Understanding the Two FGF4 Retrogenes in Dogs: CFA12 vs. CFA18

Dogs possess **two different FGF4 retrogenes** that arose independently during evolution. Both affect skeletal development, but they are **not identical** in their effects.

Feature	FGF4 Retrogene on CFA12 (CDDY)	FGF4 Retrogene on CFA18 (Chondrodysplasia)
Chromosome	Chromosome 12	Chromosome 18
Primary Effect	Chondrodystrophy (mild to moderate shortening of limbs)	Chondrodysplasia (breed-defining short legs)
IVDD Risk	**Strongly increases the risk of IVDD**	Little or no independent increase in IVDD risk
Disc Calcification	Yes	No significant effect
Premature Disc Degeneration	Yes	No
DNA Test Available	Yes	Yes

FGF4 Retrogene on Chromosome 18 (CFA18)

The chromosome 18 retrogene was discovered first.

It is responsible for the classic short-legged appearance seen in breeds such as:

- Dachshund
- Pembroke Welsh Corgi
- Cardigan Welsh Corgi
- Basset Hound

Its primary effect is on **bone growth** during development.

Dogs carrying the CFA18 retrogene develop proportionately shorter legs but **this mutation alone is not considered the major cause of IVDD.**

In other words:

CFA18 changes the dog's appearance much more than it changes spinal health.

Reference

Parker HG, VonHoldt BM, Quignon P, et al. (2009). *An expressed FGF4 retrogene is associated with breed-defaling chondrodysplasia in domestic dogs.* Science.

FGF4 Retrogene on Chromosome 12 (CFA12)

The chromosome 12 retrogene was discovered several years later.

Researchers found that it not only shortens the limbs but also causes important changes within the intervertebral discs.

Dogs carrying the CFA12 mutation develop:

- premature degeneration of the discs,
- disc calcification,
- significantly increased risk of Hansen Type I IVDD.

Unlike the CFA18 mutation, the CFA12 mutation affects **both the skeleton and the spine.**

This is the mutation now referred to as **CDDY (Chondrodystrophy with Increased Risk of IVDD).**

Reference

Brown EA, Dickinson PJ, Mansour T, et al. (2017). *FGF4 retrogene on CFA12 is responsible for chondrodystrophy and intervertebral disc disease in dogs.* Proceedings of the National Academy of Sciences.

Dogs May Carry One or Both Retrogenes

Some breeds possess:

- only the CFA18 retrogene,
- only the CFA12 retrogene,
- or both.

When both are present, the skeletal effects may be additive, producing shorter limbs, while the **CFA12 mutation contributes most of the increased IVDD risk.**

Reference

Batcher K, Dickinson P, Giuffra J, et al. (2019). *Phenotypic Effects of FGF4 Retrogenes on Intervertebral Disc Disease in Dogs.* Genes, 10(6), 435.

Why Is This Important for Bedlington Terriers?

Current evidence indicates that Bedlington Terriers possess the **CFA12 (CDDY)** retrogene and **do not possess the CFA18 retrogene**.

This means the concern in Bedlington Terriers is **not the classic dwarfism seen in breeds such as Dachshunds or Corgis.** Instead, the concern is whether the **CFA12 mutation produces measurable shortening of the limbs and increases the risk of IVDD within the breed.**

Research in many breeds demonstrates that the CFA12 mutation has these biological effects. However, **the prevalence, degree of limb shortening, penetrance, and lifetime IVDD risk have not yet been determined specifically in Bedlington Terriers.** Those unanswered questions are the focus of the current breed-specific research.

Bottom Line

The two FGF4 retrogenes are **not interchangeable**.

- **CFA18** primarily affects **limb length** and is responsible for the classic short-legged appearance of several breeds.
- **CFA12 (CDDY)** affects **both limb length and spinal disc health**, increasing the risk of Intervertebral Disc Disease (IVDD).
- The mutation identified in Bedlington Terriers is the **CFA12 (CDDY) retrogene**, making breed-specific research essential to determine how strongly it influences health and conformation in this breed.

One additional point is worth emphasizing because it is often misunderstood in discussions of CDDY. **The CFA12 mutation is considered pleiotropic**, meaning **a single gene affects more than one trait**. In this case, the same mutation influences both **skeletal development (limb length)** and **intervertebral disc biology (predisposition to IVDD)**. This is why researchers cannot separate the "dwarfism gene" from the "IVDD gene"—they are different biological effects of the same mutation. Brown et al. (2017) and Batcher et al. (2019) both emphasize this relationship.