

CMHD Pathology Report



CMHD Pathology Core

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ReportID: Report Date: Pathologist: Dr. H. Adissu

Mouse Genetics Project

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

CMHD LabID: N13-578

Relevant History:

Phenotype:

Homozygous partial lethality at P14

AnimalID: M00796558 (Male)

Histopathology Findings:

liver (MA:0000358)

Histopath Description:

diffuse lipidosis

Morphological Diagnosis:

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

Definitive Diagnosis:

hepatic steatosis

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 eosinophlic 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 reminisecent of the zona glomerulosa and zona fasciculate of the mature adrenal gland.

Morphological Diagnosis:

Distribution: focal;

Definitive Diagnosis:

accessory adrenal cortical tissue

thymus (MA:0000142)

Histopath Description:

There is a 50 um diamater epithelial cyst within the medulla.

Morphological Diagnosis:

Distribution: multifocal; **MPATH Diagnosis:** cyst MPATH:62

Definitive Diagnosis:

Epithelial cyst

Histopathology Comments:

This is a developmental abnormality commonly seen in mice.

lymph node (MA:0000139)

Histopath Description:

early lymphoma

Morphological Diagnosis:

MPATH Diagnosis: lymphoid neoplasms MPATH:513

Definitive Diagnosis:

Early lymphoma

Organ/Tissue Analyzed:

Histopathology examination included the following organs and tissues: brain, trigeminal ganglion, eyes, salivary glands, trachea, lungs, heart, thymus, thyroid gland, parathyroid gland, exocrine and endocrine pancreas, oesophagus, stomach, small intestine, large intestine, liver, gall bladder, spleen, kidneys, adrenal gland, lymph nodes, spinal cord, bone marrow, sternum, femur and tibia with associated skeletal muscles, brown fat, pinna, skin, testis, epididymis, seminal vesicle, and prostate.

AnimalID: M00796559 (Male)

Histopathology Findings:

liver (MA:0000358)

Histopath Description:

diffuse lipidosis

Morphological Diagnosis:

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

Definitive Diagnosis:

hepatic steatosis

retina (MA:0000276)

Histopath Description:

Involving one eye, there are clusters of external nuclear structures within the internal and outer plexiform layer.

Morphological Diagnosis:

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

Definitive Diagnosis:

Retinal dysplasia

Organ/Tissue Analyzed:

Histopathology examination included the following organs and tissues: brain, trigeminal ganglion, eyes, salivary glands, trachea, lungs, heart, thymus, thyroid gland, parathyroid gland, exocrine and endocrine pancreas, oesophagus, stomach, small intestine, large intestine, liver, gall bladder, spleen, kidneys, adrenal gland, lymph nodes, spinal cord, bone marrow, sternum, femur and tibia with associated skeletal muscles, brown fat, pinna, skin, testis, epididymis, seminal vesicle, and prostate.

AnimalID: M00796560 (Female)

Histopathology Findings:

liver (MA:0000358)

Histopath Description:

diffuse lipidosis

Morphological Diagnosis:

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

Definitive Diagnosis:

hepatic steatosis

spleen (MA:0000141)

Histopath Description:

marked erythroid hyperplasia

Morphological Diagnosis:

Distribution: multifocal to coalescing; **Severity:** extreme; **MPATH Diagnosis:** extramedullary

hemopoiesis MPATH:595

Definitive Diagnosis:

Splenic erythroid hyperplasia

lymph node (MA:0000139)

Histopath Description:

The mesenteric lymph node is markedly enlarged (greater than four fold). The medulla is particularly expanded by chords and sheets of plasmatoid cells. There are promient germinal centers within the medulla

Morphological Diagnosis:

Distribution: Diffuse; Severity: moderate; MPATH Diagnosis: hyperplasia MPATH:134

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. Early maginal center lymphoma is suspected.

Organ/Tissue Analyzed:

Histopathology examination included the following organs and tissues: brain, trigeminal ganglion, eyes, salivary glands, trachea, lungs, heart, thymus, thyroid gland, parathyroid gland, exocrine and endocrine pancreas, oesophagus, stomach, small intestine, large intestine, liver, gall bladder, spleen, kidneys, adrenal gland, lymph nodes, spinal cord, bone marrow, sternum, femur and tibia with associated skeletal muscles, brown fat, pinna, skin, uterus, oviduct, and ovary, and mammary gland.

AnimalID: M00796561 (Female)

Histopathology Findings:

liver (MA:0000358)

Histopath Description:

diffuse lipidosis

Morphological Diagnosis:

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

Definitive Diagnosis:

hepatic steatosis

lymph node (MA:0000139)

Histopath Description:

The mesenteric lymph node is markedly enlarged (greater than four fold). The medulla is particularly expanded by chords and sheets of plasmatoid cells. There are promient germinal centers within the medulla

Morphological Diagnosis:

Distribution: Diffuse; Severity: moderate; MPATH Diagnosis: hyperplasia MPATH:134

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. Early maginal center lymphoma is suspected.

Organ/Tissue Analyzed:

Histopathology examination included the following organs and tissues: brain, trigeminal ganglion, eyes, salivary glands, trachea, lungs, heart, thymus, thyroid gland, parathyroid gland, exocrine and endocrine pancreas, oesophagus, stomach, small intestine, large intestine, liver, gall bladder, spleen, kidneys, adrenal gland, lymph nodes, spinal cord, bone marrow, sternum, femur and tibia with associated skeletal muscles, brown fat, pinna, skin, uterus, oviduct, and ovary, and mammary gland.

Report Summary and Recommendation:

Lesions in this line are incidental or attributable to diet or strain background. There are no lesions predictive of homozygous lethality at P14. Analysis of preweaning homozygous animals is required to determine cause of mortality.

Recessive ABCA4 gene mutation in humans is associated with various retinal conditions including CONE-ROD DYSTROPHY 3#604116. We did not see any morphological abnormality in the retina except in one mouse with mild retinal dysplasia (heterotopia of external nuclear layer cells). This mild heterotopia is a background lesion C57/B6N strain and is not considered to have causal association with the genotype.