

Key Points

1. Chiari and syringomyelia were identified in cavalier King Charles spaniels in 1997 and found to be fairly common
2. This study examined the pedigrees of dogs with CM/SM and compared them to dogs without
3. Tracing back from the affected dogs, researchers identified 4 key breeding lines (labeled C, D, M, S)
4. They further traced every affected dog back to a single female who had only one litter with two offspring
5. In comparing the affected dogs to unaffected ones, found that the number of great-grandparents descended from the 4 key dogs was more important to developing CM/SM than the mother and father having the gene
6. This indicates that more than one gene may be involved in Chiari, or that the percent of dogs who actually develop Chiari from a defective gene is variable

Definitions

autosomal recessive - for a hereditary disease, type of transmission where the defective gene must be present in each parent for it to be passed on

cavalier King Charles Spaniel - toy dog initially bred in the early 1900's

dam - a puppy's female parent

occipital bone hypoplasia - underdevelopment of the bone in back of the skull, near the bottom; thought to lead to Chiari

oligenic - refers to an inheritable disease which is the result of more than one gene

penetrance - percent of people who carry a "disease" gene, who will actually develop that disease

sire - a puppy's male parent

Dogs Provide Chiari Genetic Clues

November 15, 2005 -- Since Chiari first comes to light for many people in their late 20's and early 30's - when they are beginning to start families - one of the most common questions people have is whether it will be passed on to their children. Research, by Dr. Speer at Duke, has shown that the number of families which have multiple members with Chiari makes it extremely likely that some percent of Chiari cases are genetic.

Unfortunately, that is about the extent of our knowledge. It is not yet known what gene or genes are involved, what percent of cases are indeed genetic, and of course, there is no Chiari genetic test. While Dr. Speer searches human DNA for genetic clues, across the pond (as the British say), Dr. Rusbridge, a veterinarian at Stone Lion Veterinary Center in the UK, is doing similar research with dogs.

In 1997, Dr. Rusbridge first identified that a breed of toy dogs, known as cavalier King Charles spaniels (see Figure 1), suffer from Chiari and syringomyelia, very much like their human owners. MRI's have confirmed that in certain dogs, the bones at the back of the skull are underdeveloped, the cerebellum crowds the spinal cord, sometimes the medulla is kinked, and syringomyelia develops in the upper part of the spine.

Dogs with Chiari and syringomyelia seem to have similar symptoms to people, they just express them in a different way. Affected dogs tend to scratch extensively at their neck and shoulders due to abnormal sensations. In addition, they tend to bark suddenly when changing position or when touched in certain areas, likely in response to pain. Canines with extensive syrinxes also exhibit limb weakness and trouble walking.

For treatment, some dogs are able to get by using anti-inflammatories or steroids, but others require surgery. Again, the surgery is very similar to the human decompression and involves enlarging the skull area and opening the dura to create more space.

As it became clear that Chiari was somewhat common in this breed, Dr. Rusbridge decided to study the hereditary aspects of the condition. The cavalier King Charles spaniel was first bred in 1928 and in the 1930's six dogs were extensively inbred. Because they are often show dogs and were bred fairly recently, pedigree information is available from a number of different sources.

Using this information, Rusbridge, and her colleague Knowler, traced the pedigrees of 45 dogs with Chiari and syringomyelia (30 identified by MRI, and 15 diagnosed by clinical exam), and compared them to 45 dogs without Chiari. They published their findings in the July, 2003 issue of the journal, *Veterinary Record*.

The group with Chiari represented dogs from the Britain, Canada, the US, Ireland, Finland, France, and Tasmania. The researchers were able to trace back 20 generations and compiled a database of over 1300 dogs. Because of inbreeding, the family trees did not look like regular family trees and the team ended up using special software to take into account that a single dog may have been used for generations of breeding.

From the database, the team identified that CM/SM was common in four family lines and identified 4 key dogs - which they labeled C, D, M, S - which were consistently involved in the affected family lineages. Interestingly, all four of the key dogs could further be traced back to a single female (labeled G) who died in 1958 at only 18 months of age. The female G was recorded to have had only one litter with two pups during her life.

As they studied the relationship of the affected dogs to the 4 key dogs and the "original" female, G, the team found that for the development of Chiari and syringomyelia, a dog's immediate parents were not as important as their great-grandparents (see Table 2). For example, while 100% of the affected dogs had parents whose pedigree could be traced back to the female, G; 60% of the unaffected group also had both parents who could be traced back in a similar fashion. So, just having a dam and sire from a certain line wasn't enough to predict Chiari. However, 100% of the affected dogs also had 6 of 8 great-grandparents from this lineage, whereas less than 7% of the healthy dogs did.

Since it wasn't possible to track the number of Chiari dogs versus non-Chiari dogs in each litter (because so many puppies are given away, etc.), Rusbridge was only able to conclude that there was a high rate of CM/SM in certain family lines and that it is likely due to a genetic defect. Further, she believes that the parent/great-grandparent finding, while indicative of an autosomal recessive trait - meaning a dog (or person) needs to inherit a bad gene from both their parents - also suggests a more complex mechanism is at play. For example more than one gene may be involved in the development of Chiari.

Dr. Rusbridge is continuing her work and has recently published her experiences in trying to build a DNA database of cavalier King Charles spaniels with Chiari and syringomyelia. It will be interesting to monitor the progress of genetic research on both people and dogs and see if man's best friend will provide a key piece of the

cerebellar tonsils - portion of the cerebellum located at the bottom, so named because of their shape

cerebellum - part of the brain located at the bottom of the skull, near the opening to the spinal area; important for muscle control, movement, and balance

cerebrospinal fluid (CSF) - clear liquid in the brain and spinal cord, acts as a shock absorber

Chiari malformation I - condition where the cerebellar tonsils are displaced out of the skull area into the spinal area, causing compression of brain tissue and disruption of CSF flow

decompression surgery - general term used for any of several surgical techniques employed to create more space around a Chiari malformation and to relieve compression

magnetic resonance imaging (MRI) - diagnostic device which uses a strong magnetic field to create images of the body's internal parts

syringomyelia (SM) - neurological condition where a fluid filled cyst forms in the spinal cord

syrinx - fluid filled cyst in the spinal cord

Source

Rusbridge C, Knowler SP. [Hereditary hemilect of occipital bone hypoplasia and syringomyelia \(Chiari type I malformation\) in cavalier King Charles spaniels](#). Vet Rec. 2003 Jul 26;153(4):107-12.

Chiari puzzle.

Figure 1
Cavalier King Charles Spaniel



Table 2
Percent of Dogs With and Without CM/SM Descended From Four Identified Key Lines

Descended From C, D, M, S	% With CM/SM	% Without CM/SM
Dam & sire	100	60
All grandparents	100	18
6 great grandparents	100	7
All great grandparents	69	4

Note: Every dog with CM/SM had 6 great-grandparents that were descended from 4 key dogs, whereas only 7 % of the non-affected group did

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