

# MY RESULTS



# HEALTH



Name	Results
Lundehund Syndrome	<b>Variant present</b> >
X-Linked Severe Combined Immunodeficiency (IL2RG gene, Cardigan Welsh Corgi)	<b>Variant present</b> >
Complement 3 Deficiency	Variant absent >
Cystinuria Type I-A (SLC3A1 gene)	Variant absent >
Cystinuria Type II-A (SLC3A1 gene)	Variant absent >
Cystinuria Type II-B (SLC7A9 gene)	Variant absent >
Degenerative Myelopathy (SOD1 gene)	Variant absent >
Epidermolytic Hyperkeratosis	Variant absent >
Glanzmann Thrombasthenia (ITGA2B gene, Pyrenean Mountain Dog)	Variant absent >
Globoid Cell Leukodystrophy (GALC gene, Irish Setter)	Variant absent >
Globoid Cell Leukodystrophy (GALC gene, Terriers)	Variant absent >
Glycogen Storage Disease Type IIIa (AGL gene)	Variant absent >
Hemophilia A (F8 gene, Boxer)	Variant absent >
Hemophilia A (F8 gene, German Shepherd)	Variant absent >
Hemophilia A (F8 gene, Old English Sheepdog)	Variant absent >
Hemophilia B (F9 gene)	Variant absent >
Hemophilia B (F9 gene, Rhodesian Ridgeback)	Variant absent >
Hereditary Footpad Hyperkeratosis (FAM83G gene)	Variant absent >
Hyperuricosuria (SLC2A9 gene)	Variant absent >
Hypocatalasia	Variant absent >
Intestinal Cobalamin Malabsorption (CUBN gene, Beagle)	Variant absent >
Intestinal Cobalamin Malabsorption (CUBN gene, Komondor)	Variant absent >
May-Hegglin Anomaly	Variant absent >
Mucopolysaccharidosis Type VII (GUSB gene, Brazilian Terrier)	Variant absent >
Mucopolysaccharidosis Type VII (GUSB gene, German Shepherd)	Variant absent >
Muscular Dystrophy (DMD gene, Golden Retriever)	Variant absent >

# Lundehund Syndrome

Lundehund`s intestinal syndrome is a stomach and intestinal pathology that causes malabsorption and protein deficiency.

Your result is

Variant present

Gene	Chromosome	Position	Interpretación
P3H2	34	22046092	It has one copy of the c.1849G>C mutation in the P3H2 gene associated with lundehund syndrome. A dog with one copy of the mutation would not develop the disease but can transmit the mutation to its offspring.

## Symptoms

This syndrome typical of Lundehund dogs includes symptoms such as low serum protein levels, vomiting, loss of weight and muscle mass, lack of appetite, lethargy, fluid accumulation in the abdomen and swollen paws. Symptoms can appear at any age between 6 months and 10 years. It can eventually lead to lymphoma, cancer of the intestine or stomach.

## Disease management

There is no cure for this condition, and management usually involves dietary changes, vitamin supplementation, administration of anti-inflammatory drugs and drugs to reduce the accumulation of cells in the intestines, improve lymph flow and eliminate harmful intestinal bacteria. The most common treatments are prednisone, metronidazole, cimetidine, FOS, Forti-flora and B12 injections. Treatment varies from dog to dog and the prognosis is highly variable, ranging from mild chronic diarrhea, often recurrent, to severe symptoms resistant to treatment.

## Genetic basis

This disease follows an autosomal recessive mode of inheritance. Autosomal recessive inheritance means that the dog, regardless of sex, must receive two copies of the mutation or pathogenic variant to be at risk of developing the disease. Both parents of an affected dog must carry at least one copy of the mutation. Animals with only one copy of the mutation are not at increased risk of developing the disease, but may pass the mutation on to future generations. Breeding between dogs carrying genetic variants that can cause disease, even if they do not show symptoms, is not recommended.

## Technical report

The exact cause of Lundehund syndrome is not fully understood, it is thought to be related to a combination of genetic and environmental factors. In 2016, the study by Metzger et al. was published which concluded that the c.1849G>C mutation in the P3H2 gene, which is the one we discuss here, may be the cause of the syndrome.

The P3H2 gene (previously known as LEPREL1) was expressed in certain neuroendocrine cells found in the intestinal mucosa and it has been suggested that it may play an important role in the processing and secretion of neuropeptides. Alteration of P3H2 could be related to changes in intestinal neuropeptide production that could be key in this and other inflammatory bowel diseases.

Most affected breeds

- Lunde hund

Bibliography

[Metzger J, Pfahler S, Distl O.](#) Variant detection and runs of homozygosity in next generation sequencing data elucidate the genetic background of Lunde hund syndrome. BMC Genomics. 2016 Aug 2;17:535.

## X-Linked Severe Combined Immunodeficiency (IL2RG gene, Cardigan Welsh Corgi)

Severe combined immunodeficiency or X-linked SCID is a rare genetic disease characterized by a dysfunctional or absent immune system and alterations in lymph node development that may be absent.

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Your result is

Variant present

Gene	Chromosome	Position	Interpretación
IL2RG	X	55483460	She has the c.583_584insC mutation in the IL2RG gene associated with X-linked severe combined immunodeficiency. Increased risk of developing the disease.

### Symptoms

There is a deficit in the production of T and B lymphocytes, the immune cells that fight infection and disease. This means that affected puppies are highly susceptible to infections, even those that would not normally be problematic for a dog with a healthy immune system. The first symptoms can appear from birth and are severe.

Disease management

There is currently no cure for X-linked SCID. Given the severity of the symptoms puppies usually do not survive beyond 4 months.

Genetic basis

This disease follows an X-linked recessive mode of inheritance. Recessive X-linked inheritance means that female dogs must receive two copies of the mutation or

pathogenic variant (one from each parent) to develop the disease, whereas males need only one copy of the mutated gene or variant from the dam to develop the disease. Male dogs usually show symptoms of the disease. Each male puppy born to a mother carrying the mutation has a 50% chance of inheriting the mutation and thus the risk of developing the disease. Bitches that do not carry the mutation are not at increased risk of having affected puppies. Breeding between dogs carrying genetic variants that can cause disease, even if they do not show symptoms, is not recommended.

#### Technical report

X-linked severe combined immunodeficiency is a failure of cellular and humoral immune responses caused by mutations in the IL2RG gene, resulting in the absence of T cells and dysfunctional B lymphocytes. As in humans, this disease in dogs causes affected animals to have serious immunity problems and die within a few weeks of life. The IL2RG gene encodes for the gamma subunit of the interleukin 2 receptor which is an essential molecule for the proper functioning of the immune system. The gamma subunit interacts with molecules known as cytokines that promote the proliferation, differentiation, survival and function of lymphocytes. Here we analyzed the c.583\_584insC mutation causing immunodeficiency in cardigan welsh corgi dogs (Somberg et al., 1995).

#### Most affected breeds

- Cardigan Welsh Corgi

#### Bibliography

[Somberg RL, Pullen RP, Casal ML, et al.](#) A single nucleotide insertion in the canine interleukin-2 receptor gamma chain results in X-linked severe combined immunodeficiency disease. *Vet Immunol Immunopathol.* 1995 Aug;47(3-4):203-13.

# TRAITS



Name	Results
Chocolate coat 1	Variant present >
Chocolate coat 2	Variant present >
Chocolate coat 3	Variant present >
Curly coat	Variant present >
Dark mask	Variant present >
Dominant black coat	Variant present >
Ears position	Variant present >
Grizzle coat	Variant present >
Long hair	Variant present >
Piebald colouring pattern	Variant present >
Recessive black coat	Variant present >
Recessive red coat	Variant present >
Short tail	Variant present >
Black and tan colour pattern	Variant absent >
Coat with fire points	Variant absent >
Harlequin colour pattern	Variant absent >

# Chocolate coat 1

The chocolate 1 or bc variant is responsible for providing a chocolate brown color to the coat, eye rims, nose, paws and paw pads.

Your result is

Variant present

Gene	Chromosome	Position	Interpretación
TYRP1	11	33317810	It has one copy of the c.121T>A variant in the TYRP1 gene. A dog with one copy of this variant would not have the trait but can transmit the mutation to its offspring.

## Definition

The so-called chocolate variant manifests itself in dogs as a deep brown or chocolate colouration of the coat. Dogs with this variant will also express brown pigmentation of the nose, footpads and eye rims. This phenotype results from alterations in the TYRP1 gene (also known as the B locus), which is involved in the production of the pigment responsible for black or brown colouration in dogs (eumelanin).

### Genetic basis

The chocolate 1 variant is also known as the "bc" allele. It is necessary for the dog to inherit two copies of the c.121T>A variant to produce brown pigment instead of black. It is possible that there are other variants in the TYRP1 gene involved in the development of the trait which, in combination with the variant described here, result in the chocolate coloured coat. The presence of a single copy of the c.121T>A variant would not result in the expression of the above phenotype, however, it can be passed on to the offspring of the dog with a 50% probability.

### Other relevant information

It is curious that in the Vizsla breed, most dogs have a brown nose both in the presence and absence of variants in the TYRP1 gene. There are other alternative names for this trait such as liver, brown or chestnut. Breeds commonly showing the trait are Australian Shepherd, German Shepherd Dog, Lancashire heeler, Leonberger, Miniature American Shepherd and Siberian Husky.

### Bibliography

[Schmutz SM, Berryere TG, Goldfinch AD.](#) TYRP1 and MC1R genotypes and their effects on coat color in dogs. Mamm Genome. 2002 Jul;13(7):380-7.

# Curly coat

The curly coat is a trait characterized by the presence of more or less defined curls in the hair.

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Your result is

### Variant present

Gene	Chromosome	Position	Interpretación
KRT71	27	2539211	It has one copy of the c.451C>T variant in the KRT71 gene. A dog with one copy of this variant is more likely to express a wavy coat.

#### Definition

The curly coat trait in dogs is a phenotype characterized by elastic curls that can be more or less tight. The variation responsible for this trait is located in the KRT71 gene which encodes a structural protein that confers strength and elasticity to the coat.

#### Genetic basis

The presence of one or two copies of the c.451C>T variant results in the manifestation of the trait in dogs. In the case of a single copy, the coat is likely to be more wavy in texture, whereas the presence of two copies may result in a tighter curl. If the c.451C>T variant is not found, the coat is likely to be smooth; however, the existence of other genetic variants could influence this outcome.

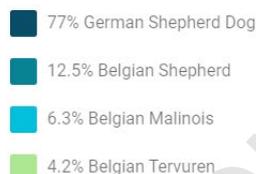
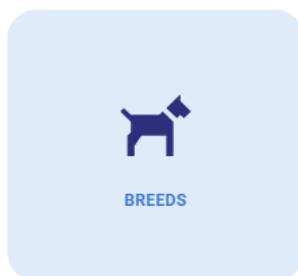
#### Other relevant information

The length of the dog's coat must be long enough to be able to curl, in fact, all the dogs analyzed in the study by Cadieu, E. et al. (2009) with curly hair presented the variant related to long hair. Breeds that commonly possess the trait are Portugese Water Dog, Airedale Terrier, Irish Water Spaniel and Bichon Frisé.

#### Bibliography

[Cadieu E, Neff MW, Quignon P, et al.](#) Coat variation in the domestic dog is governed by variants in three genes. *Science*. 2009 Oct 2;326(5949):150-3.

## BREEDS



+ German Shepherd Dog

+ Belgian Shepherd

+ Belgian Malinois

+ Belgian Tervuren

## German Shepherd

The German Shepherd is an intelligent, loyal and versatile breed of dog, originally from Germany. Physically, they are characterized by their double black coat with brown spots. In addition, they are known for their ability to perform a variety of tasks from herding to family companionship.

### General details

Male German Shepherds usually weigh between 30 and 40 kg, while females usually weigh between 22 and 32 kg. The height at the withers of males varies between 60 and 65 cm, while females usually measure between 55 and 60 cm. The average life expectancy is approximately 10-12 years. According to the Fédération Cynologique Internationale (FCI), the German Shepherd belongs to Group 1, which includes Shepherd Dogs and Cattle Dogs (except Swiss Cattle Dogs).

### Brief history of the breed

The German Shepherd was developed in Germany in the late 19th century by Max von Stephanitz, a former cavalry captain who wanted to create a highly functional and versatile working breed of dog for herding sheep. Von Stephanitz selected and crossed different

breeds of local sheepdogs, looking for dogs that had exceptional working abilities, intelligence and a strong, balanced constitution. Throughout the 20th century, the popularity of the German Shepherd grew rapidly, and the breed began to be used in roles other than herding, such as police, military, search and rescue, and as guide dogs for the visually impaired, roles in which dogs of this breed continue to participate today.

### Breed characteristics

The German Shepherd is a medium sized dog breed with an athletic build. Its head is wedge-shaped with almond-shaped eyes, medium size and dark color. Its ears are erect, pointed and medium-sized. The tail is of moderate length and, at rest, is usually carried hanging with a slight curve, while, in moments of excitement, it rises without exceeding the topline. The coat is double-coated and may be short or of medium length. The most common colors in this breed include black with reddish brown, brown and yellow markings and even light gray. They are characterized by their intelligence, loyalty, versatility and working abilities, making them suitable for a wide range of tasks, such as herding, search and rescue, and service to people with disabilities, as well as being excellent companion and family protection dogs.

### Common health problems

The German Shepherd, with optimal care and attention, can live a long healthy life, although it is predisposed to certain health problems. Among the most common diseases or disorders are hip and elbow dysplasia, degenerative myelopathy, inflammatory bowel disease, allergies and panosteitis.

### Belgian Shepherd

### Belgian Malinois

## Belgian Tervuren

The Belgian Shepherd breed originates from Belgium, where it was bred for the guarding and protection of livestock. The long coat is a hallmark of the breed, with a thick collar around the neck and bangs on its legs. Dogs of the breed are versatile and intelligent and are noted for their strong work ethic.

### General details

Dogs of the breed are medium sized and balanced. The weight ranges between 25 and 34 kilograms in males, while females weigh between 20.5 and 27 kilograms. Males measure between 61 and 66 centimeters, and females around 60 and 61 centimeters. Life expectancy is 12 to 14 years. They belong to Group 1 of the Fédération Cynologique Internationale (FCI), which groups Sheepdogs and Cattle Dogs (except Swiss Cattle Dogs).

### Brief history of the breed

The Belgian Shepherd originated in Belgium at the end of the 19th century, where they were originally bred to herd and protect livestock. At that time there were eight types of Belgian Shepherd Dogs, however, after being officially classified, there were only the four that we know today: Belgian Shepherd, Tervuren, Laekenois and Malinois. The breed is also known as Groenendael, this name was used by Nicolas Rose (an important breeder) to name the long-haired black variety that is identified with the modern Belgian Shepherd. During the First and Second World War they were used as ambulance dogs, messengers and armament

carriers. Today, it is one of the most popular breeds in Belgium and around the world for excelling in a variety of roles such as athlete, police, military, searcher, guard or show dog.

### Breed characteristics

The Belgian Shepherd breed is a square, proud and medium sized dog. The male usually shows a more imposing appearance than the female. The size of the head is proportional to the body with brown eyes, of medium size and slightly almond-shaped. The ears are triangular in shape, erect and proportional to the size of the head. The tail is held low at rest, with the tip bent backwards. When the dog is in motion, the tail is raised with a curl, but without forming a hook. The coat is double, the undercoat is usually extremely dense and adapted to climatic conditions. The outer coat is long, smooth, dense and of medium harshness. The hair is of shorter length on the outside of the ears, the head and the underside of the legs. While, the longer length hair is located on the neck (forming a collar), and has abundant fringe on the back of the forearms, hindquarters and tail. The usual coloration of the Belgian Shepherd is black, which can be solid or combined with small white patches. The coat color can turn reddish as a result of weather conditions. The Belgian Shepherd is a well-known herd guardian dog, although they also act to protect people and their owners` property. Alertness, loyalty and courage are also qualities to be highlighted in this guard dog. In addition, they are intelligent and possessive dogs, as well as friendly and affectionate with acquaintances. Loneliness and lack of exercise can affect the breed`s behavior.

### Common health problems

The Belgian Shepherd, with optimal care and attention, can enjoy a long healthy life, although it shows a predisposition to suffer from certain health problems. Among the most common diseases or disorders are epilepsy, cataracts, hip dysplasia, hypothyroidism, progressive retinal atrophy, retinal dysplasia, X-linked muscular dystrophy and chronic superficial keratitis.