



WINN FELINE FOUNDATION

For the Health and Well-being of All Cats

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MARKERS OF ABYSSINIAN/SOMALI AMYLOIDOSIS

PROJECT STUDY: Seeking genetic markers of Abyssinian/Somali amyloidosis

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Final report summary, W16-028

Familiar Amyloidosis is an unresolved diagnostic, prognostic and therapeutic problem in cats. Particularly in Abyssinians, is notorious, due to its sudden juvenile onset and to the difficulties in *intra vitam* identification of the characteristic and diagnostic amyloid deposits. The objective difficulties in the data sample collection and consequently in systematic studies, led to a lack of knowledge on the biological mechanisms that underlie the disease. Abyssinian cats develop a mortal form within the 5-6 year of age range on average by renal failure, being the most targeted organ for the pathogenic aggregates of amyloid proteins mainly the kidney. Some differences in the expressivity of the disease (age of onset, number and type of affected organs) have been recorded, suggesting the genetic background of the cat may affect the clinical presentations.

The joint project of the University of Missouri and University of Milan to discover the genetic basis of Amyloidosis started with the collection of wide cohorts of Abyssinian affected cases and healthy controls, sampled in years, also unlocking the Academic archives in Milan and worldwide and recovering DNA from the stored histological preps from the necropsies (FFPE). The genomic approach has been integrated, consisting in both a case/control association study on hundreds of thousands variant points and in two Abyssinian whole genome sequenced, carried out by Dr. Lyons at University of Missouri. The comparison of the variants identified as possible candidate with the over 100 genomes of 99 Lives Cat Genome Sequencing Project, used as controls, allowed the selection of several interesting mutations. All of them have been genotyped on all the Abyssinian samples available and on many controls of other pure breeds plus random bred cats, to a total of 273 cats. Three gene variants result strongly related to the disease. Analysis of the relationship between the genetic variants combinations and different phenotypes of the disease is on the way to provide an explanation on the disease mechanisms.

Two posters on the study have been presented to date at genetics conferences. No publications are available currently.

Summary prepared by Principal Investigators © 2017

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