

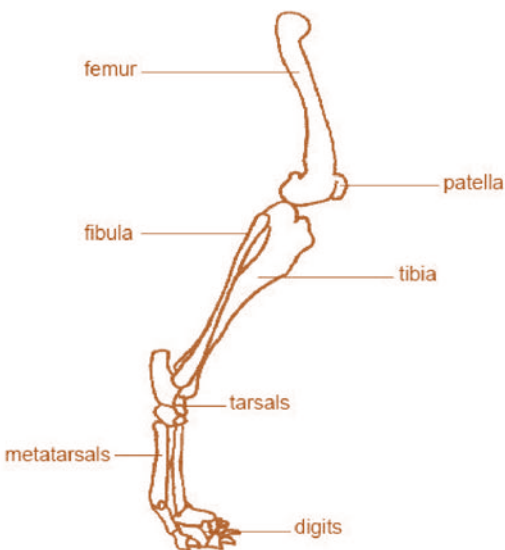
# REAR LEG LAMENESS: A Genetic Perspective

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## Introduction

Three major orthopedic conditions affecting the rear leg in West Highland White Terriers include patellar luxation, hip dysplasia, and Legg-Calve-Perthes. Each will be briefly discussed in terms of description of the condition,



treatment options, and what is known from a genetic standpoint.

As a review, let us start with some basic anatomical points of reference that will be discussed throughout the article. The normal rear leg of the dog has 3 main joints: the hip, the

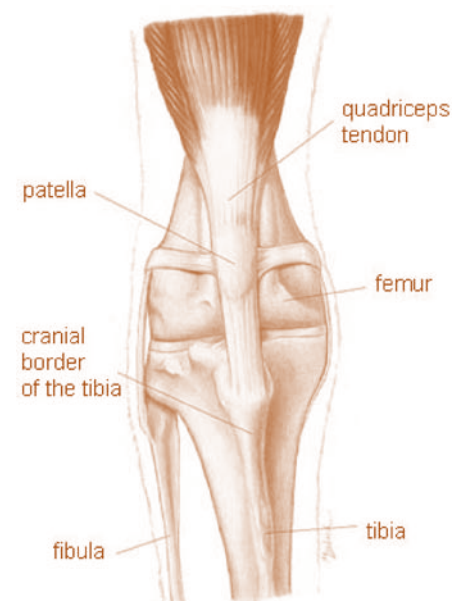
stifle, and the hock. The bones comprising the rear leg are, from the top down: femur (long bone between the hip and stifle), patella (kneecap), tibia and fibula (bones between stifle and hock), tarsals (bones in the hock, equivalent to our ankle bones), metatarsals (bones between hock and foot, equivalent to the bones in the flat of our feet), and phalanges/digits (the bones in the toes). The patella (kneecap) is maintained in proper position by the quadriceps and patellar tendons. The hip and stifle joints, and associated structures, will be the focus of this article.

## Patellar Luxation

Patellar luxation is a dislocation of the kneecap. This is one of the most common congenital anomalies in dogs. Nearly 7% of all puppies are diagnosed with a luxating patella. Small breed dogs are 10 times more likely to be affected than large breed dogs. Half of the diagnosed cases have a bilateral (both stifles) involvement.

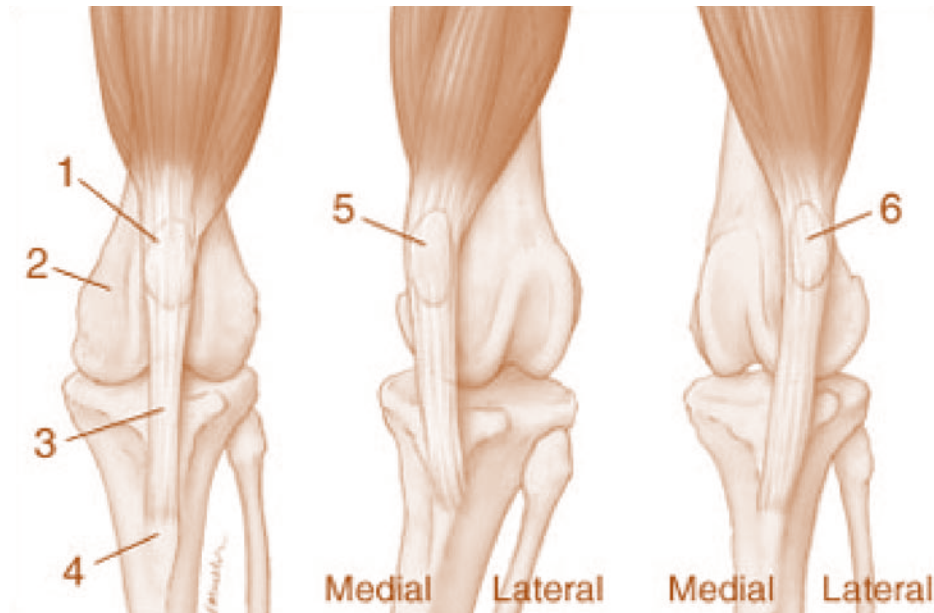
The patella can luxate (move out of place) either medially (towards the belly) or laterally (towards the outside of the leg). Medial luxations are more common than lateral conditions.

Signs that your pet may have patellar luxation include: carrying leg off the ground for several steps, shaking or extending the leg prior to full use, or intermittent rear leg lameness. Severe cases may present with a “bow-legged” appearance. This is due to an improper development of the leg(s) as a result of the luxation.



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1- Patella	4- Tibia
2- Femur	5- Medial luxation
3- Patellar ligament	6- Lateral luxation

There are four grades used in the evaluation of the severity of a luxation. Grade 1 luxations mean the patella can be manipulated out of its groove, but returns to the normal position spontaneously. Grade 2-4 luxations indicate that the patella spontaneously rides out of its groove and may or may not be able to be replaced in proper position. The higher the grade, the more time the patella resides outside of groove. Owners with dogs having a grade 2-4 may need to consider surgical treatment.

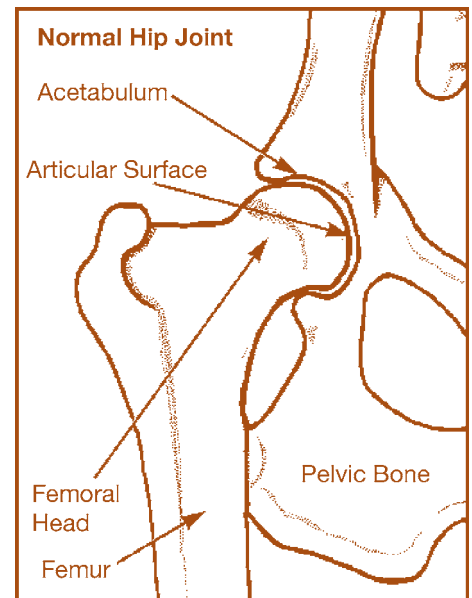
Surgical corrections aim to reconstruct the stifle joint for proper alignment and congruency of the quadriceps, femur, patella, tendons, and tibia/fibula.

Breed predispositions have been observed, indicating there is a genetic link to this condition. The Westie was one of 32 breeds identified as having an increased risk for developing patellar luxation by Drs. LaFond, Breur, and Austin. One recent study investigating patellar luxation in flat-coated retrievers suggested an autosomal recessive transmission. This was proposed by Dr. Herman Hazewinkel at Utrecht University. If luxating patellas are

determined to be a recessive condition, this is good news for eliminating the condition from breeding lines. With proper use of a genetic test, autosomal recessive conditions can be eliminated from breeding lines in two to three generations. This, of course, requires a genetic test, which is currently unavailable.

### Hip Dysplasia

Canine Hip Dysplasia (CHD) is the most common orthopedic disease of the dog. CHD is a degenerative disease characterized by an improper formation of the hip joint. Changes in hip joint laxity and conformation begin in the first few weeks after birth. CHD inevitably leads to osteoarthritis (OA), also known as degenerative joint disease (DJD). This is the result of



the femoral head having mobility in the socket, causing it to contact the edge of the acetabulum.

Clinical signs of CHD include painful hips between the ages of 6-8 months; lameness, particularly after exercise or prolonged confinement; reluctance to run, play or jump; tiring easily; exhibition of a “bunny-hop” gait; a loss of muscle mass in rear legs; a “popping” sensation felt over the hips.

The only definitive method for diagnosing CHD is the observation of arthritis in the hip joint. Unfortunately, this means that the hip joint must undergo painful structural changes before detection is possible.

Normal hips shown on left, dysplastic hips on right. Notice the dislocation of the right hip (left hip as you view picture). Femoral head is not seated properly in the acetabulum. You do not observe the typical ball-in-socket joint congruency.



There are several methods of evaluating the hip joints, which you can discuss with your veterinarian. Each method has associated pros and cons. These methods include (but are not limited to): ventrodorsal hip extended view (VHE), University of Pennsylvania Hip Improvement Program (PennHIP) method (illustrated in images to left), and dorsolateral subluxation (DLS) test.

There are many courses of treatment to consider in a dog afflicted with CHD. These options should be discussed with

your veterinarian as to which would be most suitable for you and your dog's needs.

CHD is a complex trait. There are numerous laboratories researching CHD and several



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regions of interest have been identified. Nutrition is also known to play a role in disease development. To date, no specific genes have been implicated from these studies and no mutations have been described in CHD-affected dogs.

Due to the multifactorial nature of this condition (genetics + environment), CHD has been very challenging to selectively breed away from. General recommendations for breeding programs trying to eliminate CHD from their lines include:

- Breed the best to the best
- Breed for the whole dog, not just for a single trait
- Keep detailed breeding records, including testing (OFA hips/patellas, etc.)
- Dysplastic dogs are not recommended for breeding

### Legg-Calve-Perthes Disease

Legg-Calve-Perthes Disease (LCPD) is a form of osteonecrosis of the hip. LCPD is a disease of many names: ischemic necrosis of the hip, coxa plana, osteochondritis, avascular necrosis of the femoral head, and Perthes Disease. LCPD affects puppies, generally between the ages of 4 and 11 months, and is limited to toy and miniature breeds. This disease can affect one or both hips (unilateral or bilateral).



Clinical signs of LCPD include a sudden onset of non-weight bearing lameness without previous

trauma, pain in the hip, muscle atrophy, and crepitus (crackling or grating feeling in joint) of the hip during palpation. Some cases of LCPD are subclinical, meaning they show no outward clinical signs.



Diagnosis of LCPD is typically made by evaluation of radiographs (x-rays) or magnetic resonance imaging (MRI). These visualization techniques are coupled with palpation of the hip and the evaluation of range of motion and observance of clinical signs in the patient. In the image to the right, notice that the right femoral head (left as viewed) is misshaped, has planar angles, and has a widened joint space as compared to the left.

LCPD has an unknown etiology, meaning we do not know what initiates the disease. Infection, trauma, metabolic or hormone imbalances, vascular abnormalities, and genetics have all been proposed origins of the disease.

Several treatment options are available for cases of LCPD. The goal of treatment is to (1) reduce hip irritability, (2) restore and maintain hip mobility, (3) prevent the femoral head from collapsing, and (4) regain proper spherical femoral head conformation. This can be accomplished in a manner of ways:

- **Conservative** – The primary objective in conservative treatment is to reduce the motion in the hip joint. This requires rest and limited exercise, crate rest, controlled walks on a

leash, and no vigorous play. Pain medicine and anti-inflammatory medication are given as needed.

- **Femoral Head Osteotomy (FHO)** – This surgical procedure involves the excision of the femoral head and neck. Over time, a false joint of muscle and tissue will form.
- **Total Hip Replacement (THR)** – This procedure is newly available for small breeds. Here, diseased bone is removed and replaced with an artificial joint. BioMedtrix is the sole provider of hip replacement for small breed dogs. (For more information, see BioMedtrix.com; search CFX™ Micro Hip System)

Our lab has started a research

project studying LCPD in several breeds of dog, including the Westie. We have collected

24 samples in total for the project: 14 affected and 8 unaffected samples. Of those, 7 affected and 6 unaffected samples are from Westies. We are asking for a small volume of



blood from each participating dog, a copy of the pedigree, and a copy of radiographs used to diagnose LCPD affected dogs.

One of our aims is to identify the mode of inheritance of LCPD in several breeds. There are reports of an autosomal recessive inheritance with incomplete penetrance (meaning not all dogs who have the disease alleles develop the disease) as well as a multifactorial inheritance, meaning there would be major and minor genes in addition to environmental factors which contribute to the disease. We are collecting pedigrees with each sample submitted to complete transmission studies of LCPD and identify the mode of inheritance in Westies. Our initial analyses using only a few



pedigrees were inconclusive – we did not have enough affected dogs to draw any conclusions about inheritance. We are continuing to collect pedigrees to complete this aim.

Our second goal is to identify candidate genes that may either cause the disease or be involved in the disease process. Our planned approach for the study of LCPD is a whole-genome association mapping strategy, where we examine molecular markers (think: genetic mile markers) that will identify regions of interest, or regions where all the affected dogs have the same markers in a unique pattern from the unaffected dogs.

Our ultimate goal from this work is to identify the mutation causing LCPD. This would allow us to develop a genetic test that would enable early classification of affected dogs as well as detection of carrier dogs. Breeders would then be able to make informed decisions in their breeding programs and

eliminate LCPD from their breeding lines. Any owners with questions regarding

this study or willing to participate in the study should contact Dr. Alison Starr ([astarr@clemson.edu](mailto:astarr@clemson.edu)) or refer to the Westie Foundation of America website for more information.

