

**S u r g i c a l P a t h o l o g y R e p o r t**

Collection Date/Time: 01/11/02 09:20:00

Received Date: 01/11/02 09:32:00

Accession # P-02-00218

### Clinical Diagnosis

Infantile Refsum's Disease, follow-up biopsy

### Clinical Data

Peroxisomal abnormality in 4 year old white male with infantile Refsum's.

### Specimen

(A) Liver biopsy

### Gross Description

(A) Specimen is received in formalin and consists of a liver core measuring 0.7 x 0.1 x 0.1 cm. The specimen is entirely submitted in one cassette A1.

### Microscopic Description

(A) 2 slides H&E, 1 Trichrome, 1 DPAS:

The normal architecture of liver is preserved. Zone 1 hepatocytes show glycogen inclusions in the nuclei. No hepatocyte steatosis or cholestasis is identified. Lipid vacuoles are increased and are mainly located in sinusoidal lining cells, the Ito cells. Portal triads demonstrate normal relation of vessels and bile ducts. One portal tract contains a cluster of large histiocytes with abundant granular, bluish/gray cytoplasm. These histiocytes contain DPAS positive glycoprotein. Trichrome stain also demonstrates delicate mild periportal fibrosis with incomplete septa formation and mild focal intralobular fibrosis. No lymphocytic infiltration of portal triads are identified. No ductal epithelial damage is seen. Hepatocytes do not contain glycoprotein globules. However, numerous Kupffer cells contain PAS-positive glycoprotein material intermixed with negative staining granules and rods, 1-2 micra in diameter.

### Comment

There is slight increase of peiportal and pericellular fibrosis since the previous biopsy.

### Diagnosis

(A) Liver, needle biopsy:

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**Date:** Thursday, December 21, 2000 9:17 AM  
**Subject:** Matthew's liver biopsy

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Despite his liver being slightly large on examination and his enzymes being slightly increased, his liver architecture looks quite good. There is very little active inflammation and the scarring that is present is very mild. Overall, his liver looks very good. We are awaiting the results of the electron microscopic exam of his liver.  
Have a nice holiday season.  
JE Heubi

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12/21/00