



### Notes from Noa Noa Safra DVM PhD

June 2013, An update on the research of Weimaraner inherited diseases at the Bannasch Laboratory:

The search for the genetic cause of HOD in Weimaraners is progressing at the Bannasch Laboratory, at the University of Davis, CA. We are able to continue our work for another year thanks to the most generous donation by Cindy and Bruce Cassidy and the continuous support from the WFF, WCA, and local Weimaraner clubs and members.

So far, the support from the WFF has allowed for sample collection of HOD cases and healthy control Weimaraners at adequate numbers. For the HOD causative variant search, we submitted DNA samples from affected and control Weimaraners for next-generation whole-genome sequencing. This means that the entire genome of these dogs will be sequenced base-by-base to allow a thorough investigation of DNA variants.

The sample collection effort resulted in the identification of two mutations that cause recessive disorders in Weimaraners: Shakey puppy (when the Bannasch laboratory collaborated with Ian Duncan), and Spinal Dysraphism. The later mutation causes a neural tube defect; a congenital condition also termed spina bifida, and when human patients with spina bifida were tested, they had rare damaging variants within the same gene as the Weimaraners. Hence, the research lead to a discovery that will advance the understanding of spina bifida in children as well as will lead to the development of a DNA test for Weimaraners.

The ultimate goal of Dr. Safra at the Bannasch laboratory, is to be able to offer Weimaraner breeders a combined DNA testing panel for mutations in the breed that will include: HUU (Hyperuricosuria), Shakey puppy, Spinal dysraphism, and if the research advances as planned, also HOD.

Thank you for your generous and continuous support!

Sincerely,  
Noa Safra, DVM, PhD.