GeneTech

COMPREHENSIVE

canine DNA screen

ADVANCED GENOMICS TESTING FOR YOUR DOG'S HEALTH

REPORT FOR COCO

ThermoFisher SCIENTIFIC

CANINE TRAITS & DISORDERS PANEL

POWERED BY GeneTech

GeneTech

CANINE DNA SCREEN

Report for

COCO

Basic Details

Breed:	**Labrador**	Sex:	**Male**	Age:	**6 months**
Parents:		Weight:	**25kgs**	Height:	
Known health conditions:					

Sample Details

Reference Id:	**REF21121001**	Collected:	**19/11/2021 5:00PM**	Sample Type:	**Blood (in EDTA vacutainer)**
Lab Ref Id:	**SLCTD01001**	Received:	**13/12/2021 1:00PM**	Reported:	**26/12/2021**

Reference Details

Referring Vet:	**Dr. Jacob Mathew**	Clinic's Name:	**Good shepherd veterinary polyclinic**
Clinic Address:	**# RK puram**		
Owner name:	**Swarup Chatterjee**	Email Address:	**Chatterjee.swarup68@gmail.com**
Owner address:	**Kompally**		

Genomics Test Details

Panel:	**Thermofisher Canine Genomics Panel - Canine Traits and Disorders, Version 1**		
Laboratory:	**GeneTech**	Technology:	**NGS**
Machine:	**Ion GeneStudio S5 & Ion Chef**	LibPrep Solution:	**Ampliseq based AgriSeq**
Panel size:	**154 markers** **97 SNPs, 6 MNPs, 13 Ins, 38 Del**	MarkerCoverage:	**134 Genetic Disorders** **20 Traits**

SUMMARY OF RESULTS

Disorders	0
Carrier	1
Traits	3

DISORDERS IDENTIFIED
CARRIER STATUS IDENTIFIED
Cone Rod Dystrophy 3
TRAITS IDENTIFIED
Coat Color, Agouti
Coat Color, Extension
Coat color, Grizzle
Coat Color, Dominant black

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INTRODUCTION

Dogs are true companions of humans. There are many health issues that can affect dogs starting from common infections and allergies to complex genetic conditions which may set in early or could be late onset. It is vital to be aware of the health risks to the dog and provide care necessary for its healthier life and overall well-being.

About Canine DNA Screen

Canine DNA screen is a genomics test for dogs offered to pet owners and veterinarians. It allows identification of multiple diseases in a single test and makes it possible to profile the pets in advance, andprovide better management and care for the pet. GeneTech's Canine DNA Screen genetic panel is based on Thermofisher's Canine Disorders and Traits Panel which was developed based on extensive research in Canine Genomics and Veterinary Science.

The genomics panel offers screening for diseases recommended by ISAG (International Society for Animal Genetics) and more. The panel identifies selected134 gene variants that are associated with common inherited conditions in different dog breeds along with 20 different variants associated with breed traits related to coat, hair and tail. Next generation sequencing (NGS) is used by the laboratory to identify gene variations or mutations.

Canine DNA Screen is a screening test and further confirmatory tests may be required based on clinical correlation and veterinarian's recommendations.

What is a Genetic Disorder?

A genetic disorder is a health condition caused by an abnormal genetic change usually called as mutation. Although some of these mutations occur spontaneously, a majority of them are inherited and are hereditary. Around 400 hereditary diseases known in canines are caused by mutations in 19,000 genes spread across 78 chromosomes. Genetic disorders eitherresult in multi organ defects and metabolic dysfunction or may alter only one functional system of dog's health such a vision, skin, immune system, nerve and muscular function etc.

These genetic disorders are not uncommon and are not exclusive to pure breeds although they might be extensively reported in few selected breeds.

How is a genetic disorder inherited from a parent to the pup?

Inheritanceis the process by which genetic information is passed on from parent to offspring. Nature provides two copies of gene for all bodily functions of which one copy comes from father and other from mother. Every genetic disorder inherits in a specific manner as given below:

- **Autosomal Dominant:** If a disease is expressed in a dog with one copy of abnormal gene and one copy of normal gene. The risk on passing on such a disease to offspring is 50%.
- **Autosomal Recessive:** If a disease is expressed in a dog only if both copies of the gene are abnormal. Such a pattern is common with 25% risk to offspring if both parents are unaffected carriers with one abnormal gene copy each.
- **X-linked:** The disease is expressed only in male dogs and female dogs are unaffected carriers.
- **Complex**: The inheritance of disease is called complex when it results due to multiple genes and their interaction with environmental factors.

There are some disorders for which the inheritance pattern is unknown and a few with more than one inheritance pattern associated.

How does Canine DNA Screen help?

- Early diagnosis and preventive care are possible before the symptoms set in
- Potential dog owners may choose to adopt a dog based on test result
- Carrier status for diseases can be identified, which allows choice of right parents in breeding programs to reduce disease risk in pups
- Veterinarians can offer evidence based, effective management and treatment to the dog

Introduction to genetic terms used in the report

DNA is the genetic material present in every cell containing a code that controls various functions in your pet's body.

Gene is a unit of DNA with genetic information or code to produce a specific protein or an enzyme in your pet's body to perform a specific function.

Gene Mutations or Gene Variants are specific changes of the genetic code at specific locations on the genes. These change of code result in a protein variation resulting in a health issue.

Genetic Disease or Disorderis a particular abnormal health condition affecting function of a body or apart of dog's body due to a genetic change. Based on the organ system involved, dog's genetic disorders can be one the following types:

- Ophthalmic (Vision problems related to eye)
- Dental (Teeth and denture related)
- Neurological (Disorders of the nervous system)
- Immunological (Conditions with immune dysfunction)
- Endocrine (hormonal or endocrine disorders)
- Skeletal (Bone defects)
- Respiratory (Breathing conditions)
- Hepatological (Liver disorders)
- Dermatological (Skin conditions)
- Metabolic (Abnormal body metabolism or enzyme defects)
- Hematological (Blood tissue related disorders)
- Gastrointestinal (Digestive system defects)
- Muscular (Muscle dysfunction)
- Renal (Kidney and urinary defects)

Genetic Trait is a distinguishing quality or characteristic of the dog determined by genetic factor or change.

Autosomal: Dogs have 78 chromosomes (39 pairs) with 38 pairs are autosomes (non-sex chromosomes) and one pair of sex chromosomes (X and Y).

Inheritance is the process by which genetic information is passed on from parent to offspring. Nature provide two copies of gene for all bodily functions of which one copy comes from father and other from mother.

We have designed this report carefully to provide you with as much information as possible in a simple yet comprehensive fashion. The detailed Table of Contents page guides you to various parts of the report.

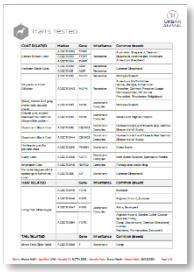
How to read Canine DNA Screen Result?

Canine DNA Screen aims at identification of the following by testing 154 genetic markers:

- Is your dog affected with any of the inherited genetic conditions tested?
- If your dog an unaffected carrier for any of these conditions?
- What are the genetic traits (among those the panel tests) that your dog has inherited?

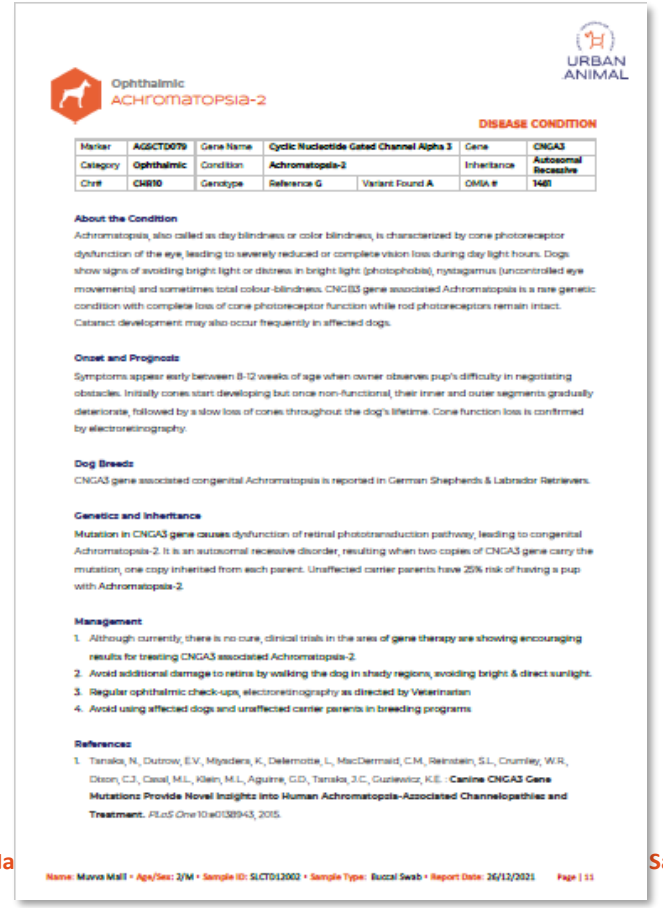
Results Summary Page

Detailed Results Page



List of Traits Tested

List of Disorders Tested



Ophthalmic
ACHROMATOPSIA-2

URBAN ANIMAL

DISEASE CONDITION

Marker	AGSCTD079	Gene Name	Cyclic Nucleotide Gated Channel Alpha 3		Gene	CNGA3
Category	Ophthalmic	Condition	Achromatopsia-2		Inheritance	Autosomal Recessive
Chr#	CHR10	Genotype	Reference G	Variant Found A	OMIA #	1481

About the Condition

Achromatopsia, also called as day blindness or color blindness, is characterized by cone photoreceptor dysfunction of the eye, leading to severely reduced or complete vision loss during day light hours. Dogs show signs of avoiding bright light or distress in bright light (photophobia), nystagamus (uncontrolled eye movements) and sometimes total colour-blindness. CNGB3 gene associated Achromatopsia is a rare genetic condition with complete loss of cone photoreceptor function while rod photoreceptors remain intact. Cataract development may also occur frequently in affected dogs.

Onset and Prognosis

Symptoms appear early between 8-12 weeks of age when owner observes pup's difficulty in negotiating obstacles. Initially cones start developing but once non-functional, their inner and outer segments gradually deteriorate, followed by a slow loss of cones throughout the dog's lifetime. Cone function loss is confirmed by electroretinography.

Dog Breeds

CNGA3 gene associated congenital Achromatopsia is reported in German Shepherds & Labrador Retrievers.

Genetics and Inheritance

Mutation in CNGA3 gene causes dysfunction of retinal phototransduction pathway, leading to congenital Achromatopsia-2. It is an autosomal recessive disorder, resulting when two copies of CNGA3 gene carry the mutation, one copy inherited from each parent. Unaffected carrier parents have 25% risk of having a pup with Achromatopsia-2.

Management

1. Although currently, there is no cure, clinical trials in the area of gene therapy are showing encouraging results for treating CNGA3 associated Achromatopsia-2.
2. Avoid additional damage to retina by walking the dog in shady regions, avoiding bright & direct sunlight.
3. Regular ophthalmic check-ups, electroretinography as directed by Veterinarian
4. Avoid using affected dogs and unaffected carrier parents in breeding programs

References

1. Tanaka, N., Dutrow, E.V., Miyadera, K., Delemotte, L., MacDermaid, C.M., Reinstein, S.L., Crumley, W.R., Dixon, C.J., Casal, M.L., Klein, M.L., Aguirre, G.D., Tanaka, J.C., Cuziewicz, K.E. : **Canine CNGA3 Gene Mutations Provide Novel Insights into Human Achromatopsia-Associated Channelopathies and Treatment.** *PLoS One* 10:e0138943, 2015.

DISORDER DETAILS PAGE

One page for each positive marker identified. Find details below on how to read/use this page.

Marker details table
Provides details about the marker that tested positive along with full gene name.

Onset and Prognosis
Not all genetic diseases express at birth and presentation of symptoms may vary within the breed or across breeds. Progression of the disease may be slow or rapid. This section describes what is known about this condition in this regard.

Dog Breeds
Some diseases are widely reported in certain breeds which are mentioned in this section.

Management
Disease management is provided in a distilled fashion in this section. Preventive measures, dietary management, medical care, and promising treatments in development are covered.

Discuss your pet's report with veterinarian before implementing any of the recommendations provided in this section.

LIMITATIONS OF *CANINE DNA SCREEN* REPORT

Product and Technology Limitations

- The test is limited to the genes and diseases tested in the panel. Genetic disorders associated with genes not included in the panel cannot be diagnosed.
- A genetic disease can be caused by multiple variants of a gene. However, only selected variants (which are commonly reportedfor certain breeds) are included in the panel for testing. In case your pet is clinically showing symptoms of any genetic condition and your Canine DNA Screen report is negative for the disease, please consult our geneticist for available confirmatory options.
- The panel is based on current research. More and more genes and their association with diseases are being discovered. Newer versions of the panel will be released periodically and shall be used. Write to us if you need more information on this.
- Complex polygenic genetic disorders caused by multiple genes are not covered in this test.
- Clinical correlation of the genetic report is strongly recommended.
- Recommendations provided in the report are general. Veterinarian consultation is mandatory.
- Next Generation Sequencing technology was used to generate the data for analysis. Next Generation Sequencing (NGS) is the latest technology for multi-gene sequencing with 99% accuracy and sensitivity.

Summary of results identified

Disorders 0

DISORDERS IDENTIFIED

Type:
Marker:
Gene:
Inheritance:

Carrier 1

CARRIER STATUS IDENTIFIED

Cone Rod dystrophy 3

Type: Ophthalmic
Marker:AGSCTD051
Gene: CNGA3
Inheritance: Autosomal Recessive

Cone Rod dystrophy 3 is an inherited eye disease, characterized by blindness due to degeneration of photoreceptors, cones and rods.

Traits 4

TRAITS IDENTIFIED

Coat Color, Agouti

Marker: AGSCTD011
Gene: ASIP
Inheritance: Autosomal

Coat color, Agouti is an inherited fur coloration displaying two or more bands of pigmentation the overall appearance of agouti fur is usually gray or dull brown and sometimes dull yellow. Agouti protein controls the release of melanin pigments (Eumelanin and Phaeomelanin) into the hair.

Coat Color, Extension

Marker: AGSCTD013
Gene: MC1R
Inheritance: Autosomal Recessive

Coat color extension is an inherited trait. The E (extension) locus is governed by the MC1R gene creates the black facial mask of many dogs as well as yellow or red coats. . A mutation of the MC1R gene can cause a dog's cells to only produce phaeomelanin in place of eumelanin.

Coat Color, Grizzle

Marker: AGSCTD014

Gene: MC1R **Inheritance**: Autosomal Grizzle is a color pattern that appears as mixed hair on the dog with no discernible pattern. The color appears blended together, making it look like just one color until you examine it closely.
Coat Color, Dominant Black
Marker: AGSCTD044 **Gene**: CBD103 **Inheritance**: Autosomal Dominant Coat color black is an inherited trait. The dominant black gene is due to a mutation in a Beta-defensin gene (CBD103). Coat coloration is controlled by several different genes in dogs.

Details for each disorder or trait are provided in following pages.

CARRIER CONDITION

Marker	**AGSCTD051**	Gene Name	ADAM Metallopeptidase Domain 9		Gene	**ADAM9**
Category	**Ophthalmic**	Condition	Cone Rod Dystrophy 3		Inheritance	**Autosomal Recessive**
Chr#	**CHR16**	Genotype	Reference **C**	Variant Found **T**	OMIA #	1520

About the Condition

Code Rod Dystrophy-3, also called progressive retinal atrophy is an inherited genetic disorder characterized by blindness due to degeneration of photoreceptors, cones and rods. The condition starts with degeneration of cone first followed by the rods eventually resulting in complete vision loss. Symptoms of affected dogs include loss of peripheral and general vision, loss of color vision and photophobia. The disease can be diagnosed by opthalmoscopy and electroretinography.

Onset and Prognosis

The age of onset of the condition can be between 7 months to 1 year beginning with peripheral vision loss followed by complete vision loss over several years.

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Dog Breeds

Code Rod Dystrophy-3 is seen reported in Glen of Imaal Terrier

Genetics and Inheritance

The gene ADAM9 codes for a ADAM metallopeptidase domain 9 protein and a non-sense mutation terminates the synthesis of the protein leading to Code Rod Dystrophy-3. It is an autosomal recessive disorder, resulting when two copies of ADAM9 gene carry the mutation, one copy inherited from each parent. Unaffected carrier parents have 25% risk of having a pup with Code Rod Dystrophy-3.

Management

1. Vitamin A and E rich foods recommended
2. Regular ophthalmic checkups, electroretinography as directed by Veterinarian
3. Avoid using affected dogs and unaffected carrier parents in breeding programs

References

1. Goldstein O, Mezey JG, Boyko AR, Gao C, Wang W, Bustamante CD, Anguish LJ, Jordan JA, Pearce-Kelling SE, Aguirre GD, Acland GM. **An ADAM9 mutation in canine cone-rod dystrophy 3 establishes homology with human cone-rod dystrophy 9**. Mol Vis. 2010 Aug 11;16:1549-69.
2. Kropatsch, R., Petrasch-Parwez, E., Seelow, D., Schlichting, A., Gerding, WM., Akkad, DA., Epplen, JT., Dekomien, G **Generalized progressive retinal atrophy in the Irish Glen of Imaal Terrier is associated with a deletion in the ADAM9 gene.** Mol Cell Probes 24:357-63, 2010

TRAIT

Marker	**AGSCTD011**	Gene Name	Aspartic Peptidase, reteroviral like 1		Gene	**ASIP**
Category	**Skin Trait**	Condition	**Coat color, Agouti**		Inheritance	**Autosomal**
Chr#	**CHR 24**	Genotype	Reference TCTCA	Variant Found GCTCG	OMIA #	**000201**

About the Trait

Coat color, Agouti is an inherited fur coloration displaying two or more bands of pigmentation. As a result the overall appearance of agouti fur is usually gray or dull brown and sometimes dull yellow. Agouti protein controls the release of melanin pigments (Eumelanin and Phaeomelanin) into the hair. A multi nucleotide variation leads to presence of display of two or more bands of pigmentation. Coat color is a complex multigene trait.

Dog Breeds

This trait is reported in German shepherd.

Genetics and Inheritance

The gene ASIP (Aspartic Peptidase, retroviral like 1) codes for Agouti-signaling protein which is responsible for the distribution of melanin pigment resulting in display of two or more bands of pigmentation. Inheritance pattern is complex, polygenic and autosomal in nature.

References

1. Dreger, D.L., Schmutz, S.M. : **A SINE insertion causes the black-and-tan and saddle tan phenotypes in domestic dogs.** J Hered :S11-8, 2011.
2. Dreger, D.L., Parker, H.G., Ostrander, E.A., Schmutz, S.M. **Identification of a mutation that is associated with the saddle tan and black-and-tan phenotypes in Basset Hounds and Pembroke Welsh Corgis.** J Hered 104:399-406, 2013.

TRAIT

Marker	**AGSCTD013**	Gene Name	Aspartic Peptidase, reteroviral like 1		Gene	**MC1R**
Category	**Skin Trait**	Condition	**Coat color, Extension**		Inheritance	**Autosomal recessive**
Chr#	**CHR 5**	Genotype	Reference C	Variant Found T	OMIA #	**001199**

About the Trait

Coat color extension is an inherited trait. The E (extension) locus is governed by the MC1R gene creates the black facial mask of many dogs as well as yellow or red coats. MC1R activation prompts the melanocyte to produce eumelanin, whereas MC1R inhibition leads to the production of pheomelanin. A mutation of the MC1R gene can cause a dog's cells to only produce phaeomelanin in place of eumelanin. Mutations in MC1R have been associated with white coloring or partial red coat in several species. Coat color is a complex multigene trait.

Dog Breeds

The trait is reported in Irish Setter, Labrador Retriever, Australian Cattle Dog, Alaskan Husky and Siberian Husky

Genetics and Inheritance

The gene MC1R gene provides instructions for making a protein called the melanocortin 1 receptor. A non sense mutation inhibits protein Melanocortin making the coat color white or dull. Inheritance is not clear with both Autosomal dominant and recessive patterns reported.

References

1. Dürig, N., Letko, A., Lepori, V., Hadji Rasouliha, S., Loechel, R., Kehl, A., Hytönen, M.K., Lohi, H., Mauri, N., Dietrich, J., Wiedmer, M., Drögemüller, M., Jagannathan, V., Schmutz, S.M., Leeb, T. **Two MC1R loss-of-function alleles in cream-coloured Australian Cattle Dogs and white Huskies.** Anim Genet 49:284-290, 2018.
2. Nowacka-Woszuk, J., Salamon, S., Gorna, A., Switonski, M. **Missense polymorphisms in the MC1R gene of the dog, red fox, arctic fox and Chinese raccoon dog.** J Anim Breed Genet 130:136-41, 2013.
3. Newton, J.M., Wilkie, A.L., He, L., Jordan, S.A., Metallinos, D.L., Holmes, N.G., Jackson, I.J., and Barsh, G.S. (2000). **Melanocortin 1 receptor variation in the domestic dog.** Mamm. Genome 11, 24–30.

TRAIT

Marker	**AGSCTD014**	Gene Name	Melanocortin 1 receptor (alpha melanocyte stimulating hormone receptor)		Gene	**MC1R**
Category	**Skin Trait**	Condition	**Coat, Grizzle**		Inheritance	**Autosomal**
Chr#	**CHR 5**	Genotype	Reference **G**	Variant Found **A**	OMIA #	**001199, 001495**

About the trait

Grizzle is a color pattern that appears as mixed hair on the dog with no discernible pattern. The color appears blended together, making it look like just one color until you examine it closely. Grizzle coloring may mix black hair with some tan or brown hairs, or white with black, making it appear grey. Coat color is a complex multigene trait.

Dog Breeds

The trait is reported in Irish Setter, Labrador Retriever, Australian Cattle Dog, Alaskan Husky, Siberian Husky and Afghan Hound

Genetics and Inheritance

The gene MC1R gene provides instructions for melanocortin 1 receptor which plays a significant role in pigmentation of cells. A missense mutation on the gene may result in Grizzle color coat.

References

1. Brancalion, L., Haase, B., Wade, C.M. : **Canine coat pigmentation genetics: a review.** *Anim Genet* :, 2021.
2. Dürig, N., Letko, A., Lepori, V., Hadji Rasouliha, S., Loechel, R., Kehl, A., Hytönen, M.K., Lohi, H., Mauri, N., Dietrich, J., Wiedmer, M., Drögemüller, M., Jagannathan, V., Schmutz, S.M., Leeb, T. **Two MC1R loss-of-function alleles in cream-coloured Australian Cattle Dogs and white Huskies.** Anim Genet 49:284-290, 2018.
3. Dreger, DL., Schmutz, SM. **A new mutation in MC1R explains a coat color phenotype in 2 "Old" breeds: Saluki and Afghan Hound.** J Hered :, 2010.

TRAIT

Marker	**AGSCTD044**	Gene Name	Beta-defensin 103		Gene	**CBD103**
Category	**Skin Trait**	Condition	**Coat color, dominant black**		Inheritance	**Autosomal dominant**
Chr#	**CHR 16**	Genotype	Reference CCC	Variant Found--	OMIA #	**001416**

About the trait

Coat color black is an inherited trait. The dominant black gene is due to a mutation in a Beta-defensin gene (CBD103). Coat coloration is controlled by several different genes in dogs. . The pigment Eumelanin can modify and create other colors such as liver (brown), blue (grey), or isabella (pale brown). Note that coat color is a complex multigene trait.

Dog Breeds

Coat color black is reported in almost all dog breeds.

Genetics and Inheritance

The gene CBD103 plays a role in pigmentation of cells and a 3 base pair deletion results in black coat. The trait follows autosomal dominant pattern with only one allele with mutation enough to express the trait.

References

1. Brancalion, L., Haase, B., Wade, C.M. : **Canine coat pigmentation genetics: a review.** Anim Genet :, 2021.
2. Candille, SI., Kaelin, CB., Cattanach, BM., Yu, B., Thompson, DA., Nix, MA., Kerns, JA., Schmutz, SM., Millhauser, GL., Barsh, GS. **A {beta}-Defensin Mutation Causes Black Coat Color in Domestic Dogs.** Science 318:1418-23, 2007. Pubmed reference: 17947548 DOI:10.1126/science.1147880

Markers for Disorders &Traits Tested

Traits **20**

TRAIT RELATED MARKERS TESTED	
Coat related traits	15
Hair related traits	4
Tail related traits	1

Disorders **134**

DISORDERS RELATED MARKERS TESTED	
Ophthalmic disorders	23
Dental and Oral disorders	2
Neurological disorders	30
Immunological disorders	6
Endocrine disorders	3
Skeletal disorders	9
Respiratory disorders	1
Hepatological disorders	2
Dermatological disorders	8
Metabolic disorders	15
Hematological disorders	20
Gastrointestinal disorders	3
Muscular disorders	8
Nephrological disorders	4

COAT RELATED	Marker	Gene	Inheritance	Common Breeds
Classic Brown Coat	AGSCTD002	TYRP1	Recessive	Australian Shepherd, German Shepherd, Leonberger, Miniature American Shepherd
	AGSCTD130	TYRP1		
	AGSCTD134	TYRP1		
Uniform Black Coat	AGSCTD010	ASIP	Recessive	German Shepherd
	AGSCTD011	ASIP	Recessive	
Alopecia or Color Dilution	AGSCTD012	MLPH	Recessive	Multiple Breeds
	AGSCTD043	MLPH	Recessive	American Staffordshire Terrier, Beagle, Doberman Pinscher, German Pinscher, Large Munsterlander, Miniature Pinnscher, Rhodesian Ridgeback
Black, brown and grey melanistic muzzle mask	AGSCTD013	MC1R	Dominant Complex	Multiple Breeds
Grizzle coat, mixed blended hair with no discernible pattern	AGSCTD014	MC1R	Dominant Complex	Saluki and Afghan Hound
Dominant Black Coat	AGSCTD015	CBD103	Dominant Complex	Domesticated wolf breeds like German Shepard and Husky
Dominant Black Coat	AGSCTD044	CBD103	Dominant Complex	Domesticated wolf breeds like German Shepard and Husky
Harlequin patchy pattern coat	AGSCTD029	PSMB7	Dominant	Great Dane
Curly Coat	AGSCTD036	KRT71	Dominant Complex	Irish Water Spaniel, Standard Poodle
Improper Coat	AGSCTD048	RSPO2	Complex	Portuguese water dog
Variable degree white spotting to full white coat	AGSCTD153	KIT	Dominant	German Shepherd
HAIR RELATED		**Gene**	**Inheritance**	**Common Breeds**
Long Hair Phenotype	AGSCTD045	FGF5	Dominant Complex	Eurasier
	AGSCTD046	FGF5		Afghan Hound, Eurasier
	AGSCTD047	FGF5		Akita, Samoyed
	AGSCTD084	FGF5		Afghan Hound, Border Collie, Cocker Spaniel, Collie, Corgi, Dachshund, German Shepherd, Golden Retriever, Pomeranian, Samoyed
TAIL RELATED		**Gene**	**Inheritance**	**Common Breeds**
Short Tails (Bob Tails)	AGSCTD006	T	Dominant	Corgi

AGSCTD001	**Progressive Retinal Atrophy**	CNGB1	Autosomal Recessive
	Progressive retinal atrophy is an inherited condition primarily affecting the dog's vision. Starting with degeneration of rod photoreceptors, leading to loss of night-vision, the disease eventually leads to complete blindness.		
AGSCTD027	**Multifocal Retinopathy 1**	BEST1	Autosomal Recessive
	Multifocal Retinopathy is characterized by fluid accumulation under the detached retina resulting in gray, tan, orange or pink "blisters" in the eye. As, the progression is slow, lesions appear to heal with little or no vision loss due to this condition.		
AGSCTD028	**Multifocal Retinopathy 2**	BEST1	Autosomal Recessive
	Multifocal Retinopathy is characterized by fluid accumulation under the detached retina resulting in gray, tan, orange or pink "blisters" in the eye. As, the progression is slow, lesions appear to heal with little or no vision loss due to this condition.		
AGSCTD033	**Cone Rod Dystrophy 1**	PDE6B	Autosomal Recessive
	Cone Rod Dystrophy is an inherited eye disorder affecting the rod and cone photoreceptors resulting in visual acuity followed by severe loss of central and color vision that often progresses to blindness due to retinal degeneration.		
AGSCTD034	**Cone Rod Dystrophy 2**	IQCB1	Autosomal Recessive
	Cone Rod Dystrophy is an inherited eye disorder affecting the rod and cone photoreceptors resulting in visual acuity followed by severe loss of central and color vision that often progresses to blindness due to retinal degeneration.		
AGSCTD035	**Congenital Stationary Night Blindness**	RPE65	Autosomal Recessive
	Congenital Stationary Night Blindness is a slow, progressive, retinal degenerative disorder which is characterized by loss of night vision, progressing to low light and also possibly day vision. Onset in early.		
AGSCTD049	**Multifocal Retinopathy 3**	BEST1	Autosomal Recessive
	Canine multifocal retinopathy is characterized by multiple areas of retinal degeneration along with generalized retinal degeneration and affected dogs may exhibit acquired post-inflammatory changes, and fundus changes		
AGSCTD050	**Multifocal Retinopathy 3**	BEST1	Autosomal Recessive
	Canine multifocal retinopathy is characterized by multiple areas of retinal degeneration along with generalized retinal degeneration and affected dogs may exhibit acquired post-inflammatory changes, and fundus changes		
AGSCTD051	**Cone Rod Dystrophy 3**	ADAM9	Autosomal Recessive
	Cone Rod Dystrophy is an inherited eye disorder affecting the rod and cone photoreceptors resulting in visual acuity followed by severe loss of central and color vision that often progresses to blindness due to retinal degeneration.		
AGSCTD061	**Rod Cone Dysplasia 3**	PDE6A	Autosomal Recessive
	Rod cone dysplasia 3 is characterized by progressive retina atrophy including changes in reflectivity and appearance of a structure behind the retina resulting in loss of peripheral vision and night vision.		
AGSCTD066	**Primary Open Angle Glaucoma**	ADAMTS10	Autosomal Recessive
	Primary Open Angle Glaucoma is a genetic condition where microfibrils of eye are affected leading to increased pressure in the eye. This leads to slow loss of vision with eye pain, watery discharge, lethargy, loss of appetite, with swelling and bulging of the eyeball.		

DISORDERS -OPHTHALMIC (CONTD.)

AGSCTD071	**Golden Retriever Progressive Retinal Atophy 2**	TTC8	Autosomal Recessive
	Progressive retinal atrophy is an inherited disease, characterized by late onset vision loss. The first sign is loss of vision in dim light that worsens progressively and culminates in total blindness. The symptoms include night blindness.		
AGSCTD073	**Progressive Retinal Atrophy**	CNGB1	Autosomal Recessive
	Progressive retinal atrophy is an inherited disease, characterized by late onset vision loss. The first sign is loss of vision in dim light that worsens progressively and culminates in total blindness. The symptoms include night blindness. .		
AGSCTD079	**Achromatopsia 2**	CNGA3	Autosomal Recessive
	Achromatopsia 2 is an inherited eye disease, characterized by early onset loss of cone photo-receptor function resulting in day blindness, total color blindness, decreased central visual acuity.		
AGSCTD080	**Achromatopsia (hemeralopia), AMAL**	CNGB3	Autosomal Recessive
	Achromatopsia, AMAL type is an inherited eye disease, characterized by the loss of cone photo-receptor function resulting in day blindness, total color blindness, decreased central visual acuity. Onset varies from early to late in different breeds.		
AGSCTD081	**Progressive Retinal Atrophy CNGA1-related**	CNGA1	Autosomal Recessive
	Progressive Retinal Atrophy is an inherited, late onset, eye disorder, characterized by vision loss. The first sign is loss of vision in dim light that worsens progressively and culminates in blindness. The symptoms include night blindness.		
AGSCTD095	**Macular Corneal Dystrophy**	LOC489707	Autosomal Recessive
	Macular Corneal Dystrophy is an inherited eye disease, characterized severe visual impairment in affected dogs. The symptoms include watery eyes, sensitivity to light glare, pain in the eye and corneal erosion.		
AGSCTD123	**Lens luxation**	ADAMTS17	Autosomal Recessive
	Lens luxation is an inherited disease, characterized by breakdown of the lens zonules in the eye of the puppy. Symptoms include pain in the eye, increased tears, inflammation, cloudiness or redness of the eye.		
AGSCTD125	**Glaucoma**	ADAMTS17	Autosomal Recessive
	Characterized by vision loss, Glaucoma is a late onset disease caused by increased intra ocular pressure. The symptoms include increase blurred vision, blue color Sclera, cloudy cornea, red eye, squinting and weak blink response.		
AGSCTD127	**Polyneuropathy, NDGR1- related**	NDRG1	Autosomal Recessive
	Degenerative Polyneuropathy results in slow wasting of muscles, exercise intolerance, gait abnormalities, ambulatory paraparesis, and difficulty in breathing due to involvement of the larynx and laryngeal folds in the throat.		
AGSCTD128	**Progressive Retinal Atrophy Basenji**	SAG	Autosomal Recessive
	Characterized by vision loss, the disease has late onset and has been described in Basenji breeds. Starting with loss of vision in dim light, the condition has slow progression and culminates in total blindness.		
AGSCTD131	**Rod Cone Dysplasia 1a**	PDE6B	Autosomal Recessive
	Rod Cone Dysplasia is characterized by early onset loss of vision loss, which becomes more severe over time. These disorders affect the retina causing night blindness, loss of visual acuity, constriction of visual fields and complete blindness		
AGSCTD132	**Rod Cone Dysplasia 1a**	PDE6B	Autosomal Recessive
	Rod Cone Dysplasia is characterized by early onset loss of vision loss, which becomes more severe over time. These disorders affect the retina causing night blindness, loss of visual acuity, constriction of visual fields and complete blindness.		

AGSCTD003	**Amelogenesis Imperfecta**	ENAM	Autosomal Recessive
	Amelogenesis Imperfecta is a common non-syndromic genetic condition caused by malfunction of enamel proteins, resulting in enamel hypoplasia or thinning, small and pointed teeth, rough surface with brown color, and greater gaps between teeth.		
AGSCTD041	**Hypomineralisation**	FAM20C	Autosomal Recessive
	Dental hypomineralization, also known as Raine Syndrome is an inherited dental disorder characterized by extensive wear of teeth, cracking of tooth enamel, brownish spots or brownish discoloration of teeth or pulpitis		

AGSCTD004	**Cerebellar ataxia**	SEL1L	Autosomal Recessive
	Cerebellar ataxia causes cerebellar shrinkage, leading to loss of balance and uncoordinated movements along with tremors, dizziness, hearing loss, weakening of limbs, head tilting, nystagmus, nausea and loss of appetite.		
AGSCTD005	**Benign Familial Juvenile Epilepsy**	LGI2	Autosomal Recessive
	Benign Familial Juvenile Epilepsy or focal epilepsy is a genetic disorder, characterized by the recurrent seizures in dogs with facial twitches, rhythmic jerks, hypersalivation, restlessness or behavioral changes with varying degree of severity.		
AGSCTD008	**Neuronal Ceroid Lipofuscinosis, 4A**	ARSG	Autosomal Recessive
	Neuronal Ceroid Lipofuscinosis is a severe neurological disorder with late-onset and slowly progressive cognitive and motor degeneration resulting in impaired vision, seizures, aggressive behavior, and increased sensitivity to loud noises.		
AGSCTD009	**Ataxia, Cerebellar, ATP1B2-related**	ATP1B2	Autosomal Recessive
	Ataxia is characterized by uncoordinated movements, loss of coordination and generalized ataxic gait starting at 4 weeks of age with seizures, showed pacing and circling and developed central blindness in affected dogs		
AGSCTD016	**Achromatopsia**	CNGB3	Autosomal Recessive
	Achromatopsia is an inherited eye disorder, characterized by cone photoreceptor dysfunction, leading to severely reduced or complete vision loss during daylight hours, photophobia, nystagamus and total colour-blindness.		
AGSCTD017	**Cerebellar Hypoplasia**	VLDLR	Autosomal Recessive
	Cerebellar hypoplasia, is non-progressive form of neurological condition resulting in inadequate development of cerebellum with poor motor skills, clumsiness, frequent falling, wide stance, tremors and vision problems.		
AGSCTD018	**Cerebellar Abiotrophy**	MUTYH	Unknown
	Cerebellar abiotrophyAlso known as neonatal cerebellar cortical degeneration (NCCD) is a neurodegenerative disorder affecting the cerebellum. Inheritance pattern is unclear.		
AGSCTD019	**Ataxia, cerebellar, ATP1B2-related**	ATP1B2 SDCA1	Autosomal Recessive
	Cerebellar Ataxia is an inherited neurological disease, characterized by degeneration of cerebellum. The symptoms include ataxia, seizures, stumbling, staggering, tremors, hopping, falling and growth impairment.		
AGSCTD020	**Cerebellar Cortical Degeneration**	SNX14	Autosomal Recessive
	Cerebellar Cortical Degenerationis an inherited neurological condition, disrupting synaptic transmission and neuronal excitability with symptoms of dysmetric ataxia, marked truncal sway, tremors with rapid progression of the disease after onset.		
AGSCTD024	**Neuronal Ceroid Lipofuscinosis 5**	CLN5	Autosomal Recessive
	The neuronal ceroid lipofuscinoses (NCLs) are a group of lysosomal storage diseases characterized by intraneuronal accumulation of fluorescent granules and early neuronal death.		

DISORDERS -NEUROLOGICAL(CONTD.)

AGSCTD052	**Degenerative myelopathy**	SOD1	Autosomal Recessive
	Degenerative myelopathy, also known aschronic degenerative radiculomyelopathy**,** is an inherited disease affecting the spinal cord, resulting in slowly progressive hind limb weakness and paralysis.		
AGSCTD053	**Gangliosidosis GM1**	GLB1	Autosomal Recessive
	GM1 gangliosidosis is a lysosomal storage disease caused by beta galactosidase deficiency and characterized by progressive neurological deteriorationincluding weight loss, ataxia, abnormal gait, tremors, strabismus and positional nystagmus.		
AGSCTD054	**Gangliosidosis GM1**	GLB1	Autosomal Recessive
	GM1 gangliosidosis is a lysosomal storage disease caused by beta galactosidase deficiency and characterized by progressive neurological deteriorationincluding weight loss, ataxia, abnormal gait, tremors, strabismus and positional nystagmus.		
AGSCTD059	**Neuronal Ceroid Lipofuscinosis, 12**	ATP13A2	Autosomal Recessive
	Neuronal Ceroid Lipofuscinosis 12 is an inherited disease, characterized by degeneration of Central Nervous System with affected dogs showing loss of vision, behavioral changes, cerebellar ataxia, tremors, and decline of cognitive and motor functions.		
AGSCTD060	**Neuronal Ceroid Lipofuscinosis, 6**	CLN6	Autosomal Recessive
	Neuronal Ceroid Lipofuscinosis 6 is an inherited disease, characterized by degeneration of Central Nervous System with affected dogs showing loss of vision, behavioral changes, cerebellar ataxia, tremors, and decline of cognitive and motor functions.		
AGSCTD062	**Spongy Degenerative Cerebellar Ataxia**	KCNJ10	Autosomal Recessive
	Spongy Degenerative Cerebellar Ataxia, is an early onset inherited neurodegenerative disorder characterized by ataxiac gait, lack of co-ordination, poor balance, seizures, accompanied by stumbling, staggering, tremors, hopping, and falling.		
AGSCTD065	**Krabbe Disease**	GALC	Autosomal Recessive
	Krabbe Disease is an inherited neurological disorder with clinical signs of cerebellar ataxia/intention tremor, postural reaction deficit, stiffness, spastic paresis/paralysis, hearing loss, vision loss, motor and sensory neuropathy,		
AGSCTD075	**Polyneuropathy, ARHGEF10 related**	ARHGEF10	Autosomal Recessive
	Polyneuropathy is an inherited neurological disease, with symptoms including change in gait, loss of bark, limb weakness, tremors, lack of coordination, difficulty in swallowing and atrophy.		
AGSCTD090	**Polyneuropathy, ARHGEF10 related**	FNIP2	Autosomal Recessive
	Polyneuropathy is an inherited neurological disease, with symptoms including change in gait, loss of bark, limb weakness, tremors, lack of coordination, difficulty in swallowing and atrophy.		
AGSCTD093	**Polyneuropathy, RAB3GAP1-related**	RAB3GAP1	Autosomal Recessive
	Polyneuropathy is an inherited, early onset neurological disease, characterized by dilated myelin sheaths of nerve cells. Symptoms include visual problems, laryngeal paralysis, regurgitation, gait abnormalities, ataxia and visual defects.		

DISORDERS -NEUROLOGICAL(CONTD.)

AGSCTD107	**Necrolapsy**	HCRTR2	Autosomal Recessive
	Necrolapsy is an inherited neurological disorder, characterized by abnormal nervous system. Affected dogs may exhibit sudden collapse and loss of movement with recovery in some time, excessive daytime sleepiness and sleep paralysis		
AGSCTD108	**Necrolapsy**	HCRTR2	Autosomal Recessive
	Necrolapsy is an inherited neurological disorder, characterized by abnormal nervous system. Affected dogs may exhibit sudden collapse and loss of movement with recovery in some time, excessive daytime sleepiness and sleep paralysis		
AGSCTD109	**Neuronal Ceroid Lipofuscinosis-1**	PPT1	Autosomal Recessive
	Neuronal Ceroid Lipofuscinosis, 1 is an inherited neurological disorder, characterized by progressive degeneration of central nervous system. Common symptoms include partial or total vision loss, behavior changes, abnormal gait, and seizures		
AGSCTD110	**Neuronal Ceroid Lipofuscinosis-10**	CTSD	Autosomal Recessive
	Neuronal Ceroid Lipofuscinosis, 10 is an inherited neurological disorder, characterized by progressive degeneration of central nervous system. Common symptoms include partial or total vision loss, behavior changes, abnormal gait, and seizures.		
AGSCTD111	**Neuronal Ceroid Lipofuscinosis-8**	CLN8	Autosomal Recessive
	Neuronal Ceroid Lipofuscinosis, 8 is an inherited neurological disorder, characterized by progressive degeneration of central nervous system. Common symptoms include partial or total vision loss, behavior changes, abnormal gait, and seizures.		
AGSCTD112	**Neonatal encephalopathy with seizures**	ATF2	Autosomal Recessive
	Neonatal encephalopathy with seizures is a serious inherited progressive brain disorder, characterized by weakness, mobility issues, and seizures in affected pups with uncoordinated movements and mental dullness.		
AGSCTD114	**Neuroaxonal dystrophy**	MFN2	Autosomal Recessive
	Neuroaxonal dystrophy is a neurological genetic disorder, characterized by progressive degeneration of the nerve cells. Symptoms include high stepping gait, poor coordination, stumbling, head tremors, incontinence, Vit E deficiency and secondary pneumonia.		
AGSCTD124	**Polyneuropathy**	NDRG1	Autosomal Recessive
	Degenerative Polyneuropathy results in slow wasting of muscles, exercise intolerance, gait abnormalities, ambulatory paraparesis, and difficulty in breathing due to involvement of the larynx and laryngeal folds in the throat.		
AGSCTD139	**Ataxia, Spinocerebellar, CAPN1-related**	CAPN1	Autosomal Recessive
	Spinocerebellar Ataxia is an inherited disease characterizedby abnormal coordination. The symptoms include ataxia, uncoordinated walk with stilted "toy soldier" leg movements, involuntary eye movement and growth impairment.		
AGSCTD147	**Tremors X-linked**	PLP1	X-linked Recessive
	Tremors X-linked is an inherited disorder characterized loss of coordination between brain and body. The male puppies have difficulties standing, eating and moving and show uncoordinated gait, leading to scuffing or dragging of the paws.		

AGSCTD007	**C3 Deficiency**	C3	Autosomal Recessive
	C3 deficiency results in recurrent bacterial infections such as pneumonia, urinary tract infections and uterine infections due to compromised immunity wuth increased risk for muscle disease and glomerulonephropathy, a kidney disease.		
AGSCTD025	**Canine Leukocyte Adhesion Deficiency**	ITGB2	Autosomal Recessive
	Canine leukocyte adhesion deficiency is a primary immunodeficiency disorder characterized by recurrent bacterial infections in the presence of marked leukocytosis, impaired wound healing, fever, gingivitis, lameness, and enlarged lymph nodes.		
AGSCTD026	**Canine Leukocyte Adhesion Deficiency**	FERMT3	Autosomal Recessive
	Canine leukocyte adhesion deficiency is a primary immunodeficiency disorder characterized by recurrent bacterial infections in the presence of marked leukocytosis, impaired wound healing, fever, gingivitis, lameness, and enlarged lymph nodes.		
AGSCTD135	**Severe combined immunodeficiency**	RAG1	Autosomal Recessive
	Severe combined immunodeficiencyautosomal, T-cell negative, B-cell negative, NK-positive is an immunological defect characterized by recurrent infections (Oral and respiratory), low immunity, chronic diarrhea and failure to thrive.		
AGSCTD137	**Severe combined immunodeficiency**	PRKDC	Autosomal Recessive
	Severe combined immunodeficiencyautosomal, is an immunological defect characterized by recurrent infections (Oral and respiratory), low immunity, chronic diarrhea, reduced levels of immunoglobulins and lymphocytes and failure to thrive.		
AGSCTD154	**Severe combined immunodeficiency**	IL2RG	X-linked Recessive
	Severe combined immunodeficiency X-linked is an inherited disorder characterized by inability to fight recurrent systemic or localized infections of the eye, ear or respiratory system showing symptoms of chronic diarrhea, serious infections and failure to thrive.		

DISORDERS -ENDOCRINE

AGSCTD021	**Hypothyroidism**	TPO	Autosomal Recessive
	Hypothyroidism or deficiency in thyroid hormone, has early onset with varied clinical including dwarfism, mental retardation, skeletal development abnormalities, Lethargy, protruding eyes, constipation, tremors and spasticity.		
AGSCTD058	**Persistent Mullerian Duct Syndrome**	AMHR2	Autosomal Recessive
	PMDS is a type of XY disorder of sexual development (XY DSD), characterized by the presence of Müllerian duct derivatives in otherwise normal males with 50% of affected dogs are unilaterally or bilaterally cryptorchid		
AGSCTD083	**Dwarfism, Pituitary**	LHX3	Autosomal Recessive
	Dwarfism, Pituitary symptoms include body being longer than normal, legs shorter, bulging eyes, swollen abdomen, tongue sticking out and High-pitched puppy bark with respiratory and coordination issues.		

AGSCTD022	**Chondrodysplasia**	ITGA10	Autosomal Recessive
	Chondrodysplais is an inherited skeletal disorder associated with abnormalities in the development of cartilage tissues and symptoms include small stature, disproportionately short arms and legs, shortness of fingers and toes, broad short hands and feet.		
AGSCTD032	**Craniomandibular osteopathy**	SLC37A1	Autosomal Recessive
	Craniomandibular osteopathy is an early onset skeletal genetic disease, characterized by abnormalities in jaws of dogs. The symptoms are Loss of appetite, bulging eyes, Jaw swelling, difficulty opening mouth and difficulty picking up food with mouth.		
AGSCTD076	**Osteogenesis Imperfecta, type III, COL1A1-related**	COL1A1	Autosomal Recessive
	Osteogenesis Imperfecta is an inherited skeletal disorder, characterized by fragile bones. The symptoms include spontaneous fracturing of the bones and teeth, loose joints, difficulty walking, pain, osteopenia and stunted growth.		
AGSCTD102	**Musladin-lueke syndrome**	ADAMTSL2	Autosomal Recessive
	Musladin-lueke syndrome is a congenital genetic defect characterized by defects in skeleton, heart, skin, and muscle. Dogs exhibit short outer toes, high set creased ears, flat skull, slant narrowed eyes, stiff gait, and seizures Severity is mild, moderate to severe.		
AGSCTD115	**Osteogenesis imperfecta**	COL1A2	Autosomal Recessive
	Osteogenesis imperfecta is an inherited disease, characterized by fragile bones (soft bones). Affected dogs show spontaneous and frequent fractures of bone and teeth, stunned growth, difficulty in walking, pain and hearing loss.		
AGSCTD116	**Osteogenesis imperfecta Dachshund**	SERPINH1	Autosomal Recessive
	Osteogenesis imperfecta is an inherited disease, characterized by fragile bones (soft bones). Affected dogs show spontaneous and frequent fractures of bone and teeth, stunned growth, difficulty in walking, pain and hearing loss.		
AGSCTD136	**Skeletal dysplasia (SD2)**	COL1A1	Autosomal Recessive
	This is an inherited disease, characterized by dwarfism. Affecetd dogs have shoetened limbs but with normal body length and width. Radiological findings typically include shortened and sometimes slightly curved long bones with front legs more affected than hind.		
AGSCTD138	**Brachycephaly**	SMOC2	Multifactorial
	Brachycephalic dogs tend to have extremely shortened snouts that make them almost appear flat-faced and dogs with brachycephaly have a history of loud snoring and noisy breathing.		
AGSCTD148	**Vitamin D Deficiency, Rickets Type II**	VDR	Autosomal Recessive
	Vitamin D-deficency, rickets type II is an inherited disorder with end-organ resistance to the active Vitamin D hormone. The disorder is characterized by hypocalcemia, secondary hyperparathyroidism, hypomineralization of bones rickets and in some cases alopecia.		

AGSCTD023	**Primary ciliary dyskinesia**	CCDC39	Autosomal Recessive
	Primary Ciliary Dyskinesia results in early onset progressively respiratory distress with nasal discharge, chronic sneezing, coughing, exercise intolerance, respiratory distress, cyanosis and infertility in male. Acute bronchopneumonia may occur.		

AGSCTD030	**Menkes Disease**	ATP7A	X-linked Recessive
	Menkes disease is an inherited, fatal, neurodegenerative disorder of copper deficiency with accumulation of copper and characterized by liver and brain degeneration, connective tissue abnormalities, coarse hair and failure to thrive.		
AGSCTD031	**Wilson Disease**	ATP7B	Autosomal Recessive
	Wilson disease, an inherited genetic disorder associated with copper accumulation resulting in hepatic cirrhosis and neuronal degeneration with fatigue, lack of appetite, jaundice, speech, poor coordination and muscle stiffness.		

DISORDERS -DERMATOLOGICAL

AGSCTD057	**Ichthyosis, SLC27A4-related**	SLC27A4	Autosomal Recessive
	Ichthyosis is a rare congenital hereditary disorderof skin, characterized by hyperkeratoses of different severity levels characterized by dry, wrinkled, thickened skin especially in the region of the eyes and nose.		
AGSCTD074	**Ichthyosis, SLC27A4-related**	SLC27A4	Autosomal Recessive
	Icthyosis is an inherited disease, characterized by abnormal lesions on skin. The symptoms include scaly skin, thickening of the skin and footpads, thick, greasy flakes/scales and dandruff. Severity ranges from moderate to severe.		
AGSCTD077	**Ichthyosis, ASPRV1-related**	ASPRV1	Autosomal Recessive
	Icthyosis is an inherited disease, characterized by abnormal lesions on skin. The symptoms include scaly skin, thickening of the skin and footpads, thick, greasy flakes/scales and dandruff. Severity ranges from moderate to severe.		
AGSCTD078	**Ectodermal Dysplasia**	FOX13	X-Linked Recessive
	Ectodermal Dysplasia is an inherited skin condition, with lack of hair on the forehead and back near the tail. Symptoms include abnormal nails, decreased skin color, large forehead, low nasal bridge, sparse hair and learning disabilities and frequent eye infections.		
AGSCTD088	**Hyperkeratosis,Palmoplantar**	FAM83G	Autosomal Recessive
	Hyperkeratosis,Palmoplantar is an inherited skin disease, characterized by early onset, abnormal scaling of skin, horny protrusions on the rims of the footpads, hard pad surface, cracks, hard nails resulting in discomfort while walking.		
AGSCTD089	**Hyperkeratosis, Epidermolytic**	KRT10	Autosomal Recessive
	Epidermolytic Hyperkeratosis is an inherited skin disorder, characterized by abnormal scaling of skin with display of sloughing and blistering of the skin with rubbing. The footpads, claws, teeth and hair of affected dogs are typically normal.		
AGSCTD133	**Epidermolysis Bullosa, Dystrophic**	COL7A1	Autosomal Recessive
	Epidermolysis Bullosa, Dystrophic is a genetic disordercharacterized by extreme fragility of the skin and mucosa, leading to unremitting blisters and erosion, bleeding in mouth and upper digestive system and defective wound healing.		
AGSCTD140	**Ichtyyosis Golden Retriever, TGM1-related**	TGM1	Autosomal Recessive
	Ichthyosis is a rare congenital hereditary disorder of skin, characterized by hyperkeratoses of different severity levels characterized by dry, wrinkled, thickened skin especially in the region of the eyes and nose.		

AGSCTD037	**Cystinuria Type I-A**	SLC3A1	Autosomal Recessive
	Cystinuria Type I-A is an inherited metabolic disorder that affects kidney with increased urinary calculi formation, blockage of the urinary tract, inflammation of the bladder, blood in the urine, renal colic and kidney failure		
AGSCTD038	**Cystinuria Type II-A**	SLC3A1	Autosomal Dominant
	Cystinuria Type II-A is an inherited metabolic disorder that affects kidney with increased urinary calculi formation, blockage of the urinary tract, inflammation of the bladder, blood in the urine, renal colic and kidney failure		
AGSCTD039	**Cystinuria Type II-A**	SLC3A1	Autosomal Dominant
	Cystinuria Type II-A is an inherited metabolic disorder that affects kidney with increased urinary calculi formation, blockage of the urinary tract, inflammation of the bladder, blood in the urine, renal colic and kidney failure		
AGSCTD040	**Cystinuria Type II-B**	SLC7A9	Autosomal Dominant
	Cystinuria Type I-A is an inherited metabolic disorder that affects kidney with increased urinary calculi formation, blockage of the urinary tract, inflammation of the bladder, blood in the urine, renal colic and kidney failure		
AGSCTD055	**Glycogen Storage Disease**	GAA	Autosomal Recessive
	Glycogen storage diseases (GSD) is a severe inherited disorder with defective carbohydrate metabolism, leading to accumulation of glycogen in tissues, resulting in vomiting, progressive muscular weakness, heart disease and myocardial hypertrophy.		
AGSCTD067	**Glycogen Storage Disease-1a**	G6PC	Autosomal Recessive
	Glycogen Storage Disease-1a is a metabolic disorder characterized by inability to convert glucose-6-phosphate to glucose, resulting in weakness, chronic low blood sugar, lethargy, enlarged liver and anorexia with varied severity.		
AGSCTD068	**Gangliosidosis, GM1**	GLB1	Autosomal Recessive
	Gangliosidosis is an inherited metabolic disease due to defective carbohydrate metabolism. The symptoms include vision loss, difficulties walking, loss of balance, head tremors, lethargy and weight loss.		
AGSCTD069	**Gangliosidosis, GM2, type II**	HEXB	Autosomal Recessive
	Glycogen Storage Disease-11 is an early onset, inherited metabolic disease with, characterized by partial or total vision loss, behavior changes, abnormal gait, and seizures.		
AGSCTD072	**Glycogen Storage Disease IIIa**	AGL	Autosomal Recessive
	Glycogen storage disease IIIa is an inherited disease, characterized by defective glycogen metabolism. Affected dog may not grow fast enough, and may have heat intolerance, bruising, hypoglycemia, enlarged liver, swollen belly and weak muscle tone.		
AGSCTD094	**L-2-Hydroxyglutaric Academia**	L2HGDH	Autosomal Recessive
	L-2-Hydroxyglutaric Academia is an inherited neurological and metabolic disease, characterized by early onset neurological traits such as psychomotor impairment, seizures, ataxia, dementia, and tremors.		
AGSCTD098	**Mucopolysaccharidosis VII**	GUSB	Autosomal Recessive
	Mucopolysaccharidosis VII is an inherited disease, characterized by skeletal abnormalities, retarded development, excessively lax joints, broad chests, low set ears, short muzzle, broad face, crooked legs with difficulty standing and walking difficulties.		

DISORDERS -METABOLIC(CONTD.)

AGSCTD099	**Mucopolysaccharidosis IIIa**	SGSH	Autosomal Recessive
	Mucopolysaccharidosis IIIa is an inherited disease, characterized by lysosomal accumulation and urinary excretion of heparan sulfate. The symptoms include growth retardation, skeletal deformities, corneal cloudiness, facial dysmorphia and neurological problems.		
AGSCTD100	**Mucopolysaccharidosis VII**	GUSB	Autosomal Recessive
	Mucopolysaccharidosis VII is an inherited disease, characterized by skeletal abnormalities, retarded development, excessively lax joints, broad chests, low set ears, short muzzle, broad face, crooked legs with difficulty standing and walking difficulties.		
AGSCTD118	**Pyruvate Dehydrogenase Deficiency**	PDP1	Autosomal Recessive
	Pyruvate Dehydrogenase Deficiencyleads to failure of the expelling waste products from the body efficiently. Affected dogs may show symptoms of nausea, vomiting, severe breathing problems, and an abnormal heartbeat.		
AGSCTD119	**Glycogen storage disease VII**	PFKM	Autosomal Recessive
	Glycogen storage disease VII is an inherited disease, characterized by defective glycogen metabolism. Affected dog may not grow fast enough, and may have heat intolerance, bruising, hypoglycemia, enlarged liver, swollen belly and weak muscle tone.		

AGSCTD056	**Hemophilia B**	F9	Autosomal Recessive
	Hemophilia is an inherited hematological disorder characterized by uncontrolled bleeding due to absence of clotting factor IX in blood and affected dogs suffer from spontaneous and prolonged bleeding.		
AGSCTD063	**Factor VII Deficiency**	F7	Autosomal Recessive
	Factor VII deficiency, is an inherited blood clotting disorder, characterized by a deficiency or reduced activity of clotting factor VII protein with dogs exhibiting uncontrolled bleeding episodes with varying age of onset.		
AGSCTD082	**Scott Syndrome**	ANO6	Autosomal Recessive
	Scott Syndrome is an inherited blood disease, characterized by excessive bleeding due to clotting factor deficiency. The symptoms include spontaneous, non-traumatic bleeding, nosebleeds, bruising, bleeding in joints, and excessive bleeding during teething.		
AGSCTD087	**Hypocatalasia**	CAT	Autosomal
	Hypocatalasia is an inherited disease, due to deficiency of catalase enzyme activity in red blood cells. It results in progressive gangrene in the oral cavity with oral ulcers, difficulty in eating and frequent infections.		
AGSCTD117	**Bleeding disorder due to P2RY12**	P2RY12	Autosomal Recessive
	Bleeding disorder due to P2RY12 is characterized by excessive bleeding due to defective platelet function. Although spontaneous bleeding is uncommon, excessive bleeding following a trauma or surgery is reported in affected dogs.		
AGSCTD120	**Pyruvate kinase deficiency of erythrocyte**	PKLR	Autosomal Recessive
	Pyruvate kinase deficiency of erythrocyte is an inherited disease, characterized by decreased number of red blood cells leading to inadequate supply of oxygen. Dog shows fatigue, lethargy, recurrent gallstones, jaundice and pale skin.		
AGSCTD121	**Pyruvate kinase deficiency of erythrocyte**	PKLR	Autosomal Recessive
	Pyruvate kinase deficiency of erythrocyte is an inherited disease, characterized by decreased number of red blood cells leading to inadequate supply of oxygen. Dog shows fatigue, lethargy, recurrent gallstones, jaundice and pale skin.		
AGSCTD122	**Pyruvate kinase deficiency of erythrocyte**	PKLR	Autosomal Recessive
	Pyruvate kinase deficiency of erythrocyte is an inherited disease, characterized by decreased number of red blood cells leading to inadequate supply of oxygen. Dog shows fatigue, lethargy, recurrent gallstones, jaundice and pale skin.		
AGSCTD126	**Polycythemia**	JAK2	Autosomal Dominant
	Polycythemia is an inherited blood disorder characterized by abnormal increase in count of red blood cells (RBC). The affected dog shows enlarged liver and spleen, fatigue, dizziness, shortness of breath, vision problems, night sweats and flushed face.		
AGSCTD129	**Prekallikrein Deficiency**	KLKB1	Autosomal Recessive
	Prekallikrein deficiency is a rare inherited disorder characterized by prolonged clotting time due to deficient Prekallikrein protein. Affected dogs may show presence of blood in the urine, gastrointestinal hemorrhage and excessive postoperative bleeding can occur.		

DISORDERS -HEMATOLOGICAL(CONTD.)

AGSCTD141	**Thrombasthenia**	ITGA2B	Autosomal Recessive
	Thrombasthenia is an inherited blood disorder characterized by excessive bleeding due to defective platelet aggregation. Symptoms include hemorrhage, skin bruising, gum and nose bleeding, blood in urine and faeces or black faeces.		
AGSCTD142	**Thrombasthenia**	ITGA2B	Autosomal Recessive
	Thrombasthenia is an inherited blood disorder characterized by excessive bleeding due to defective platelet aggregation. Symptoms include hemorrhage, skin bruising, gum and nose bleeding, blood in urine and faeces or black faeces.		
AGSCTD143	**Thrombocytopaenia**	TUBB1	Autosomal Recessive
	Thrombocytopaenia is charecterized by excessive bleeding due to low platelet count. Affected dogs may show fever, lethargy, loss of appetite, weakness, heart murmur, bleeding of gums, skin bruises and blood in urine.		
AGSCTD144	**Thrombopathia**	RASGRP1	Autosomal Recessive
	Thrombopathia is characterized by excessive bleeding with abnormal platelets. The decreased platelet function can cause dogs to bleed excessively during an injury or surgery. Other symptoms are gingival bleeding and hematomas.		
AGSCTD145	**Thrombopathia**	RASGRP1	Autosomal Recessive
	Thrombopathia is characterized by excessive bleeding with abnormal platelets. The decreased platelet function can cause dogs to bleed excessively during an injury or surgery. Other symptoms are gingival bleeding and hematomas.		
AGSCTD146	**Trapped Neutrophil Syndrome**	VPS13B	Autosomal Recessive
	Trapped Neutrophil Syndrome is an inherited hematological disease where neutrophils are affected and ability to fight illnesses decreases. Along with lack of immunity, slow weight gain and slow growth, dogs have depression, swollen joints and failure to thrive.		
AGSCTD149	**Von Willebrand disease I**	VWF	Autosomal Dominant and Recessive
	Von Willebrand disease III is an inherited bleeding disorder characterized by total or near-total absence of Willebrand factor (VWF) in the plasma, leading to a prolonged and excessive bleeding after injury, bleeding gums, blood in stool and skin bruising.		
AGSCTD150	**Von Willebrand disease II**	VWF	Autosomal Recessive
	Von Willebrand disease III is an inherited bleeding disorder characterized by total or near-total absence of Willebrand factor (VWF) in the plasma, leading to a prolonged and excessive bleeding after injury, bleeding gums, blood in stool and skin bruising.		
AGSCTD151	**Von Willebrand Disease III**	VWF	Autosomal Recessive
	Von Willebrand disease III is an inherited bleeding disorder characterized by total or near-total absence of Willebrand factor (VWF) in the plasma, leading to a prolonged and excessive bleeding after injury, bleeding gums, blood in stool and skin bruising.		
AGSCTD152	**Von Willebrand Disease III**	VWF	Autosomal Recessive
	Von Willebrand disease III is an inherited bleeding disorder characterized by total or near-total absence of Willebrand factor (VWF) in the plasma, leading to a prolonged and excessive bleeding after injury, bleeding gums, blood in stool and skin bruising.		

AGSCTD064	**Gallbladder Mucoceles**	ABCB4	Autosomal Dominant, Complex
	Gall bladder Mucocele results when there is a blockage within the fundus of gallbladder resulting in episodic symptoms of lack of appetite, vomiting and abdominal pain. Severe cases show gall bladder extension, necrosis of gallbladder and peritonitis.		
AGSCTD091	**Intestinal cobalamin malabsorption, CUBN-related**	CUBN	Autosomal Recessive
	Intestinal cobalamin malabsorption is an inherited disease affecting bone marrow and gastric system. It is characterized by failure to thrive, neutropenia, decreased serum cobalamin, non-regenerative anemia, methylmalonic aciduria, and homocysteinemia.		
AGSCTD092	**Intestinal cobalamin malabsorption, CUBN-related**	CUBN	Autosomal Recessive
	Intestinal cobalamin malabsorption is an inherited disease affecting bone marrow and gastric system. It is characterized by failure to thrive, neutropenia, decreased serum cobalamin, non-regenerative anemia, methylmalonic aciduria, and homocysteinemia.		

AGSCTD070	**Muscular Dystrophy, Duchenne type**	DMD	X-linked Recessive
	Duchene Muscular dystrophy is an inherited muscle disease, characterized by stiff gait, weakening of muscles, difficulty swallowing, progressive generalized weakness, plantigrade stance, and muscle atrophy.		
AGSCTD096	**Myotonia**	CLCN1	Autosomal Recessive
	Myotonia is an inherited muscle disease, characterized abnormal muscles. Affected dogs exhibit a stiff gait, experience trouble when rising, often suffer swollen tongues and may have difficulty swallowing		
AGSCTD097	**Muscular dystrophy, Duchenne type**	DMD	X-linked Recessive
	Duchene Muscular dystrophy is an inherited muscle disease, characterized by stiff gait, weakening of muscles, difficulty swallowing, progressive generalized weakness, plantigrade stance, and muscle atrophy.		
AGSCTD101	**Muscular hypertrophy**	MSTN	Autosomal Recessive
	Muscular hypertrophy, also called as Bully Whippet – Whippet Double Muscling, is an inherited muscular disorder characterized by increased muscle mass with broad chest, strongly developed leg and neck musculature		
AGSCTD103	**Myasthenic syndrome, congenital**	CHAT	Autosomal Recessive
	Congenital Myasthenic is an inherited muscle disease, characterized by severe generalized skeletal muscle weakness and fatigue, usually induced by exercise. Puppies also show inability to close the eyes, excessive drooling and difficulty breathing.		
AGSCTD104	**Myotonia**	CLCN1	Autosomal Recessive
	Myotonia is an inherited muscle disease, characterized by muscle stiffness and weakness. Affected dogs exhibit a stiff gait, experience trouble when rising, suffer with swollen tongues and may have difficulty swallowing.		
AGSCTD105	**Myopathy, Great Dane**	BIN1	Autosomal Recessive
	Myopathy is an inherited muscle disease, characterized by severe, progressive muscle atrophy in puppies. Symptoms include general weakness, exercise intolerance, muscle pain, limited joint movement and ventroflexion of head and neck.		
AGSCTD106	**Myotubular Myopathy**	MTM1	X-linked Recessive
	Myotubular Myopathy manifests with severe, progressive muscle atrophy in puppies. Symptoms include general weakness, exercise intolerance, muscle pain, limited joint movement and ventroflexion of head and neck.		

AGSCTD042	**Renal Cystadenocarcinoma & Nodular Dermatofibrosis**	FLCN	Autosomal Dominant
	Renal Cystadenocarcinoma and Nodular Dermatofibrosis is a genetic disorder, which is inherited, that can lead to uterine, kidney, and dermal cancer with onset usually at around 6yrs		
AGSCTD085	**Polycystic Kidney Disease**	PKD1	Autosomal Dominant
	Polycystic Kidney Disease is an inherited kidney disease, with high susceptibility for formation of cysts in kidneys. Symptoms include enlarged kidneys, thirst, frequent urination, lethargy, lack of appetite, weight loss, vomiting and high blood pressure.		
AGSCTD086	**Urolithiasis**	SLC2A9	Autosomal Recessive
	Urolithiasis is an inherited renal condition, with high susceptibility for formation of bladder stones (calculi). Symptoms include frequent urination, blood in the urine, lethargy, depression, reduced appetite, pain, vomiting and difficulty in urination.		
AGSCTD113	**Nephritis, X-linked**	COl4A5	X-linked Recessive
	Nephritis is a renal defect caused by defective collagen. Affected males exhibit proteinuria and develop rapidly progressive renal failure, which is usually fatal. Carrier females rarely exhibit mild symptoms.		