

#### **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

# **CMHD Pathology Report**

## 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: March 12, 2013

Pathologist: H. Adissu



## **Mouse Genetics Project**

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

email:

MGPenguiries@sanger.ac.uk Mouse Portal Europhenome

**CMHD LabID: N12-1521** 

## **Relevant History:**

Phenotype:

increased susceptibility to bacterial infection decreased circulating glucose level increased circulating alkaline phosphatase level hyperphosphatemia decreased circulating LDL cholesterol level decreased circulating thyroxine level

## AnimalID: M00617598 (Male)

## **Tissue Preservation and Staining:**

The skin is fragmented with marked separation of dermis and hypodermis

## **Histopathology Findings:**

liver (MA:0000358)

## **Histopath Description:**

mild lipidosis

#### **Morphological Diagnosis:**

**Distribution:** multifocal; **Severity:** mild; **MPATH Diagnosis:** steatosis MPATH:622

## **Definitive Diagnosis:**

hepatic lipidosis

## retina (MA:0000276)

## **Histopath Description:**

There are multifocal mild folds of the nuclear layers with occasional clusters of external nuclear structures within the layer of rods and cons.

#### **Morphological Diagnosis:**

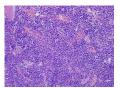
Distribution: Focal; Severity: moderate;

#### **Definitive Diagnosis:**

Retinal folding (dysplasia)



Retinal dysplasia



Bone marrow, normal

## **AnimalID: M00617597 (Male)**

## **Tissue Preservation and Staining:**

The skin is fragmented with marked separation of dermis and hypodermis

## **Histopathology Findings:**

liver (MA:0000358)

# **Histopath Description:**

Diffuse lipidosis

## **Morphological Diagnosis:**

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

#### **Definitive Diagnosis:**

Diffuse hepatic steatosis

## retina (MA:0000276)

## **Histopath Description:**

There are multifocal mild folds along the retina clusters of external nuclear structures within the layer of rods and cons.

## **Morphological Diagnosis:**

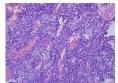
Distribution: Focal; Severity: moderate;

## **Definitive Diagnosis:**

Retinal folding (dysplasia)



Retinal dysplasia



Bone marrow,

## AnimalID: M00340377 (Female)

## **Histopathology Findings:**

liver (MA:0000358)

## **Histopath Description:**

Diffuse lipidosis

## **Morphological Diagnosis:**

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

## **Definitive Diagnosis:**

Diffuse hepatic steatosis

## spleen (MA:0000141)

## **Histopath Description:**

mild erythroid hyperplasia

## **Morphological Diagnosis:**

Distribution: multifocal; Severity: mild; MPATH Diagnosis: extramedullary hemopoiesis

MPATH: 595

#### **Definitive Diagnosis:**

Splenic erythroid hyperplasia

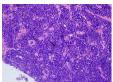
## bone marrow (MA:0000134)

## **Histopath Description:**

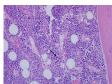
There are increased numbers of erythroid cells with erythroid myeloid ration of 1:2 to 1:3 (normal EM ratio in WT is 1:4 or above). Coarse iron deposites are abundant.

## **Morphological Diagnosis:**

**Severity:** mild; **Definitive Diagnosis:** Erythroid hyperplasia



Erythroid hyperplasia with abundant coarse iron deposites (arrow)



Wild type control: normal marrow, note myeloid dominance and few iron deposites (arrow)

## AnimalID: M00340378 (Female)

## **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

## neck muscle (MA:0000587)

#### **Histopath Description:**

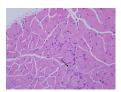
Within the neck muscle are occasional myofibers with hyalinized sarcoplasm and centralized nuclei.

## **Morphological Diagnosis:**

Distribution: multifocal; Severity: mild;

## **Definitive Diagnosis:**

Myodegeneration



Muscle degeneration

## liver (MA:0000358)

## **Histopath Description:**

Diffuse lipidosis

# **Morphological Diagnosis:**

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

## **Definitive Diagnosis:**

Diffuse hepatic steatosis

## retina (MA:0000276)

## **Histopath Description:**

There are multifocal mild folds of the nuclear layers.

## **Morphological Diagnosis:**

Distribution: multifocal; Severity: moderate;

# **Definitive Diagnosis:**

Retinal folding (dysplasia)



Retinal dysplasia

## bone marrow (MA:0000134)

## **Histopath Description:**

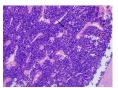
There are increased numbers of erythroid cells with erythroid myeloid ration of 1:2 to 1:3 (normal EM ratio in WT is 1:4 or above). Coarse iron deposites are abundant.

## **Morphological Diagnosis:**

Severity: mild;

## **Definitive Diagnosis:**

Erythroid hyperplasia



Erythroid hyperplasia

## **Report Summary and Recommendation:**

Retinal dysplasia was obsreved in three mice in this line. Erythroid hyperplasia with abundant iron deposites was observed in two female mice. No lesions were found to explain the clinical phenotypes. The thyroid glands and parathyroid glands were unremarkable. Other lesions are considered incidental and attributable to diet or strain background.