

# Bulldogs' screw tails linked to human genetic disease

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Moxie, a 3-year-old French bulldog, took part in a study of the genetics of 'screwtail' dog breeds (bulldogs, French bulldogs and Boston terriers). A common mutation in these dogs is similar to genetic changes in a rare human disease, Robinow syndrome. Credit: Katy Robertson

With their small size, stubby faces and wide-set eyes, bulldogs, French bulldogs and Boston terriers are among the most popular of domestic dog breeds. Now researchers at the University of California, Davis, School of Veterinary Medicine have found the genetic basis for these dogs' appearance, and linked it to a rare inherited syndrome in humans.

Bulldogs, French bulldogs and Boston terriers aren't the only dogs with short, wide heads, but they do share another feature not found in other breeds: a short, kinked tail or "screwtail," said Professor Danika Bannasch, Department of Population Health and Reproduction in the UC Davis School of Veterinary Medicine. These three breeds all lack the vertebrae that make up the tail bone, she said.

The researchers sequenced the whole genome—the entire DNA sequence—of 100 dogs, including 10 from screwtail breeds. All the participating dogs were privately owned pets seen at the UC Davis Veterinary Medical Teaching Hospital, whose owners agreed to participate. Graduate students Tamer Mansour and Katherine Lucot, with C. Titus Brown, associate professor at the School of Veterinary Medicine and Genome Center, searched through the DNA sequences to find changes associated with screwtail breeds.

From more than 12 million individual differences they were able to identify one mutation, in a gene called DISHEVELLED 2 or DVL2. This variant was found in 100 percent of the bulldogs and French bulldogs sampled, and was very common in Boston terriers.

This kind of whole genome comparison is relatively new, Bannasch said.

"Normally, we would have first had to identify a region DNA and work from there," she said. "We could look at [breed](#)-specific traits, but not as

well as we can now."

## Link to Robinow syndrome

Professor Henry Ho at the UC Davis School of Medicine studies similar genes in humans. Mutations in the related DVL1 and DVL3 genes are known to cause Robinow syndrome, a rare inherited disorder in humans characterized by strikingly similar anatomical changes—a short, wide "babyface," short limbs and spinal deformities. In addition, Robinow patients and the screwtail breeds also share other disease traits, such as cleft palate. In both humans and dogs, DVL genes are part of a signaling pathway called WNT involved in development of the skeleton and nervous system, among other things, said Peter Dickinson, professor of surgical and radiological sciences at the School of Veterinary Medicine. By characterizing the screwtail DVL2 protein product, Sara Konopelski, a [graduate student](#) the Ho lab, pinpointed a key biochemical step in the WNT pathway that is disrupted by the mutation. This finding further suggests that a common molecular defect is responsible for the distinct appearances of both Robinow patients and screwtail dog breeds.

The DVL2 screwtail mutation is so common in these breeds, and so closely tied to the breed appearance, that it would be difficult to remove it by breeding, Dickinson said. Other genes are known to contribute to short, wide "brachycephalic" heads in dogs, and there are likely multiple genes that contribute both to appearance and to chronic health problems in these breeds.

Understanding a common mutation in popular dog breeds may, however, give more insight into the rare Robinow syndrome in humans. Only a few hundred cases have been documented since the syndrome was identified in 1969.

"It's a very rare human disease but very common in [dogs](#), so that could

be a model for the human syndrome," Bannasch said.

**More information:** Tamer A. Mansour et al, Whole genome variant association across 100 dogs identifies a frame shift mutation in DISHEVELLED 2 which contributes to Robinow-like syndrome in Bulldogs and related screw tail dog breeds, *PLOS Genetics* (2018). [DOI: 10.1371/journal.pgen.1007850](https://doi.org/10.1371/journal.pgen.1007850)

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