



CMHD Pathology Report



CMHD Pathology Core

Toronto Centre for
Phenogenomics
25 Orde St. 3rd fl.
Toronto, Ont. M5T 3H7
Tel.(416) 586-8375
Fax (416) 586-5993

contact: Dr. Susan
Newbigging
email:
newbigging@lunenfeld.ca

Principle Investigator: Dr. Jacqui White

Institute: Wellcome Trust Sanger Institute
Address: Attn: Linda Read Wellcome Trust
Genome Campus Hinxton Cambridge CB10
1SA, UK

ReportID: Report Date: April 24, 2013
Pathologist: H. Adissu

Mouse Genetics Project

Wellcome Trust Sanger
Institute
Wellcome Trust Genome
Campus
Hinxton, Cambridge
CB10 1SA
UK

email:
MGPenquiries@sanger.ac.uk
[Mouse Portal](#)
[Europhenome](#)

CMHD LabID: N13-238

Relevant History:

Clinical phenotype:
- preweaning lethality

AnimalID: M00270555

Histopathology Findings:

liver (MA:0000358)

Histopath Description:

severe lipidosis

Morphological Diagnosis:

Distribution: diffuse; **Severity:** severe; **MPATH Diagnosis:** steatosis MPATH:622

Definitive Diagnosis:

Hepatic lipidosis

pancreatic islet (MA:0000127)

Histopath Description:

There is mild enlargement of the pancreatic islets.

Morphological Diagnosis:

Distribution: multifocal; **Severity:** mild;

Definitive Diagnosis:

Pancreatic islet hypertrophy

AnimalID: M00270556

Histopathology Findings:

lymph node (MA:0000139)

Histopath Description:

The mesenteric lymph node is enlarged (greater than three-fold). There are multiple follicles with large germinal centers. The sinuses contain large numbers of mature lymphocytes and plasma cells

Morphological Diagnosis:

Duration: Sub-acute; **Distribution:** Diffuse; **Severity:** moderate;

Definitive Diagnosis:

Lymphoid hyperplasia.

Histopathology Comments:

The changes in the mesenteric lymph node are suggestive of draining of a regional inflammatory process. However, such a process was not observed in the tissues examined.

liver (MA:0000358)

Histopath Description:

lipidosis is not seen

pancreas (MA:0000120)**Morphological Diagnosis:**

Duration: chronic-active; **Distribution:** focally extensive; **Severity:** mild;

Definitive Diagnosis:

Lobular atrophic pancreatitis

Histopathology Comments:

This lesion is rarely seen in mice

retina (MA:0000276)**Histopath Description:**

There is a focally extensive folding of the retina at the optic nerve.

Morphological Diagnosis:

Distribution: Focal; **Severity:** mild;

Definitive Diagnosis:

Retinal folding (dysplasia)

AnimalID: M00262023**Tissue Preservation and Staining:**

The barin is sectioned obliquely and the olfactory lobe is not in view.

Histopathology Findings:**brown fat (MA:0000057)****Histopath Description:**

There is focally extensive hyperplasia of brown fat infiltrated with low numbers of inflammatory cells (lymphocytes, macrophages and rare netrophils)

Morphological Diagnosis:

Duration: Chronic-active; **Distribution:** Focal; **Severity:** mild; **MPATH Diagnosis:** steatitis
MPATH:636

Definitive Diagnosis:

Steatitis with hyperplasia

lymph node (MA:0000139)**Histopath Description:**

The mesenteric lymph node is enlarged more than two-fold. Its architecture is altered by large numbers of monomorphic lymphocytes that fill and distend all the sinuses and elevate the capsule. The neoplastic cells have generally a scant amount of eosinophilic cytoplasm, medium sized round central nucleus with granular chromatin, and single variably distinct amphophilic nucleoli. Mitotic figures are (less than 1/HPF).

Definitive Diagnosis:

Lymphoma

Histopathology Comments:

The lesion is suggestive of an early lymphoma of the mesenteric lymph node.

liver (MA:0000358)**Histopath Description:**

severe lipidosis

Morphological Diagnosis:

Distribution: diffuse; **Severity:** severe; **MPATH Diagnosis:** steatosis MPATH:622

Definitive Diagnosis:

Hepatic lipidosis

spinal cord (MA:0000216)**Histopath Description:**

Located within the leptomeninges and mildly compressing the dorsal spinal column is a 100 µm

round keratin-filled cystic structure that is largely lined by a well-differentiated stratified squamous epithelium and by simple squamous epithelium that is devoid of basal layer at one margin. The apical cells abutting the lumen contain cytoplasmic keratohyaline granules. There are few (up to 15) neutrophils at the stratified margin.

Morphological Diagnosis:

Distribution: Focal; **Severity:** mild; **MPATH Diagnosis:** dermoid cyst MPATH:311

Definitive Diagnosis:

Spinal leptomeningeal dermoid cyst (dermoid sinus)

Histopathology Comments:

Dermoid cyst is caused by defective epidermal closure along embryonic fissures isolating an island of ectoderm in the dermis or subcutis. Spinal leptomeningeal dermoid cyst has been described various inbred mouse strains (Stroop, 1984). Dermoid cysts have been associated with multiple vertebral and spinal malformations (Ginn et al., 2007). The cyst was small with minimal compression of the dorsal spinal column; hence considered insignificant.

AnimalID: M00266767**Histopathology Findings:****brown fat (MA:0000057)****Histopath Description:**

There is focally extensive hyperplasia of brown fat infiltrated with low numbers of inflammatory cells (lymphocytes, macrophages and rare neutrophils)

Morphological Diagnosis:

Duration: Chronic-active; **Distribution:** Focal; **Severity:** mild; **MPATH Diagnosis:** steatitis MPATH:636

Definitive Diagnosis:

Steatitis with hyperplasia

liver (MA:0000358)**Histopath Description:**

moderate lipidosis

Morphological Diagnosis:

Distribution: diffuse; **Severity:** severe;

Definitive Diagnosis:

Hepatic lipidosis

lymph node (MA:0000139)**Histopath Description:**

The mesenteric lymph node is enlarged more than two-fold. Its architecture is altered by large numbers of monomorphic lymphocytes that fill and distend all the sinuses and elevate the capsule. The neoplastic cells have generally a scant amount of eosinophilic cytoplasm, medium sized round central nucleus with granular chromatin, and single variably distinct amphophilic nucleoli. Mitotic figures are (less than 1/HPF).

Definitive Diagnosis:

Lymphoma

Histopathology Comments:

The lesion is suggestive of an early lymphoma of the mesenteric lymph node.

adrenal gland (MA:0000116)**Histopath Description:**

There is a small, well-circumscribed mass in the cortex. It is encapsulated by a thin layer of pale eosinophilic material and fusiform cells (connective tissue with fibroblasts) and is made of nests of polygonal cells interspersed by a very thin fibrovascular membrane. The architecture is reminiscent of the zona glomerulosa and zona fasciculata of the mature adrenal gland.

Morphological Diagnosis:

Distribution: focal;

Definitive Diagnosis:

accessory adrenal cortical tissue

Report Summary and Recommendation:

Lesions in this line are considered incidental and/or attributable to strain background. Analysis of mice at or before preweaning age would be helpful to determine the cause of preweaning lethality in this line.

Dhcr24 gene is associated with desmosterolosis, a rare autosomal recessive disorder characterized by multiple congenital anomalies and elevated levels of the cholesterol precursor desmosterol in plasma, tissue, and cultured cells in humans. We did not find skeletal and brain abnormalities that are described in this condition (<http://omim.org/entry/602398>).