



CMHD Pathology Report



CMHD Pathology Core

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ReportID: Report Date: June 18, 2013
Pathologist: Dr. H. Adissu

Mouse Genetics Project

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

CMHD LabID: N13-466

Relevant History:

narrow eye opening
corneal opacity
abnormal cornea morphology
vertebral fusion
abnormal spine curvature
decreased platelet cell number
decreased heart weight
abnormal eye pigmentation
eye opacity [mp:0009859]
abnormal eye size
narrow eye opening
decreased body length
decreased body weight
decreased lean body mass
decreased body weight
kyphosis
scoliosis
partial lethality
abnormal fertility/fecundity
chromosomal instability

AnimalID: M00217387 (Male)

Histopathology Findings:

parathyroid gland (MA:0000128)

Morphological Diagnosis:

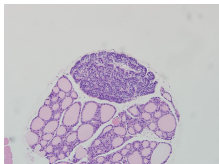
Distribution: unilateral; **Severity:** moderate; **MPATH Diagnosis:** cyst MPATH:62

Definitive Diagnosis:

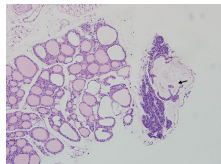
Unilateral Parathyroid cystic degeneration

Histopathology Comments:

The significance of this lesion is uncertain in presence of a normal contralateral gland



Parathyroid, cyst,
20x, HE



Parathyroid,
contralateral, 20x,
HE

liver (MA:0000358)

Histopath Description:

diffuse lipidosis

Morphological Diagnosis:

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

Definitive Diagnosis:

Hepatic lipidosis

testis (MA:0000411)**Histopath Description:**

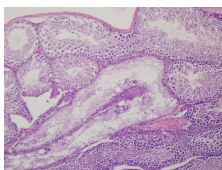
Bilateral testicular degeneration and atrophy affecting 20% of the seminiferous tubule profiles

Morphological Diagnosis:**Distribution:** bilateral; **Severity:** moderate;**Definitive Diagnosis:**

Seminiferous tubules degeneration and atrophy

Histopathology Comments:

Testis is not available from the other male mouse



Testis, seminiferous tubules degeneration and atrophy, 20x, HE

brain (MA:0000168)**Histopath Description:**

There is mild dilation of the lateral ventricles

Morphological Diagnosis:**Distribution:** bilateral; **Severity:** mild;**Definitive Diagnosis:**

Dilation of the brain ventricles

Histopathology Comments:

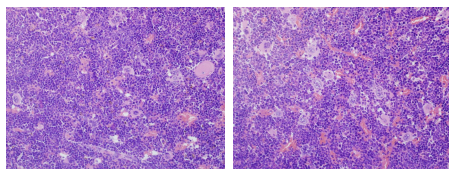
Mild dilation of the lateral ventricles is a background condition in mice of C57BL/6N background (Brayton et al., 2004).

bone marrow (MA:0000134)**Histopath Description:**

The number of megakaryocytes is reduced (compared to wild type).

Morphological Diagnosis:**Severity:** mild;**Definitive Diagnosis:**

Megakaryocyte hypoplasia



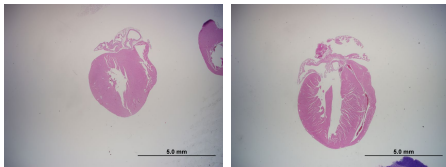
Bonemarrow, megakaryocyte hypoplasia, 40x, HE Bonemarrow, wildtype control, normal, 40x, HE

heart (MA:0000072)**Histopath Description:**

The heart is small compared to that of WT control; there is no evidence of morphological abnormalities in any of the structures examined.

Morphological Diagnosis:**Distribution:** generalized; **Severity:** moderate;**Definitive Diagnosis:**

Cardiac hypoplasia



Heart, hypoplasia,
1.25x, HE

Heart, wildtype
control, normal,
1.25x, HE

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: M00660858 (Female)

Histopathology Findings:

liver (MA:0000358)

Histopath Description:

diffuse lipidosis

Morphological Diagnosis:

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

Definitive Diagnosis:

Hepatic lipidosis

brain (MA:0000168)

Histopath Description:

There is mild dilation of the lateral ventricles

Morphological Diagnosis:

Distribution: bilateral; **Severity:** mild;

Definitive Diagnosis:

Dilation of the brain ventricles

Histopathology Comments:

Mild dilation of the lateral ventricles is a background condition in mice of C57BL/6N background (Brayton et al., 2004).

stomach (MA:0000353)

Histopath Description:

moderate neutrophilic gastritis

Morphological Diagnosis:

Distribution: multifocal to coalescing; **Severity:** moderate;

Definitive Diagnosis:

Gastritis, neutrophilic

bone marrow (MA:0000134)

Histopath Description:

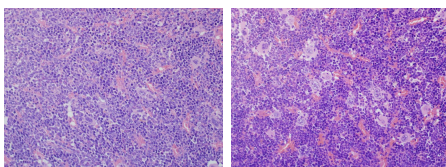
The number of megakaryocytes is reduced (compared to wild type).

Morphological Diagnosis:

Severity: mild;

Definitive Diagnosis:

Megakaryocyte hypoplasia



Bonemarrow,
megakaryocyte
hypoplasia, 40x, HE

Bonemarrow,
wildtype control,
normal, 40x, HE

heart (MA:0000072)**Histopath Description:**

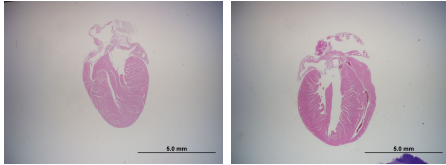
The heart is small compared to that of WT control; there is no evidence of morphological abnormalities in any of the structures examined.

Morphological Diagnosis:

Distribution: generalized; **Severity:** moderate;

Definitive Diagnosis:

Cardiac hypoplasia



Heart, hypoplasia,
1.25x, HE

Heart, wildtype
control, normal,
1.25x, HE

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: M00217389 (Female)**Histopathology Findings:****liver (MA:0000358)****Histopath Description:**

diffuse lipidosis

Morphological Diagnosis:

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

Definitive Diagnosis:

Hepatic lipidosis

bone marrow (MA:0000134)**Histopath Description:**

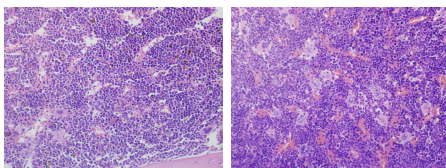
The number of megakaryocytes is reduced (compared to wild type).

Morphological Diagnosis:

Severity: mild;

Definitive Diagnosis:

Megakaryocyte hypoplasia



Bonemarrow,
megakaryocyte
hypoplasia, 40x, HE

Bonemarrow,
wildtype control,
normal, 40x, HE

heart (MA:0000072)**Histopath Description:**

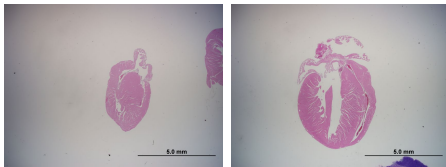
The heart is small compared to that of WT control; there is no evidence of morphological abnormalities in any of the structures examined.

Morphological Diagnosis:

Distribution: generalized; **Severity:** moderate;

Definitive Diagnosis:

Cardiac hypoplasia



Heart, hypoplasia,
1.25x, HE

Heart, wildtype
control, normal,
1.25x, HE

brain (MA:0000168)

Histopath Description:

There is mild dilation of the lateral ventricles

Morphological Diagnosis:

Distribution: bilateral; **Severity:** mild;

Definitive Diagnosis:

Dilation of the brain ventricles

Histopathology Comments:

Mild dilation of the lateral ventricles is a background condition in mice of C57BL/6N background (Brayton et al., 2004).

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: M00217384 (Male)

Histopathology Findings:

liver (MA:0000358)

Histopath Description:

diffuse lipidosis

Morphological Diagnosis:

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

Definitive Diagnosis:

Hepatic lipidosis

brain (MA:0000168)

Histopath Description:

There is more marked dilation of the lateral ventricles; there is rarefaction of the periventricular neuropil

Morphological Diagnosis:

Distribution: bilateral; **Severity:** mild;

Definitive Diagnosis:

Dilation of the brain ventricles

Histopathology Comments:

Mild dilation of the lateral ventricles is a background condition in mice of C57BL/6N background (Brayton et al., 2004).

eye (MA:0000261)

Histopath Description:

There is extensive adhesion of the iris with the cornea (anterior synechia). The cornea is overall thin and is irregular in thickness; the corneal epithelium is reduced to a single cuboidal layer.

Morphological Diagnosis:

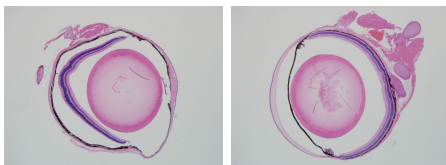
Distribution: unilateral; **Severity:** severe;

Definitive Diagnosis:

Corneal atrophy and anterior synechia

Histopathology Comments:

The lesion may explain the ocular abnormalities detected. Only one eye is present for examination



Eye, corneal atrophy and anterior synechia, 4x, HE

Eye, wildtype control, normal, 40x, HE

bone marrow (MA:0000134)

Histopath Description:

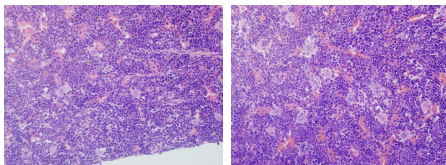
The number of megakaryocytes is reduced (compared to wild type).

Morphological Diagnosis:

Severity: mild;

Definitive Diagnosis:

Megakaryocyte hypoplasia



Bonemarrow, megakaryocyte hypoplasia, 40x, HE

Bonemarrow, wildtype control, normal, 40x, HE

heart (MA:0000072)

Histopath Description:

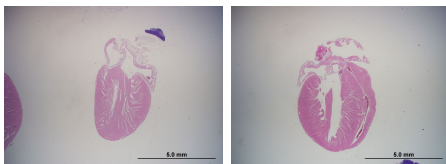
The heart is small compared to that of WT control; there is no evidence of morphological abnormalities in any of the structures examined.

Morphological Diagnosis:

Distribution: generalized; **Severity:** moderate;

Definitive Diagnosis:

Cardiac hypoplasia



Heart, hypoplasia, 1.25x, HE

Heart, wildtype control, normal, 1.25x, HE

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.

Report Summary and Recommendation:

Various lesions that correlate with some of the observed phenotypes are noted in this line.

Corneal lesion may explain the ocular abnormalities. Testicular degeneration and atrophy is consistent with infertility. Megakaryocyte hypoplasia may explain reduced platelet count. Small heart size is consistent with decreased heart weight.

Line summary: Corneal atrophy and anterior synechia (1/4); Testicular degeneration and atrophy (2/2); Megakaryocyte hypoplasia (4/4); Cardiac hypoplasia (4/4)