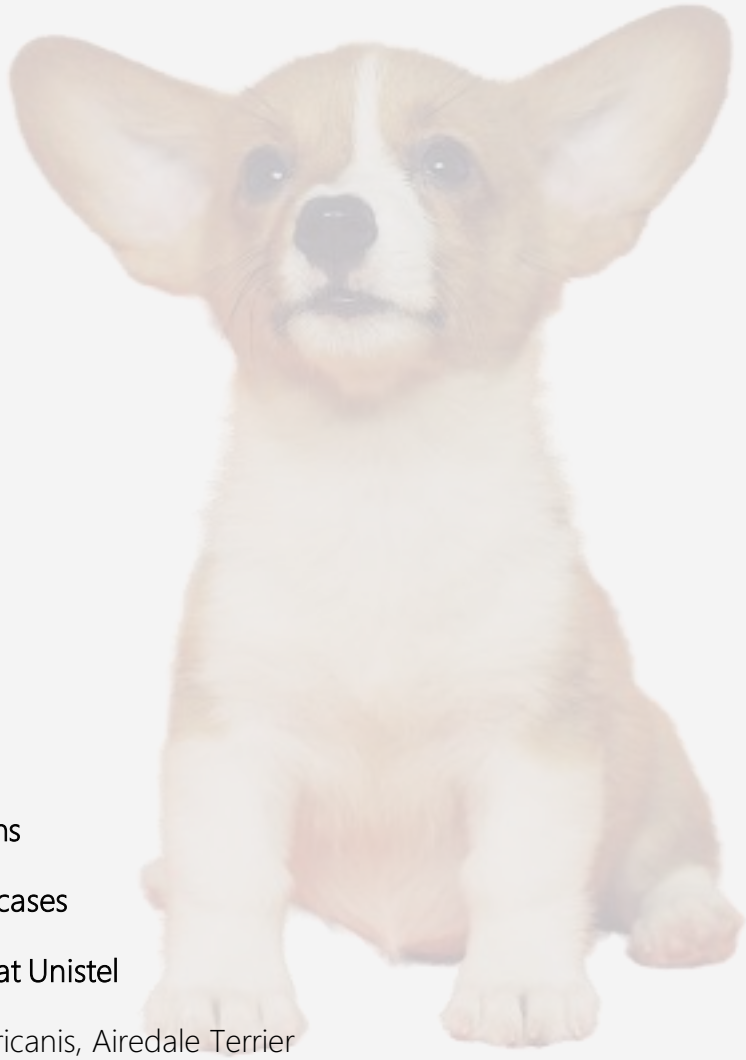
Unistel® PETS

Paws | Passion | Precision

CANINE TESTING INFORMATION HANDBOOK

Contents:



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Sample Taking Guidelines:

Buccal swabs – Unistel DNA kit:

1. Wait approximately 2 hours after meal or treat before collecting DNA samples.
2. Twist open the swab encasing.
3. Obtain cheek cells by firmly rolling swab between the cheek and gums for approximately 15 seconds. Pets can be kept comfortable by swaddling them in a blanket/towel.
4. Allow the swab to air dry for 5 minutes. Repeat steps 2 & 3 for the second swab.
5. Reinsert the dried swabs into their protective covers.
6. Label the casings with your pet's details.
7. Complete the sample request forms provided.
8. Place the swabs and completed forms in the DNA kit box.
9. Mail/courier the samples to Unistel Medical Laboratories.

Pulled hairs:

A minimum of 15 - 20 hairs with follicles/roots are needed for the successful isolation of DNA. The best places to sample are respectively the shoulders, flank and forehead although any body hair is functionally suitable. Hair can be gripped between the thumb and forefinger as close to the skin as possible and pulled against the grain. This will minimize the risk of breakage and losing the follicle, in which the DNA of the hair is stored. Please take note that certain flea and tick treatments may inhibit downstream processing of your sample. It is advised to wait 2 – 3 weeks after the application of flea and tick treatments before taking a hair sample.

Blood spots:

It is advised that a blood spot no smaller than the size of a forefinger nail should be blotted onto the Guthrie card. For best results the blood should soak through the card. Allow the blood to dry on the Guthrie cards before placing them in the envelopes.

Please take note:

Samples should preferably be packaged in a paper envelope. If possible, avoid plastic bags as moisture build-up can lead to fungal growth on the samples which will render it unusable. Please only place one animal's sample per envelope/bag to avoid cross-contamination of samples. Samples should be clearly labelled with the information of the animal on the envelope/bag corresponding to the information provided on the sample submission form. For repeat samples, please attach the email requesting the samples for repeat to the new sample submission paperwork.

Coat Colour & Traits offered at Unistel

Coat Colour and Trait Testing Offered:

Trait	Normal	Mutation	Chr	Location on chormosome	Gene
Coat colour, agouti ressessive black	C	T	CHR24	23393552	ASIP
Coat colour, agouti	GCTCG	TCTCA	CHR24	23393510	ASIP
Coat colour, agouti SINE insertion	No SINE	SINE insertion	CHR24	Not provided	ASIP
Coat colour, dominant black	CCC	del	CHR16	58965449	CBD103
Coat colour, brown - Bd type	CCT	del	CHR11	33326727	TYRP1
Coat colour, brown - Bs type	C	T	CHR11	33326685	TYRP1
Coat colour, brown - Bc type	T	A	CHR11	33317810	TYRP1
Coat colour, dilute - D1 type	G	A	CHR25	48121642	MLPH
Coat colour, extension melanistic mask	C	T	CHR5	63694460	MC1R
Coat colour, extension - E1 type (red/yellow)	G	A	CHR5	63694334	MC1R
Coat colour, harlequin	T	G	CHR9	58530295	PSMB7
Coat colour, Merle	No SINE insertion	SINE insertion	CHR10	Not provided	PMEL
Coat colour, white spotting, KIT-related	-/-	ins A	CHR13	47144514	KIT
Alopecia, colour mutant	G	A	CHR25	48150678	MLPH
Hair, long - Eurasier type	GCGGGCCGGGAGTGGT	del	CHR32	4528617	FGF5
Hair, long - Afghan, Eurasier, Frenchies	-/-	GG	CHR32	4528622	FGF5
Hair, long - Akita, Samoyed, Eurasier, Husky type	C	T	CHR32	4528634	FGF5
Hair length - most breeds	G	T	CHR32	4509367	FGF5
Curly coat	C	T	CHR27	2539211	KRT71
Improper coat	ATGGCTTCTTATCTCATAGCAATAG	del	CHR13	8610420	RSPO2
Tail, short - Corgi type (Dominant, homozygous lethal)	C	G	CHR1	54192143	T

Canine Coat Colouration

Coat colour in canines is determined by several genes within the canine genome. These genes are able to produce the wide variety of coat colours observed by affecting the expression of the two primary coat colour pigments, **Eumelanin (black)** and **Phaeomelanin (red)**. How much, and where, these pigments are expressed forms the basis for all coat colours and patterns that have been observed within canines.

Genes associated with Canine coat colour diversity

Five genes in particular are known to have a large effect on coat colour, and are thus the most commonly tested for. These include the **Pigment-type switching genes** (A Locus, E Locus and K Locus), as well as the **Pigment dilution genes** (B Locus and D Locus).

Pigment-type switching:

As the name suggests, the pigment-type switching genes determine which of the pigment types, black or red, are expressed. The **K Locus** controls the production of black pigment, and is largely dominant over the expression of the other coat colour genes when in its dominant form. Alternatively, the **A Locus** is responsible for excessive red/yellow pigment production in its most dominant form, and black in its most recessive form. In its dominant form, the **E Locus** facilitates the synthesis of black hairs, while its recessive form allows only the synthesis of red/yellow hairs.

Pigment dilution genes:

Rather than determining pigment type, the two pigment dilution genes are responsible for the dilution, or lightening, of the base coat colour. The **B locus** dilutes black coats to brown, while the **D locus** dilutes black coats to blue, brown coats to isabella, and tan coats to champagne.

Pigmentation *vs* non-pigmentation:

As mentioned above, the black and red pigments are responsible for producing the wide variety of dog coat colours present within the population. However, when no pigment is produced, the hairs appear white. When no black pigment is produced in the nose, the dog may present with a pink nose, and if not in the eyes, with blue eyes. In rare cases, the entire body of the dog is affected with a lack of pigment, which results in an albino.

Canine Coat Colouration

K Locus: Dominant black (*CBD103* gene)

The K locus is the predominant locus responsible for producing black coat colour in canines, and as the name suggests, tends to be expressed in a dominant fashion relative to the other coat colour genes. The dominant allele, **K**, will result in a black coat, while the recessive wild type allele, **N**, will allow recessive red/yellow or non-solid black to be expressed (see Table 1).

Rules for expression:

- The E locus will affect the expression of the K locus:
    - The K locus will not be expressed in dogs that are **e/e** at the E Locus .

Table 1: Interpretation of each mutation region within the K locus.

Mutation tested:	Variant possible at mutation region and their interpretation		
K/N mutation: c.231_233del	CCC/CCC **N/N** Red/yellow or non-solid black coat	CCC/ - **N/K** Solid black coat colour	- / - **K/K** Solid black coat colour

Canine Coat Colouration

A Locus: Agouti (*ASIP* gene)

When activated, the A locus influences coat colour by causing a switch from the production of black pigment to red/yellow. This switching can occur uniformly across the entire body, or in a particular pattern, depending on the variant present.

There are **4 mutations** at the A locus, which, when combined, correspond to **4 agouti alleles** (see Table 2). These are expressed in the coat according to the following dominance hierarchy: $\mathbf{a^y} > \mathbf{a^w} > \mathbf{a^t} > \mathbf{a}$.

Rules for expression:

- The E locus and K locus will both affect the expression of the A locus:
  - The A locus (with the exception of **a/a**) will not be expressed in dogs that are **e/e** at the E Locus.
  - The A locus will not be expressed in dogs that are **K/K** or **K/N** at the K locus.

Table 2: In order for one of the four A locus alleles to be scored, they require all mutations listed for the specific allele to be present.

Allele	Colour produced	Mutation 1 A82S	Mutation 2 R83H	Mutation 3 SINE	Mutation 4 R96C
a^y	Fawn/sable	c.246T	c.250A	No SINE insertion	c.286C or c.286C/T
a^w	Agouti wolf*	c.246G	c.250G	No SINE insertion	c.286C or c.286C/T
a^t	Black & tan	c.246G	c.250G	SINE insertion present	c.286C or c.286C/T
a	Recessive black	c.246G or T	c.250G or A	SINE insertion present	c.286T

* It is often difficult to distinguish between agouti wolf and a shaded sable. The main difference is that a shaded sable will not have the visible banding pattern on the hairs.

E Locus: Extension (*MC1R* gene)

Similarly to the A and K loci, the E locus influences the production of pigment. However, rather than determining the pigment type directly, the E locus simply encodes the ability or inability to produce darker pigment. The wild type dominant allele, **E**, facilitates the production of black pigment, while the most recessive allele, **e**, allows only the production of red/yellow pigment. Other alleles at the E locus, such as **E^m** (melanistic mask), affect the expression of coat colour patterns on the face. These are expressed in the coat according to the following dominance hierarchy: **E^m** > **E** > **e**

There are **two mutation regions** within the E locus, with 2 known alleles at each region that are responsible for pigment alterations (see Table 3).

Rules for expression:

- The A locus will affect the expression of the E locus:
    - The E locus will not be expressed in dogs that are **a/a** at the A Locus.

Table 3: Interpretation of each mutation region within the E locus.

Mutation tested:	Variant possible at mutation region and their interpretation		
E^m/E mutation: c.790A>G g.63694460T>C	T/T **E/E** No mask	T/C **E/E^m** Melanistic mask	C/C **E^m/E^m** Melanistic mask
E/e mutation: c.916C>T g.63694334G>A	G/G **E/E** Black coat possible	G/A **E/e** Black coat possible	A/A **e/e** Red/yellow coat

Canine Coat Colouration

B Locus: Brown (*TYRP1* gene)

The B locus is responsible for the dilution of black pigment to brown (also referred to as liver, or chocolate), and has 3 mutation regions, **B^{d}**, **B^{s}** and **B^{c}**, with 2 known alleles at each region. The dominant allele, **B**, produces a black coat and nose, while the recessive allele, **b**, results in a brown coat and nose. The 3 mutation regions should always be viewed together in order to interpret the B locus (see Table 4).

Rules for expression:

- The E locus and K locus will both affect the expression of the B locus:
    - The B locus will not be expressed in dogs that are **e/e** at the E Locus.
    - The brown pigment is only produced when any two recessive **b** alleles are present at any of the 3 mutation regions. *E.g.* A dog with **B/b** at **B^{d}** and **B/b** at **B^{c}** will be brown.

Table 4: Interpretation of each mutation region within the B locus. Dogs need only 2 copies of the recessive allele from any of the mutation regions in order to produce a brown coat.

Mutation tested:	Variant possible at mutation region and their interpretation		
B^{d} region: c.1033_1035del	CCT/CCT **B/B** Black coat colour	CCT/ - **B/b** Can produce brown if paired with another recessive allele	- / - **b/b** Brown coat colour
B^{s} region: c.991C>T g.33326685C>T	C/C **B/B** Black coat colour	C/T **B/b** Can produce brown if paired with another recessive allele	T/T **b/b** Brown coat colour
B^{c} region: c.121T>A g.33317810T>A	T/T **B/B** Black coat colour	T/A **B/b** Can produce brown if paired with another recessive allele	A/A **b/b** Brown coat colour

D locus: Dilution (*MLPH* gene)

The D locus is responsible for producing lighter, or diluted, coat colours other than brown. The dominant allele at this locus, **D**, produces dogs with no dilution, while the recessive allele, **d**, results in a paler version of the base coat colour.

Rules for expression:

- Dogs must have 2 copies of the recessive allele, **d/d**, in order for the coat colour to be diluted (see Table 5).

Table 5: Interpretation of the effect of the D locus on the respective pigment types. Dogs need 2 copies of the recessive allele in order to produce a dilution of the base coat colour.

Base coat colour	D/D	D/d	d/d
Black	Black	Black	Blue
Brown	Brown	Brown	Isabella
Tan	Tan	Tan	Champagne

Interpreting the A, E, K, B and D loci

Grouped on the following pages are tables indicating the interpretation of all 5 coat colour genes together. Starting with the K Locus (far left), each column to the right indicates the contribution of an additional locus, as well as the effect that this locus will have on the coat colour based on the loci before it. The colours indicated on the far right are the final coat colour.

Co Locus: Cocoa (*HPS3* gene)

Brown, or chocolate, coat colour is produced by variants at the B locus (*TYRP1* gene) in many animal species. In dogs, five different B locus alleles have been described, which explain the vast majority of dogs with brown coats. Recently, however, a novel variant has been identified within brown French Bulldogs that did not carry any of the known Brown alleles. Instead, the new allele was located in the *HPS3* gene, with the dominant wild type allele, **N**, resulting in a non-brown coat, and the recessive allele, **Co**, resulting in a brown coat, referred to as "Cocoa" (see Table 11).

Rules for expression:

- Dogs must have 2 copies of the recessive allele, **Co/Co,** in order for the coat colour to be Cocoa.

Table 11: Interpretation of each combination of variants at the Co locus. Dogs need 2 copies of the recessive allele in order to produce a Cocoa coat.

Mutation tested	Variant possible at mutation region and interpretation			Breeds affected
N/Co mutation: c.2420G>A	G / G N/**N** Non-Cocoa coat	G / A **N/Co** Non-Cocoa coat	A / A **Co/ Co** Cocoa coat	French Bulldog

Canine Coat Colouration

M Locus: Merle (*SILV* gene)

In canines, the *SILV* gene is responsible for causing the highly variable Merle phenotype. The Merle variant contains a short interspersed nucleotide element (SINE) insertion, which disrupts the normal expression of pigment and causes the production of a solid base coat colour with lighter blue/grey or reddish patches. Blue or heterochromic eyes are also possible, as well as mottled pink/black paw pads and noses. The dominant allele, **M**, produces dogs with the Merle phenotype, while the recessive wild type allele, **m**, results in a solid coat. The extent to which the phenotype is expressed, however, is dependent on which Merle allele/s are present (see Table 12). Two Merle alleles can result in a much more extreme phenotype, with "double Merle" dogs potentially suffering from hearing and eye defects. The occurrence of these defects is significantly more likely in dogs that possess the larger alleles (M and Mh), but caution should still be used when breeding with the intermediary alleles (Ma and Ma+).

Table 12: Interpretation of the major differences in phenotype caused by the 7 different alleles at the Merle locus. For the larger M alleles, dogs need only one copy to produce the Merle phenotype.

Allele	Phenotype	
	One copy (Heterozygous with m allele)[1]	Two copies
m	No *SILV* SINE insertion - No Merle pattern, solid coat	
Mc	No Merle pattern, solid coat	No Merle pattern, Possible: Faded/brownish[2] coat
Mc+	No Merle pattern, solid coat	No Merle pattern, Possible: Faded/brownish[2] coat
Ma	No Merle pattern, Possible: Brownish[2]/diluted[3] coat, lighter shaded areas, blue eyes	No Merle pattern, Common: Brownish[2]/diluted[3] coat, Possible: More extensive lighter shading, blue eyes
Ma+	Common: Brownish[2]/diluted[3] coat, Possible: **Muted Merle** pattern, lighter shaded areas, blue eyes	Common: **Muted Merle** pattern, brownish[2]/diluted[3] coat, Possible: More extensive lighter shading, blue eyes, Tweed pattern[4], areas of deleted pigment (white)
M	**Classic Merle** pattern, Possible: Blue eyes	**Classic Merle** pattern, Possible: Blue eyes, areas of deleted pigment (white), health defects
Mh	**"Minimal Merle"**[5] / **"Herding Harlequin"**[6] patterns, Possible: Blue eyes, health defects	**"Minimal Merle"**[5] / **"Herding Harlequin"**[6] patterns, Possible: More extensive white areas, blue eyes, health defects

[1] Two alleles with different SINE insertions are possible. Phenotypes may vary from what is presented here.
[2] Independent of b/b genotype at Brown locus (*TYRP1* gene).
[3] Independent of d/d genotype at Dilution locus (*MLPH* gene).
[4] Patchwork pattern of brown, grey, white, and tan shades.
[5] Mostly solid coat with small, random Merle patches. Extensive white areas possible.
[6] Random areas of Merle pigment deleted to white, Tweed pattern possible. Extensive white areas possible.

Canine Coat Colouration

Table 6: Interpretation of coat colours for dogs carrying the **dominant black** (K) allele at the K locus.

K locus	E locus	A locus $a^y/a^w/a^t$	B locus		D locus	
K/K; K/N Dominant Black	E^m/E^m; E^m/E	**Black *** *Inhibited by dominant K locus*	B/B; B/b	Black	D/D; D/d	Black
					d/d	Blue
	E/E; E/e		b/b	Brown	D/D; D/d	Brown
					d/d	Isabella
	e/e	**Tan** *Inhibited by recessive E locus*	B/B; B/b; b/b	**Tan** *Inhibited by recessive E locus*	D/D; D/d	Tan
					d/d	Champagne

* Black mask present, but not visible.

Table 7: Interpretation of coat colours for dogs carrying only the **normal allele** (N) at the K locus, paired with a^y at the A locus.

K locus	E locus	A locus a^y/ -	B locus		D locus	
N/N	E^m/E^m; E^m/E	Sable* **Black mask**	B/B; B/b	Clear/ **Black** tipped/ shaded **Black mask**	D/D; D/d	Clear/**Black** tipped/shaded **Black mask**
					d/d	Clear/**Blue** tipped/shaded **Blue mask**
			b/b	Clear/ **Brown** tipped/ shaded **Brown mask**	D/D; D/d	Clear/**Brown** tipped/shaded **Brown mask**
					d/d	Clear/**Isabella** tipped/shaded **Isabella mask**
	E/E; E/e	Sable* **No mask**	B/B; B/b	Clear/ **Black** tipped/ shaded**	D/D; D/d	Clear/**Black** tipped/shaded**
					d/d	Clear/**Blue** tipped/shaded**
			b/b	Clear/ **Brown** tipped/ shaded**	D/D; D/d	Clear/**Brown** tipped/shaded**
					d/d	Clear/**Isabella** tipped/shaded**
	e/e	**Tan coat** *Inhibited by recessive E locus*	B/B; B/b; b/b	**Tan** *Inhibited by recessive E locus*	D/D; D/d	Tan
					d/d	Champagne

* Also referred to as Fawn.
** Most tipped dogs tend to have masks (E^m), although the link between the two is not genetically confirmed.

Canine Coat Colouration

Table 8: Interpretation of coat colours for dogs carrying only the **normal allele** (N) at the K locus, paired with **a^w** at the A locus.

K locus	E locus	A locus a^w/ -	B locus		D locus	
N/N	E^m/E^m; E^m/E	Wolf* **Black mask**	B/B; B/b	Wolf* **Black mask**	D/D; D/d	Wolf* **Black mask**
					d/d	Blue Wolf* **Blue mask**
			b/b	Brown Wolf* **Brown mask**	D/D; D/d	Brown Wolf* **Brown mask**
					d/d	Isabella Wolf* **Isabella mask**
	E/E; E/e	Wolf* **No mask**	B/B; B/b	Wolf*	D/D; D/d	Wolf*
					d/d	Blue Wolf*
			b/b	Brown Wolf*	D/D; D/d	Brown Wolf*
					d/d	Isabella Wolf*
	e/e	**Tan coat** *Inhibited by recessive E locus*	B/B; B/b; b/b	**Tan** *Inhibited by recessive E locus*	D/D; D/d	Tan
					d/d	Champagne

* Also referred to as Agouti or Wolf Sable.

Canine Coat Colouration

Table 9: Interpretation of coat colours for dogs carrying only the **normal allele** (N) at the K locus, paired with **a^t** at the A locus.

K locus	E locus	A locus a^t/ -	B locus		D locus	
N/N	E^m/E^m; E^m/E	Black with tan points Black mask	B/B; B/b	Black with tan points Black mask	D/D; D/d	Black with tan points Black mask
					d/d	Blue with tan points **Blue mask**
			b/b	Brown with tan points **Brown mask**	D/D; D/d	Brown with tan points **Brown mask**
					d/d	Isabella with tan points **Isabella mask**
	E/E; E/e	Black with tan points **No mask**	B/B; B/b	Black with tan points	D/D; D/d	Black with tan points
					d/d	Blue with tan points
			b/b	Brown with tan points	D/D; D/d	Brown with tan points
					d/d	Isabella with tan points
	e/e	**Tan coat** *Inhibited by recessive E locus*	B/B; B/b; b/b	**Tan** *Inhibited by recessive E locus*	D/D; D/d	Tan
					d/d	Champagne

Table 10: Interpretation of coat colours for dogs carrying only the **normal allele** (N) at the K locus, paired with **recessive black a/a** at the A locus.

K locus	E locus	A locus a/a	B locus		D locus	
N/N	E^m/E^m; E^m/E E/E; E/e	Black *	B/B; B/b	Black	D/D; D/d	Black
					d/d	Blue
			b/b	Brown	D/D; D/d	Brown
					d/d	Isabella
	e/e	**Black *** *Inhibited by recessive A locus – In some breeds, can cause a white coat*	B/B; B/b; b/b	**Black** *Inhibited by recessive A locus – In some breeds, can cause a white coat*	D/D; D/d	Black/ White in some breeds
					d/d	Blue/ White in some breeds

* Black mask present, but not visible.

Canine Coat Colouration

In addition to coat colour, several genes relating to coat type are also known. Three of the most commonly tested for include the genes for **long/fluffy hair** (L Locus), **curly hair** (Cu Locus), and **furnishings** (F Locus).

L Locus: Hair length (*FGF5* gene)

In canines, variation in hair length is predominantly determined by the L Locus, with two known alleles at 5 possible mutation regions (see Table 13). In each case, the dominant allele, **L**, produces short hair, while the recessive allele, **l**, results in long hair.

Rules for expression:

- Dogs must have 2 copies of the recessive allele, **l**, in order for the coat to be long.
- Dogs with one copy of the recessive allele at two or more of the mutation regions can have long hair, but the respective mutations must be present on different copies of the L locus, *i.e.* one must have been inherited from the mother, and the other from the father.

Table 13: Interpretation of each mutation region within the L locus. Dogs need 2 copies of the recessive allele from any of the mutation regions in order to produce a long coat.

Mutation tested	Variant possible at mutation region and interpretation			Breeds affected
Region 1: c.284G>T	G/G L/L Short coat	G/T L/l^1 Short coat	T/T l^1/ l^1 Long coat	Common to many
Region 2: c.578C>T	C/C L/L Short coat	C/T L/l^2 Short coat	T/T l^2/ l^2 Long coat	Akita, Samoyed, Eurasier, Siberian Husky
Region 3: c.556_571del16	GCGGGCCG GGAGTGGT/ GCGGGCCG GGAGTGGT L/L Short coat	GCGGGCCG GGAGTGGT/ - L/l^3 Short coat	- / - l^3/ l^3 Long coat	Eurasier
Region 4: c.559_560dupGG	- / - L/L Short coat	- / GG L/l^4 Short coat	GG/GG l^4/ l^4 Long coat	Afghan Hound, Eurasier, French Bulldog
Region 5: g.8193T>A	T/T L/L Short coat	T/A L/l^5 Short coat	A/A l^5/ l^5 Long coat	Afghan Hound

Canine Coat Colouration

Cu Locus: Curl (*KRT71* gene)

The presence or absence of a curly or wavy coat is associated with the Cu Locus. In canines, two alleles at two different mutation regions have been identified, which are more or less prevalent depending on the breed. In both cases, the dominant allele, **C**, results in hair that is either curly (2 copies) or wavy (1 copy), while the recessive allele, **N**, results in straight hair (see Table 14).

Rules for expression:

- The curly allele, C, has incomplete dominance, and will therefore be expressed differently depending on the number of copies present. One copy (**N/C**) will result in a wavy, or moderately curly coat, while two copies (**C/C**) will result in a more tightly curled coat.
- Dogs with one copy of the recessive allele at both mutation regions will have curly hair. *E.g.* A dog that has **N/C**^{1} at Region 1, and **N/C**^{2} at Region 2, will have curly hair.

Table 14: Interpretation of each mutation region within the Cu locus. Dogs need 1 copy of the dominant allele from any of the mutation regions in order to produce a wavy coat. Two copies produce a curly coat.

Mutation tested	Variant possible at mutation region and interpretation			Breeds affected
Region 1: c.451C>T	C/C **N/N** Straight coat	C/T **N/C**^{1} Wavy coat	T/T **C**^{1}**/ C**^{1} Curly coat	Common to many
Region 2: c.1266_1273delACCA	ACA/ACA **N/N** Straight coat	ACA/ - **N/C**^{2} Wavy coat	- / - **C**^{2}**/ C**^{2} Curly coat	Bichon Frise, Chesapeake Bay Retriever, Curly-Coated Retriever, Irish Terrier, Lagotto Romagnolo, Mudi, Poodle, Spanish Water Dog

F Locus: Furnishings/Improper coat (*RSPO2* gene)

The Furnishings trait, indicating the presence or absence of longer hair around the muzzle and eyebrows, has been associated with variants at the F locus in canines. In breeds for which furnishings is considered a breed standard (e.g. Portuguese Water Dogs and wire-haired breeds), the lack of furnishings is referred to as Improper Coat and is not favourable. The dominant allele, **F**, results in the presence of furnishings, while the recessive wild type allele, **IC**, results in the lack of furnishings, or an improper coat (see Table 15).

Rules for expression:

- The Furnishings allele, **F**, is dominant over the wild type/Improper Coat allele, **IC**. Therefore, only one copy of the dominant allele is needed to produce a coat with furnishings.

Table 15: Interpretation of each combination of variants at the F locus. Dogs need 2 copies of the recessive wild type allele in order to produce an improper coat, but only one copy of the dominant F allele to produce a coat with furnishings.

Mutation tested	Variant possible at mutation region and interpretation			Breeds affected
F/IC mutation: g. 8610419_8610420ins167	- / - IC/**IC** No furnishings / Improper coat	- / INS **IC/F** Furnishings present	INS / INS **F/ F** Furnishings present	Common to many

Why is it important to screen puppies for health issues?

There will always be negative mutations present in every population. It is an unavoidable part of life, as these mutations occur naturally during cell replication. In theory, when a cell replicates, all the information in that cell is copied over exactly into the next cell. However, as nobody is perfect, mistakes sometimes happen during the replication process. These mutations are either deletions, insertions, inversions or substitutions of the genetic code. Most of the time, these changes to the code will have little or no effect, but at other times, these changes will give rise to a physiological change within the body. Not all mutations are harmful however; as some actually help the animal adapt better to their environment, out-competing others within their population. But there are also mutations that give rise to harmful physiologies. Normally, when you have a very diverse population, the frequency of the occurrence of these harmful mutations will be kept to a minimum, as there is a massive gene pool to be selected from and not many animals will end up with this harmful mutation. But what happens when you start isolating certain groups within a population based on how they look (i.e. assigning dogs who look alike to a specific breed)?

Not allowing dogs from different breeds to interbreed dramatically reduced the amount of variation present within the gene pools of these established breeds. When you reduce the amount of variation possible within a group, you start increasing the frequency at which the already established mutations occur within the group. The more you breed within a population with limited variation, the more you establish those mutations within the group. This is why we see so many harmful mutations within specific breeds of dogs. Of course, these mutations are also still present within your mixed breed populations, but at a much, much lower frequency than the pure bred groups.

The only way to really combat the prevalence of these mutations within established breeds, is to try and breed with dogs who will not produce affected puppies. It must be noted, however, that only mating with clear dogs is also not advised within breeds that already suffer from low genetic diversity, as removing carriers from the population entirely will only further reduce the genetic diversity available in that group. The best approach is to pair carriers to clear dogs so that the diversity within the population is maintained without the risk of producing affected puppies. This is where genetic screening plays a vital role. If a breeder can know the genetic status of their dogs before a mating, they can actively start pairing up dogs who won't produce affected offspring. For most of these mutations, a puppy would require two copies of the mutated gene in order to be affected. The screenings are not only beneficial for breeders however, but for pet parents as well. Knowing which mutations your dog carries will help the owner make decisions regarding their dogs' health. Screenings are not only a valuable tool for preventing the following generations of pups from developing these diseases in the first place, but can also be a valuable tool for informed healthcare of family pets.

What are the typical tests done and how does it work?

There are currently over 300 known genetically inherited traits/disorders in dogs. Unistel includes the mutations most commonly found in a specific breed and only test for those mutations. Owners can ask to receive a home testing kit containing 2 buccal swabs. They will then need to swab the inside of their dog's cheek for about 30 seconds, let the swabs dry and then ship it for testing. The lab then extracts the DNA from these swabs and uses it for testing. Each test offered has been specifically designed to zero in on whichever region of the genome contains the mutation of interest, and to identify whether the individual possesses one, or two, or no mutated copies of the gene. Once this has been determined, the dog can be given a status of "clear", "carrier" or "affected" for each trait/condition.

Who can screen dogs for inherited diseases?

Breeders are encouraged to screen for genetic diseases within their breeding stock, however, if you as an owner have already purchased a puppy, there is nothing preventing you from getting the test done yourself.

Does one have to work through a veterinarian to get the test done?

Owners and breeders can work through a veterinary service, and many breeders do ask the vet to take blood samples for testing their puppies while going for their routine check-ups and inoculations; however, this is not essential. We offer a testing kit which can be used in the comfort of the client's home. In short, dog owners can use vets as their proxy, but you do not need a vet referral in order to have your dog tested.

What typically happens when a puppy tests positive for a serious disease?

This will all depend on the severity of the disease. Many of these diseases can be managed with either the correct diet, medication or adaptation of your dogs' routine. For dogs who are allergic to certain drugs, alternative drugs can be prescribed. For dogs who collapse after exercise, or too much excitement, a vet might prescribe a more subdued playing regimen. A positive result should never be equated with a death sentence for your dog. Even when your pup is affected, it is almost impossible to know how the mutation will manifest itself in the dog. The pup might develop severe symptoms and need to be euthanized to prevent suffering, but could also develop a milder, more manageable form of the disease. For breeders, it will mean removing the dog from their breeding pool and finding them an alternative home. For pet owners, the best advice would be to discuss the findings with a vet. Explore the options available for management of the symptoms and take the dog for regular check-ups to monitor the progression of the disease.

Health Screening offered at Unistel

Afghan Hound

Disorder	Normal	Mutation	Chr	Location	Gene
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Africanis dog

Disorder	Normal	Mutation	Chr	Location	Gene
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Airedale Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Factor VII Deficiency	G	A	CHR22	60578895	F7
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Factor VII Deficiency:
Mild to moderate inherited blood clotting disorder.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Alaskan Malamute

Disorder	Normal	Mutation	Chr	Location	Gene
Polyneuropathy	G	T	CHR13	29714606	NDRG1
Achromatopsia 3	CCA	del	CHR10	44234198	CNGB3
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Achromatopsia:
Canine day blindness or achromatopsia (ACHM) is a congenital eye disorder. The disorder can vary among affected dog by its severity, but common trait of all affected ones is the hemeralopa, or blindness in full sun. For this reason, it is also known as *day blindness* since the affected dog sees better in dim light.

Polyneuropathy:
Neurological disorder characterized by a dysfunction of multiple peripheral nerves. The etiology of the disease is diverse; it may occur in cases of infectious, immune-mediated, or hereditary conditions or in association with endocrinopathy, neoplasm, or chemical intoxication.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Akita

Disorder	Normal	Mutation	Chr	Location	Gene
Gangliosidosis GM1	C	del	CHR23	3796315	GLB1
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Gangliosidosis GM1:
Metabolic disorder in dogs. It is commonly known as storage disease because dogs that suffer from it lack an enzyme in their brain that helps with the breakdown of old molecules.

Hyperuricosuria:
Excess of uric acid in urine.

American Bulldog

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Canine Multifocal Retinopathy 1	C	T	CHR18	54478586	BEST1
Neuronal ceroid lipofuscinosis, 10	G	A	CHR18	46013354	CTSD
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9

Canine Multifocal Retinopathy:
Eye disease that results in a shortened, dysfunctional protein. Affected dogs typically present with multiple, discrete circular areas of retinal detachment between 11 and 16 weeks of age.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Neuronal ceroid lipofuscinosis:
Group of progressive degenerative diseases of the central nervous system. Signs of disease in affected dogs begin between one and two years of age and include behaviour issues such as: anxiety, constant circling, aggression, compulsive behaviours, and loss of learned skills.

American Bully

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
prcd-PRA	G	A	CHR9	4188663	PRCD
crd4-PRA	-	44bp ins	CHR15	18332036	RPGRIP1
Canine Multifocal Retinopathy 1	C	T	CHR18	54478586	BEST1
Cone rod dystrophy 1	GTT	del	CHR3	91747728	PDE68
Cone rod dystrophy 2	-	C	CHR33	25078909	IQCB1
Neuronal ceroid lipofuscinosis, 4	G	A	CHR9	15071276	ARSG
Neuronal ceroid lipofuscinosis, 10	G	A	CHR18	46013354	CTSD

Canine Multifocal Retinopathy:
Eye disease that results in a shortened, dysfunctional protein. Affected dogs typically present with multiple, discrete circular areas of retinal detachment between 11 and 16 weeks of age.

crd4PRA and Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Neuronal ceroid lipofuscinosis:
Group of progressive degenerative diseases of the central nervous system. Signs of disease in affected dogs begin between one and two years of age and include behaviour issues such as: anxiety, constant circling, aggression, compulsive behaviours, and loss of learned skills.

American Cocker Spaniel

Disorder	Normal	Mutation	Chr	Location	Gene
Exercise induced Collapse	G	T	CHR9	55282762	DNM1
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Familial nephropathy	A	T	CHR25	39953906	COL4A4
Glycogen Storage Disease (PFK Def)	G	A	CHR27	6620819	PFKM

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Exercise Induced Collapse:
Characterized by muscle weakness and lack of coordination. Affected dogs tolerate mild to moderate activity but will display signs of EIC after 5-20 minutes of strenuous exercise.

Familial nephropathy:
Familial nephropathy describes a disease in which young dogs suffer from early-onset kidney failure.

Glycogen Storage Disease:
Disorder is characterized by excessive accumulation of glycogen in the liver and other organs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

American English Coonhound

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

American Pitbull Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
Neuronal ceroid lipofuscinosis, 4	G	A	CHR9	15071276	ARSG
Cone rod dystrophy 1	GTT	del	CHR3	91747728	PDE68
Cone rod dystrophy 2	-	ins C	CHR33	25078909	IQCB1

Cone rod dystrophy:
Inherited eye disease affecting dogs. Affected dogs typically have abnormal thinning and degeneration of the retina by 11 weeks of age.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Neuronal ceroid lipofuscinosis:
Disorder is characterized by excessive accumulation of glycogen in the liver and other organs.

American Staffordshire terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
Neuronal ceroid lipofuscinosis, 4	G	A	CHR9	15071276	ARSG
Cone rod dystrophy 1	GTT	del	CHR3	91747728	PDE68
Cone rod dystrophy 2	-	C	CHR33	25078909	IQCB1
prcd-PRA	G	A	CHR9	4188663	PRCD

Cone rod dystrophy:
Inherited eye disease affecting dogs. Affected dogs typically have abnormal thinning and degeneration of the retina by 11 weeks of age.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Neuronal ceroid lipofuscinosis:
Disorder is characterized by excessive accumulation of glycogen in the liver and other organs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Anatolian Shepherd

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Australian Cattle dog

Disorder	Normal	Mutation	Chr	Location	Gene
Collie Eye Anomaly	-	7.8kb del	CHR37	25698028	NHEJ1
Cystinuria	ACCACC	del	CHR10	46725149	SLC3A1
Multiple Drug Resistance MDR1	AGAT	del	CHR14	13726596	ABCB1
Myotonia congenita	-	A	CHR16	6344730	CLCN1
Neuronal ceroid lipofuscinosis, 12	C	T	CHR2	81208162	ATP13A2
Neuronal ceroid lipofuscinosis, 5	C	T	CHR22	30574637	CLCN5
Primary lens luxation	G	A	CHR37	40782144	ADAMTS17
prcd-PRA	G	A	CHR9	4188663	PRCD

Collie eye anomaly:
Cause vision defects. The disease occurs in both eyes, but each eye may be affected differently.

Cystinuria:
Cystine bladder stones appear to be the result of a genetic abnormality that prevents a dog from reabsorbing cystine from the kidneys.

Multiple Drug Resistance:
A mutation of this gene causes sensitivity to Ivermectin and a number of other drugs. Dogs with the mutation will react to those drugs. Whether a dog reacts depends on the dosage of the drug.

Myotonia congenita:
Skeletal muscle disorder characterized by prolonged contraction or delayed relaxation of a muscle after voluntary or stimulated contraction.

Neuronal ceroid lipofuscinosis:
Group of progressive degenerative diseases of the central nervous system. Signs of disease in affected dogs begin between one and two years of age and include behaviour issues such as: anxiety, constant circling, aggression, compulsive behaviours, and loss of learned skills.

Primary lens luxation:
Results in a breakdown of the lens zonules – which usually occurs at a young age.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Australian Shepherd

Disorder	Normal	Mutation	Chr	Location	Gene
Collie Eye Anomaly	-	7.8kb del	CHR37	25698028	NHEJ1
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
Intestinal cobalamin malabsorption	C	del	CHR2	19974334	CUBN
Multiple Drug Resistance	AGAT	del	CHR14	13726596	ABCB1
Canine Multifocal Retinopathy	C	T	CHR18	54478586	BEST1
prcd-PRA	G	A	CHR9	4188663	PRCD
Factor VII Deficiency	G	A	CHR22	60578895	F7
Exercise induced Collapse	G	T	CHR9	55282762	DNM1
Neuronal ceroid lipofuscinosis, 6	T	C	CHR30	32247875	CLN6
Von Willebrandt Type 1	G	A	CHR27	38951839	VWF

Canine Multifocal Retinoapthy:
Eye disease that results in a shortened, dysfunctional protein. Affected dogs typically present with multiple, discrete circular areas of retinal detachment between 11 and 16 weeks of age.

Collie eye anomaly:
Cause vision defects. The disease occurs in both eyes, but each eye may be affected differently.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Exercise Induced Collapse:
Characterized by muscle weakness and lack of coordination. Affected dogs tolerate mild to moderate activity but will display signs of EIC after 5-20 minutes of strenuous exercise.

Factor VII Deficiency:
Mild to moderate inherited blood clotting disorder.

Hyperuricosuria:
Excess of uric acid in urine.

Intestinal cobalamin malabsorption:
Affected dogs are unable to make adequate amounts of a protein that plays a role in absorption of certain nutrients from the intestinal tract and kidneys, including the B vitamin, cobalamin. Affected dogs have increased levels of methylmalonic acid in their urine.

Multiple Drug Resistance:
A mutation of this gene causes sensitivity to Ivermectin and a number of other drugs. Dogs with the mutation will react to those drugs. Whether a dog reacts depends on the dosage of the drug.

Neuronal ceroid lipofuscinosis:
Group of progressive degenerative diseases of the central nervous system. Signs of disease in affected dogs begin between one and two years of age and include behaviour issues such as: anxiety, constant circling, aggression, compulsive behaviours, and loss of learned skills.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Von Willebrandt:
Blood clotting disorder.

Australian Silky Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Basenji

Disorder	Normal	Mutation	Chr	Location	Gene
Progressive retinal atrophy	T	C	CHR25	4483440	SAG
Spinocerebellar ataxia	C	G	CHR38	22140300	KCNJ10

Progressive retinal atrophy:
Affects the photoreceptor cells in the eye involved in both night and day vision.

Spinocerebellar ataxia:
Ataxia in dogs refers to a loss of coordination or unbalanced gait due to sensory dysfunction.

Basset Artesian Normand

Disorder	Normal	Mutation	Chr	Location	Gene
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Basset Hound

Disorder	Normal	Mutation	Chr	Location	Gene
Primary open angle glaucoma	C	T	CHR20	53096339	ADAMTS17
Thrombopathia	TCT	del	CHR18	52417313	RASGRP1
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Von Willebrandt Type 1	G	A	CHR27	38951839	VWF

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Primary Open Angle Glaucoma:
Open-angle glaucoma is a painless and gradual development of blind spots or loss of vision over a long period of time.

Thrombopathia:
Affected dogs have abnormal platelet function. Platelets are blood cells that are responsible for the normal clotting of blood.

Von Willebrandt:
Blood clotting disorder.

Bearded Collie

Disorder	Normal	Mutation	Chr	Location	Gene
Collie Eye Anomaly	-	7.8kb del	CHR37	25698028	NHEJ1
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Collie eye anomaly:
Cause vision defects. The disease occurs in both eyes, but each eye may be affected differently.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Beagle

Disorder	Normal	Mutation	Chr	Location	Gene
Hypocatalasia	C	T	CHR18	33397548	CAT
Factor VII Deficiency	G	A	CHR22	60578895	F7
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Musladin Leuke syndrome	C	T	CHR9	49931561	ADAMTSL2
Cerebellar abiotrophy	TCAAGGCA	del	CHR18	50666027	SPTBN2
Osteogenesis imperfecta	CTGA	del	CHR14	19918265	COL1A2
Primary open angle glaucoma	G	A	CHR20	53096339	ADAMTS10
Pyruvate kinase deficiency	G	A	CHR7	42268927	PKLR
Intestinal cobalamin malabsorption	C	del	CHR2	19796293	CUBN

Cerebellar abiotrophy:
The term is used to describe premature degeneration of fully formed cerebellar neurons caused by an intrinsic metabolic defect. Cerebellar abiotrophies typically involve a primary degeneration or loss of Purkinje neurons, variable loss of granule cells, and cortical astrogliosis.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Factor VII Deficiency:
Mild to moderate inherited blood clotting disorder.

Hypocatalasia:
Can suffer from mouth ulcers leading to difficulty eating and increased susceptibility to oral infections.

Intestinal cobalamin malabsorption:
Affected dogs are unable to make adequate amounts of a protein that plays a role in absorption of certain nutrients from the intestinal tract and kidneys, including the B vitamin, cobalamin. Affected dogs have increased levels of methylmalonic acid in their urine.

Musladin Leuke syndrome:
Affects the development and structure of connective tissue. It is multi-systemic, with involvement of multiple organs, including bone, heart, skin, and muscle. MLS is inherited as a recessive trait.

Osteogenesis imperfecta:
Disease which causes defective collagen, leading to extremely fragile bones and teeth. Affected puppies suffer from bone fractures after minor trauma which can result from playing.

Primary Open Angle Glaucoma:
Open-angle glaucoma is a painless and gradual development of blind spots or loss of vision over a long period of time.

Pyruvate kinase deficiency:
Haemolytic anaemia caused by a defect in the enzyme pyruvate kinase. Loss of function of this enzyme results in premature death of red blood cells. Affected dogs do not have sufficient quantities of red blood cells to adequately supply the body with oxygen.

Bedlington Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Belgian Shepherd

Disorder	Normal	Mutation	Chr	Location	Gene
Cerebellar Ataxia	T	C	CHR38	22140659	SOCA1
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
Mucopolysaccharidosis VII	G	A	CHR6	741429	GUSB
prcd-PRA	G	A	CHR9	4188663	PRCD

Cerebellar Ataxia:
Ataxia in dogs refers to a loss of coordination or unbalanced gait due to sensory dysfunction.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Mucopolysaccharidosis VII:
Deficiency of the lysosomal enzyme, alpha-L-iduronidase, causing physical deformities and organ dysfunction in dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Bernese Mountain

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Von Willebrandt Type 1	G	A	CHR27	38951839	VWF
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Von Willebrandt:
Blood clotting disorder.

Biro Yorkshire Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Craniomandibular osteopathy	C	T	CHR5	9387327	SLC37A2
Primary lens luxation	G	A	CHR37	40782144	ADAMTS17
prcd-PRA	G	A	CHR9	4188663	PRCD
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
L2 Hydroxyglutaric aciduria	A	G	CHR8	26760351	L2HGDH

Craniomandibular osteopathy:
Craniomandibular Osteopathy (CMO) is the result of bone swelling during the growth of the bones of the skull and jaw. Sometimes, only the jaw is involved. As such, young dogs between the ages of three and eight months are the most commonly afflicted.

Hyperuricosuria:
Excess of uric acid in urine.

L2-Hydroxyglutaric aciduria:
Autosomal recessive encephalopathy including neurological traits such as psychomotor impairment, seizures and ataxia. The onset of the disease occurs predominantly in dogs less than one year old, although it has been identified in older dogs.

Primary lens luxation:
Results in a breakdown of the lens zonules – which usually occurs at a young age..

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Biewer Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Craniomandibular osteopathy	C	T	CHR5	9387327	SLC37A2
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Primary lens luxation	G	A	CHR37	40782144	ADAMTS17
prcd-PRA	G	A	CHR9	4188663	PRCD
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
L2 Hydroxyglutaric aciduria	A	G	CHR8	26760351	L2HGDH

Craniomandibular osteopathy:
Craniomandibular Osteopathy (CMO) is the result of bone swelling during the growth of the bones of the skull and jaw. Sometimes, only the jaw is involved. As such, young dogs between the ages of three and eight months are the most commonly afflicted.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

L2-Hydroxyglutaric aciduria:
Autosomal recessive encephalopathy including neurological traits such as psychomotor impairment, seizures and ataxia. The onset of the disease occurs predominantly in dogs less than one year old, although it has been identified in older dogs.

Primary lens luxation:
Results in a breakdown of the lens zonules – which usually occurs at a young age.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

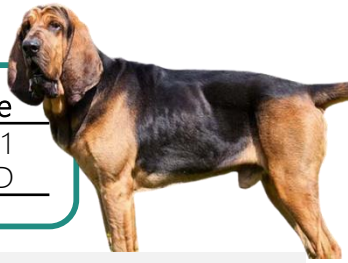
Bichon Frise

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Bloodhound



Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Boerboel

Disorder	Normal	Mutation	Chr	Location	Gene
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
Canine Multifocal Retinopathy 1	C	T	CHR18	54478586	BEST1
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Canine Multifocal Retinopathy:
Eye disease that results in a shortened, dysfunctional protein. Affected dogs typically present with multiple, discrete circular areas of retinal detachment between 11 and 16 weeks of age.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Border Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Border Collie

Disorder	Normal	Mutation	Chr	Location	Gene
Collie Eye Anomaly	-	7.8kb del	CHR37	25698028	NHEJ1
Dental hypomineralization	C	T	CHR6	16452327	FAM20C
Myotonia congenita	-	A	CHR16	6344730	CLCN1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
Intestinal cobalamin malabsorption	C	del	CHR2	19974334	CUBN
Multiple Drug Resistance	AGAT	del	CHR14	13726596	ABCB1
Neuronal ceroid lipofuscinosis, 5	C	T	CHR22	30574637	CLCN5
Trapped neutrophil syndrome	GTTT	del	CHR13	1412654	VPS13B
Exercise induced Collapse	G	T	CHR9	55282762	DNM1
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Collie eye anomaly:
Cause vision defects. The disease occurs in both eyes, but each eye may be affected differently.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Dental hypomineralization:
Affected dogs may develop brown discoloured teeth, smooth enamel, signs of wear, cracked teeth and inflammation.

Exercise Induced Collapse:
Characterized by muscle weakness and lack of coordination. Affected dogs tolerate mild to moderate activity but will display signs of EIC after 5-20 minutes of strenuous exercise.

Factor VII Deficiency:
Mild to moderate inherited blood clotting disorder.

Hyperuricosuria:
Excess of uric acid in urine.

Intestinal cobalamin malabsorption:
Affected dogs are unable to make adequate amounts of a protein that plays a role in absorption of certain nutrients from the intestinal tract and kidneys, including the B vitamin, cobalamin. Affected dogs have increased levels of methylmalonic acid in their urine.

Myotonia congenital:
Skeletal muscle disorder characterized by prolonged contraction or delayed relaxation of a muscle after voluntary or stimulated contraction.

Multiple Drug Resistance:
A mutation of this gene causes sensitivity to Ivermectin and a number of other drugs. Dogs with the mutation will react to those drugs. Whether a dog reacts depends on the dosage of the drug.

Neuronal ceroid lipofuscinosis:
Group of progressive degenerative diseases of the central nervous system. Signs of disease in affected dogs begin between one and two years of age and include behaviour issues such as: anxiety, constant circling, aggression, compulsive behaviours, and loss of learned skills.

Trapped neutrophil syndrome:
Inherited neutropenia that compromises the immune system, leading to chronic infection in affected dogs.

Borzoi

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Boston Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Bouvier Des Flandres

Disorder	Normal	Mutation	Chr	Location	Gene
Exercise induced Collapse	G	T	CHR9	55282762	DNM1
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Exercise Induced Collapse:
Characterized by muscle weakness and lack of coordination. Affected dogs tolerate mild to moderate activity but will display signs of EIC after 5-20 minutes of strenuous exercise.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Boxer

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Cardiomyopathy	CATACACA	del	CHR17	32373916	CFA17

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Dilated cardiomyopathy:
Disease of predominantly large and giant breed dogs that results in progressive heart muscle dysfunction, chamber dilation, and eventual congestive heart failure or death of affected patients.

Brittany

Disorder	Normal	Mutation	Chr	Location	Gene
C3 deficiency	C	del	CHR20	53573746	C3
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

C3 deficiency:
Occurs when there is a lack of the protein complement component C3. Generally, C3 is characterized by an increased vulnerability to renal disease and infection. Dogs that are affected with complement deficiency are also predisposed to the development of familial juvenile glomerulonephropathy and hereditary canine spinal muscular atrophy.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Bulldog

Disorder	Normal	Mutation	Chr	Location	Gene
Cystinuria Type 3	G	A	CHR1	119209134	SLC7A9
Canine Multifocal Retinopathy 1	C	T	CHR18	54478586	BEST1
crd4-PRA	-	44bp ins	CHR15	18332036	RPGRIP1

Canine multifocal retinopathy:
Eye disease that results in a shortened, dysfunctional protein. Affected dogs typically present with multiple, discrete circular areas of retinal detachment between 11 and 16 weeks of age.

Cystinuria:
Cystine bladder stones appear to be the result of a genetic abnormality that prevents a dog from reabsorbing cystine from the kidneys.

Crd4-PRA:
Inherited eye disease affecting dogs. PRA-crd4 occurs as a result of degeneration of both rod and cone type Photoreceptor Cells of the Retina, which are important for vision in dim and bright light, respectively.

Bullmastiff

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Canine Multifocal Retinopathy 1	C	T	CHR18	54478586	BEST1
prcd-PRA	G	A	CHR9	4188663	PRCD

Canine multifocal retinopathy:
Eye disease that results in a shortened, dysfunctional protein. Affected dogs typically present with multiple, discrete circular areas of retinal detachment between 11 and 16 weeks of age.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Bull Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Neuronal ceroid lipofuscinosis, 4	G	A	CHR9	15071276	ARSG
Polcystic kidney disease	G	A	CHR6	38856816	PKD1
Primary lens luxation	G	A	CHR37	40782144	ADAMTS17
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Exercise Induced Collapse:
Characterized by muscle weakness and lack of coordination. Affected dogs tolerate mild to moderate activity but will display signs of EIC after 5-20 minutes of strenuous exercise.

Neuronal ceroid lipofuscinosis:
Group of progressive degenerative diseases of the central nervous system. Signs of disease in affected dogs begin between one and two years of age and include behaviour issues such as: anxiety, constant circling, aggression, compulsive behaviours, and loss of learned skills.

Polycystic kidney disease:
Affected dogs begin to develop multiple small cysts within both kidneys early in life. As the dog ages, these cysts gradually begin to increase in both size and number.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Primary lens luxation:
Results in a breakdown of the lens zonules – which usually occurs at a young age.

Cairn Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Craniomandibular osteopathy	C	T	CHR5	9387327	SLC37A2
Krabbe disease	T	G	CHR8	59311801	GALC
Haemophilia B	G	A	CHRX	109532018	F9
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Craniomandibular Osteopathy:
Result of bone swelling during the growth of the bones of the skull and jaw. Sometimes, only the jaw is involved. As such, young dogs between the ages of three and eight months are the most commonly afflicted.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Haemophilia B:
Caused by a deficiency of coagulation Factor IX, causing a bleeding disorder.

Krabbe disease:
It results in abnormal processing and storage of an enzyme that is critical in the production of myelin, the substance that coats and protects the nerves throughout the brain and spinal cord.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Cane Corso

Disorder	Normal	Mutation	Chr	Location	Gene
Canine Multifocal Retinopathy 1	C	T	CHR18	54478586	BEST1
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Canine multifocal retinopathy:
Eye disease that results in a shortened, dysfunctional protein. Affected dogs typically present with multiple, discrete circular areas of retinal detachment between 11 and 16 weeks of age.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Cavalier King Charles Spaniel

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Episodic Falling syndrome	-	AAGGC	CHR7	41325010	BCAN
prcd-PRA	G	A	CHR9	4188663	PRCD
Muscular dystrophy, Duchenne	C	A	CHRX	26956239	DMD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Episodic falling syndrome:
Paroxysmal disorder. Episodes begin between fourteen weeks and four years of age and are triggered by exercise, stress, apprehension or excitement.

Muscular dystrophy, Duchenne type:
Stiff gait or a dog that "bunny hops" awkwardly with his back legs. These dogs also have decreased agility and exercise tolerance, compared to normal dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Chesapeake Bay Retriever

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Exercise induced Collapse	G	T	CHR9	55282762	DNM1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Exercise Induced Collapse:
Characterized by muscle weakness and lack of coordination. Affected dogs tolerate mild to moderate activity but will display signs of EIC after 5-20 minutes of strenuous exercise.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Chihuahua

Disorder	Normal	Mutation	Chr	Location	Gene
crd4-PRA	-	44bp ins	CHR15	18332036	RPGRIP1
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Crd4-PRA:
Inherited eye disease affecting dogs. PRA-crd4 occurs as a result of degeneration of both rod and cone type Photoreceptor Cells of the Retina, which are important for vision in dim and bright light, respectively.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Chinese Crested

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Primary lens luxation	G	A	CHR37	40782144	ADAMTS17
prcd-PRA	G	A	CHR9	4188663	PRCD
Ectodermal dysplasia	G	A	CHRX	54511433	FOXI3

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Ectodermal dysplasia:
Inherited disorder that has been reported in several breeds and mixed-breed dogs. Affected pups are born lacking hair on the forehead and back near the tail. Pups have a high frequency of eye infections prior to opening.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Primary lens luxation:
Results in a breakdown of the lens zonules – which usually occurs at a young age.

Chow chow

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Clumber Spaniel

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Pyruvate dehydrogenase	C	T	CHR29	38788096	PDP1
prcd-PRA	G	A	CHR9	4188663	PRCD
Exercise induced Collapse	G	T	CHR9	55282762	DNM1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Exercise Induced Collapse:
Characterized by muscle weakness and lack of coordination. Affected dogs tolerate mild to moderate activity but will display signs of EIC after 5-20 minutes of strenuous exercise.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Pyruvate dehydrogenase:
Metabolic disorder affecting dogs. Affected dogs may present as early as 15 weeks of age with an inability to play as long as littermates. Pyruvate is an important metabolic component needed for energy production in muscle.

Cocker Spaniel

Disorder	Normal	Mutation	Chr	Location	Gene
Exercise induced Collapse	G	T	CHR9	55282762	DNM1
Familial nephropathy	A	T	CHR25	39953906	COL4A4
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Exercise Induced Collapse:
Characterized by muscle weakness and lack of coordination. Affected dogs tolerate mild to moderate activity but will display signs of EIC after 5-20 minutes of strenuous exercise.

Familial nephropathy:
Disease in which young dogs suffer from early-onset kidney failure.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Collie (Rough)

Disorder	Normal	Mutation	Chr	Location	Gene
Collie Eye Anomaly	-	7.8kb del	CHR37	25698028	NHEJ1
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Multiple Drug Resistance	AGAT	del	CHR14	13726596	ABCB1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9

Collie eye anomaly:
Cause vision defects. The disease occurs in both eyes, but each eye may be affected differently.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Multiple Drug Resistance:
A mutation of this gene causes sensitivity to Ivermectin and a number of other drugs. Dogs with the mutation will react to those drugs. Whether a dog reacts depends on the dosage of the drug.

Corgi

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
rcd3 PRA	A	del	CHRX	59145361	PDE6A
Exercise induced Collapse	G	T	CHR9	55282762	DNM1
Severe combined immunodeficiency	-	C	CHRX	55483459	IL2RG
Von Willebrandt Type 1	G	A	CHR27	38951839	VWF
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Exercise Induced Collapse:
Characterized by muscle weakness and lack of coordination. Affected dogs tolerate mild to moderate activity but will display signs of EIC after 5-20 minutes of strenuous exercise.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Rcd3-PRA:
Rod cells of a young Corgi begin to die, often before the age of 1 year old. At 2-3 years old, the dog's cone cells also degenerate and die off. This leads to a loss of colour vision as well as well as vision in bright light. This ultimately results in complete blindness.

Severe combined immunodeficiency:
Mutations in the common gamma (gamma c) subunit of the IL-2, IL-4, IL-7, IL-9 and IL-15 receptors. The most striking clinical feature is a failure to thrive or 'stunted' growth.

Von Willebrandt:
Blood clotting disorder.

Coton De Tulear

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Canine Multifocal retinopathy 2	G	A	CHR18	54476143	BEST1
Von Willebrandt Type 1	G	A	CHR27	38951839	VWF
prcd-PRA	G	A	CHR9	4188663	PRCD

Canine multifocal retinopathy:
Eye disease that results in a shortened, dysfunctional protein. Affected dogs typically present with multiple, discrete circular areas of retinal detachment between 11 and 16 weeks of age.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Von Willebrandt:
Blood clotting disorder.

Curly-coated Retriever

Disorder	Normal	Mutation	Chr	Location	Gene
Exercise induced Collapse	G	T	CHR9	55282762	DNM1
prcd-PRA	G	A	CHR9	4188663	PRCD
crd4-PRA	-	44bp ins	CHR15	18332036	RPGRIP1
Glycogen storage disease	A	del	CHR6	50050451	AGL
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Crd4-PRA:
Inherited eye disease affecting dogs. PRA-crd4 occurs as a result of degeneration of both rod and cone type Photoreceptor Cells of the Retina, which are important for vision in dim and bright light, respectively.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Exercise Induced Collapse:
Characterized by muscle weakness and lack of coordination. Affected dogs tolerate mild to moderate activity but will display signs of EIC after 5-20 minutes of strenuous exercise.

Glycogen storage disease:
Disorder is characterized by excessive accumulation of glycogen in the liver and other organs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Dalmatian

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
prcd-PRA	G	A	CHR9	4188663	PRCD
Urolithiasis	G	T	CHR3	69456869	SLC2A9

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Urolithiasis:
Common condition responsible for lower urinary tract disease in dogs and cats. The formation of bladder stones (calculi) is associated with precipitation and crystal formation of a variety of minerals.

Dachshund

Disorder	Normal	Mutation	Chr	Location	Gene
Mucopolysaccharidosis IIIA	CCA	del	CHR9	1544373	SGSH
Narcolepsy	G	A	CHR12	22517939	HCRTR2
Neuronal ceroid lipofuscinosis, 1	-	C	CHR15	2883477	PPT1
Osteogenesis imperfecta	T	C	CHR21	23033735	SERPINH1
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Mucopolysaccharidosis:
Mucopolysaccharidoses are a rare group of genetic metabolic disorders that cause physical deformities and organ dysfunction in dogs.

Narcolepsy:
Disorder of the nervous system, affecting primarily young dogs and cats. A narcoleptic episode involves sudden collapse and loss of movement. The pet literally falls asleep, often while physically active, then wakes up abruptly and proceeds as if nothing happened.

Neuronal ceroid lipofuscinosis:
Group of progressive degenerative diseases of the central nervous system. Signs of disease in affected dogs begin between one and two years of age and include behaviour issues such as: anxiety, constant circling, aggression, compulsive behaviours, and loss of learned skills.

Osteogenesis imperfecta:
Disease which causes defective collagen, leading to extremely fragile bones and teeth. Affected puppies suffer from bone fractures after minor trauma which can result from playing.

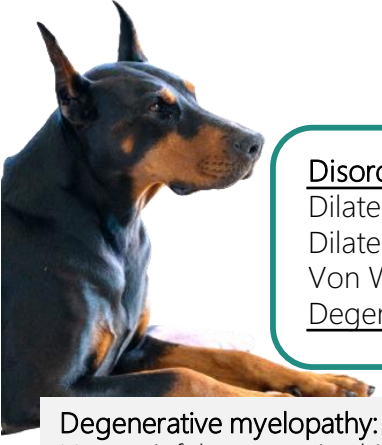
Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Dogo Argentino

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.



Dobermann

Disorder	Normal	Mutation	Chr	Location	Gene
Dilated cardiomyopathy	-	A	CHR15	22989894	PTPRQ
Dilated cardiomyopathy	-	16bp del	CHR14	20829667	PKD4
Von Willebrandt Type 1	G	A	CHR27	38951839	VWF
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Dilated cardiomyopathy:
Disease of predominantly large and giant breed dogs that results in progressive heart muscle dysfunction, chamber dilation, and eventual congestive heart failure or death of affected patients.

Von Willebrandt:
Blood clotting disorder.

Dogue de Bordeaux (French Mastiff)

Disorder	Normal	Mutation	Chr	Location	Gene
Canine Multifocal Retinopathy 1	C	T	CHR18	54478586	BEST1
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Canine multifocal retinopathy:
Eye disease that results in a shortened, dysfunctional protein. Affected dogs typically present with multiple, discrete circular areas of retinal detachment between 11 and 16 weeks of age.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

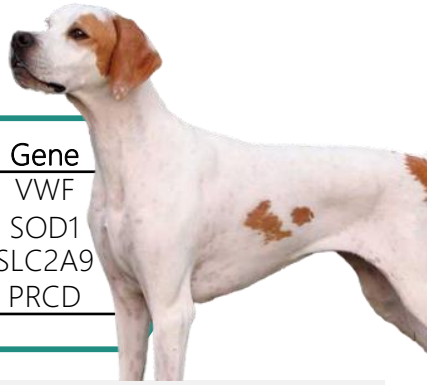
English Foxhound

Disorder	Normal	Mutation	Chr	Location	Gene
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

English Pointer



Disorder	Normal	Mutation	Chr	Location	Gene
Von Willebrandt Type 2	A	G	CHR27	38924099	VWF
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Von Willebrandt:
Blood clotting disorder.

English Setter

Disorder	Normal	Mutation	Chr	Location	Gene
Neuronal ceroid lipofuscinosis, 8	T	C	CHR37	30874779	CLN8
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Neuronal ceroid lipofuscinosis:
Group of progressive degenerative diseases of the central nervous system. Signs of disease in affected dogs begin between one and two years of age and include behaviour issues such as: anxiety, constant circling, aggression, compulsive behaviours, and loss of learned skills.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

English Springer Spaniel

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Familial nephropathy	A	T	CHR25	39953906	COL4A4
Glycogen Storage Disease	G	A	CHR27	6620819	PFKM
crd4-PRA	-	44bp ins	CHR15	18332036	RPGRIP1
prcd-PRA	G	A	CHR9	4188663	PRCD
Tremor, X-linked	A	C	CHRX	77200833	PLP1

Crd4-PRA:
Inherited eye disease affecting dogs. PRA-crd4 occurs as a result of degeneration of both rod and cone type Photoreceptor Cells of the Retina, which are important for vision in dim and bright light, respectively.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Dilated cardiomyopathy:
Disease of predominantly large and giant breed dogs that results in progressive heart muscle dysfunction, chamber dilation, and eventual congestive heart failure or death of affected patients.

Familial nephropathy:
Familial nephropathy describes a disease in which young dogs suffer from early-onset kidney failure.

Glycogen Storage Disease:
Disorder is characterized by excessive accumulation of glycogen in the liver and other organs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Tremor, X-linked:
Neurological disease that affects normal movement and proprioception. Hereditary transmission is via the X chromosome so males are the animals that are severely affected.

Eskimo Dog

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Primary lens luxation	G	A	CHR37	40782144	ADAMTS17
prcd-PRA	G	A	CHR9	4188663	PRCD
Thrombopathia	-	A	CHR18	52417256	RASGRP1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Primary lens luxation:
Results in a breakdown of the lens zonules – which usually occurs at a young age.

Thrombopathia:
Affected dogs have abnormal platelet function. Platelets are blood cells that are responsible for the normal clotting of blood.

Flat-coated Retriever

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Fox Terrier (Smooth and Wired)

Disorder	Normal	Mutation	Chr	Location	Gene
Congenital hypothyroidism	C	T	CHR17	784660	TPO
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Primary lens luxation	G	A	CHR37	40782144	ADAMTS17
Spinocerebellar ataxia	C	G	CHR38	22140300	KCNJ10
prcd-PRA	G	A	CHR9	4188663	PRCD

Congenital hypothyroidism:
Results in an underactive thyroid gland.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Primary lens luxation:
Results in a breakdown of the lens zonules – which usually occurs at a young age.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Spinocerebellar ataxia:
Characterized by progressive incoordination of gait, loss of balance, hypermetric and spastic movements. This condition may be associated with changes of brainstem auditory evoked potentials, myokymia, neuromyotonia and muscle fasciculation or seizures.

French Bulldog

Disorder	Normal	Mutation	Chr	Location	Gene
Brachycephaly	C	A	CHR32	5231894	SMOC2
Cystinuria Type 3	G	A	CHR1	119209134	SLC7A9
Canine Multifocal Retinopathy 1	C	T	CHR18	54478586	BEST1
crd4-PRA	-	44bp ins	CHR15	18332036	RPGRIP1
Congenital hypothyroidism	C	T	CHR17	784660	TPO
prcd-PRA	G	A	CHR9	4188663	PRCD
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Brachycephaly:
The term, brachycephalic, defines short-muzzled dog with a flattened face. It is the result of a genetic mutation which alters the way that the bones in their skulls grow. As a result, the shape of their skull is wide and short.

Canine multifocal retinopathy:
Eye disease that results in a shortened, dysfunctional protein. Affected dogs typically present with multiple, discrete circular areas of retinal detachment between 11 and 16 weeks of age.

Congenital hypothyroidism:
Results in an underactive thyroid gland.

Crd4-PRA:
Inherited eye disease affecting dogs. PRA-crd4 occurs as a result of degeneration of both rod and cone type Photoreceptor Cells of the Retina, which are important for vision in dim and bright light, respectively.

Cystinuria:
Cystine bladder stones appear to be the result of a genetic abnormality that prevents a dog from reabsorbing cystine from the kidneys.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

German Shepherd

Disorder	Normal	Mutation	Chr	Location	Gene
Scott syndrome	C	T	CHR27	8912219	ANO6
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Ichthyosis	A	G	CHR10	685587027	ASPRV1
Leukocyte adhesion Deficiency	-	12bp ins	CHR18	52835932	FERMT3
Renal cystadenocarcinoma	A	G	CHR5	42186445	FLCN
Cone degeneration	C	T	CHR10	44234861	CNGA3
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
Multiple Drug Resistance	AGAT	del	CHR14	13726596	ABCB1

Cone degeneration:
Inherited eye disease affecting dogs. Affected dogs develop day blindness (blindness in bright light) and Photophobia (light sensitivity) between 8 to 12 weeks after birth due to degeneration of cells in the eye called cone photoreceptors which are responsible for vision in bright light.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Ichthyosis:
The mutation prevents the outer layer of skin from developing properly. Affected skin is rough and covered with thick, greasy flakes that stick to the hair.

Leukocyte adhesion Deficiency:
Primary immunodeficiency disease characterized by recurrent bacterial infections in the presence of marked leukocytosis.

Multiple Drug Resistance:
A mutation of this gene causes sensitivity to Ivermectin and a number of other drugs. Dogs with the mutation will react to those drugs. Whether a dog reacts depends on the dosage of the drug.

Renal cystadenocarcinoma:
Inherited cancer. The disease has late onset and is progressive. Affected animals develop multiple firm nodules (dermatofibrosis) in the skin typically by 6 years of age.

Scott syndrome:
Defect in platelet function leading to impaired secondary hemostasis. Secondary hemostasis occurs after a platelet "plug" has formed. Its role is to make the plug stable by adding fibrin to the clot.

German Shorthaired Pointer

Disorder	Normal	Mutation	Chr	Location	Gene
Factor VII Deficiency	G	A	CHR22	60578895	F7
Cone degeneration	G	A	CHR22	32837065	CNGB3
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
Von Willebrandt Type 2	A	G	CHR27	38924099	VWF
prcd-PRA	G	A	CHR9	4188663	PRCD

Cone degeneration:
Inherited eye disease affecting dogs. Affected dogs develop day blindness (blindness in bright light) and Photophobia (light sensitivity) between 8 to 12 weeks after birth due to degeneration of cells in the eye called cone photoreceptors which are responsible for vision in bright light.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Factor VII Deficiency:
Mild to moderate inherited blood clotting disorder.

Hyperuricosuria:
Excess of uric acid in urine.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Von Willebrandt:
Blood clotting disorder.

German Wirehaired Pointer

Disorder	Normal	Mutation	Chr	Location	Gene
Factor VII Deficiency	G	A	CHR22	60578895	F7
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Exercise induced Collapse	G	T	CHR9	55282762	DNM1
Von Willebrandt Type 2	A	G	CHR27	38924099	VWF
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Exercise Induced Collapse:
Characterized by muscle weakness and lack of coordination. Affected dogs tolerate mild to moderate activity but will display signs of EIC after 5-20 minutes of strenuous exercise.

Factor VII Deficiency:
Mild to moderate inherited blood clotting disorder.

Hyperuricosuria:
Excess of uric acid in urine.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Von Willebrandt:
Blood clotting disorder.

Golden Retriever

Disorder	Normal	Mutation	Chr	Location	Gene
Ichthyosis	ACC	del	CHR12	5417388	PNPLA1
Golden Retriever PRA 2	A	del	CHR8	60090185	TTC8
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Exercise induced Collapse	G	T	CHR9	55282762	DNM1
Dystropic epidermolysis bullosa	G	A	CHR20	40538034	COL7A1
Muscular dystrophy	A	G	CHRX	27926946	DMD
Osteogenesis imperfecta	G	C	CHR9	26193593	COL1A1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Dystropic epidermolysis bullosa:
It is characterized by cleavage at the dermo-epidermal junction, leading to fragility of the skin and mucus membranes, blisters, and skin erosions.

Exercise Induced Collapse:
Characterized by muscle weakness and lack of coordination. Affected dogs tolerate mild to moderate activity but will display signs of EIC after 5-20 minutes of strenuous exercise.

Golden Retriever PRA 2:
Progressive retinal atrophy is characterized by bilateral degeneration of the retina resulting in progressive vision loss leading to total blindness. Clinical symptoms of PRA2 appear around 4 years of age.

Hyperuricosuria:
Excess of uric acid in urine.

Ichthyosis:
The mutation prevents the outer layer of skin from developing properly. Affected skin is rough and covered with thick, greasy flakes that stick to the hair.

Muscular dystrophy:
Stiff gait or a dog that "bunny hops" awkwardly with his back legs. These dogs also have decreased agility and exercise tolerance, compared to normal dogs.

Osteogenesis imperfecta:
Disease which causes defective collagen, leading to extremely fragile bones and teeth. Affected puppies suffer from bone fractures after minor trauma which can result from playing.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Gordon Setter

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
crd4-PRA	-	44bp ins	CHR15	18332036	RPGRIP1
Neuronal ceroid lipofuscinosis, 8	T	C	CHR37	30874779	CLN8

Crd4-PRA:
Inherited eye disease affecting dogs. PRA-crd4 occurs as a result of degeneration of both rod and cone type Photoreceptor Cells of the Retina, which are important for vision in dim and bright light, respectively.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Neuronal ceroid lipofuscinosis:
Group of progressive degenerative diseases of the central nervous system. Signs of disease in affected dogs begin between one and two years of age and include behaviour issues such as: anxiety, constant circling, aggression, compulsive behaviours, and loss of learned skills.

Great Dane

Disorder	Normal	Mutation	Chr	Location	Gene
Ichthyosis	G	A	CHR9	55168916	SLC27A4
Myopathy	A	G	CHR19	23522400	BIN1
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Ichthyosis:
The mutation prevents the outer layer of skin from developing properly. Affected skin is rough and covered with thick, greasy flakes that stick to the hair.

Myopathy:
Inherited myopathy is a rapidly progressive muscle myopathy characterized by exercise intolerance and progressive muscle atrophy.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Greyhound

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Polyneuropathy	-	10bp del	CHR13	29691070	NDRG1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Polyneuropathy:
Neurological disorder characterized by a dysfunction of multiple peripheral nerves. The etiology of the disease is diverse; it may occur in cases of infectious, immune-mediated, or hereditary conditions or in association with endocrinopathy, neoplasm, or chemical intoxication.

Great Swiss mountain dog

Disorder	Normal	Mutation	Chr	Location	Gene
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Bleeding disorder	GAG	del	CHR23	45909987	P2RY12

Bleeding disorder:
Platelet disorder. This disorder is particularly troublesome because spontaneous haemorrhage is absent to mild in affected dogs; however, following routine surgical procedures or trauma, excessive bleeding could occur and have possible fatal consequences.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Havanese

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Canine Multifocal Retinopathy 1	C	T	CHR18	54478586	BEST1
prcd-PRA	G	A	CHR9	4188663	PRCD

Canine Multifocal Retinopathy:
Eye disease that results in a shortened, dysfunctional protein. Affected dogs typically present with multiple, discrete circular areas of retinal detachment between 11 and 16 weeks of age.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Hungarian Vizsla

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD
Exercise induced Collapse	G	T	CHR9	55282762	DNM1
Cerebellar degeneration	C	T	CHR12	45530566	SNX14

Cerebellar degeneration:
Inherited condition can cause the neurons in the cerebellum to degenerate and die at a young age. This is called cortical cerebellar abiotrophy or often just cerebellar abiotrophy (CA).

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Exercise Induced Collapse:
Characterized by muscle weakness and lack of coordination. Affected dogs tolerate mild to moderate activity but will display signs of EIC after 5-20 minutes of strenuous exercise.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Ibizan Hound

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Irish Wolfhound

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Irish Setter

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Leukocyte adhesion deficiency	G	C	CHR31	38537012	ITGB2
rcd1 PRA rod cone dysplasia	G	A	CHR3	91747714	PDE68
crd4-PRA	-	44bp ins	CHR15	18332036	RPGRIP1
Von Willebrandt Type 1	G	A	CHR27	38951839	VWF

Crd4-PRA:
Inherited eye disease affecting dogs. PRA-crd4 occurs as a result of degeneration of both rod and cone type Photoreceptor Cells of the Retina, which are important for vision in dim and bright light, respectively.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Leukocyte adhesion Deficiency:
Primary immunodeficiency disease characterized by recurrent bacterial infections in the presence of marked leukocytosis.

rcd1 PRA rod cone dysplasia:
Progressive retinal atrophy (PRA) is a late onset inherited rod-cone dysplasia (type 1-rcd1. Dogs with this disease typically start showing signs of visual impairment between 2-3 years of age.

Von Willebrandt:
Blood clotting disorder.

Irish Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD
Hyperkeratosis, palmoplantar	G	C	CHR5	41055619	FAM83G

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperkeratosis, palmoplantar:
skin condition in which excess keratin is produced, in particular in the nose and/or paw pads; causing skin thickening and hardening, sometimes to the point of cracking, thus leading to the emergence of secondary infections.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Irish Water spaniel

Disorder	Normal	Mutation	Chr	Location	Gene
Factor VII Deficiency	G	A	CHR22	60578895	F7
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Factor VII Deficiency:
Mild to moderate inherited blood clotting disorder.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Italian Greyhound

Disorder	Normal	Mutation	Chr	Location	Gene
Amelogenesis imperfecta	TTTCC	del	CHR13	59946494	ENAM
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
Primary lens luxation	G	A	CHR37	40782144	ADAMTS17
prcd-PRA	G	A	CHR9	4188663	PRCD

Amelogenesis imperfecta:
Amelogenesis imperfecta is an inherited disease of enamel formation. Enamel formation starts before the eruption of the first teeth and involves many different genes. Defects in any one of these genes can result in defects in the formation of baby and permanent teeth.

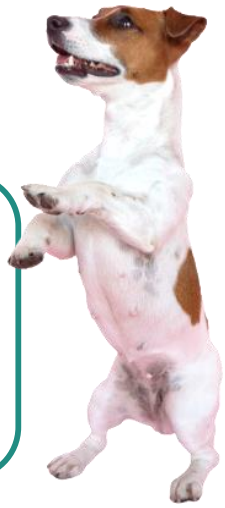
Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Primary lens luxation:
Results in a breakdown of the lens zonules – which usually occurs at a young age.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Jack Russel Terrier



Disorder	Normal	Mutation	Chr	Location	Gene
Craniomandibular osteopathy	C	T	CHR5	9387327	SLC37A2
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
Primary lens luxation	G	A	CHR37	40782144	ADAMTS17
Severe Immunodeficiency	G	T	CHR29	49588	PRKDC
prcd-PRA	G	A	CHR9	4188663	PRCD

Craniomandibular osteopathy:
Result of bone swelling during the growth of the bones of the skull and jaw. Sometimes, only the jaw is involved. As such, young dogs between the ages of three and eight months are the most commonly afflicted.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Primary lens luxation:
Results in a breakdown of the lens zonules – which usually occurs at a young age.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Severe combined immunodeficiency:
Mutations in the common gamma (gamma c) subunit of the IL-2, IL-4, IL-7, IL-9 and IL-15 receptors. The most striking clinical feature is a failure to thrive or 'stunted' growth.

Japanese Chin

Disorder	Normal	Mutation	Chr	Location	Gene
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Japanese Spitz

Disorder	Normal	Mutation	Chr	Location	Gene
Factor VII Deficiency	G	A	CHR22	60578895	F7
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Factor VII Deficiency:
Mild to moderate inherited blood clotting disorder.

Kerry Blue Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Von Willebrandt Type 1	G	A	CHR27	38951839	VWF
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Von Willebrandt:
Blood clotting disorder.

King Charles Spaniel

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Exercise induced Collapse	G	T	CHR9	55282762	DNM1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Exercise Induced Collapse:
Characterized by muscle weakness and lack of coordination. Affected dogs tolerate mild to moderate activity but will display signs of EIC after 5-20 minutes of strenuous exercise.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Kooikerhondje

Disorder	Normal	Mutation	Chr	Location	Gene
Von Willebrandt Type III	G	A	CHR27	38892182	VWF
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Von Willebrandt:
Blood clotting disorder.

Leonberger

Disorder	Normal	Mutation	Chr	Location	Gene
Polyneuropathy	-	10bp del	CHR16	54349199	ARHGEF10
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Polyneuropathy:
Neurological disorder characterized by a dysfunction of multiple peripheral nerves. The etiology of the disease is diverse; it may occur in cases of infectious, immune-mediated, or hereditary conditions or in association with endocrinopathy, neoplasm, or chemical intoxication.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Labrador Retriever

Disorder	Normal	Mutation	Chr	Location	Gene
Myotubular myopathy 1	C	A	CHRX	11885117	MTM1
Narcolepsy	G	A	CHR12	22620881	ADAMTSL2
Exercise induced Collapse	G	T	CHR9	55282762	DNM1
Hereditary nasal parakeratosis	T	G	CHR2	21731842	SUV39H2
Macular Corneal dystrophy	C	A	CHR5	75279762	CHST6
prcd-PRA	G	A	CHR9	4188663	PRCD
Skeletal dysplasia 2	G	C	CHR12	2652874	COLIIA2
Cystinuria	G	del	CHR10	46700948	SLC3A1
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
Ichthyosis	ACC	del	CHR12	5417388	PNPLA1
Pyruvate kinase deficiency	C	T	CHR7	42268632	PKLR
Wilson disease	G	A	CHR22	225112	ATP7B
Menkes disease	C	T	CHRX	60279237	ATP7A

Cystinuria:
Cystine bladder stones appear to be the result of a genetic abnormality that prevents a dog from reabsorbing cystine from the kidneys.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Exercise Induced Collapse:
Characterized by muscle weakness and lack of coordination. Affected dogs tolerate mild to moderate activity but will display signs of EIC after 5-20 minutes of strenuous exercise.

Hereditary nasal parakeratosis:
Genetic defect caused by a mutation in a gene that regulates differentiation of nose skin cells. The mutation affects specialized cells of the nose resulting in the formation of a crust with cracks over the nasal area of young dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Ichthyosis:
The mutation prevents the outer layer of skin from developing properly. Affected skin is rough and covered with thick, greasy flakes that stick to the hair.

Macular Corneal dystrophy:
Progressive eye disease. Affected dogs frequently present around 4 to 6 years of age with clouding of their corneas accompanied by pinpoint white to gray spots made up of an accumulation of a carbohydrate known as glycosaminoglycan.

Menkes disease:
Fatal neurodegenerative disorder of copper deficiency.

Myotubular myopathy 1:
X-linked myotubular myopathy in Labrador Retrievers is an inherited muscle disease that manifests with generalized muscle weakness and progressive muscle atrophy in puppies.

Narcolepsy:
Disorder of the nervous system, affecting primarily young dogs and cats. A narcoleptic episode involves sudden collapse and loss of movement. The pet literally falls asleep, often while physically active, then wakes up abruptly and proceeds as if nothing happened.

Labrador Retriever (continued)

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Pyruvate kinase deficiency:
Haemolytic anaemia caused by a defect in the enzyme pyruvate kinase. Loss of function of this enzyme results in premature death of red blood cells. Affected dogs do not have sufficient quantities of red blood cells to adequately supply the body with oxygen.

Skeletal dysplasia 2:
Musculoskeletal disorder that causes a form of mild disproportionate dwarfism in affected dogs – their body length and width are normal, but their legs are shorter than normal.

Wilson disease:
Biliary excretion is prevented by lack of an active form of ATP7B, which permits copper entry into bile. This leads to copper accumulation and severe liver toxicosis. Dogs tend to have the same problem, with much higher liver copper levels than other mammals.

Maltese

Disorder	Normal	Mutation	Chr	Location	Gene
Glycogen storage disease Ia	G	C	CHR9	20138777	G6PC
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Glycogen Storage Disease:
Disorder is characterized by excessive accumulation of glycogen in the liver and other organs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Manchester Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Von Willebrandt Type 1	G	A	CHR27	38951839	VWF
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Von Willebrandt:
Blood clotting disorder.

Mastino Napoletano (Neapolitan Mastiff)

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD
Canine Multifocal Retinopathy 1	C	T	CHR18	54478586	BEST1

Canine Multifocal Retinopathy:
Eye disease that results in a shortened, dysfunctional protein. Affected dogs typically present with multiple, discrete circular areas of retinal detachment between 11 and 16 weeks of age.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Miniature Pinscher

Disorder	Normal	Mutation	Chr	Location	Gene
Cystinuria Type 3	G	A	CHR1	119209134	SLC7A9
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Cystinuria:
Cystine bladder stones appear to be the result of a genetic abnormality that prevents a dog from reabsorbing cystine from the kidneys.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

New Foundland

Disorder	Normal	Mutation	Chr	Location	Gene
Cystinuria	C	T	CHR10	46706001	SCL3A1
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Cystinuria:
Cystine bladder stones appear to be the result of a genetic abnormality that prevents a dog from reabsorbing cystine from the kidneys.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Norfolk Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Epidermolytic hyperkerotosis	G	T	CHR9	21866234	KRT10
Primary lens luxation	G	A	CHR37	40782144	ADAMTS17
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Epidermolytic hyperkerotosis:
It is characterized by cleavage at the dermo-epidermal junction, leading to fragility of the skin and mucus membranes, blisters, and skin erosions.

Primary lens luxation:
Results in a breakdown of the lens zonules – which usually occurs at a young age.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Old English Sheepdog

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Exercise induced Collapse	G	T	CHR9	55282762	DNM1
Multiple Drug Resistance MDR1	AGAT	del	CHR14	13726596	ABCB1
Primary cilliary dyskinesia	C	T	CHR34	13952270	CCDC39
Cerebellar degeneration	A	C	CHR4	36055678	RAB24
prcd-PRA	G	A	CHR9	4188663	PRCD

Cerebellar degeneration:
Inherited condition can cause the neurons in the cerebellum to degenerate and die at a young age. This is called cortical cerebellar abiotrophy or often just cerebellar abiotrophy (CA).

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Exercise Induced Collapse:
Characterized by muscle weakness and lack of coordination. Affected dogs tolerate mild to moderate activity but will display signs of EIC after 5-20 minutes of strenuous exercise.

Multiple Drug Resistance:
A mutation of this gene causes sensitivity to Ivermectin and a number of other drugs. Dogs with the mutation will react to those drugs. Whether a dog reacts depends on the dosage of the drug.

Primary ciliary dyskinesia:
Congenital defect where the ciliary throughout the body do not function properly. This can cause respiratory issues such as coughing, nasal congestion and exercise intolerance. In older male dogs, it can lead to infertility.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Papillon

Disorder	Normal	Mutation	Chr	Location	Gene
Factor VII Deficiency	G	A	CHR22	60578895	F7
PRA	A	del	CHR2	5862673	CNGB1
PRA	-	AGCTAC	CHR2	58622675	CNGB1
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Von Willebrandt Type 1	G	A	CHR27	38951839	VWF

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Factor VII Deficiency:
Mild to moderate inherited blood clotting disorder.

PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Von Willebrandt:
Blood clotting disorder.

Pekingese

Disorder	Normal	Mutation	Chr	Location	Gene
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Pharoah Hound

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Parson Russell Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Craniomandibular osteopathy	C	T	CHR5	9387327	SLC37A2
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
Late onset ataxia	G	A	CHR18	52009339	CAPN1
Primary lens luxation	G	A	CHR37	40782144	ADAMTS17
Severe Immunodeficiency	G	T	CHR29	49588	PRKDC
Spinocerebellar ataxia	C	G	CHR38	22140300	KCNJ10

Craniomandibular osteopathy:
Craniomandibular Osteopathy (CMO) is the result of bone swelling during the growth of the bones of the skull and jaw. Sometimes, only the jaw is involved. As such, young dogs between the ages of three and eight months are the most commonly afflicted.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Late onset ataxia:
Characterised by worsening incoordination and loss of balance, which can make everyday tasks and moving difficult. There is no treatment for this condition and affected dogs are often put to sleep around two years after onset.

Primary lens luxation:
Results in a breakdown of the lens zonules – which usually occurs at a young age.

Severe combined immunodeficiency:
Mutations in the common gamma (gamma c) subunit of the IL-2, IL-4, IL-7, IL-9 and IL-15 receptors. The most striking clinical feature is a failure to thrive or 'stunted' growth.

Spinocerebellar ataxia:
Ataxia in dogs refers to a loss of coordination or unbalanced gait due to sensory dysfunction.

Polish Lowland Sheepdog

Disorder	Normal	Mutation	Chr	Location	Gene
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Pomeranian

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Von Willebrandt Type 1	G	A	CHR27	38951839	VWF
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
rcd3 PRA	A	del	CHRX	59145361	PDE6A
Vitamin D Deficiency	G	del	CHR27	6895069	VDR

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

Rcd3-PRA:
In this form of PRA, the rod cells of a young dog begin to die, often before the age of 1 year old. The rod cells are responsible for vision in low-light or night settings and the dog quickly becomes night-blind.

Vitamin D deficiency:
Malabsorption of vitamin D, leading to bridle or malformed bones.

Von Willebrandt:
Blood clotting disorder.

Portuguese Water Dog

Disorder	Normal	Mutation	Chr	Location	Gene
Gangliosidosis GM1	G	A	CHR23	3754313	GLB1
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Gangliosidosis GM1:
Metabolic disorder in dogs. It is commonly known as storage disease because dogs that suffer from it lack an enzyme in their brain that helps with the breakdown of old molecules.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Poodle

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Neonatal encephalopathy w/ seizures	T	G	CHR36	19078954	ATF2
prcd-PRA	G	A	CHR9	4188663	PRCD
Von Willebrandt Type 1	G	A	CHR27	38951839	VWF
Gangliosidosis GM2	G	del	CHR2	57225684	HEXB
Multiple Drug Resistance	AGAT	del	CHR14	13726596	ABCB1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Gangliosidosis GM2:
GM2-gangliosidosis is a fatal neurodegenerative lysosomal storage disease (LSD) caused by deficiency of either β-hexosaminidase A (Hex-A) and β-hexosaminidase B (Hex-B) together, or the GM2 activator protein. Clinical signs can be variable and are not pathognomonic for the specific, causal deficiency.

Hyperuricosuria:
Excess of uric acid in urine.

Late onset ataxia:
Characterised by worsening incoordination and loss of balance, which can make everyday tasks and moving difficult. There is no treatment for this condition and affected dogs are often put to sleep around two years after onset.

Multiple Drug Resistance:
A mutation of this gene causes sensitivity to Ivermectin and a number of other drugs. Dogs with the mutation will react to those drugs. Whether a dog reacts depends on the dosage of the drug.

Neonatal encephalopathy w/ seizures:
Affected puppies are small and weak at birth. Cerebella from affected puppies have been found to be reduced in size. Many die in their first week of life.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Von Willebrandt:
Blood clotting disorder.

Pug

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Pyruvate kinase deficiency of erythrocyte	T	C	CHR7	42268681	PKLR

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Pyruvate kinase deficiency of erythrocyte:
Haemolytic anaemia caused by a defect in the enzyme pyruvate kinase. Loss of function of this enzyme results in premature death of red blood cells. Affected dogs do not have sufficient quantities of red blood cells to adequately supply the body with oxygen.

Puli

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Primary lens luxation	G	A	CHR37	40782144	ADAMTS17
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Primary lens luxation:
Results in a breakdown of the lens zonules – which usually occurs at a young age.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Pyrenean Mountain Dog

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Canine Multifocal Retinopathy 1	C	T	CHR18	54478586	BEST1
prcd-PRA	G	A	CHR9	4188663	PRCD
Thrombasthenia	-	14bp dup	CHR9	19057144	ITGA2B

Canine Multifocal Retinopathy:
Eye disease that results in a shortened, dysfunctional protein. Affected dogs typically present with multiple, discrete circular areas of retinal detachment between 11 and 16 weeks of age.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Thrombasthenia:
A disease of defective platelet aggregation, this causes affected dogs to bleed excessively in response to mild tissue trauma. Platelets are the first responders to tissue trauma: first, they clump together ("aggregate") by virtue of specific molecules in their membranes.

Pyrenean Sheepdog

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Red and white setter

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
rcd1 PRA rod cone dysplasia	G	A	CHR3	91747714	PDE68
Von Willebrandt Type 1	G	A	CHR27	38951839	VWF
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

rcd1 PRA rod cone dysplasia:
Progressive retinal atrophy (PRA) is a late onset inherited rod-cone dysplasia (type 1-rcd1. Dogs with this disease typically start showing signs of visual impairment between 2-3 years of age.

Von Willebrandt:
Blood clotting disorder.

Rhodesian Ridgeback

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Haemophilia B	G	A	CHRX	109532018	F9
Juvenile myoclonic epilepsy	-	del	CHR20	56474668	DIRAS1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Haemophilia B:
Caused by a deficiency of coagulation Factor IX, causing a bleeding disorder.

Juvenile myoclonic epilepsy:
Form of epilepsy with sudden, electroshock-like muscle twitches and seizures. These often occur during the initial sleep phase or can also be triggered by sudden bright light. Most dogs have daily seizures.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Rottweiler

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Von Willebrandt Type 1	G	A	CHR27	38951839	VWF
Cystinuria	ACCACC	del	CHR10	46725149	SLC3A1

Cystinuria:
Cystine bladder stones appear to be the result of a genetic abnormality that prevents a dog from reabsorbing cystine from the kidneys.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Von Willebrandt:
Blood clotting disorder.

Saint Bernard

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Leukocyte adhesion deficiency	G	C	CHR31	38537012	ITGB2
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Leukocyte adhesion Deficiency:
Primary immunodeficiency disease characterized by recurrent bacterial infections in the presence of marked leukocytosis.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Saluki

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD
Neuronal ceroid lipofuscinosis, 8	T	C	CHR37	30874779	CLN8

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Neuronal ceroid lipofuscinosis:
Group of progressive degenerative diseases of the central nervous system. Signs of disease in affected dogs begin between one and two years of age and include behaviour issues such as: anxiety, constant circling, aggression, compulsive behaviours, and loss of learned skills.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Samoyed

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Hereditary nephritis	G	T	CHRX	82196868	COL4A5
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hereditary nephritis:
Disease that is inherited from a carrier female. Males are more likely to be affected by it because they only have one X chromosome. Male puppies will begin showing symptoms of the kidney condition at an early age and will generally die from kidney failure by 15 months of age.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Schipperke

Disorder	Normal	Mutation	Chr	Location	Gene
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Schnauzer

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Myotonia congenita	C	T	CHR16	6366383	CLCN1
prcd-PRA	G	A	CHR9	4188663	PRCD
Persistent Mullerian duct syndrome	C	T	CHR27	1794738	AMHR2
Factor VII Deficiency	G	A	CHR22	60578895	F7

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Factor VII Deficiency:
Mild to moderate inherited blood clotting disorder.

Myotonia congenita:
Skeletal muscle disorder characterized by prolonged contraction or delayed relaxation of a muscle after voluntary or stimulated contraction.

Persistent Mullerian duct syndrome:
Form of male pseudohermaphroditism in dogs. It is an abnormal sexual phenotype in males that is characterized by the existence of a hypoplastic oviduct, uterus, and cranial part of the vagina. Dogs suffering from PMDS are often accompanied by cryptorchidism.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Scottish Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Craniomandibular osteopathy	C	T	CHR5	9387327	SLC37A2
Von Willebrandt Type 3	C	del	CHR27	38848107	VWF
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Craniomandibular osteopathy:
Craniomandibular Osteopathy (CMO) is the result of bone swelling during the growth of the bones of the skull and jaw. Sometimes, only the jaw is involved. As such, young dogs between the ages of three and eight months are the most commonly afflicted.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Von Willebrandt:
Blood clotting disorder.

Sealyham Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Primary lens luxation	G	A	CHR37	40782144	ADAMTS17
Factor VII Deficiency	G	A	CHR22	60578895	F7
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Factor VII Deficiency:
Mild to moderate inherited blood clotting disorder.

Primary lens luxation:
Results in a breakdown of the lens zonules – which usually occurs at a young age.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Shetland Sheepdog

Disorder	Normal	Mutation	Chr	Location	Gene
Collie Eye Anomaly	-	7.8kb del	CHR37	25698028	NHEJ1
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Multiple Drug Resistance	AGAT	del	CHR14	13726596	ABCB1
Progressive retinal atrophy	AGTT	del	CHR13	43831897	CNGA1
Von Willebrandt Type III	G	A	CHR27	38892182	VWF
Gallbladder mucoceles	-	C	CHR14	13584929	ABCB4

Collie eye anomaly:
Cause vision defects. The disease occurs in both eyes, but each eye may be affected differently.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Gallbladder mucoceles:
Characterized by progressive accumulation of tenacious, pale yellow to dark green, mucin-laden bile, which may extend into the cystic, hepatic, and common bile ducts, resulting in variable degrees of bile duct obstruction.

Multiple Drug Resistance:
A mutation of this gene causes sensitivity to Ivermectin and a number of other drugs. Dogs with the mutation will react to those drugs. Whether a dog reacts depends on the dosage of the drug.

Progressive retinal atrophy:
Affects the photoreceptor cells in the eye involved in both night and day vision.

Von Willebrandt:
Blood clotting disorder.

Shiba Inu

Disorder	Normal	Mutation	Chr	Location	Gene
Gangliosidosis GM1	C	del	CHR23	3796315	GLB1
Gangliosidosis GM2	G	del	CHR2	57225684	HEXB
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Gangliosidosis:
Metabolic disorder in dogs. It is commonly known as storage disease because dogs that suffer from it lack an enzyme in their brain that helps with the breakdown of old molecules.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Shih Tzu

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Prekallikrein Deficiency	T	A	CHR16	44501415	KLKB1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prekallikrein Deficiency:
This is a benign blood disorder characterized by low levels of prekallikrein; affected dogs typically suffer no ill effects. Prekallikrein is an enzyme necessary for activation of other clotting proteins.Progressive retinal atrophy.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Shorty Bull

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
L2 Hydroxyglutaric aciduria	T	C	CHR8	26723470	L2HGDH
Canine Multifocal Retinopathy	C	T	CHR18	54478586	BEST1
crd4-PRA	-	44bp ins	CHR15	18332036	RPGRIP1

Canine Multifocal Retinopathy:
Eye disease that results in a shortened, dysfunctional protein. Affected dogs typically present with multiple, discrete circular areas of retinal detachment between 11 and 16 weeks of age.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

L2 Hydroxyglutaric aciduria:
L-2-hydroxyglutaric aciduria is an autosomal recessive encephalopathy including neurological traits such as psychomotor impairment, seizures and ataxia. The onset of the disease occurs predominantly in dogs less than one year old, although it has been identified in older dogs.

Crd4-PRA:
Inherited eye disease affecting dogs. PRA-crd4 occurs as a result of degeneration of both rod and cone type Photoreceptor Cells of the Retina, which are important for vision in dim and bright light, respectively.

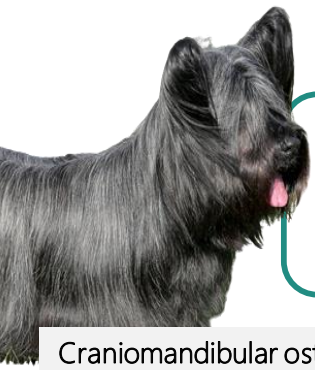
Siberian Husky

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD
Gangliosidosis GM1	-	dup	CHR23	3796356	GLB1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.
with the mutation will react to those drugs. Whether a dog reacts depends on the dosage of the drug.

Gangliosidosis:
Metabolic disorder in dogs. It is commonly known as storage disease because dogs that suffer from it lack an enzyme in their brain that helps with the breakdown of old molecules.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.



Skye Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Craniomandibular osteopathy	C	T	CHR5	9387327	SLC37A2
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Craniomandibular osteopathy:
Craniomandibular Osteopathy (CMO) is the result of bone swelling during the growth of the bones of the skull and jaw. Sometimes, only the jaw is involved. As such, young dogs between the ages of three and eight months are the most commonly afflicted.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Soft coated wheaten Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Staffordshire Bull Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
L2 Hydroxyglutaric aciduria	T	C	CHR8	26723470	L2HGDH
L2 Hydroxyglutaric aciduria	A	G	CHR8	26760351	L2HGDH
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

L2 Hydroxyglutaric aciduria:
L-2-hydroxyglutaric aciduria is an autosomal recessive encephalopathy including neurological traits such as psychomotor impairment, seizures and ataxia. The onset of the disease occurs predominantly in dogs less than one year old, although it has been identified in older dogs.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Tibetan Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
crd4-PRA	-	44bp ins	CHR15	18332036	RPGRIP1
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Primary lens luxation	G	A	CHR37	40782144	ADAMTS17
Neuronal ceroid lipofuscinosis, 12	C	T	CHR2	81208162	ATP13A2
prcd-PRA	G	A	CHR9	4188663	PRCD

Crd4-PRA:
Inherited eye disease affecting dogs. PRA-crd4 occurs as a result of degeneration of both rod and cone type Photoreceptor Cells of the Retina, which are important for vision in dim and bright light, respectively.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Neuronal ceroid lipofuscinosis:
Group of progressive degenerative diseases of the central nervous system. Signs of disease in affected dogs begin between one and two years of age and include behaviour issues such as: anxiety, constant circling, aggression, compulsive behaviours, and loss of learned skills.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Primary lens luxation:
Results in a breakdown of the lens zonules – which usually occurs at a young age.

Weimaraner

Disorder	Normal	Mutation	Chr	Location	Gene
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
Hypomyelination	A	del	CHR15	5592827	FNIP2
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.
with the mutation will react to those drugs. Whether a dog reacts depends on the dosage of the drug.

Hyperuricosuria:
Excess of uric acid in urine.

Hypomyelination:
Congenital condition caused by insufficient myelin production in the body. This condition affects the central nervous system (CNS), with related tremors that are most apparent when a dog is active.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Welsh Springer Spaniel

Disorder	Normal	Mutation	Chr	Location	Gene
Factor VII Deficiency	G	A	CHR22	60578895	F7
Familial nephropathy	A	T	CHR25	39953906	COL4A4
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
prcd-PRA	G	A	CHR9	4188663	PRCD
Dilated cardiomyopathy	G	A	CHR1	58588129	

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Dilated cardiomyopathy:
Disease of predominantly large and giant breed dogs that results in progressive heart muscle dysfunction, chamber dilation, and eventual congestive heart failure or death of affected patients.

Factor VII Deficiency:
Mild to moderate inherited blood clotting disorder.

Familial nephropathy:
Familial nephropathy describes a disease in which young dogs suffer from early-onset kidney failure.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Welsh Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Primary lens luxation	G	A	CHR37	40782144	ADAMTS17
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Primary lens luxation:
Results in a breakdown of the lens zonules – which usually occurs at a young age.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

West Highland White Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Craniomandibular osteopathy	C	T	CHR5	9387327	SLC37A2
Von Willebrandt Type 1	G	A	CHR27	38951839	VWF
prcd-PRA	G	A	CHR9	4188663	PRCD
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Krabbe disease	T	G	CHR8	39311801	GALC

Craniomandibular osteopathy:
Craniomandibular Osteopathy (CMO) is the result of bone swelling during the growth of the bones of the skull and jaw. Sometimes, only the jaw is involved. As such, young dogs between the ages of three and eight months are the most commonly afflicted.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Krabbe disease:
It results in abnormal processing and storage of an enzyme that is critical in the production of myelin, the substance that coats and protects the nerves throughout the brain and spinal cord.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Von Willebrandt:
Blood clotting disorder.

Whippet

Disorder	Normal	Mutation	Chr	Location	Gene
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Glycogen Storage Disease	G	A	CHR27	6620819	PFKM
Myostatin deficiency	TG	del	CHR37	729360	MSTN
prcd-PRA	G	A	CHR9	4188663	PRCD

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.
with the mutation will react to those drugs. Whether a dog reacts depends on the dosage of the drug.

Glycogen Storage Disease:
Disorder is characterized by excessive accumulation of glycogen in the liver and other organs.

Myostatin deficiency (whippet type):
Inherited muscular disorder affecting dogs. Affected dogs have broad chests and overly developed muscles especially of the neck and legs as well as an overbite.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Yorkshire Terrier

Disorder	Normal	Mutation	Chr	Location	Gene
Craniomandibular osteopathy	C	T	CHR5	9387327	SLC37A2
Degenerative myelopathy	G	A	CHR31	26540348	SOD1
Primary lens luxation	G	A	CHR37	40782144	ADAMTS17
prcd-PRA	G	A	CHR9	4188663	PRCD
Hyperuricosuria	G	T	CHR3	69456869	SLC2A9
L2 Hydroxyglutaric aciduria	A	G	CHR8	26760351	L2HGDH

Craniomandibular osteopathy:
Craniomandibular Osteopathy (CMO) is the result of bone swelling during the growth of the bones of the skull and jaw. Sometimes, only the jaw is involved. As such, young dogs between the ages of three and eight months are the most commonly afflicted.

Degenerative myelopathy:
Non-painful progressive hind limb paralysis in older dogs.

Hyperuricosuria:
Excess of uric acid in urine.

L2-Hydroxyglutaric aciduria:
Autosomal recessive encephalopathy including neurological traits such as psychomotor impairment, seizures and ataxia. The onset of the disease occurs predominantly in dogs less than one year old, although it has been identified in older dogs.

Primary lens luxation:
Results in a breakdown of the lens zonules – which usually occurs at a young age.

Prcd-PRA:
Affects the photoreceptor cells in the eye involved in both night and day vision. PRCD-affected dogs have noticeable visual impairment by 4 years of age.

Certificates for health screening and coat colour testing

Unistel Medical Laboratories

Genetic Report

Canis lupus familiaris

Name:
Microchip number:
Registration ID:
Sample type: Buccal swab
Laboratory number: U
Date of birth:
Gender:
Breed: French Bulldog

Disorders Status

Potential health risks detected	0
Carrier of disorder detected	0

Test performed:	Status:
Cystinuria Type 3	Clear
Congenital hypothyroidism	Clear
Degenerative myelopathy	Clear
Hyperuricosuria	Clear
Multifocal Retinopathy	Clear
PRA- prcd	Clear
PRA-crd4	Clear
Brachycephaly	Double copy

Coat Colour & Traits*

Test performed:	Status:
A Locus: Agouti TCTCA; C>T Black-and-tan. Expression inhibited by K locus.	at/at
B Locus: Brown C>T; T>A; CCT/del Carries allele for brown. Coat colour will be dependent on results at A, E and K locus.	B/b
D Locus: Dilute G>A No dilution possible	D/D
E Locus: Extension C>T **E Locus: Extension** G>A Red/yellow coat colouration. Inhibits expression of A, B and K locus.	Em/Em e/e
K Locus: Dominant black CCC/del Inhibits expression of A and E locus.	K/N
Co Locus: Coco G>A Normal copies of gene. Cannot produce cocoa coat.	N/N
Coat Length	Short
Curly Coat	Non-curly

Unistel Medical Laboratories

Genetic Report

Health and Traits

Canis lupus familiaris

Name:
Microchip:
Registration ID:
Sample type: Buccal swab
Lab Number: U
Date of birth:
Gender:
Breed: French Bulldog

Disorder Status

Hypothyroidism: Results in an underactive thyroid gland. Mutation tested: C>T
Status: CLEAR

Multifocal Retinopathy: Eye disease. Affected dogs typically present with multiple, discrete circular areas of retinal detachment between 11 -16 weeks of age. Mutation tested: G>A
Status: CLEAR

Cystinuria: Cystine bladder stones appear to be the result of a genetic abnormality that prevents a dog from reabsorbing cystine from the kidneys. Mutation tested: G>A
Status: CLEAR

Degenerative myelopathy: Degeneration of the white matter of the spinal cord. Mutation tested: G>A
Status: CLEAR

Hyperuricemia: Results in an abnormally high level of uric acid in the blood. Mutation tested: G>T
Status: CLEAR

Progressive retinal atrophy: Inherited disease of dogs that causes slowly progressive blindness over a period of months or years. Mutation tested: G>A & small insertion
Status: CLEAR for prcd-PRA and crd4-PRA

Unistel Medical Laboratories

Genetic Report

Canis lupus familiaris

Name:
Microchip number:
Registration ID:
Sample type: Buccal swab
Laboratory number: U
Date of birth:
Gender:
Breed: French Bulldog

Coat Colour & Traits

Test performed:	Status:
A Locus: Agouti TCTCA; C > T Fawn/Sable possible	ay/ay
B Locus: Brown C > T	B/B
B Locus: Brown T > A	B/B
B Locus: Brown CCT/del Does not carry brown	B/B
D Locus: Dilute No dilution possible	D/D
E Locus: Extension C > T;G > A Carries black pigment. Inhibited by the expression of K locus.	E/E
K Locus: Dominant black Allows expression of A locus. Colour dependent on its genotypes at the E, A and B genes.	N/N
Coat Length Short coat length	N/N
Curly Coat Non-curly coat	N/N
Furnishing No furnishing present	N/N

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