PRESS RELEASE

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ISMRD is Pleased to Announce the Outcome of Two ISMRD-Sponsored Mucolipidosis Research Projects

1. *To evaluate AAV Gene Therapy in the Feline Model of ML II.* Allison M Bradbury, Abigail Wexner Research Institute and Ohio State University; and Steven J Gray, University of North Carolina.

The goal of this research project was to evaluate adeno-associated virus (AAV)-mediated gene therapy in the feline model of MLll.

Different doses of AAV gene therapy were tried at different ages, and it was found that treatment with the highest dose of AAV gene therapy at four weeks of age resulted in the greatest correction of visual, skeletal, and cardiac system disease, critically with no evidence of congenital heart disease which is often the cause of fatality in MLII. See the full report here.

2. *To evaluate the cardiovascular manifestations of MLII in a feline colony through genetic sequencing, cardiac ultrasound and pathology.* Allison M Bradbury, Abigail Wexner Research Institute and Ohio State University; Heather Flanagan-Steet, Greenwood Genetic Center; and Joshua Stern, North Carolina State University.

The goal of this study was to identify disease-modifying variants to further characterize the genetics of MLII cardiovascular pathology.

Whole genome sequencing for the renewed MLII colony at the University of California Davis and North Carolina State University has been completed for 15 cats.

The findings of the research underscore the value of the model and the need to clearly outbreed the colony to maintain disease status that is not impacted by inbreeding.
Kittens produced in the present-day MLII colony have cardiovascular lesions of valve thickening and regurgitation consistent with those described in humans.

At this time, a stable, outbred colony of cats that harbor the mutation for MLII in GNPTAB is thriving and available for future research endeavors. See the full report here.

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