Genetic Diversity Testing for Giant Schnauzers

Overview

The Veterinary Genetics Laboratory (VGL), in collaboration with Dr. Niels C. Pedersen and staff, has developed a panel of short tandem repeat (STR) markers that will determine genetic diversity across most of the genome and in the Dog Leukocyte Antigen (DLA) class I and II regions. This test panel will be useful to breeders who wish to track and increase genetic diversity of their breed as a long-term goal.

Genetic diversity testing of Giant Schnauzer is now in the data collection phase. During this phase, we will continue to test more registered dogs to build genetic data necessary to provide breeders with an accurate assessment of genetic diversity in their breed. We are accepting dogs from the USA and Canada, as well as from other regions of the world. At the time of writing the report below we had tested 133 Giant Schnauzers – 106 from the US and Canada, and 27 from Europe (mainly UK). Although this number of dogs will probably cover 95% or more of the genetic diversity that exists in dogs from North America, the report will be updated when no new genomic alleles or DLA haplotypes are being recognized.

Price: $80
Price reduced to $70 when combined with a diagnostic test.

ORDER TEST KITS
Allow 5-10 business days for results.

Results reported as:
Short tandem repeat (STR) loci: A total of 33 STR loci from across the genome were used to gauge genetic diversity within an individual and across the breed. The alleles inherited from each parent are displayed graphically to highlight heterozygosity, and breed-wide allele frequency is provided.

DLA haplotypes: STR loci linked to the DLA class I and II genes were used to identify genetic differences in regions regulating immune responses and self/non-self-recognition. Problems with self/non-self-recognition, along with non-genetic factors in the environment, are responsible for autoimmune disease, allergies, and immunodeficiency.

Internal Relatedness: The IR value is a measure of genetic diversity within an individual that takes into consideration both heterozygosity of alleles at each STR loci and their relative frequency in the population. It is also an estimate of the genetic relatedness of a dog's parents. Unlike standard genetic assessments, IR puts more emphasis on heterozygosity over homozygosity and uncommon over common alleles. IR values are unique to each dog and cannot be compared between dogs. Two dogs may have identical IR values but with very different genetic makeups

I. Introduction

A. Breed history

The Giant Schnauzer is the largest of the three breeds of Schnauzer, the two other breeds being the Standard and Miniature Schnauzer. The breed is said to arise from Swabia in the German state of Bavaria, and Württemberg in the 17th century. Numerous breeds were purportedly used by key breeders to develop the Giant Schnauzer, including Oberlanders, black Great Danes, Rottweilers, Dobermans, Boxers, Bouvier des Flandres, Thuringian Shepherds, and Standard Schnauzer. However, the original Giant Schnauzers were considered a rough-coated version of the German pinscher breeds, a necessity against the harsh German winters.

The first Giant Schnauzers were imported to America in the 1930s, mainly from German stock, but the breed remained uncommon until the 1960s, their popularity being superseded by a peak of interest in German shepherd dogs. In 1962, there were 23 new Giant Schnauzers registered with the American Kennel Club; in 1974 this number was 386; in 1984 it was over 800; and at the highpoint in 1987, it was around 1000 dogs. The breed was ranked 94th in AKC registrations in 2011 and 80th in 2017.

The breed was used in olden times on farms for herding and guarding livestock and in cities as personal and property guard dogs. In modern times, the breed has become more dual in function. In Europe, the breed is considered to be more of a working than show dog and used for police work, obedience and agility trials, herding, carting, search and rescue, and personal protection and companionship (Schatzhund). However, the German Shepherd is the prized breed for Schutzhund training.

The decades between the 1930s and 1960s was a quiet period lacking the breeding practices that plague any breed that becomes too popular too fast. The few breeders of Giant Schnauzers during this period concentrated on the dogs that were in the country and made little to publicize and promote the breed. The Giant Schnauzer Club of America was founded in 1962 and because of interest in the breed their number has steadily increased ever since. This increased interest has led to the importation of new dogs from Europe. This renewed interest was concentrated mainly in the show ring, where the breed has performed very well. However, as with many working breeds, interest in obedience and police work has risen and an American-bred dog has even become a Schutzhund III titleholder.

The rapid increase in the numbers of Giant Schnauzers being registered by the AKC has caused many breeders of the quiet period to voice concern that the quality of the breed cannot be sustained. This concern is one reason for the effort to document the genetic diversity of the breed with DNA testing. This information will hopefully provide a baseline on which to guide breeding practices into the future. The minimum goal should be to maintain present genetic diversity and to seek out small pockets of additional diversity wherever it may exist in the world.

B. Breed standard and appearance

According to AKC standards, a well-bred Giant Schnauzer closely resembles the Standard Schnauzer, but only bigger. Their size should be imposing, as indicated by their breed name. Males stand as high as 27.5 inches at the shoulder and can weigh 95 pounds. The body, as befitting giants, should be substantial and muscular. Giant Schnauzers are supposed to present as bold and valiant. The double coat is either solid black or "pepper and salt." A harsh beard and eyebrows are features shared by Mini, Standard, and Giant Schnauzers. The Giant Schnauzer, as both a working and show dog, is both compelling to look at and smart. Giant Schnauzers were originally bred to be an all-around worker, helping on the farm with carting, herding, protecting its territory and humans.

II. Baseline genetic diversity testing and what it tells us about the Giant Schnauzer

A. Population genetics based on 33 STR loci on 25 chromosomes

1. Allele and allele frequencies for each of the 33 STR loci

STR markers are highly polymorphic and have great power to determine genetic differences among individuals and breeds. The routine test panel contains 33 STRs that are recommended for universal parentage determination for domestic dogs by the International Society of Animal Genetics (ISAG) with additional markers developed by the VGL for forensic purposes

Thirty-three STRs and their alleles were studied in 133 Giant Schnauzers (Table 1). Allele and allele frequencies were used to determine basic genetic parameters such as the number of alleles found at each STR locus (Na), the number of effective alleles (Ne) per locus, i.e., the number of alleles that contribute most to genetic differences, the observed or actual heterozygosity (Ho) that was found, and the heterozygosity that would be expected (He) if the existing population is in Hardy Weinberg equilibrium (HWE). HWE is achieved when the selection of mates is entirely random and subject to no positive or negative human selection pressure. The value F is a coefficient of inbreeding derived from the Ho and He values. A value of +1.0 would occur only if every individual were genetically indistinguishable at each of the 33 STR loci, while a value of -1.0 would be seen when all the dogs were completely different at each of the 33 loci.

The 33 STR loci chosen from 25/38 different autosomes were quite polymorphic with 3-12 alleles per locus (Table 1). The frequency that each allele occurs is an indication of the degree of artificial selection. If dogs are not subject to any selection pressures by people, the number of alleles at each locus would be higher and the frequency of each allele more equal. However, this is not the case with pure breeds of dogs, which are subject almost entirely to human selection. This human selection is indicated by the comparative frequency of individual alleles at each locus. In the case of pure breed dogs, one or two alleles will always occur at a much higher frequency than others. These high frequency alleles are highlighted in Table 1. A single allele is found to be shared by one-third to one-half or more of the dogs. A single allele at locus LEI004 and REN105L03 occured at a frequency of 82 and 91%, respectively, indicating a region of the genome that has been under strong positive selection since founding of the breed (Table 1).

Table 1. Allele designation and frequency at 33 STR loci for Giant Schnauzers. The allele that occurs at the highest frequency at each locus is highlighted.

Table 1 Link

2. Standard genetic assessment for each of the 33 STR loci

Heterozygosity is a measure of how often two different alleles occur at the same locus on the same chromosome- one chromosome being inherited by the sire and one by the dam. Observed heterozygosity (Ho) is based on the actual allele frequencies at each STR locus and their distribution, while the expected heterozygosity (He) of a locus is the value that would be predicted if allele frequencies at a specific locus were in Hardy-Weinberg equilibrium (HWE). HWE is achieved when all alleles at a specific locus are segregating randomly. A Ho value of 1.0 would be observed when alleles at each locus are unique to everyone in the population. A Ho value of 0.00 would occur if there is no heterozygosity, e.g. every individual has the same alleles at a given locus.

Ho in each of STR locus ranged from 0.165 to 0.850, indicating a large range of genetic diversity from one locus to another, while the He ranged from 0.176 to 0.851 (Table 2). The Ho and He values were used to calculate the F value (1-Ho/He), a measure of deviation from HWE. F values ranged from -0.105 to +0.280. Nine loci had F values greater than 0.100 (AHTh171-A, AHTh171-A, AHTk211, AHTk253, INU030, REN169O18, REN247M23, REN54P11 and REN64E19), while only AHT137 and AHTh260 had F values less than 0.100. This indicated that many of the loci were under strong positive selection for the same allele.

Table 2. Standard Genetic Assessment for Giant Schnauzers using 33 STR loci (Updated October 09, 2019)

#	Locus	N	Na	Ne	Ho	He	F
1	**AHT121**	213	13	5.575	0.761	0.821	0.073
2	**AHT137**	213	8	2.018	0.526	0.504	-0.042
3	**AHTH130**	213	9	4.353	0.765	0.770	0.007
4	**AHTh171-A**	213	7	3.466	0.742	0.711	-0.043
5	**AHTh260**	213	7	3.218	0.775	0.689	-0.124
6	**AHTk211**	213	4	2.550	0.474	0.608	0.220
7	**AHTk253**	213	6	3.816	0.704	0.738	0.046
8	**C22.279**	213	5	2.701	0.582	0.630	0.076
9	**FH2001**	213	7	3.200	0.592	0.688	0.140
10	**FH2054**	213	7	4.231	0.775	0.764	-0.014
11	**FH2848**	213	8	2.880	0.648	0.653	0.007
12	**INRA21**	213	6	3.412	0.634	0.707	0.103
13	**INU005**	213	7	1.608	0.338	0.378	0.106

14	**INU030**	213	5	4.147	0.718	0.759	0.053
15	**INU055**	213	6	3.334	0.620	0.700	0.115
16	**LEI004**	213	5	1.459	0.310	0.315	0.015
17	**REN105L03**	213	5	1.196	0.146	0.164	0.111
18	**REN162C04**	213	5	3.970	0.690	0.748	0.077
19	**REN169D01**	213	6	1.334	0.183	0.250	0.269
20	**REN169O18**	213	7	4.649	0.746	0.785	0.049
21	**REN247M23**	213	4	2.557	0.606	0.609	0.005
22	**REN54P11**	213	7	3.156	0.615	0.683	0.100
23	**REN64E19**	213	4	2.930	0.634	0.659	0.038
24	**VGL0760**	213	9	2.073	0.451	0.518	0.129
25	**VGL0910**	213	11	5.455	0.831	0.817	-0.018
26	**VGL1063**	213	13	4.711	0.728	0.788	0.076
27	**VGL1165**	213	13	6.238	0.808	0.840	0.038
28	**VGL1828**	213	10	5.018	0.770	0.801	0.038
29	**VGL2009**	213	7	2.455	0.563	0.593	0.049
30	**VGL2409**	213	9	4.412	0.761	0.773	0.017
31	**VGL2918**	213	11	5.981	0.831	0.833	0.002
32	**VGL3008**	213	9	5.408	0.779	0.815	0.044
33	**VGL3235**	213	5	3.152	0.634	0.683	0.072

3. Using allele frequency data to do standard genetic assessments of the entire population.

A standard genetic assessment was made from allele frequency data for all 33 STR loci (Table 1) for all 133 Giant Schnauzers that were tested (Table 3). The average number of alleles (Na) per loci was 6.85 and the number of effective alleles (Ne) was 3.56. These values were within the range of most pure breeds that have been tested. The observed heterozygosity of alleles across the 33 STR loci was 0.632 and the heterozygosity expected (He) if the alleles were in Hardy-Weinberg equilibrium (HWE) was 0.663 (i.e., if the population was randomly breeding). An inbreeding coefficient was calculated based on the differences in He and Ho and in this case, F was 0.052. A value of -1.0 would mean that no dog in the population shared alleles, while a value of +1.0 would mean that all the dogs were genetically the same. This F value was only slightly positive, indicating that the population was in a reasonably random bred state. However, this is an average of the entire population and does not measure the degree to which an individual dog is inbred (see IR values).

Table 3. Summary of Standard Genetic Assessment for Giant Schnauzer using 33 STR loci (Updated October 09, 2019)

	N	Na	Ne	Ho	He	F
Mean	213	7.424	3.535	0.628	0.660	0.056
SE		0.445	0.235	0.030	0.029	0.013

B. Differences in population structure as determined by principal coordinate analysis (PCoA)

1. Genetic relationships of Giant Schnauzers from across the USA

Principal coordinate analysis (PCoA) uses genetic distance based on allele sharing to graph genetic differentiation between individuals in related or unrelated populations. The resulting data is multi-dimensional (spherical) but is usually portrayed in two dimensions by selecting the two coordinates (planes of the sphere) that represent the greatest proportions of individuals. This usually includes coordinates 1 and 2. We tested 133 Giant Schnauzers from several countries in North America and Europe. The Giant Schnauzers that were tested belong to a single breed based on their position within the larger square, but they are divided into two closely related populations (varieties or bloodlines) by the four quadrants.

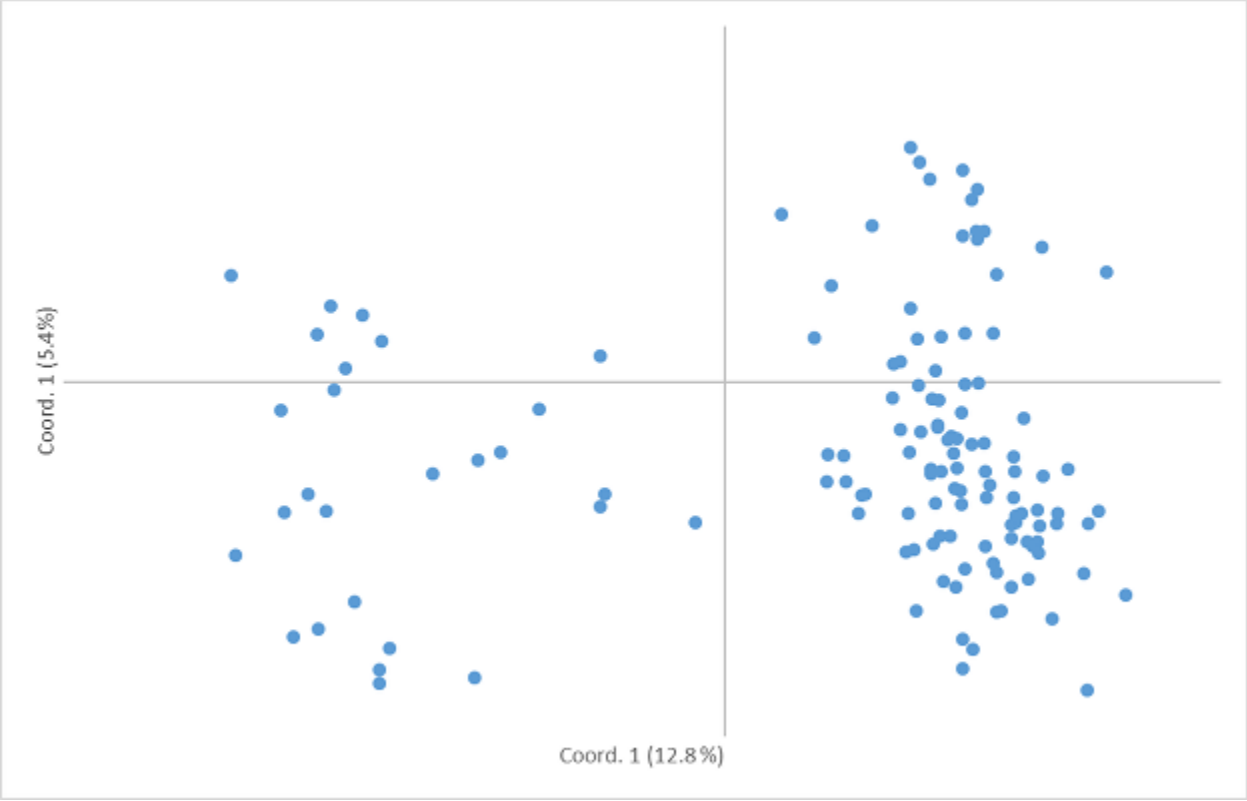

Fig. 1. PCoA of Giant Schnauzer (n=133) based on the 33 STRs

We wanted to see if this segregation was geographical by identifying North American and European dogs in the same PCOA as shown above in figure 1. The main population of Giant Schnauzers contained equal proportions of European and North American dogs (Fig. 2). This tended to be true to the smaller population as well, although the number of European dogs in this minor variety was small.

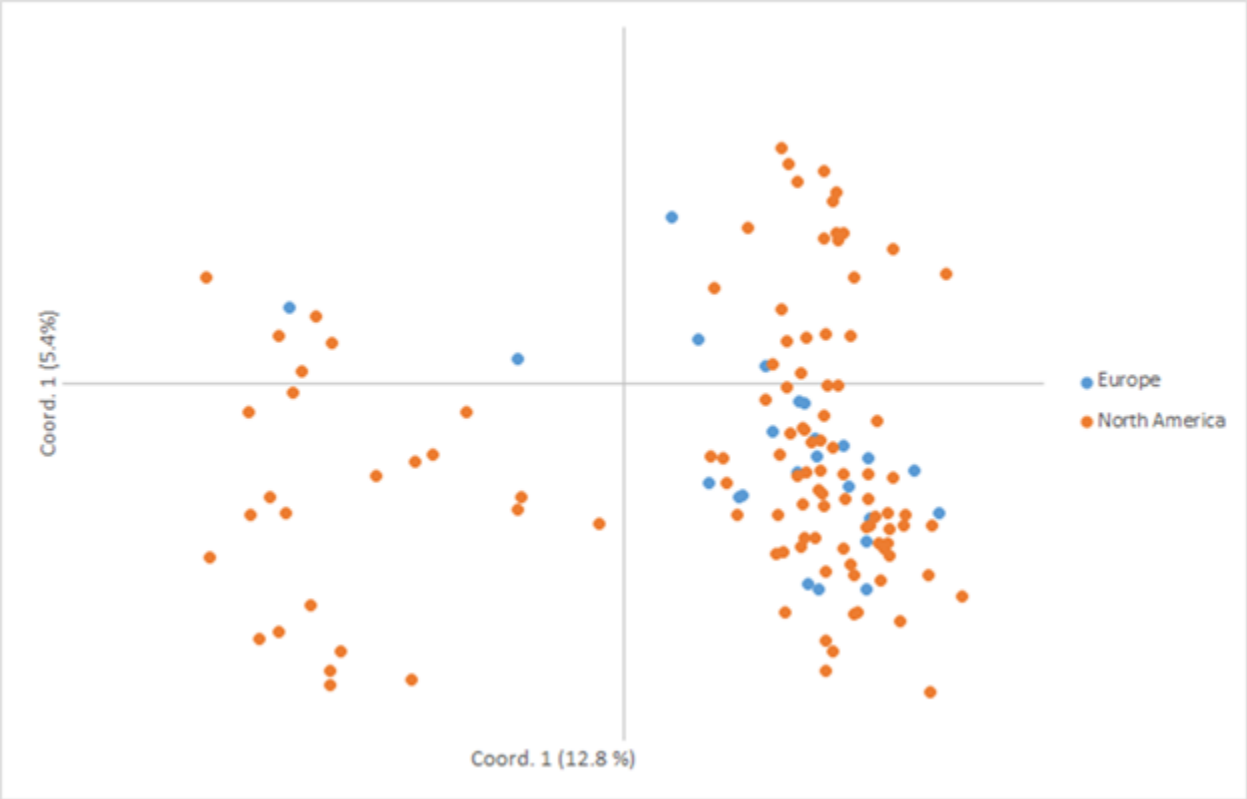

Fig. 2. PCoA of Giant Schnauzer (n=133) based on the 33 STRs. Dogs from Europe (blue) and North America (orange) were identified.

The next comparison was between Black and Pepper and Salt dogs, which are known to be bred somewhat separately (Fig. 3). This PCOA plot clearly demonstrated that black and pepper/salt colored dogs were distinct varieties that have been closely maintained as separate “breeds.”

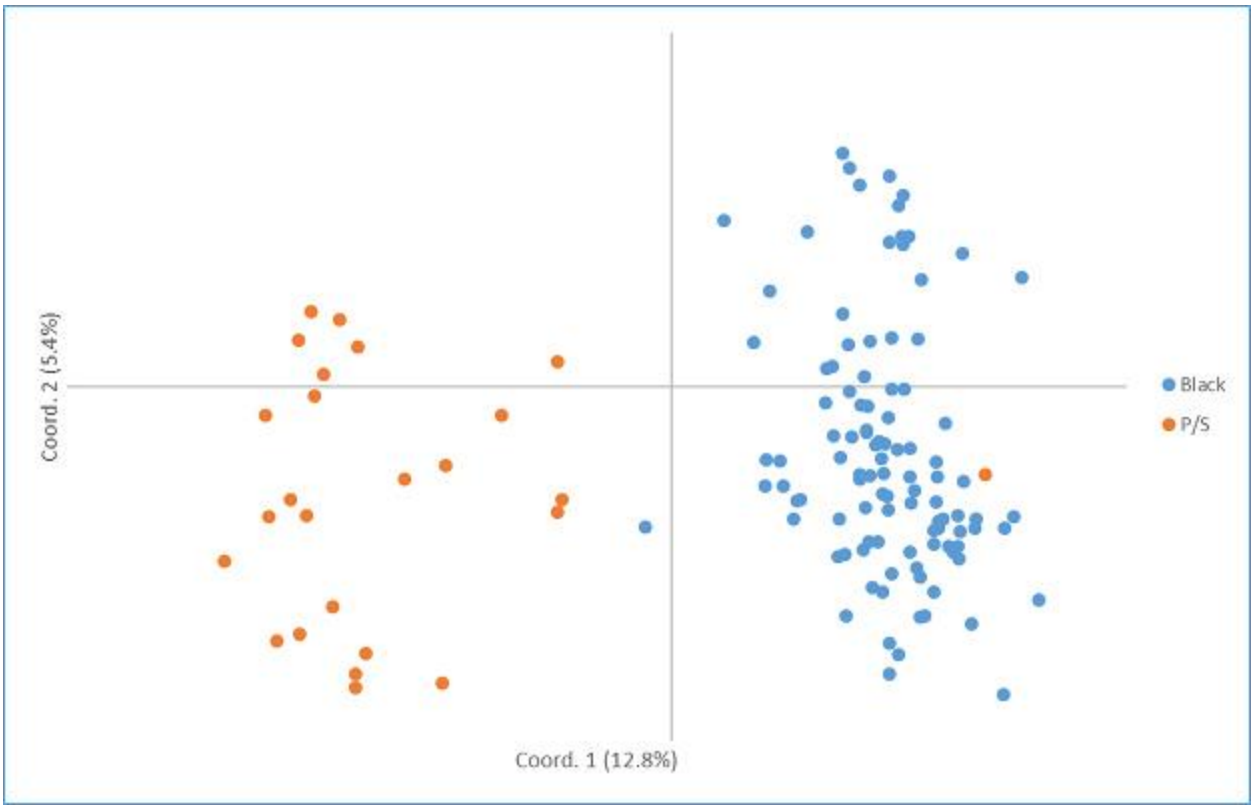

Fig. 3. PCoA of Giant Schnauzer (n=129, 4 did not report color) based on the 33 STRs showing dogs identified as Black versus those identified as Pepper and Salt.

III. The use of genomic allele frequencies to determine internal relatedness

A. Internal relatedness (IR) of individuals and the population as a whole

1. IR values

Genetic assessments such as those presented in Tables 1-3 are indicators of population-wide heterozygosity and do not reflect the genetic diversity of individuals within the population. The genetic diversity of an individual dog is largely determined by the diversity inherited from each of its parents. Internal Relatedness (IR) is a calculation that has been used to determine the degree to which the two parents were related (Table 4). The IR calculation takes into consideration homozygosity at each locus and gives more importance to rare and uncommon alleles. Rare and uncommon alleles would presumably be present in less related individuals. IR scores of all individuals in a population can be graphed to form a curve ranging from -1.0 to +1.0. A dog with a value of -1.0 would have parents that were totally unrelated at all 33 STR loci, while a dog with an IR value of +1.0 has parents that were genetically identical at all loci. An IR value of +0.25 would be equivalent to offspring of full sibling parents from a random breeding population. IR values >0.25 occur when the parents of the full sibling parents were themselves highly inbred.

IR scores ranged from a low of -0.181 (parents least related) to a high of 0.486 (parents most related), with a mean (average) value of 0.047. Therefore, one fourth of the population had IR scores from -0.041 to -0.181, and one fourth +0.139 to 0.486. Although the standard genetic assessments made from allele frequencies indicated that the population was randomly breeding, the values were an average of all dogs and may be misleading. The IR value looks at allele frequency for each individual and gives more weight to dogs that have rarer (less frequent) alleles. Standard genetic assessment weights all alleles the same. IR values show that there are three populations among the 133 Giant Schnauzers, one quarter containing individuals from very unrelated parents, one half with parents of average relatedness, and one fourth with parents that are quite related.

Table 4. IR vs IRVD comparison for Giant Schnauzers (n=133)

	IR	**IRVD**
Min	-0.1808	-0.0169
1st Qu	-0.0412	0.1743
Mean	0.0469	0.2643
Median	0.0445	0.2610
3rd Qu	0.1387	0.3505
Max	0.4855	0.6746

2. Estimation of genetic diversity lost during breed creation using village dogs as a gold standard

The IR values can also be used to give an approximation of how much genetic diversity has been lost during breed development and subsequent evolution. This is done by comparing the frequency of a given allele in Giant Schnauzers with the frequency of the same alleles in a population of village dogs from the Middle East, SE Asia, Taiwan and other Pacific island nations such as Brunei and the Philippines. Contemporary village dogs are largely unchanged from the ancestors of almost all modern dog breeds. The resultant frequencies are then used to calculate the IRVD.

A comparison of IR values (red curve) and IRVD values (blue curve) can be used as a rough estimate of how much of the genetic diversity available in contemporary village dogs still exists in contemporary Giant Schnauzers. A rough estimate based on areas under the curve (black), indicates that Giant Schnauzers possess 43% of the genetic diversity still present among indigenous dogs. This value is like many other pure breeds of dogs.

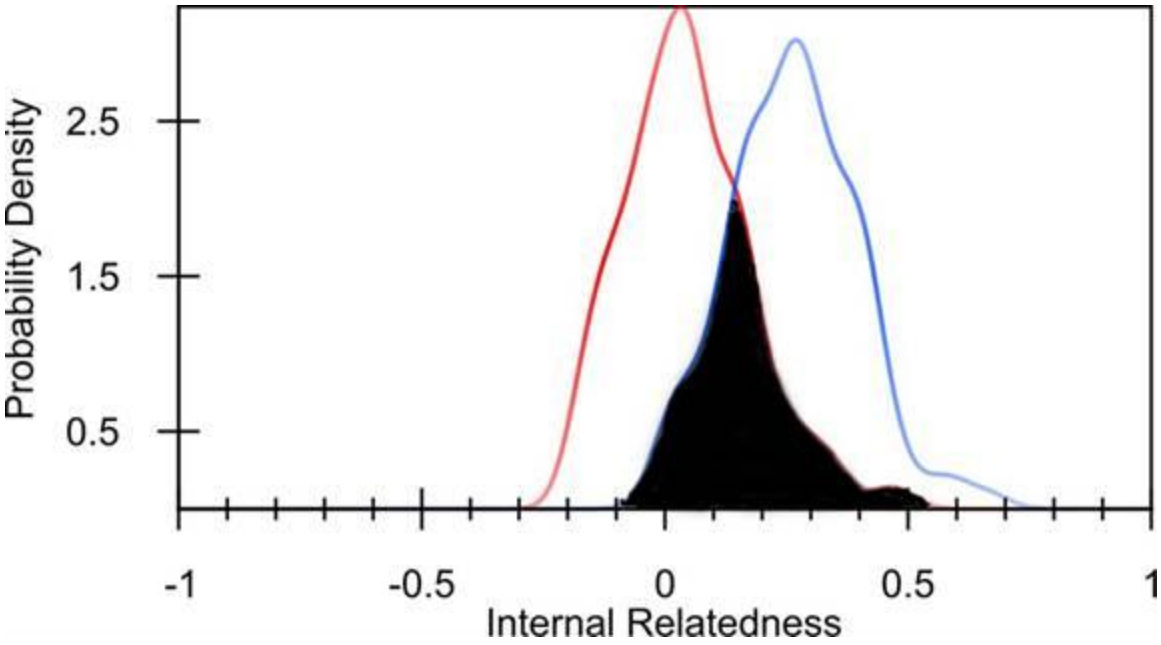

Fig. 2. Distribution of IR estimated in Giant Schnauzer (n=133) based on intra-breed diversity (red), compared with IR adjusted to diversity lost during breed development (blue). Lost diversity was determined by comparing allele frequencies at the same loci between Giant Schnauzers and village dogs from the Middle East, SE Asia, and the Pacific Islands. Village dogs were the most diverse population studied. The black-shaded area represents genetic diversity in village dog that is still found in Giant Schnauzers.

IV. DLA Class I and II Haplotype frequencies and genetic diversity

The DLA consists of four gene rich regions (classes I-IV) making up a small part of canine chromosome 12. Two of these regions contain genes that help regulate normal cell- (Class I) and antibody-mediated (Class II) immunity. Polymorphisms in these regions have also been associated with abnormal immune responses responsible for autoimmune diseases. The Class I region contains several genes, but only one, DLA-88, is highly polymorphic (with many allelic forms) and is therefore most important for immune regulation. Specific alleles at the four STR loci associated with the DLA88 are linked together in various combinations, forming specific haplotypes (Table 4). Groups of genes and their alleles inherited as a block, rather than singly, are called haplotypes. The class II region also contains several genes, three of which are highly polymorphic, DLA-DRB1, DLA-DQB1 and DLA-DQA1. Specific alleles at STR loci associated with each of the three Class II genes are strongly linked and inherited as a single block or haplotype (Table 5). One haplotype comes from each of the parents. Specific class I and II haplotypes are often linked to each other and inherited as a genetic block with limited recombination over time. Therefore, DLA class I and II haplotypes can be viewed as reasonable surrogate markers for breed founders.

The STR-based haplotype nomenclature used in this breed diversity analysis is based on numerical ranking with the first haplotypes identified in Standard Poodles being named 1001, 1002, ... for class I haplotypes and 2001, 2002, ... for class II haplotypes. It is common for various dog breeds to share common and even rare haplotypes, depending on common ancestry.

1. DLA class I and II haplotypes existing in Giant Schnauzers

Giant Schnauzers possess 14 DLA class I haplotypes and 15 class II haplotypes. The frequencies for the various class I and II haplotypes vary widely, but none appear to dominate all others as is so common in other purebreds. The three most common DLA class I haplotypes are 1014 (0.252), 1092 (0.233) and 1159 (0.173) and are collectively found in 66% of the dogs. The three most common DLA class II haplotypes are 2006 (0.162), 2037 (0.320), 2050 (0.165) and occur collectively in 65% of dogs. A dog or dogs with these haplotypes were important founders of the breed.

Tables 4 & 5. DLA class I and Class II haplotype and their frequencies

DLA Class I Haplotype Frequencies (Updated Oct 9, 2019)		
DLA1 #	**STR types**	**Giant Schnauzer (n=213)**
1006	387 375 293 180	0.052
1008	386 373 289 182	0.047
1011	376 365 281 180	0.012
1014	375 373 287 178	0.254
1016	382 371 277 178	0.035
1017	386 373 289 178	0.094
1054	382 379 277 184	0.005
1065	380 371 277 181	0.002
1068	380 373 287 181	0.035
1091	381 371 277 181	0.045
1092	376 379 277 181	0.249
1129	382 371 277 181	0.002
1159	395 379 277 181	0.167
1181	381 371 277 182	0.002

DLA Class II Haplotype Frequencies (Updated Oct 9, 2019)		
DLA2 #	**STR types**	**Giant Schnauzer (n=213)**
2001	343 324 284	0.007
2003	343 324 282	0.038
2005	339 322 280	0.009
2006	339 325 280	0.153
2007	351 327 280	0.052
2012	345 322 280	0.012
2014	339 322 284	0.002
2022	339 327 282	0.005

2031	339 322 282	0.035
2033	339 323 282	0.047
2037	341 327 280	0.347
2050	341 327 284	0.153
2053	343 324 280	0.042
2060	343 323 284	0.002
2083	339 324 282	0.002
2090	339 322 278	0.094

2. DLA class I and II haplotype sharing between Giant Schnauzers and other breeds

DLA haplotypes are in strong linkage disequilibrium and represent a large block of genes that undergo limited recombination and inherited by descent from one generation to another. Therefore, they have some value in determining breed evolution. All the DLA class I and II haplotypes observed in this group of Giant Schnauzers have also been found in other breeds. Therefore, there are no haplotypes unique to Giant Schnauzers, which is unlike most other breeds. There is also no single dominant class I and II haplotype in Giant Schnauzers that points to a single important founder or founder line, as is the case with many other smaller breeds. This extensive sharing of DLA haplotypes with other breeds indicates that Giant Schnauzers were derived from many breeds during the period from earliest conception to registration and theoretical closure to further introgressions.

Many of the DLA class I and II haplotypes found in Giant Schnauzers are also found in several other breeds at low frequency (Table 6). However, the major 1014 DLA class I haplotype (0.252) is also very common in the Alaskan Klee Kai (0.366), which was unexpected. The common 1092 DLA class I haplotype is very common in the Black Russian Terrier and Japanese Akita, which might be expected. The DLA class II 2006 haplotype is common in both Giant Schnauzer (0.162) and the Magyar Agar (0.240), while the 2037 (0.320) haplotype is common in Alaskan Klee Kai (0.377), Black Russian Terrier (0.289), American (0.165) and Japanese Akita (0.261). The dominant 2050 haplotype (0.320) is found in many breeds at low frequency

Table 6. A chart of DLA class I and II haplotypes that have been identified in many dog breeds. These haplotypes are limited in number among all dogs, wolves and coyotes and are inherited by descent through generations. Various breeds will inherit a portion of these haplotypes from their founders. Some haplotypes will be unique to a breed, but most are shared at different incidences between breeds.

DLA Class I Haplotype Frequencies (Updated Apr 27, 2018)

DLA1 #	STR types	Alaskan Klee Kai (n=451)	Black Russian Terrier (n=109)	Shiloh Shepherd (n=115)	Labrador Retriever (n=125)	Magyar Agar (n=33)	American Akita (n=79)	Japanese Akita (n=305)	Blend Akita (n=49)	Golden Retriever (n=690)	Doberman Pinscher (n=410)	Flat Coated Retriever (n=338)	Havanese (n=367)	Samoyed (n=278)	Miniature Poodle (n=242)	Swedish Vallhund (n=102)	Poodle (n=2050)	Giant Schnauzer (n=133)	Polish Lowland Sheepdog (n=16)	English Bulldog (n=163)	Biewer (n=93)	Biewer Yorshire Terrier (n=50)	Biewer Terrier (n=49)	Yorkshire Terrier (n=16)	Italian Greyhound (n=679)
1006	387 375 293 180	--	0.032	--	0.028	--	0.044	--	--	0.0145	--	--	0.045	0.004	0.002	0.265	0.0478	0.056	--	0.003	--	--	--	--	--
1008	386 373 289 182	0.061	--	--	0.072	--	--	--	--	0.0014	--	--	--	--	--	--	0.0198	0.034	--	0.006	0.011	--	--	0.06	0.1355
1011	376 365 281 180	0.059	--	--	--	--	--	--	--	0.0007	--	--	--	0.246	0.004	--	0.021	0.015	--	--	--	--	--	--	--
1014	375 373 287 178	0.366	0.041	--	--	--	--	--	--	0.0399	--	--	0.033	0.002	0.004	--	0.0076	0.252	--	--	0.016	0.01	0.08	0.09	--
1016	382 371 277 178	--	0.009	--	0.012	--	--	--	--	0.0014	0.015	--	0.222	--	0.023	--	0.0146	0.038	--	0.095	0.027	0.02	0.01	0.03	0.0633
1017	386 373 289 178	--	--	--	0.036	--	--	--	--	--	0.087	0.454	--	--	--	--	0.0034	0.09	--	--	--	--	--	--	--
1054	382 379 277 184	--	--	--	0.088	0.14	--	--	--	--	--	0.12	0.109	--	0.002	--	--	0.008	--	--	--	--	--	--	0.0162
1065	380 371 277 181	--	--	--	0.384	--	--	--	--	0.2609	--	0.001	--	--	--	--	--	0.004	--	--	--	--	--	--	0.0007
1068	380 373 287 181	--	--	0.248	0.044	--	--	--	--	0.05	--	0.265	0.016	0.029	0.014	0.358	--	0.038	--	--	--	0.01	--	--	--
1091	381 371 277 181	--	0.505	--	--	--	--	--	--	--	0.001	--	--	--	--	--	--	0.053	--	--	--	--	--	--	--
1092	376 379 277 181	--	0.252	--	--	--	--	0.287	0.01	--	--	--	0.086	--	--	--	0.0002	0.233	--	0.006	--	0.04	--	0.06	--
1129	382 371 277 181	--	--	--	--	--	--	--	--	0.0007	--	--	--	--	--	--	--	0.004	--	--	--	--	--	--	--
1159	395 379 277 181	--	--	--	--	--	--	--	--	--	0.001	--	--	--	--	--	--	0.173	--	--	--	--	--	--	--
1181	381 371 277 182	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	0.004	--	--	--	--	--	--	--

DLA Class II Haplotype Frequencies (Updated Apr 27, 2018)

DLA2 #	STR types	Alaskan Klee Kai (n=451)	Black Russian Terrier (n=109)	Shiloh Shepherd (n=115)	Labrador Retriever (n=125)	Magyar Agar (n=33)	American Akita (n=79)	Japanese Akita (n=305)	Blend Akita (n=49)	Golden Retriever (n=690)	Doberman Pinscher (n=410)	Flat Coated Retriever (n=338)	Havanese (n=367)	Samoyed (n=278)	Miniature Poodle (n=242)	Swedish Vallhund (n=102)	Poodle (n=2050)	Giant Schnauzer (n=133)	Polish Lowland Sheepdog (n=16)	English Bulldog (n=163)	Biewer (n=93)	Biewer Yorshire Terrier (n=50)	Biewer Terrier (n=49)	Yorkshire Terrier (n=16)	Italian Greyhound (n=679)
2001	343 324 284	--	--	--	0.004	0.14	--	--	--	0.1406	--	--	0.041	--	0.017	--	0.6032	0.008	--	--	--	--	--	--	--
2003	343 324 282	--	--	--	0.02	--	--	--	--	0.0225	--	0.129	0.223	0.013	0.504	--	0.091	0.03	--	0.598	0.226	0.2	0.21	0.38	0.0066
2005	339 322 280	--	0.018	--	0.06	--	--	--	--	0.0159	--	0.422	0.003	--	--	--	0.0232	0.004	--	0.015	0.027	0.01	0.09	--	--
2006	339 325 280	--	--	--	--	0.24	--	--	--	--	--	--	0.004	--	--	--	0.0312	0.162	--	--	--	--	--	--	--
2007	351 327 280	0.01	0.032	--	0.032	--	0.044	--	--	0.0145	--	--	0.049	0.004	--	0.275	0.0146	0.056	--	--	--	--	--	--	--
2012	345 322 280	0.063	--	--	--	0.12	--	0.002	--	0.0007	--	--	0.005	--	0.066	--	0.0049	0.015	--	--	--	--	--	--	--
2014	339 322 284	0.074	--	--	--	--	--	--	--	--	--	0.033	0.01	--	0.023	--	0.0134	0.004	0.59	0.092	--	--	--	--	--
2022	339 327 282	--	--	0.057	0.088	--	--	--	--	0.0007	0.002	0.124	0.108	0.092	0.002	0.054	0.0002	0.008	--	0.015	--	--	--	--	--
2031	339 322 282	--	0.023	--	0.012	--	--	--	--	--	--	--	--	--	--	--	--	0.038	--	--	--	--	--	--	0.0626
2033	339 323 282	--	0.509	--	--	--	0.013	--	--	--	0.001	--	0.003	--	--	--	--	0.056	--	--	--	--	--	--	0.0066
2037	341 327 280	0.366	0.289	--	--	--	0.165	0.261	0.14	--	--	--	0.033	--	0.008	--	--	0.32	--	--	0.016	0.01	0.08	0.09	0.011
2050	341 327 284	--	--	--	--	--	--	--	--	0.0399	--	--	--	0.002	--	--	--	0.165	--	--	--	--	--	--	--
2053	343 324 280	--	--	0.317	0.04	--	--	--	--	0.029	--	0.138	0.04	0.613	0.014	0.5	--	0.041	--	--	--	0.01	--	--	--
2060	343 323 284	--	0.032	--	--	--	0.006	--	--	--	--	--	--	--	--	--	--	0.004	--	--	--	--	--	--	--
2090	339 322 278	--	--	--	--	--	--	--	--	--	0.082	--	--	--	--	--	--	0.09	--	--	--	--	--	--	--

The common class I and II haplotypes are frequently in linkage, i.e. inherited together (Table 7). Interestingly, these class I/II haplotypes vary in occurrence and frequency in each of the color varieties. The 1006/2007, 1016/2031, 1014/2050, and 1092/2037 are common in Black Giant Schnauzers but of low frequency or absent from the Pepper and Salt colored dogs. Conversely, 1014/2037 and 1091/2033 are common in Pepper and Salt Giant Schnauzers but absent or low frequency in Black dogs. This suggests that the various breed founders contributed unequally to the genetics of Black and Pepper/Salt Giant Schnauzers. It is also noteworthy that several class I/II recombinants are present within the breed, an uncommon event in other breeds. DLA class I 1016 is linked with 2014 in one dog of unknown color and with 2031 in nine black dogs. The class I haplotype 1014 is linked with 2037 in 52% of Pepper and Salt Giant Schnauzers but only 6% of Black Giant Schnauzers. Conversely, 1014 is linked to 2050 in 27% of black dogs and only 4% of Pepper and Salt Giant Schnauzers. It is noteworthy that the 1014 haplotype is usually linked with 2037 in other breeds such as the Golden Retriever and Alaskan Klee Kai, and that the 1014/2050 haplotype has only been seen at this point in the Giant Schnauzer. This suggests that the 1014/2050 recombination occurred either as a mutation in a black dog that became a popular sire of that variety, or that it was introduced by a subsequent introgression.

Table 7. A comparison of frequencies for extended DLA class I/II haplotypes in Black and Pepper and Salt Giant Schnauzers

DLA1	DLA2	# dogs	BK	PS
1008	2005	1	0.01	
1006	2007	15	0.13	
1016	2014	1	?	?
1016	2031	9	0.09	
1014	2037	22	0.06	0.52
1011	2012	4		0.08
1014	2050	45	0.27	0.04

1068	2053	10	0.04	0.08
1091	2033	14		0.24
1092	2037	62	0.26	
1054	2022	2		
1008	2003	8	0.04	
1017	2090	24	0.06	

V. What does this assessment of genetic diversity tell us about contemporary Giant Schnauzer

Giant Schnauzers, although still a relatively small breed, possess an average degree of genetic diversity compared to other breeds as measured by the 33 STR markers. This may change a little with the addition of more dogs, but 133 dogs from diverse countries in North America and Europe would be expected to define 95% or more of diversity that currently exists in the breed. Any additional diversity would likely exist in more obscure and isolated geographic regions. Therefore, the current study should provide an adequate baseline on which to guide future decisions on how the breed should be genetically managed from this time forward.

It appears that breeders have done a good job in balancing genetic diversity across distant geographic regions, like breeds such as the Standard Poodle, but very different from breeds like the Italian greyhound. However, there is evidence of genetic differentiation within the breed at the level of variety or bloodline both by the 33 autosomal and 7 DLA STR markers. The two resulting varieties were clearly linked to black vs. pepper and salt coat colors. This is the first example in a breed that we have studied where coat color played such an important role in genetic diversity. However, studies of the breed confirm that the two coat-color variants are vigorously maintained more as “breeds” than varieties.

A standard genetic assessment of the allele frequencies of 33 STRs indicated that the average Giant Schnauzer was a product of random selection. However, IR values for individual dogs indicated that there was about one-fourth of dogs that were offspring of more closely related parents and that this population was balanced by an equal number of dogs born to quite unrelated parents. This would suggest that not all breeders of Giant Schnauzers are careful in how they select mates for their dogs and/or that pedigrees are not as accurate as perceived or that three generation pedigrees are not accurate indicators of relatedness. These are reasons to consider DNA testing as an adjunct to pedigrees.

VI. Health problems of Giant Schnauzer

A. Lifespan

Giant Schnauzers are reported in some sources to have a life span of 12-15 years (3, 5), which is surprisingly long for a large breed dog. Another source lists the lifespan as between 10-12 years compared to the median lifespan of 10-13 years of purebred dogs in general (6), a more realistic figure. A UKC breed health survey from 2014 lists disease conditions found in 82 Giant Schnauzers and found the median age in this group to be 4 years, with progressively declining numbers of animals up to 11.5 years of age (6).

B. Common disorders intrinsic to dogs and of complex genetic origin

Several conditions of complex genetic origin occur in Giant Schnauzers, but none occurs at an unexpectedly high incidence compared to other large breeds. Hip and elbow dysplasia are common in the breed and of complex genetic origins that appear to have been inherited by descent in many purebred dogs, especially those that are larger and fast growing. These problems lead to osteoarthritis of the hips and elbows with time and can be major problems in older, and occasionally younger, dogs.

Cancer is a common cause of death in all dogs, as it is in people and other species. The incidence in Giant Schnauzers is about average for other breeds. The types of cancers that occur in the breed are varied. Non-cancerous tumors of the skin such as follicular cysts and lipoma are common, while cancers that tend to occur in dark-pigmented dogs such as melanomas of the limbs and digits are less common. Squamous cell carcinoma of the digit has been described in the breed. Lymphoma is a common cancer, as it is in all dogs. Bone cancers, a problem in many large breeds, occur in Giant Schnauzers. Liver cancer may be more common than in most other breeds.

Heart problems are said to be one of the more common causes of death besides lymphoma. Conditions predisposing to heart failure include mitral valve disease and dilated cardiomyopathy. As a large, deep-chested dog, the Giant Schnauzer is prone to acute gastric torsion and bloat, a severe medical/surgical emergency.

Autoimmune disorders occur in all breeds of dogs and are also seen at similar incidences in Giant Schnauzers. The most common autoimmune disorder in all breeds is hypothyroidism secondary to chronic thryroiditis. Autoimmune hemolytic anemia is the second most common autoimmune condition in pure bred dogs including Giant Schnauzer. Keratoconjunctivitis sicca is a common autoimmune condition found in breeds predisposed to other autoimmune conditions such as hypothyroidism. Panosteitis is an auto-inflammatory disorder seen in adolescent dogs of several larger breeds. Central diabetes insipidus occurs in the breed and is also of autoimmune origin. Onchyodystrophy is another autoimmune disorder. Epilepsy is a problem in many pure breeds and may also have an autoimmune origin. Giant Schnauzers are reportedly more apt to develop drug allergies to things like sulfonamides and gold.

C. Genetic disorders involving Mendelian (simple) inheritance

Simple Mendelian traits involve single mutations in specific genes and over 600 have been described in dogs. Each breed averages 5 or more heritable diseases, most commonly simple autosomal recessive in origin. These mutations occur spontaneously within the breed are inherited by descent from founders or subsequent introgressions. They will often go unnoticed unless their incidence is inadvertently amplified by artificial selection for some desired conformation or performance trait that is unknowingly in genetic linkage. Popular sire effects are the most common reason for their amplification. Autosomal recessive traits are often not noticed until 1-2% or more of the population is affected, at which time 20% of the population may be carriers.

1. Cobalamin malabsorption (non-regenerative anemia)*
2. Factor VII Deficiency*
3. Progressive Retinal Atrophy (prcd-PRA)*
4. Neuroaxonal Dystrophy (NAD)*
5. Degenerative myelopathy*
6. Hyperuricosuria (HUU)*
7. Familial dilated cardiomyopathy*
8. Heritable cataracts**
9. Multifocal retinal dysplasia**
10. Glaucoma**

*DNA based tests available

**Inheritance not determined-simple autosomal recessives in other breeds. Screened by ophthalmoscopic examination.

VII. Information sources

A. Breed history

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