# CMHD Pathology Report

**CMHD Pathology Core**
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**Mouse Genetics Project**
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ReportID: Report Date:
Pathologist: Dr. H. Adissu

**CMHD LabID: N13-579**

**Relevant History:**
Phenotype:

Homozygous viability at P14
Recessive Lethal Study

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**AnimalID: M00895238 (Male)**

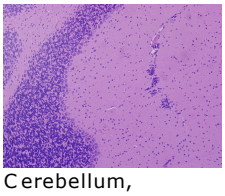
**Histopathology Findings:**

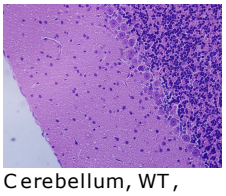
**brain (MA:0000168)**

**Histopath Description:**
There are rare foci of aggregates of granular cells in the within the outer aspect of the molecular layer.

**Morphological Diagnosis:**
**Distribution:** multifocal; **Severity:** mild;

**Definitive Diagnosis:**
Cerebellar granular cell heterotopia


Cerebellum, heterotopic granular cells, 40x, HE


Cerebellum, WT, normal, 40x, HE

**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

**Histopathology Comments:**
Retinal dysplasia is reported as a background lesion in C57BL/6N lines ( Mattapallil et al., 2012).

**liver (MA:0000358)**

**Histopath Description:**
diffuse lipidosis

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

**Definitive Diagnosis:**

hepatic steatosis

### salivary gland (MA:0000346)

**Histopath Description:**
There is focal perivascular mononuclear inflammatory cells.

**Morphological Diagnosis:**
**Distribution:** focal; **Severity:** mild; **MPATH Diagnosis:** inflammation MPATH:212

**Definitive Diagnosis:**
Perivascular inflammatory aggregate

**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.

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## AnimalID: M00895239 (Male)

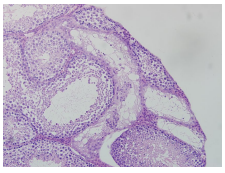
**Histopathology Findings:**

### testis (MA:0000411)

**Histopath Description:**
There is focally extensive vacuolar degeneration and atrophy of the seminiferous tubules.

**Morphological Diagnosis:**
**Distribution:** multifocal; **Severity:** mild;

**Definitive Diagnosis:**
Testicular degeneration and atrophy



Testis, degeneration and atrophy, multifocal, 20x, HE

### 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:**
There is focally extensive retinal folding at the posterior aspect. There are also clusters of external nuclear structures within the internal and outer plexiform layer.

**Morphological Diagnosis:**
**Distribution:** Focal; **Severity:** mild;

**Definitive Diagnosis:**
Retinal dysplasia

**Histopathology Comments:**
Retinal dysplasia is reported as a background lesion in C57BL/6N lines ( Mattapallil et al., 2012).

### lymph node (MA:0000139)

**Histopath Description:**
The mesenteric lymph node is markedly enlarged (greater than five-fold). The medulla is expanded by chords and sheets of plasmatoid cells.

**Morphological Diagnosis:**
**Distribution:** Diffuse; **Severity:** severe; **MPATH Diagnosis:** hyperplasia MPATH:134

**Definitive Diagnosis:**
Lymphoid hyperplasia with medullary plasmacytosis.

**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.

**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.

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**AnimalID: M00895235 (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

**ovary (MA:0000384)**

**Histopath Description:**
There ovarian stroma is markedly expanded.

**Definitive Diagnosis:**
Ovarian stromal hyperplasia

**pancreas (MA:0000120)**

**Histopath Description:**
There is a focal glandular necrosis and chronic active inflammation centered on the necrotic focus.

**Morphological Diagnosis:**
**Distribution:** focal; **Severity:** mild;

**Definitive Diagnosis:**
Focal necrotic pancreatitis

**Histopathology Comments:**
The lesion is considered incidental

**spleen (MA:0000141)**

**Histopath Description:**
Mild erythropoiesis

**Morphological Diagnosis:**
**Distribution:** multifocal to coalescing; **Severity:** mild; **MPATH Diagnosis:** extramedullary hemopoiesis MPATH:595

**Definitive Diagnosis:**
Mild erythropoiesis

**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.

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**AnimalID: M00895236 (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:**
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**Report Summary and Recommendation:**
Focally extensive testicular degeneration and atrophy is observed in one male mouse. Heterotopia of granular cells of the cerebellum is observed in one mouse. 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 mutation in ESCO2 gene in humans is associated with Roberts syndrome & SC Phocomelia Syndrome (#268300). Roberts syndrome (RBS) is characterized by pre- and postnatal growth retardation, severe symmetric limb reduction defects, craniofacial anomalies and severe intellectual deficit. SC phocomelia is a milder form of RBS. Examination of homozygous animals may reveal skeletal abnormalities described in this condition.

Summary:
Testis: Testicular degeneration and atrophy (1/2 males);
Brain, Cerebellum: Granular cell heterotopia (1/4)