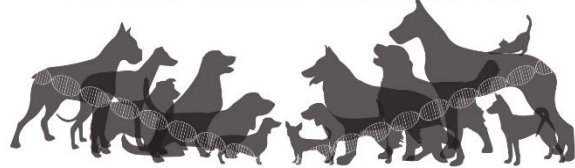




## Purdue Canine Genetics Lab



Animal Disease Diagnostic Laboratory

# Basset Hound Glycogen Storage Disease (BHGS)

## Genetic Test Result Interpretation

*We have designated the letter D to indicate the form of the GSD gene that contains this variant, and N to indicate the reference (normal) form of the gene. A dog's particular combination of N or D forms of the gene is known as its genotype.*

**Clear (GSD-N/N):** A **clear** dog has no copies of the GSD variant (this is also referred to as being homozygous normal or free of the variant associated with this form of glycogen storage disease). A clear dog cannot produce GSD affected (D/D) offspring.

**Carrier (GSD-D/N):** A **carrier** dog has one copy of the GSD variant (this is also referred to as being heterozygous). Based on current data, a **carrier dog is not at risk of developing GSD**. GSD carriers will, on average, pass the GSD variant on to half of their offspring; they can produce **GSD-D/D** (affected/susceptible) offspring if mated to another carrier (**GSD-D/N**) or affected (**GSD-D/D**) dog.

**Affected/Susceptible (GSD-D/D):** An **affected/susceptible** dog has two copies of the GSD variant (this is also referred to as being homozygous affected). Based on current data, **all GSD-D/D dogs develop clinical signs within the first several years of life**; clinical signs are difficult to detect, and may be absent, but include lethargy and exercise intolerance, labored breathing, and enlarged heart on imaging (X-ray or echo). The first sign something is wrong can be sudden death. All affected dogs died or were euthanized as relatively young adults. GSD-D/D dogs will pass one copy of this variant on to all of their offspring.

## Further Information on GSD

Based on current data, GSD is most likely inherited in a **fully penetrant autosomal recessive** manner. Fully penetrant means that all genetically affected dogs (**GSD-D/D**) will develop the disease during their lifetime. Autosomal recessive means that two copies of the mutation are required to show signs of disease; the genotype of GSD affected / susceptible dogs is D/D. Both clear (GSD-N/N) and carrier (GSD-D/N) dogs do not develop the GSD disease but could still show clinical signs of other unrelated cardiac diseases (e.g., heart murmurs, congestive heart failure, valve degeneration, etc.) or metabolic diseases.

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Below are the chances any given puppy in a litter from the indicated mating will have the genotype of N/N, D/N, or D/D. **Matings that produce an affected (GSD-D/D) dog are not recommended, and are shown in red.**

**Matings that will not produce GSD affected dogs:**

- Clear (N/N) x Clear (N/N) = 100% Clear (N/N)
- Clear (N/N) x Carrier (D/N) = 50% Clear (N/N), 50% Carrier (D/N)  
(This is an average, individual litters may see anywhere from 100% Clear to 100% Carrier)
- Clear (N/N) x Affected (D/D) = 100% Carrier (D/N)

**Matings that can produce GSD affected (D/D) dogs and are NOT recommended:**

- Carrier (D/N) x Carrier (D/N) = 25% Clear (N/N), 50% Carrier (D/N), **25% Affected (D/D)**  
(This is an average, individual litters may see more or less of any result)
- Carrier (D/N) x Affected (D/D) = 50% Carrier (D/N), **50% Affected (D/D)**  
(This is an average, individual litters may see anywhere from 100% Carrier to 100% Affected)
- Affected (D/D) x Affected (D/D) = **100% Affected (D/D)**

*We do not recommend exclusion of GSD carrier (D/N) dogs from the breeding population. We do recommend avoiding matings that have the potential to produce GSD affected (D/D) offspring as detailed above. As long as one of the two parents is GSD clear (N/N), GSD affected offspring will not be produced.*

**Immediately eliminating all GSD D/N (carrier) dogs from breeding may have negative consequences for the genetic diversity of the breed.**

**Test Limitations**

While we have identified this GSD variant associated with glycogen storage disease in Basset Hounds, other forms of genetic cardiac or metabolic disease may exist in the breed. It is therefore important to remember that this GSD test is diagnostic for only one form of GSD and will not identify other forms. Thus, it is still possible that offspring could be affected with a different genetic form of GSD, even if both parents have tested GSD-N/N (clear). To that end, we recommend that both dogs in a breeding pair be free of any past or present signs of metabolic and cardiac diseases, regardless of genotype. Nonetheless, this GSD test will help to prevent this form of glycogen storage disease and therefore significantly reduce the frequency of this disorder in the Basset Hound breed.