CONGENITAL DEAFNESS

By Dr. George M. Strain,
Louisiana State University Comparative Biomedical Sciences,
School of Veterinary Medicine, Baton Rouge, Louisiana 70803

[Every effort has been made to ensure accuracy of information. However, this is not a substitute for prompt veterinary care. Any similarity to other publications is unintentional. Published online at Sealyhealthguard.org, 11/23/10]

NOTE: For an explanation of "congenital" see http://www.medterms.com/script/main/art.asp?articlekey=10766

Congenital deafness in dogs (or other animals) can be acquired [caused by intrauterine infections, ototoxic drugs like gentamicin, liver disorders, or other toxic exposures before or soon after birth] or inherited. Inherited deafness can be caused by a gene defect that is autosomal dominant, recessive, sex-linked, or may involve multiple genes (more on this later). It is usually impossible to determine the cause of congenital deafness unless a clear problem has been observed in the breed, or carefully planned breedings are performed. In this article I will discuss what is currently known about the genetics of deafness in dogs so that breeders can make the best informed decisions possible when attempting to reduce or eliminate deafness.

Congenital deafness has been reported for approximately 80 breeds, with the list growing at a regular rate; it can potentially appear in any breed but **especially those with white pigmentation**. Deafness may have been long-established in a breed but kept hidden from outsiders to protect reputations.

The disorder is usually associated with pigmentation patterns, where the presence of white in the hair coat increases the likelihood of deafness. Two pigmentation genes in particular are often associated with deafness in dogs: the merle gene (seen in the Collie, Shetland Sheepdog, Dappled Dachshund, Harlequin Great Dane, American Foxhound, Old English Sheepdog, and Norwegian Dunkerhound among others) and the **piebald gene** (Bull Terrier, Samoyed, Greyhound, Great Pyrenees, **Sealyham Terrier**, Beagle, Bulldog, Dalmatian, English Setter).

However, not all breeds with these genes have been reported to be affected. The deafness, which usually develops in the first few weeks after birth while the ear canal is still closed, usually results from the degeneration of part of the blood supply to the cochlea (the stria vascularis). The nerve cells of the cochlea subsequently die and permanent deafness results. The cause of the vascular degeneration is not known, but appears to be associated with the absence of pigment producing cells (melanocytes) in the blood vessels. All of the function of these cells are not known, but one role is to maintain high potassium concentrations in the fluid surrounding the hair cells of the cochlea; these pigment cells are critical for survival of the stria.

Deafness in the Doberman, which is also accompanied by vestibular (balance) disturbance, results from a different mechanism, where hair cell death is not the result of degeneration of the stria. Deafness may also occur later in life in dogs from other causes such as toxicities, infections, or injuries, or due to aging (presbycusis); these forms of deafness almost never have a genetic cause in animals and thus do not present a concern in breeding decisions.

The prevalence of congenital deafness in different breeds is seldom known because of the limited number of studies In the Dalmatian, where the incidence is highest, 8 percent of all dogs in the US are bilaterally deaf and 22 percent are unilaterally deaf. In the English Setter, English Cocker Spaniel, Australian Cattle Dog, and Bull Terrier, where fewer numbers of dogs have been hearing tested, the incidence appears to be about one third to one half that of Dalmatians.

Unilateral or bilateral deafness is found in 75percent of all white Norwegian Dunkerhounds, but the incidence in normal-color dogs is unknown. Other breeds with a high incidence are the Catahoula and Australian Shepherd. The incidence of all types of deafness in the general dog population is low, reported to be 2.56 to 6.5 cases per 10,000 dogs seen at veterinary school teaching hospitals, but these data predate the availability of hearing testing devices and so are much lower that actual values. Recognition of affected cases is often difficult, because unilaterally deaf dogs appear to hear normally unless a special test (the

brainstem auditory evoked response, BAER) is performed; facilities to perform the BAER are usually only available at veterinary schools (see list of BAER Testers). It should be noted that a unilaterally deaf dog can be as great a genetic risk for transmission of deafness to its offspring as is a bilaterally deaf dog.

The method of genetic transmission of deafness in dogs is usually not known. There are no recognized forms of sex-linked deafness in dogs, although this does occur in humans. The disorder has been reported to have an autosomal recessive mechanism in the Rottweiler, Bull Terrier, and Pointer, but this suggestion is not reliable because the reports were before the availability of BAER testing and the ability to detect unilaterally deaf dogs. References usually state that deafness transmission in most other breeds is autosomal dominant, but this is false, as will be discussed below. Pigment-associated inherited deafness is not restricted to dogs. Similar defects have been reported for mice, mink, pigs, horses, cattle, cats, and humans. Deafness in blue-eyed white cats is common and is known to be passed on as an autosomal dominant defect. Blue eyes, resulting from an absence of pigment in the iris, is common with pigment-associated deafness but is not, in and of itself, an indication of deafness or the presence of a deafness gene; however, in several breeds dogs (Dalmatian, English Setter, English Cocker Spaniel, Bull Terrier) with blue eyes are statistically more likely to be deaf. Waardenburg's syndrome, a human condition, presents with deafness, a stripe of white in the hair and beard, blue or different colored eyes (even in blacks and asians), no pigment behind the retina, and minor structural deformities around the nose and eyes. This is an autosomal dominant disorder with incomplete penetrance, which means that individuals that inherit the disorder may not show all components of the syndrome - i.e., they may not be deaf. Incomplete penetrance of a defect greatly complicates the determination of mode of inheritance. At present there is no documentation that incomplete penetrance is a factor in any canine deafness.

In simple Mendelian genetics, each dog carries two copies of each gene, one from each parent. The possible outcomes of breedings can be demonstrated with tables showing the genotype of both parents and the possible combinations in their offspring. If deafness is carried as a theoretical simple autosomal recessive gene (d), the breeding of two hearing carriers (Dd) will result, on average, in 25 percent affected dogs (dd), 50 percent hearing carriers (Dd), and 25 percent free of the defect (DD). The breeding of a carrier to a dog free of the defect (Table 2) will result in no affected dogs but 50 percent carriers and 50 percent free. The breeding of an affected dog to a carrier will result in 50 percent affected, 50 percent carriers, and no free. Finally, the breeding of an affected dog to a dog free of the defect will result in 100 percent carriers and no affected or free.

If instead the deafness is carried as a simple autosomal dominant gene (D), the breeding of an affected dog (Dd) to a free dog (dd) would result on average in 50 percent affected and 50 percent free. Dogs with the genotype DD would be unlikely to occur unless two deaf dogs had been bred. All of the above assumes that incomplete penetrance is not acting. If more than one gene (recessive and/or dominant) is involved in producing the deafness, the possible combinations become much more complicated. In humans more than 50 different autosomal recessive or dominant deafness genes or loci have been identified. The children of two deaf parents with two different recessive deafness genes can be unaffected but carry both genes. If deafness in dogs results from more than one recessive gene, the possible outcomes of breedings are more numerous and determination of the mechanisms of transmission will be difficult.

As stated above, deafness is often associated with the merle (dapple) gene, which produces a mingled or patchwork combination of dark and light areas. This gene (M) is dominant so that affected dogs (Mm) show the pattern, which is desirable in many breeds. However, when two dogs with merle are bred, 25 percent will end up with the MM genotype. These dogs usually have a solid white coat and blue irises, are often deaf and/or blind, and are sterile. Breeders of these dogs breeds know not to breed merle to merle. In this case the deafness is neither dominant nor recessive, but is linked to a dominant gene that disrupts pigmentation and secondarily produces deaf dogs.

Genetic transmission of deafness in dogs with the piebald (sp) and extreme white piebald (sw) pigment genes, such as the Dalmatian, is less clear. These genes affect the amount and distribution of white areas on the body. Deafness in Dalmatians does not appear to be autosomal dominant, since deaf puppies result from hearing parents. It does not appear to be a simple recessive disorder: we have bred pairs of deaf Dalmatians and obtained bilaterally hearing and unilaterally hearing puppies, when all should have been deaf if the disorder was recessive.

These findings might be explained by a multi-gene cause, the presence of two different autosomal recessive deafness genes, or a syndrome with incomplete penetrance. Further studies (in progress) will be required to determine the mechanisms. Several candidate genes known to cause pigment-related deafness in humans or mice have been eliminated as the possible cause of pigment-associated deafness in Dalmatians. Whole-genome screens will hopefully identify the cause in this and other breeds.

Recent studies have shown that deafness in Dobermans, which do not carry the merle or piebald genes, results from direct loss of cochlear hair cells without any effects on the stria vascularis. Vestibular (balance) system signs, including head tilt and circling, are seen, and the deafness is transmitted by a simple autosomal recessive mechanism. A similar pathology has been described for the Shropshire Terrier.

So what should breeders do when deafness crops up? The most conservative approach would be to not breed the affected animal and not repeat the breeding that produced deafness. It is frequently recommended (Le. Dalmatian Club of America) that bilaterally deaf puppies should be euthanized, since they make poor pets, are difficult to train, are prone to startle biting, frequently die from misadventure (cars), and require excessive care.

There is considerable controversy on this point, and there is no question that many people have successfully raised deaf dogs. For every story of a problem deaf dog there seems to be a story of one that was successfully raised. Unfortunately, there is no way to predict how a deaf puppy will turn out. Unilaterally deaf dogs can make good pets but should never be bred.

When deafness is uncommon in a breed, affected dogs should not be bred, but this does not mean that all related dogs are a risk and must be retired from breeding. An understanding of simple autosomal recessive and dominant patterns, as explained above, can allow the breeder to make better informed decisions and likely avoid future deaf animals without sacrificing a breeding line that has been shaped over many years.

However, extreme caution must be used when line breeding of dogs related to deaf dogs, whether the deafness is unilateral or bilateral. To make these decisions in an informed manner for breeds with known deafness, it is important that advantage be taken of hearing testing facilities at veterinary schools. Unilaterally deaf dogs cannot be detected by other means, and these dogs will pass on their deafness genes.

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