanks

Yong

From: yong.xie@outlook.com
To: victoria.kuiper@seattlechildrens.org
CC: yong.xie@outlook.com
Subject: RE: contact info
Date: Fri, 10 May 2013 10:25:49 -0700

Vickie,

While we are happy to see that Jianhua has been very stable at home waiting for his liver transplant, there are some big concerns that we want to ask Dr. Horslen and Dr. Hsu. It's difficult for any parents to wait every two weeks for an answer.

Our biggest concern is the reason of his liver damage. Without the reason, the damage in his body may not be limited to his liver only (spleen is still big while liver data are improving), and there might be other things happening while he is waiting. "Fat in Stool" is still observed everyday at home, could this be a serious problem?

His blood test is continuously improving slowly while taking his medicine and Pregestimil, which is given to babies with metabolic diseases. We didn't see many metabolic testings in the lab list, how can we be certain that it's not related to metabolic things? Maybe he is not a common case, some symptoms are not so apparent on him? We checked his pictures, jaundice is never apparent on him until several days before being accepted in ER on Apr 9.

Before the biopsy, Dr. Horslen told us that the biopsy could either telling us the damage source or pointing to the right direction for further testings. We just wonder what testings are we going to do and when.

Vickie, please re-word my questions if they are not clear or not so nice. We just cannot stand still long while Jianhua's condition could get worse for some unknown reason.

Thanks