What Is G6S?

G6-Sulfatase deficiency is an inherited metabolic defect that occurs in Nubian goats and related crosses. A mutation in the G6-S gene renders the enzyme incapable of degrading complex polysaccharides known as heparin-sulfate glycosaminoglycans (HS-GAGs) which then abnormally accumulate in tissues such as central nervous system and internal organs. Affected goats have been known to exhibit delayed motor development, growth retardation, and/or early death. The disease is inherited in an autosomal recessive fashion. This means that both sexes are equally affected and two copies of the defective gene must be present for signs of the disorder to be observed. Breeding two carrier goats, which appear normal but each possesses a single copy of the mutation, is predicted to produce 25% affected offspring. (Table 2)

What is the Prevalence of This Defect?

In the last decade, there has been a raised awareness of this defect, and testing rates have increased. Research indicates that the overall prevalence of this defect in the Nubian population is 74.2% Normal, 23.9% Carrier, 1.9% Affected.

When Can G6S Testing be done?

G6S is a genetic defect. Animals can be tested at any age.

Why Should I Care?

Carrier animals can live long and productive lives. However, breeding carrier to carrier animals can result in affected animals.
What Does a “Normal,” “Carrier,” or “Affected” Result Mean?

**Normal** result means that the goat does not possess the genetic mutation and therefore does not have the G6S deficiency. Breeding two normal individuals results in normal offspring.

**Carrier** result means that the goat carries one copy of the mutation, inherited from one parent. A carrier will possess one copy of the normal gene and one copy of the defective gene. Carrier animals appear normal since the disorder occurs when the mutation is inherited from both parents. If one parent possesses a copy of the defective gene there is a 50% chance of passing it to their offspring. Thus, 50% of the offspring can be expected to carry the mutation. However, 100% will appear normal and be unaffected. (Table I)

**Affected** result should be interpreted to mean that the individual possesses two copies of the defective gene, and therefore, the goat has the G6S deficiency. Even affected animals may not show symptoms of the disease until they are sexually mature. The mutation may be unknowingly passed on to offspring, unless carriers and affected animals are identified through testing. Additionally, because symptoms of this disease may mimic those of other conditions or diseases, identification of affected animals is very likely underreported.

Table 1 shows the potential results (probability) when breeding a NN (normal) to a NG (carrier). 50% of the offspring will be normal (NN) and 50% will be carriers.

### TABLE 1

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<tr>
<td>N</td>
<td>NN (25%) normal</td>
<td>NN (25%) normal</td>
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<tr>
<td>G</td>
<td>GN (25%) carrier</td>
<td>GN (25%) carrier</td>
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Table 2 shows the potential results (probability) when breeding two carrier goats (NG). Remember, both animals appear normal but carry the mutation (NG). In this case, there is a 75% chance that offspring will appear normal. However 50% of offspring are potentially carriers (NG) and only 25% have a chance of being truly normal (NN). Moreover, there is a 25% chance that the offspring of two carriers will inherit the mutation from both parents and be affected (GG).

### TABLE 2

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<tr>
<td>N</td>
<td>NN (25%) normal</td>
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</table>

**How Do I Test My Goats?**

Currently, two laboratories offer genetic testing for G6S. UC Davis Veterinary Genetics Laboratory (VGL) and Texas A&M Diagnostic Laboratory (TVMDL).

VGL is internationally recognized as a pioneer and expert in DNA-based animal testing. ADGA has negotiated a discounted rate for ADGA Members/ADGA Plus Members for all genetic testing through VGL. G6S testing can be done with the same hair sample that owners provide for DNA Typing (required for valid semen collections on bucks). The lab can use the same sample to test for other genetic traits such as alpha S1 Casein. Go to the ADGA website: [http://adga.org/?s=G6s](http://adga.org/?s=G6s) for additional information. Test results done through the ADGA program are automatically reported to the submitter (owner) as well as to ADGA.

TVMDL also offers testing for G6S. Results can be reported by TVMDL to ADGA upon submitter request. More information can be found on the TVMDL website. 
[https://tvmdl.tamu.edu/2015/04/30/faqs-on-g6s-diagnostic-testing/](https://tvmdl.tamu.edu/2015/04/30/faqs-on-g6s-diagnostic-testing/)

**How are Results Reported?**

G6S DNA profiles are reported by VGL as N/N for Normal, N/G for Carrier and G/G for Affected goats. Results from TVMDL are reported as Normal, Carrier, and Affected.