

TYPE I IVDD/CDDY: What is it? What does it mean?

Congratulations on your new canine companion. It is exciting, and understandably there may be questions. Your breeder has taken the time to use the Embark DNA test to screen for genetic health risks for over 215+ conditions.

This includes the variant associated with **Type I Intervertebral Disc Disease** and **Chondrodystrophy**. This resource aims to provide a better understanding of what this result means, encourage proactive medical care, and aid in initiating a conversation with your dog's veterinarian.

1. What is Type I Intervertebral Disc Disease/ Chondrodystrophy (IVDD/CDDY)?

The FGF4 retrogene insertion on chromosome 12 may be responsible for a portion of cases of chondrodystrophy and increased risk for Type I IVDD in certain populations of dogs.

Chondrodystrophy is a physical trait that is known to occur in many breeds and includes a short-legged appearance.

- This is the accepted breed standard for many breeds (e.g. Dachshunds, Corgis, French Bulldogs).
- CDDY can be caused by other variants alone or in combination with the CDDY/IVDD variant.
- This is different from Chondrodysplasia (CDPA) which is found on chromosome 18, and not tested with Embark. This refers to the short-legged phenotype, but is not associated with the risk of Type I IVDD like CDDY.

Type I IVDD is one form of a medical condition also known as a "slipped disc."

- Type I IVDD is characterized by the rupture of the center part of the disc (nucleus pulposus) through tears in the outer part of the disc (annulus fibrosus). The previously noted variant in FGF4 is known to increase the risk of Type I IVDD.
- There is another form of IVDD, known as Type II. Type II IVDD is typically more age-related and is most commonly diagnosed in larger breed dogs (non chondrodystrophic breeds). This type is caused by chronic bulging of the outer part of the disc on the spinal cord. It is typically slowly progressive and may or may not be painful.



2. Why did my breeder breed dogs knowing they could produce "at-risk" offspring?

Many dog breeds, due to human selection for a desired appearance, or phenotype, have a high frequency of this variant in the FGF4 retrogene, meaning most or all dogs of the breed have at least one copy of the variant. Some breeds are "fixed" for the FGF4 retrogene (all dogs have two copies of the variant).

When considering whether a dog is right for a breeding program, multiple factors should be considered for the health and longevity of the breed, such as:

- Non-genetic health testing (physical exam, blood work, x-rays)
- Health and longevity of the lines (e.g. heart disease), and especially in relation to IVDD risk, occurrence of spinal or neurologic problems, or changes on spinal imaging (x-rays)
- Adherence to the breed standard and other physical conformation parameters
- Temperament
- Overall goals of the breeding program and puppy placement (e.g. pets versus working dogs)

If all other health and temperament parameters are desired, then breeding dogs with one or two copies of the CDDY/IVDD variant to dogs with no copies is a step towards reducing the frequency of this variant within certain breeds. However, this may not always be possible in breeds where the FGF4 retrogene occurs at high frequency, such as when chondrodystrophy represents the desired phenotype. In addition to changing the physical appearance, attempting to eliminate the FGF4 variant through selective breeding could have negative side effects for many breeds. In breeds where the variant occurs frequently, removing all dogs with one or two copies of the variant from the breeding pool generally reduces the size of the

breeding population substantially, and can lead to other health issues associated with inbreeding and reduced genetic diversity. This can actually harm the overall health and longevity of a breed.

3. Common breeds where the variant is fixed or occurs at high frequency:

- Beagles
- Cavalier King Charles Spaniel
- Cocker Spaniels
- Corgis (Pembroke Welsh Corgis & Cardigan Welsh Corgis)
- Dachshunds
- French Bulldogs
- Miniature Poodles
- Pekingese

A more comprehensive list of breeds with their FGF4 variant frequencies can be found here:

<https://vgl.ucdavis.edu/test/cddy-cdpa>

4. Will my dog develop Type I IVDD?

The presence of the variant is not a diagnosis of disease, nor determines whether or not a dog will 100% develop IVDD.

There is active research assessing the risk of this variant and its association with IVDD, within and among dog breeds, but these data are not available yet to predict lifetime risk. Many dogs that have this variant do not develop Type I IVDD. However, knowing the risk and making lifestyle modifications may contribute to a reduction in occurrence.

While the variant is known to increase the risk of Type I IVDD, there are other factors that contribute to the overall risk of developing Type I IVDD, including:

- Other (unknown) genetic factors
- Lifestyle and other environmental factors

5. What steps can I take to reduce my dog's risk?

Early intervention provides the best outcome once clinical IVDD occurs: Discuss with your dog's veterinarian to assess their overall risk, develop a monitoring plan, and learn the signs that warrant a vet visit. Some measures that may reduce the risk of IVDD include:

- Maintain an ideal body condition (weight)
- Modifying activities to put less stress on the spine (e.g: provide steps or ramps to reduce jumping on and off objects.)
- When a harness is used, low-stress options such as front clip options may be recommended
- These recommendations may also reduce the risk of other health concerns, and therefore can be a part of a healthy lifestyle in general

6. Signs and symptoms?

The degree and severity of clinical IVDD is determined by your dog's veterinarian. This is graded on a scale of 1-5, with 1 being the least severe, and 5 being the most severe. Treatment will range from conservative medical management and rest to surgery.

* The inability to move or feel the limbs is a veterinary emergency

Dogs that are diagnosed with IVDD may show:

- Sudden or progressive onset of hind leg weakness
- Paw dragging
- An ataxic (often compared to a drunken) gait
- Signs of back pain which may include reluctance to jump or walk, vocalization when moving, reluctance to be pet or lifted, unusual aggression towards being touched

7. Does this mean my dog is unhealthy or cannot be active?

Absolutely not! Breeders strive to produce the best dogs they can for the betterment of the breed they work with, and that includes using their time, knowledge, dedication, and expertise. Through genetic and other health testing, providing preventative veterinary care, proper husbandry, and knowledge of their lines, breeders are taking remarkable steps to produce happy and healthy puppies. Physical activity is also important to long term happiness and health for dogs, just like it is for people. Knowing the potential risks and having an open dialogue with your veterinarian will enable you to customize activities for your dog.

Additional Resources:

Breeding schemes for intervertebral disc disease in dachshunds: Is disc calcification score preferable to genotyping of the FGF4 retrogene insertion on CFA12:

<https://cgejournal.biomedcentral.com/articles/10.1186/s40575-020-00096-6>

Current Insights Into the Pathology of Canine Intervertebral Disc Extrusion-Induced Spinal Cord Injury: <https://www.frontiersin.org/articles/10.3389/fvets.2020.595796/full>

VIN: Intervertebral Disk Disease (IVDD) in Dogs: <https://veterinarypartner.vin.com/default.aspx?pid=19239&id=4953012>

VIN: Chondrodystrophy In Dogs: <https://veterinarypartner.vin.com/default.aspx?pid=19239&catId=102899&id=9756702>

ACVS: Intervertebral Disc Disease: <https://www.acvs.org/small-animal/intervertebral-disc-disease>

About the Embark test: <https://embarkvet.com/breeders/embark-approach/>