# Ridgeback International Genetic Profile CERTIFICATE

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Name dog: Aaron Aus Dem Artland

Registrationnumber: IRV363459

MicroChipnumber: 276093400451584

Date of birth: 11-05-2013

Gender : Rüde

Owner : Stephan Gruber Address : Flechtenweg 11

Town : Meppen Country : Germany

### **R.I.GE.P.** (Ridgeback International Genetic Profile) results:

Disorder	Result
Neurological disorders	
L-2-Hydroxyglutaric aciduria (L2HGA); mutation 1	Free
L-2-Hydroxyglutaric aciduria (L2HGA); mutation 2 originally found in Staffordshire Bull Terrier	Unknown
Neonatal Encephalopathy with Seizures (NEWS)	Free
Cerebellar abiotrophy or neonatal cerebellar cortical degeneration (NCCD)	Free
Polyneuropathy; mutation originally found in Alaskan Malamute	Free
Bandera's Neonatal Ataxia (BNAt)	Unknown
Progressive early-onset cerebellar ataxia; mutation originally found in Finnish Hound	Free
Benign Familial Juvenile Epilepsy or Remitting Focal Epilepsy	Free
Polyneuropathy; mutation originally found in Greyhound	Free
Neuronal Ceroid Lipofuscinosis 1 (NCL1)	Free
Neuronal Ceroid Lipofuscinosis, type 12, mutation originally found in Tibetan terrier	Free
Neuronal Ceroid Lipofuscinosis 10 (NCL10)	Free
Neuronal Ceroid Lipofuscinosis 2 (NCL2)	Unknown
Neuronal Ceroid Lipofuscinosis 4A (NCL4)	Unknown
Neuronal Ceroid Lipofuscinosis 5 (NCL5)	Free
Neuronal Ceroid Lipofuscinosis 6 (NCL6)	Unknown
Fetal-onset Neuroaxonal Dystrophy (FNAD)	Free
L-2-hydroxyglutaric Aciduria (L2HGA), Yorkshire Terrier mutation	Unknown
Bandera's Neonatal Ataxia (BNAt)	Free
Alaskan Husky Encephalopathy (AHE)	Free
Neuronal Ceroid Lipofuscinosis 8 (NCL8)	Free
Neuronal Ceroid Lipofuscinosis 8 (NCL8), rare variant	Free
Spinocerebellar ataksia (SCA); mutation originally found in Parson Russell Terrier	Free
Shaking Puppy (X-linked Generalized Tremor); mutation originally found in English Springer Spaniel	Free
Hypomyelination and Tremor; mutation originally found in Weimaraner	Free
Lagotto Storage Disease	Free
Spinocerebellar ataxia with myokymia and/or seizures	Free
Spinal Dysraphism	Free
Dandy-Walker-Like Malformation (DWLM); mutation originally found in Eurasier	Free
Neuronal Ceroid Lipofuscinosis, (NCL7); mutation originally found in Chinese Crested Dog and Chihuahua	Free
Cerebral Dysfunction; mutation originally found in Friesian Stabyhoun	Free
Neuroaxonal Dystrophy (NAD); mutation originally found in Spanish Water Dog	Free

#### Eye disorders

Canine Multifocal Retinopathy 1 (CMR1), Eye disorder. Collie Eye Anomaly (CEA)

Free Unknown

Cone and Display 1 (count DDA)	Гиол
Congenital Stationary Night Blindness (CSNR)	Free Free
Congenital Stationary Night Blindness (CSNB) Autosomal Dominant Progressive Retinal Atrophy (ADPRA)	Free
Golden Retriever Progressive Retinal Atrophy 1 (GR_PRA 1)	Free
Primary Hereditary Cataract (PHC); mutation originally found in Australian Shepherd	Free
Primary Lens Luxation (PLL)	Free
Rod-Cone Dysplasia 1 (rcd1); mutation originally found in Irish Setter	Free
Rod-Cone Dysplasia 1a (rcd1a); mutation originally found in Sloughi	Free
Rod-Cone Dysplasia 3 (rcd3)	Free
X-Linked Progressive Retinal Atrophy 1 (XLPRA1)	Free
Cone-rod dystrophy (crd SWD); mutation originally found in Standard Wire-haired Dachshund	Free
Primary Open Angle Glaucoma; mutation originally found in Beagle	Free
Achromatopsia or Cone Degeneration (CD); mutation originally found in German Shorthaired Pointer	Free
OSD2 (Oculoskeletal Dysplasia 2) or DRD2 (Dwarfism-Retinal Dysplasia 2)	Unknown
Canine Multifocal Retinopathy 3 (cmr3); mutation originally found in Lapponian Herder	Free
Canine Multifocal Retinopathy 2 (cmr2); mutation originally found in Coton de Tulear	Free
Generalized Progressive Retinal Atrophy; mutation originally found in Schapendoes	Free
Golden Retriever Progressive Retinal Atrophy 2 (GR_PRA 2)	Unknown
Early Retinal Degeneration; mutation originally found in Norwegian Elkhound	Unknown
Primary Hereditary Cataract (PHC); mutation originally found in Terriers	Unknown
Progressive Retinal Atrophy (PAP1_PRA); mutation originally found in Papillon and Phalene	Free
Progressive Retinal Atrophy - adult onset; mutation originally found in Basenji	Free
Progressive Retinal Atrophy (PRA), type III; mutation originally found in Tibetan Spaniel and Tibetan Terrier	Free
Cone-Rod Dystrophy 2 (crd2); mutation originally found in Pit Bull Terrier	Free
Glaucoma; mutation originally found in Norwegian Elkhound	Free
PRA Progressive Retinal Atrophy; Swedish Vallhund marker test	Free
X-Linked Progressive Retinal Atrophy 2 (XLPRA2)	Free
Achromatopsia or Cone Degeneration (CD); CNGB3 gene deletion	Free
Cone-Rod Dystrophy 1 (crd1); mutation originally found in American Staffordshire Terrier	Free
Progressive Retinal Atrophy, (CNGA1-PRA); mutation originally found in Shetland Sheepdog	Free
Neuromuscular disorders	
Degenerative Myelopathy (DM)	Free
Exercise-Induced Collapse (EIC)	Unknown
Globoid Cell Leukodystrophy (GLD) or Krabbe's disease, Terrier mutation	Free
Alpha Fucosidosis	Unknown
Globoid Cell Leukodystrophy (GLD) or Krabbe's disease; mutation originally found in Irish Setter	Unknown
GM1 Gangliosidosis; mutation originally found in Portuguese Water Dog	Free
GM1 Gangliosidosis; mutation originally found in Alaskan Husky	Free
GM1 Gangliosidosis; mutation originally found in Shiba Dog	Free
Episodic falling (EF)	Unknown
GM2 Gangliosidosis	Free
Hyperekplexia or Startle Disease	Free
GM2 Gangliosidosis, mutation originally found in Japanese Chin	Free
Congenital Myasthenic Syndrome (CMS)	Free
Globoid Cell Leukodystrophy (GLD) or Krabbe's disease; mutation originally found in Irish Setter	Free
Congenital Myasthenic Syndrome, (CMS); mutation originally found in Jack Russell Terrier	Free
Metabolic disorders	
Glycogen Storage Disease, type IIIa (GSDIIIa)	Free
Mucopolysaccharidosis Type VII (MPSVII); mutation originally found in Brazilian Terrier	Free
Pyruvate Dehydrogenase Deficiency	Free
Glycogen Storage Disease, type II or Pompe's disease	Free
Glycogen Storage Disease, Type Ia (GSDIa)	Free
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	Unknown
Mucopolysaccharidosis Type I (MPSI)	
	Unknown
Mucopolysaccharidosis Type I (MPSI) Mucopolysaccharidosis Type IIIA (MPSIIIA); mutation originally found in Dachshund	Unknown Free
Mucopolysaccharidosis Type I (MPSI) Mucopolysaccharidosis Type IIIA (MPSIIIA); mutation originally found in Dachshund Mucopolysaccharidosis Type IIIA (MPSIIIA); mutation originally found in New Zealand Huntaway	Unknown Free Unknown
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Mucopolysaccharidosis Type I (MPSI)  Mucopolysaccharidosis Type IIIA (MPSIIIA); mutation originally found in Dachshund  Mucopolysaccharidosis Type IIIA (MPSIIIA); mutation originally found in New Zealand Huntaway  Mucopolysaccharidosis Type VI (MPSVI); mutation originally found in Poodle  Hypocatalasia or Acatalasemia	Unknown Free Unknown Unknown Free
Mucopolysaccharidosis Type I (MPSI)  Mucopolysaccharidosis Type IIIA (MPSIIIA); mutation originally found in Dachshund  Mucopolysaccharidosis Type IIIA (MPSIIIA); mutation originally found in New Zealand Huntaway  Mucopolysaccharidosis Type VI (MPSVI); mutation originally found in Poodle  Hypocatalasia or Acatalasemia  Mucopolysaccharidosis Type VII (MPS VII); mutation originally found in German Shepherd	Unknown Free Unknown Unknown Free Free Free

#### **Blood disorders**

Trapped Neutrophil Syndrome (TNS)  Von Willebrand's Disease (WDD) Type III; mutation originally found in Kooikerhondje  Free Canine Leukocyte Adhesion Deficiency (CLAD), type I  Unknown May-Hegglin Anomaly (MHA)  Free Factor IX Deficiency or Haemophilia B, mutation originally found in Lhasa Apso Free Factor IX Deficiency or Haemophilia B, Gly379Glu mutation Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in West Highland White Terrier Free Glanzmann Thrombasthenia (GT), Type I; mutation originally found in Pyrenean Mountain Dog Free Glycogen Storage Disease VII or Hereditary Phosphofructokinase (PFK) Deficiency Free Bleeding disorder due to PZRV12 defect Free Von Willebrand's Disease (wWD) Type III; mutation originally found in Shetland Sheepdog Free Factor VII Deficiency Free Factor VIII Deficiency or Haemophilia A; mutation originally found in German Shepherd Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Pug Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Pug Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Beagle Canine Leucocyte Adhesion Deficiency (CLAD), type III Free Factor IX Deficiency or Haemophilia B; mutation originally found in Rhodesian Ridgeback Free Factor IX Deficiency or Haemophilia B; mutation originally found in Miredale Terrier Factor IX Deficiency or Haemophilia B; mutation originally found in Miredale Terrier Factor IX Deficiency or Haemophilia B; mutation originally found in Basenji Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Basenji Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Basenji Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Basenji Free Pyruvate Kinase Deficiency of Frethrocyte; mutation originally found in Beannyi Free Pyruvate Kinase Deficiency of Frythrocyte; mutation originally found in Beannyi Free Pyruvate Kinase Deficiency of Free Pyruvate Kinase Deficiency of Free Py	Canine Cyclic Neutropenia (Gray Collie Syndrome)	Free
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Factor VII Deficiency Factor VIII deficiency or Haemophilia A; mutation originally found in German Shepherd Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Labrador Retriever Unknown Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Pug Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Beagle Canine Leucocyte Adhesion Deficiency (CLAD), type III Free Factor IX Deficiency or Haemophilia B; mutation originally found in Rhodesian Ridgeback Free Factor IX Deficiency or Haemophilia B; mutation originally found in Airedale Terrier Free Factor IX Deficiency or Haemophilia B; mutation originally found in German Wirehaired Pointer Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Basenji Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in West Highland White Terrier Unknown Von Willebrand's Disease (wWD) Type II Free Congenital Macrothrombocytopenia; disease-linked SNP originally found in Norfolk and Cairn Terrier Free Thrombopathia; mutation originally found in Basset Hound Free Thrombopathia; mutation originally found in Landseer Free Thrombopathia; mutation originally found in Eskimo Spitz Free Prekallikrein Deficiency Free Fliptocytosis Free Fliptocytosis Free Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd Free Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer Free Von Willebrand's Disease (wWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (wWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (wWD) Type III; mutation originally found in Scottish Terrier Free	Bleeding disorder due to P2RY12 defect	Free
Factor VIII deficiency or Haemophilia A; mutation originally found in German Shepherd Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Labrador Retriever Unknown Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Pug Prevate Kinase Deficiency of Erythrocyte; mutation originally found in Beagle Canine Leucocyte Adhesion Deficiency (CLAD), type III Free Factor IX Deficiency or Haemophilia B; mutation originally found in Rhodesian Ridgeback Free Factor IX Deficiency or Haemophilia B; mutation originally found in Airedale Terrier Factor IX Deficiency or Haemophilia B; mutation originally found in German Wirehaired Pointer Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Basenji Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in West Highland White Terrier Unknown Von Willebrand's Disease (vWD) Type II Free Congenital Macrothrombocytopenia; disease-linked SNP originally found in Norfolk and Cairn Terrier Free Thrombopathia; mutation originally found in Basset Hound Free Thrombopathia; mutation originally found in Landseer Free Thrombopathia; mutation originally found in Eskimo Spitz Free Prekallikrein Deficiency Free Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd Free Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Found in Scottish Terrier Free	Von Willebrand's Disease (vWD) Type III; mutation originally found in Shetland Sheepdog	Free
Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Labrador Retriever Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Pug Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Beagle Canine Leucocyte Adhesion Deficiency (CLAD), type III Free Factor IX Deficiency or Haemophilia B; mutation originally found in Rhodesian Ridgeback Free Factor IX Deficiency or Haemophilia B; mutation originally found in Airedale Terrier Frec Factor IX Deficiency or Haemophilia B; mutation originally found in German Wirehaired Pointer Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Basenji Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in West Highland White Terrier Unknown Von Willebrand's Disease (vWD) Type II Free Congenital Macrothrombocytopenia; disease-linked SNP originally found in Norfolk and Cairn Terrier Free Thrombopathia; mutation originally found in Basset Hound Free Thrombopathia; mutation originally found in Landseer Free Prekallikrein Deficiency Free Prekallikrein Deficiency Free Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd Free Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free	Factor VII Deficiency	Free
Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Pug Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Beagle Canine Leucocyte Adhesion Deficiency (CLAD), type III Free Factor IX Deficiency or Haemophilia B; mutation originally found in Rhodesian Ridgeback Free Factor IX Deficiency or Haemophilia B; mutation originally found in Airedale Terrier Free Factor IX Deficiency or Haemophilia B; mutation originally found in German Wirehaired Pointer Free Factor IX Deficiency of Erythrocyte; mutation originally found in Basenji Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in West Highland White Terrier Unknown Von Willebrand's Disease (VWD) Type II Free Congenital Macrothrombocytopenia; disease-linked SNP originally found in Norfolk and Cairn Terrier Free Thrombopathia; mutation originally found in Basset Hound Free Thrombopathia; mutation originally found in Landseer Free Thrombopathia; mutation originally found in Eskimo Spitz Free Prekallikrein Deficiency Free Fliptocytosis Free Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd Free Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free	Factor VIII deficiency or Haemophilia A; mutation originally found in German Shepherd	Free
Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Beagle Canine Leucocyte Adhesion Deficiency (CLAD), type III Free Factor IX Deficiency or Haemophilia B; mutation originally found in Rhodesian Ridgeback Free Factor IX Deficiency or Haemophilia B; mutation originally found in Airedale Terrier Free Factor IX Deficiency or Haemophilia B; mutation originally found in German Wirehaired Pointer Free Factor IX Deficiency of Haemophilia B; mutation originally found in German Wirehaired Pointer Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Basenji Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in West Highland White Terrier Unknown Von Willebrand's Disease (vWD) Type II Free Congenital Macrothrombocytopenia; disease-linked SNP originally found in Norfolk and Cairn Terrier Thrombopathia; mutation originally found in Basset Hound Free Thrombopathia; mutation originally found in Landseer Thrombopathia; mutation originally found in Eskimo Spitz Free Prekallikrein Deficiency Elliptocytosis Free Elliptocytosis Free Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd Free Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free	Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Labrador Retriever	Unknown
Canine Leucocyte Adhesion Deficiency (CLAD), type III Free Factor IX Deficiency or Haemophilia B; mutation originally found in Rhodesian Ridgeback Free Factor IX Deficiency or Haemophilia B; mutation originally found in Airedale Terrier Free Factor IX Deficiency or Haemophilia B; mutation originally found in German Wirehaired Pointer Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Basenji Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in West Highland White Terrier Unknown Von Willebrand's Disease (vWD) Type II Free Congenital Macrothrombocytopenia; disease-linked SNP originally found in Norfolk and Cairn Terrier Free Thrombopathia; mutation originally found in Basset Hound Free Thrombopathia; mutation originally found in Landseer Free Thrombopathia; mutation originally found in Eskimo Spitz Free Prekallikrein Deficiency Free Elliptocytosis Free Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in Boxer Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free	Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Pug	Free
Factor IX Deficiency or Haemophilia B; mutation originally found in Rhodesian Ridgeback Free Factor IX Deficiency or Haemophilia B; mutation originally found in Airedale Terrier Free Factor IX Deficiency or Haemophilia B; mutation originally found in German Wirehaired Pointer Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Basenji Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in West Highland White Terrier Unknown Von Willebrand's Disease (vWD) Type II Free Congenital Macrothrombocytopenia; disease-linked SNP originally found in Norfolk and Cairn Terrier Free Thrombopathia; mutation originally found in Basset Hound Free Thrombopathia; mutation originally found in Landseer Free Thrombopathia; mutation originally found in Eskimo Spitz Free Prekallikrein Deficiency Free Elliptocytosis Free Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd Free Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type II; mutation originally found in Scottish Terrier Free	Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Beagle	Free
Factor IX Deficiency or Haemophilia B; mutation originally found in Airedale Terrier Factor IX Deficiency or Haemophilia B; mutation originally found in German Wirehaired Pointer Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Basenji Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in West Highland White Terrier Unknown Von Willebrand's Disease (vWD) Type II Free Congenital Macrothrombocytopenia; disease-linked SNP originally found in Norfolk and Cairn Terrier Free Thrombopathia; mutation originally found in Basset Hound Free Thrombopathia; mutation originally found in Landseer Free Thrombopathia; mutation originally found in Eskimo Spitz Free Prekallikrein Deficiency Free Elliptocytosis Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type I	Canine Leucocyte Adhesion Deficiency (CLAD), type III	Free
Factor IX Deficiency or Haemophilia B; mutation originally found in German Wirehaired Pointer  Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Basenji  Free Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in West Highland White Terrier  Unknown Von Willebrand's Disease (vWD) Type II  Free Congenital Macrothrombocytopenia; disease-linked SNP originally found in Norfolk and Cairn Terrier  Thrombopathia; mutation originally found in Basset Hound  Free Thrombopathia; mutation originally found in Landseer  Thrombopathia; mutation originally found in Eskimo Spitz  Free Prekallikrein Deficiency  Elliptocytosis  Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd  Free Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer  Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier  Free Von Willebrand's Disease (vWD) Type I Free	Factor IX Deficiency or Haemophilia B; mutation originally found in Rhodesian Ridgeback	Free
Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Basenji Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in West Highland White Terrier Unknown Willebrand's Disease (vWD) Type II Free Congenital Macrothrombocytopenia; disease-linked SNP originally found in Norfolk and Cairn Terrier Free Thrombopathia; mutation originally found in Basset Hound Free Thrombopathia; mutation originally found in Landseer Free Thrombopathia; mutation originally found in Eskimo Spitz Free Prekallikrein Deficiency Free Fliiptocytosis Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type I	Factor IX Deficiency or Haemophilia B; mutation originally found in Airedale Terrier	Free
Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in West Highland White Terrier  Von Willebrand's Disease (vWD) Type II  Congenital Macrothrombocytopenia; disease-linked SNP originally found in Norfolk and Cairn Terrier  Free Thrombopathia; mutation originally found in Basset Hound  Free Thrombopathia; mutation originally found in Landseer  Thrombopathia; mutation originally found in Eskimo Spitz  Free Prekallikrein Deficiency  Elliptocytosis  Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd  Free Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer  Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier  Von Willebrand's Disease (vWD) Type I  Free	Factor IX Deficiency or Haemophilia B; mutation originally found in German Wirehaired Pointer	Free
Von Willebrand's Disease (vWD) Type II  Congenital Macrothrombocytopenia; disease-linked SNP originally found in Norfolk and Cairn Terrier  Free Thrombopathia; mutation originally found in Basset Hound  Free Thrombopathia; mutation originally found in Landseer  Free Thrombopathia; mutation originally found in Eskimo Spitz  Free Prekallikrein Deficiency  Elliptocytosis  Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd  Free Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer  Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier  Von Willebrand's Disease (vWD) Type I  Free	Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Basenji	Free
Congenital Macrothrombocytopenia; disease-linked SNP originally found in Norfolk and Cairn Terrier  Thrombopathia; mutation originally found in Basset Hound  Free Thrombopathia; mutation originally found in Landseer  Thrombopathia; mutation originally found in Eskimo Spitz  Free Prekallikrein Deficiency  Elliptocytosis  Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd  Free Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer  Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier  Free Von Willebrand's Disease (vWD) Type I	Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in West Highland White Terrier	Unknown
Thrombopathia; mutation originally found in Basset Hound Thrombopathia; mutation originally found in Landseer Thrombopathia; mutation originally found in Eskimo Spitz Free Prekallikrein Deficiency Elliptocytosis Free Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd Free Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type I	Von Willebrand's Disease (vWD) Type II	Free
Thrombopathia; mutation originally found in Landseer Thrombopathia; mutation originally found in Eskimo Spitz Free Prekallikrein Deficiency Elliptocytosis Free Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd Free Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type I	Congenital Macrothrombocytopenia; disease-linked SNP originally found in Norfolk and Cairn Terrier	Free
Thrombopathia; mutation originally found in Eskimo Spitz  Prekallikrein Deficiency  Elliptocytosis  Free Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd  Free Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer  Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier  Free Von Willebrand's Disease (vWD) Type I	Thrombopathia; mutation originally found in Basset Hound	Free
Prekallikrein Deficiency  Elliptocytosis Free Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd Free Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type I	Thrombopathia; mutation originally found in Landseer	Free
Elliptocytosis Free Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd Free Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type I	Thrombopathia; mutation originally found in Eskimo Spitz	Free
Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type I Free	Prekallikrein Deficiency	Free
Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer Free Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type I Free	Elliptocytosis	Free
Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier Free Von Willebrand's Disease (vWD) Type I Free	Factor VIII deficiency or Haemophilia A; p.Cys548Tyr mutation originally found in German Shepherd	Free
Von Willebrand's Disease (vWD) Type I	Factor VIII deficiency or Haemophilia A; mutation originally found in Boxer	Free
· · · · · ·	Von Willebrand's Disease (vWD) Type III; mutation originally found in Scottish Terrier	Free
Canine Scott Syndrome, (CSS)	Von Willebrand's Disease (vWD) Type I	Free
	Canine Scott Syndrome, (CSS)	Free

## **Kidney disorders**

Unknown
Free
Free
Free
Unknown
Free
Unknown

#### Other disorders

Malignant Hyperthermia (MH)	Free
Ivermectin sensitivity (MDR1)	Unknown
Narcolepsy; mutation originally found in Dobermann	Free
Primary Ciliary Dyskinesia (PCD)	Free
Congenital Keratoconjuctivitis Sicca and Ichthyosiform Dermatosis (CKCSID) or Dry Eye Curly Coat Syndrome	Free
Gallbladder Mucocele Formation	Unknown
Persistant Mullerian Duct Syndrome (PMDS), mutation originally found in Miniature Schnauzer	Free
Narcolepsy; mutation originally found in Labrador Retriever	Free

Narcolepsy; mutation originally found in Dachshund	Free
Autosomal Recessive Amelogenesis Imperfecta (ARAI)	Free
Hereditary Nasal Parakeratosis (HNPK)	Unknown
Cleft palate; mutation originally found in Nova Scotia Duck Tolling Retriever, reverse assay	Free
Muscular disorders  Muscular Dystrophy, Duchenne type or Golden Retriever Muscular Dystrophy (GRMD)	Free
Myotonia; mutation originally found in Miniature Schnauzer	Free
Cavalier King Charles Spaniel Muscular Dystrophy (CKCS-MD)	Free
Duchenne-like Muscular Dystrophy, Pembroke Welsh Corgi-type	Unknown
Myotubular Myopathy 1 or X-linked Myotubular Myopathy	Free
Centronuclear Myopathy; mutation originally found in Great Dane	Free
Myotonia; mutation originally found in Australian Cattle Dog	Free
Centronuclear Myopathy; mutation originally found in Labrador Retriever	Free
Cerebellar ataxia; mutation originally found in Old English Sheepdog and Gordon Setter	Free
Muscular Hypertrophy (Double Muscling)	Free
Muscular Dystrophy (MDL), Ullrich-type; mutation originally found in Landseer	Free
Duchenne or Dystrophin Muscular Dystrophy, (DMD); mutation originally found in Norfolk Terrier	Free
Immunological disorders	
ARSCID (Autosomal Recessive Severe Combined Immunodeficiency)	Unknown
X-linked Severe Combined Immunodeficiency (XSCID); mutation originally found in Basset Hound	Free
X-linked Severe Combined Immunodeficiency (XSCID); mutation originally found in Cardigan Welsh Corgi	Free
C3 deficiency	Free
Severe Combined Immunodeficiency (SCID); mutation originally found in Frisian Water Dog	Free -
Autosomal Recessive Severe Combined Immunodeficiency (ARSCID)	Free
Myeloperoxidase Deficiency	Free
Skeletal disorders	
Osteogenesis imperfecta (OI) or Brittle Bone Disease; mutation originally found in Dachshund	Free
Chondrodysplasia (dwarfism); mutation originally found in Norwegian Elkhound and Karelian Bear Dog	Free
Skeletal Dysplasia 2 (SD2)	Free
Craniomandibular Osteopathy (CMO)	Free
Hereditary Vitamin D-Resistant Rickets (HVDRR)	Free
Osteogenesis imperfecta; mutation originally found in Golden Retriever	Unknown
Oculoskeletal Dysplasia 2 or Dwarfism-Retinal Dysplasia 2	Free
Osteogenesis imperfecta; mutation originally found in Beagle	Free
Osteochondrodysplasia, mutation originally found in Miniature Poodle	Free
Cleft Palate; Cleft Lip and Palate with Syndactyly; ADAMTS20 gene mutation originally found in Nova Scotia Duck	TolliFree
Spondylocostal Dysostosis	Free
Van den Ende-Gupta Syndrome, (VDEGS)	Free
Skin disorders	
Golden Retriever Ichtyosis	Unknown
Anhidrotic Ectodermal Dysplasia or X-linked Ectodermal Dysplasia (XHED)	Unknown
Musladin-Lueke syndrome (MLS)	Free
Lamellar Ichthyosis (LI)	Unknown
Epidermolysis bullosa, dystrophic	Free
Epidermolytic Hyperkeratosis or Ichthyosis in Norfolk Terrier	Free
Ectodermal dysplasia or Skin Fragility Syndrome (ED-SFS)	Unknown
Lamellar Ichthyosis (LI)	Free
Anhidrotic Ectodermal Dysplasia or X-linked Ectodermal Dysplasia (XHED)	Free
Hereditary Footpad Hyperkeratosis (HFH)	Free
Focal Non-Epidermolytic Palmoplantar Keratoderma (FNEPPK/DH)	Free
Ligneous Membranitis	Free -
Ichthyosis; mutation originally found in Great Dane	Free
Cardiological disorders	
Dilated Cardiomyopathy. DCM SAS Heart disorder only ULTRASOUND for Ridgebacks	Unknown
Endocrine disorders  Hypothyroidisms mutation originally found in Tenterfield Terrior	Fron

Free

Hypothyroidism; mutation originally found in Tenterfield Terrier

Hypothyroidism; mutation originally found in Toy Fox- and Rat Terrier

Free

**Cardiac disorders** 

Long QT Syndrome Free

**Pharmacogenetics** 

Glucocorticoid receptor alpha (NR3C1) gene variant c.2111T>C Unknown

**Teeth disorders** 

Dental Hypomineralization; mutation originally found in Border Collie Free

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P. Schubert, Studbook Manager



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