

# Optimal Selection™

POWERED BY GENOSCOPE LABORATORIES

BR11 837  
Cash, Mixed breed

**Nickname:** Cash

**Owner:** Brooke Dent

**Microchip:** 933000320253600

**Country:** United States

**Breed:** Mixed breed

**Testing date:** 2020/1/29

**Gender:** Male

## Test results - Known disorders in the breed - page 1

| Disorder  | Type                    | Mode of Inheritance | Result |
|---|-------------------------|---------------------|--------|
| 2,8-Dihydroxyadenine (2,8-DHA) urolithiasis   | Renal Disorders         | Autosomal Recessive | Clear  |
| Alaskan Husky Encephalopathy, (AHE)   | Neurological Disorders  | Autosomal Recessive | Clear  |
| Amelogenesis Imperfecta, (AI); mutation originally found in Italian Greyhound           | Other Disorders         | Autosomal Recessive | Clear  |
| Bleeding disorder due to P2RY12 defect  | Blood Disorders         | Autosomal Recessive | Clear  |
| Canine Cyclic Neutropenia, Cyclic Hematopoiesis, Grey Collie Syndrome, (CN)             | Blood Disorders         | Autosomal Recessive | Clear  |
| Canine Leukocyte Adhesion Deficiency (CLAD), type III                                   | Blood Disorders         | Autosomal Recessive | Clear  |
| Centronuclear Myopathy, (CNM); mutation originally found in Labrador Retriever          | Muscular Disorders      | Autosomal Recessive | Clear  |
| Chondrodysplasia; mutation originally found in Norwegian Elkhound and Karelian Bear Dog | Skeletal Disorders      | Autosomal Recessive | Clear  |
| Complement 3 (C3) Deficiency  | Immunological Disorders | Autosomal Recessive | Clear  |

On behalf of Genoscooper Laboratories,



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|---|------------------------|---|--------|
| Cone Degeneration, (CD) or Achromatopsia; mutation originally found in Alaskan Malamute   | Ocular Disorders       | Autosomal Recessive                         | Clear  |
| Cone-Rod Dystrophy 1, (crd1); mutation originally found in American Staffordshire Terrier | Ocular Disorders       | Autosomal Recessive                         | Clear  |
| Cone-Rod Dystrophy 2, (crd2); mutation originally found in American Pit Bull Terrier      | Ocular Disorders       | Autosomal Recessive                         | Clear  |
| Congenital Hypothyroidism; mutation originally found in Toy Fox and Rat Terrier           | Endocrine Disorders    | Autosomal Recessive                         | Clear  |
| Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatitis, (CKCSID)              | Other Disorders        | Autosomal Recessive                         | Clear  |
| Craniomandibular Osteopathy, (CMO); mutation associated with terrier breeds               | Skeletal Disorders     | Autosomal Dominant (Incomplete Penetrance)  | Clear  |
| Cystinuria Type II-A; mutation originally found in Australian Cattle Dog                  | Renal Disorders        | Autosomal Dominant                          | Clear  |
| Degenerative Myelopathy, (DM; SOD1A)  | Neurological Disorders | Autosomal Recessive (Incomplete Penetrance) | Clear  |
| Dominant Progressive Retinal Atrophy, (DPRA)  | Ocular Disorders       | Autosomal Dominant                          | Clear  |

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| Disorder   | Type                    | Mode of Inheritance                         | Result |
|--|-------------------------|---|--------|
| Duchenne or Dystrophin Muscular Dystrophy, (DMD); mutation originally found in Golden Retriever  | Muscular Disorders      | X-linked Recessive                          | Clear  |
| Early Retinal Degeneration, (erd); mutation originally found in Norwegian Elkhound               | Ocular Disorders        | Autosomal Recessive                         | Clear  |
| Early-Onset Progressive Polyneuropathy; mutation originally found in Alaskan Malamute            | Neurological Disorders  | Autosomal Recessive                         | Clear  |
| Episodic Falling Syndrome, (EFS)   | Neuromuscular Disorders | Autosomal Recessive                         | Clear  |
| Exercise-Induced Collapse, (EIC)   | Neuromuscular Disorders | Autosomal Recessive (Incomplete Penetrance) | Clear  |
| Factor VII Deficiency  | Blood Disorders         | Autosomal Recessive                         | Clear  |
| Factor VIII Deficiency or Hemophilia A; p.Cys548Tyr mutation originally found in German Shepherd | Blood Disorders         | X-linked Recessive                          | Clear  |
| Fetal Onset Neuroaxonal Dystrophy, (FNAD)  | Neurological Disorders  | Autosomal Recessive                         | Clear  |
| GM2 Gangliosidosis, mutation originally found in Japanese Chin                                   | Neuromuscular Disorders | Autosomal Recessive                         | Clear  |

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## Test results - Known disorders in the breed - page 4

| Disorder  | Type                    | Mode of Inheritance | Result |
|---|-------------------------|---------------------|--------|
| Glanzmann Thrombasthenia Type I, (GT); mutation originally found in Pyrenean Mountain Dog                   | Blood Disorders         | Autosomal Recessive | Clear  |
| Glanzmann Thrombasthenia Type I, (GT); mutation originally found in mixed breed dogs                        | Blood Disorders         | Autosomal Recessive | Clear  |
| Globoid Cell Leukodystrophy or Krabbe Disease, (GLD); mutation originally found in Terriers                 | Neuromuscular Disorders | Autosomal Recessive | Clear  |
| Golden Retriever Progressive Retinal Atrophy 1, (GR_PRA 1)  | Ocular Disorders        | Autosomal Recessive | Clear  |
| Hereditary Ataxia or Cerebellar Ataxia; mutation originally found in Old English Sheepdog and Gordon Setter | Neurological Disorders  | Autosomal Recessive | Clear  |
| Hereditary Footpad Hyperkeratosis, (HFH)  | Dermal Disorders        | Autosomal Recessive | Clear  |
| Hereditary Phosphofructokinase (PFK) Deficiency   | Blood Disorders         | Autosomal Recessive | Clear  |
| Hereditary Vitamin D-Resistant Rickets, (HVDRR)   | Skeletal Disorders      | Autosomal Recessive | Clear  |
| Hyperekplexia or Startle Disease  | Neurological Disorders  | Autosomal Recessive | Clear  |

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## Test results - Known disorders in the breed - page 5

| Disorder   | Type                   | Mode of Inheritance | Result |
|--|------------------------|---------------------|--------|
| Hyperuricosuria, (HUU)   | Renal Disorders        | Autosomal Recessive | Clear  |
| Hypocatalasia or Acatlasemia   | Metabolic Disorders    | Autosomal Recessive | Clear  |
| Hypomyelination; mutation originally found in Weimaraner   | Neurological Disorders | Autosomal Recessive | Clear  |
| Intestinal Cobalamin Malabsorption or Imerslund-Gräsbeck Syndrome, (IGS); mutation originally found in Beagle        | Metabolic Disorders    | Autosomal Recessive | Clear  |
| Intestinal Cobalamin Malabsorption or Imerslund-Gräsbeck Syndrome, (IGS); mutation originally found in Border Collie | Metabolic Disorders    | Autosomal Recessive | Clear  |
| L-2-Hydroxyglutaric aciduria, (L2HGA); mutation originally found in Staffordshire Bull Terrier                       | Neurological Disorders | Autosomal Recessive | Clear  |
| Lamellar Ichthyosis, (LI)  | Dermal Disorders       | Autosomal Recessive | Clear  |
| Macrothrombocytopenia; disease-linked variant originally found in Norfolk and Cairn Terrier                          | Blood Disorders        | Autosomal Recessive | Clear  |
| May-Hegglin Anomaly (MHA)  | Blood Disorders        | Autosomal Dominant  | Clear  |

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## Test results - Known disorders in the breed - page 6

| Disorder   | Type                   | Mode of Inheritance | Result |
|--|------------------------|---------------------|--------|
| Mucopolysaccharidosis Type VII, (MPS VII); mutation originally found in German Shepherd      | Metabolic Disorders    | Autosomal Recessive | Clear  |
| Musladin-Lueke syndrome, (MLS)   | Dermal Disorders       | Autosomal Recessive | Clear  |
| Myostatin deficiency (Double Muscling, "Bully")  | Muscular Disorders     | Autosomal Recessive | Clear  |
| Narcolepsy; mutation originally found in Labrador Retriever                                  | Other Disorders        | Autosomal Recessive | Clear  |
| Neuronal Ceroid Lipofuscinosis 10, (NCL10); mutation originally found in American Bulldog    | Neurological Disorders | Autosomal Recessive | Clear  |
| Neuronal Ceroid Lipofuscinosis 8, (NCL8); mutation originally found in Australian Shepherd   | Neurological Disorders | Autosomal Recessive | Clear  |
| Osteochondrodysplasia; mutation originally found in Miniature Poodle                         | Skeletal Disorders     | Autosomal Recessive | Clear  |
| Osteogenesis Imperfecta, (OI); mutation originally found in Dachshund                        | Skeletal Disorders     | Autosomal Recessive | Clear  |
| Persistent Müllerian Duct Syndrome, (PMDS); mutation originally found in Miniature Schnauzer | Other Disorders        | Autosomal Recessive | Clear  |

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## Test results - Known disorders in the breed - page 7

| Disorder   | Type             | Mode of Inheritance                        | Result |
|--|------------------|--|--------|
| Prekallikrein Deficiency   | Blood Disorders  | Autosomal Recessive                        | Clear  |
| Primary Ciliary Dyskinesia, (PCD)  | Other Disorders  | Autosomal Recessive                        | Clear  |
| Primary Hereditary Cataract, (PHC); mutation originally found in Australian Shepherd | Ocular Disorders | Autosomal Dominant (Incomplete Penetrance) | Clear  |
| Primary Lens Luxation, (PLL)   | Ocular Disorders | Autosomal Recessive                        | Clear  |
| Primary Open Angle Glaucoma, (POAG); mutation originally found in Beagle             | Ocular Disorders | Autosomal Recessive                        | Clear  |
| Primary Open Angle Glaucoma, (POAG); mutation originally found in Norwegian Elkhound | Ocular Disorders | Autosomal Recessive                        | Clear  |
| Progressive Retinal Atrophy, (PRA); mutation originally found in Basenji             | Ocular Disorders | Autosomal Recessive                        | Clear  |
| Protein Losing Nephropathy, (PLN); NPHS1 gene variant                                | Renal Disorders  |  | Clear  |
| Pyruvate Kinase Deficiency; mutation originally found in Beagle                      | Blood Disorders  | Autosomal Recessive                        | Clear  |

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## Test results - Known disorders in the breed - page 8

| Disorder   | Type                   | Mode of Inheritance | Result |
|--|------------------------|---------------------|--------|
| Pyruvate Kinase Deficiency; mutation originally found in West Highland White Terrier                   | Blood Disorders        | Autosomal Recessive | Clear  |
| Renal Cystadenocarcinoma and Nodular Dermatofibrosis, (RCND)   | Renal Disorders        | Autosomal Dominant  | Clear  |
| Rod-Cone Dysplasia 1, (rcd1); mutation originally found in Irish Setter                                | Ocular Disorders       | Autosomal Recessive | Clear  |
| Rod-Cone Dysplasia 3, (rcd3)   | Ocular Disorders       | Autosomal Recessive | Clear  |
| Sensory Neuropathy; mutation originally found in Border Collie   | Neurological Disorders | Autosomal Recessive | Clear  |
| Skeletal Dysplasia 2, (SD2)  | Skeletal Disorders     | Autosomal Recessive | Clear  |
| Spinocerebellar Ataxia with Myokymia and/or Seizures (SCA)   | Neurological Disorders | Autosomal Recessive | Clear  |
| Spinocerebellar Ataxia/ Late-Onset Ataxia (SCA, LOA)   | Neurological Disorders | Autosomal Recessive | Clear  |
| Spongy Degeneration with Cerebellar Ataxia, (SDCA1); mutation originally found in Belgian Shepherd Dog | Neurological Disorders | Autosomal Recessive | Clear  |

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## Test results - Known disorders in the breed - page 9

| Disorder  | Type               | Mode of Inheritance | Result |
|---|--------------------|---------------------|--------|
| Trapped Neutrophil Syndrome, (TNS)  | Blood Disorders    | Autosomal Recessive | Clear  |
| Von Willebrand's Disease (vWD) Type 1   | Blood Disorders    | Autosomal Recessive | Clear  |
| Von Willebrand's Disease (vWD) Type 3; mutation originally found in Shetland Sheepdog | Blood Disorders    | Autosomal Recessive | Clear  |
| X-Linked Myotubular Myopathy  | Muscular Disorders | X-linked Recessive  | Clear  |
| X-Linked Progressive Retinal Atrophy 2, (XLPRA2; Type A PRA)                          | Ocular Disorders   | X-linked Recessive  | Clear  |
| Xanthinuria, Type 1a; mutation originally found in mixed breed dogs                   | Renal Disorders    | Autosomal Recessive | Clear  |

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## Test results for pharmacogenetics

| Disorder                        | Mode of Inheritance | Result |
|---------------------------------|---------------------|--------|
| Multi-Drug Resistance 1, (MDR1) | Autosomal Dominant  | Clear  |

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## Test results - Traits - page 1

### Coat Type

| Trait  | Genotype | Description   |
|--|----------|---|
| Coat Length  | I/I      | The dog is genetically long-haired.   |
| Furnishings / Improper Coat in Portuguese Water Dogs (marker test) | AG/TC    | The dog is likely genetically heterozygous at the furnishings locus, but may express the phenotype.   |
| KRT71 c.451C>T (p.Arg151Trp)                                       | C/T      | The dog is likely to have curly hair, if it is long-haired. The dog carries one copy of the tested allele causing curly coat, and may also pass on the non-curly allele to its offspring. |
| MC5R c.237A>T  | T/T      | The dog has two copies of the allele associated with low shedding. The dog is likely average or low shedder.  |
| SGK3 (p.Val96Glyfs)  | I/I      | The dog does not carry the tested hairlessness allele of the American Hairless Terrier.   |
| SGK3 c.137_138insT (p.Glu47Glyfs)                                  | D/D      | The dog does not carry the tested hairlessness allele of the Scottish Deerhound.  |

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## Test results - Traits - page 2

### Coat Colour

| Trait  | Genotype                                   | Description   |
|--|--|---|
| Colour Locus E - Extensions                        | Em/e                                       | The dog is likely to have a dark mask. The dog carries recessive red.   |
| Colour Locus B - Brown                             | b/b    bb/B                                | Dog with this genotype may express brown or black pigment. The dog has black pigment, if the b alleles causing brown colour are located on the same chromosome. |
| Colour Locus K - Dominant Black                    | KB/ky    KB/kbr<br>   kbr/ky   <br>kbr/kbr | The dog is genetically dominant black or brindle.   |
| Colour Locus A - Agouti                            | at/at                                      | The dog has genetically tan points or saddle tan pattern.   |
| Colour Locus S - Piebald or extreme white spotting | sp/sp                                      | The dog is likely to have piebald spotting or to be extreme white.  |
| Colour Locus H - Harlequin                         | h/h  | The dog doesn't have harlequin pattern.   |
| Dilution (d <sup>2</sup> allele)                   | D/D  | The dog does not carry any copies of the rare d <sup>2</sup> allele associated with dilution in Chow Chow, French Bulldog, Sloughi and Thai Ridgeback.          |
| Merle (M allele)                                   | m/m  | The dog is genetically non-merle and does not carry a <i>S/LV</i> gene SINE insertion.  |
| Saddle Tan ( <i>RALY</i> gene dupl.)               | dup/dup                                    | The dog may have tan points if it has tan point genotype at the A locus.  |
| Albinism (caL-allele)                              | C/C  | The dog does not carry the tested mutation for albinism.  |

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## Test results - Traits - page 3

### Body Size

| Trait                                  | Genotype | Description  |
|--|----------|--|
| <i>IGF1</i><br>(chr15:41221438)        | A/A      | The dog is homozygous for the derived allele typically associated with small body mass.  |
| <i>IGF1R</i> c.611G>A<br>(p.Arg204His) | A/A      | The dog has two derived alleles typically found in small and medium-sized breeds.  |
| <i>ACSL4</i><br>chrX:82919525C>T       | C/C      | The dog doesn't have the allele associated with large skeletal size and heavy muscling with considerable back fat thickness.           |
| <i>IGSF1</i> p.Asp768Glu               | C/C      | The dog doesn't have the allele associated with heavy muscling   |
| <i>IRS4</i><br>chrX:82296039           | A/A      | The dog has two copies of the allele associated with large body size.  |
| <i>FGF4</i> insertion                  | D/I      | The dog is heterozygous to the <i>fgf4</i> insertion associated with breed-defining Chondrodysplasia.                                  |
| <i>STC2</i><br>(chr4:39182836)         | A/T      | The dog carries one copy of the allele associated with reduced body size and one copy of the allele associated with no size reduction. |
| <i>GHR1</i><br>(p.Glu191Lys)           | A/A      | The dog is homozygous for the derived allele associated with reduced body size.  |
| <i>GHR2</i><br>(p.Pro177Leu)           | T/T      | The dog is homozygous for the derived allele associated with reduced body size.  |
| <i>HMGA2</i><br>(chr10:8348804)        | A/A      | The dog has two copies of the derived allele associated with reduced body size.  |

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## Test results - Traits - page 4

### Morphology

| Trait                               | Genotype | Description   |
|-------------------------------------|----------|---|
| <i>BMP3</i> c.1344C>A (p.Phe448Leu) | C/C      | The dog does not carry the tested allele typically associated with shortened head (brachycephaly). The dog is more likely to have an elongated head (dolichocephaly). |
| <i>SMOC2</i>                        | A/A      |   |
| chr10:11072007                      | C/T      | The dog carries one copy of an allele typically associated with floppy ears, and one copy of an allele typically associated with pricked ears.                        |
| <i>T</i> c.189C>G (p.Ile63Met)      | C/C      | The dog does not carry the tested bobtail-causing genetic variant. The dog is most likely long-tailed.  |
| <i>EPAS1</i> (p.Gly305Ser)          | G/G      | The dog does not carry the tested variant associated with adaptation to high altitudes.   |
| <i>LIMBR1</i> DC-1                  | G/G      | The dog does not carry the tested allele associated with hind dewclaws in Asian breeds. The dog is not likely to have hind dewclaws.                                  |
| <i>LIMBR1</i> DC-2                  | A/G      | The dog carries one copy of the allele associated with hind dewclaws in western breeds. About 50% of the dogs with this genotype have hind dewclaws.                  |
| <i>AXL4</i>                         | D/D      | The dog does not have the tested allele typically associated with blue eyes in Siberian Huskies. The dog is likely to have brown eyes.                                |

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## Test results - Additional disorders found in other breeds - page 1

### Blood Disorders

| Disorder  | Mode of Inheritance                        | Result |
|---|--|--------|
| Canine Scott Syndrome, (CSS)  | Autosomal Recessive                        | Clear  |
| Factor IX Deficiency or Hemophilia B; mutation Gly379Glu                                  | X-linked Recessive                         | Clear  |
| Factor IX Deficiency or Hemophilia B; mutation originally found in Airedale Terrier       | X-linked Recessive                         | Clear  |
| Factor IX Deficiency or Hemophilia B; mutation originally found in Lhasa Apso             | X-linked Recessive                         | Clear  |
| Factor VIII Deficiency or Hemophilia A; mutation originally found in Boxer                | X-linked Recessive                         | Clear  |
| Factor VIII Deficiency or Hemophilia A; mutation originally found in German Shepherd Dog  | X-linked Recessive                         | Clear  |
| Factor VIII Deficiency or Hemophilia A; mutation originally found in Havanaese            | X-linked Recessive                         | Clear  |
| Factor VIII Deficiency or Hemophilia A; mutation originally found in Old English Sheepdog | X-linked Recessive                         | Clear  |
| Factor XI Deficiency  | Autosomal Dominant (Incomplete Penetrance) | Clear  |
| Familial Congenital Methemoglobinemia; mutation originally found in Pomeranian            | Autosomal Recessive                        | Clear  |
| Hereditary Elliptocytosis   |  | Clear  |
| Pyruvate Kinase Deficiency; mutation originally found in Basenji                          | Autosomal Recessive                        | Clear  |
| Pyruvate Kinase Deficiency; mutation originally found in Pug                              | Autosomal Recessive                        | Clear  |
| Von Willebrand's Disease (vWD) Type 2   | Autosomal Recessive                        | Clear  |
| Von Willebrand's Disease (vWD) Type 3; mutation originally found in Kooikerhondje         | Autosomal Recessive                        | Clear  |
| Von Willebrand's Disease (vWD) Type 3; mutation originally found in Scottish Terrier      | Autosomal Recessive                        | Clear  |

## Test results - Additional disorders found in other breeds - page 2

### Ocular Disorders - page 1

| Disorder   | Mode of Inheritance                         | Result |
|--|---|--------|
| Canine Multifocal Retinopathy 1, (CMR1); mutation originally found in Mastiff-related breeds                           | Autosomal Recessive                         | Clear  |
| Canine Multifocal Retinopathy 2, (CMR2); mutation originally found in Coton de Tulear                                  | Autosomal Recessive                         | Clear  |
| Canine Multifocal Retinopathy 3, (CMR3); mutation originally found in Laponian Herder                                  | Autosomal Recessive                         | Clear  |
| Cone Degeneration, (CD) or Achromatopsia; mutation originally found in German Shepherd Dog                             | Autosomal Recessive                         | Clear  |
| Cone Degeneration, (CD) or Achromatopsia; mutation originally found in German Shorthaired Pointer                      | Autosomal Recessive                         | Clear  |
| Cone-Rod Dystrophy, (cord1-PRA / crd4)   | Autosomal Recessive (Incomplete Penetrance) | Clear  |
| Cone-Rod Dystrophy, Standard Wirehaired Dachshund, (crd SWD)   | Autosomal Recessive                         | Clear  |
| Congenital Eye Disease; mutation originally found in Irish Soft-Coated Wheaten Terrier                                 | Autosomal Recessive                         | Clear  |
| Early Onset PRA (EOPRA); mutation originally found in Portuguese Water Dog   | Autosomal Recessive                         | Clear  |
| Generalized Progressive Retinal Atrophy  | Autosomal Recessive                         | Clear  |
| Goniodysgenesis and glaucoma; mutation originally found in Border Collie   | Autosomal Recessive                         | Clear  |
| Italian Greyhound Progressive Retinal Atrophy 1 (IG-PRA1)  | Autosomal Recessive                         | Clear  |
| Primary Open Angle Glaucoma, (POAG); mutation originally found in Basset Fauve de Bretagne                             | Autosomal Recessive                         | Clear  |
| Primary Open Angle Glaucoma, (POAG); mutation originally found in Petit Basset Griffon Vendéen                         | Autosomal Recessive                         | Clear  |
| Primary lens luxation (PLL) and glaucoma; mutation originally found in Shar Pei  | Autosomal Recessive                         | Clear  |
| Progressive Retinal Atrophy (PRA4); mutation originally found in Lhasa Apso  | Autosomal Recessive                         | Clear  |
| Progressive Retinal Atrophy Type III, (PRA type III); mutation originally found in Tibetan Spaniel and Tibetan Terrier | Autosomal Recessive                         | Clear  |
| Progressive Retinal Atrophy, (CNGA1-PRA); mutation originally found in Shetland Sheepdog                               | Autosomal Recessive                         | Clear  |
| Progressive Retinal Atrophy, (PAP1_PRA); mutation originally found in Papillon and Phalene                             | Autosomal Recessive                         | Clear  |
| Progressive Retinal Atrophy, (PRA); mutation originally found in Swedish Vallhund                                      | Autosomal Recessive                         | Clear  |
| Rod-Cone Dysplasia 1a, (rdc1a); mutation originally found in Sloughi   | Autosomal Recessive                         | Clear  |



## Test results - Additional disorders found in other breeds - page 3

### Ocular Disorders - page 2

| Disorder  | Mode of Inheritance | Result |
|---|---------------------|--------|
| X-Linked Progressive Retinal Atrophy 1, (XLPR1) | X-linked Recessive  | Clear  |

### Cardiac Disorders

| Disorder  | Mode of Inheritance | Result |
|---|---------------------|--------|
| Dilated Cardiomyopathy, (DCM); mutation originally found in Schnauzer | Autosomal Recessive | Clear  |
| Long QT Syndrome  | Autosomal Dominant  | Clear  |

### Endocrine Disorders

| Disorder  | Mode of Inheritance | Result |
|---|---------------------|--------|
| Congenital Dysmaturagenic Hypothyroidism with Goiter; mutation originally found in Shih Tzu | Autosomal Recessive | Clear  |
| Congenital Hypothyroidism; mutation originally found in Tenterfield Terrier                 | Autosomal Recessive | Clear  |

### Immunological Disorders

| Disorder   | Mode of Inheritance | Result |
|--|---------------------|--------|
| Autosomal Recessive Severe Combined Immunodeficiency, (ARSCID)                                       | Autosomal Recessive | Clear  |
| Myeloperoxidase Deficiency   | Autosomal Recessive | Clear  |
| Severe Combined Immunodeficiency in Frisian Water Dogs, (SCID)                                       | Autosomal Recessive | Clear  |
| X-Linked Severe Combined Immunodeficiency (XSCID); mutation originally found in Basset Hound         | X-linked Recessive  | Clear  |
| X-Linked Severe Combined Immunodeficiency (XSCID); mutation originally found in Cardigan Welsh Corgi | X-linked Recessive  | Clear  |

## Test results - Additional disorders found in other breeds - page 4

### Renal Disorders

| Disorder  | Mode of Inheritance | Result |
|---|---------------------|--------|
| Cystic Renal Dysplasia and Hepatic Fibrosis; mutation originally found in Norwich Terrier                   | Autosomal Recessive | Clear  |
| Cystinuria Type I-A; mutation originally found in Newfoundland Dog  | Autosomal Recessive | Clear  |
| Fanconi Syndrome  | Autosomal Recessive | Clear  |
| Polycystic Kidney Disease in Bull Terriers, (BTPKD)   | Autosomal Dominant  | Clear  |
| Primary Hyperoxaluria, (PH); mutation originally found in Coton de Tulear                                   | Autosomal Recessive | Clear  |
| X-Linked Hereditary Nephropathy, (XLHN)   | X-linked Recessive  | Clear  |
| X-Linked Hereditary Nephropathy, (XLHN); mutation originally found in Navasota Dog                          | X-linked Recessive  | Clear  |
| Xanthinuria, Type 2a; mutation originally found in Toy Manchester Terrier                                   | Autosomal Recessive | Clear  |
| Xanthinuria, Type 2b; mutation originally found in Cavalier King Charles Spaniel and English Cocker Spaniel | Autosomal Recessive | Clear  |

### Metabolic Disorders

| Disorder   | Mode of Inheritance | Result |
|--|---------------------|--------|
| Glycogen Storage Disease Type II or Pompe's Disease, (GSD II)                                  | Autosomal Recessive | Clear  |
| Glycogen Storage Disease Type IIIa, (GSD IIIa)   | Autosomal Recessive | Clear  |
| Glycogen Storage Disease Type Ia, (GSD Ia)   | Autosomal Recessive | Clear  |
| Mucopolysaccharidosis Type IIIA, (MPS IIIA); mutation originally found in Dachshund            | Autosomal Recessive | Clear  |
| Mucopolysaccharidosis Type IIIA, (MPS IIIA); mutation originally found in New Zealand Huntaway | Autosomal Recessive | Clear  |
| Mucopolysaccharidosis Type VII, (MPS VII); mutation originally found in Brazilian Terrier      | Autosomal Recessive | Clear  |
| Pyruvate Dehydrogenase Phosphatase 1 (PDP1) Deficiency   | Autosomal Recessive | Clear  |

## Test results - Additional disorders found in other breeds - page 5

### Muscular Disorders

| Disorder   | Mode of Inheritance | Result |
|--|---------------------|--------|
| Cavalier King Charles Spaniel Muscular Dystrophy, (CKCS-MD)                                    | X-linked Recessive  | Clear  |
| Centronuclear Myopathy, (CNM); mutation originally found in Great Dane                         | Autosomal Recessive | Clear  |
| Duchenne or Dystrophin Muscular Dystrophy, (DMD); mutation originally found in Norfolk Terrier | X-linked Recessive  | Clear  |
| Muscular Dystrophy, Ullrich-type; mutation originally found in Landseer                        | Autosomal Recessive | Clear  |
| Myotonia Congenita; mutation originally found in Australian Cattle Dog                         | Autosomal Recessive | Clear  |
| Myotubular Myopathy; mutation originally found in Rottweiler                                   | X-linked Recessive  | Clear  |
| Nemaline Myopathy; mutation originally found in American Bulldog                               | Autosomal Recessive | Clear  |

## Test results - Additional disorders found in other breeds - page 6

### Neurological Disorders - page 1

| Disorder   | Mode of Inheritance | Result |
|--|---------------------|--------|
| Acral Mutilation Syndrome, (AMS)   | Autosomal Recessive | Clear  |
| Alexander Disease (AxD); mutation originally found in Labrador Retriever                               | Autosomal Dominant  | Clear  |
| Bandera's Neonatal Ataxia, (BNAt)  | Autosomal Recessive | Clear  |
| Benign Familial Juvenile Epilepsy or Remitting Focal Epilepsy  | Autosomal Recessive | Clear  |
| Cerebellar Cortical Degeneration, (CCD); mutation originally found in Vizsla                           | Autosomal Recessive | Clear  |
| Cerebral Dysfunction; mutation originally found in Friesian Stabyhoun                                  | Autosomal Recessive | Clear  |
| Dandy-Walker-Like Malformation (DWLM); mutation originally found in Eurasier                           | Autosomal Recessive | Clear  |
| Hereditary Ataxia; mutation originally found in in Norwegian Buhund                                    | Autosomal Recessive | Clear  |
| Juvenile Myoclonic Epilepsy, (JME); mutation originally found in Rhodesian Ridgeback                   | Autosomal Recessive | Clear  |
| Juvenile encephalopathy; mutation originally found in Parson Russell Terrier                           | Autosomal Recessive | Clear  |
| L-2-Hydroxyglutaric aciduria, (L2HGA); mutation originally found in West Highland White Terrier        | Autosomal Recessive | Clear  |
| Lagotto Storage Disease, (LSD)   | Autosomal Recessive | Clear  |
| Neonatal Cerebellar Cortical Degeneration or Cerebellar Abiotrophy, (NCCD)                             | Autosomal Recessive | Clear  |
| Neonatal Encephalopathy with Seizures, (NEWS)  | Autosomal Recessive | Clear  |
| Neuroaxonal Dystrophy (NAD); mutation originally found in Rottweiler                                   | Autosomal Recessive | Clear  |
| Neuroaxonal Dystrophy (NAD); mutation originally found in Spanish Water Dog                            | Autosomal Recessive | Clear  |
| Neuroaxonal Dystrophy, (NAD); mutation originally found in Papillon                                    | Autosomal Recessive | Clear  |
| Neuronal Ceroid Lipofuscinosis 1, (NCL1); mutation originally found in Dachshund                       | Autosomal Recessive | Clear  |
| Neuronal Ceroid Lipofuscinosis 8, (NCL8); mutation originally found in Alpine Dachsbracke              | Autosomal Recessive | Clear  |
| Neuronal Ceroid Lipofuscinosis 8, (NCL8); mutation originally found in English Setter                  | Autosomal Recessive | Clear  |
| Neuronal Ceroid Lipofuscinosis, (NCL7); mutation originally found in Chinese Crested Dog and Chihuahua | Autosomal Recessive | Clear  |

## Test results - Additional disorders found in other breeds - page 7

### Neurological Disorders - page 2

| Disorder   | Mode of Inheritance | Result |
|--|---------------------|--------|
| Polyneuropathy with ocular abnormalities and neuronal vacuolation, (POANV); mutation originally found in Black Russian Terrier | Autosomal Recessive | Clear  |
| Progressive Early-Onset Cerebellar Ataxia; mutation originally found in Finnish Hound  | Autosomal Recessive | Clear  |
| Shaking Puppy Spongiform LeucoEncephaloMyelopathy, (SLEM); mutation originally found in Border Terrier                         | Autosomal Recessive | Clear  |
| Spongy Degeneration with Cerebellar Ataxia, (SDCA2); mutation originally found in Belgian Shepherd Dog                         | Autosomal Recessive | Clear  |
| X-Linked Tremors; mutation originally found in English Springer Spaniel  | X-linked Recessive  | Clear  |

### Neuromuscular Disorders

| Disorder  | Mode of Inheritance | Result |
|---|---------------------|--------|
| Congenital Myasthenic Syndrome (CMS); mutation originally found in Labrador Retriever           | Autosomal Recessive | Clear  |
| Congenital Myasthenic Syndrome, (CMS); mutation originally found in Jack Russell Terrier        | Autosomal Recessive | Clear  |
| Congenital Myasthenic Syndrome, (CMS); mutation originally found in Old Danish Pointing Dog     | Autosomal Recessive | Clear  |
| GM1 Gangliosidosis; mutation originally found in Portuguese Water Dog                           | Autosomal Recessive | Clear  |
| GM2 Gangliosidosis; mutation originally found in Toy Poodle                                     | Autosomal Recessive | Clear  |
| Globoid Cell Leukodystrophy or Krabbe Disease, (GLD); mutation originally found in Irish Setter | Autosomal Recessive | Clear  |
| Paroxysmal Dyskinesia, (PxD); mutation originally found in Irish Soft Coated Wheaten Terrier    | Autosomal Recessive | Clear  |

## Test results - Additional disorders found in other breeds - page 8

### Skeletal Disorders

| Disorder  | Mode of Inheritance | Result |
|---|---------------------|--------|
| Cleft Palate; Cleft Lip and Palate with Syndactyly; ADAMTS20 gene mutation originally found in Nova Scotia Duck Tolling Retriever | Autosomal Recessive | Clear  |
| Cleft Palate; DLX6 gene mutation originally found in Nova Scotia Duck Tolling Retriever   | Autosomal Recessive | Clear  |
| Osteochondromatosis; mutation originally found in American Staffordshire Terrier  | Autosomal Dominant  | Clear  |
| Osteogenesis Imperfecta, (OI); mutation originally found in Beagle  | Autosomal Dominant  | Clear  |
| Skeletal Disease (Hypophosphatasia); mutation originally found in Karelian Bear Dog   | Autosomal Recessive | Clear  |
| Spondylocostal Dysostosis   | Autosomal Recessive | Clear  |
| Van den Ende-Gupta Syndrome, (VDEGS)  | Autosomal Recessive | Clear  |

### Dermal Disorders

| Disorder   | Mode of Inheritance | Result |
|--|---------------------|--------|
| Dystrophic Epidermolysis Bullosa; mutation originally found in Central Asian Ovcharka                      | Autosomal Recessive | Clear  |
| Dystrophic Epidermolysis Bullosa; mutation originally found in Golden Retriever                            | Autosomal Recessive | Clear  |
| Epidermolytic Hyperkeratosis   | Autosomal Recessive | Clear  |
| Focal Non-Epidermolytic Palmoplantar Keratoderma, (FNEPPK); mutation originally found in Dogue de Bordeaux | Autosomal Recessive | Clear  |
| Hereditary Nasal Parakeratosis, (HNPK); mutation originally found in Greyhound                             | Autosomal Recessive | Clear  |
| Ichthyosis; mutation originally found in American Bulldog  | Autosomal Recessive | Clear  |
| Ichthyosis; mutation originally found in Great Dane  | Autosomal Recessive | Clear  |
| Lethal Acrodermatitis, (LAD); mutation originally found in in Bull Terrier and Miniature Bull Terrier      | Autosomal Recessive | Clear  |
| Ligneous Membranitis   | Autosomal Recessive | Clear  |
| X-Linked Ectodermal Dysplasia, (XHED)  | X-linked Recessive  | Clear  |

## Test results - Additional disorders found in other breeds - page 9

### Other Disorders

| Disorder  | Mode of Inheritance | Result |
|---|---------------------|--------|
| Acute Respiratory Distress Syndrome, (ARDS); mutation originally found in Dalmatian | Autosomal Recessive | Clear  |
| Amelogenesis Imperfecta, (AI); mutation originally found in Parson Russell Terrier  | Autosomal Recessive | Clear  |
| Dental Hypomineralisation; mutation originally found in Border Collie               | Autosomal Recessive | Clear  |
| Lung Developmental Disease; mutation originally found in in Airedale Terrier        | Autosomal Recessive | Clear  |
| Narcolepsy; mutation originally found in Dachshund                                  | Autosomal Recessive | Clear  |

## APPENDIX Explanation of the results of the tested disorders

### Autosomal recessive inheritance (ARI)

Clear - A dog carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

Carrier - A dog carries one copy of the tested mutation. Carriers typically have a normal, healthy appearance but pass on the mutation to approximately 50% of their offspring.

At risk - A dog carries two copies of the tested mutation and is at high or increased risk of developing the disease/condition.

### Autosomal dominant inheritance (ADI)

Clear - A dog carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

At risk - A dog carries one or two copies of the tested mutation and is at high or increased risk of developing the disease/condition.

### X-linked recessive inheritance (X-linked)

Clear - A dog carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

Carrier - Female carriers typically have a normal, healthy appearance but carry one copy of the tested mutation on one of their X chromosomes. As males only have one X chromosome, there are no male carriers.

At risk - Female dogs at risk carry two mutated copies of the tested mutation. Males carry one copy of the tested mutation on their single X chromosome. Dogs at risk are at high or increased risk of developing the disease/condition.

Please note that the descriptions above are generalized based on typically observed inheritance patterns. When obtaining a 'carrier' or 'at risk' test result, always refer to the corresponding online test documentation for more detailed information on the condition and any exceptions.





BR11 837  
Cash, Mixed breed

## Terms and Conditions

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