



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

ReportID: Report Date: February 21,
2014
Pathologist: Dr. H. Adissu

Mouse Genetics Project

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

CMHD LabID: N13-925

Relevant History:

Phenotypes:
decreased circulating alkaline phosphatase level
decreased circulating glucose level
vertebral fusion
abnormal cranium morphology
increased mean platelet volume
increased circulating alanine transaminase level
increased circulating aspartate transaminase level
partial lethality
decreased bone mineral content
decreased bone trabecula number
abnormal craniofacial development
abnormal eye development
fetal edema
abnormal brainstem auditory evoked potential

AnimalID: M00205003 (Male)

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: M00204998 (Male)

Histopathology Findings:

liver (MA:0000358)

Histopath Description:

Severe lipidosis

Morphological Diagnosis:

Distribution: diffuse; **Severity:** extreme; **MPATH Diagnosis:** steatosis MPATH:622; **MPATH Process Term:** lipid deposition MPATH:42

Definitive Diagnosis:

Hepatic lipidosis

Histopathology Comments:

This is dietary steatosis

brain (MA:0000168)

Histopath Description:

There is marked dilation of the lateral ventricles

Morphological Diagnosis:

Distribution: diffuse; **Severity:** severe; **MPATH Diagnosis:** hydrocephalus MPATH:639;

MPATH Process Term: degenerative change MPATH:14

Definitive Diagnosis:

Dilation of the brain ventricles

Histopathology Comments:

Mild to moderate dilation of the ventricles is a background condition in mice of C57BL/6N background

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: M00215317 (Female)

Histopathology Findings:

sternum (MA:0001331)

Histopath Description:

The sternum is moderately curved outwardly

Morphological Diagnosis:

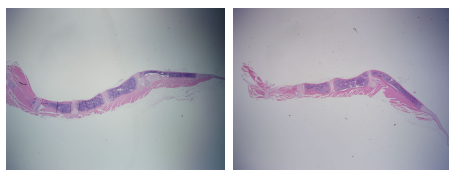
Severity: moderate; **MPATH Process Term:** developmental and structural abnormality MPATH:55

Definitive Diagnosis:

Curved sternum

Histopathology Comments:

This lesion is suggestive of pectus carinatum (outward protrusion of the sternum)



Sternum, outward curving (protrusion), 1.25x, HE. Sternum, Wt, normal, 1.25x, HE.

liver (MA:0000358)

Histopath Description:

Severe lipidosi

Morphological Diagnosis:

Distribution: diffuse; **Severity:** extreme; **MPATH Diagnosis:** steatosis MPATH:622; **MPATH Process Term:** lipid deposition MPATH:42

Definitive Diagnosis:

Hepatic lipidosi

Histopathology Comments:

This is dietary steatosis

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: M00217355 (Female)

Histopathology Findings:

liver (MA:0000358)

Histopath Description:

Severe lipidosi

Morphological Diagnosis:

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

Process Term: lipid deposition MPATH:42

Definitive Diagnosis:

Hepatic lipidosis

Histopathology Comments:

This is dietary steatosis

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:

Outward protrusion or curving of the sternum is present in one mouse. This sternal deformity is consistent with pectus carinatum. In humans, this deformity is associated with vertebral deformities such as scoliosis (as is the case in this mouse line). We did not see morphological correlate to the rest of the phenotypes documented by clinical phenotyping. The auricular structures were not available to rule out otitis or other causes of conductive hearing loss that could explain the abnormal brainstem auditory evoked potential in this line.

Line summary:

Sternum: Sternal deformity (pectus carinatum) - 1/4