**Question 5 – September 2**

An infant presents with jaundice and white stools at 6 weeks of age. Bili is 6 with a direct of 4; alkaline phosphatase 600, GGT 400. Physical exam shows hepatosplenomegaly. Liver biopsy shows marked cholestasis, mild fibrosis, and paucity of bile ducts.

What would be the next steps in management?

A. Schedule operative cholangiogram with possible Kasai portoenterostomy
B. Request PAS staining seeking positive globules
C. Request an echocardiogram, an eye exam, and spine films
D. Order an acylcarnitine profile
E. Arrange ERCP

**Answer: C**

The child probably has Alagille's, an autosomal dominant disorder in which the bile duct paucity is accompanied by pulmonic stenosis, butterfly vertebrae, and posterior embryotoxon on eye exam. (Biliary atresia would show bile duct *proliferation.* PAS staining is done when alpha 1 is suspected. Acylcarnitines help with mitochondrial disease. ERCP is technically difficult to do in children and would not be diagnostic in Alagille's.)

**Reference:**

1. PD Turnpenny et al. *European Journal of Human Genetics* advance online publication 21 September 2011